

## EURO-NMD Registry DEMO (PID: 80)

03.04.2024 09:47

Instruments		Events	Languages						
#	Variable / Field Name	Field Label <i>Field Note</i>	Field Attributes (Field Type, Validation, Choices, Calculations, etc.)						
<b>Instrument: Consent (consent)</b>									
Active languages: None									
1	[record_id]	Patient ID	text Field Annotation: @HIDDEN						
2	[consent_note1]	The entry of data is only possible with the written informed consent of the patient.	descriptive						
3	[consent_note2]	Patient ID [record-name] Please document the Patient ID within your local patient file, otherwise you may not be able to identify the patient at a later point. No personally identifying information about the patient is stored in this registry.	descriptive						
4	[consent_h1]	CONSENT	descriptive						
5	[date_of_consent]	Date of consent yyyy-mm-dd	text (date_ymd), Required						
6	[has_consent_commercial]	"I CONSENT that my pseudonymized data [my child/the patient pseudonymized data] may also be used to support commercial projects aimed to improve healthcare."	radio, Required <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
7	[has_consent_non_eu]	"I CONSENT that my pseudonymized data [my child/the patient pseudonymized data] may be transferred to non-EU countries, in compliance with GDPR, to support projects aimed to improve healthcare."	radio, Required <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
8	[has_consent_linkage]	"I CONSENT that my pseudonymized data [my child/the patient pseudonymized data] may be linked to existing databases/registries to improve healthcare."	radio, Required <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
9	[has_consent_be_contacted]	Agreement to be contacted for research purposes "I WOULD LIKE TO BE CONTACTED by my medical doctor [my child/the patient medical doctor] about any research project and/or clinical study related to my condition [my child/the patient condition]." <i>Patient's permission exists for being contacted for research purposes (Element 7.1. in the Set of common data elements for Rare Diseases Registration (link))</i>	radio, Required <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
10	[has_consent_email] Show the field ONLY if: [has_consent_be_contacted] = 'yes'	"I CONSENT to provide my email address and be contacted to complete questionnaires about my health [the health of my child/the patient] and receive news and updates about the registry."	radio, Required <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
11	[consent_complete]	Section Header: <i>Form Status</i> Complete?	dropdown <table border="1"> <tr> <td>0</td> <td>Incomplete</td> </tr> <tr> <td>1</td> <td>Unverified</td> </tr> <tr> <td>2</td> <td>Complete</td> </tr> </table>	0	Incomplete	1	Unverified	2	Complete
0	Incomplete								
1	Unverified								
2	Complete								
<b>Instrument: Baseline (baseline)</b>									
Active languages: None									
12	[disease_group]	disease_group	dropdown <table border="1"> <tr> <td>enrolment_and_firs_arm_1</td> <td>Neuropathies</td> </tr> <tr> <td>enrolment_and_firs_arm_2</td> <td>Myopathies</td> </tr> </table>	enrolment_and_firs_arm_1	Neuropathies	enrolment_and_firs_arm_2	Myopathies		
enrolment_and_firs_arm_1	Neuropathies								
enrolment_and_firs_arm_2	Myopathies								

			<table border="1"> <tr> <td>enrolment_and_firs_arm_3</td> <td>Mitochondrial Diseases</td> </tr> <tr> <td>enrolment_and_firs_arm_4</td> <td>Neuromuscular Junction Disorders</td> </tr> <tr> <td>enrolment_and_firs_arm_5</td> <td>Motoneuron Diseases</td> </tr> <tr> <td>enrolment_and_firs_arm_6</td> <td>Undiagnosed Cases</td> </tr> </table> <p>Field Annotation: @READONLY @DEFAULT='[first-event-name]'</p>	enrolment_and_firs_arm_3	Mitochondrial Diseases	enrolment_and_firs_arm_4	Neuromuscular Junction Disorders	enrolment_and_firs_arm_5	Motoneuron Diseases	enrolment_and_firs_arm_6	Undiagnosed Cases						
enrolment_and_firs_arm_3	Mitochondrial Diseases																
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enrolment_and_firs_arm_5	Motoneuron Diseases																
enrolment_and_firs_arm_6	Undiagnosed Cases																
13	[baseline_h1]	BASELINE	descriptive														
14	[internat_pat_id]	International patient identifier <i>Element 1.1. in the Set of common data elements for Rare Diseases Registration (link); more information available here</i>	text, Required Field Annotation: @HIDDEN @HIDDEN-PDF														
15	[sex_at_birth]	Sex at birth <i>Patient's sex at birth (Element 2.2. in the Set of common data elements for Rare Diseases Registration (link))</i>	radio, Required <table border="1"> <tr> <td>female</td> <td>Female</td> </tr> <tr> <td>male</td> <td>Male</td> </tr> <tr> <td>undetermined</td> <td>Undetermined</td> </tr> <tr> <td>foetus_unknown</td> <td>Foetus (Unknown)</td> </tr> </table>	female	Female	male	Male	undetermined	Undetermined	foetus_unknown	Foetus (Unknown)						
female	Female																
male	Male																
undetermined	Undetermined																
foetus_unknown	Foetus (Unknown)																
16	[date_of_birth]	Date of birth yyyy-mm-dd <i>Patient's date of birth (Element 2.1. in the Set of common data elements for Rare Diseases Registration (link))</i>	text (date_ymd), Required, Identifier														
17	[date_of_visit_bl]	Date of baseline yyyy-mm-dd	text (date_ymd), Required														
18	[age_at_baseline]	Age at baseline (years) <i>Age of patient (years)</i>	calc Calculation: rounddown(datediff([date_of_birth], [date_of_visit_bl], "y"))														
19	[age_at_baseline_months]	Age at baseline (months) <i>Age of patient (months)</i>	calc Calculation: rounddown(datediff([date_of_birth], [date_of_visit_bl], "M"))														
20	[date_of_first_contact_known]	Section Header: DISEASE HISTORY Date of first contact with specialised centre known <i>Element 4.1. in the Set of common data elements for Rare Diseases Registration (link)</i>	radio, Required <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> </table> <p>Field Annotation: @DEFAULT = 'yes'</p>	yes	Yes	no	No										
yes	Yes																
no	No																
21	[date_of_first_contact]	Date of first contact with specialised centre yyyy-mm-dd <i>Date of first contact with specialised centre (Element 4.1. in the Set of common data elements for Rare Diseases Registration (link)). If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.</i>	text (date_ymd), Required														
22	[age_at_onset]	Age at onset of the neuromuscular disease <i>Age at which symptoms/signs first appeared (Element 5.1. in the Set of common data elements for Rare Diseases Registration (link)) Definitions: - Antenatal onset (accessed: 25.04.2022) - Congenital onset (accessed: 25.04.2022) - Neonatal onset (accessed: 25.04.2022) - Pediatric onset (accessed: 25.04.2022) - Adult onset (accessed: 25.04.2022)</i>	radio, Required <table border="1"> <tr> <td>date</td> <td>Date</td> </tr> <tr> <td>antenatal</td> <td>Antenatal onset</td> </tr> <tr> <td>congenital</td> <td>Congenital onset</td> </tr> <tr> <td>neonatal</td> <td>Neonatal onset</td> </tr> <tr> <td>pediatric</td> <td>Pediatric onset</td> </tr> <tr> <td>adult</td> <td>Adult onset</td> </tr> <tr> <td>undetermined</td> <td>Undetermined</td> </tr> </table> <p>Field Annotation: @DEFAULT = 'date'</p>	date	Date	antenatal	Antenatal onset	congenital	Congenital onset	neonatal	Neonatal onset	pediatric	Pediatric onset	adult	Adult onset	undetermined	Undetermined
date	Date																
antenatal	Antenatal onset																
congenital	Congenital onset																
neonatal	Neonatal onset																
pediatric	Pediatric onset																
adult	Adult onset																
undetermined	Undetermined																
23	[date_for_age_at_onset]	Date of onset of the neuromuscular disease yyyy-mm-dd <i>If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year. (Element 5.1. in the Set of common data elements for Rare Diseases Registration (link))</i>	text (date_ymd), Required														
24	[consanguinity]	Consanguinity	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown								
yes	Yes																
no	No																
unknown	Unknown																

25	[affected_family]	Affected family members	<table border="1"> <tr><td colspan="2">radio</td></tr> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	radio		yes	Yes	no	No	unknown	Unknown																
radio																											
yes	Yes																										
no	No																										
unknown	Unknown																										
26	[which_affected_family] Show the field ONLY if: [affected_family] = 'yes'	Affected family members (Click all that apply)	<table border="1"> <tr><td colspan="2">checkbox, Required</td></tr> <tr><td>mother</td><td>which_affected_family__mother</td></tr> <tr><td>father</td><td>which_affected_family__father</td></tr> <tr><td>uncle</td><td>which_affected_family__uncle</td></tr> <tr><td>aunt</td><td>which_affected_family__aunt</td></tr> <tr><td>grandfather</td><td>which_affected_family__grandfather</td></tr> <tr><td>grandmother</td><td>which_affected_family__grandmother</td></tr> <tr><td>sister</td><td>which_affected_family__sister</td></tr> <tr><td>brother</td><td>which_affected_family__brother</td></tr> <tr><td>son</td><td>which_affected_family__son</td></tr> <tr><td>daughter</td><td>which_affected_family__daughter</td></tr> <tr><td>other</td><td>which_affected_family__other</td></tr> </table>	checkbox, Required		mother	which_affected_family__mother	father	which_affected_family__father	uncle	which_affected_family__uncle	aunt	which_affected_family__aunt	grandfather	which_affected_family__grandfather	grandmother	which_affected_family__grandmother	sister	which_affected_family__sister	brother	which_affected_family__brother	son	which_affected_family__son	daughter	which_affected_family__daughter	other	which_affected_family__other
checkbox, Required																											
mother	which_affected_family__mother																										
father	which_affected_family__father																										
uncle	which_affected_family__uncle																										
aunt	which_affected_family__aunt																										
grandfather	which_affected_family__grandfather																										
grandmother	which_affected_family__grandmother																										
sister	which_affected_family__sister																										
brother	which_affected_family__brother																										
son	which_affected_family__son																										
daughter	which_affected_family__daughter																										
other	which_affected_family__other																										
27	[had_reduced_foetal_movement] Show the field ONLY if: (([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6) AND [age_at_baseline] < 16)	Reduced foetal movements	<table border="1"> <tr><td colspan="2">radio</td></tr> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	radio		yes	Yes	no	No	unknown	Unknown																
radio																											
yes	Yes																										
no	No																										
unknown	Unknown																										
28	[had_polyhydramnios] Show the field ONLY if: (([arm-number]=2 OR [arm-number]=4)	Polyhydramnios	<table border="1"> <tr><td colspan="2">radio</td></tr> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	radio		yes	Yes	no	No	unknown	Unknown																
radio																											
yes	Yes																										
no	No																										
unknown	Unknown																										
29	[s4_had_symp_fluc] Show the field ONLY if: (([arm-number]=4 OR [arm-number]=6)	Symptom fluctuation	<table border="1"> <tr><td colspan="2">radio</td></tr> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	radio		yes	Yes	no	No	unknown	Unknown																
radio																											
yes	Yes																										
no	No																										
unknown	Unknown																										
30	[s4_had_exacerb_infection] Show the field ONLY if: [arm-number]=4	Exacerbation with infection	<table border="1"> <tr><td colspan="2">radio</td></tr> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	radio		yes	Yes	no	No	unknown	Unknown																
radio																											
yes	Yes																										
no	No																										
unknown	Unknown																										
31	[s4_had_change_sev_puberty] Show the field ONLY if: [arm-number]=4 AND [age_at_baseline] >= 12	Change of severity in puberty	<table border="1"> <tr><td colspan="2">radio</td></tr> <tr><td>improvement</td><td>Improvement</td></tr> <tr><td>deterioration</td><td>Deterioration</td></tr> <tr><td>no_change</td><td>No change</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> <tr><td>not_applicable</td><td>Not applicable</td></tr> </table>	radio		improvement	Improvement	deterioration	Deterioration	no_change	No change	unknown	Unknown	not_applicable	Not applicable												
radio																											
improvement	Improvement																										
deterioration	Deterioration																										
no_change	No change																										
unknown	Unknown																										
not_applicable	Not applicable																										
32	[s4_had_change_sev_menstr] Show the field ONLY if: [arm-number]=4 AND ([sex_at_birth] = 'female' AND [age_at_baseline] >= 12)	Change of severity with menstrual cycle	<table border="1"> <tr><td colspan="2">radio</td></tr> <tr><td>improvement</td><td>Improvement</td></tr> <tr><td>deterioration</td><td>Deterioration</td></tr> <tr><td>no_change</td><td>No change</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> <tr><td>not_applicable</td><td>Not applicable</td></tr> </table>	radio		improvement	Improvement	deterioration	Deterioration	no_change	No change	unknown	Unknown	not_applicable	Not applicable												
radio																											
improvement	Improvement																										
deterioration	Deterioration																										
no_change	No change																										
unknown	Unknown																										
not_applicable	Not applicable																										
33	[s4_had_change_sev_horm] Show the field ONLY if:	Change of severity with hormonal therapy	<table border="1"> <tr><td colspan="2">radio</td></tr> <tr><td>improvement</td><td>Improvement</td></tr> <tr><td>deterioration</td><td>Deterioration</td></tr> </table>	radio		improvement	Improvement	deterioration	Deterioration																		
radio																											
improvement	Improvement																										
deterioration	Deterioration																										

	[arm-number]=4 AND ([sex_at_birth] = 'female' AND [age_at_baseline] >= 12)		<table border="1"> <tr> <td>no_change</td> <td>No change</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> <tr> <td>not_applicable</td> <td>Not applicable</td> </tr> </table>	no_change	No change	unknown	Unknown	not_applicable	Not applicable				
no_change	No change												
unknown	Unknown												
not_applicable	Not applicable												
34	[s4_had_change_sev_preg] Show the field ONLY if: [arm-number]=4 AND ([sex_at_birth] = 'female' AND [age_at_baseline] >= 12)	Change of severity with pregnancy or childbirth	radio <table border="1"> <tr> <td>improvement</td> <td>Improvement</td> </tr> <tr> <td>deterioration</td> <td>Deterioration</td> </tr> <tr> <td>no_change</td> <td>No change</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> <tr> <td>not_applicable</td> <td>Not applicable</td> </tr> </table>	improvement	Improvement	deterioration	Deterioration	no_change	No change	unknown	Unknown	not_applicable	Not applicable
improvement	Improvement												
deterioration	Deterioration												
no_change	No change												
unknown	Unknown												
not_applicable	Not applicable												
35	[diagnosis_rd] Show the field ONLY if: [arm-number]=1 OR [arm-number]=2 OR [arm-number]=3 OR [arm-number]=4 OR [arm-number]=5	Section Header: <i>DIAGNOSIS</i> Diagnosis of the neuromuscular disease <i>Please answer "Neuromuscular disease diagnosed" if this patient has a known clinical diagnosis, even if the genetic cause has not been established. This is then the diagnosis retained by the specialised centre (Element 6.1. in the Set of common data elements for Rare Diseases Registration (link))</i>	radio, Required <table border="1"> <tr> <td>diagnosed</td> <td>Neuromuscular disease diagnosed</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table> Field Annotation: @DEFAULT = 'diagnosed'	diagnosed	Neuromuscular disease diagnosed	unknown	Unknown						
diagnosed	Neuromuscular disease diagnosed												
unknown	Unknown												
36	[diagnosis_rd_orpha] Show the field ONLY if: [diagnosis_rd] = 'diagnosed'	Diagnosis of the neuromuscular disease (ORPHAcode) <i>Diagnosis retained by the specialised centre (Element 6.1. in the Set of common data elements for Rare Diseases Registration (link))</i>	text, Required <table border="1"> <tr> <td>BIOPORTAL:ORDO</td> <td>BIOPORTAL:ORDO</td> </tr> </table>	BIOPORTAL:ORDO	BIOPORTAL:ORDO								
BIOPORTAL:ORDO	BIOPORTAL:ORDO												
37	[diagnosis_rd_orpha_value] Show the field ONLY if: [diagnosis_rd] = 'diagnosed'	diagnosis_rd_orpha_value	text Field Annotation: @HIDDEN @HIDDEN-PDF @READONLY @CALCTEXT(if([isblankormissingcode]([diagnosis_rd_orpha_mid]([diagnosis_rd_orpha], 7, length([diagnosis_rd_orpha])), "")))										
38	[diagnosis_rd_orpha_uri] Show the field ONLY if: [diagnosis_rd] = 'diagnosed'	diagnosis_rd_orpha_uri	text Field Annotation: @HIDDEN @HIDDEN-PDF @READONLY @CALCTEXT(if([isblankormissingcode]([diagnosis_rd_orpha_concat]('http://www.orpha.net/ORDO/Orphanet_', [diagnosis_rd_orpha_value]), "")))										
39	[diagnosis_rd_orpha_disp] Show the field ONLY if: [diagnosis_rd] = 'diagnosed'	Diagnosis of the neuromuscular disease (ORDO-URI): [diagnosis_rd_orpha_uri]	descriptive										
40	[pabpn1_v_report_def]	pabpn1_v_report_def	text Field Annotation: @READONLY @CALCTEXT(if([diagnosis_rd_orpha:value]='ORPHA:270' 'distinct_number', "")) @HIDDEN @HIDDEN-PDF										
41	[dmpk_v_report_def]	dmpk_v_report_def	text Field Annotation: @READONLY @CALCTEXT(if([diagnosis_rd_orpha:value]='ORPHA:273' 'distinct_number', "")) @HIDDEN @HIDDEN-PDF										
42	[cnbp_v_report_def]	cnbp_v_report_def	text Field Annotation: @READONLY @CALCTEXT(if([diagnosis_rd_orpha:value]='ORPHA:606' 'distinct_number', "")) @HIDDEN @HIDDEN-PDF										
43	[ar_v1_report_def]	ar_v1_report_def	text Field Annotation: @READONLY @CALCTEXT(if([diagnosis_rd_orpha:value]='ORPHA:431' 'distinct_number', "")) @HIDDEN @HIDDEN-PDF										
44	[age_at_diagnosis] Show the field ONLY if: [diagnosis_rd] = 'diagnosed'	Age at diagnosis of the neuromuscular disease <i>Age at which diagnosis was made (Element 5.2. in the Set of common data elements for Rare Diseases Registration (link))</i>	radio, Required <table border="1"> <tr> <td>antenatal</td> <td>Antenatal (Prenatal)</td> </tr> <tr> <td>postnatal</td> <td>Postnatal</td> </tr> <tr> <td>date</td> <td>Date</td> </tr> <tr> <td>undetermined</td> <td>Undetermined</td> </tr> </table> Field Annotation: @DEFAULT = 'date'	antenatal	Antenatal (Prenatal)	postnatal	Postnatal	date	Date	undetermined	Undetermined		
antenatal	Antenatal (Prenatal)												
postnatal	Postnatal												
date	Date												
undetermined	Undetermined												
45	[date_for_age_at_diagno]	Date of diagnosis of the neuromuscular disease yyyy-mm-	text (date_ymd), Required										

	[ sis ] Show the field ONLY if: [age_at_diagnosis]='date'	dd <i>If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year. (Element 5.2. in the Set of common data elements for Rare Diseases Registration (link))</i>											
46	[ diagnosis_rd_method_known ] Show the field ONLY if: [diagnosis_rd] = 'diagnosed'	Diagnostic method known	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> </table> Field Annotation: @DEFAULT = 'yes'	yes	Yes	no	No						
yes	Yes												
no	No												
47	[ diagnosis_rd_method ] Show the field ONLY if: [diagnosis_rd] = 'diagnosed' AND [diagnosis_rd_method_known] = 'yes'	How was the diagnosis established? (select all that apply)	checkbox <table border="1"> <tr> <td>clinical_history</td> <td>diagnosis_rd_method__clinical_h</td> </tr> <tr> <td>family_history</td> <td>diagnosis_rd_method__family_hi</td> </tr> <tr> <td>clinical_exam</td> <td>diagnosis_rd_method__clinical_e</td> </tr> <tr> <td>genetic_screening</td> <td>diagnosis_rd_method__genetic_s</td> </tr> <tr> <td>other</td> <td>diagnosis_rd_method__other</td> </tr> </table>	clinical_history	diagnosis_rd_method__clinical_h	family_history	diagnosis_rd_method__family_hi	clinical_exam	diagnosis_rd_method__clinical_e	genetic_screening	diagnosis_rd_method__genetic_s	other	diagnosis_rd_method__other
clinical_history	diagnosis_rd_method__clinical_h												
family_history	diagnosis_rd_method__family_hi												
clinical_exam	diagnosis_rd_method__clinical_e												
genetic_screening	diagnosis_rd_method__genetic_s												
other	diagnosis_rd_method__other												
48	[ diagnosis_rd_2_exists ] Show the field ONLY if: [diagnosis_rd] = 'diagnosed'	Section Header: <i>Second diagnosis</i> Does the patient have a second neuromuscular diagnosis?	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown				
yes	Yes												
no	No												
unknown	Unknown												
49	[ diagnosis_rd_2_orpha ] Show the field ONLY if: [diagnosis_rd_2_exists] = 'yes'	Diagnosis of the second neuromuscular disease (ORPHAcode)	text <table border="1"> <tr> <td>BIOPORTAL:ORDO</td> <td>BIOPORTAL:ORDO</td> </tr> </table>	BIOPORTAL:ORDO	BIOPORTAL:ORDO								
BIOPORTAL:ORDO	BIOPORTAL:ORDO												
50	[ s3_main_mito_phenot_known ] Show the field ONLY if: [arm-number] = 3	Section Header: <i>Disease specific</i> Main mitochondrial phenotype known	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> </table> Field Annotation: @DEFAULT = if([arm-number] = 3, 'yes', '')	yes	Yes	no	No						
yes	Yes												
no	No												
51	[ s3_main_mito_phenot ] Show the field ONLY if: [arm-number]=3 AND [s3_main_mito_phenot_known]='yes'	Main mitochondrial phenotype	text, Required <table border="1"> <tr> <td>BIOPORTAL:HP</td> <td>BIOPORTAL:HP</td> </tr> </table>	BIOPORTAL:HP	BIOPORTAL:HP								
BIOPORTAL:HP	BIOPORTAL:HP												
52	[ s4_tensilon_test_resp ] Show the field ONLY if: [arm-number]=4	Tensilon/prostigmin test response	radio <table border="1"> <tr> <td>improvement</td> <td>Improvement</td> </tr> <tr> <td>deterioration</td> <td>Deterioration</td> </tr> <tr> <td>no_change</td> <td>No change</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	improvement	Improvement	deterioration	Deterioration	no_change	No change	not_done	Not done		
improvement	Improvement												
deterioration	Deterioration												
no_change	No change												
not_done	Not done												
53	[ s4_chest_ct_findings ] Show the field ONLY if: [arm-number]=4	Chest Computed Tomography (CT) findings	radio <table border="1"> <tr> <td>thymoma</td> <td>Thymoma</td> </tr> <tr> <td>residual_thymus_tissue</td> <td>Residual thymus tissue</td> </tr> <tr> <td>absent_thymus</td> <td>Absent thymus</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	thymoma	Thymoma	residual_thymus_tissue	Residual thymus tissue	absent_thymus	Absent thymus	not_done	Not done		
thymoma	Thymoma												
residual_thymus_tissue	Residual thymus tissue												
absent_thymus	Absent thymus												
not_done	Not done												
54	[ s5_sma_presymptomatic ] Show the field ONLY if: [diagnosis_rd_orpha]='ORPHA:70'	Spinal muscular atrophy: Presymptomatic?	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown				
yes	Yes												
no	No												
unknown	Unknown												
55	[ s5 CGI-S score ] Show the field ONLY if:	Clinical Global Impression of Severity (CGI-S)	radio <table border="1"> <tr> <td>normal</td> <td>Normal, not at all ill</td> </tr> </table>	normal	Normal, not at all ill								
normal	Normal, not at all ill												

	[arm-number]=5		<table border="1"> <tr> <td>borderline_ill</td> <td>Borderline ill</td> </tr> <tr> <td>mildly_ill</td> <td>Mildly ill</td> </tr> <tr> <td>moderately_ill</td> <td>Moderately ill</td> </tr> <tr> <td>markedly_ill</td> <td>Markedly ill</td> </tr> <tr> <td>severly_ill</td> <td>Severely ill</td> </tr> <tr> <td>most_extreme</td> <td>Among the most extremely ill patients</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	borderline_ill	Borderline ill	mildly_ill	Mildly ill	moderately_ill	Moderately ill	markedly_ill	Markedly ill	severly_ill	Severely ill	most_extreme	Among the most extremely ill patients	unknown	Unknown
borderline_ill	Borderline ill																
mildly_ill	Mildly ill																
moderately_ill	Moderately ill																
markedly_ill	Markedly ill																
severly_ill	Severely ill																
most_extreme	Among the most extremely ill patients																
unknown	Unknown																
56	<p>[s5_esco_present_form]</p> <p>Show the field ONLY if: [diagnosis_rd_orpha]='ORPHA:803' OR [diagnosis_rd_orpha]='ORPHA:357043' OR [diagnosis_rd_orpha]='ORPHA:275872' OR [diagnosis_rd_orpha]='ORPHA:300605'</p>	El Escorial presentation form	<p>radio</p> <table border="1"> <tr> <td>bulbar</td> <td>Bulbar</td> </tr> <tr> <td>limb</td> <td>Limb</td> </tr> <tr> <td>combined</td> <td>Combined</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	bulbar	Bulbar	limb	Limb	combined	Combined	unknown	Unknown						
bulbar	Bulbar																
limb	Limb																
combined	Combined																
unknown	Unknown																
57	<p>[s5_onset_weight_loss]</p> <p>Show the field ONLY if: [arm-number]=5</p>	>10% weight loss in 6 months prior to time of disease onset	<p>radio</p> <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>presymptomatic</td> <td>Presymptomatic (no disease onset so far at the time of this Baseline-form)</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	presymptomatic	Presymptomatic (no disease onset so far at the time of this Baseline-form)	unknown	Unknown						
yes	Yes																
no	No																
presymptomatic	Presymptomatic (no disease onset so far at the time of this Baseline-form)																
unknown	Unknown																
58	<p>[s5_onset_cognitive]</p> <p>Show the field ONLY if: [arm-number]=5</p>	Cognitive symptoms or signs in 6 months prior to time of disease onset	<p>radio</p> <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>presymptomatic</td> <td>Presymptomatic (no disease onset so far at the time of this Baseline-form)</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	presymptomatic	Presymptomatic (no disease onset so far at the time of this Baseline-form)	unknown	Unknown						
yes	Yes																
no	No																
presymptomatic	Presymptomatic (no disease onset so far at the time of this Baseline-form)																
unknown	Unknown																
59	<p>[s5_onset_behavioural]</p> <p>Show the field ONLY if: [arm-number]=5</p>	Behavioural symptoms or signs in 6 months prior to time of disease onset	<p>radio</p> <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>presymptomatic</td> <td>Presymptomatic (no disease onset so far at the time of this Baseline-form)</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	presymptomatic	Presymptomatic (no disease onset so far at the time of this Baseline-form)	unknown	Unknown						
yes	Yes																
no	No																
presymptomatic	Presymptomatic (no disease onset so far at the time of this Baseline-form)																
unknown	Unknown																
60	[bio_sample_available]	<p>Section Header: <i>BIOBANKING</i></p> <p>Biological sample available for research <i>Patient's biological sample available for research (Element 7.3. in the Set of common data elements for Rare Diseases Registration (link))</i></p>	<p>radio, Required</p> <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown								
yes	Yes																
no	No																
unknown	Unknown																
61	<p>[bio_sample_biobank]</p> <p>Show the field ONLY if: [bio_sample_available]='yes'</p>	<p>Biological sample stored in a biobank <i>Biological sample stored in a biobank (Element 7.4. in the Set of common data elements for Rare Diseases Registration (link))</i></p>	<p>radio, Required</p> <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown								
yes	Yes																
no	No																
unknown	Unknown																
62	<p>[link_to_biobank]</p> <p>Show the field ONLY if: [bio_sample_biobank] = 'yes'</p>	<p>Link to a biobank <i>Link to a biobank (Element 7.4. in the Set of common data elements for Rare Diseases Registration (link))</i></p>	notes, Required														
63	<p>[has_bulbar_weakness]</p> <p>Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)</p>	<p>Section Header: <i>MOBILITY AND GENERAL PHYSICAL FUNCTIONING</i></p> <p>Bulbar weakness</p>	<p>radio</p> <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown								
yes	Yes																
no	No																
unknown	Unknown																
64	[has_tongue_wasting]	Tongue wasting	radio														

	Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)		<table border="1"> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
65	[has_muscle_wasting] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)	Muscle wasting	radio <table border="1"> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
66	[has_ptosis] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)	Section Header: <i>Muscular Functioning: Eye</i> Ptosis	radio <table border="1"> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
67	[has_ophthalmoparesis] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)	Ophthalmoparesis	radio <table border="1"> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
68	[has_diplopia] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)	Diplopia	radio <table border="1"> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
69	[s4_has_episodic_apnoea] Show the field ONLY if: ([arm-number]=4 OR [arm-number]=6)	Section Header: <i>Respiratory / Ventilation</i> Episodic apnoea	radio <table border="1"> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
70	[s4_has_stridor] Show the field ONLY if: ([arm-number]=4 OR [arm-number]=6)	Stridor	radio <table border="1"> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
71	[s4_has_rec_resp_infection] Show the field ONLY if: ([arm-number]=4 OR [arm-number]=6)	Recurrent respiratory tract infection	radio <table border="1"> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
72	[has_resp_weakness] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)	Respiratory weakness	radio <table border="1"> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
73	[s5_has_resp_diff] Show the field ONLY if: [arm-number]=5	Respiratory difficulties	radio <table border="1"> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
74	[has_high_arch_palate] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)	Section Header: <i>Skeletal</i> High arched palate	radio <table border="1"> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
75	[has_rigid_spine] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)	Rigid spine	radio <table border="1"> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
76	[has_scapular_winging]	Scapular winging	radio <table border="1"> <tr><td></td><td></td></tr> </table>						

	Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)		<table border="1"> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	yes	Yes	no	No	unknown	Unknown												
yes	Yes																				
no	No																				
unknown	Unknown																				
77	[has_joint_contr] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=5 OR [arm-number]=6)	Joint contractures	radio <table border="1"> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	yes	Yes	no	No	unknown	Unknown												
yes	Yes																				
no	No																				
unknown	Unknown																				
78	[which_joint_contr] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=5 OR [arm-number]=6) AND [has_joint_contr] = 'yes'	Joint contractures (select all that apply)	checkbox <table border="1"> <tr><td>elbow</td><td>which_joint_contr__elbow</td><td>Elbow</td></tr> <tr><td>wrist</td><td>which_joint_contr__wrist</td><td>Wrist</td></tr> <tr><td>fingers</td><td>which_joint_contr__fingers</td><td>Fingers</td></tr> <tr><td>hip</td><td>which_joint_contr__hip</td><td>Hip</td></tr> <tr><td>knee</td><td>which_joint_contr__knee</td><td>Knee</td></tr> <tr><td>ankle</td><td>which_joint_contr__ankle</td><td>Ankle</td></tr> </table>	elbow	which_joint_contr__elbow	Elbow	wrist	which_joint_contr__wrist	Wrist	fingers	which_joint_contr__fingers	Fingers	hip	which_joint_contr__hip	Hip	knee	which_joint_contr__knee	Knee	ankle	which_joint_contr__ankle	Ankle
elbow	which_joint_contr__elbow	Elbow																			
wrist	which_joint_contr__wrist	Wrist																			
fingers	which_joint_contr__fingers	Fingers																			
hip	which_joint_contr__hip	Hip																			
knee	which_joint_contr__knee	Knee																			
ankle	which_joint_contr__ankle	Ankle																			
79	[has_arthrogryposis] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6) AND [has_joint_contr] = 'yes'	Arthrogryposis	radio <table border="1"> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	yes	Yes	no	No	unknown	Unknown												
yes	Yes																				
no	No																				
unknown	Unknown																				
80	[had_delay_recov_anesth] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)	Section Header: <i>TREATMENT</i> Delayed recovery from anesthetics	radio <table border="1"> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	yes	Yes	no	No	unknown	Unknown												
yes	Yes																				
no	No																				
unknown	Unknown																				
81	[s4_pyridost_treat_resp] Show the field ONLY if: [arm-number]=4	Pyridostigmine treatment response	radio <table border="1"> <tr><td>improvement</td><td>Improvement</td></tr> <tr><td>deterioration</td><td>Deterioration</td></tr> <tr><td>no_change</td><td>No change</td></tr> <tr><td>not_done</td><td>Not done</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	improvement	Improvement	deterioration	Deterioration	no_change	No change	not_done	Not done	unknown	Unknown								
improvement	Improvement																				
deterioration	Deterioration																				
no_change	No change																				
not_done	Not done																				
unknown	Unknown																				
82	[baseline_desc2]	Section Header: <i>NOTE</i> To complete the baseline data collection, please remember to fill the following forms: - "Consent"- "Visit Data"- "Medication"- "Genetic Diagnosis" (or "Undiagnosed")	descriptive																		
83	[kpi_onset_contact_hcp_d]	Time from onset (first symptoms) to first HCP visit [days]	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([date_for_age_at_onset]) AND !isblankormissingcode([date_of_first_contact]) AND [date_for_age_at_onset] <= [date_of_first_contact], datediff([date_for_age_at_onset], [date_of_first_contact], "d","ymd", true), ""))																		
84	[kpi_onset_contact_hcp_y]	Time from onset (first symptoms) to first HCP visit [years]	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([date_for_age_at_onset]) AND !isblankormissingcode([date_of_first_contact]) AND [date_for_age_at_onset] <= [date_of_first_contact], datediff([date_for_age_at_onset], [date_of_first_contact], "y","ymd", true), ""))																		
85	[kpi_contact_hcp_onset_d]	Time from first HCP visit to onset (first symptoms) [days]	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([date_of_first_contact]) AND !isblankormissingcode([date_for_age_at_onset]) AND [date_of_first_contact] < [date_for_age_at_onset],																		



			datediff([date_of_first_contact], [date_for_age_at_onset], "d","ymd", true, ""))
86	[kpi_contact_hcp_onset_y]	Time from first HCP visit to onset (first symptoms) [years]	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([date_of_first_contact]) AND !isblankormissingcode([date_for_age_at_onset]) AND [date_of_first_contact] < [date_for_age_at_onset], datediff([date_of_first_contact], [date_for_age_at_onset], "y","ymd", true, ""))
87	[kpi_onset_diaclin_d]	Time from onset (first symptoms) to specific diagnosis [days]	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([date_for_age_at_onset]) AND !isblankormissingcode([date_for_age_at_diagnosis]) AND [date_for_age_at_onset] <= [date_for_age_at_diagnosis], datediff([date_for_age_at_onset], [date_for_age_at_diagnosis], "d","ymd", true, ""))
88	[kpi_onset_diaclin_y]	Time from onset (first symptoms) to specific diagnosis [years]	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([date_for_age_at_onset]) AND !isblankormissingcode([date_for_age_at_diagnosis]) AND [date_for_age_at_onset] <= [date_for_age_at_diagnosis], datediff([date_for_age_at_onset], [date_for_age_at_diagnosis], "y","ymd", true, ""))
89	[kpi_diaclin_onset_d]	Time from specific diagnosis to onset (first symptoms) [days]	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([date_for_age_at_onset]) AND !isblankormissingcode([date_for_age_at_diagnosis]) AND [date_for_age_at_diagnosis] < [date_for_age_at_onset], datediff([date_for_age_at_diagnosis], [date_for_age_at_onset], "d","ymd", true, ""))
90	[kpi_diaclin_onset_y]	Time from specific diagnosis to onset (first symptoms) [years]	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([date_for_age_at_onset]) AND !isblankormissingcode([date_for_age_at_diagnosis]) AND [date_for_age_at_diagnosis] < [date_for_age_at_onset], datediff([date_for_age_at_diagnosis], [date_for_age_at_onset], "y","ymd", true, ""))
91	[kpi_contact_hcp_diaclin_d]	Time from first HCP visit to specific diagnosis [days]	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([date_of_first_contact]) AND !isblankormissingcode([date_for_age_at_diagnosis]) AND [date_of_first_contact] <= [date_for_age_at_diagnosis], datediff([date_of_first_contact], [date_for_age_at_diagnosis], "d","ymd", true, ""))
92	[kpi_contact_hcp_diaclin_y]	Time from first HCP visit to specific diagnosis [years]	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([date_of_first_contact]) AND !isblankormissingcode([date_for_age_at_diagnosis]) AND [date_of_first_contact] <= [date_for_age_at_diagnosis], datediff([date_of_first_contact], [date_for_age_at_diagnosis], "y","ymd", true, ""))
93	[kpi_diaclin_contact_hcp_d]	Time from specific diagnosis to first HCP visit [days]	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([date_for_age_at_onset]) AND !isblankormissingcode([date_of_first_contact])

			AND [date_for_age_at_diagnosis] < [date_of_first_contact], datediff([date_for_age_at_diagnosis], [date_of_first_contact], "d","ymd", true, ""))						
94	[kpi_diaclin_contact_hcp_y]	Time from specific diagnosis to first HCP visit [years]	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([date_for_age_at_diagnosis]) AND !isblankormissingcode([date_of_first_contact]) AND [date_for_age_at_diagnosis] < [date_of_first_contact], datediff([date_for_age_at_diagnosis], [date_of_first_contact], "y","ymd", true, ""))						
95	[baseline_complete]	Section Header: <i>Form Status</i> Complete?	dropdown <table border="1"> <tr> <td>0</td> <td>Incomplete</td> </tr> <tr> <td>1</td> <td>Unverified</td> </tr> <tr> <td>2</td> <td>Complete</td> </tr> </table>	0	Incomplete	1	Unverified	2	Complete
0	Incomplete								
1	Unverified								
2	Complete								

**Instrument: Visit Data (visit\_data)**

Active languages: None

96	[visit_data_h1]	VISIT DATA	descriptive				
97	[date_of_visit_prev]	date_of_visit_prev: Value of date_of_visit in previous report. Is needed for piping for other fields (e.g. s5_hands_up_gain_d_known)	text Field Annotation: @CALCTEXT(if(starts_with([event-name], 'enrolment'), "", if(starts_with([event-name], 'visit') AND [current-instance]=1, [first-event-name] [date_of_visit], if(starts_with([event-name], 'visit') AND [current-instance]>1, [event-name] [date_of_visit][previous-instance], ""))) @READONLY @HIDDEN @HIDDEN-PDF				
98	[fu_instance_id]	fu_instance_id	text Field Annotation: @CALCTEXT([current-instance]) @READONLY @HIDDEN @HIDDEN-PDF				
99	[sex_at_birth_v]	sex_at_birth_v (needed for branching logic within this form)	text Field Annotation: @CALCTEXT(if(starts_with([event-name], 'visit'), [first-event-name][sex_at_birth], [event-name][sex_at_birth])) @READONLY @HIDDEN @HIDDEN-PDF				
100	[diagnosis_rd_orpha_v]	diagnosis_rd_orpha_v (needed for branching logic within this form)	text Field Annotation: @CALCTEXT(if(starts_with([event-name], 'visit'), [first-event-name] [diagnosis_rd_orpha], [event-name] [diagnosis_rd_orpha])) @READONLY @HIDDEN @HIDDEN-PDF				
101	[date_of_visit]	Section Header: <i>General</i> Date of visit yyyy-mm-dd	text (date_ymd), Required				
102	[age_at_visit]	Age of patient (years)	calc Calculation: if(starts_with([event-name], 'visit'), rounddown(datediff([first-event-name] [date_of_birth], [date_of_visit], "y")), rounddown(datediff([event-name][date_of_birth], [date_of_visit], "y")))				
103	[age_at_visit_months]	Age of patient (months)	calc Calculation: if(starts_with([event-name], 'visit'), rounddown(datediff([first-event-name] [date_of_birth], [date_of_visit], "M")), rounddown(datediff([event-name][date_of_birth], [date_of_visit], "M")))				
104	[body_height_cm_known]	Body Height (cm) known	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> </table> Field Annotation: @DEFAULT = 'yes'	yes	Yes	no	No
yes	Yes						
no	No						
105	[body_height_cm] Show the field ONLY if:	Body Height (cm)	text (number), Required				

	[body_height_cm_known] = 'yes'								
106	[body_weight_kg_known]	Body Weight (kg) known	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> </table>	yes	Yes	no	No		
yes	Yes								
no	No								
107	[body_weight_kg] Show the field ONLY if: [body_weight_kg_known] = 'yes'	Body Weight (kg)	text (number), Required						
108	[affil_registries_desc1]	Section Header: <i>Research</i> Participation in affiliated registries* *registries affiliated to the EURO-NMD Registry Hub for Rare Neuromuscular Diseases	descriptive						
109	[affil_registries_desc2] Show the field ONLY if: [pat_in_dmscope] = 'yes' OR [pat_in_cramp] = 'yes' OR [pat_in_ddp] = 'yes' OR [pat_in_smartcare] = 'yes' OR [pat_in_other_reg] = 'yes'	NOTE: The ID of the patient in an other registry may only be entered here if the patient has consented to this procedure in writing.	descriptive						
110	[pat_in_dmscope] Show the field ONLY if: [arm-number]=2	Patient registered in DM-Scope (French Observatory of Myotonic Dystrophies)	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table> Field Annotation: @DEFAULT='[event-name][pat_in_dmscope:value][previous-instance]'	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
111	[pat_id_dmscope] Show the field ONLY if: [arm-number]=2 AND [pat_in_dmscope]='yes'	Patient ID in DM-Scope <a href="http://www.dmscope.fr/french-registry-of-myotonic-dystrophies/">http://www.dmscope.fr/french-registry-of-myotonic-dystrophies/</a>	text Field Annotation: @DEFAULT='[event-name][pat_id_dmscope:value][previous-instance]'						
112	[pat_in_cramp] Show the field ONLY if: ([arm-number]=1 OR [arm-number]=2)	Patient registered in CRAMP (Computer Registry of All Myopathies and Polyneuropathies)	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table> Field Annotation: @DEFAULT='[event-name][pat_in_cramp:value][previous-instance]'	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
113	[pat_id_cramp] Show the field ONLY if: ([arm-number]=1 OR [arm-number]=2) AND [pat_in_cramp] = 'yes'	Patient ID in CRAMP <i>CRAMP on Orphanet</i>	text Field Annotation: @DEFAULT='[event-name][pat_id_cramp:value][previous-instance]'						
114	[pat_in_ddp] Show the field ONLY if: [arm-number]=2	Patient registered in DDP (Duchenne Data Platform)	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table> Field Annotation: @DEFAULT='[event-name][pat_in_ddp:value][previous-instance]'	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
115	[pat_id_ddp] Show the field ONLY if: [arm-number]=2 AND [pat_in_ddp] = 'yes'	Patient ID in DDP <a href="https://duchenne.nl/duchenne-data-platform/">https://duchenne.nl/duchenne-data-platform/</a>	text Field Annotation: @DEFAULT='[event-name][pat_id_ddp:value][previous-instance]'						
116	[pat_in_smartcare] Show the field ONLY if: [arm-number]=5	Patient registered in SMARTCARE (Spinal Muscular Atrophy)	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> </table>	yes	Yes	no	No		
yes	Yes								
no	No								

			<table border="1"> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table> <p>Field Annotation: @DEFAULT='[event-name][pat_in_smartcare:value][previous-instance]'</p>	unknown	Unknown				
unknown	Unknown								
117	<p>[pat_id_smartcare]</p> <p>Show the field ONLY if: [arm-number]=5 AND [pat_id_smartcare] = 'yes'</p>	<p>Patient ID in SMARtCARE</p> <p><a href="https://www.smartcare.de/en/">https://www.smartcare.de/en/</a></p>	<p>text</p> <p>Field Annotation: @DEFAULT='[event-name][pat_id_smartcare:value][previous-instance]'</p>						
118	<p>[pat_in_other_reg]</p>	<p>Patient registered in a registry that is not listed above</p>	<p>radio</p> <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table> <p>Field Annotation: @DEFAULT='[event-name][pat_in_other_reg:value][previous-instance]'</p>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
119	<p>[link_to_patient_registry]</p> <p>Show the field ONLY if: [pat_in_other_reg] = 'yes'</p>	<p>Link to other patient registry (please provide a comma-separated list of URIs if this patient is registered in more than one other registry)</p>	<p>notes</p> <p>Field Annotation: @DEFAULT='[event-name][link_to_patient_registry:value][previous-instance]'</p>						
120	<p>[pat_in_clinical_trial]</p>	<p>Patient in a clinical trial?</p>	<p>radio</p> <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table> <p>Field Annotation: @DEFAULT='[event-name][pat_in_clinical_trial:value][previous-instance]'</p>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
121	<p>[link_to_clinical_trial]</p> <p>Show the field ONLY if: [pat_in_clinical_trial] = 'yes'</p>	<p>Link to clinical trial (please provide a comma-separated list of URIs if this patient participates in more than one clinical trial)</p>	<p>notes</p> <p>Field Annotation: @DEFAULT='[event-name][link_to_clinical_trial:value][previous-instance]'</p>						
122	<p>[number_of_preg_known]</p> <p>Show the field ONLY if: [sex_at_birth_v] = 'female' AND [age_at_visit] &gt;= 16</p>	<p>Section Header: <i>Gestation</i></p> <p>Number of pregnancies known</p>	<p>radio</p> <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> </table>	yes	Yes	no	No		
yes	Yes								
no	No								
123	<p>[number_of_preg]</p> <p>Show the field ONLY if: [sex_at_birth_v] = 'female' AND [number_of_preg_known] = 'yes'</p>	<p>Number of pregnancies</p>	<p>text (integer), Required</p>						
124	<p>[number_of_child_known]</p> <p>Show the field ONLY if: [sex_at_birth_v] = 'female' AND [age_at_visit] &gt;= 16</p>	<p>Number of children known</p>	<p>radio</p> <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> </table>	yes	Yes	no	No		
yes	Yes								
no	No								
125	<p>[number_of_child]</p> <p>Show the field ONLY if: [sex_at_birth_v] = 'female' AND [number_of_child_known] = 'yes'</p>	<p>Number of children</p>	<p>text (integer), Required</p>						
126	<p>[had_genetic_couns]</p> <p>Show the field ONLY if: [arm-number]=1 OR [arm-number]=2 OR [arm-number]=3 OR [arm-number]=4 OR [arm-number]=5</p>	<p>Section Header: <i>Care pathway</i></p> <p>Patient had genetic counselling</p> <p><i>Has a genetic counselling taken place since the last registered contact?</i></p>	<p>radio</p> <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
127	<p>[genetic_couns_purp]</p> <p>Show the field ONLY if: [had_genetic_couns] = 'yes'</p>	<p>Genetic counselling (purpose)</p>	<p>checkbox</p> <table border="1"> <tr> <td>post_test_report_re</td> <td>genetic_couns_purp__post_test</td> </tr> </table>	post_test_report_re	genetic_couns_purp__post_test				
post_test_report_re	genetic_couns_purp__post_test								

			<table border="1"> <tr> <td>pre_natal</td> <td>genetic_couns_purp__pre_nata</td> </tr> <tr> <td>post_natal</td> <td>genetic_couns_purp__post_nata</td> </tr> <tr> <td>pre_symp</td> <td>genetic_couns_purp__pre_symp</td> </tr> <tr> <td>carrier</td> <td>genetic_couns_purp__carrier</td> </tr> <tr> <td>pre_conception</td> <td>genetic_couns_purp__pre_conc</td> </tr> <tr> <td>other</td> <td>genetic_couns_purp__other</td> </tr> </table>	pre_natal	genetic_couns_purp__pre_nata	post_natal	genetic_couns_purp__post_nata	pre_symp	genetic_couns_purp__pre_symp	carrier	genetic_couns_purp__carrier	pre_conception	genetic_couns_purp__pre_conc	other	genetic_couns_purp__other						
pre_natal	genetic_couns_purp__pre_nata																				
post_natal	genetic_couns_purp__post_nata																				
pre_symp	genetic_couns_purp__pre_symp																				
carrier	genetic_couns_purp__carrier																				
pre_conception	genetic_couns_purp__pre_conc																				
other	genetic_couns_purp__other																				
128	<p>[s2_had_malig_screening]</p> <p>Show the field ONLY if: [arm-number]=2</p>	Patient had malignancy screening	<table border="1"> <tr> <td colspan="2">radio</td> </tr> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	radio		yes	Yes	no	No	unknown	Unknown										
radio																					
yes	Yes																				
no	No																				
unknown	Unknown																				
129	<p>[has_access_to_phys_therapy]</p>	Does the patient have access to physical therapy?	<table border="1"> <tr> <td colspan="2">radio</td> </tr> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	radio		yes	Yes	no	No	unknown	Unknown										
radio																					
yes	Yes																				
no	No																				
unknown	Unknown																				
130	<p>[has_access_to_occ_therapy]</p>	Does the patient have access to occupational therapy?	<table border="1"> <tr> <td colspan="2">radio</td> </tr> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	radio		yes	Yes	no	No	unknown	Unknown										
radio																					
yes	Yes																				
no	No																				
unknown	Unknown																				
131	<p>[s2_has_speech_therapy]</p> <p>Show the field ONLY if: [arm-number]=2</p>	Does the patient have access to speech therapy?	<table border="1"> <tr> <td colspan="2">radio</td> </tr> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	radio		yes	Yes	no	No	unknown	Unknown										
radio																					
yes	Yes																				
no	No																				
unknown	Unknown																				
132	<p>[s2_has_dietitian_advice]</p> <p>Show the field ONLY if: [arm-number]=2</p>	Does the patient have access to dietitian advice?	<table border="1"> <tr> <td colspan="2">radio</td> </tr> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	radio		yes	Yes	no	No	unknown	Unknown										
radio																					
yes	Yes																				
no	No																				
unknown	Unknown																				
133	<p>[delayed_motor_milestones]</p> <p>Show the field ONLY if: [age_at_visit] &lt; 16</p>	<p>Section Header: <i>Developmental</i></p> <p>Delayed motor milestones</p>	<table border="1"> <tr> <td colspan="2">radio</td> </tr> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	radio		yes	Yes	no	No	unknown	Unknown										
radio																					
yes	Yes																				
no	No																				
unknown	Unknown																				
134	<p>[cgii]</p> <p>Show the field ONLY if: starts_with([event-name], 'visit')</p>	<p>Section Header: <i>Clinical Global Impression - Improvement scale (CGI-I)</i></p> <p>Compared to the patient's condition at baseline, this patient's [average] condition has/is...?</p>	<table border="1"> <tr> <td colspan="2">radio</td> </tr> <tr> <td>0</td> <td>Not assessed</td> </tr> <tr> <td>1</td> <td>Very Much Improved</td> </tr> <tr> <td>2</td> <td>Much Improved</td> </tr> <tr> <td>3</td> <td>Minimally Improved</td> </tr> <tr> <td>4</td> <td>No Change</td> </tr> <tr> <td>5</td> <td>Minimally Worse</td> </tr> <tr> <td>6</td> <td>Much Worse</td> </tr> <tr> <td>7</td> <td>Very Much Worse</td> </tr> </table>	radio		0	Not assessed	1	Very Much Improved	2	Much Improved	3	Minimally Improved	4	No Change	5	Minimally Worse	6	Much Worse	7	Very Much Worse
radio																					
0	Not assessed																				
1	Very Much Improved																				
2	Much Improved																				
3	Minimally Improved																				
4	No Change																				
5	Minimally Worse																				
6	Much Worse																				
7	Very Much Worse																				
135	<p>[s3_was_disc_in_boards]</p> <p>Show the field ONLY if: ([arm-number]=3 OR [arm-number]=6)</p>	<p>Section Header: <i>Events in the past 12 months</i></p> <p>Patient discussed in MDT/CPMS/Board within the last 12 months?</p> <p><i>Was the patient discussed in Multidisciplinary Team (MDT) / Clinical Patient Management System (CPMS) / Board within the last 12 months?</i></p>	<table border="1"> <tr> <td colspan="2">radio</td> </tr> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	radio		yes	Yes	no	No	unknown	Unknown										
radio																					
yes	Yes																				
no	No																				
unknown	Unknown																				
136	<p>[unpl_hosp_12m]</p>	Have there been any unplanned hospitalizations in the last	<table border="1"> <tr> <td colspan="2">radio</td> </tr> <tr> <td></td> <td></td> </tr> </table>	radio																	
radio																					

		12 months?	<table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown						
yes	Yes														
no	No														
unknown	Unknown														
137	[unpl_hosp_12m_n] Show the field ONLY if: [unpl_hosp_12m] = 'yes'	Number of unplanned hospitalizations in the last 12 months	text (integer), Required												
138	[unpl_hosp_12m_ndays_known] Show the field ONLY if: [unpl_hosp_12m] = 'yes'	Unplanned hospitalization in the last 12 months: Duration in days known	radio, Required <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> </table>	yes	Yes	no	No								
yes	Yes														
no	No														
139	[unpl_hosp_12m_ndays] Show the field ONLY if: [unpl_hosp_12m_ndays_known] = 'yes'	Unplanned hospitalization in the last 12 months: Duration in days	text (number), Required												
140	[had_comp_remi] Show the field ONLY if: ([arm-number]=1 OR [arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)	Section Header: <i>Events since last visit</i> <b>Complete remission</b> <i>Has the patient had a complete remission since the last recorded contact in this registry?</i>	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> <tr> <td>not_applicable</td> <td>Not applicable</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown	not_applicable	Not applicable				
yes	Yes														
no	No														
unknown	Unknown														
not_applicable	Not applicable														
141	[date_of_comp_remi] Show the field ONLY if: ([arm-number]=1 OR [arm-number]=2 OR [arm-number]=4 OR [arm-number]=6) AND [had_comp_remi]='yes'	Date of complete remission yyyy-mm-dd <i>If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.</i>	text (date_ymd)												
142	[had_part_remi] Show the field ONLY if: ([arm-number]=1 OR [arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)	Partial remission <i>Has the patient had a partial remission since the last recorded contact in this registry?</i>	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> <tr> <td>not_applicable</td> <td>Not applicable</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown	not_applicable	Not applicable				
yes	Yes														
no	No														
unknown	Unknown														
not_applicable	Not applicable														
143	[date_of_part_remi] Show the field ONLY if: ([arm-number]=1 OR [arm-number]=2 OR [arm-number]=4 OR [arm-number]=6) AND [had_part_remi]='yes'	Date of partial remission yyyy-mm-dd <i>If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.</i>	text (date_ymd)												
144	[s5_esco_diag_grade] Show the field ONLY if: [diagnosis_rd_orpha_v]='ORPHA:803' OR [diagnosis_rd_orpha_v]='ORPHA:357043' OR [diagnosis_rd_orpha_v]='ORPHA:275872' OR [diagnosis_rd_orpha_v]='ORPHA:300605'	Section Header: <i>Diagnosis</i> El Escorial diagnosis grade	radio <table border="1"> <tr> <td>suspected_als</td> <td>Suspected ALS</td> </tr> <tr> <td>possible_als</td> <td>Possible ALS</td> </tr> <tr> <td>probable_als_lab</td> <td>Probable ALS lab supported</td> </tr> <tr> <td>probable_als</td> <td>Probable ALS</td> </tr> <tr> <td>definitive_als</td> <td>Definitive ALS</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	suspected_als	Suspected ALS	possible_als	Possible ALS	probable_als_lab	Probable ALS lab supported	probable_als	Probable ALS	definitive_als	Definitive ALS	unknown	Unknown
suspected_als	Suspected ALS														
possible_als	Possible ALS														
probable_als_lab	Probable ALS lab supported														
probable_als	Probable ALS														
definitive_als	Definitive ALS														
unknown	Unknown														
145	[s1_antibodies_present] Show the field ONLY if: [arm-number]=1	Section Header: <i>Inflammatory neuropathy auto-antibodies</i> Presence of inflammatory neuropathy auto-antibodies	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>not_done</td> <td>Not tested (not done)</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	not_done	Not tested (not done)	unknown	Unknown				
yes	Yes														
no	No														
not_done	Not tested (not done)														
unknown	Unknown														
146	[s1_aa_paraprot_result] Show the field ONLY if: [s1_antibodies_present] = 'yes'	Paraproteins test-result <i>e.g. in case of Chronic inflammatory demyelinating polyradiculoneuropathy (CIDP)</i>	radio <table border="1"> <tr> <td>positive</td> <td>Positive</td> </tr> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	positive	Positive	negative	Negative	not_done	Not done						
positive	Positive														
negative	Negative														
not_done	Not done														

147	[s1_aa_antimag_result] Show the field ONLY if: [s1_antibodies_present] = 'yes'	Anti-MAG antibodies test-result	radio <table border="1"> <tr><td>positive</td><td>Positive</td></tr> <tr><td>negative</td><td>Negative</td></tr> <tr><td>not_done</td><td>Not done</td></tr> </table>	positive	Positive	negative	Negative	not_done	Not done		
positive	Positive										
negative	Negative										
not_done	Not done										
148	[s1_aa_antigm1_result] Show the field ONLY if: [s1_antibodies_present] = 'yes'	Anti-GM1 antibodies test-result <i>e.g. in case of Multifocal motor neuropathy (MMN)</i>	radio <table border="1"> <tr><td>positive</td><td>Positive</td></tr> <tr><td>negative</td><td>Negative</td></tr> <tr><td>not_done</td><td>Not done</td></tr> </table>	positive	Positive	negative	Negative	not_done	Not done		
positive	Positive										
negative	Negative										
not_done	Not done										
149	[s1_aa_antigm1_igm_result] Show the field ONLY if: [s1_aa_antigm1_result] = 'positive'	IgM antibodies test-result	radio <table border="1"> <tr><td>positive</td><td>Positive</td></tr> <tr><td>negative</td><td>Negative</td></tr> <tr><td>not_done</td><td>Not done</td></tr> </table>	positive	Positive	negative	Negative	not_done	Not done		
positive	Positive										
negative	Negative										
not_done	Not done										
150	[s1_aa_antigm1_igm_od] Show the field ONLY if: [s1_aa_antigm1_igm_result] = 'positive'	IgM antibodies ELISA OD value	text (number)								
151	[s1_aa_antigm1_igg_result] Show the field ONLY if: [s1_aa_antigm1_result] = 'positive'	IgG antibodies test-result	radio <table border="1"> <tr><td>positive</td><td>Positive</td></tr> <tr><td>negative</td><td>Negative</td></tr> <tr><td>not_done</td><td>Not done</td></tr> </table>	positive	Positive	negative	Negative	not_done	Not done		
positive	Positive										
negative	Negative										
not_done	Not done										
152	[s1_aa_antigm1_igg_od] Show the field ONLY if: [s1_aa_antigm1_igg_result] = 'positive'	IgG antibodies ELISA OD value	text (number)								
153	[s1_aa_antigang_result] Show the field ONLY if: [s1_antibodies_present] = 'yes'	Other antiganglioside antibodies test-result	radio <table border="1"> <tr><td>positive</td><td>Positive</td></tr> <tr><td>negative</td><td>Negative</td></tr> <tr><td>not_done</td><td>Not done</td></tr> </table>	positive	Positive	negative	Negative	not_done	Not done		
positive	Positive										
negative	Negative										
not_done	Not done										
154	[s1_aa_antigang_which] Show the field ONLY if: [s1_aa_antigang_result] = 'positive'	Which other antiganglioside antibodies? Please enter the names of other antiganglioside antibodies tested positive as a comma-separated list.	notes								
155	[s1_aa_nodal_result] Show the field ONLY if: [s1_antibodies_present] = 'yes'	Anti-nodal/paranodal protein antibodies test-result <i>e.g. in case of Chronic inflammatory demyelinating polyradiculoneuropathy (CIDP)</i>	radio <table border="1"> <tr><td>positive</td><td>Positive</td></tr> <tr><td>negative</td><td>Negative</td></tr> <tr><td>not_done</td><td>Not done</td></tr> </table>	positive	Positive	negative	Negative	not_done	Not done		
positive	Positive										
negative	Negative										
not_done	Not done										
156	[s1_aa_nodal_which] Show the field ONLY if: [s1_aa_nodal_result] = 'positive'	Which anti-nodal/paranodal protein antibodies? Please enter the names of anti-nodal/paranodal protein antibodies tested positive as a comma-separated list. <i>e.g.: anti-contactin, anti-CASPR, anti-neurofascin</i>	notes								
157	[s2_antibodies_present] Show the field ONLY if: [arm-number]=2	Section Header: <i>Myositis auto-antibodies</i> Presence of myositis auto-antibodies	radio <table border="1"> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> <tr><td>not_done</td><td>Not tested (not done)</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	yes	Yes	no	No	not_done	Not tested (not done)	unknown	Unknown
yes	Yes										
no	No										
not_done	Not tested (not done)										
unknown	Unknown										
158	[s2_aa_mi_2alpha] Show the field ONLY if: [s2_antibodies_present] = 'yes'	Mi-2 $\alpha$ test-result	radio <table border="1"> <tr><td>positive</td><td>Positive</td></tr> <tr><td>negative</td><td>Negative</td></tr> <tr><td>not_done</td><td>Not done</td></tr> </table>	positive	Positive	negative	Negative	not_done	Not done		
positive	Positive										
negative	Negative										
not_done	Not done										
159	[s2_aa_mi_2beta] Show the field ONLY if:	Mi-2 $\beta$ test-result	radio <table border="1"> <tr><td>positive</td><td>Positive</td></tr> </table>	positive	Positive						
positive	Positive										

	[s2_antibodies_present] = 'yes'		<table border="1"> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	negative	Negative	not_done	Not done		
negative	Negative								
not_done	Not done								
160	[s2_aa_mup44] Show the field ONLY if: [s2_antibodies_present] = 'yes'	Mup44 test-result	radio <table border="1"> <tr> <td>positive</td> <td>Positive</td> </tr> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	positive	Positive	negative	Negative	not_done	Not done
positive	Positive								
negative	Negative								
not_done	Not done								
161	[s2_aa_tif1gamma] Show the field ONLY if: [s2_antibodies_present] = 'yes'	TIF1γ test-result	radio <table border="1"> <tr> <td>positive</td> <td>Positive</td> </tr> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	positive	Positive	negative	Negative	not_done	Not done
positive	Positive								
negative	Negative								
not_done	Not done								
162	[s2_aa_mda5] Show the field ONLY if: [s2_antibodies_present] = 'yes'	MDA5 test-result	radio <table border="1"> <tr> <td>positive</td> <td>Positive</td> </tr> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	positive	Positive	negative	Negative	not_done	Not done
positive	Positive								
negative	Negative								
not_done	Not done								
163	[s2_aa_nxp2] Show the field ONLY if: [s2_antibodies_present] = 'yes'	NPX2 test-result	radio <table border="1"> <tr> <td>positive</td> <td>Positive</td> </tr> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	positive	Positive	negative	Negative	not_done	Not done
positive	Positive								
negative	Negative								
not_done	Not done								
164	[s2_aa_sae1] Show the field ONLY if: [s2_antibodies_present] = 'yes'	SAE1 test-result	radio <table border="1"> <tr> <td>positive</td> <td>Positive</td> </tr> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	positive	Positive	negative	Negative	not_done	Not done
positive	Positive								
negative	Negative								
not_done	Not done								
165	[s2_aa_ku] Show the field ONLY if: [s2_antibodies_present] = 'yes'	Ku test-result	radio <table border="1"> <tr> <td>positive</td> <td>Positive</td> </tr> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	positive	Positive	negative	Negative	not_done	Not done
positive	Positive								
negative	Negative								
not_done	Not done								
166	[s2_aa_pm_scl100] Show the field ONLY if: [s2_antibodies_present] = 'yes'	PM-Scl100 test-result	radio <table border="1"> <tr> <td>positive</td> <td>Positive</td> </tr> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	positive	Positive	negative	Negative	not_done	Not done
positive	Positive								
negative	Negative								
not_done	Not done								
167	[s2_aa_pm_scl75] Show the field ONLY if: [s2_antibodies_present] = 'yes'	PM-Scl75 test-result	radio <table border="1"> <tr> <td>positive</td> <td>Positive</td> </tr> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	positive	Positive	negative	Negative	not_done	Not done
positive	Positive								
negative	Negative								
not_done	Not done								
168	[s2_aa_jo_1] Show the field ONLY if: [s2_antibodies_present] = 'yes'	Jo-1 test-result	radio <table border="1"> <tr> <td>positive</td> <td>Positive</td> </tr> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	positive	Positive	negative	Negative	not_done	Not done
positive	Positive								
negative	Negative								
not_done	Not done								
169	[s2_aa_srp] Show the field ONLY if: [s2_antibodies_present] = 'yes'	SRP test-result	radio <table border="1"> <tr> <td>positive</td> <td>Positive</td> </tr> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	positive	Positive	negative	Negative	not_done	Not done
positive	Positive								
negative	Negative								
not_done	Not done								
170	[s2_aa_pl_7] Show the field ONLY if: [s2_antibodies_present] = 'yes'	PL-7 test-result	radio <table border="1"> <tr> <td>positive</td> <td>Positive</td> </tr> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	positive	Positive	negative	Negative	not_done	Not done
positive	Positive								
negative	Negative								
not_done	Not done								
171	[s2_aa_pl_12] Show the field ONLY if:	PL-12 test-result	radio <table border="1"> <tr> <td>positive</td> <td>Positive</td> </tr> </table>	positive	Positive				
positive	Positive								



	[s2_antibodies_present] = 'yes'		<table border="1"> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	negative	Negative	not_done	Not done						
negative	Negative												
not_done	Not done												
172	[s2_aa_ej] Show the field ONLY if: [s2_antibodies_present] = 'yes'	EJ test-result	<table border="1"> <tr> <td colspan="2">radio</td> </tr> <tr> <td>positive</td> <td>Positive</td> </tr> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	radio		positive	Positive	negative	Negative	not_done	Not done		
radio													
positive	Positive												
negative	Negative												
not_done	Not done												
173	[s2_aa_oj] Show the field ONLY if: [s2_antibodies_present] = 'yes'	OJ test-result	<table border="1"> <tr> <td colspan="2">radio</td> </tr> <tr> <td>positive</td> <td>Positive</td> </tr> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	radio		positive	Positive	negative	Negative	not_done	Not done		
radio													
positive	Positive												
negative	Negative												
not_done	Not done												
174	[s2_aa_ro_52] Show the field ONLY if: [s2_antibodies_present] = 'yes'	Ro-52 test-result	<table border="1"> <tr> <td colspan="2">radio</td> </tr> <tr> <td>positive</td> <td>Positive</td> </tr> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	radio		positive	Positive	negative	Negative	not_done	Not done		
radio													
positive	Positive												
negative	Negative												
not_done	Not done												
175	[s2_aa_anti_srp] Show the field ONLY if: [s2_antibodies_present] = 'yes'	anti-SRP test-result	<table border="1"> <tr> <td colspan="2">radio</td> </tr> <tr> <td>positive</td> <td>Positive</td> </tr> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	radio		positive	Positive	negative	Negative	not_done	Not done		
radio													
positive	Positive												
negative	Negative												
not_done	Not done												
176	[s2_aa_anti_hmgcr] Show the field ONLY if: [s2_antibodies_present] = 'yes'	anti-HMGCR test-result	<table border="1"> <tr> <td colspan="2">radio</td> </tr> <tr> <td>positive</td> <td>Positive</td> </tr> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	radio		positive	Positive	negative	Negative	not_done	Not done		
radio													
positive	Positive												
negative	Negative												
not_done	Not done												
177	[s4_antibodies_present] Show the field ONLY if: [arm-number]=4	Section Header: <i>Myasthenia auto-antibodies</i> Presence of myasthenia auto-antibodies	<table border="1"> <tr> <td colspan="2">radio</td> </tr> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>not_done</td> <td>Not tested (not done)</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	radio		yes	Yes	no	No	not_done	Not tested (not done)	unknown	Unknown
radio													
yes	Yes												
no	No												
not_done	Not tested (not done)												
unknown	Unknown												
178	[s4_aa_achr_ria_result] Show the field ONLY if: [s4_antibodies_present] = 'yes'	AChR (standard, RIA) test-result	<table border="1"> <tr> <td colspan="2">radio</td> </tr> <tr> <td>positive</td> <td>Positive</td> </tr> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	radio		positive	Positive	negative	Negative	not_done	Not done		
radio													
positive	Positive												
negative	Negative												
not_done	Not done												
179	[s4_aa_achr_cba_result] Show the field ONLY if: [s4_antibodies_present] = 'yes'	AChR (low-affinity, CBA) test-result	<table border="1"> <tr> <td colspan="2">radio</td> </tr> <tr> <td>positive</td> <td>Positive</td> </tr> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	radio		positive	Positive	negative	Negative	not_done	Not done		
radio													
positive	Positive												
negative	Negative												
not_done	Not done												
180	[s4_aa_musk_ria_result] Show the field ONLY if: [s4_antibodies_present] = 'yes'	Musk (standard,RIA) test-result	<table border="1"> <tr> <td colspan="2">radio</td> </tr> <tr> <td>positive</td> <td>Positive</td> </tr> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	radio		positive	Positive	negative	Negative	not_done	Not done		
radio													
positive	Positive												
negative	Negative												
not_done	Not done												
181	[s4_aa_musk_cba_result] Show the field ONLY if: [s4_antibodies_present] = 'yes'	MuSK (low-affinity, CBA) test-result	<table border="1"> <tr> <td colspan="2">radio</td> </tr> <tr> <td>positive</td> <td>Positive</td> </tr> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	radio		positive	Positive	negative	Negative	not_done	Not done		
radio													
positive	Positive												
negative	Negative												
not_done	Not done												
182	[s4_aa_titin_result] Show the field ONLY if: [s4_antibodies_present] = 'yes'	Titin test-result	<table border="1"> <tr> <td colspan="2">radio</td> </tr> <tr> <td>positive</td> <td>Positive</td> </tr> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	radio		positive	Positive	negative	Negative	not_done	Not done		
radio													
positive	Positive												
negative	Negative												
not_done	Not done												
183	[s4_aa_ryr1_result]	Ryanodine receptor (RYR1) test-result	<table border="1"> <tr> <td colspan="2">radio</td> </tr> <tr> <td></td> <td></td> </tr> </table>	radio									
radio													

	Show the field ONLY if: [s4_antibodies_present] = 'yes'		<table border="1"> <tr> <td>positive</td> <td>Positive</td> </tr> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	positive	Positive	negative	Negative	not_done	Not done		
positive	Positive										
negative	Negative										
not_done	Not done										
184	[s4_aa_vgcc_result] Show the field ONLY if: [s4_antibodies_present] = 'yes'	Voltage-gated calcium channel antibodies (VGCC) test-result	radio <table border="1"> <tr> <td>positive</td> <td>Positive</td> </tr> <tr> <td>negative</td> <td>Negative</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	positive	Positive	negative	Negative	not_done	Not done		
positive	Positive										
negative	Negative										
not_done	Not done										
185	[s2_ck_level] Show the field ONLY if: [arm-number]=2	Section Header: <i>Creatine Kinase</i> Creatine kinase level	radio <table border="1"> <tr> <td>normal</td> <td>Normal</td> </tr> <tr> <td>elevated_up_to_10_times</td> <td>Elevated - less than 10x normal or 10x normal</td> </tr> <tr> <td>elevated_10_times_plus</td> <td>Elevated - more than 10x normal</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> </table>	normal	Normal	elevated_up_to_10_times	Elevated - less than 10x normal or 10x normal	elevated_10_times_plus	Elevated - more than 10x normal	not_done	Not done
normal	Normal										
elevated_up_to_10_times	Elevated - less than 10x normal or 10x normal										
elevated_10_times_plus	Elevated - more than 10x normal										
not_done	Not done										
186	[s2_ck_exact_val_known] Show the field ONLY if: [arm-number]=2 AND ([s2_ck_level]='elevated_up_to_10_times' OR [s2_ck_level]='elevated_10_times_plus')	Exact creatine kinase value known	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> </table>	yes	Yes	no	No				
yes	Yes										
no	No										
187	[s2_ck_exact_val] Show the field ONLY if: [arm-number]=2 AND ([s2_ck_level]='elevated_up_to_10_times' OR [s2_ck_level]='elevated_10_times_plus') AND [s2_ck_exact_val_known] = 'yes'	Exact creatine kinase value [U/l]	text (number)								
188	[s2_ck_value_ref_report] Show the field ONLY if: [arm-number]=2 AND ([s2_ck_level]='elevated_up_to_10_times' OR [s2_ck_level]='elevated_10_times_plus')	Referenced normal value for creatine kinase level known	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> </table>	yes	Yes	no	No				
yes	Yes										
no	No										
189	[s2_ck_value_ref] Show the field ONLY if: [arm-number]=2 AND [s2_ck_value_ref_report] = 'yes'	Upper level of normal (ULN) for reference creatine kinase level (in enzyme units per litre [U/l]):	text (number)								
190	[s2_ck_value_report] Show the field ONLY if: [arm-number]=2 AND ([s2_ck_level]='elevated_up_to_10_times' OR [s2_ck_level]='elevated_10_times_plus')	Report value of creatine kinase	radio <table border="1"> <tr> <td>distinct_number</td> <td>Report a distinct number</td> </tr> <tr> <td>range</td> <td>Report a range</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	distinct_number	Report a distinct number	range	Report a range	unknown	Unknown		
distinct_number	Report a distinct number										
range	Report a range										
unknown	Unknown										
191	[s2_ck_value] Show the field ONLY if: [s2_ck_value_report] = 'distinct_number'	Creatine Kinase value (in enzyme units per litre [U/l]):	text (number)								
192	[s2_ck_value_r1] Show the field ONLY if: [s2_ck_value_report] = 'range'	Start of range for Creatine Kinase value (in enzyme units per litre [U/l]):	text (number)								
193	[s2_ck_value_r2] Show the field ONLY if: [s2_ck_value_report] = 'range'	End of range for Creatine Kinase value (in enzyme units per litre [U/l]):	text (number)								
194	[s2_musc_biopsy_result]	Section Header: <i>Muscular</i>	radio								

	Show the field ONLY if: [arm-number]=2	Muscle biopsy result	<table border="1"> <tr><td>normal</td><td>Normal</td></tr> <tr><td>abnormal</td><td>Abnormal</td></tr> <tr><td>not_done</td><td>Not done</td></tr> </table>	normal	Normal	abnormal	Abnormal	not_done	Not done				
normal	Normal												
abnormal	Abnormal												
not_done	Not done												
195	[s2_musc_biopsy_abnorm_type]  Show the field ONLY if: [arm-number]=2 AND [s2_musc_biopsy_result]='abnormal'	Muscle biopsy abnormal type	radio <table border="1"> <tr><td>neurogenic</td><td>Neurogenic</td></tr> <tr><td>myopathic</td><td>Myopathic</td></tr> <tr><td>dystrophic</td><td>Dystrophic</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	neurogenic	Neurogenic	myopathic	Myopathic	dystrophic	Dystrophic	unknown	Unknown		
neurogenic	Neurogenic												
myopathic	Myopathic												
dystrophic	Dystrophic												
unknown	Unknown												
196	[s2_musc_mri]  Show the field ONLY if: [arm-number]=2	Muscle MRI	radio <table border="1"> <tr><td>normal</td><td>Normal</td></tr> <tr><td>abnormal</td><td>Abnormal</td></tr> <tr><td>not_done</td><td>Not done</td></tr> </table>	normal	Normal	abnormal	Abnormal	not_done	Not done				
normal	Normal												
abnormal	Abnormal												
not_done	Not done												
197	[emg_result_type]	Section Header: <i>Neuromuscular</i> Electromyography (EMG): result type	radio <table border="1"> <tr><td>normal</td><td>Normal</td></tr> <tr><td>abnormal</td><td>Abnormal</td></tr> <tr><td>equivocal</td><td>Equivocal</td></tr> <tr><td>not_done</td><td>Not done</td></tr> </table>	normal	Normal	abnormal	Abnormal	equivocal	Equivocal	not_done	Not done		
normal	Normal												
abnormal	Abnormal												
equivocal	Equivocal												
not_done	Not done												
198	[emg_abnormality_type]  Show the field ONLY if: [emg_result_type]='abnormal'	Electromyography (EMG): abnormality type	checkbox <table border="1"> <tr><td>myopathic</td><td>emg_abnormality_type__myopathic</td></tr> <tr><td>neurogenic</td><td>emg_abnormality_type__neurogenic</td></tr> </table>	myopathic	emg_abnormality_type__myopathic	neurogenic	emg_abnormality_type__neurogenic						
myopathic	emg_abnormality_type__myopathic												
neurogenic	emg_abnormality_type__neurogenic												
199	[s4_sfemg_result_type]  Show the field ONLY if: [arm-number]=4	Single Fiber Electromyography (SFEMG): result type	radio <table border="1"> <tr><td>normal</td><td>Normal</td></tr> <tr><td>abnormal</td><td>Abnormal</td></tr> <tr><td>equivocal</td><td>Equivocal</td></tr> <tr><td>not_done</td><td>Not done</td></tr> </table>	normal	Normal	abnormal	Abnormal	equivocal	Equivocal	not_done	Not done		
normal	Normal												
abnormal	Abnormal												
equivocal	Equivocal												
not_done	Not done												
200	[s4_rep_nerve_stimu]  Show the field ONLY if: ([arm-number]=4 OR [arm-number]=6)	Repetitive nerve stimulation	radio <table border="1"> <tr><td>normal</td><td>Normal</td></tr> <tr><td>abnormal</td><td>Abnormal</td></tr> <tr><td>not_done</td><td>Not done</td></tr> </table>	normal	Normal	abnormal	Abnormal	not_done	Not done				
normal	Normal												
abnormal	Abnormal												
not_done	Not done												
201	[s4_rep_nerve_stimu_decr]  Show the field ONLY if: ([arm-number]=4 OR [arm-number]=6) AND [s4_rep_nerve_stimu]='abnormal'	Repetitive nerve stimulation: Decrement percentage <i>expected: number, minimum: 0, maximum: 100</i>	text (number, Min: 0, Max: 100)										
202	[s4_rep_nerve_stimu_incr]  Show the field ONLY if: ([arm-number]=4 OR [arm-number]=6) AND [s4_rep_nerve_stimu]='abnormal'	Repetitive nerve stimulation: Increment percentage <i>expected: number, minimum: 0</i>	text (number, Min: 0)										
203	[ambulatory_status]	Section Header: <i>Mobility and General Physical Functioning</i> Current ambulatory status	radio <table border="1"> <tr><td>walk_unsupported</td><td>Walk without support</td></tr> <tr><td>walk_supported</td><td>Walk with support</td></tr> <tr><td>wheelchair_pt</td><td>Wheelchair parttime</td></tr> <tr><td>wheelchair_ft</td><td>Wheelchair fulltime</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	walk_unsupported	Walk without support	walk_supported	Walk with support	wheelchair_pt	Wheelchair parttime	wheelchair_ft	Wheelchair fulltime	unknown	Unknown
walk_unsupported	Walk without support												
walk_supported	Walk with support												
wheelchair_pt	Wheelchair parttime												
wheelchair_ft	Wheelchair fulltime												
unknown	Unknown												
204	[ambulatory_status_prev]	ambulatory_status_prev: Value of ambulatory_status in previous report. Is needed for piping and branching for other fields.	text Field Annotation: @CALCTEXT(if(starts_with([event-name], 'enrolment'), ', ', if(starts_with([event-name], 'visit') AND [current-instance]=1, [first-event-name]										

			[ambulatory_status], if(starts_with([event-name], 'visit') AND [current-instance]>1, [event-name] [ambulatory_status][previous-instance], "")) @READONLY @HIDDEN @HIDDEN-PDF						
205	[ambulation_change_d_known]  Show the field ONLY if: [ambulatory_status_prev] != 'unknown' AND [ambulatory_status] != 'unknown' AND !isblankormissingcode([ambulatory_status_prev]) AND !isblankormissingcode([ambulatory_status]) AND [ambulatory_status_prev] != [ambulatory_status]	When comparing this report to the last report, there was a change in in terms of the patients ambulatory status. Last time (date: [date_of_visit_prev]) you reported: [ambulatory_status_prev] Here (date: [date_of_visit]) you reported: [ambulatory_status:value]  Do you know when this ability changed?	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> </table>	yes	Yes	no	No		
yes	Yes								
no	No								
206	[date_of_change_of_ambulation]  Show the field ONLY if: [ambulation_change_d_known] = 'yes'	Date of change of ambulation yyyy-mm-dd <i>If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.</i>	text (date_ymd)						
207	[s2_max_walking_m]  Show the field ONLY if: [arm-number]=2	Maximum walking distance estimate (m)	text (number)						
208	[has_limb_weakness]	Limb weakness	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
209	[has_neck_weakness]	Neck muscle weakness	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
210	[has_facial_weakness]	Weakness of facial musculature	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
211	[s5_hands_up_curr]  Show the field ONLY if: [arm-number]=5	Can the patient currently raise hands to mouth in a sitting position?	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
212	[s5_hands_up_curr_prev]	s5_hands_up_curr_prev: Value of s5_hands_up_curr in previous report. Is needed for piping and branching for other fields.	text Field Annotation: @CALCTEXT(if(starts_with([event-name], 'enrolment'), ", if(starts_with([event-name], 'visit') AND [current-instance]=1, [first-event-name] [s5_hands_up_curr], if(starts_with([event-name], 'visit') AND [current-instance]>1, [event-name] [s5_hands_up_curr][previous-instance], "")) @READONLY @HIDDEN @HIDDEN-PDF						
213	[s5_hands_up_d_known]  Show the field ONLY if: [arm-number]=5 AND ([s5_hands_up_curr]='yes' AND [s5_hands_up_curr_prev]='no' OR [s5_hands_up_curr]='no' AND [s5_hands_up_curr_prev]='yes')	When comparing this report to the last report, there was a change in in terms of the patients ability to raise hands to mouth in a sitting position. Last time (date: [date_of_visit_prev]) you reported: Is patient able? [s5_hands_up_curr_prev] Here (date: [date_of_visit]) you reported: Is patient able? [s5_hands_up_curr:value]  Do you know when this ability changed?	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> </table>	yes	Yes	no	No		
yes	Yes								
no	No								
214	[s5_date_of_hands_up_gain]	Date of gain of raising hands ability yyyy-mm-dd <i>If the exact day is not known, please enter 15th. If the month is not known,</i>	text (date_ymd)						

	Show the field ONLY if: [s5_hands_up_curr]='yes' AND D [s5_hands_up_curr_prev]= 'no' AND [s5_hands_up_d_kn own]='yes'	<i>please enter 1st of July of that year.</i>							
215	[s5_date_of_hands_up_lo ss]  Show the field ONLY if: [s5_hands_up_curr]='no' AN D [s5_hands_up_curr_prev]= 'yes' AND [s5_hands_up_d_k nown]='yes'	Date of loss of raising hands ability yyyy-mm-dd <i>If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.</i>	text (date_ymd)						
216	[s5_hold_head_curr]  Show the field ONLY if: [arm-number]=5 AND [age_ at_visit] < 16	Can the patient currently hold their head up without support?	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
217	[s5_hold_head_curr_prev ]	s5_hold_head_curr_prev: Value of s5_hold_head_curr in previous report. Is needed for piping and branching for other fields.	text Field Annotation: @CALCTEXT(if(starts_with([event- name], 'enrolment'), ", if(starts_with([event-name], 'visit') AND [current-instance]=1, [first-event-name] [s5_hold_head_curr], if(starts_with([event-name], 'visit') AND [current-instance]>1, [event-name] [s5_hold_head_curr][previous-instance], ""))) @READONLY @HIDDEN @HIDDEN-PDF						
218	[s5_hold_head_d_known]  Show the field ONLY if: [arm-number]=5 AND ([s5_h old_head_curr]='yes' AND [s 5_hold_head_curr_prev]='no' OR [s5_hold_head_curr]='no' AND [s5_hold_head_curr_pr ev]='yes')	When comparing this report to the last report, there was a change in in terms of the patients ability to hold their head up without support. Last time (date: [date_of_visit_prev]) you reported: Is patient able? [s5_hold_head_curr_prev] Here (date: [date_of_visit]) you reported: Is patient able? [s5_hold_head_curr:value]  Do you know when this ability changed?	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> </table>	yes	Yes	no	No		
yes	Yes								
no	No								
219	[s5_date_of_hold_head_g ain]  Show the field ONLY if: [s5_hold_head_curr]='yes' A ND [s5_hold_head_curr_prev ]='no' AND [s5_hold_head_d _known]='yes'	Date of gain of holding head up yyyy-mm-dd <i>If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.</i>	text (date_ymd)						
220	[s5_date_of_hold_head_l oss]  Show the field ONLY if: [s5_hold_head_curr]='no' AN D [s5_hold_head_curr_prev] ='yes' AND [s5_hold_head_d _known]='yes'	Date of loss of holding head up yyyy-mm-dd <i>If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.</i>	text (date_ymd)						
221	[s5_sitting_curr]  Show the field ONLY if: [arm-number]=5 AND [age_ at_visit] < 16	Can the patient currently sit without support?	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
222	[s5_sitting_curr_prev]	s5_sitting_curr_prev: Value of s5_sitting_curr in previous report. Is needed for piping and branching for other fields.	text Field Annotation: @CALCTEXT(if(starts_with([event- name], 'enrolment'), ", if(starts_with([event-name], 'visit') AND [current-instance]=1, [first-event-name] [s5_sitting_curr], if(starts_with([event-name], 'visit') AND [current-instance]>1, [event-name] [s5_sitting_curr][previous-instance], ""))) @READONLY @HIDDEN @HIDDEN-PDF						
223	[s5_sitting_d_known]  Show the field ONLY if: [arm-number]=5 AND ([s5_si tting_curr]='yes' AND [s5_sitt	When comparing this report to the last report, there was a change in in terms of the patients ability to sit without support. Last time (date: [date_of_visit_prev]) you reported: Is patient	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> </table>	yes	Yes	no	No		
yes	Yes								
no	No								

	ing_curr_prev]='no' OR [s5_sitting_curr]='no' AND [s5_sitting_curr_prev]='yes')	able? [s5_sitting_curr_prev] Here (date: [date_of_visit]) you reported: Is patient able? [s5_sitting_curr:value]  Do you know when this ability changed?							
224	[s5_date_of_sitting_gain]  Show the field ONLY if: [s5_sitting_curr]='yes' AND [s5_sitting_curr_prev]='no' AND [s5_sitting_d_known]='yes'	Date of gain of sitting ability yyyy-mm-dd <i>If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.</i>	text (date_ymd)						
225	[s5_date_of_sitting_loss]  Show the field ONLY if: [s5_sitting_curr]='no' AND [s5_sitting_curr_prev]='yes' AND [s5_sitting_d_known]='yes'	Date of loss of sitting ability yyyy-mm-dd <i>If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.</i>	text (date_ymd)						
226	[has_cognitive_impairment]	Section Header: <i>Cognitive Impairment</i> Cognitive impairment	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
227	[cognitive_impairment_type]  Show the field ONLY if: [has_cognitive_impairment]='yes'	Type of cognitive impairment	radio <table border="1"> <tr> <td>developmental</td> <td>Developmental</td> </tr> <tr> <td>acquired</td> <td>Acquired</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	developmental	Developmental	acquired	Acquired	unknown	Unknown
developmental	Developmental								
acquired	Acquired								
unknown	Unknown								
228	[has_breathing_difficulties]	Section Header: <i>Respiratory / Ventilation</i> Breathing difficulties	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
229	[has_noct_hypovent]	Symptoms of nocturnal hypoventilation	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
230	[fvc_seated_known]  Show the field ONLY if: [has_breathing_difficulties]='yes'	Forced Vital Capacity (FVC) seated known	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> </table>	yes	Yes	no	No		
yes	Yes								
no	No								
231	[fvc_seated]  Show the field ONLY if: [fvc_seated_known]='yes'	Forced Vital Capacity (FVC) seated % of estimated	text (number)						
232	[fvc_lying_known]  Show the field ONLY if: [has_breathing_difficulties]='yes'	Forced Vital Capacity (FVC) lying known	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> </table>	yes	Yes	no	No		
yes	Yes								
no	No								
233	[fvc_lying]  Show the field ONLY if: [fvc_lying_known]='yes'	Forced Vital Capacity (FVC) lying % of estimated	text (number)						
234	[ventilation_required]  Show the field ONLY if: [has_breathing_difficulties]='yes'	Assisted ventilation required	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
235	[ventilation_times_noninv]  Show the field ONLY if:	Noninvasive ventilation	radio <table border="1"> <tr> <td>night</td> <td>Night</td> </tr> <tr> <td>day_night</td> <td>Day &amp; Night</td> </tr> </table>	night	Night	day_night	Day & Night		
night	Night								
day_night	Day & Night								

	[ventilation_required] = 'yes'		<table border="1"> <tr> <td>during_exacerb</td> <td>During exacerbations</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> <tr> <td>not_applicable</td> <td>Not applicable</td> </tr> </table>	during_exacerb	During exacerbations	unknown	Unknown	not_applicable	Not applicable				
during_exacerb	During exacerbations												
unknown	Unknown												
not_applicable	Not applicable												
236	[ventilation_times_inv] Show the field ONLY if: [ventilation_required] = 'yes'	Invasive Ventilation	radio <table border="1"> <tr> <td>night</td> <td>Night</td> </tr> <tr> <td>day_night</td> <td>Day &amp; Night</td> </tr> <tr> <td>during_exacerb</td> <td>During exacerbations</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> <tr> <td>not_applicable</td> <td>Not applicable</td> </tr> </table>	night	Night	day_night	Day & Night	during_exacerb	During exacerbations	unknown	Unknown	not_applicable	Not applicable
night	Night												
day_night	Day & Night												
during_exacerb	During exacerbations												
unknown	Unknown												
not_applicable	Not applicable												
237	[s4_vent_days_py_known] Show the field ONLY if: [arm-number]=4	Number of ventilation days in the last 12 months known	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> </table>	yes	Yes	no	No						
yes	Yes												
no	No												
238	[s4_vent_days_py] Show the field ONLY if: [arm-number]=4	Number of ventilation days in the last 12 months	text (integer), Required										
239	[has_feed_swal_difficulties]	Section Header: <i>Feeding</i> Feeding or swallowing difficulties	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown				
yes	Yes												
no	No												
unknown	Unknown												
240	[has_feeding_difficulties] Show the field ONLY if: [has_feed_swal_difficulties]='yes'	Feeding difficulties	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown				
yes	Yes												
no	No												
unknown	Unknown												
241	[has_swallow_difficulties] Show the field ONLY if: [has_feed_swal_difficulties]='yes'	Swallowing difficulties	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown				
yes	Yes												
no	No												
unknown	Unknown												
242	[independent_feeding] Show the field ONLY if: isblankormissingcode([age_at_visit]) OR [age_at_visit] >= 3	Independent feeding (hand to mouth)	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown				
yes	Yes												
no	No												
unknown	Unknown												
243	[has_tube_feeding_gastro] Show the field ONLY if: [has_feed_swal_difficulties]='yes'	Gastrostomy tube feeding	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown				
yes	Yes												
no	No												
unknown	Unknown												
244	[has_tube_feeding_nasog] Show the field ONLY if: [has_feed_swal_difficulties]='yes'	Nasogastric tube feeding	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown				
yes	Yes												
no	No												
unknown	Unknown												
245	[s3_receives_mitococktail] Show the field ONLY if: [arm-number]=3	"Mitococktail" or food supplements? <i>Does the patient receive "Mitococktail" or food supplements?</i>	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown				
yes	Yes												
no	No												
unknown	Unknown												
246	[has_cardiac_involvement]	Section Header: <i>Cardiac</i> Cardiac involvement	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown				
yes	Yes												
no	No												
unknown	Unknown												
247	[which_cardiac_syndrome]	Cardiac syndrome (ORPHAcodes)	text										

	Show the field ONLY if: [has_cardiac_involvement] = 'yes'		<table border="1"> <tr> <td>BIOPORTAL:ORDO</td> <td>BIOPORTAL:ORDO</td> </tr> </table>	BIOPORTAL:ORDO	BIOPORTAL:ORDO				
BIOPORTAL:ORDO	BIOPORTAL:ORDO								
248	[has_impl_cardio_defib] Show the field ONLY if: [has_cardiac_involvement]='yes'	Does the patient have a cardioverter defibrillator?	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
249	[had_heart_transplantation] Show the field ONLY if: [has_cardiac_involvement]='yes'	Has the patient had a heart transplantation?	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
250	[has_pacemaker] Show the field ONLY if: [has_cardiac_involvement]='yes'	Does the patient have a pacemaker?	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
251	[had_cardiac_resync_therapy] Show the field ONLY if: [has_cardiac_involvement]='yes'	Has the patient had Cardiac Resynchronization Therapy (CRT)?	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
252	[has_scoliosis]	Section Header: <i>Scoliosis</i> Scoliosis	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
253	[had_scoliosis_surgery] Show the field ONLY if: [has_scoliosis] = 'yes'	Has the patient had a scoliosis surgery?	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
254	[scoliosis_cobb_angle_known] Show the field ONLY if: [has_scoliosis] = 'yes'	Scoliosis Cobb Angle known	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> </table>	yes	Yes	no	No		
yes	Yes								
no	No								
255	[scoliosis_cobb_angle] Show the field ONLY if: [has_scoliosis] = 'yes' AND [scoliosis_cobb_angle_known] = 'yes'	Scoliosis Cobb Angle	text (number), Required						
256	[s3_has_mito_asso_epilepsy] Show the field ONLY if: [arm-number]=3	Section Header: <i>Epilepsy</i> Does the patient have Mito-associated epilepsy?	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
257	[phenotypes_description_1]	Section Header: <i>PHENOTYPES</i> Please enter the most important symptom(s) for the neuromuscular clinic appointment here. Some of the following fields may be prepopulated with data from a previous visit. Please make sure that all values are correct. Phenotypes are to be coded with the Human Phenotype Ontology (HPO). Element 6.3. in the Set of common data elements for Rare Diseases Registration (link)	descriptive						
258	[phenotype_matrix]	No Provide phenotype Phenotype (HPO) 1 {pheno_yes_1} {pheno_hpo_1} 2 {pheno_yes_2} {pheno_hpo_2} 3 {pheno_yes_3} {pheno_hpo_3} 4 {pheno_yes_4} {pheno_hpo_4} 5 {pheno_yes_5} {pheno_hpo_5} 6 {pheno_yes_6} {pheno_hpo_6} 7 {pheno_yes_7} {pheno_hpo_7} 8 {pheno_yes_8} {pheno_hpo_8} 9	descriptive Field Annotation: @HIDDEN-PDF						



		{pheno_yes_9} {pheno_hpo_9} 10 {pheno_yes_10} {pheno_hpo_10}					
259	[pheno_yes_1]	Phenotype 1 yes/no	yesno, Required <table border="1"> <tr><td>1</td><td>Yes</td></tr> <tr><td>0</td><td>No</td></tr> </table> Custom alignment: LV Field Annotation: @DEFAULT='1' @READONLY	1	Yes	0	No
1	Yes						
0	No						
260	[pheno_hpo_1] Show the field ONLY if: [pheno_yes_1]='1'	Phenotype: HPO-code (1)	text, Required <table border="1"> <tr><td>BIOPORTAL:HP</td><td>BIOPORTAL:HP</td></tr> </table> Field Annotation: @DEFAULT='[event-name] [pheno_hpo_1:value][previous-instance]'	BIOPORTAL:HP	BIOPORTAL:HP		
BIOPORTAL:HP	BIOPORTAL:HP						
261	[pheno_yes_2]	Phenotype 2 yes/no	yesno <table border="1"> <tr><td>1</td><td>Yes</td></tr> <tr><td>0</td><td>No</td></tr> </table> Custom alignment: LV Field Annotation: @DEFAULT='[event-name] [pheno_yes_2:value][previous-instance]'	1	Yes	0	No
1	Yes						
0	No						
262	[pheno_hpo_2] Show the field ONLY if: [pheno_yes_2]='1'	Phenotype: HPO-code (2)	text, Required <table border="1"> <tr><td>BIOPORTAL:HP</td><td>BIOPORTAL:HP</td></tr> </table> Field Annotation: @DEFAULT='[event-name] [pheno_hpo_2:value][previous-instance]'	BIOPORTAL:HP	BIOPORTAL:HP		
BIOPORTAL:HP	BIOPORTAL:HP						
263	[pheno_yes_3]	Phenotype 3 yes/no	yesno <table border="1"> <tr><td>1</td><td>Yes</td></tr> <tr><td>0</td><td>No</td></tr> </table> Custom alignment: LV Field Annotation: @DEFAULT='[event-name] [pheno_yes_3:value][previous-instance]'	1	Yes	0	No
1	Yes						
0	No						
264	[pheno_hpo_3] Show the field ONLY if: [pheno_yes_3]='1'	Phenotype: HPO-code (3)	text, Required <table border="1"> <tr><td>BIOPORTAL:HP</td><td>BIOPORTAL:HP</td></tr> </table> Field Annotation: @DEFAULT='[event-name] [pheno_hpo_3:value][previous-instance]'	BIOPORTAL:HP	BIOPORTAL:HP		
BIOPORTAL:HP	BIOPORTAL:HP						
265	[pheno_yes_4]	Phenotype 4 yes/no	yesno <table border="1"> <tr><td>1</td><td>Yes</td></tr> <tr><td>0</td><td>No</td></tr> </table> Custom alignment: LV Field Annotation: @DEFAULT='[event-name] [pheno_yes_4:value][previous-instance]'	1	Yes	0	No
1	Yes						
0	No						
266	[pheno_hpo_4] Show the field ONLY if: [pheno_yes_4]='1'	Phenotype: HPO-code (4)	text, Required <table border="1"> <tr><td>BIOPORTAL:HP</td><td>BIOPORTAL:HP</td></tr> </table> Field Annotation: @DEFAULT='[event-name] [pheno_hpo_4:value][previous-instance]'	BIOPORTAL:HP	BIOPORTAL:HP		
BIOPORTAL:HP	BIOPORTAL:HP						
267	[pheno_yes_5]	Phenotype 5 yes/no	yesno <table border="1"> <tr><td>1</td><td>Yes</td></tr> <tr><td>0</td><td>No</td></tr> </table> Custom alignment: LV Field Annotation: @DEFAULT='[event-name] [pheno_yes_5:value][previous-instance]'	1	Yes	0	No
1	Yes						
0	No						
268	[pheno_hpo_5] Show the field ONLY if: [pheno_yes_5]='1'	Phenotype: HPO-code (5)	text, Required <table border="1"> <tr><td>BIOPORTAL:HP</td><td>BIOPORTAL:HP</td></tr> </table> Field Annotation: @DEFAULT='[event-name]'	BIOPORTAL:HP	BIOPORTAL:HP		
BIOPORTAL:HP	BIOPORTAL:HP						

			[pheno_hpo_5:value][previous-instance]'				
269	[pheno_yes_6]	Phenotype 6 yes/no	<p>yesno</p> <table border="1"> <tr><td>1</td><td>Yes</td></tr> <tr><td>0</td><td>No</td></tr> </table> <p>Custom alignment: LV Field Annotation: @DEFAULT='[event-name] [pheno_yes_6:value][previous-instance]'</p>	1	Yes	0	No
1	Yes						
0	No						
270	[pheno_hpo_6] Show the field ONLY if: [pheno_yes_6]='1'	Phenotype: HPO-code (6)	<p>text, Required</p> <table border="1"> <tr><td>BIOPORTAL:HP</td><td>BIOPORTAL:HP</td></tr> </table> <p>Field Annotation: @DEFAULT='[event-name] [pheno_hpo_6:value][previous-instance]'</p>	BIOPORTAL:HP	BIOPORTAL:HP		
BIOPORTAL:HP	BIOPORTAL:HP						
271	[pheno_yes_7]	Phenotype 7 yes/no	<p>yesno</p> <table border="1"> <tr><td>1</td><td>Yes</td></tr> <tr><td>0</td><td>No</td></tr> </table> <p>Custom alignment: LV Field Annotation: @DEFAULT='[event-name] [pheno_yes_7:value][previous-instance]'</p>	1	Yes	0	No
1	Yes						
0	No						
272	[pheno_hpo_7] Show the field ONLY if: [pheno_yes_7]='1'	Phenotype: HPO-code (7)	<p>text, Required</p> <table border="1"> <tr><td>BIOPORTAL:HP</td><td>BIOPORTAL:HP</td></tr> </table> <p>Field Annotation: @DEFAULT='[event-name] [pheno_hpo_7:value][previous-instance]'</p>	BIOPORTAL:HP	BIOPORTAL:HP		
BIOPORTAL:HP	BIOPORTAL:HP						
273	[pheno_yes_8]	Phenotype 8 yes/no	<p>yesno</p> <table border="1"> <tr><td>1</td><td>Yes</td></tr> <tr><td>0</td><td>No</td></tr> </table> <p>Custom alignment: LV Field Annotation: @DEFAULT='[event-name] [pheno_yes_8:value][previous-instance]'</p>	1	Yes	0	No
1	Yes						
0	No						
274	[pheno_hpo_8] Show the field ONLY if: [pheno_yes_8]='1'	Phenotype: HPO-code (8)	<p>text, Required</p> <table border="1"> <tr><td>BIOPORTAL:HP</td><td>BIOPORTAL:HP</td></tr> </table> <p>Field Annotation: @DEFAULT='[event-name] [pheno_hpo_8:value][previous-instance]'</p>	BIOPORTAL:HP	BIOPORTAL:HP		
BIOPORTAL:HP	BIOPORTAL:HP						
275	[pheno_yes_9]	Phenotype 9 yes/no	<p>yesno</p> <table border="1"> <tr><td>1</td><td>Yes</td></tr> <tr><td>0</td><td>No</td></tr> </table> <p>Custom alignment: LV Field Annotation: @DEFAULT='[event-name] [pheno_yes_9:value][previous-instance]'</p>	1	Yes	0	No
1	Yes						
0	No						
276	[pheno_hpo_9] Show the field ONLY if: [pheno_yes_9]='1'	Phenotype: HPO-code (9)	<p>text, Required</p> <table border="1"> <tr><td>BIOPORTAL:HP</td><td>BIOPORTAL:HP</td></tr> </table> <p>Field Annotation: @DEFAULT='[event-name] [pheno_hpo_9:value][previous-instance]'</p>	BIOPORTAL:HP	BIOPORTAL:HP		
BIOPORTAL:HP	BIOPORTAL:HP						
277	[pheno_yes_10]	Phenotype 10 yes/no	<p>yesno</p> <table border="1"> <tr><td>1</td><td>Yes</td></tr> <tr><td>0</td><td>No</td></tr> </table> <p>Custom alignment: LV Field Annotation: @DEFAULT='[event-name] [pheno_yes_10:value][previous-instance]'</p>	1	Yes	0	No
1	Yes						
0	No						
278	[pheno_hpo_10] Show the field ONLY if: [pheno_yes_10]='1'	Phenotype: HPO-code (10)	<p>text, Required</p> <table border="1"> <tr><td>BIOPORTAL:HP</td><td>BIOPORTAL:HP</td></tr> </table> <p>Field Annotation: @DEFAULT='[event-name]</p>	BIOPORTAL:HP	BIOPORTAL:HP		
BIOPORTAL:HP	BIOPORTAL:HP						

			[pheno_hpo_10:value][previous-instance']						
279	[review_medi_desc1]	Section Header: <i>Medication Review</i> This section is intended to review the recorded information on this patient's medications. An overview of all recorded medications is also visible on the patient's Record Home Page.	descriptive						
280	[review_medi_all]	Show all recorded medications for this patient	sql SELECT a.value FROM redcap_data a WHERE a.project_id = [project-id] and a.record = [record-name] and a.field_name = 'medication_label_display'						
281	[review_medi_ongoing]	Show all recorded ongoing medications for this patient	sql SELECT a.value FROM redcap_data a LEFT JOIN redcap_data b ON a.project_id = b.project_id AND a.record = b.record AND a.event_id = b.event_id AND a.instance = b.instance WHERE a.project_id = [project-id] AND a.record = [record-name] AND a.field_name = 'medication_label_display' AND a.instance > 0 AND b.field_name = 'medi_status' AND b.value = 'ongoing' ORDER BY a.instance  Field Annotation: @HIDDEN @HIDDEN-PDF						
282	[medi_change_yes] Show the field ONLY if: lstarts_with([event-name], 'enrolment')	Are there any changes in the patient's medications compared to the available data listed in the dropdowns above?	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
283	[review_medi_desc2] Show the field ONLY if: [medi_change_yes]=1	Please complete the medication questionnaire(s) with the updated medication details (accessible from the patient's Record Home Page).	descriptive						
284	[visit_data_complete]	Section Header: <i>Form Status</i> Complete?	dropdown <table border="1"> <tr> <td>0</td> <td>Incomplete</td> </tr> <tr> <td>1</td> <td>Unverified</td> </tr> <tr> <td>2</td> <td>Complete</td> </tr> </table>	0	Incomplete	1	Unverified	2	Complete
0	Incomplete								
1	Unverified								
2	Complete								

**Instrument: NJD: Thymus (njd\_thymus)**

Active languages: None

285	[s4_thymus_h1]	NJD: THYMUS	descriptive								
286	[date_of_assess_thym]	Date of assessment yyyy-mm-dd	text (date_ymd), Required								
287	[s4_thymectomy_type] Show the field ONLY if: [arm-number]=4	Thymectomy	radio <table border="1"> <tr> <td>open_surgery</td> <td>Open surgery</td> </tr> <tr> <td>endoscopic</td> <td>Endoscopic</td> </tr> <tr> <td>not_done</td> <td>Not done</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	open_surgery	Open surgery	endoscopic	Endoscopic	not_done	Not done	unknown	Unknown
open_surgery	Open surgery										
endoscopic	Endoscopic										
not_done	Not done										
unknown	Unknown										
288	[s4_date_of_thymectomy] Show the field ONLY if: [arm-number]=4 AND ([s4_thymectomy_type] = 'open_surgery' OR [s4_thymectomy_type] = 'endoscopic')	Date of thymectomy yyyy-mm-dd <i>If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.</i>	text (date_ymd)								
289	[s4_thym_histology] Show the field ONLY if: [arm-number] = 4 AND ([s4_thymectomy_type] = 'open_surgery' OR [s4_thymectomy_type] = 'endoscopic')	Thymus histology	radio <table border="1"> <tr> <td>thymoma</td> <td>Thymoma</td> </tr> <tr> <td>other</td> <td>Other</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	thymoma	Thymoma	other	Other	unknown	Unknown		
thymoma	Thymoma										
other	Other										
unknown	Unknown										
290	[s4_thym_mk_stage] Show the field ONLY if: [arm-number]=4 AND [s4_th	Masaoka-Koga stage	radio <table border="1"> <tr> <td>I</td> <td>I</td> </tr> <tr> <td></td> <td></td> </tr> </table>	I	I						
I	I										

	ym_histology] = 'thymoma'		<table border="1"> <tr><td>Ila</td><td>Ila</td></tr> <tr><td>IIb</td><td>IIb</td></tr> <tr><td>III</td><td>III</td></tr> <tr><td>IVa</td><td>IVa</td></tr> <tr><td>IVb</td><td>IVb</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	Ila	Ila	IIb	IIb	III	III	IVa	IVa	IVb	IVb	unknown	Unknown		
Ila	Ila																
IIb	IIb																
III	III																
IVa	IVa																
IVb	IVb																
unknown	Unknown																
291	[s4_thym_who_class] Show the field ONLY if: [arm-number]=4 AND [s4_thym_histology] = 'thymoma'	WHO classification	radio <table border="1"> <tr><td>A</td><td>A</td></tr> <tr><td>AB</td><td>AB</td></tr> <tr><td>B1</td><td>B1</td></tr> <tr><td>B2</td><td>B2</td></tr> <tr><td>B3</td><td>B3</td></tr> <tr><td>C</td><td>C</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	A	A	AB	AB	B1	B1	B2	B2	B3	B3	C	C	unknown	Unknown
A	A																
AB	AB																
B1	B1																
B2	B2																
B3	B3																
C	C																
unknown	Unknown																
292	[s4_thym_ther_type_known] Show the field ONLY if: [arm-number]=4	Thymoma therapy known	radio <table border="1"> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> <tr><td>no_thymoma</td><td>No Thymoma</td></tr> </table> Field Annotation: @DEFAULT = 'yes'	yes	Yes	no	No	no_thymoma	No Thymoma								
yes	Yes																
no	No																
no_thymoma	No Thymoma																
293	[s4_thym_ther_type] Show the field ONLY if: [arm-number]=4 AND [s4_thym_ther_type_known] = 'yes'	Thymoma therapy	checkbox, Required <table border="1"> <tr><td>surgery</td><td>s4_thym_ther_type__surgery</td></tr> <tr><td>chemotherapy</td><td>s4_thym_ther_type__chemotherapy</td></tr> <tr><td>radiotherapy</td><td>s4_thym_ther_type__radiotherapy</td></tr> </table>	surgery	s4_thym_ther_type__surgery	chemotherapy	s4_thym_ther_type__chemotherapy	radiotherapy	s4_thym_ther_type__radiotherapy								
surgery	s4_thym_ther_type__surgery																
chemotherapy	s4_thym_ther_type__chemotherapy																
radiotherapy	s4_thym_ther_type__radiotherapy																
294	[s4_had_thym_relapse] Show the field ONLY if: [arm-number]=4	Thymoma relapse	radio <table border="1"> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	yes	Yes	no	No	unknown	Unknown								
yes	Yes																
no	No																
unknown	Unknown																
295	[s4_date_of_thym_relapse] Show the field ONLY if: [arm-number]=4 AND [s4_had_thym_relapse]='yes'	Date of thymoma relapse yyyy-mm-dd <i>If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.</i>	text (date_ymd)														
296	[njd_thymus_complete]	Section Header: <i>Form Status</i> Complete?	dropdown <table border="1"> <tr><td>0</td><td>Incomplete</td></tr> <tr><td>1</td><td>Unverified</td></tr> <tr><td>2</td><td>Complete</td></tr> </table>	0	Incomplete	1	Unverified	2	Complete								
0	Incomplete																
1	Unverified																
2	Complete																
<b>Instrument: NJD: Quantitative Myasthenia Gravis Test (QMG) (njd_quantitative_myasthenia_gravis_test_qmg)</b>																	
Active languages: None																	
297	[cms_qmg_h1]	Quantitative Myasthenia Gravis Test (QMG)	descriptive														
298	[date_of_assess_qmg]	Date of assessment yyyy-mm-dd	text (date_ymd), Required														
299	[cms_qmg_subscores_known] Show the field ONLY if: [arm-number]=4	Score of the individual data items is known	radio <table border="1"> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> </table>	yes	Yes	no	No										
yes	Yes																
no	No																
300	[cms_qmg_score_manual] Show the field ONLY if: [cms_qmg_subscores_known] = 'no'	Total QMG Score (manual) <i>Please enter a number in the range [0, 39]</i>	text (number, Min: 0, Max: 39)														
301	[cms_qmg1] Show the field ONLY if:	1. Double vision on lateral gaze right or left (circle one), seconds	radio <table border="1"> <tr><td>0</td><td>61</td></tr> </table>	0	61												
0	61																

	[cms_qmg_subscores_known] = 'yes'		<table border="1"> <tr><td>1</td><td>11-60</td></tr> <tr><td>2</td><td>1-10</td></tr> <tr><td>3</td><td>Spontaneous</td></tr> </table>	1	11-60	2	1-10	3	Spontaneous		
1	11-60										
2	1-10										
3	Spontaneous										
302	[ cms_qmg1_score ] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	Item 1 Score	<p>calc</p> <p>Calculation: if([cms_qmg_subscores_known] = 'yes', if([cms_qmg1] = '0', 0, 0) or if([cms_qmg1] = '1', 1, 0) or if([cms_qmg1] = '2', 2, 0) or if([cms_qmg1] = '3', 3, 0), "")</p> <p>Field Annotation: @HIDDEN @HIDDEN-PDF</p>								
303	[ cms_qmg2 ] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	2. Ptosis (upward gaze), seconds	<p>radio</p> <table border="1"> <tr><td>0</td><td>61</td></tr> <tr><td>1</td><td>11-60</td></tr> <tr><td>2</td><td>1-10</td></tr> <tr><td>3</td><td>Spontaneous</td></tr> </table>	0	61	1	11-60	2	1-10	3	Spontaneous
0	61										
1	11-60										
2	1-10										
3	Spontaneous										
304	[ cms_qmg2_score ] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	Item 2 Score	<p>calc</p> <p>Calculation: if([cms_qmg_subscores_known] = 'yes', if([cms_qmg2] = '0', 0, 0) or if([cms_qmg2] = '1', 1, 0) or if([cms_qmg2] = '2', 2, 0) or if([cms_qmg2] = '3', 3, 0), "")</p> <p>Field Annotation: @HIDDEN @HIDDEN-PDF</p>								
305	[ cms_qmg3 ] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	3. Facial Muscles	<p>radio</p> <table border="1"> <tr><td>0</td><td>Normal lid closure</td></tr> <tr><td>1</td><td>Complete weak, some resistance</td></tr> <tr><td>2</td><td>Complete, without resistance</td></tr> <tr><td>3</td><td>Incomplete</td></tr> </table>	0	Normal lid closure	1	Complete weak, some resistance	2	Complete, without resistance	3	Incomplete
0	Normal lid closure										
1	Complete weak, some resistance										
2	Complete, without resistance										
3	Incomplete										
306	[ cms_qmg3_score ] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	Item 3 Score	<p>calc</p> <p>Calculation: if([cms_qmg_subscores_known] = 'yes', if([cms_qmg3] = '0', 0, 0) or if([cms_qmg3] = '1', 1, 0) or if([cms_qmg3] = '2', 2, 0) or if([cms_qmg3] = '3', 3, 0), "")</p> <p>Field Annotation: @HIDDEN @HIDDEN-PDF</p>								
307	[ cms_qmg4 ] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	4. Swallowing 4 oz. / 120 mL water	<p>radio</p> <table border="1"> <tr><td>0</td><td>Normal</td></tr> <tr><td>1</td><td>Minimal coughing or throat clearing</td></tr> <tr><td>2</td><td>Severe coughing/choking or nasal regurgitation</td></tr> <tr><td>3</td><td>Cannot swallow (test not attempted)</td></tr> </table>	0	Normal	1	Minimal coughing or throat clearing	2	Severe coughing/choking or nasal regurgitation	3	Cannot swallow (test not attempted)
0	Normal										
1	Minimal coughing or throat clearing										
2	Severe coughing/choking or nasal regurgitation										
3	Cannot swallow (test not attempted)										
308	[ cms_qmg4_score ] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	Item 4 Score	<p>calc</p> <p>Calculation: if([cms_qmg_subscores_known] = 'yes', if([cms_qmg4] = '0', 0, 0) or if([cms_qmg4] = '1', 1, 0) or if([cms_qmg4] = '2', 2, 0) or if([cms_qmg4] = '3', 3, 0), "")</p> <p>Field Annotation: @HIDDEN @HIDDEN-PDF</p>								
309	[ cms_qmg5 ] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	5. Speech following counting aloud from 1 to 50 (onset of dysarthria)	<p>radio</p> <table border="1"> <tr><td>0</td><td>None at #50</td></tr> <tr><td>1</td><td>Dysarthria at #30-49</td></tr> <tr><td>2</td><td>Dysarthria at #10-29</td></tr> <tr><td>3</td><td>Dysarthria at #9</td></tr> </table>	0	None at #50	1	Dysarthria at #30-49	2	Dysarthria at #10-29	3	Dysarthria at #9
0	None at #50										
1	Dysarthria at #30-49										
2	Dysarthria at #10-29										
3	Dysarthria at #9										
310	[ cms_qmg5_score ] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	Item 5 Score	<p>calc</p> <p>Calculation: if([cms_qmg_subscores_known] = 'yes', if([cms_qmg5] = '0', 0, 0) or if([cms_qmg5] = '1', 1, 0) or if([cms_qmg5] = '2', 2, 0) or if([cms_qmg5] = '3', 3, 0), "")</p> <p>Field Annotation: @HIDDEN @HIDDEN-PDF</p>								
311	[ cms_qmg6 ] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	6. Right arm outstretched (90 degrees sitting), seconds	<p>radio</p> <table border="1"> <tr><td>0</td><td>240</td></tr> <tr><td>1</td><td>90-239</td></tr> <tr><td></td><td></td></tr> </table>	0	240	1	90-239				
0	240										
1	90-239										

			<table border="1"> <tr> <td>2</td> <td>10-89</td> </tr> <tr> <td>3</td> <td>0-9</td> </tr> </table>	2	10-89	3	0-9				
2	10-89										
3	0-9										
312	<p>[ cms_qmg6_score ]</p> <p>Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'</p>	Item 6 Score	<p>calc</p> <p>Calculation: if([cms_qmg_subscores_known] = 'yes', if([cms_qmg6] = '0', 0, 0) or if([cms_qmg6] = '1', 1, 0) or if([cms_qmg6] = '2', 2, 0) or if([cms_qmg6] = '3', 3, 0), "")</p> <p>Field Annotation: @HIDDEN @HIDDEN-PDF</p>								
313	<p>[ cms_qmg7 ]</p> <p>Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'</p>	7. Left arm outstretched (90 degrees sitting), seconds	<p>radio</p> <table border="1"> <tr> <td>0</td> <td>240</td> </tr> <tr> <td>1</td> <td>90-239</td> </tr> <tr> <td>2</td> <td>10-89</td> </tr> <tr> <td>3</td> <td>0-9</td> </tr> </table>	0	240	1	90-239	2	10-89	3	0-9
0	240										
1	90-239										
2	10-89										
3	0-9										
314	<p>[ cms_qmg7_score ]</p> <p>Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'</p>	Item 7 Score	<p>calc</p> <p>Calculation: if([cms_qmg_subscores_known] = 'yes', if([cms_qmg7] = '0', 0, 0) or if([cms_qmg7] = '1', 1, 0) or if([cms_qmg7] = '2', 2, 0) or if([cms_qmg7] = '3', 3, 0), "")</p> <p>Field Annotation: @HIDDEN @HIDDEN-PDF</p>								
315	<p>[ cms_qmg8 ]</p> <p>Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'</p>	8. Vital capacity (% predicted) mouthpiece or facemask (circle 1, best of 3)	<p>radio</p> <table border="1"> <tr> <td>0</td> <td>greater than or equal to 80%</td> </tr> <tr> <td>1</td> <td>65-79%</td> </tr> <tr> <td>2</td> <td>50-64%</td> </tr> <tr> <td>3</td> <td>&lt; 50%</td> </tr> </table>	0	greater than or equal to 80%	1	65-79%	2	50-64%	3	< 50%
0	greater than or equal to 80%										
1	65-79%										
2	50-64%										
3	< 50%										
316	<p>[ cms_qmg8_score ]</p> <p>Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'</p>	Item 8 Score	<p>calc</p> <p>Calculation: if([cms_qmg_subscores_known] = 'yes', if([cms_qmg8] = '0', 0, 0) or if([cms_qmg8] = '1', 1, 0) or if([cms_qmg8] = '2', 2, 0) or if([cms_qmg8] = '3', 3, 0), "")</p> <p>Field Annotation: @HIDDEN @HIDDEN-PDF</p>								
317	<p>[ cms_qmg9 ]</p> <p>Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'</p>	9. Right hand grip: (best of 2), kgW	<p>radio</p> <table border="1"> <tr> <td>0</td> <td>Male 45+, Female 30+</td> </tr> <tr> <td>1</td> <td>Male 15-44, Female 10-29</td> </tr> <tr> <td>2</td> <td>Male 5-14, Female 5-9</td> </tr> <tr> <td>3</td> <td>0-4</td> </tr> </table>	0	Male 45+, Female 30+	1	Male 15-44, Female 10-29	2	Male 5-14, Female 5-9	3	0-4
0	Male 45+, Female 30+										
1	Male 15-44, Female 10-29										
2	Male 5-14, Female 5-9										
3	0-4										
318	<p>[ cms_qmg9_score ]</p> <p>Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'</p>	Item 9 Score	<p>calc</p> <p>Calculation: if([cms_qmg_subscores_known] = 'yes', if([cms_qmg9] = '0', 0, 0) or if([cms_qmg9] = '1', 1, 0) or if([cms_qmg9] = '2', 2, 0) or if([cms_qmg9] = '3', 3, 0), "")</p> <p>Field Annotation: @HIDDEN @HIDDEN-PDF</p>								
319	<p>[ cms_qmg10 ]</p> <p>Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'</p>	10. Left hand grip: (best of 2), kgW	<p>radio</p> <table border="1"> <tr> <td>0</td> <td>Male 45+, Female 30+</td> </tr> <tr> <td>1</td> <td>Male 15-44, Female 10-29</td> </tr> <tr> <td>2</td> <td>Male 5-14, Female 5-9</td> </tr> <tr> <td>3</td> <td>0-4</td> </tr> </table>	0	Male 45+, Female 30+	1	Male 15-44, Female 10-29	2	Male 5-14, Female 5-9	3	0-4
0	Male 45+, Female 30+										
1	Male 15-44, Female 10-29										
2	Male 5-14, Female 5-9										
3	0-4										
320	<p>[ cms_qmg10_score ]</p> <p>Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'</p>	Item 10 Score	<p>calc</p> <p>Calculation: if([cms_qmg_subscores_known] = 'yes', if([cms_qmg10] = '0', 0, 0) or if([cms_qmg10] = '1', 1, 0) or if([cms_qmg10] = '2', 2, 0) or if([cms_qmg10] = '3', 3, 0), "")</p> <p>Field Annotation: @HIDDEN @HIDDEN-PDF</p>								
321	<p>[ cms_qmg11 ]</p> <p>Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'</p>	11. Head, lifted (45 degrees supine), seconds	<p>radio</p> <table border="1"> <tr> <td>0</td> <td>120</td> </tr> <tr> <td>1</td> <td>30-119</td> </tr> <tr> <td>2</td> <td>1-29</td> </tr> <tr> <td>3</td> <td>0</td> </tr> </table>	0	120	1	30-119	2	1-29	3	0
0	120										
1	30-119										
2	1-29										
3	0										

322	[ cms_qmg11_score ] Show the field ONLY if: [ cms_qmg_subscores_known ] = 'yes'	Item 11 Score	calc Calculation: if([ cms_qmg_subscores_known ] = 'yes', if([ cms_qmg11 ] = '0', 0, 0) or if([ cms_qmg11 ] = '1', 1, 0) or if([ cms_qmg11 ] = '2', 2, 0) or if([ cms_qmg11 ] = '3', 3, 0), "") Field Annotation: @HIDDEN @HIDDEN-PDF								
323	[ cms_qmg12 ] Show the field ONLY if: [ cms_qmg_subscores_known ] = 'yes'	12. Right leg outstretched (45 degrees supine), seconds	radio <table border="1"> <tr><td>0</td><td>100</td></tr> <tr><td>1</td><td>31-99</td></tr> <tr><td>2</td><td>1-30</td></tr> <tr><td>3</td><td>0</td></tr> </table>	0	100	1	31-99	2	1-30	3	0
0	100										
1	31-99										
2	1-30										
3	0										
324	[ cms_qmg12_score ] Show the field ONLY if: [ cms_qmg_subscores_known ] = 'yes'	Item 12 Score	calc Calculation: if([ cms_qmg_subscores_known ] = 'yes', if([ cms_qmg12 ] = '0', 0, 0) or if([ cms_qmg12 ] = '1', 1, 0) or if([ cms_qmg12 ] = '2', 2, 0) or if([ cms_qmg12 ] = '3', 3, 0), "") Field Annotation: @HIDDEN @HIDDEN-PDF								
325	[ cms_qmg13 ] Show the field ONLY if: [ cms_qmg_subscores_known ] = 'yes'	13. Left leg outstretched (45 degrees supine), seconds	radio <table border="1"> <tr><td>0</td><td>100</td></tr> <tr><td>1</td><td>31-99</td></tr> <tr><td>2</td><td>1-30</td></tr> <tr><td>3</td><td>0</td></tr> </table>	0	100	1	31-99	2	1-30	3	0
0	100										
1	31-99										
2	1-30										
3	0										
326	[ cms_qmg13_score ] Show the field ONLY if: [ cms_qmg_subscores_known ] = 'yes'	Item 13 Score	calc Calculation: if([ cms_qmg_subscores_known ] = 'yes', if([ cms_qmg13 ] = '0', 0, 0) or if([ cms_qmg13 ] = '1', 1, 0) or if([ cms_qmg13 ] = '2', 2, 0) or if([ cms_qmg13 ] = '3', 3, 0), "") Field Annotation: @HIDDEN @HIDDEN-PDF								
327	[ cms_qmg_score_calc ] Show the field ONLY if: [ cms_qmg_subscores_known ] = 'yes'	Total QMG Score (calculated)	calc Calculation: if([ cms_qmg_subscores_known ] = 'yes', sum([ cms_qmg1_score ], [ cms_qmg2_score ], [ cms_qmg3_score ], [ cms_qmg4_score ], [ cms_qmg5_score ], [ cms_qmg6_score ], [ cms_qmg7_score ], [ cms_qmg8_score ], [ cms_qmg9_score ], [ cms_qmg10_score ], [ cms_qmg11_score ], [ cms_qmg12_score ], [ cms_qmg13_score ]), "") Field Annotation: @READONLY								
328	[ njd_quantitative_myasthenia_gravis_test_qmg_complete ]	Section Header: <i>Form Status</i> Complete?	dropdown <table border="1"> <tr><td>0</td><td>Incomplete</td></tr> <tr><td>1</td><td>Unverified</td></tr> <tr><td>2</td><td>Complete</td></tr> </table>	0	Incomplete	1	Unverified	2	Complete		
0	Incomplete										
1	Unverified										
2	Complete										

**Instrument: Motorneuron FRS-R (Age 16+ Only) (motorneuron\_frsr\_age\_16\_only)**

Active languages: None

329	[ s5_als_frs_h1 ]	MOTORNEURON: FRS-R (AGE >= 16 ONLY)	descriptive										
330	[ s5_als_frs_description 1 ]	Section Header: <i>Notes on editing</i> ALS Functional Rating Scale - Revised (ALS-FRS-R) must be completed for adults only (age at vist >= 16).	descriptive										
331	[ date_of_assess_frsr ]	Date of assessment yyyy-mm-dd	text (date_ymd), Required										
332	[ s5_als_frs_r_speech ]	Section Header: <i>ALS Functional Rating Scale - Revised (ALS-FRS-R)</i> 1. Speech	radio <table border="1"> <tr><td>0</td><td>Loss of useful speech</td></tr> <tr><td>1</td><td>Speech combined with nonvocal communication</td></tr> <tr><td>2</td><td>Intelligible with repeating</td></tr> <tr><td>3</td><td>Detectable speech disturbance</td></tr> <tr><td>4</td><td>Normal speech process</td></tr> </table>	0	Loss of useful speech	1	Speech combined with nonvocal communication	2	Intelligible with repeating	3	Detectable speech disturbance	4	Normal speech process
0	Loss of useful speech												
1	Speech combined with nonvocal communication												
2	Intelligible with repeating												
3	Detectable speech disturbance												
4	Normal speech process												

333	[s5_als_frs_r_saliv]	2. Salivation	radio <table border="1"> <tr><td>0</td><td>Marked drooling; requires constant tissue or handkerchief</td></tr> <tr><td>1</td><td>Marked excessive saliva wth some drooling</td></tr> <tr><td>2</td><td>Moderately excessive saliva; may have minimal drooling</td></tr> <tr><td>3</td><td>Slight but definitive excessive saliva in mouth; may have nighttime drooling</td></tr> <tr><td>4</td><td>Normal</td></tr> </table>	0	Marked drooling; requires constant tissue or handkerchief	1	Marked excessive saliva wth some drooling	2	Moderately excessive saliva; may have minimal drooling	3	Slight but definitive excessive saliva in mouth; may have nighttime drooling	4	Normal
0	Marked drooling; requires constant tissue or handkerchief												
1	Marked excessive saliva wth some drooling												
2	Moderately excessive saliva; may have minimal drooling												
3	Slight but definitive excessive saliva in mouth; may have nighttime drooling												
4	Normal												
334	[s5_als_frs_r_swallow]	3. Swallowing	radio <table border="1"> <tr><td>0</td><td>Nothing by mouth; exclusively parenteral or enteral feeding</td></tr> <tr><td>1</td><td>Needs supplemental tube feeding</td></tr> <tr><td>2</td><td>Dietary consistency changes</td></tr> <tr><td>3</td><td>Early eating problems; occasional chocking</td></tr> <tr><td>4</td><td>Normal eating habits</td></tr> </table>	0	Nothing by mouth; exclusively parenteral or enteral feeding	1	Needs supplemental tube feeding	2	Dietary consistency changes	3	Early eating problems; occasional chocking	4	Normal eating habits
0	Nothing by mouth; exclusively parenteral or enteral feeding												
1	Needs supplemental tube feeding												
2	Dietary consistency changes												
3	Early eating problems; occasional chocking												
4	Normal eating habits												
335	[s5_als_frs_r_handwr]	4. Handwriting	radio <table border="1"> <tr><td>0</td><td>Unable to grip pen</td></tr> <tr><td>1</td><td>Able to grip pen but unable to write</td></tr> <tr><td>2</td><td>Not all words are legible</td></tr> <tr><td>3</td><td>Slow or sloppy; all words are legible</td></tr> <tr><td>4</td><td>Normal</td></tr> </table>	0	Unable to grip pen	1	Able to grip pen but unable to write	2	Not all words are legible	3	Slow or sloppy; all words are legible	4	Normal
0	Unable to grip pen												
1	Able to grip pen but unable to write												
2	Not all words are legible												
3	Slow or sloppy; all words are legible												
4	Normal												
336	[s5_als_frs_r_gastron]	ALS-FRS-R Patient with gastrostomy and >50% daily nutrition intake via G-tube	yesno <table border="1"> <tr><td>1</td><td>Yes</td></tr> <tr><td>0</td><td>No</td></tr> </table> <p>Field Annotation: @DEFAULT = 0</p>	1	Yes	0	No						
1	Yes												
0	No												
337	[s5_als_frs_r_utensils_a]  Show the field ONLY if: [s5_als_frs_r_gastron] = 0	5. Cutting food and handling utensils (patients without gastrostomy)	radio <table border="1"> <tr><td>0</td><td>Needs to be fed</td></tr> <tr><td>1</td><td>Food must be cut by someone but can still feed slowly</td></tr> <tr><td>2</td><td>Can cut most foods although clumsy and slow; some help needed</td></tr> <tr><td>3</td><td>Somewhat slow and clumsy but no help needed</td></tr> <tr><td>4</td><td>Normal</td></tr> </table>	0	Needs to be fed	1	Food must be cut by someone but can still feed slowly	2	Can cut most foods although clumsy and slow; some help needed	3	Somewhat slow and clumsy but no help needed	4	Normal
0	Needs to be fed												
1	Food must be cut by someone but can still feed slowly												
2	Can cut most foods although clumsy and slow; some help needed												
3	Somewhat slow and clumsy but no help needed												
4	Normal												
338	[s5_als_frs_r_utensils_b]  Show the field ONLY if: [s5_als_frs_r_gastron] = 1	5. Cutting food and handling utensils (alternate scale for patients with gastrostomy)	radio <table border="1"> <tr><td>0</td><td>Unable to perform any aspect of task</td></tr> <tr><td>1</td><td>Provides minimal assistance to caregiver</td></tr> <tr><td>2</td><td>Some help needed with closures and fasteners</td></tr> <tr><td>3</td><td>Clumsy but able to perform all manipulations independently</td></tr> <tr><td>4</td><td>Normal</td></tr> </table>	0	Unable to perform any aspect of task	1	Provides minimal assistance to caregiver	2	Some help needed with closures and fasteners	3	Clumsy but able to perform all manipulations independently	4	Normal
0	Unable to perform any aspect of task												
1	Provides minimal assistance to caregiver												
2	Some help needed with closures and fasteners												
3	Clumsy but able to perform all manipulations independently												
4	Normal												
339	[s5_als_frs_r_dressing]	6. Dressing and hygiene	radio <table border="1"> <tr><td>0</td><td>Total dependence</td></tr> <tr><td>1</td><td>Needs attendant for self-care</td></tr> <tr><td>2</td><td>Intermittent assistance or substitute methods</td></tr> <tr><td>3</td><td>Independent and complete self-care with effort or decreased efficiency</td></tr> <tr><td>4</td><td>Normal function</td></tr> </table>	0	Total dependence	1	Needs attendant for self-care	2	Intermittent assistance or substitute methods	3	Independent and complete self-care with effort or decreased efficiency	4	Normal function
0	Total dependence												
1	Needs attendant for self-care												
2	Intermittent assistance or substitute methods												
3	Independent and complete self-care with effort or decreased efficiency												
4	Normal function												
340	[s5_als_frs_r_bed]	7. Turning in bed and adjusting bed clothes	radio <table border="1"> <tr><td>0</td><td>Helpless</td></tr> </table>	0	Helpless								
0	Helpless												



			<table border="1"> <tr> <td>1</td> <td>Can initiate but not turn or adjust sheets alone</td> </tr> <tr> <td>2</td> <td>Can turn alone or adjust sheets but with great difficulty</td> </tr> <tr> <td>3</td> <td>Somewhat slow and clumsy but no help needed</td> </tr> <tr> <td>4</td> <td>Normal</td> </tr> </table>	1	Can initiate but not turn or adjust sheets alone	2	Can turn alone or adjust sheets but with great difficulty	3	Somewhat slow and clumsy but no help needed	4	Normal		
1	Can initiate but not turn or adjust sheets alone												
2	Can turn alone or adjust sheets but with great difficulty												
3	Somewhat slow and clumsy but no help needed												
4	Normal												
341	[s5_als_frs_r_walking]	8. Walking	radio <table border="1"> <tr> <td>0</td> <td>No purposeful leg movement</td> </tr> <tr> <td>1</td> <td>Nonambulatory functional movement</td> </tr> <tr> <td>2</td> <td>Walks with assistance</td> </tr> <tr> <td>3</td> <td>Early ambulation difficulties</td> </tr> <tr> <td>4</td> <td>Normal</td> </tr> </table>	0	No purposeful leg movement	1	Nonambulatory functional movement	2	Walks with assistance	3	Early ambulation difficulties	4	Normal
0	No purposeful leg movement												
1	Nonambulatory functional movement												
2	Walks with assistance												
3	Early ambulation difficulties												
4	Normal												
342	[s5_als_frs_r_stairs]	9. Climbing stairs	radio <table border="1"> <tr> <td>0</td> <td>Cannot do</td> </tr> <tr> <td>1</td> <td>Needs assistance</td> </tr> <tr> <td>2</td> <td>Mild unsteadiness or fatigue</td> </tr> <tr> <td>3</td> <td>Slow</td> </tr> <tr> <td>4</td> <td>Normal</td> </tr> </table>	0	Cannot do	1	Needs assistance	2	Mild unsteadiness or fatigue	3	Slow	4	Normal
0	Cannot do												
1	Needs assistance												
2	Mild unsteadiness or fatigue												
3	Slow												
4	Normal												
343	[s5_als_frs_r_dyspnea]	10. Dyspnea	radio <table border="1"> <tr> <td>0</td> <td>Significant difficulty; considering using mechanical respiratory support</td> </tr> <tr> <td>1</td> <td>Occurs at rest; difficulty breathing when either sitting or lying</td> </tr> <tr> <td>2</td> <td>Occurs with one or more of the following: eating, bathing, dressing (ADL)</td> </tr> <tr> <td>3</td> <td>Occurs when walking</td> </tr> <tr> <td>4</td> <td>None</td> </tr> </table>	0	Significant difficulty; considering using mechanical respiratory support	1	Occurs at rest; difficulty breathing when either sitting or lying	2	Occurs with one or more of the following: eating, bathing, dressing (ADL)	3	Occurs when walking	4	None
0	Significant difficulty; considering using mechanical respiratory support												
1	Occurs at rest; difficulty breathing when either sitting or lying												
2	Occurs with one or more of the following: eating, bathing, dressing (ADL)												
3	Occurs when walking												
4	None												
344	[s5_als_frs_r_orthopnea]	11. Orthopnea	radio <table border="1"> <tr> <td>0</td> <td>Unable to sleep</td> </tr> <tr> <td>1</td> <td>Can only sleep sitting up</td> </tr> <tr> <td>2</td> <td>Needs extra pillows in order to sleep (more than two)</td> </tr> <tr> <td>3</td> <td>Some difficulty sleeping at night due to shortness of breath; does not routinely use more than two pillows</td> </tr> <tr> <td>4</td> <td>None</td> </tr> </table>	0	Unable to sleep	1	Can only sleep sitting up	2	Needs extra pillows in order to sleep (more than two)	3	Some difficulty sleeping at night due to shortness of breath; does not routinely use more than two pillows	4	None
0	Unable to sleep												
1	Can only sleep sitting up												
2	Needs extra pillows in order to sleep (more than two)												
3	Some difficulty sleeping at night due to shortness of breath; does not routinely use more than two pillows												
4	None												
345	[s5_als_frs_r_resp_insuff]	12. Respiratory insufficiency	radio <table border="1"> <tr> <td>0</td> <td>Invasive mechanical ventilation by intubation or trachostomy</td> </tr> <tr> <td>1</td> <td>Continuous use of BiPAP during night and day</td> </tr> <tr> <td>2</td> <td>Continuous use of BiPAP during the night</td> </tr> <tr> <td>3</td> <td>Intermittent use of BiPAP</td> </tr> <tr> <td>4</td> <td>None</td> </tr> </table>	0	Invasive mechanical ventilation by intubation or trachostomy	1	Continuous use of BiPAP during night and day	2	Continuous use of BiPAP during the night	3	Intermittent use of BiPAP	4	None
0	Invasive mechanical ventilation by intubation or trachostomy												
1	Continuous use of BiPAP during night and day												
2	Continuous use of BiPAP during the night												
3	Intermittent use of BiPAP												
4	None												
346	[s5_als_frs_description2]	Section Header: <i>ALS-FRS-R Total Score</i> Please note: ALS-FRS-R Total score will only be calculated if all 12 questions have been answered.	descriptive										
347	[s5_als_frs_r_total_score]	ALS-FRS-R total score	calc Calculation: $if([s5\_als\_frs\_r\_gastron]=0, ([s5\_als\_frs\_r\_speech]+ [s5\_als\_frs\_r\_saliv]+ [s5\_als\_frs\_r\_swallow]+ [s5\_als\_frs\_r\_handwr]+ [s5\_als\_frs\_r\_utensils\_a]+ [s5\_als\_frs\_r\_dressing]+ [s5\_als\_frs\_r\_bed]+ [s5\_als\_frs\_r\_walking]+ [s5\_als\_frs\_r\_stairs]+ [s5\_als\_frs\_r\_dyspnea]+ [s5\_als\_frs\_r\_orthopnea]+ [s5\_als\_frs\_r\_resp\_insuf]), ([s5\_als\_frs\_r\_speech]+ [s5\_als\_frs\_r\_saliv]+$										

			[s5_als_frs_r_swallow]+ [s5_als_frs_r_handwr]+ [s5_als_frs_r_utensils_b]+ [s5_als_frs_r_dressing]+ [s5_als_frs_r_bed]+ [s5_als_frs_r_walking]+ [s5_als_frs_r_stairs]+ [s5_als_frs_r_dyspnea]+ [s5_als_frs_r_orthopnea]+ [s5_als_frs_r_resp_insuf] ) )						
348	[ <a href="#">motorneuron_frsr_age_16_only_complete</a> ]	Section Header: <i>Form Status</i> Complete?	dropdown <table border="1"> <tr><td>0</td><td>Incomplete</td></tr> <tr><td>1</td><td>Unverified</td></tr> <tr><td>2</td><td>Complete</td></tr> </table>	0	Incomplete	1	Unverified	2	Complete
0	Incomplete								
1	Unverified								
2	Complete								

**Instrument: Motor Function Measures (motor\_function\_measures)**

Active languages: None

349	[ <a href="#">motor_measures_h1</a> ]	Motor Function Measures	descriptive				
350	[ <a href="#">date_of_assess_mm</a> ]	Date of assessment yyyy-mm-dd	text (date_ymd), Required				
351	[ <a href="#">age_at_visit_mm</a> ] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2)	Age of patient (years)	calc Calculation: if([arm-number]=5, rounddown(datediff([enrolment_and_firs_arm_5][date_of_birth], [date_of_assess_mm], "y")), if([arm-number]=2, rounddown(datediff([enrolment_and_firs_arm_2][date_of_birth], [date_of_assess_mm], "y")), -999))				
352	[ <a href="#">show_mm_child</a> ] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [age_at_visit_mm] < 16	Section Header: <i>Children only motor measures (age &lt; 16)</i> Motor Measure Performed? Value / Result Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders (CHOP-INTEND) {s5_chop_intend_taken} {s5_chop_intend_score} Hammersmith Functional Motor Scale (HFMS) {hfms_taken} {hfms_score} Hammersmith Functional Motor Scale Expanded (HFMS-E) {hfmse_taken} {hfmse_score} Hammersmith Infant Neurological Examination (HINE) Section 2 {hine_taken} {hine_score}	descriptive				
353	[ <a href="#">s5_chop_intend_taken</a> ] Show the field ONLY if: [arm-number]=5 AND [age_at_visit_mm] < 16	s5_chop_intend_taken	yesno <table border="1"> <tr><td>1</td><td>Yes</td></tr> <tr><td>0</td><td>No</td></tr> </table>	1	Yes	0	No
1	Yes						
0	No						
354	[ <a href="#">s5_chop_intend_score</a> ] Show the field ONLY if: [arm-number]=5 AND [age_at_visit_mm] < 16 AND [s5_chop_intend_taken] = 1	CHOP-INTEND-Score <i>Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders</i>	text (number)				
355	[ <a href="#">hfms_taken</a> ] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [age_at_visit_mm] < 16	hfms_taken	yesno <table border="1"> <tr><td>1</td><td>Yes</td></tr> <tr><td>0</td><td>No</td></tr> </table>	1	Yes	0	No
1	Yes						
0	No						
356	[ <a href="#">hfms_score</a> ] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [age_at_visit_mm] < 16 AND [hfms_taken] = 1	HFMS-Score <i>Hammersmith Functional Motor Scale</i>	text (number)				
357	[ <a href="#">hfmse_taken</a> ] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [age_at_visit_mm] < 16	hfmse_taken	yesno <table border="1"> <tr><td>1</td><td>Yes</td></tr> <tr><td>0</td><td>No</td></tr> </table>	1	Yes	0	No
1	Yes						
0	No						
358	[ <a href="#">hfmse_score</a> ] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [age_at_visit_mm] < 16 AND [hfmse_taken] = 1	HFMS-E-Score <i>Hammersmith Functional Motor Scale Expanded</i>	text (number)				

359	[hine_taken] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [age_at_visit_mm] < 16	hine_taken	yesno 1 Yes 0 No
360	[hine_score] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [age_at_visit_mm] < 16 AND [hine_taken] = 1	HINE-Section2-Score <i>Hammersmith Infant Neurological Examination (HINE) Section 2</i>	text (number)
361	[s5_show_mm_adult] Show the field ONLY if: [arm-number]=5 AND [age_at_visit_mm] >= 16	Section Header: <i>Adult only motor mesures (age &gt;= 16)</i> Motor Measure Performed? Value / Result Adult Test of Neuromuscular Disorders (CHOP-ATEND) {s5_chop_atend_taken} {s5_chop_atend_score}	descriptive
362	[s5_chop_atend_taken] Show the field ONLY if: [arm-number]=5 AND [age_at_visit_mm] >= 16	s5_chop_atend_taken	yesno 1 Yes 0 No
363	[s5_chop_atend_score] Show the field ONLY if: [arm-number]=5 AND [age_at_visit_mm] >= 16 AND [s5_chop_atend_taken] = 1	CHOP-ATEND-Score <i>Adult Test of Neuromuscular Disorders (CHOP-ATEND)</i>	text (number)
364	[show_mm_general] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2)	Section Header: <i>Age independent motor measures</i> Motor Measure Performed? Value / Result Revised Upper Limb Modul (RULM) {rulm_taken} {rulm_score} Motor Function Measurement (MFM) {mfm_taken} {mfm_result} 6 Minute Walk Test: Result in metres (m) {six_mwt_taken} {six_mwt_result} 10 Minute Walk Test: Result in metres (m) {ten_mwt_taken} {ten_mwt_result} Timed "Up and Go" (TUG) {tug_taken} {tug_result} Other motor measure {other_mm_taken} {which_other_mm}	descriptive
365	[rulm_taken] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2)	rulm_taken	yesno 1 Yes 0 No
366	[rulm_score] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [rulm_taken] = 1	RULM-Score <i>Revised Upper Limb Measure (RULM)</i>	text (number)
367	[mfm_taken] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2)	mfm_taken	yesno 1 Yes 0 No
368	[mfm_result] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [mfm_taken] = 1	MFM-Result <i>Motor Function Measurement (MFM)</i>	text (number)
369	[six_mwt_taken] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2)	six_mwt_taken	yesno 1 Yes 0 No
370	[six_mwt_result] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [six_mwt_taken] = 1	6 Minute Walk Test (6MWT)	text (number)
371	[ten_mwt_taken]	ten_mwt_taken	yesno

	Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2)		<table border="1"> <tr><td>1</td><td>Yes</td></tr> <tr><td>0</td><td>No</td></tr> </table>	1	Yes	0	No		
1	Yes								
0	No								
372	[ten_mwt_result]  Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [ten_mwt_taken] = 1	10 Minute Walk Test (10MWT)	text (number)						
373	[tug_taken]  Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2)	tug_taken	yesno <table border="1"> <tr><td>1</td><td>Yes</td></tr> <tr><td>0</td><td>No</td></tr> </table>	1	Yes	0	No		
1	Yes								
0	No								
374	[tug_result]  Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [tug_taken] = 1	TUG-Result <i>Timed "Up &amp; Go" (TUG)</i>	text (number)						
375	[other_mm_taken]  Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2)	s5_other_mm_taken	yesno <table border="1"> <tr><td>1</td><td>Yes</td></tr> <tr><td>0</td><td>No</td></tr> </table>	1	Yes	0	No		
1	Yes								
0	No								
376	[which_other_mm]  Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [other_mm_taken] = 1	Other motor measure <i>Which other motor measure has been performed?</i>	text						
377	[motor_function_measures_complete]	Section Header: <i>Form Status</i> Complete?	dropdown <table border="1"> <tr><td>0</td><td>Incomplete</td></tr> <tr><td>1</td><td>Unverified</td></tr> <tr><td>2</td><td>Complete</td></tr> </table>	0	Incomplete	1	Unverified	2	Complete
0	Incomplete								
1	Unverified								
2	Complete								

**Instrument: Genetic Diagnosis: Changes in nuclear DNA (genetic\_diagnosis\_changes\_in\_nuclear\_dna)**

Active languages: None

378	[gendia_h1]	Genetic Diagnosis: Changes in nuclear DNA	descriptive				
379	[date_of_gendia]	Date of genetic diagnosis [or if no genetic diagnosis: date of report] yyyy-mm-dd	text (date_ymd), Required				
380	[gendia_known]	Do you have a genetic diagnosis for this patient?	radio, Required <table border="1"> <tr><td>yes</td><td>Yes</td></tr> <tr><td>no</td><td>No</td></tr> </table>	yes	Yes	no	No
yes	Yes						
no	No						
381	[gendia_label]  Show the field ONLY if: [gendia_known] = 'yes'	Genetic diagnosis name <i>This is a label to identify this diagnostic record to the registry user. It will be displayed on the record home page together with the date of diagnosis. You can name it anything you find helpful - consider something like the gene name and inheritance pattern or similar.</i>	text, Required				
382	[gendia_label_disp]	gendia_label_disp	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_known]='yes',concat([gendia_label'],' (', [date_of_gendia], ')'),'[date_of_gendia]'))				
383	[gendia_desc1]  Show the field ONLY if: [gendia_known] = 'yes'	The disease-group of this patient is: [arm-label]The clinical diagnosis of the neuromuscular disease for this patient is (from Baseline-form): [first-event-name] [diagnosis_rd_orpha] ([first-event-name] [diagnosis_rd_orpha:value])	descriptive				
384	[gendia_listpref]  Show the field ONLY if: [gendia_known] = 'yes'	Which gene is affected? <i>Genetic diagnosis retained by the specialised centre (Element 6.2. in the Set of common data elements for Rare Diseases Registration (link))</i>	radio, Required <table border="1"> <tr><td>all_nmd</td><td>Search in full neuromuscular disease gene list</td></tr> <tr><td>ontology</td><td>Gene is not in neuromuscular disease gene list</td></tr> </table>	all_nmd	Search in full neuromuscular disease gene list	ontology	Gene is not in neuromuscular disease gene list
all_nmd	Search in full neuromuscular disease gene list						
ontology	Gene is not in neuromuscular disease gene list						

			Field Annotation: @DEFAULT=if([show_snv_fields]=1,'disease_group','')																																																
385	<p>[gendia_omim_list]</p> <p>Show the field ONLY if: [gendia_listpref]='all_nmd'</p>	<p>Gene Symbol (HGNC) for all neuromuscular diseases</p> <p><i>Genetic diagnosis retained by the specialised centre (Element 6.2. in the Set of common data elements for Rare Diseases Registration (link))</i></p>	<p>dropdown (autocomplete), Required</p> <table border="1"> <tr> <td>601065</td> <td>AARS1 / HGNC:20 / OMIM:601065 (alanyl-tRNA synthetase 1)</td> </tr> <tr> <td>612035</td> <td>AARS2 / HGNC:21022 / OMIM:612035 (alanyl-tRNA synthetase 2, mitochondrial)</td> </tr> <tr> <td>601439</td> <td>ABCC9 / HGNC:60 / OMIM:601439 (ATP binding cassette subfamily C member 9)</td> </tr> <tr> <td>604780</td> <td>ABHD5 / HGNC:21396 / OMIM:604780 (abhydrolase domain containing 5, lysophosphatidic acid acyltransferase)</td> </tr> <tr> <td>611103</td> <td>ACAD9 / HGNC:21497 / OMIM:611103 (acyl-CoA dehydrogenase family member 9)</td> </tr> <tr> <td>609575</td> <td>ACADVL / HGNC:92 / OMIM:609575 (acyl-CoA dehydrogenase very long chain)</td> </tr> <tr> <td>617036</td> <td>ACER3 / HGNC:16066 / OMIM:617036 (alkaline ceramidase 3)</td> </tr> <tr> <td>102610</td> <td>ACTA1 / HGNC:129 / OMIM:102610 (actin alpha 1, skeletal muscle)</td> </tr> <tr> <td>102540</td> <td>ACTC1 / HGNC:143 / OMIM:102540 (actin alpha cardiac muscle 1)</td> </tr> <tr> <td>102573</td> <td>ACTN2 / HGNC:164 / OMIM:102573 (actinin alpha 2)</td> </tr> <tr> <td>102576</td> <td>ACVR1 / HGNC:171 / OMIM:102576 (activin A receptor type 1)</td> </tr> <tr> <td>606980</td> <td>COQ8A / HGNC:16812 / OMIM:606980 (coenzyme Q8A)</td> </tr> <tr> <td>600294</td> <td>ADCY6 / HGNC:237 / OMIM:600294 (adenylate cyclase 6)</td> </tr> <tr> <td>612243</td> <td>ADGRG6 / HGNC:13841 / OMIM:612243 (adhesion G protein-coupled receptor G6)</td> </tr> <tr> <td>612498</td> <td>ADSS1 / HGNC:20093 / OMIM:612498 (adenylosuccinate synthase 1)</td> </tr> <tr> <td>604581</td> <td>AFG3L2 / HGNC:315 / OMIM:604581 (AFG3 like matrix AAA peptidase subunit 2)</td> </tr> <tr> <td>610860</td> <td>AGL / HGNC:321 / OMIM:610860 (amylase-like 1, 6-glucosidase, 4-alpha-glucanotransferase)</td> </tr> <tr> <td>103320</td> <td>AGRN / HGNC:329 / OMIM:103320 (agrin)</td> </tr> <tr> <td>606830</td> <td>AGTPBP1 / HGNC:17258 / OMIM:606830 (ATP/GTP binding carboxypeptidase 1)</td> </tr> <tr> <td>608570</td> <td>AHNAK2 / HGNC:20125 / OMIM:608570 (AHNAK nucleoprotein 2)</td> </tr> <tr> <td>300169</td> <td>AIFM1 / HGNC:8768 / OMIM:300169 (apoptosis inducing factor mitochondria associated 1)</td> </tr> <tr> <td>604001</td> <td>AKAP9 / HGNC:379 / OMIM:604001 (A-kinase anchoring protein 9)</td> </tr> <tr> <td>138250</td> <td>ALDH18A1 / HGNC:9722 / OMIM:138250 (aldehyde dehydrogenase 18 family member A1)</td> </tr> <tr> <td>609523</td> <td>ALDH3A2 / HGNC:403 / OMIM:609523 (aldehyde dehydrogenase 3 family member A2)</td> </tr> </table>	601065	AARS1 / HGNC:20 / OMIM:601065 (alanyl-tRNA synthetase 1)	612035	AARS2 / HGNC:21022 / OMIM:612035 (alanyl-tRNA synthetase 2, mitochondrial)	601439	ABCC9 / HGNC:60 / OMIM:601439 (ATP binding cassette subfamily C member 9)	604780	ABHD5 / HGNC:21396 / OMIM:604780 (abhydrolase domain containing 5, lysophosphatidic acid acyltransferase)	611103	ACAD9 / HGNC:21497 / OMIM:611103 (acyl-CoA dehydrogenase family member 9)	609575	ACADVL / HGNC:92 / OMIM:609575 (acyl-CoA dehydrogenase very long chain)	617036	ACER3 / HGNC:16066 / OMIM:617036 (alkaline ceramidase 3)	102610	ACTA1 / HGNC:129 / OMIM:102610 (actin alpha 1, skeletal muscle)	102540	ACTC1 / HGNC:143 / OMIM:102540 (actin alpha cardiac muscle 1)	102573	ACTN2 / HGNC:164 / OMIM:102573 (actinin alpha 2)	102576	ACVR1 / HGNC:171 / OMIM:102576 (activin A receptor type 1)	606980	COQ8A / HGNC:16812 / OMIM:606980 (coenzyme Q8A)	600294	ADCY6 / HGNC:237 / OMIM:600294 (adenylate cyclase 6)	612243	ADGRG6 / HGNC:13841 / OMIM:612243 (adhesion G protein-coupled receptor G6)	612498	ADSS1 / HGNC:20093 / OMIM:612498 (adenylosuccinate synthase 1)	604581	AFG3L2 / HGNC:315 / OMIM:604581 (AFG3 like matrix AAA peptidase subunit 2)	610860	AGL / HGNC:321 / OMIM:610860 (amylase-like 1, 6-glucosidase, 4-alpha-glucanotransferase)	103320	AGRN / HGNC:329 / OMIM:103320 (agrin)	606830	AGTPBP1 / HGNC:17258 / OMIM:606830 (ATP/GTP binding carboxypeptidase 1)	608570	AHNAK2 / HGNC:20125 / OMIM:608570 (AHNAK nucleoprotein 2)	300169	AIFM1 / HGNC:8768 / OMIM:300169 (apoptosis inducing factor mitochondria associated 1)	604001	AKAP9 / HGNC:379 / OMIM:604001 (A-kinase anchoring protein 9)	138250	ALDH18A1 / HGNC:9722 / OMIM:138250 (aldehyde dehydrogenase 18 family member A1)	609523	ALDH3A2 / HGNC:403 / OMIM:609523 (aldehyde dehydrogenase 3 family member A2)
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300776	ALG13 / HGNC:30881 / OMIM:300776 (ALG13 UDP-N-acetylglucosaminyltransferase subunit)
612866	ALG14 / HGNC:28287 / OMIM:612866 (ALG14 UDP-N-acetylglucosaminyltransferase subunit)
607905	ALG2 / HGNC:23159 / OMIM:607905 (ALG2 alpha-1,3/1,6-mannosyltransferase)
617608	ALPK3 / HGNC:17574 / OMIM:617608 (alpha kinase 3)
606352	ALS2 / HGNC:443 / OMIM:606352 (alsin Rho guanine nucleotide exchange factor ALS2)
102771	AMPD2 / HGNC:469 / OMIM:102771 (adenosine monophosphate deaminase 2)
105850	ANG / HGNC:483 / OMIM:105850 (angiogenin)
106410	ANK2 / HGNC:493 / OMIM:106410 (ankyrin 2)
609599	ANKRD1 / HGNC:15819 / OMIM:609599 (ankyrin repeat domain 1)
613726	ANO10 / HGNC:25519 / OMIM:613726 (anoctamin 10)
608662	ANO5 / HGNC:27337 / OMIM:608662 (anoctamin 5)
602572	ANXA11 / HGNC:535 / OMIM:602572 (annexin A11)
607245	AP4B1 / HGNC:572 / OMIM:607245 (adaptor related protein complex 4 subunit beta 1)
607244	AP4E1 / HGNC:573 / OMIM:607244 (adaptor related protein complex 4 subunit epsilon 1)
602296	AP4M1 / HGNC:574 / OMIM:602296 (adaptor related protein complex 4 subunit mu 1)
607243	AP4S1 / HGNC:575 / OMIM:607243 (adaptor related protein complex 4 subunit sigma 1)
613653	AP5Z1 / HGNC:22197 / OMIM:613653 (adaptor related protein complex 5 subunit zeta 1)
606350	APTX / HGNC:15984 / OMIM:606350 (aprataxin)
313700	AR / HGNC:644 / OMIM:313700 (androgen receptor)
608136	ARHGEF10 / HGNC:14103 / OMIM:608136 (Rho guanine nucleotide exchange factor 10)
607669	ARL6IP1 / HGNC:697 / OMIM:607669 (ADP ribosylation factor like GTPase 6 interacting protein 1)
613468	ASAH1 / HGNC:735 / OMIM:613468 (N- acylsphingosine amidohydrolase 1)
614215	ASCC1 / HGNC:24268 / OMIM:614215 (activating signal cointegrator 1 complex subunit 1)
604261	ATG5 / HGNC:589 / OMIM:604261 (autophagy related 5)

606439	ATL1 / HGNC:11231 / OMIM:606439 (atlastin GTPase 1)
609369	ATL3 / HGNC:24526 / OMIM:609369 (atlastin GTPase 3)
607585	ATM / HGNC:795 / OMIM:607585 (ATM serine/threonine kinase)
610513	ATP13A2 / HGNC:30213 / OMIM:610513 (ATPase cation transporting 13A2)
182310	ATP1A1 / HGNC:799 / OMIM:182310 (ATPase Na <sup>+</sup> /K <sup>+</sup> transporting subunit alpha 1)
182340	ATP1A2 / HGNC:800 / OMIM:182340 (ATPase Na <sup>+</sup> /K <sup>+</sup> transporting subunit alpha 2)
108730	ATP2A1 / HGNC:811 / OMIM:108730 (ATPase sarcoplasmic/endoplasmic reticulum Ca <sup>2+</sup> transporting 1)
300011	ATP7A / HGNC:869 / OMIM:300011 (ATPase copper transporting alpha)
601556	ATXN1 / HGNC:10548 / OMIM:601556 (ataxin 1)
611150	ATXN10 / HGNC:10549 / OMIM:611150 (ataxin 10)
601517	ATXN2 / HGNC:10555 / OMIM:601517 (ataxin 2)
607047	ATXN3 / HGNC:7106 / OMIM:607047 (ataxin 3)
607640	ATXN7 / HGNC:10560 / OMIM:607640 (ataxin 7)
603680	ATXN8OS / HGNC:10561 / OMIM:603680 (ATXN8 opposite strand lncRNA)
610194	B3GALNT2 / HGNC:28596 / OMIM:610194 (beta-1,3-N- acetylgalactosaminyltransferase 2)
605517	B4GAT1 / HGNC:15685 / OMIM:605517 (beta-1,4-glucuronyltransferase 1)
601873	B4GALNT1 / HGNC:4117 / OMIM:601873 (beta-1,4-N-acetyl- galactosaminyltransferase 1)
603883	BAG3 / HGNC:939 / OMIM:603883 (BAG cochaperone 3)
612051	BEAN1 / HGNC:24160 / OMIM:612051 (brain expressed associated with NEDD4 1)
609797	BICD2 / HGNC:17208 / OMIM:609797 (BICD cargo adaptor 2)
601248	BIN1 / HGNC:1052 / OMIM:601248 (bridging integrator 1)
606158	BSCL2 / HGNC:15832 / OMIM:606158 (BSCL2 lipid droplet biogenesis associated, seipin)
604577	BVES / HGNC:1152 / OMIM:604577 (blood vessel epicardial substance)
613541	MTRFR / HGNC:26784 / OMIM:613541 (mitochondrial translation release factor in rescue)
614297	C19orf12 / HGNC:25443 / OMIM:614297 (chromosome 19 open reading frame 12)
618682	C1orf194 / HGNC:32331 / OMIM:618682 (chromosome 1 open reading frame

	194)
601269	C1QBP / HGNC:1243 / OMIM:601269 (complement C1q binding protein)
614260	C9orf72 / HGNC:28337 / OMIM:614260 (C9orf72-SMCR8 complex subunit)
601011	CACNA1A / HGNC:1388 / OMIM:601011 (calcium voltage-gated channel subunit alpha1 A)
114205	CACNA1C / HGNC:1390 / OMIM:114205 (calcium voltage-gated channel subunit alpha1 C)
604065	CACNA1G / HGNC:1394 / OMIM:604065 (calcium voltage-gated channel subunit alpha1 G)
607904	CACNA1H / HGNC:1395 / OMIM:607904 (calcium voltage-gated channel subunit alpha1 H)
114208	CACNA1S / HGNC:1397 / OMIM:114208 (calcium voltage-gated channel subunit alpha1 S)
600003	CACNB2 / HGNC:1402 / OMIM:600003 (calcium voltage-gated channel auxiliary subunit beta 2)
601949	CACNB4 / HGNC:1404 / OMIM:601949 (calcium voltage-gated channel auxiliary subunit beta 4)
114180	CALM1 / HGNC:1442 / OMIM:114180 (calmodulin 1)
114182	CALM2 / HGNC:1445 / OMIM:114182 (calmodulin 2)
611414	CALR3 / HGNC:20407 / OMIM:611414 (calreticulin 3)
114220	CAPN1 / HGNC:1476 / OMIM:114220 (calpain 1)
114240	CAPN3 / HGNC:1480 / OMIM:114240 (calpain 3)
114250	CASQ1 / HGNC:1512 / OMIM:114250 (calsequestrin 1)
114251	CASQ2 / HGNC:1513 / OMIM:114251 (calsequestrin 2)
601253	CAV3 / HGNC:1529 / OMIM:601253 (caveolin 3)
603198	CAVIN1 / HGNC:9688 / OMIM:603198 (caveolae associated protein 1)
617714	CAVIN4 / HGNC:33742 / OMIM:617714 (caveolae associated protein 4)
614666	CCDC78 / HGNC:14153 / OMIM:614666 (coiled-coil domain containing 78)
611204	CCDC88C / HGNC:19967 / OMIM:611204 (coiled-coil domain containing 88C)
610150	CCT5 / HGNC:1618 / OMIM:610150 (chaperonin containing TCP1 subunit 5)
601443	CFL2 / HGNC:1875 / OMIM:601443 (cofilin 2)
118490	CHAT / HGNC:1912 / OMIM:118490 (choline O-acetyltransferase)
615903	CHCHD10 / HGNC:15559 / OMIM:615903 (coiled-coil-helix-coiled-coil-helix domain containing 10)
612395	CHKB / HGNC:1938 / OMIM:612395



	(choline kinase beta)
609512	CHMP2B / HGNC:24537 / OMIM:609512 (charged multivesicular body protein 2B)
606988	CHP1 / HGNC:17433 / OMIM:606988 (calcineurin like EF-hand protein 1)
100690	CHRNA1 / HGNC:1955 / OMIM:100690 (cholinergic receptor nicotinic alpha 1 subunit)
100710	CHRN1 / HGNC:1961 / OMIM:100710 (cholinergic receptor nicotinic beta 1 subunit)
100720	CHRNA1 / HGNC:1965 / OMIM:100720 (cholinergic receptor nicotinic delta subunit)
100725	CHRNE / HGNC:1966 / OMIM:100725 (cholinergic receptor nicotinic epsilon subunit)
100730	CHRNA1 / HGNC:1967 / OMIM:100730 (cholinergic receptor nicotinic gamma subunit)
118425	CLCN1 / HGNC:2019 / OMIM:118425 (chloride voltage-gated channel 1)
607042	CLN3 / HGNC:2074 / OMIM:607042 (CLN3 lysosomal/endosomal transmembrane protein, battenin)
601273	CLTCL1 / HGNC:2093 / OMIM:601273 (clathrin heavy chain like 1)
116955	CNBP / HGNC:13164 / OMIM:116955 (CCHC-type zinc finger nucleic acid binding protein)
600016	CNTN1 / HGNC:2171 / OMIM:600016 (contactin 1)
602346	CNTNAP1 / HGNC:8011 / OMIM:602346 (contactin associated protein 1)
615623	COA7 / HGNC:25716 / OMIM:615623 (cytochrome c oxidase assembly factor 7)
120320	COL12A1 / HGNC:2188 / OMIM:120320 (collagen type XII alpha 1 chain)
120350	COL13A1 / HGNC:2190 / OMIM:120350 (collagen type XIII alpha 1 chain)
610004	COL25A1 / HGNC:18603 / OMIM:610004 (collagen type XXV alpha 1 chain)
120215	COL5A1 / HGNC:2209 / OMIM:120215 (collagen type V alpha 1 chain)
120220	COL6A1 / HGNC:2211 / OMIM:120220 (collagen type VI alpha 1 chain)
120240	COL6A2 / HGNC:2212 / OMIM:120240 (collagen type VI alpha 2 chain)
120250	COL6A3 / HGNC:2213 / OMIM:120250 (collagen type VI alpha 3 chain)
603033	COLQ / HGNC:2226 / OMIM:603033 (collagen like tail subunit of asymmetric acetylcholinesterase)
609825	COQ2 / HGNC:25223 / OMIM:609825 (coenzyme Q2, polyprenyltransferase)
612898	COQ4 / HGNC:19693 / OMIM:612898 (coenzyme Q4)
614647	COQ6 / HGNC:20233 / OMIM:614647 (coenzyme Q6, monooxygenase)

601683	COQ7 / HGNC:2244 / OMIM:601683 (coenzyme Q7, hydroxylase)
612837	COQ9 / HGNC:25302 / OMIM:612837 (coenzyme Q9)
603646	COX15 / HGNC:2263 / OMIM:603646 (cytochrome c oxidase assembly homolog COX15)
602072	COX6A1 / HGNC:2277 / OMIM:602072 (cytochrome c oxidase subunit 6A1)
602009	COX6A2 / HGNC:2279 / OMIM:602009 (cytochrome c oxidase subunit 6A2)
608846	CPT1C / HGNC:18540 / OMIM:608846 (carnitine palmitoyltransferase 1C)
600650	CPT2 / HGNC:2330 / OMIM:600650 (carnitine palmitoyltransferase 2)
614631	CRPPA / HGNC:37276 / OMIM:614631 (CDP-L-ribitol pyrophosphorylase A)
123590	CRYAB / HGNC:2389 / OMIM:123590 (crystallin alpha B)
600824	CSRP3 / HGNC:2472 / OMIM:600824 (cysteine and glycine rich protein 3)
604927	CTDP1 / HGNC:2498 / OMIM:604927 (CTD phosphatase subunit 1)
607667	CTNNA3 / HGNC:2511 / OMIM:607667 (catenin alpha 3)
616120	CWF19L1 / HGNC:25613 / OMIM:616120 (CWF19 like cell cycle control factor 1)
610670	CYP2U1 / HGNC:20582 / OMIM:610670 (cytochrome P450 family 2 subfamily U member 1)
603711	CYP7B1 / HGNC:2652 / OMIM:603711 (cytochrome P450 family 7 subfamily B member 1)
128239	DAG1 / HGNC:2666 / OMIM:128239 (dystroglycan 1)
615820	DCAF8 / HGNC:24891 / OMIM:615820 (DDB1 and CUL4 associated factor 8)
601143	DCTN1 / HGNC:2711 / OMIM:601143 (dynactin subunit 1)
614603	DDHD1 / HGNC:19714 / OMIM:614603 (DDHD domain containing 1)
615003	DDHD2 / HGNC:29106 / OMIM:615003 (DDHD domain containing 2)
125660	DES / HGNC:2770 / OMIM:125660 (desmin)
606983	DGAT2 / HGNC:16940 / OMIM:606983 (diacylglycerol O-acyltransferase 2)
601465	DGUOK / HGNC:2858 / OMIM:601465 (deoxyguanosine kinase)
614984	DHTKD1 / HGNC:23537 / OMIM:614984 (dehydrogenase E1 and transketolase domain containing 1)
300377	DMD / HGNC:2928 / OMIM:300377 (dystrophin)
605377	DMPK / HGNC:2933 / OMIM:605377 (DM1 protein kinase)
601810	DNA2 / HGNC:2939 / OMIM:601810 (DNA replication helicase/nuclease 2)
604139	DNAJB2 / HGNC:5228 / OMIM:604139

	(DnaJ heat shock protein family (Hsp40) member B2)
611332	DNAJB6 / HGNC:14888 / OMIM:611332 (DnaJ heat shock protein family (Hsp40) member B6)
602378	DNM2 / HGNC:2974 / OMIM:602378 (dynamin 2)
126375	DNMT1 / HGNC:2976 / OMIM:126375 (DNA methyltransferase 1)
610285	DOK7 / HGNC:26594 / OMIM:610285 (docking protein 7)
610746	DOLK / HGNC:23406 / OMIM:610746 (dolichol kinase)
191350	DPAGT1 / HGNC:2995 / OMIM:191350 (dolichyl-phosphate N-acetylglucosaminophosphotransferase 1)
603503	DPM1 / HGNC:3005 / OMIM:603503 (dolichyl-phosphate mannosyltransferase subunit 1, catalytic)
603564	DPM2 / HGNC:3006 / OMIM:603564 (dolichyl-phosphate mannosyltransferase subunit 2, regulatory)
605951	DPM3 / HGNC:3007 / OMIM:605951 (dolichyl-phosphate mannosyltransferase subunit 3, regulatory)
125645	DSC2 / HGNC:3036 / OMIM:125645 (desmocollin 2)
125671	DSG2 / HGNC:3049 / OMIM:125671 (desmoglein 2)
125647	DSP / HGNC:3052 / OMIM:125647 (desmoplakin)
113810	DST / HGNC:1090 / OMIM:113810 (dystonin)
601239	DTNA / HGNC:3057 / OMIM:601239 (dystrobrevin alpha)
606009	DUX4L1 / HGNC:3082 / OMIM:606009 (double homeobox 4 like 1 (pseudogene))
600112	DYNC1H1 / HGNC:2961 / OMIM:600112 (dynein cytoplasmic 1 heavy chain 1)
603009	DYSF / HGNC:3097 / OMIM:603009 (dysferlin)
130610	EEF2 / HGNC:3214 / OMIM:130610 (eukaryotic translation elongation factor 2)
129010	EGR2 / HGNC:3239 / OMIM:129010 (early growth response 2)
605512	ELOVL4 / HGNC:14415 / OMIM:605512 (ELOVL fatty acid elongase 4)
611805	ELOVL5 / HGNC:21308 / OMIM:611805 (ELOVL fatty acid elongase 5)
603722	ELP1 / HGNC:5959 / OMIM:603722 (elongator acetyltransferase complex subunit 1)
300384	EMD / HGNC:3331 / OMIM:300384 (emerin)
131370	ENO3 / HGNC:3354 / OMIM:131370

	(enolase 3)
601752	ENTPD1 / HGNC:3363 / OMIM:601752 (ectonucleoside triphosphate diphosphohydrolase 1)
190151	ERBB3 / HGNC:3431 / OMIM:190151 (erb-b2 receptor tyrosine kinase 3)
600543	ERBB4 / HGNC:3432 / OMIM:600543 (erb-b2 receptor tyrosine kinase 4)
611604	ERLIN1 / HGNC:16947 / OMIM:611604 (ER lipid raft associated 1)
611605	ERLIN2 / HGNC:1356 / OMIM:611605 (ER lipid raft associated 2)
608053	ETFA / HGNC:3481 / OMIM:608053 (electron transfer flavoprotein subunit alpha)
231675	ETFDH / HGNC:3483 / OMIM:231675 (electron transfer flavoprotein dehydrogenase)
606489	EXOSC3 / HGNC:17944 / OMIM:606489 (exosome component 3)
606019	EXOSC8 / HGNC:17035 / OMIM:606019 (exosome component 8)
603550	EYA4 / HGNC:3522 / OMIM:603550 (EYA transcriptional coactivator and phosphatase 4)
611026	FA2H / HGNC:21197 / OMIM:611026 (fatty acid 2-hydroxylase)
615584	FAM111B / HGNC:24200 / OMIM:615584 (FAM111 trypsin like peptidase B)
611592	FARS2 / HGNC:21062 / OMIM:611592 (phenylalanyl-tRNA synthetase 2, mitochondrial)
612322	FASTKD2 / HGNC:29160 / OMIM:612322 (FAST kinase domains 2)
604269	FAT2 / HGNC:3596 / OMIM:604269 (FAT atypical cadherin 2)
604580	FBLN5 / HGNC:3602 / OMIM:604580 (fibulin 5)
605654	FBXL4 / HGNC:13601 / OMIM:605654 (F-box and leucine rich repeat protein 4)
608533	FBXO38 / HGNC:28844 / OMIM:608533 (F-box protein 38)
614585	FDX2 / HGNC:30546 / OMIM:614585 (ferredoxin 2)
611104	FGD4 / HGNC:19125 / OMIM:611104 (FYVE, RhoGEF and PH domain containing 4)
601515	FGF14 / HGNC:3671 / OMIM:601515 (fibroblast growth factor 14)
300163	FHL1 / HGNC:3702 / OMIM:300163 (four and a half LIM domains 1)
609390	FIG4 / HGNC:16873 / OMIM:609390 (FIG4 phosphoinositide 5-phosphatase)
606596	FKRP / HGNC:17997 / OMIM:606596 (fukutin related protein)
607440	FKTN / HGNC:3622 / OMIM:607440 (fukutin)
610595	FLAD1 / HGNC:24671 / OMIM:610595 (flavin adenine dinucleotide synthetase 1)

300017	FLNA / HGNC:3754 / OMIM:300017 (filamin A)
102565	FLNC / HGNC:3756 / OMIM:102565 (filamin C)
609144	FLVCR1 / HGNC:24682 / OMIM:609144 (FLVCR heme transporter 1)
601278	FRG1 / HGNC:3954 / OMIM:601278 (FSD region gene 1)
137070	FUS / HGNC:4010 / OMIM:137070 (FUS RNA binding protein)
606829	FXN / HGNC:3951 / OMIM:606829 (frataxin)
600819	FXR1 / HGNC:4023 / OMIM:600819 (FMR1 autosomal homolog 1)
606800	GAA / HGNC:4065 / OMIM:606800 (alpha glucosidase)
605379	GAN / HGNC:4137 / OMIM:605379 (gigaxonin)
600287	GARS1 / HGNC:4162 / OMIM:600287 (glycyl-tRNA synthetase 1)
614518	GATAD1 / HGNC:29941 / OMIM:614518 (GATA zinc finger domain containing 1)
606463	GBA / HGNC:4177 / OMIM:606463 (glucosylceramidase beta)
609471	GBA2 / HGNC:18986 / OMIM:609471 (glucosylceramidase beta 2)
607839	GBE1 / HGNC:4180 / OMIM:607839 (1,4- alpha-glucan branching enzyme 1)
603698	GBF1 / HGNC:4181 / OMIM:603698 (golgi brefeldin A resistant guanine nucleotide exchange factor 1)
606598	GDAP1 / HGNC:15968 / OMIM:606598 (ganglioside induced differentiation associated protein 1)
618128	GDAP2 / HGNC:18010 / OMIM:618128 (ganglioside induced differentiation associated protein 2)
138292	GFPT1 / HGNC:4241 / OMIM:138292 (glutamine--fructose-6-phosphate transaminase 1)
605072	GIPC1 / HGNC:1226 / OMIM:605072 (GIPC PDZ domain containing family member 1)
121013	GJA5 / HGNC:4279 / OMIM:121013 (gap junction protein alpha 5)
304040	GJB1 / HGNC:4283 / OMIM:304040 (gap junction protein beta 1)
603324	GJB3 / HGNC:4285 / OMIM:603324 (gap junction protein beta 3)
608803	GJC2 / HGNC:17494 / OMIM:608803 (gap junction protein gamma 2)
608603	GLDN / HGNC:29514 / OMIM:608603 (gliomedin)
603371	GLE1 / HGNC:4315 / OMIM:603371 (GLE1 RNA export mediator)
615320	GMPPB / HGNC:22932 / OMIM:615320 (GDP-mannose pyrophosphorylase B)
610863	GNB4 / HGNC:20731 / OMIM:610863 (G protein subunit beta 4)
603824	GNE / HGNC:23657 / OMIM:603824

	(glucosamine (UDP-N-acetyl)-2-epimerase/N-acetylmannosamine kinase)
602580	GOLGA2 / HGNC:4425 / OMIM:602580 (golgin A2)
604027	GOSR2 / HGNC:4431 / OMIM:604027 (golgi SNAP receptor complex member 2)
611778	GPD1L / HGNC:28956 / OMIM:611778 (glycerol-3-phosphate dehydrogenase 1 like)
602368	GRID2 / HGNC:4576 / OMIM:602368 (glutamate ionotropic receptor delta type subunit 2)
604473	GRM1 / HGNC:4593 / OMIM:604473 (glutamate metabotropic receptor 1)
603942	GYG1 / HGNC:4699 / OMIM:603942 (glycogenin 1)
138570	GYS1 / HGNC:4706 / OMIM:138570 (glycogen synthase 1)
610467	HACD1 / HGNC:9639 / OMIM:610467 (3-hydroxyacyl-CoA dehydratase 1)
610876	HACE1 / HGNC:21033 / OMIM:610876 (HECT domain and ankyrin repeat containing E3 ubiquitin protein ligase 1)
142810	HARS1 / HGNC:4816 / OMIM:142810 (histidyl-tRNA synthetase 1)
605206	HCN4 / HGNC:16882 / OMIM:605206 (hyperpolarization activated cyclic nucleotide gated potassium channel 4)
606873	HEXB / HGNC:4879 / OMIM:606873 (hexosaminidase subunit beta)
601314	HINT1 / HGNC:4912 / OMIM:601314 (histidine triad nucleotide binding protein 1)
142600	HK1 / HGNC:4922 / OMIM:142600 (hexokinase 1)
164017	HNRNPA1 / HGNC:5031 / OMIM:164017 (heterogeneous nuclear ribonucleoprotein A1)
600124	HNRNPA2B1 / HGNC:5033 / OMIM:600124 (heterogeneous nuclear ribonucleoprotein A2/B1)
607137	HNRNPDL / HGNC:5037 / OMIM:607137 (heterogeneous nuclear ribonucleoprotein D like)
142984	HOXD10 / HGNC:5133 / OMIM:142984 (homeobox D10)
190020	HRAS / HGNC:5173 / OMIM:190020 (HRas proto-oncogene, GTPase)
602195	HSPB1 / HGNC:5246 / OMIM:602195 (heat shock protein family B (small) member 1)
604624	HSPB3 / HGNC:5248 / OMIM:604624 (heat shock protein family B (small) member 3)
608014	HSPB8 / HGNC:30171 / OMIM:608014 (heat shock protein family B (small) member 8)
118190	HSPD1 / HGNC:5261 / OMIM:118190 (heat shock protein family D (Hsp60) member 1)

142461	HSPG2 / HGNC:5273 / OMIM:142461 (heparan sulfate proteoglycan 2)
615316	IBA57 / HGNC:27302 / OMIM:615316 (iron-sulfur cluster assembly factor IBA57)
603502	IFRD1 / HGNC:5456 / OMIM:603502 (interferon related developmental regulator 1)
600502	IGHMBP2 / HGNC:5542 / OMIM:600502 (immunoglobulin mu DNA binding protein 2)
602366	ILK / HGNC:6040 / OMIM:602366 (integrin linked kinase)
610982	INF2 / HGNC:23791 / OMIM:610982 (inverted formin 2)
607875	INPP5K / HGNC:33882 / OMIM:607875 (inositol polyphosphate-5-phosphatase K)
611911	ISCU / HGNC:29882 / OMIM:611911 (iron-sulfur cluster assembly enzyme)
600536	ITGA7 / HGNC:6143 / OMIM:600536 (integrin subunit alpha 7)
147265	ITPR1 / HGNC:6180 / OMIM:147265 (inositol 1,4,5-trisphosphate receptor type 1)
147267	ITPR3 / HGNC:6182 / OMIM:147267 (inositol 1,4,5-trisphosphate receptor type 3)
601920	JAG1 / HGNC:6188 / OMIM:601920 (jagged canonical Notch ligand 1)
605267	JPH2 / HGNC:14202 / OMIM:605267 (junctophilin 2)
173325	JUP / HGNC:6207 / OMIM:173325 (junction plakoglobin)
601421	KARS1 / HGNC:6215 / OMIM:601421 (lysyl-tRNA synthetase 1)
613727	KBTBD13 / HGNC:37227 / OMIM:613727 (kelch repeat and BTB domain containing 13)
176260	KCNA1 / HGNC:6218 / OMIM:176260 (potassium voltage-gated channel subfamily A member 1)
176267	KCNA5 / HGNC:6224 / OMIM:176267 (potassium voltage-gated channel subfamily A member 5)
176264	KCNC3 / HGNC:6235 / OMIM:176264 (potassium voltage-gated channel subfamily C member 3)
605411	KCND3 / HGNC:6239 / OMIM:605411 (potassium voltage-gated channel subfamily D member 3)
176261	KCNE1 / HGNC:6240 / OMIM:176261 (potassium voltage-gated channel subfamily E regulatory subunit 1)
603796	KCNE2 / HGNC:6242 / OMIM:603796 (potassium voltage-gated channel subfamily E regulatory subunit 2)
604433	KCNE3 / HGNC:6243 / OMIM:604433 (potassium voltage-gated channel subfamily E regulatory subunit 3)
152427	KCNH2 / HGNC:6251 / OMIM:152427

	(potassium voltage-gated channel subfamily H member 2)
602323	KCNJ12 / HGNC:6258 / OMIM:602323 (potassium inwardly rectifying channel subfamily J member 12)
600681	KCNJ2 / HGNC:6263 / OMIM:600681 (potassium inwardly rectifying channel subfamily J member 2)
600734	KCNJ5 / HGNC:6266 / OMIM:600734 (potassium inwardly rectifying channel subfamily J member 5)
607542	KCNQ1 / HGNC:6294 / OMIM:607542 (potassium voltage-gated channel subfamily Q member 1)
610657	WASHC5 / HGNC:28984 / OMIM:610657 (WASH complex subunit 5)
615759	KIDINS220 / HGNC:29508 / OMIM:615759 (kinase D interacting substrate 220)
601255	KIF1A / HGNC:888 / OMIM:601255 (kinesin family member 1A)
605995	KIF1B / HGNC:16636 / OMIM:605995 (kinesin family member 1B)
603060	KIF1C / HGNC:6317 / OMIM:603060 (kinesin family member 1C)
608283	KIF21A / HGNC:19349 / OMIM:608283 (kinesin family member 21A)
614026	KIF26B / HGNC:25484 / OMIM:614026 (kinesin family member 26B)
602821	KIF5A / HGNC:6323 / OMIM:602821 (kinesin family member 5A)
611729	KLC2 / HGNC:20716 / OMIM:611729 (kinesin light chain 2)
615340	KLHL40 / HGNC:30372 / OMIM:615340 (kelch like family member 40)
607701	KLHL41 / HGNC:16905 / OMIM:607701 (kelch like family member 41)
611201	KLHL9 / HGNC:18732 / OMIM:611201 (kelch like family member 9)
605739	KY / HGNC:26576 / OMIM:605739 (kyphoscoliosis peptidase)
308840	L1CAM / HGNC:6470 / OMIM:308840 (L1 cell adhesion molecule)
156225	LAMA2 / HGNC:6482 / OMIM:156225 (laminin subunit alpha 2)
600133	LAMA4 / HGNC:6484 / OMIM:600133 (laminin subunit alpha 4)
601033	LAMA5 / HGNC:6485 / OMIM:601033 (laminin subunit alpha 5)
150325	LAMB2 / HGNC:6487 / OMIM:150325 (laminin subunit beta 2)
309060	LAMP2 / HGNC:6501 / OMIM:309060 (lysosomal associated membrane protein 2)
603590	LARGE1 / HGNC:6511 / OMIM:603590 (LARGE xylosyl- and glucuronyltransferase 1)
605906	LDB3 / HGNC:15710 / OMIM:605906 (LIM domain binding 3)
150000	LDHA / HGNC:6535 / OMIM:150000



	(lactate dehydrogenase A)
607908	LIMS2 / HGNC:16084 / OMIM:607908 (LIM zinc finger domain containing 2)
603795	LITAF / HGNC:16841 / OMIM:603795 (lipopolysaccharide induced TNF factor)
150330	LMNA / HGNC:6636 / OMIM:150330 (lamin A/C)
616112	LMOD3 / HGNC:6649 / OMIM:616112 (leiomodlin 3)
605518	LPIN1 / HGNC:13345 / OMIM:605518 (lipin 1)
618299	LRP12 / HGNC:31708 / OMIM:618299 (LDL receptor related protein 12)
604270	LRP4 / HGNC:6696 / OMIM:604270 (LDL receptor related protein 4)
610933	LRSAM1 / HGNC:25135 / OMIM:610933 (leucine rich repeat and sterile alpha motif containing 1)
159460	MAG / HGNC:6783 / OMIM:159460 (myelin associated glycoprotein)
609479	MAP3K20 / HGNC:17797 / OMIM:609479 (mitogen-activated protein kinase kinase kinase 20)
157140	MAPT / HGNC:6893 / OMIM:157140 (microtubule associated protein tau)
156560	MARS1 / HGNC:6898 / OMIM:156560 (methionyl-tRNA synthetase 1)
609728	MARS2 / HGNC:25133 / OMIM:609728 (methionyl-tRNA synthetase 2, mitochondrial)
164015	MATR3 / HGNC:6912 / OMIM:164015 (matrin 3)
160000	MB / HGNC:6915 / OMIM:160000 (myoglobin)
603294	MCM3AP / HGNC:6946 / OMIM:603294 (minichromosome maintenance complex component 3 associated protein)
612453	MEGF10 / HGNC:29634 / OMIM:612453 (multiple EGF like domains 10)
608507	MFN2 / HGNC:16877 / OMIM:608507 (mitofusin 2)
615076	MGME1 / HGNC:16205 / OMIM:615076 (mitochondrial genome maintenance exonuclease 1)
608677	MIB1 / HGNC:21086 / OMIM:608677 (MIB E3 ubiquitin protein ligase 1)
120520	MME / HGNC:7154 / OMIM:120520 (membrane metalloendopeptidase)
616661	MORC2 / HGNC:23573 / OMIM:616661 (MORC family CW-type zinc finger 2)
604041	MPDU1 / HGNC:7207 / OMIM:604041 (mannose-P-dolichol utilization defect 1)
137960	MPV17 / HGNC:7224 / OMIM:137960 (mitochondrial inner membrane protein MPV17)
159440	MPZ / HGNC:7225 / OMIM:159440 (myelin protein zero)
600814	MRE11 / HGNC:7230 / OMIM:600814 (MRE11 homolog, double strand break

	repair nuclease)
607118	MRPL3 / HGNC:10379 / OMIM:607118 (mitochondrial ribosomal protein L3)
611849	MRPL44 / HGNC:16650 / OMIM:611849 (mitochondrial ribosomal protein L44)
611987	MRPS25 / HGNC:14511 / OMIM:611987 (mitochondrial ribosomal protein S25)
601788	MSTN / HGNC:4223 / OMIM:601788 (myostatin)
617619	MSTO1 / HGNC:29678 / OMIM:617619 (misato mitochondrial distribution and morphology regulator 1)
590065	MT-TM / HGNC:7492 / OMIM:590065 (mitochondrially encoded tRNA-Met (AUA/G))
590090	MT-TT / HGNC:7499 / OMIM:590090 (mitochondrially encoded tRNA-Thr (ACN))
300415	MTM1 / HGNC:7448 / OMIM:300415 (myotubularin 1)
603557	MTMR2 / HGNC:7450 / OMIM:603557 (myotubularin related protein 2)
614667	MTO1 / HGNC:19261 / OMIM:614667 (mitochondrial tRNA translation optimization 1)
613669	MTPAP / HGNC:25532 / OMIM:613669 (mitochondrial poly(A) polymerase)
601296	MUSK / HGNC:7525 / OMIM:601296 (muscle associated receptor tyrosine kinase)
160794	MYBPC1 / HGNC:7549 / OMIM:160794 (myosin binding protein C1)
600958	MYBPC3 / HGNC:7551 / OMIM:600958 (myosin binding protein C3)
608568	MYH14 / HGNC:23212 / OMIM:608568 (myosin heavy chain 14)
160740	MYH2 / HGNC:7572 / OMIM:160740 (myosin heavy chain 2)
160720	MYH3 / HGNC:7573 / OMIM:160720 (myosin heavy chain 3)
160710	MYH6 / HGNC:7576 / OMIM:160710 (myosin heavy chain 6)
160760	MYH7 / HGNC:7577 / OMIM:160760 (myosin heavy chain 7)
160741	MYH8 / HGNC:7578 / OMIM:160741 (myosin heavy chain 8)
160780	MYL1 / HGNC:7582 / OMIM:160780 (myosin light chain 1)
160781	MYL2 / HGNC:7583 / OMIM:160781 (myosin light chain 2)
160790	MYL3 / HGNC:7584 / OMIM:160790 (myosin light chain 3)
160770	MYL4 / HGNC:7585 / OMIM:160770 (myosin light chain 4)
606566	MYLK2 / HGNC:16243 / OMIM:606566 (myosin light chain kinase 2)
615345	MYMK / HGNC:33778 / OMIM:615345 (myomaker, myoblast fusion factor)
607295	MYO18B / HGNC:18150 / OMIM:607295 (myosin XVIIIIB)

604875	MYO9A / HGNC:7608 / OMIM:604875 (myosin IXA)
604103	MYOT / HGNC:12399 / OMIM:604103 (myotilin)
605602	MYOZ2 / HGNC:1330 / OMIM:605602 (myozenin 2)
608517	MYPN / HGNC:23246 / OMIM:608517 (myopalladin)
609701	NAGLU / HGNC:7632 / OMIM:609701 (N- acetyl-alpha-glucosaminidase)
612803	NARS2 / HGNC:26274 / OMIM:612803 (asparaginyl-tRNA synthetase 2, mitochondrial)
605262	NDRG1 / HGNC:7679 / OMIM:605262 (N-myc downstream regulated 1)
606934	NDUFAF1 / HGNC:18828 / OMIM:606934 (NADH:ubiquinone oxidoreductase complex assembly factor 1)
161650	NEB / HGNC:7720 / OMIM:161650 (nebulin)
162230	NEFH / HGNC:7737 / OMIM:162230 (neurofilament heavy chain)
162280	NEFL / HGNC:7739 / OMIM:162280 (neurofilament light chain)
604588	NEK1 / HGNC:7744 / OMIM:604588 (NIMA related kinase 1)
609798	NEK9 / HGNC:18591 / OMIM:609798 (NIMA related kinase 9)
613121	NEXN / HGNC:29557 / OMIM:613121 (nexilin F-actin binding protein)
162030	NGF / HGNC:7808 / OMIM:162030 (nerve growth factor)
608145	NIPA1 / HGNC:17043 / OMIM:608145 (NIPA magnesium transporter 1)
605955	NKX6-2 / HGNC:19321 / OMIM:605955 (NK6 homeobox 2)
608701	NMNAT2 / HGNC:16789 / OMIM:608701 (nicotinamide nucleotide adenylyltransferase 2)
614154	NOP56 / HGNC:15911 / OMIM:614154 (NOP56 ribonucleoprotein)
108780	NPPA / HGNC:7939 / OMIM:108780 (natriuretic peptide A)
600417	NT5C2 / HGNC:8022 / OMIM:600417 (5'- nucleotidase, cytosolic II)
191315	NTRK1 / HGNC:8031 / OMIM:191315 (neurotrophic receptor tyrosine kinase 1)
606694	NUP155 / HGNC:8063 / OMIM:606694 (nucleoporin 155)
602552	NUP88 / HGNC:8067 / OMIM:602552 (nucleoporin 88)
605290	OPA1 / HGNC:8140 / OMIM:605290 (OPA1 mitochondrial dynamin like GTPase)
602432	OPTN / HGNC:17142 / OMIM:602432 (optineurin)
610277	ORAI1 / HGNC:25896 / OMIM:610277 (ORAI calcium release-activated calcium

	modulator 1)
602279	PABPN1 / HGNC:8565 / OMIM:602279 (poly(A) binding protein nuclear 1)
167410	PAX7 / HGNC:8621 / OMIM:167410 (paired box 7)
176740	PCNA / HGNC:8729 / OMIM:176740 (proliferating cell nuclear antigen)
300906	PDK3 / HGNC:8811 / OMIM:300906 (pyruvate dehydrogenase kinase 3)
131340	PDYN / HGNC:8820 / OMIM:131340 (prodynorphin)
601757	PEX7 / HGNC:8860 / OMIM:601757 (peroxisomal biogenesis factor 7)
610681	PFKM / HGNC:8877 / OMIM:610681 (phosphofructokinase, muscle)
176610	PFN1 / HGNC:8881 / OMIM:176610 (profilin 1)
612931	PGAM2 / HGNC:8889 / OMIM:612931 (phosphoglycerate mutase 2)
311800	PGK1 / HGNC:8896 / OMIM:311800 (phosphoglycerate kinase 1)
171900	PGM1 / HGNC:8905 / OMIM:171900 (phosphoglucomutase 1)
311870	PHKA1 / HGNC:8925 / OMIM:311870 (phosphorylase kinase regulatory subunit alpha 1)
602753	PHOX2A / HGNC:691 / OMIM:602753 (paired like homeobox 2A)
602026	PHYH / HGNC:8940 / OMIM:602026 (phytanoyl-CoA 2-hydroxylase)
613629	PIEZO2 / HGNC:26270 / OMIM:613629 (piezo type mechanosensitive ion channel component 2)
606102	PIP5K1C / HGNC:8996 / OMIM:606102 (phosphatidylinositol-4-phosphate 5-kinase type 1 gamma)
602861	PKP2 / HGNC:9024 / OMIM:602861 (plakophilin 2)
615698	PLD3 / HGNC:17158 / OMIM:615698 (phospholipase D family member 3)
601282	PLEC / HGNC:9069 / OMIM:601282 (plectin)
611101	PLEKHG5 / HGNC:29105 / OMIM:611101 (pleckstrin homology and RhoGEF domain containing G5)
172405	PLN / HGNC:9080 / OMIM:172405 (phospholamban)
300401	PLP1 / HGNC:9086 / OMIM:300401 (proteolipid protein 1)
170715	PMP2 / HGNC:9117 / OMIM:170715 (peripheral myelin protein 2)
601097	PMP22 / HGNC:9118 / OMIM:601097 (peripheral myelin protein 22)
605610	PNKP / HGNC:9154 / OMIM:605610 (polynucleotide kinase 3'-phosphatase)
609059	PNPLA2 / HGNC:30802 / OMIM:609059 (patatin like phospholipase domain containing 2)
603197	PNPLA6 / HGNC:16268 / OMIM:603197 (patatin like phospholipase domain

	containing 6)
612123	PNPLA8 / HGNC:28900 / OMIM:612123 (patatin like phospholipase domain containing 8)
615618	POGLUT1 / HGNC:22954 / OMIM:615618 (protein O-glucosyltransferase 1)
174763	POLG / HGNC:9179 / OMIM:174763 (DNA polymerase gamma, catalytic subunit)
604983	POLG2 / HGNC:9180 / OMIM:604983 (DNA polymerase gamma 2, accessory subunit)
606822	POMGNT1 / HGNC:19139 / OMIM:606822 (protein O-linked mannose N-acetylglucosaminyltransferase 1 (beta 1,2-))
614828	POMGNT2 / HGNC:25902 / OMIM:614828 (protein O-linked mannose N-acetylglucosaminyltransferase 2 (beta 1,4-))
615247	POMK / HGNC:26267 / OMIM:615247 (protein O-mannose kinase)
607423	POMT1 / HGNC:9202 / OMIM:607423 (protein O-mannosyltransferase 1)
607439	POMT2 / HGNC:19743 / OMIM:607439 (protein O-mannosyltransferase 2)
605824	POPDC3 / HGNC:17649 / OMIM:605824 (popeye domain containing 3)
604325	PPP2R2B / HGNC:9305 / OMIM:604325 (protein phosphatase 2 regulatory subunit Bbeta)
616458	PRDM12 / HGNC:13997 / OMIM:616458 (PR/SET domain 12)
605557	PRDM16 / HGNC:14000 / OMIM:605557 (PR/SET domain 16)
609557	PREPL / HGNC:30228 / OMIM:609557 (prolyl endopeptidase like)
602743	PRKAG2 / HGNC:9386 / OMIM:602743 (protein kinase AMP-activated non-catalytic subunit gamma 2)
176980	PRKCG / HGNC:9402 / OMIM:176980 (protein kinase C gamma)
170710	PRPH / HGNC:9461 / OMIM:170710 (peripherin)
311850	PRPS1 / HGNC:9462 / OMIM:311850 (phosphoribosyl pyrophosphate synthetase 1)
617413	PRUNE1 / HGNC:13420 / OMIM:617413 (prune exopolyphosphatase 1)
605725	PRX / HGNC:13797 / OMIM:605725 (periaxin)
104311	PSEN1 / HGNC:9508 / OMIM:104311 (presenilin 1)
600759	PSEN2 / HGNC:9509 / OMIM:600759 (presenilin 2)
608625	PTRH2 / HGNC:24265 / OMIM:608625 (peptidyl-tRNA hydrolase 2)
607204	PUM1 / HGNC:14957 / OMIM:607204

	(pumilio RNA binding family member 1)
600473	PURA / HGNC:9701 / OMIM:600473 (purine rich element binding protein A)
608109	PUS1 / HGNC:15508 / OMIM:608109 (pseudouridine synthase 1)
608455	PYGM / HGNC:9726 / OMIM:608455 (glycogen phosphorylase, muscle associated)
617220	PYROXD1 / HGNC:26162 / OMIM:617220 (pyridine nucleotide-disulphide oxidoreductase domain 1)
602298	RAB7A / HGNC:9788 / OMIM:602298 (RAB7A, member RAS oncogene family)
164760	RAF1 / HGNC:9829 / OMIM:164760 (Raf-1 proto-oncogene, serine/threonine kinase)
601592	RAPSN / HGNC:9863 / OMIM:601592 (receptor associated protein of the synapse)
610924	RBCK1 / HGNC:15864 / OMIM:610924 (RANBP2-type and C3HC4-type zinc finger containing 1)
613171	RBM20 / HGNC:27424 / OMIM:613171 (RNA binding motif protein 20)
612413	RBM7 / HGNC:9904 / OMIM:612413 (RNA binding motif protein 7)
609139	REEP1 / HGNC:25786 / OMIM:609139 (receptor accessory protein 1)
609347	REEP2 / HGNC:17975 / OMIM:609347 (receptor accessory protein 2)
613114	RETREG1 / HGNC:25964 / OMIM:613114 (reticulophagy regulator 1)
102579	RFC1 / HGNC:9969 / OMIM:102579 (replication factor C subunit 1)
604123	RNASEH1 / HGNC:18466 / OMIM:604123 (ribonuclease H1)
609948	RNF216 / HGNC:21698 / OMIM:609948 (ring finger protein 216)
612159	RPH3A / HGNC:17056 / OMIM:612159 (rabphilin 3A)
604712	RRM2B / HGNC:17296 / OMIM:604712 (ribonucleotide reductase regulatory TP53 inducible subunit M2B)
603183	RTN2 / HGNC:10468 / OMIM:603183 (reticulon 2)
613516	RUBCN / HGNC:28991 / OMIM:613516 (rubicon autophagy regulator)
605862	RXYLT1 / HGNC:13530 / OMIM:605862 (ribitol xylosyltransferase 1)
180901	RYR1 / HGNC:10483 / OMIM:180901 (ryanodine receptor 1)
180902	RYR2 / HGNC:10484 / OMIM:180902 (ryanodine receptor 2)
180903	RYR3 / HGNC:10485 / OMIM:180903 (ryanodine receptor 3)
604490	SACS / HGNC:10519 / OMIM:604490 (sacsin molecular chaperone)
603560	SBF1 / HGNC:10542 / OMIM:603560 (SET binding factor 1)

607697	SBF2 / HGNC:2135 / OMIM:607697 (SET binding factor 2)
604385	SCN11A / HGNC:10583 / OMIM:604385 (sodium voltage-gated channel alpha subunit 11)
600235	SCN1B / HGNC:10586 / OMIM:600235 (sodium voltage-gated channel beta subunit 1)
601327	SCN2B / HGNC:10589 / OMIM:601327 (sodium voltage-gated channel beta subunit 2)
608214	SCN3B / HGNC:20665 / OMIM:608214 (sodium voltage-gated channel beta subunit 3)
603967	SCN4A / HGNC:10591 / OMIM:603967 (sodium voltage-gated channel alpha subunit 4)
608256	SCN4B / HGNC:10592 / OMIM:608256 (sodium voltage-gated channel beta subunit 4)
600163	SCN5A / HGNC:10593 / OMIM:600163 (sodium voltage-gated channel alpha subunit 5)
603415	SCN9A / HGNC:10597 / OMIM:603415 (sodium voltage-gated channel alpha subunit 9)
604272	SCO2 / HGNC:10604 / OMIM:604272 (synthesis of cytochrome C oxidase 2)
607982	SCYL1 / HGNC:14372 / OMIM:607982 (SCY1 like pseudokinase 1)
600857	SDHA / HGNC:10680 / OMIM:600857 (succinate dehydrogenase complex flavoprotein subunit A)
606210	SELENON / HGNC:15999 / OMIM:606210 (selenoprotein N)
604061	SEPTIN9 / HGNC:7323 / OMIM:604061 (septin 9)
608465	SETX / HGNC:445 / OMIM:608465 (senataxin)
600119	SGCA / HGNC:10805 / OMIM:600119 (sarcoglycan alpha)
600900	SGCB / HGNC:10806 / OMIM:600900 (sarcoglycan beta)
601411	SGCD / HGNC:10807 / OMIM:601411 (sarcoglycan delta)
608896	SGCG / HGNC:10809 / OMIM:608896 (sarcoglycan gamma)
603729	SGPL1 / HGNC:10817 / OMIM:603729 (sphingosine-1-phosphate lyase 1)
608206	SH3TC2 / HGNC:29427 / OMIM:608206 (SH3 domain and tetratricopeptide repeats 2)
601978	SIGMAR1 / HGNC:8157 / OMIM:601978 (sigma non-opioid intracellular receptor 1)
608005	SIL1 / HGNC:24624 / OMIM:608005 (SIL1 nucleotide exchange factor)
604878	SLC12A6 / HGNC:10914 / OMIM:604878 (solute carrier family 12 member 6)
600682	SLC16A1 / HGNC:10922 / OMIM:600682 (solute carrier family 16 member 1)

600336	SLC18A3 / HGNC:10936 / OMIM:600336 (solute carrier family 18 member A3)
600111	SLC1A3 / HGNC:10941 / OMIM:600111 (solute carrier family 1 member 3)
603377	SLC22A5 / HGNC:10969 / OMIM:603377 (solute carrier family 22 member 5)
190315	SLC25A1 / HGNC:10979 / OMIM:190315 (solute carrier family 25 member 1)
606521	SLC25A19 / HGNC:14409 / OMIM:606521 (solute carrier family 25 member 19)
613698	SLC25A20 / HGNC:1421 / OMIM:613698 (solute carrier family 25 member 20)
103220	SLC25A4 / HGNC:10990 / OMIM:103220 (solute carrier family 25 member 4)
610823	SLC25A42 / HGNC:28380 / OMIM:610823 (solute carrier family 25 member 42)
610826	SLC25A46 / HGNC:25198 / OMIM:610826 (solute carrier family 25 member 46)
603690	SLC33A1 / HGNC:95 / OMIM:603690 (solute carrier family 33 member 1)
607882	SLC52A2 / HGNC:30224 / OMIM:607882 (solute carrier family 52 member 2)
613350	SLC52A3 / HGNC:16187 / OMIM:613350 (solute carrier family 52 member 3)
608761	SLC5A7 / HGNC:14025 / OMIM:608761 (solute carrier family 5 member 7)
107310	SLC9A1 / HGNC:11071 / OMIM:107310 (solute carrier family 9 member A1)
604990	SLC9A3R1 / HGNC:11075 / OMIM:604990 (SLC9A3 regulator 1)
614982	SMCHD1 / HGNC:29090 / OMIM:614982 (structural maintenance of chromosomes flexible hinge domain containing 1)
600354	SMN1 / HGNC:11117 / OMIM:600354 (survival of motor neuron 1, telomeric)
601627	SMN2 / HGNC:11118 / OMIM:601627 (survival of motor neuron 2, centromeric)
610457	SMPD4 / HGNC:32949 / OMIM:610457 (sphingomyelin phosphodiesterase 4)
600322	SNAP25 / HGNC:11132 / OMIM:600322 (synaptosome associated protein 25)
601017	SNTA1 / HGNC:11167 / OMIM:601017 (syntrophin alpha 1)
616105	SNX14 / HGNC:14977 / OMIM:616105 (sorting nexin 14)
147450	SOD1 / HGNC:11179 / OMIM:147450 (superoxide dismutase 1)
182500	SORD / HGNC:11184 / OMIM:182500 (sorbitol dehydrogenase)
607111	SPART / HGNC:18514 / OMIM:607111 (spartin)
604277	SPAST / HGNC:11233 / OMIM:604277 (spastin)
615950	SPEG / HGNC:16901 / OMIM:615950 (striated muscle enriched protein)



	kinase)
610844	SPG11 / HGNC:11226 / OMIM:610844 (SPG11 vesicle trafficking associated, spatacsin)
608181	SPG21 / HGNC:20373 / OMIM:608181 (SPG21 abhydrolase domain containing, maspardin)
602783	SPG7 / HGNC:11237 / OMIM:602783 (SPG7 matrix AAA peptidase subunit, paraplegin)
182810	SPTAN1 / HGNC:11273 / OMIM:182810 (spectrin alpha, non-erythrocytic 1)
604985	SPTBN2 / HGNC:11276 / OMIM:604985 (spectrin beta, non-erythrocytic 2)
606214	SPTBN4 / HGNC:14896 / OMIM:606214 (spectrin beta, non-erythrocytic 4)
605712	SPTLC1 / HGNC:11277 / OMIM:605712 (serine palmitoyltransferase long chain base subunit 1)
605713	SPTLC2 / HGNC:11278 / OMIM:605713 (serine palmitoyltransferase long chain base subunit 2)
601530	SQSTM1 / HGNC:11280 / OMIM:601530 (sequestosome 1)
615521	STAC3 / HGNC:28423 / OMIM:615521 (SH3 and cysteine rich domain 3)
605921	STIM1 / HGNC:11386 / OMIM:605921 (stromal interaction molecule 1)
607207	STUB1 / HGNC:11427 / OMIM:607207 (STIP1 homology and U-box containing protein 1)
603921	SUCLA2 / HGNC:11448 / OMIM:603921 (succinate-CoA ligase ADP-forming subunit beta)
611224	SUCLG1 / HGNC:11449 / OMIM:611224 (succinate-CoA ligase GDP/ADP-forming subunit alpha)
185620	SURF1 / HGNC:11474 / OMIM:185620 (SURF1 cytochrome c oxidase assembly factor)
604126	SVIL / HGNC:11480 / OMIM:604126 (supervillin)
608441	SYNE1 / HGNC:17089 / OMIM:608441 (spectrin repeat containing nuclear envelope protein 1)
608442	SYNE2 / HGNC:17084 / OMIM:608442 (spectrin repeat containing nuclear envelope protein 2)
610949	SYT14 / HGNC:23143 / OMIM:610949 (synaptotagmin 14)
600104	SYT2 / HGNC:11510 / OMIM:600104 (synaptotagmin 2)
605078	TARDBP / HGNC:11571 / OMIM:605078 (TAR DNA binding protein)
300394	TFAZZIN / HGNC:11577 / OMIM:300394 (tafazzin, phospholipid-lysophospholipid transacylase)
604649	TBCD / HGNC:11581 / OMIM:604649 (tubulin folding cofactor D)
604834	TBK1 / HGNC:11584 / OMIM:604834 (TANK binding kinase 1)

600075	TBP / HGNC:11588 / OMIM:600075 (TATA-box binding protein)
604488	TCAP / HGNC:11610 / OMIM:604488 (titin-cap)
607198	TDP1 / HGNC:18884 / OMIM:607198 (tyrosyl-DNA phosphodiesterase 1)
605764	TDP2 / HGNC:17768 / OMIM:605764 (tyrosyl-DNA phosphodiesterase 2)
615000	TECPR2 / HGNC:19957 / OMIM:615000 (tectonin beta-propeller repeat containing 2)
617242	TECRL / HGNC:27365 / OMIM:617242 (trans-2,3-enoyl-CoA reductase like)
602498	TFG / HGNC:11758 / OMIM:602498 (trafficking from ER to golgi regulator)
190230	TGFB3 / HGNC:11769 / OMIM:190230 (transforming growth factor beta 3)
613900	TGM6 / HGNC:16255 / OMIM:613900 (transglutaminase 6)
603518	TIA1 / HGNC:11802 / OMIM:603518 (TIA1 cytotoxic granule associated RNA binding protein)
607251	TIMM22 / HGNC:17317 / OMIM:607251 (translocase of inner mitochondrial membrane 22)
188250	TK2 / HGNC:11831 / OMIM:188250 (thymidine kinase 2)
616101	TMEM240 / HGNC:25186 / OMIM:616101 (transmembrane protein 240)
612048	TMEM43 / HGNC:28472 / OMIM:612048 (transmembrane protein 43)
616609	TMEM65 / HGNC:25203 / OMIM:616609 (transmembrane protein 65)
188380	TMPO / HGNC:11875 / OMIM:188380 (thymopoietin)
191040	TNNC1 / HGNC:11943 / OMIM:191040 (troponin C1, slow skeletal and cardiac type)
191043	TNNI2 / HGNC:11946 / OMIM:191043 (troponin I2, fast skeletal type)
191044	TNNI3 / HGNC:11947 / OMIM:191044 (troponin I3, cardiac type)
191041	TNNT1 / HGNC:11948 / OMIM:191041 (troponin T1, slow skeletal type)
191045	TNNT2 / HGNC:11949 / OMIM:191045 (troponin T2, cardiac type)
600692	TNNT3 / HGNC:11950 / OMIM:600692 (troponin T3, fast skeletal type)
610032	TNPO3 / HGNC:17103 / OMIM:610032 (transportin 3)
601243	TOP3A / HGNC:11992 / OMIM:601243 (DNA topoisomerase III alpha)
605204	TOR1A / HGNC:3098 / OMIM:605204 (torsin family 1 member A)
614512	TOR1AIP1 / HGNC:29456 / OMIM:614512 (torsin 1A interacting protein 1)
191010	TPM1 / HGNC:12010 / OMIM:191010 (tropomyosin 1)

190990	TPM2 / HGNC:12011 / OMIM:190990 (tropomyosin 2)
191030	TPM3 / HGNC:12012 / OMIM:191030 (tropomyosin 3)
614138	TRAPPC11 / HGNC:25751 / OMIM:614138 (trafficking protein particle complex subunit 11)
603283	TRDN / HGNC:12261 / OMIM:603283 (triadin)
614141	TRIM2 / HGNC:15974 / OMIM:614141 (tripartite motif containing 2)
602290	TRIM32 / HGNC:16380 / OMIM:602290 (tripartite motif containing 32)
606474	TRIM54 / HGNC:16008 / OMIM:606474 (tripartite motif containing 54)
606131	TRIM63 / HGNC:16007 / OMIM:606131 (tripartite motif containing 63)
604501	TRIP4 / HGNC:12310 / OMIM:604501 (thyroid hormone receptor interactor 4)
602345	TRPC3 / HGNC:12335 / OMIM:602345 (transient receptor potential cation channel subfamily C member 3)
605427	TRPV4 / HGNC:18083 / OMIM:605427 (transient receptor potential cation channel subfamily V member 4)
604723	TSFM / HGNC:12367 / OMIM:604723 (Ts translation elongation factor, mitochondrial)
611695	TTBK2 / HGNC:19141 / OMIM:611695 (tau tubulin kinase 2)
188840	TTN / HGNC:12403 / OMIM:188840 (titin)
600415	TTPA / HGNC:12404 / OMIM:600415 (alpha tocopherol transfer protein)
176300	TTR / HGNC:12405 / OMIM:176300 (transthyretin)
191110	TUBA4A / HGNC:12407 / OMIM:191110 (tubulin alpha 4a)
602661	TUBB3 / HGNC:20772 / OMIM:602661 (tubulin beta 3 class III)
606075	TWINK / HGNC:1160 / OMIM:606075 (twinkle mtDNA helicase)
131222	TYMP / HGNC:3148 / OMIM:131222 (thymidine phosphorylase)
314370	UBA1 / HGNC:12469 / OMIM:314370 (ubiquitin like modifier activating enzyme 1)
609787	UBAP1 / HGNC:12461 / OMIM:609787 (ubiquitin associated protein 1)
300264	UBQLN2 / HGNC:12509 / OMIM:300264 (ubiquilin 2)
191342	UCHL1 / HGNC:12513 / OMIM:191342 (ubiquitin C-terminal hydrolase L1)
605836	UNC13B / HGNC:12566 / OMIM:605836 (unc-13 homolog B)
611220	UNC45B / HGNC:14304 / OMIM:611220 (unc-45 myosin chaperone B)
185880	VAMP1 / HGNC:12642 / OMIM:185880 (vesicle associated membrane protein 1)

605704	VAPB / HGNC:12649 / OMIM:605704 (VAMP associated protein B and C)
193065	VCL / HGNC:12665 / OMIM:193065 (vinculin)
601023	VCP / HGNC:12666 / OMIM:601023 (valosin containing protein)
300913	VMA21 / HGNC:22082 / OMIM:300913 (vacuolar ATPase assembly factor VMA21)
608877	VPS13D / HGNC:23595 / OMIM:608877 (vacuolar protein sorting 13 homolog D)
609927	VPS37A / HGNC:24928 / OMIM:609927 (VPS37A subunit of ESCRT-I)
602168	VRK1 / HGNC:12718 / OMIM:602168 (VRK serine/threonine kinase 1)
614884	VWA3B / HGNC:28385 / OMIM:614884 (von Willebrand factor A domain containing 3B)
191050	WARS1 / HGNC:12729 / OMIM:191050 (tryptophanyl-tRNA synthetase 1)
605232	WNK1 / HGNC:14540 / OMIM:605232 (WNK lysine deficient protein kinase 1)
605131	WWOX / HGNC:12799 / OMIM:605131 (WW domain containing oxidoreductase)
194360	XRCC1 / HGNC:12828 / OMIM:194360 (X-ray repair cross complementing 1)
603623	YARS1 / HGNC:12840 / OMIM:603623 (tyrosyl-tRNA synthetase 1)
610957	YARS2 / HGNC:24249 / OMIM:610957 (tyrosyl-tRNA synthetase 2)
613915	ZBTB42 / HGNC:32550 / OMIM:613915 (zinc finger and BTB domain containing 42)
617828	ZFH2 / HGNC:20152 / OMIM:617828 (zinc finger homeobox 2)
612012	ZFYVE26 / HGNC:20761 / OMIM:612012 (zinc finger FYVE-type containing 26)
610243	ZFYVE27 / HGNC:26559 / OMIM:610243 (zinc finger FYVE-type containing 27)

386	[gendia_hgnc_ont] Show the field ONLY if: [gendia_listpref]='ontology'	Gene HGNC-code (browse ontology)If the affected gene is not present in the neuromuscular gene list above (e.g. a new gene not previously associated with neuromuscular disease) please search for the gene at <a href="https://www.genenames.org/tools/search/">https://www.genenames.org/tools/search/</a> and enter the numerical HGNC ID here (e.g. for DMD HGNC:2928 you would enter HGNC_2928) <i>Genetic diagnosis retained by the specialised centre (Element 6.2. in the Set of common data elements for Rare Diseases Registration (link))</i>	text, Required <table border="1"> <tr> <td>BIOPORTAL:HGNC</td> <td>BIOPORTAL:HGNC</td> </tr> </table>	BIOPORTAL:HGNC	BIOPORTAL:HGNC
BIOPORTAL:HGNC	BIOPORTAL:HGNC				
387	[gendia_omim_ont] Show the field ONLY if: [gendia_listpref]='ontology'	Gene OMIM-code (browse ontology)If the affected gene is not present in the neuromuscular gene list above (e.g. a new gene not previously associated with neuromuscular disease) please search for the gene at <a href="https://www.omim.org/">https://www.omim.org/</a> and enter the numerical OMIM ID here (e.g. for DMD you would enter 300377; you also find the code for this example if you type 'Dystrophin' to the box on the right) <i>Genetic diagnosis retained by the specialised centre (Element 6.2. in the Set of common data elements for Rare Diseases Registration (link))</i>	text, Required <table border="1"> <tr> <td>BIOPORTAL:OMIM</td> <td>BIOPORTAL:OMIM</td> </tr> </table>	BIOPORTAL:OMIM	BIOPORTAL:OMIM
BIOPORTAL:OMIM	BIOPORTAL:OMIM				
388	[gendia_omim]	Gene OMIM code calculated field	calc Calculation: if([gendia_listpref]='ontology', [gendia_omim_ont], if([gendia_listpref]='all_nmd',		

			[gendia_omim_list], ") Field Annotation: @HIDDEN @HIDDEN-PDF @READONLY																												
389	[gendia_inher]  Show the field ONLY if: [gendia_known] = 'yes'	Inheritance	dropdown <table border="1"> <tr><td>autosomal_dominant</td><td>Autosomal dominant</td></tr> <tr><td>autosomal_dominant_dn</td><td>Autosomal dominant - de novo</td></tr> <tr><td>sporadic</td><td>Sporadic</td></tr> <tr><td>autosomal_recessive</td><td>Autosomal recessive</td></tr> <tr><td>x_linked</td><td>X-linked</td></tr> <tr><td>x_linked_dom</td><td>X-linked dominant</td></tr> <tr><td>x_linked_rec</td><td>X-linked recessive</td></tr> <tr><td>y_linked</td><td>Y-linked inheritance</td></tr> <tr><td>digentic</td><td>Digentic inheritance</td></tr> <tr><td>mitochondrial</td><td>Mitochondrial inheritance</td></tr> <tr><td>oligogenic</td><td>Oligogenic inheritance</td></tr> <tr><td>autosomal_dominant_pat</td><td>Autosomal dominant with paternal imprinting</td></tr> <tr><td>autosomal_dominant_mat</td><td>Autosomal dominant with maternal imprinting</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	autosomal_dominant	Autosomal dominant	autosomal_dominant_dn	Autosomal dominant - de novo	sporadic	Sporadic	autosomal_recessive	Autosomal recessive	x_linked	X-linked	x_linked_dom	X-linked dominant	x_linked_rec	X-linked recessive	y_linked	Y-linked inheritance	digentic	Digentic inheritance	mitochondrial	Mitochondrial inheritance	oligogenic	Oligogenic inheritance	autosomal_dominant_pat	Autosomal dominant with paternal imprinting	autosomal_dominant_mat	Autosomal dominant with maternal imprinting	unknown	Unknown
autosomal_dominant	Autosomal dominant																														
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x_linked_rec	X-linked recessive																														
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390	[gendia_square_bracket]	Section Header: <i>DISEASE-SPECIFIC GENETIC DETAILS</i> gendia_square_bracket	text Field Annotation: @DEFAULT='[' @HIDDEN @HIDDEN-PDF																												
391	[gendia_desc4]  Show the field ONLY if: [gendia_known] = 'yes'	The genetic reporting methods in this section are displayed based on the disease group ([arm-label]) or the gene selected above (OMIM:[gendia_omim]) or the clinical diagnosis indicated in the "Baseline"-form ([first-event-name][diagnosis_rd_orpha] / [first-event-name][diagnosis_rd_orpha:value]).	descriptive																												
392	[helper_var1_v2]	#SECTION Oculopharyngeal Muscular Dystrophy (OPMD, ORPHA:270)	descriptive Field Annotation: @HIDDEN @HIDDEN-PDF																												
393	[gendia_pabpn1_report_v1]  Show the field ONLY if: [gendia_omim] = 602279	Section Header: <i>PABPN1 (OMIM:602279; HGNC:8565; e.g. for diagnosis ORPHA:270)</i>  Report number of GCG-repeats in PABPN1-gene (OMIM:602279; HGNC:8565) at NM_004643.3:c.4_6GCG	radio, Required <table border="1"> <tr><td>distinct_number</td><td>Report a distinct number</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> <tr><td>not_applicable</td><td>Not applicable</td></tr> <tr><td>not_present</td><td>Not present</td></tr> </table> Field Annotation: @DEFAULT='[pabpn1_v_report_def]'	distinct_number	Report a distinct number	unknown	Unknown	not_applicable	Not applicable	not_present	Not present																				
distinct_number	Report a distinct number																														
unknown	Unknown																														
not_applicable	Not applicable																														
not_present	Not present																														
394	[gendia_pabpn1_nrep_v1]  Show the field ONLY if: [gendia_omim] = 602279 AND [gendia_pabpn1_report_v1] = 'distinct_number'	Number of GCG repeats in PABPN1-gene (OMIM:602279; HGNC:8565) at NM_004643.3:c.4_6GCG <i>expected: integer between 8 and 13</i>	text (integer, Min: 8, Max: 13), Required																												
395	[gendia_pabpn1_hgvs_v1]  Show the field ONLY if: [gendia_omim] = 602279 AND [gendia_pabpn1_report_v1] = 'distinct_number'	gendia_pabpn1_hgvs_v1	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_pabpn1_nrep_v1]!="",concat("NM", [gendia_square_bracket], [gendia_pabpn1_nrep_v1], ']), ""))																												
396	[gendia_pabpn1_hgvs_display]  Show the field ONLY if: [gendia_omim] = 602279 AND	Resulting HGVS variant notation:[gendia_pabpn1_hgvs_v1]	descriptive																												

	D [gencia_pabpn1_report_v1] = 'distinct_number'																																																				
397	[gencia_pabpn1_gen_build_v1]  Show the field ONLY if: [gencia_omim] = 602279 AND [gencia_pabpn1_report_v1] = 'distinct_number'	Genomic build	dropdown <table border="1"> <tr> <td>GRCh38</td> <td>GRCh38 (hg38)</td> </tr> <tr> <td>GRCh37</td> <td>GRCh37 (hg19)</td> </tr> </table>	GRCh38	GRCh38 (hg38)	GRCh37	GRCh37 (hg19)																																														
GRCh38	GRCh38 (hg38)																																																				
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398	[gencia_pabpn1_gen_approach_v1]  Show the field ONLY if: [gencia_omim] = 602279 AND [gencia_pabpn1_report_v1] = 'distinct_number'	Genetic approach	dropdown <table border="1"> <tr> <td>single_gene_screening</td> <td>Single Gene Screening</td> </tr> <tr> <td>acgh</td> <td>aCGH</td> </tr> <tr> <td>snp_array</td> <td>SNP array</td> </tr> <tr> <td>mlpa</td> <td>MLPA</td> </tr> <tr> <td>karyotype</td> <td>Karyotype</td> </tr> <tr> <td>ngs_panel</td> <td>NGS panel</td> </tr> <tr> <td>wes</td> <td>WES</td> </tr> <tr> <td>wgs</td> <td>WGS</td> </tr> </table>	single_gene_screening	Single Gene Screening	acgh	aCGH	snp_array	SNP array	mlpa	MLPA	karyotype	Karyotype	ngs_panel	NGS panel	wes	WES	wgs	WGS																																		
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399	[gencia_pabpn1_chrom_v1]  Show the field ONLY if: [gencia_omim] = 602279 AND [gencia_pabpn1_report_v1] = 'distinct_number'	Chromosome	dropdown <table border="1"> <tr><td>1</td><td>1</td></tr> <tr><td>2</td><td>2</td></tr> <tr><td>3</td><td>3</td></tr> <tr><td>4</td><td>4</td></tr> <tr><td>5</td><td>5</td></tr> <tr><td>6</td><td>6</td></tr> <tr><td>7</td><td>7</td></tr> <tr><td>8</td><td>8</td></tr> <tr><td>9</td><td>9</td></tr> <tr><td>10</td><td>10</td></tr> <tr><td>11</td><td>11</td></tr> <tr><td>12</td><td>12</td></tr> <tr><td>13</td><td>13</td></tr> <tr><td>14</td><td>14</td></tr> <tr><td>15</td><td>15</td></tr> <tr><td>16</td><td>16</td></tr> <tr><td>17</td><td>17</td></tr> <tr><td>18</td><td>18</td></tr> <tr><td>19</td><td>19</td></tr> <tr><td>20</td><td>20</td></tr> <tr><td>21</td><td>21</td></tr> <tr><td>22</td><td>22</td></tr> <tr><td>X</td><td>X</td></tr> <tr><td>Y</td><td>Y</td></tr> <tr><td>MT</td><td>MT</td></tr> </table>	1	1	2	2	3	3	4	4	5	5	6	6	7	7	8	8	9	9	10	10	11	11	12	12	13	13	14	14	15	15	16	16	17	17	18	18	19	19	20	20	21	21	22	22	X	X	Y	Y	MT	MT
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400	[gencia_pabpn1_zygosity_v1]  Show the field ONLY if: [gencia_omim] = 602279 AND [gencia_pabpn1_report_v1] = 'distinct_number'	Zygosity	dropdown <table border="1"> <tr> <td>heterozygous</td> <td>Heterozygous</td> </tr> <tr> <td>compound_heterozygous</td> <td>Compound heterozygous</td> </tr> <tr> <td>homozygous</td> <td>Homozygous</td> </tr> <tr> <td>hemizygous</td> <td>Hemizygous</td> </tr> <tr> <td>mosaic</td> <td>Mosaic</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	heterozygous	Heterozygous	compound_heterozygous	Compound heterozygous	homozygous	Homozygous	hemizygous	Hemizygous	mosaic	Mosaic	unknown	Unknown																																						
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mosaic	Mosaic																																																				
unknown	Unknown																																																				

401	[helper_var2_v2]	#SECTION Myotonic Dystrophy type 1 (DM1, ORPHA:273)	descriptive Field Annotation: @HIDDEN @HIDDEN-PDF										
402	[gendia_dmpk_report_v1] Show the field ONLY if: [gendia_omim] = 605377	Section Header: DMPK (OMIM:605377; HGNC:2933; e.g. for diagnosis ORPHA:273) Report number of CTG-repeats in DMPK-gene (OMIM:605377; HGNC:2933) at NM_001081563.1:c.*224_226CTG	radio, Required <table border="1"> <tr> <td>distinct_number</td> <td>Report a distinct number</td> </tr> <tr> <td>range</td> <td>[for higher repeats]: Report a range</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> <tr> <td>not_applicable</td> <td>Not applicable</td> </tr> <tr> <td>not_present</td> <td>Not present</td> </tr> </table> Field Annotation: @DEFAULT='[dmpk_v_report_def]'	distinct_number	Report a distinct number	range	[for higher repeats]: Report a range	unknown	Unknown	not_applicable	Not applicable	not_present	Not present
distinct_number	Report a distinct number												
range	[for higher repeats]: Report a range												
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not_applicable	Not applicable												
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403	[gendia_dmpk_nrep_v1] Show the field ONLY if: [gendia_omim] = 605377 AND [gendia_dmpk_report_v1] = 'distinct_number'	Number of CTG-repeats in DMPK-gene (OMIM:605377; HGNC:2933) at NM_001081563.1:c.*224_226CTG <i>expected: integer, minimum: 50</i>	text (integer, Min: 50), Required										
404	[gendia_dmpk_nrep_r1_v1] Show the field ONLY if: [gendia_omim] = 605377 AND [gendia_dmpk_report_v1] = 'range'	Inclusive lower bound of range for number of CTG-repeats in DMPK-gene (OMIM:605377; HGNC:2933) at NM_001081563.1:c.*224_226CTG <i>expected: integer, minimum: 50</i>	text (integer, Min: 50), Required										
405	[gendia_dmpk_nrep_r2_v1] Show the field ONLY if: [gendia_omim] = 605377 AND [gendia_dmpk_report_v1] = 'range'	Inclusive upper bound of range for number of CTG-repeats in DMPK-gene (OMIM:605377; HGNC:2933) at NM_001081563.1:c.*224_226CTG <i>expected: integer</i>	text (integer, Min: 51), Required										
406	[gendia_dmpk_hgvs_v1] Show the field ONLY if: [gendia_omim] = 605377 AND ([gendia_dmpk_report_v1] = 'distinct_number' OR [gendia_dmpk_report_v1] = 'range')	gendia_dmpk_hgvs_v1	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_dmpk_report_v1] = 'distinct_number', concat('NM_001081563.1:c.*224_226CTG', [gendia_square_bracket], [gendia_dmpk_nrep_v1], ']'), if([gendia_dmpk_report_v1]='range', concat('NM_001081563.1:c.*224_226CTG', [gendia_square_bracket], [gendia_dmpk_nrep_r1_v1], '_', [gendia_dmpk_nrep_r2_v1], ']'), '')))										
407	[gendia_dmpk_hgvs_disp_v1] Show the field ONLY if: [gendia_omim] = 605377 AND ([gendia_dmpk_report_v1] = 'distinct_number' OR [gendia_dmpk_report_v1] = 'range')	Resulting variant notation:[gendia_dmpk_hgvs_v1]	descriptive										
408	[gendia_dmpk_gen_build_v1] Show the field ONLY if: [gendia_omim] = 605377 AND ([gendia_dmpk_report_v1] = 'distinct_number' OR [gendia_dmpk_report_v1] = 'range')	Genomic build	dropdown <table border="1"> <tr> <td>GRCh38</td> <td>GRCh38 (hg38)</td> </tr> <tr> <td>GRCh37</td> <td>GRCh37 (hg19)</td> </tr> </table>	GRCh38	GRCh38 (hg38)	GRCh37	GRCh37 (hg19)						
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409	[gendia_dmpk_gen_app_v1] Show the field ONLY if: [gendia_omim] = 605377 AND ([gendia_dmpk_report_v1] = 'distinct_number' OR [gendia_dmpk_report_v1] = 'range')	Genetic approach	dropdown <table border="1"> <tr> <td>single_gene_screening</td> <td>Single Gene Screening</td> </tr> <tr> <td>acgh</td> <td>aCGH</td> </tr> <tr> <td>snp_array</td> <td>SNP array</td> </tr> <tr> <td>mlpa</td> <td>MLPA</td> </tr> <tr> <td>karyotype</td> <td>Karyotype</td> </tr> </table>	single_gene_screening	Single Gene Screening	acgh	aCGH	snp_array	SNP array	mlpa	MLPA	karyotype	Karyotype
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410	<p>[gendia_dmpk_chrom_v1]</p> <p>Show the field ONLY if: [gendia_omim] = 605377 AND ([gendia_dmpk_report_v1] = 'distinct_number' OR [gendia_dmpk_report_v1] = 'range')</p>	Chromosome	dropdown <table border="1"> <tr><td>1</td><td>1</td></tr> <tr><td>2</td><td>2</td></tr> <tr><td>3</td><td>3</td></tr> <tr><td>4</td><td>4</td></tr> <tr><td>5</td><td>5</td></tr> <tr><td>6</td><td>6</td></tr> <tr><td>7</td><td>7</td></tr> <tr><td>8</td><td>8</td></tr> <tr><td>9</td><td>9</td></tr> <tr><td>10</td><td>10</td></tr> <tr><td>11</td><td>11</td></tr> <tr><td>12</td><td>12</td></tr> <tr><td>13</td><td>13</td></tr> <tr><td>14</td><td>14</td></tr> <tr><td>15</td><td>15</td></tr> <tr><td>16</td><td>16</td></tr> <tr><td>17</td><td>17</td></tr> <tr><td>18</td><td>18</td></tr> <tr><td>19</td><td>19</td></tr> <tr><td>20</td><td>20</td></tr> <tr><td>21</td><td>21</td></tr> <tr><td>22</td><td>22</td></tr> <tr><td>X</td><td>X</td></tr> <tr><td>Y</td><td>Y</td></tr> <tr><td>MT</td><td>MT</td></tr> </table>		1	1	2	2	3	3	4	4	5	5	6	6	7	7	8	8	9	9	10	10	11	11	12	12	13	13	14	14	15	15	16	16	17	17	18	18	19	19	20	20	21	21	22	22	X	X	Y	Y	MT	MT
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411	<p>[gendia_dmpk_zygos_v1]</p> <p>Show the field ONLY if: [gendia_omim] = 605377 AND ([gendia_dmpk_report_v1] = 'distinct_number' OR [gendia_dmpk_report_v1] = 'range')</p>	Zygosity	dropdown <table border="1"> <tr><td>heterozygous</td><td>Heterozygous</td></tr> <tr><td>compound_heterozygous</td><td>Compound heterozygous</td></tr> <tr><td>homozygous</td><td>Homozygous</td></tr> <tr><td>hemizygous</td><td>Hemizygous</td></tr> <tr><td>mosaic</td><td>Mosaic</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>		heterozygous	Heterozygous	compound_heterozygous	Compound heterozygous	homozygous	Homozygous	hemizygous	Hemizygous	mosaic	Mosaic	unknown	Unknown																																						
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mosaic	Mosaic																																																					
unknown	Unknown																																																					
412	[helper_var3_v2]	#SECTION Myotonic Dystrophy type 2 (DM2, ORPHA:606)	descriptive Field Annotation: @HIDDEN @HIDDEN-PDF																																																			
413	<p>[gendia_cnbp_report_v1]</p> <p>Show the field ONLY if: [gendia_omim] = 116955</p>	Section Header: <i>CNBP (OMIM:116955; HGNC:13164; e.g. for diagnosis ORPHA:606)</i> Report number of CCTG-repeats in CNBP-gene (OMIM:116955; HGNC:13164) at NM_003418.5(CNBP):c.-14-833_-14-806CCTG	radio, Required <table border="1"> <tr><td>distinct_number</td><td>Report a distinct number</td></tr> <tr><td>range</td><td>[for higher repeats]: Report a range</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> <tr><td>not_applicable</td><td>Not applicable</td></tr> <tr><td>not_present</td><td>Not present</td></tr> </table> Field Annotation: @DEFAULT=[cnbp_v_report_def]		distinct_number	Report a distinct number	range	[for higher repeats]: Report a range	unknown	Unknown	not_applicable	Not applicable	not_present	Not present																																								
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414	<p>[gendia_cnbp_nrep_v1]</p> <p>Show the field ONLY if: [gendia_omim] = 116955 AND</p>	Number of CCTG-repeats in CNBP-gene (OMIM:116955; HGNC:13164) at NM_003418.5(CNBP):c.-14-833_-14-806CCTG <i>expected: integer, minimum: 75</i>	text (integer, Min: 75), Required																																																			



	D [genda_cnbp_report_v1] = 'distinct_number'																		
415	[genda_cnbp_nrep_r1_v1] Show the field ONLY if: [genda_omim] = 116955 AND [genda_cnbp_report_v1] = 'range'	Inclusive lower bound of range for number of CCTG-repeats in CNBP-gene (OMIM:116955; HGNC:13164) at NM_003418.5(CNBP):c.-14-833_-14-806CCTG <i>expected: integer, minimum: 75</i>	text (integer, Min: 75), Required																
416	[genda_cnbp_nrep_r2_v1] Show the field ONLY if: [genda_omim] = 116955 AND [genda_cnbp_report_v1] = 'range'	Inclusive upper bound of range for number of CCTG-repeats in CNBP-gene (OMIM:116955; HGNC:13164) at NM_003418.5(CNBP):c.-14-833_-14-806CCTG <i>expected: integer</i>	text (integer, Min: 76), Required																
417	[genda_cnbp_hgvs_v1] Show the field ONLY if: [genda_omim] = 116955 AND ([genda_cnbp_report_v1] = 'distinct_number' OR [genda_cnbp_report_v1]='range')	cnbp_v1_hgvs	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([genda_cnbp_report_v1]='distinct_number', concat('NM_003418.5(CNBP):c.-14-833_-14-806CCTG', [genda_square_bracket], [genda_cnbp_nrep_v1], ','), if([genda_cnbp_report_v1]='range', concat('NM_003418.5(CNBP):c.-14-833_-14-806CCTG', [genda_square_bracket], [genda_cnbp_nrep_r1_v1], ',', [genda_cnbp_nrep_r2_v1], ','), '')))																
418	[genda_cnbp_hgvs_disp_v1] Show the field ONLY if: [genda_omim] = 116955 AND ([genda_cnbp_report_v1] = 'distinct_number' OR [genda_cnbp_report_v1]='range')	Resulting variant notation:[genda_cnbp_hgvs_v1]	descriptive																
419	[genda_cnbp_gen_build_v1] Show the field ONLY if: [genda_omim] = 116955 AND ([genda_cnbp_report_v1] = 'distinct_number' OR [genda_cnbp_report_v1]='range')	Genomic build	dropdown <table border="1"> <tr> <td>GRCh38</td> <td>GRCh38 (hg38)</td> </tr> <tr> <td>GRCh37</td> <td>GRCh37 (hg19)</td> </tr> </table>	GRCh38	GRCh38 (hg38)	GRCh37	GRCh37 (hg19)												
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420	[genda_cnbp_gen_app_v1] Show the field ONLY if: [genda_omim] = 116955 AND ([genda_cnbp_report_v1] = 'distinct_number' OR [genda_cnbp_report_v1]='range')	Genetic approach	dropdown <table border="1"> <tr> <td>single_gene_screening</td> <td>Single Gene Screening</td> </tr> <tr> <td>acgh</td> <td>aCGH</td> </tr> <tr> <td>snp_array</td> <td>SNP array</td> </tr> <tr> <td>mlpa</td> <td>MLPA</td> </tr> <tr> <td>karyotype</td> <td>Karyotype</td> </tr> <tr> <td>ngs_panel</td> <td>NGS panel</td> </tr> <tr> <td>wes</td> <td>WES</td> </tr> <tr> <td>wgs</td> <td>WGS</td> </tr> </table>	single_gene_screening	Single Gene Screening	acgh	aCGH	snp_array	SNP array	mlpa	MLPA	karyotype	Karyotype	ngs_panel	NGS panel	wes	WES	wgs	WGS
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421	[genda_cnbp_chrom_v1] Show the field ONLY if: [genda_omim] = 116955 AND ([genda_cnbp_report_v1] = 'distinct_number' OR [genda_cnbp_report_v1]='range')	Chromosome	dropdown <table border="1"> <tr><td>1</td><td>1</td></tr> <tr><td>2</td><td>2</td></tr> <tr><td>3</td><td>3</td></tr> <tr><td>4</td><td>4</td></tr> <tr><td>5</td><td>5</td></tr> <tr><td>6</td><td>6</td></tr> <tr><td>7</td><td>7</td></tr> <tr><td>8</td><td>8</td></tr> </table>	1	1	2	2	3	3	4	4	5	5	6	6	7	7	8	8
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422	<p>[gencia_cnbp_zygos_v1]</p> <p>Show the field ONLY if:  [gencia_omim] = 116955 AND  D ([gencia_cnbp_report_v1]  = 'distinct_number' OR [gen  dia_cnbp_report_v1]='range'  )</p>	Zygosity	<p>dropdown</p> <table border="1"> <tr><td>heterozygous</td><td>Heterozygous</td></tr> <tr><td>compound_heterozygous</td><td>Compound heterozygous</td></tr> <tr><td>homozygous</td><td>Homozygous</td></tr> <tr><td>hemizygous</td><td>Hemizygous</td></tr> <tr><td>mosaic</td><td>Mosaic</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	heterozygous	Heterozygous	compound_heterozygous	Compound heterozygous	homozygous	Homozygous	hemizygous	Hemizygous	mosaic	Mosaic	unknown	Unknown																						
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423	[helper_var8_v2]	#SECTION Facioscapulohumeral Muscular Dystrophy type 1 (FSDH, OMIM:158900, ORPHA:269 - orphacode not for type1 only)	<p>descriptive</p> <p>Field Annotation: @HIDDEN @HIDDEN-PDF</p>																																		
424	<p>[gencia_frag_4q35_report]</p> <p>Show the field ONLY if:  [gencia_known] = 'yes' AND  [gencia_omim] = 601278</p>	<p>Section Header: <i>FRG1 (OMIM:601278; HGNC:3954; e.g. for diagnosis ORPHA:269): 4q35 fragment</i></p> <p>4q35 fragment:  Report size of remaining EcoRI/BlnI fragment in kB</p>	<p>radio</p> <table border="1"> <tr><td>distinct_number</td><td>Report a distinct number</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> <tr><td>not_applicable</td><td>Not applicable</td></tr> <tr><td>not_present</td><td>Not present</td></tr> </table> <p>Field Annotation:  @DEFAULT='[fragment_4q35_report_def]'</p>	distinct_number	Report a distinct number	unknown	Unknown	not_applicable	Not applicable	not_present	Not present																										
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425	<p>[gencia_frag_4q35_size]</p> <p>Show the field ONLY if:  [gencia_known] = 'yes' AND  [gencia_omim] = 601278 AND  D [gencia_frag_4q35_report]  ='distinct_number'</p>	<p>4q35 fragment:  Size of remaining EcoRI/BlnI fragment in kB  <i>expected: integer between 0 and 40</i></p>	text (integer, Min: 0, Max: 40), Required																																		
426	<p>[gencia_4q35_gen_build]</p> <p>Show the field ONLY if:  [gencia_known] = 'yes' AND  [gencia_omim] = 601278 AND  D [gencia_frag_4q35_report]  ='distinct_number'</p>	Genomic build	<p>dropdown</p> <table border="1"> <tr><td>GRCh38</td><td>GRCh38 (hg38)</td></tr> <tr><td>GRCh37</td><td>GRCh37 (hg19)</td></tr> </table>	GRCh38	GRCh38 (hg38)	GRCh37	GRCh37 (hg19)																														
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428	<p>[ <a href="#">gencia_4q35_chrom</a> ]</p> <p>Show the field ONLY if:  [gencia_known] = 'yes' AND  [gencia_omim] = 601278 AND  D [gencia_frag_4q35_report]  ='distinct_number'</p>	Chromosome	<p>dropdown</p> <table border="1"> <tr><td>1</td><td>1</td></tr> <tr><td>2</td><td>2</td></tr> <tr><td>3</td><td>3</td></tr> <tr><td>4</td><td>4</td></tr> <tr><td>5</td><td>5</td></tr> <tr><td>6</td><td>6</td></tr> <tr><td>7</td><td>7</td></tr> <tr><td>8</td><td>8</td></tr> <tr><td>9</td><td>9</td></tr> <tr><td>10</td><td>10</td></tr> <tr><td>11</td><td>11</td></tr> <tr><td>12</td><td>12</td></tr> <tr><td>13</td><td>13</td></tr> <tr><td>14</td><td>14</td></tr> <tr><td>15</td><td>15</td></tr> <tr><td>16</td><td>16</td></tr> <tr><td>17</td><td>17</td></tr> <tr><td>18</td><td>18</td></tr> <tr><td>19</td><td>19</td></tr> <tr><td>20</td><td>20</td></tr> <tr><td>21</td><td>21</td></tr> <tr><td>22</td><td>22</td></tr> <tr><td>X</td><td>X</td></tr> <tr><td>Y</td><td>Y</td></tr> <tr><td>MT</td><td>MT</td></tr> </table>	1	1	2	2	3	3	4	4	5	5	6	6	7	7	8	8	9	9	10	10	11	11	12	12	13	13	14	14	15	15	16	16	17	17	18	18	19	19	20	20	21	21	22	22	X	X	Y	Y	MT	MT
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430	[ <a href="#">helper_var4_v2</a> ]	#SECTION Charcot-Marie-Tooth 1A (CMT1A, ORPHA:101081) #SECTION Hereditary neuropathy with liability to pressure palsies (ORPHA:640)	descriptive Field Annotation: @HIDDEN @HIDDEN-PDF																																																		
431	<p>[ <a href="#">gd_pmp22_dup_del</a> ]</p> <p>Show the field ONLY if:  [gencia_omim] = 601097</p>	<p>Section Header: Duplication of PMP22 (OMIM:601097; HGNC:9118; e.g. for diagnosis ORPHA:101081) OR Deletion of PMP22 (OMIM:601097; HGNC:9118; e.g. for diagnosis ORPHA:640)</p> <p>Which change applies to PMP22-gene (OMIM:601097; HGNC:9118)?</p>	<p>radio</p> <table border="1"> <tr> <td>duplication</td> <td>Duplication of the gene</td> </tr> <tr> <td>deletion</td> <td>Deletion of the gene</td> </tr> <tr> <td>not_present</td> <td>Not present (none of the above)</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	duplication	Duplication of the gene	deletion	Deletion of the gene	not_present	Not present (none of the above)	unknown	Unknown																																										
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432	<p>[ <a href="#">gd_pmp22_dup_hgvs_v1</a> ]</p> <p>Show the field ONLY if:  [gencia_omim] = 601097 AND  D [gd_pmp22_dup_del] = 'duplication'</p>	gd_pmp22_dup_hgvs_v1	<p>text</p> <p>Field Annotation: @HIDDEN @HIDDEN-PDF  @CALCTEXT(if([gencia_omim] = 601097 AND [gd_pmp22_dup_del] = 'duplication', 'NM_000304.3:c.-238_*1140{2}', ''))</p>																																																		

433	[gd_pmp22_dup_hgvs_v2] Show the field ONLY if: [genda_omim] = 601097 AND [gd_pmp22_dup_del] = 'duplication'	gd_pmp22_dup_hgvs_v2	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([genda_omim] = 601097 AND [gd_pmp22_dup_del] = 'duplication', 'NC_000017.10:g.(?_15133094)_(15168674_?)dup', ''))																
434	[gd_pmp22_del_hgvs_v1] Show the field ONLY if: [genda_omim] = 601097 AND [gd_pmp22_dup_del] = 'deletion'	gd_pmp22_del_hgvs_v1	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([genda_omim] = 601097 AND [gd_pmp22_dup_del] = 'deletion', 'NM_000304.3:c.-238_*1140{0}', ''))																
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443	[gd_pmp22_zygos_v1] Show the field ONLY if: [genda_omim] = 601097 AND ([gd_pmp22_dup_del] = 'duplication' OR [gd_pmp22_dup_del] = 'deletion')	Zygoty	dropdown <table border="1"> <tr><td>heterozygous</td><td>Heterozygous</td></tr> <tr><td>compound_heterozygous</td><td>Compound heterozygous</td></tr> <tr><td>homozygous</td><td>Homozygous</td></tr> <tr><td>hemizygous</td><td>Hemizygous</td></tr> <tr><td>mosaic</td><td>Mosaic</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	heterozygous	Heterozygous	compound_heterozygous	Compound heterozygous	homozygous	Homozygous	hemizygous	Hemizygous	mosaic	Mosaic	unknown	Unknown																																
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444	[helper_var6_v2]	#SECTION Spinobulbar Muscular Atrophy (SBMA, ORPHA:481)	descriptive Field Annotation: @HIDDEN @HIDDEN-PDF																																												
445	[genda_ar_report_v1] Show the field ONLY if: [genda_omim] = 313700	Section Header: AR (OMIM:313700; HGNC:644; e.g. for diagnosis ORPHA:481) Report number of CAG-repeats in AR-gene (OMIM:313700; HGNC:644) at NM_000044.3:c.172_174CAG	radio, Required <table border="1"> <tr><td>distinct_number</td><td>Report a distinct number</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> <tr><td>not_applicable</td><td>Not applicable</td></tr> <tr><td>not_present</td><td>Not present</td></tr> </table> Field Annotation: @DEFAULT='[ar_v1_report_def]'	distinct_number	Report a distinct number	unknown	Unknown	not_applicable	Not applicable	not_present	Not present																																				
distinct_number	Report a distinct number																																														
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not_present	Not present																																														
446	[genda_ar_nrep_v1] Show the field ONLY if: [genda_omim] = 313700 AND [genda_ar_report_v1] = 'distinct_number'	Number of CAG-repeats in AR-gene (OMIM:313700; HGNC:644) at NM_000044.3:c.172_174CAG <i>expected: integer between 38 and 62</i>	text (integer, Min: 38, Max: 62)																																												
447	[genda_ar_hgvs_v1] Show the field ONLY if: [genda_omim] = 313700 AND [genda_ar_report_v1] = 'distinct_number'	genda_ar_hgvs_v1	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([genda_ar_nrep_v1]!="",concat("NM_000044.3:c.",[genda_square_bracket], [genda_ar_nrep_v1], "]"), ""))																																												
448	[genda_ar_hgvs_disp_v1]	Resulting variant notation:[genda_ar_hgvs_v1]	descriptive																																												

	Show the field ONLY if: [gendia_omim] = 313700 AND [gendia_ar_report_v1] = 'distinct_number'																																																				
449	[gendia_ar_gen_build_v1]  Show the field ONLY if: [gendia_omim] = 313700 AND [gendia_ar_report_v1] = 'distinct_number'	Genomic build	dropdown <table border="1"> <tr> <td>GRCh38</td> <td>GRCh38 (hg38)</td> </tr> <tr> <td>GRCh37</td> <td>GRCh37 (hg19)</td> </tr> </table>	GRCh38	GRCh38 (hg38)	GRCh37	GRCh37 (hg19)																																														
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451	[gendia_ar_chrom_v1]  Show the field ONLY if: [gendia_omim] = 313700 AND [gendia_ar_report_v1] = 'distinct_number'	Chromosome	dropdown <table border="1"> <tr><td>1</td><td>1</td></tr> <tr><td>2</td><td>2</td></tr> <tr><td>3</td><td>3</td></tr> <tr><td>4</td><td>4</td></tr> <tr><td>5</td><td>5</td></tr> <tr><td>6</td><td>6</td></tr> <tr><td>7</td><td>7</td></tr> <tr><td>8</td><td>8</td></tr> <tr><td>9</td><td>9</td></tr> <tr><td>10</td><td>10</td></tr> <tr><td>11</td><td>11</td></tr> <tr><td>12</td><td>12</td></tr> <tr><td>13</td><td>13</td></tr> <tr><td>14</td><td>14</td></tr> <tr><td>15</td><td>15</td></tr> <tr><td>16</td><td>16</td></tr> <tr><td>17</td><td>17</td></tr> <tr><td>18</td><td>18</td></tr> <tr><td>19</td><td>19</td></tr> <tr><td>20</td><td>20</td></tr> <tr><td>21</td><td>21</td></tr> <tr><td>22</td><td>22</td></tr> <tr><td>X</td><td>X</td></tr> <tr><td>Y</td><td>Y</td></tr> <tr><td>MT</td><td>MT</td></tr> </table>	1	1	2	2	3	3	4	4	5	5	6	6	7	7	8	8	9	9	10	10	11	11	12	12	13	13	14	14	15	15	16	16	17	17	18	18	19	19	20	20	21	21	22	22	X	X	Y	Y	MT	MT
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453	[helper_var9_v2]	#SECTION Spinal Muscular Atrophy (SMA types 1, 2, 3)	descriptive Field Annotation: @HIDDEN @HIDDEN-PDF																																														
454	[gendia_smn2_copy_n_known]	Section Header: <i>SMN2 (OMIM:601627; HGNC:11118) copy number</i> SMN2 (OMIM:601627; HGNC:11118) copy number known  Show the field ONLY if: [gendia_known] = 'yes' AND ([arm-number] = 5 OR [gendia_omim] = 601627)	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> </table>	yes	Yes	no	No																																										
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455	[gendia_smn2_copy_n]	SMN2 copy number <i>expected: integer between 1 and 6</i>	text (number, Min: 1, Max: 6), Required																																														
456	[gendia_smn2_gen_build_v1]	Genomic build  Show the field ONLY if: [gendia_smn2_copy_n] >= 1	dropdown <table border="1"> <tr> <td>GRCh38</td> <td>GRCh38 (hg38)</td> </tr> <tr> <td>GRCh37</td> <td>GRCh37 (hg19)</td> </tr> </table>	GRCh38	GRCh38 (hg38)	GRCh37	GRCh37 (hg19)																																										
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457	[gendia_smn2_gen_app_v1]	Genetic approach  Show the field ONLY if: [gendia_smn2_copy_n] >= 1	dropdown <table border="1"> <tr> <td>single_gene_screening</td> <td>Single Gene Screening</td> </tr> <tr> <td>acgh</td> <td>aCGH</td> </tr> <tr> <td>snp_array</td> <td>SNP array</td> </tr> <tr> <td>mlpa</td> <td>MLPA</td> </tr> <tr> <td>karyotype</td> <td>Karyotype</td> </tr> <tr> <td>ngs_panel</td> <td>NGS panel</td> </tr> <tr> <td>wes</td> <td>WES</td> </tr> <tr> <td>wgs</td> <td>WGS</td> </tr> </table>	single_gene_screening	Single Gene Screening	acgh	aCGH	snp_array	SNP array	mlpa	MLPA	karyotype	Karyotype	ngs_panel	NGS panel	wes	WES	wgs	WGS																														
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458	[gendia_smn2_chrom_v1]	Chromosome  Show the field ONLY if: [gendia_smn2_copy_n] >= 1	dropdown <table border="1"> <tr><td>1</td><td>1</td></tr> <tr><td>2</td><td>2</td></tr> <tr><td>3</td><td>3</td></tr> <tr><td>4</td><td>4</td></tr> <tr><td>5</td><td>5</td></tr> <tr><td>6</td><td>6</td></tr> <tr><td>7</td><td>7</td></tr> <tr><td>8</td><td>8</td></tr> <tr><td>9</td><td>9</td></tr> <tr><td>10</td><td>10</td></tr> <tr><td>11</td><td>11</td></tr> <tr><td>12</td><td>12</td></tr> <tr><td>13</td><td>13</td></tr> <tr><td>14</td><td>14</td></tr> <tr><td>15</td><td>15</td></tr> <tr><td>16</td><td>16</td></tr> <tr><td>17</td><td>17</td></tr> <tr><td>18</td><td>18</td></tr> <tr><td>19</td><td>19</td></tr> <tr><td>20</td><td>20</td></tr> <tr><td>21</td><td>21</td></tr> <tr><td></td><td></td></tr> <tr><td></td><td></td></tr> </table>	1	1	2	2	3	3	4	4	5	5	6	6	7	7	8	8	9	9	10	10	11	11	12	12	13	13	14	14	15	15	16	16	17	17	18	18	19	19	20	20	21	21				
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459	<p>[gendia_smn2_zygos_v1]</p> <p>Show the field ONLY if: [gendia_smn2_copy_n] &gt;= 1</p>	Zygosity	<p>dropdown</p> <table border="1"> <tr><td>heterozygous</td><td>Heterozygous</td></tr> <tr><td>compound_heterozygous</td><td>Compound heterozygous</td></tr> <tr><td>homozygous</td><td>Homozygous</td></tr> <tr><td>hemizygous</td><td>Hemizygous</td></tr> <tr><td>mosaic</td><td>Mosaic</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	heterozygous	Heterozygous	compound_heterozygous	Compound heterozygous	homozygous	Homozygous	hemizygous	Hemizygous	mosaic	Mosaic	unknown	Unknown
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460	<p>[gendia_smn1_variant_v1]</p> <p>Show the field ONLY if: [gendia_known] = 'yes' AND ([arm-number]=5 OR [gendia_omim]=600354)</p>	<p>Section Header: SMN1 (OMIM:600354; HGNC:11117)</p> <p>SMN1 (OMIM:600354; HGNC:11117) variant</p>	<p>radio</p> <table border="1"> <tr><td>homoz_del_ex7</td><td>Homozygous deletion of exon 7 of SMN1</td></tr> <tr><td>comp_heteroz_del_ex7</td><td>Compound heterozygous for a deletion of exon 7 and a point mutation in SMN1</td></tr> <tr><td>comp_heteroz_2sub</td><td>Compound heterozygous for two substitutions (point mutations) in SMN1</td></tr> <tr><td>homoz_1sub</td><td>Homozygous for a single substitution (point mutation) in SMN1 (rare, may occur in consanguinous families)</td></tr> <tr><td>none_of_above</td><td>None of the above</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	homoz_del_ex7	Homozygous deletion of exon 7 of SMN1	comp_heteroz_del_ex7	Compound heterozygous for a deletion of exon 7 and a point mutation in SMN1	comp_heteroz_2sub	Compound heterozygous for two substitutions (point mutations) in SMN1	homoz_1sub	Homozygous for a single substitution (point mutation) in SMN1 (rare, may occur in consanguinous families)	none_of_above	None of the above	unknown	Unknown
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461	<p>[gendia_smn1_variant_desc2]</p> <p>Show the field ONLY if: [gendia_smn1_variant_v1] = 'comp_heteroz_del_ex7'</p>	Please fill in the point mutation in variant 1 of "REPORTING CAUSATIVE VARIANT(S)" below.	descriptive												
462	<p>[gendia_smn1_variant_desc3]</p> <p>Show the field ONLY if: [gendia_smn1_variant_v1] = 'comp_heteroz_2sub'</p>	Please fill in both variants in "REPORTING CAUSATIVE VARIANT(S)" below.	descriptive												
463	<p>[gendia_smn1_variant_desc4]</p> <p>Show the field ONLY if: [gendia_smn1_variant_v1] = 'homoz_1sub'</p>	Please fill in same variant in both variant 1 and variant 2 in "REPORTING CAUSATIVE VARIANT(S)" below.	descriptive												
464	<p>[gendia_smn1_gen_build_v1]</p> <p>Show the field ONLY if: [gendia_known] = 'yes' AND ([arm-number]=5 OR [gendia_omim]=600354) AND ([gendia_smn1_variant_v1] = 'homoz_del_ex7' OR [gendia_smn1_variant_v1] = 'comp_heteroz_del_ex7' OR [gendia_smn1_variant_v1] = 'comp_heteroz_2sub' OR [gendia_smn1_variant_v1] = 'homoz_1sub')</p>	Genomic build	<p>dropdown</p> <table border="1"> <tr><td>GRCh38</td><td>GRCh38 (hg38)</td></tr> <tr><td>GRCh37</td><td>GRCh37 (hg19)</td></tr> </table>	GRCh38	GRCh38 (hg38)	GRCh37	GRCh37 (hg19)								
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465	<p>[ <a href="#">gendia_smn1_gen_app_v1</a> ]</p> <p>Show the field ONLY if:  [gendia_known] = 'yes' AND  ([arm-number]=5 OR [gendia_omim]=600354) AND ([gendia_smn1_variant_v1] = 'homozygous_deletion' OR [gendia_smn1_variant_v1] = 'heterozygous_deletion' OR [gendia_smn1_variant_v1] = 'heterozygous_insertion' OR [gendia_smn1_variant_v1] = 'homozygous_insertion')</p>	Genetic approach	<p>dropdown</p> <table border="1"> <tr><td>single_gene_screening</td><td>Single Gene Screening</td></tr> <tr><td>acgh</td><td>aCGH</td></tr> <tr><td>snp_array</td><td>SNP array</td></tr> <tr><td>mlpa</td><td>MLPA</td></tr> <tr><td>karyotype</td><td>Karyotype</td></tr> <tr><td>ngs_panel</td><td>NGS panel</td></tr> <tr><td>wes</td><td>WES</td></tr> <tr><td>wgs</td><td>WGS</td></tr> </table>	single_gene_screening	Single Gene Screening	acgh	aCGH	snp_array	SNP array	mlpa	MLPA	karyotype	Karyotype	ngs_panel	NGS panel	wes	WES	wgs	WGS																																		
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467	[ <a href="#">helpervar7_v2</a> ]	#SECTION DMD/BMD	<p>descriptive</p> <p>Field Annotation: @HIDDEN @HIDDEN-PDF</p>																																																		
468	<p>[ <a href="#">gendia_dmd_desc1</a> ]</p> <p>Show the field ONLY if:  [gendia_known] = 'yes' AND  [gendia_omim]='300377'</p>	<p>Section Header: <i>DMD (OMIM:300377; HGNC:2928; e.g. for diagnoses ORPHA:98895, ORPHA:98896, ORPHA:206546)</i></p> <p>If... the change is described in the NCBI ClinVar database OR you can describe the change in HGVS-simple notation OR there is a point mutation in the DMD-gene as well ...please fill the section "REPORTING CAUSATIVE VARIANT(S)" below. The LOVD-tool (accessed: 01.03.2022) may be helpful to convert exon deletions in the DMD gene to HGVS nomenclature.</p>	descriptive																																																		
469	<p>[ <a href="#">gendia_dmd_mutation_type</a> ]</p> <p>Show the field ONLY if:  [gendia_known] = 'yes' AND  [gendia_omim]='300377'</p>	Mutation type of DMD-gene (OMIM:300377; HGNC:2928)	<p>radio, Required</p> <table border="1"> <tr><td>deletion</td><td>Deletion of one or several exons</td></tr> <tr><td>duplication</td><td>Duplication of one or several exons</td></tr> <tr><td>triplication</td><td>Triplication of one or several exons</td></tr> <tr><td>not_present</td><td>Not present (none of the above)</td></tr> </table>	deletion	Deletion of one or several exons	duplication	Duplication of one or several exons	triplication	Triplication of one or several exons	not_present	Not present (none of the above)																																										
deletion	Deletion of one or several exons																																																				
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not_present	Not present (none of the above)																																																				

			unknown	Unknown
470	[ <a href="#">gendia_dmd_exon_del_first</a> ]  Show the field ONLY if: [gendia_dmd_mutation_type] = 'deletion'	Section Header: <i>Deletion</i> First deleted exon	text (integer), Required	
471	[ <a href="#">gendia_dmd_exon_del_last</a> ]  Show the field ONLY if: [gendia_dmd_mutation_type] = 'deletion'	Last deleted exon	text (integer), Required	
472	[ <a href="#">gendia_dmd_exon_del</a> ]  Show the field ONLY if: [gendia_dmd_mutation_type] = 'deletion'	gendia_dmd_exon_del	text Field Annotation: @CALCTEXT(if([gendia_dmd_mutation_type] = 'deletion', if([gendia_dmd_exon_del_first]=[gendia_dmd_exon_del_last], concat('del ex', [gendia_dmd_exon_del_first]), concat('del ex', [gendia_dmd_exon_del_first], '-', [gendia_dmd_exon_del_last])), '')) @READONLY @HIDDEN @HIDDEN-PDF	
473	[ <a href="#">exon_v_del_disp_v2</a> ]  Show the field ONLY if: [gendia_dmd_mutation_type] = 'deletion'	Resulting exon-based description of the variant: [gendia_dmd_exon_del] (see guidelines for curators on mutation entries in DMD registries, accessed 18.11.2021)	descriptive	
474	[ <a href="#">gendia_dmd_exon_dup_first</a> ]  Show the field ONLY if: [gendia_dmd_mutation_type] = 'duplication'	Section Header: <i>Duplication</i> First duplicated exon	text (integer), Required	
475	[ <a href="#">gendia_dmd_exon_dup_last</a> ]  Show the field ONLY if: [gendia_dmd_mutation_type] = 'duplication'	Last duplicated exon	text (integer), Required	
476	[ <a href="#">gendia_dmd_exon_dup</a> ]  Show the field ONLY if: [gendia_dmd_mutation_type] = 'duplication'	gendia_dmd_exon_dup	text Field Annotation: @CALCTEXT(if([gendia_dmd_mutation_type] = 'duplication', if([gendia_dmd_exon_dup_first]=[gendia_dmd_exon_dup_last], concat('dup ex', [gendia_dmd_exon_dup_first]), concat('dup ex', [gendia_dmd_exon_dup_first], '-', [gendia_dmd_exon_dup_last])), '')) @READONLY @HIDDEN @HIDDEN-PDF	
477	[ <a href="#">gendia_dmd_exon_dup_disp</a> ]  Show the field ONLY if: [gendia_dmd_mutation_type] = 'duplication'	Resulting exon-based description of the variant: [gendia_dmd_exon_dup] (see guidelines for curators on mutation entries in DMD registries, accessed 18.11.2021)	descriptive	
478	[ <a href="#">gendia_dmd_exon_tri_first</a> ]  Show the field ONLY if: [gendia_dmd_mutation_type] = 'triplication'	Section Header: <i>Triplication</i> First triplicated exon	text (integer), Required	
479	[ <a href="#">gendia_dmd_exon_tri_last</a> ]  Show the field ONLY if: [gendia_dmd_mutation_type] = 'triplication'	Last triplicated exon	text (integer), Required	
480	[ <a href="#">gendia_dmd_exon_tri</a> ]  Show the field ONLY if: [gendia_dmd_mutation_type] = 'triplication'	gendia_dmd_exon_tri	text Field Annotation: @CALCTEXT(if([gendia_dmd_mutation_type] = 'triplication', if([gendia_dmd_exon_tri_first]=[gendia_dmd_exon_tri_last], concat('tri ex',	

			[gendia_dmd_exon_tri_first]), concat('tri ex', [gendia_dmd_exon_tri_first], '-', [gendia_dmd_exon_tri_last]), '') @READONLY @HIDDEN @HIDDEN-PDF																																																		
481	[gendia_dmd_exon_tri_display] Show the field ONLY if: [gendia_dmd_mutation_type] = 'triplication'	Resulting exon-based description of the variant: [gendia_dmd_exon_tri] (see guidelines for curators on mutation entries in DMD registries, accessed 18.11.2021)	descriptive																																																		
482	[gendia_dmd_gen_build_v1] Show the field ONLY if: [gendia_dmd_mutation_type] = 'deletion' OR [gendia_dmd_mutation_type] = 'duplication' OR [gendia_dmd_mutation_type] = 'triplication'	Section Header: <i>Additional Information</i> Genomic build	dropdown <table border="1"> <tr> <td>GRCh38</td> <td>GRCh38 (hg38)</td> </tr> <tr> <td>GRCh37</td> <td>GRCh37 (hg19)</td> </tr> </table>	GRCh38	GRCh38 (hg38)	GRCh37	GRCh37 (hg19)																																														
GRCh38	GRCh38 (hg38)																																																				
GRCh37	GRCh37 (hg19)																																																				
483	[gendia_dmd_gen_app_v1] Show the field ONLY if: [gendia_dmd_mutation_type] = 'deletion' OR [gendia_dmd_mutation_type] = 'duplication' OR [gendia_dmd_mutation_type] = 'triplication'	Genetic approach	dropdown <table border="1"> <tr> <td>single_gene_screening</td> <td>Single Gene Screening</td> </tr> <tr> <td>acgh</td> <td>aCGH</td> </tr> <tr> <td>snp_array</td> <td>SNP array</td> </tr> <tr> <td>mlpa</td> <td>MLPA</td> </tr> <tr> <td>karyotype</td> <td>Karyotype</td> </tr> <tr> <td>ngs_panel</td> <td>NGS panel</td> </tr> <tr> <td>wes</td> <td>WES</td> </tr> <tr> <td>wgs</td> <td>WGS</td> </tr> </table>	single_gene_screening	Single Gene Screening	acgh	aCGH	snp_array	SNP array	mlpa	MLPA	karyotype	Karyotype	ngs_panel	NGS panel	wes	WES	wgs	WGS																																		
single_gene_screening	Single Gene Screening																																																				
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484	[gendia_dmd_chrom_v1] Show the field ONLY if: [gendia_dmd_mutation_type] = 'deletion' OR [gendia_dmd_mutation_type] = 'duplication' OR [gendia_dmd_mutation_type] = 'triplication'	Chromosome	dropdown <table border="1"> <tr><td>1</td><td>1</td></tr> <tr><td>2</td><td>2</td></tr> <tr><td>3</td><td>3</td></tr> <tr><td>4</td><td>4</td></tr> <tr><td>5</td><td>5</td></tr> <tr><td>6</td><td>6</td></tr> <tr><td>7</td><td>7</td></tr> <tr><td>8</td><td>8</td></tr> <tr><td>9</td><td>9</td></tr> <tr><td>10</td><td>10</td></tr> <tr><td>11</td><td>11</td></tr> <tr><td>12</td><td>12</td></tr> <tr><td>13</td><td>13</td></tr> <tr><td>14</td><td>14</td></tr> <tr><td>15</td><td>15</td></tr> <tr><td>16</td><td>16</td></tr> <tr><td>17</td><td>17</td></tr> <tr><td>18</td><td>18</td></tr> <tr><td>19</td><td>19</td></tr> <tr><td>20</td><td>20</td></tr> <tr><td>21</td><td>21</td></tr> <tr><td>22</td><td>22</td></tr> <tr><td>X</td><td>X</td></tr> <tr><td>Y</td><td>Y</td></tr> <tr><td>MT</td><td>MT</td></tr> </table>	1	1	2	2	3	3	4	4	5	5	6	6	7	7	8	8	9	9	10	10	11	11	12	12	13	13	14	14	15	15	16	16	17	17	18	18	19	19	20	20	21	21	22	22	X	X	Y	Y	MT	MT
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485	[gendia_dmd_zygos_v1]	Zygosity	dropdown																																																		

	Show the field ONLY if: [gendia_dmd_mutation_type ] = 'deletion' OR [gendia_dmd_mutation_type] = 'duplication' OR [gendia_dmd_mutation_type] = 'triplication'		<table border="1"> <tr> <td>heterozygous</td> <td>Heterozygous</td> </tr> <tr> <td>compound_heterozygous</td> <td>Compound heterozygous</td> </tr> <tr> <td>homozygous</td> <td>Homozygous</td> </tr> <tr> <td>hemizygous</td> <td>Hemizygous</td> </tr> <tr> <td>mosaic</td> <td>Mosaic</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	heterozygous	Heterozygous	compound_heterozygous	Compound heterozygous	homozygous	Homozygous	hemizygous	Hemizygous	mosaic	Mosaic	unknown	Unknown
heterozygous	Heterozygous														
compound_heterozygous	Compound heterozygous														
homozygous	Homozygous														
hemizygous	Hemizygous														
mosaic	Mosaic														
unknown	Unknown														
486	[gendia_desc2]  Show the field ONLY if: [gendia_known] = 'yes'	Section Header: <i>REPORTING CAUSATIVE VARIANT(S)</i>  Please note: You only need to fill in this section if a) if no data entry fields are shown in the above section "DISEASE-SPECIFIC GENETIC DETAILS". In this case, none of the more frequent variants for which we have already stored a correspondingly simplified data entry applies to this patient. b) if you want to report variant(s) in the selected gene (OMIM:[gendia_omim]) that are not covered by the variant(s) described under "DISEASE-SPECIFIC GENETIC DETAILS" above Please report only an additional variant for the gene selected above (OMIM:[gendia_omim]). If you wish to report a variant for another gene, please report that in a separate 'Genetic Diagnosis'-form for that gene. This section is intended to record the change(s) in DNA, RNA or protein sequences according to the standards of the Sequence Variant Nomenclature "HGVS simple" (accessed: 18.13.22). HGVS-codes can be checked with Mutalyzer (accessed: 18.03.22). Please enter all the details that you have available from the report. For autosomal recessive cases, you must enter both variant 1 and variant 2. If the variant is homozygous, enter the identical details twice. For autosomal dominant and X-linked, only one is required.	descriptive												
487	[gendia_report_causa_variant]  Show the field ONLY if: [gendia_known] = 'yes'	Report (additional) variant(s) / variant(s) not covered by the predefined variant(s) under "DISEASE-SPECIFIC GENETIC DETAILS" above	yesno, Required <table border="1"> <tr> <td>1</td> <td>Yes</td> </tr> <tr> <td>0</td> <td>No</td> </tr> </table>	1	Yes	0	No								
1	Yes														
0	No														
488	[gendia_v1_report]  Show the field ONLY if: [gendia_report_causa_variant] = 1	Section Header: <i>Variant 1</i>  How would you like to describe variant 1? Note: We prefer NCBI ClinVar but if the change in question is not yet described there, you can give a dbSNP ID if available, or you can type in the protein or cDNA change given in the genetic report.	radio, Required <table border="1"> <tr> <td>clinvar</td> <td>NCBI ClinVar</td> </tr> <tr> <td>dbSNP</td> <td>dbSNP</td> </tr> <tr> <td>custom_hgvs</td> <td>Custom HGVS-simple notation</td> </tr> </table>	clinvar	NCBI ClinVar	dbSNP	dbSNP	custom_hgvs	Custom HGVS-simple notation						
clinvar	NCBI ClinVar														
dbSNP	dbSNP														
custom_hgvs	Custom HGVS-simple notation														
489	[gendia_v1_clinvar_uri]  Show the field ONLY if: [gendia_v1_report] = 'clinvar'	Variant 1: NCBI ClinVar URI (please provide website-link) <i>If the change is described in the NCBI ClinVar database, please provide website-link (e.g. <a href="https://www.ncbi.nlm.nih.gov/clinvar/variation/4886/">https://www.ncbi.nlm.nih.gov/clinvar/variation/4886/</a>)</i>	text, Required												
490	[gendia_v1_dbSNP_id]  Show the field ONLY if: [gendia_v1_report] = 'dbSNP'	Variant 1: dbSNP RS ID <i>If the change has a dbSNP ID, enter it here, e.g. rs606231128</i>	text, Required												
491	[gendia_v1_hgvs_opts]  Show the field ONLY if: [gendia_v1_report] = 'custom_hgvs'	Variant 1: What kind of change(s) would you like to report in "HGVS simple"-notation? Please enter all the details that you have available from the report – e.g. if the report lists both the cDNA and protein changes, please provide both.	checkbox, Required <table border="1"> <tr> <td>cdna</td> <td>gendia_v1_hgvs_opts__cdna</td> <td>Report cDNA change (c.)</td> </tr> <tr> <td>nucleotide</td> <td>gendia_v1_hgvs_opts__nucleotide</td> <td>Report nucleotide change (g.)</td> </tr> <tr> <td>protein</td> <td>gendia_v1_hgvs_opts__protein</td> <td>Report protein change (p.)</td> </tr> <tr> <td>other</td> <td>gendia_v1_hgvs_opts__other</td> <td>Report other change (e.g. l.)</td> </tr> </table>	cdna	gendia_v1_hgvs_opts__cdna	Report cDNA change (c.)	nucleotide	gendia_v1_hgvs_opts__nucleotide	Report nucleotide change (g.)	protein	gendia_v1_hgvs_opts__protein	Report protein change (p.)	other	gendia_v1_hgvs_opts__other	Report other change (e.g. l.)
cdna	gendia_v1_hgvs_opts__cdna	Report cDNA change (c.)													
nucleotide	gendia_v1_hgvs_opts__nucleotide	Report nucleotide change (g.)													
protein	gendia_v1_hgvs_opts__protein	Report protein change (p.)													
other	gendia_v1_hgvs_opts__other	Report other change (e.g. l.)													

				(r.) non- codi
492	[ <a href="#">gendia_v1_refseq_c</a> ] Show the field ONLY if: [gendia_report_causa_varia nt]=1 AND [gendia_v1_hgvs_ opts(cdna)] = 1	Section Header: <i>Variant 1: cDNA change</i> Reference sequence for cDNA change <i>Enter the RefSeq transcript ID with the prefix NM_ or NR_ etc., e.g. 'NM_173660.5'. More information and examples see here and here</i>	text, Required	
493	[ <a href="#">gendia_v1_desc_c</a> ] Show the field ONLY if: [gendia_report_causa_varia nt]=1 AND [gendia_v1_hgvs_ opts(cdna)] = 1	Description of cDNA change <i>Enter the cDNA change in HGVS nomenclature, e.g. c.1124_1127dupTGCC or c.346G&gt;A More information and examples see here</i>	text, Required	
494	[ <a href="#">gendia_v1_hgvs_c</a> ] Show the field ONLY if: [gendia_report_causa_varia nt]=1 AND [gendia_v1_hgvs_ opts(cdna)] = 1	gendia_v1_hgvs_c	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_v1_refseq_c]!=" and [gendia_v1_desc_c]!=" , concat([gendia_v1_refseq_c], ':', [gendia_v1_desc_c], ''))	
495	[ <a href="#">gendia_v1_hgvs_c_disp</a> ] Show the field ONLY if: [gendia_report_causa_varia nt]=1 AND [gendia_v1_hgvs_ opts(cdna)] = 1	Resulting HGVS notation for variant 1 cDNA change: [gendia_v1_hgvs_c]	descriptive	
496	[ <a href="#">gendia_v1_refseq_g</a> ] Show the field ONLY if: [gendia_report_causa_varia nt]=1 AND [gendia_v1_hgvs_ opts(nucleotide)] = 1	Section Header: <i>Variant 1: Nucleotide (genomic) change</i> Reference sequence for nucleotide (genomic) change <i>Enter the RefSeq transcript ID with the prefix NC_ or NG_ etc., e.g. 'NC_000023.10'. More information and examples see here and here</i>	text, Required	
497	[ <a href="#">gendia_v1_desc_g</a> ] Show the field ONLY if: [gendia_report_causa_varia nt]=1 AND [gendia_v1_hgvs_ opts(nucleotide)] = 1	Description of nucleotide (genomic) change <i>Enter the nucleotide change in HGVS nomenclature, e.g. g.34805_34808dup More information and examples see here</i>	text, Required	
498	[ <a href="#">gendia_v1_hgvs_g</a> ] Show the field ONLY if: [gendia_report_causa_varia nt]=1 AND [gendia_v1_hgvs_ opts(nucleotide)] = 1	gendia_v1_hgvs_g	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_v1_refseq_g]!=" and [gendia_v1_desc_g]!=" , concat([gendia_v1_refseq_g], ':', [gendia_v1_desc_g], ''))	
499	[ <a href="#">gendia_v1_hgvs_g_disp</a> ] Show the field ONLY if: [gendia_report_causa_varia nt]=1 AND [gendia_v1_hgvs_ opts(nucleotide)] = 1	Resulting HGVS notation for variant 1 nucleotide (genomic) change:[gendia_v1_hgvs_g]	descriptive	
500	[ <a href="#">gendia_v1_refseq_p</a> ] Show the field ONLY if: [gendia_report_causa_varia nt]=1 AND [gendia_v1_hgvs_ opts(protein)] = 1	Section Header: <i>Variant 1: Protein change</i> Reference sequence for protein change <i>Enter the RefSeq transcript ID with the prefix NP_ , e.g. NP_003997.1 More information and examples see here and here</i>	text, Required	
501	[ <a href="#">gendia_v1_desc_p</a> ] Show the field ONLY if: [gendia_report_causa_varia nt]=1 AND [gendia_v1_hgvs_ opts(protein)] = 1	Description of protein change <i>Enter the protein change in HGVS nomenclature, e.g. p.Ala378fs More information and examples see here</i>	text, Required	
502	[ <a href="#">gendia_v1_hgvs_p</a> ] Show the field ONLY if: [gendia_report_causa_varia nt]=1 AND [gendia_v1_hgvs_ opts(protein)] = 1	gendia_v1_hgvs_p	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_v1_refseq_p]!=" and [gendia_v1_desc_p]!=" , concat([gendia_v1_refseq_p], ':', [gendia_v1_desc_p], ''))	
503	[ <a href="#">gendia_v1_hgvs_p_disp</a> ] Show the field ONLY if: [gendia_report_causa_varia nt]=1 AND [gendia_v1_hgvs_ opts(protein)] = 1	Resulting HGVS notation for variant 1 protein change: [gendia_v1_hgvs_p]	descriptive	

	Show the field ONLY if: [genda_report_causa_variant]=1 AND [genda_v1_hgvs_opts(protein)] = 1																		
504	[genda_v1_refseq_o]  Show the field ONLY if: [genda_report_causa_variant]=1 AND [genda_v1_hgvs_opts(other)] = 1	Section Header: <i>Variant 1: Other change</i>  Reference sequence for other change <i>Examples for RNA reference sequences: NC_000023.10(NM_004006.2) or LRG_199t1. More information and examples see here and here</i>	text, Required																
505	[genda_v1_desc_o]  Show the field ONLY if: [genda_report_causa_variant]=1 AND [genda_v1_hgvs_opts(other)] = 1	Description of other change <i>Examples for descriptions of RNA changes: r.357_358ins357+1_357+12 or r.11u&gt;g More information and examples see here</i>	text, Required																
506	[genda_v1_hgvs_o]  Show the field ONLY if: [genda_report_causa_variant]=1 AND [genda_v1_hgvs_opts(other)] = 1	genda_v1_hgvs_o	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([genda_v1_refseq_o]!=" and [genda_v1_desc_o]!=", concat([genda_v1_refseq_o], ':', [genda_v1_desc_o]), ''))																
507	[genda_v1_hgvs_o_disp]  Show the field ONLY if: [genda_report_causa_variant]=1 AND [genda_v1_hgvs_opts(other)] = 1	Resulting HGVS notation for variant 1 other change: [genda_v1_hgvs_o]	descriptive																
508	[genda_v1_zygos]  Show the field ONLY if: [genda_known] = 'yes' AND [genda_report_causa_variant] = 1	Section Header: <i>Variant 1: Additional Information</i>  Zygoty	dropdown <table border="1"> <tr><td>heterozygous</td><td>Heterozygous</td></tr> <tr><td>compound_heterozygous</td><td>Compound heterozygous</td></tr> <tr><td>homozygous</td><td>Homozygous</td></tr> <tr><td>hemizygous</td><td>Hemizygous</td></tr> <tr><td>mosaic</td><td>Mosaic</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	heterozygous	Heterozygous	compound_heterozygous	Compound heterozygous	homozygous	Homozygous	hemizygous	Hemizygous	mosaic	Mosaic	unknown	Unknown				
heterozygous	Heterozygous																		
compound_heterozygous	Compound heterozygous																		
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hemizygous	Hemizygous																		
mosaic	Mosaic																		
unknown	Unknown																		
509	[genda_v1_gen_build]  Show the field ONLY if: [genda_known] = 'yes' AND [genda_report_causa_variant] = 1 AND [genda_v1_repo] = 'custom_hgvs'	Genomic build	dropdown <table border="1"> <tr><td>GRCh38</td><td>GRCh38 (hg38)</td></tr> <tr><td>GRCh37</td><td>GRCh37 (hg19)</td></tr> </table>	GRCh38	GRCh38 (hg38)	GRCh37	GRCh37 (hg19)												
GRCh38	GRCh38 (hg38)																		
GRCh37	GRCh37 (hg19)																		
510	[genda_v1_gen_app]  Show the field ONLY if: [genda_known] = 'yes' AND [genda_report_causa_variant] = 1 AND [genda_v1_repo] = 'custom_hgvs'	Genetic approach	dropdown <table border="1"> <tr><td>single_gene_screening</td><td>Single Gene Screening</td></tr> <tr><td>acgh</td><td>aCGH</td></tr> <tr><td>snp_array</td><td>SNP array</td></tr> <tr><td>mlpa</td><td>MLPA</td></tr> <tr><td>karyotype</td><td>Karyotype</td></tr> <tr><td>ngs_panel</td><td>NGS panel</td></tr> <tr><td>wes</td><td>WES</td></tr> <tr><td>wgs</td><td>WGS</td></tr> </table>	single_gene_screening	Single Gene Screening	acgh	aCGH	snp_array	SNP array	mlpa	MLPA	karyotype	Karyotype	ngs_panel	NGS panel	wes	WES	wgs	WGS
single_gene_screening	Single Gene Screening																		
acgh	aCGH																		
snp_array	SNP array																		
mlpa	MLPA																		
karyotype	Karyotype																		
ngs_panel	NGS panel																		
wes	WES																		
wgs	WGS																		
511	[genda_v1_chrom]  Show the field ONLY if: [genda_known] = 'yes' AND [genda_report_causa_variant] = 1 AND [genda_v1_repo] = 'custom_hgvs'	Chromosome	dropdown <table border="1"> <tr><td>1</td><td>1</td></tr> <tr><td>2</td><td>2</td></tr> <tr><td>3</td><td>3</td></tr> <tr><td>4</td><td>4</td></tr> <tr><td>5</td><td>5</td></tr> <tr><td>6</td><td>6</td></tr> <tr><td>7</td><td>7</td></tr> </table>	1	1	2	2	3	3	4	4	5	5	6	6	7	7		
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4	4																		
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7	7																		

			<table border="1"> <tr><td>8</td><td>8</td></tr> <tr><td>9</td><td>9</td></tr> <tr><td>10</td><td>10</td></tr> <tr><td>11</td><td>11</td></tr> <tr><td>12</td><td>12</td></tr> <tr><td>13</td><td>13</td></tr> <tr><td>14</td><td>14</td></tr> <tr><td>15</td><td>15</td></tr> <tr><td>16</td><td>16</td></tr> <tr><td>17</td><td>17</td></tr> <tr><td>18</td><td>18</td></tr> <tr><td>19</td><td>19</td></tr> <tr><td>20</td><td>20</td></tr> <tr><td>21</td><td>21</td></tr> <tr><td>22</td><td>22</td></tr> <tr><td>X</td><td>X</td></tr> <tr><td>Y</td><td>Y</td></tr> <tr><td>MT</td><td>MT</td></tr> </table>	8	8	9	9	10	10	11	11	12	12	13	13	14	14	15	15	16	16	17	17	18	18	19	19	20	20	21	21	22	22	X	X	Y	Y	MT	MT
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X	X																																						
Y	Y																																						
MT	MT																																						
512	<p><b>[gendia_desc3]</b></p> <p>Show the field ONLY if: [gendia_known] = 'yes' AND [gendia_report_causa_variant] = 1</p>	<p>Section Header: <i>Variant 2 (if applicable)</i></p> <p>Please report only an additional variant for the gene selected above (OMIM:[gendia_omim]). If you wish to report a variant for another gene, please report that in a separate 'Genetic Diagnosis'-form for that gene. Please enter all the details that you have available from the report. For autosomal recessive cases, you must enter both variant 1 and variant 2. If the variant is homozygous, enter the identical details twice. For autosomal dominant and X-linked, only one is required.</p>	descriptive																																				
513	<p><b>[gendia_v2_yes]</b></p> <p>Show the field ONLY if: [gendia_known] = 'yes' AND [gendia_report_causa_variant] = 1</p>	Report variant 2	<p>yesno</p> <table border="1"> <tr><td>1</td><td>Yes</td></tr> <tr><td>0</td><td>No</td></tr> </table>	1	Yes	0	No																																
1	Yes																																						
0	No																																						
514	<p><b>[gendia_v2_report]</b></p> <p>Show the field ONLY if: [gendia_v2_yes] = 1</p>	How would you like to describe variant 2? Note: We prefer NCBI ClinVar but if the change in question is not yet described there, you can give a dbSNP ID if available, or you can type in the protein or cDNA change given in the genetic report.	<p>radio, Required</p> <table border="1"> <tr><td>clinvar</td><td>NCBI ClinVar</td></tr> <tr><td>dbSNP</td><td>dbSNP</td></tr> <tr><td>custom_hgvs</td><td>Custom HGVS-simple notation</td></tr> </table>	clinvar	NCBI ClinVar	dbSNP	dbSNP	custom_hgvs	Custom HGVS-simple notation																														
clinvar	NCBI ClinVar																																						
dbSNP	dbSNP																																						
custom_hgvs	Custom HGVS-simple notation																																						
515	<p><b>[gendia_v2_clinvar_uri]</b></p> <p>Show the field ONLY if: [gendia_v2_report] = 'clinvar'</p>	<p>Variant 2: NCBI ClinVar URI (please provide website-link)</p> <p><i>If the change is described in the NCBI ClinVar database, please provide website-link (e.g. <a href="https://www.ncbi.nlm.nih.gov/clinvar/variation/4886/">https://www.ncbi.nlm.nih.gov/clinvar/variation/4886/</a>)</i></p>	text, Required																																				
516	<p><b>[gendia_v2_dbSNP_id]</b></p> <p>Show the field ONLY if: [gendia_v2_report] = 'dbSNP'</p>	<p>Variant 2: dbSNP RS ID</p> <p><i>If the change has a dbSNP ID, enter it here, e.g. rs606231128</i></p>	text, Required																																				
517	<p><b>[gendia_v2_hgvs_opts]</b></p> <p>Show the field ONLY if: [gendia_v2_report] = 'custom_hgvs'</p>	Variant 2: What kind of change(s) would you like to report in "HGVS simple"-notation? Please enter all the details that you have available from the report – e.g. if the report lists both the cDNA and protein changes, please provide both.	<p>checkbox, Required</p> <table border="1"> <tr> <td>cdna</td> <td>gendia_v2_hgvs_opts__cdna</td> <td>Report cDN char (c.)</td> </tr> <tr> <td>nucleotide</td> <td>gendia_v2_hgvs_opts__nucleotide</td> <td>Report nucleic acid char (g.)</td> </tr> <tr> <td>protein</td> <td>gendia_v2_hgvs_opts__protein</td> <td>Report protein char</td> </tr> </table>	cdna	gendia_v2_hgvs_opts__cdna	Report cDN char (c.)	nucleotide	gendia_v2_hgvs_opts__nucleotide	Report nucleic acid char (g.)	protein	gendia_v2_hgvs_opts__protein	Report protein char																											
cdna	gendia_v2_hgvs_opts__cdna	Report cDN char (c.)																																					
nucleotide	gendia_v2_hgvs_opts__nucleotide	Report nucleic acid char (g.)																																					
protein	gendia_v2_hgvs_opts__protein	Report protein char																																					

				(p.)
			other	gendia_v2_hgvs_opts__other
				Rebo othe of ch e.g. l (r.) o non- codi
518	[ gendia_v2_refseq_c ] Show the field ONLY if: [gendia_report_causa_varia nt]=1 AND [gendia_v2_hgvs_ opts(cdna)] = 1	Section Header: <i>Variant 2: cDNA change</i> Reference sequence for cDNA change <i>Enter the RefSeq transcript ID with the prefix NM_ or NR_ etc., e.g. 'NM_173660.5'. More information and examples see here and here</i>	text, Required	
519	[ gendia_v2_desc_c ] Show the field ONLY if: [gendia_report_causa_varia nt]=1 AND [gendia_v2_hgvs_ opts(cdna)] = 1	Description of cDNA change <i>Enter the cDNA change in HGVS nomenclature, e.g. c.1124_1127dupTGCC or c.346G&gt;A More information and examples see here</i>	text, Required	
520	[ gendia_v2_hgvs_c ] Show the field ONLY if: [gendia_report_causa_varia nt]=1 AND [gendia_v2_hgvs_ opts(cdna)] = 1	gendia_v1_hgvs_c	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_v2_refseq_c]!=" and [gendia_v2_desc_c]!=" , concat([gendia_v2_refseq_c], ';', [gendia_v2_desc_c], ''))	
521	[ gendia_v2_hgvs_c_disp ] Show the field ONLY if: [gendia_report_causa_varia nt]=1 AND [gendia_v2_hgvs_ opts(cdna)] = 1	Resulting HGVS notation for variant 2 cDNA change: [gendia_v2_hgvs_c]	descriptive	
522	[ gendia_v2_refseq_g ] Show the field ONLY if: [gendia_report_causa_varia nt]=1 AND [gendia_v2_hgvs_ opts(nucleotide)] = 1	Section Header: <i>Variant 2: Nucleotide (genomic) change</i> Reference sequence for nucleotide (genomic) change <i>Enter the RefSeq transcript ID with the prefix NC_ or NG_ etc., e.g. 'NC_000023.10'. More information and examples see here and here</i>	text, Required	
523	[ gendia_v2_desc_g ] Show the field ONLY if: [gendia_report_causa_varia nt]=1 AND [gendia_v2_hgvs_ opts(nucleotide)] = 1	Description of nucleotide (genomic) change <i>Enter the nucleotide change in HGVS nomenclature, e.g. g.34805_34808dup More information and examples see here</i>	text, Required	
524	[ gendia_v2_hgvs_g ] Show the field ONLY if: [gendia_report_causa_varia nt]=1 AND [gendia_v2_hgvs_ opts(nucleotide)] = 1	gendia_v2_hgvs_g	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_v2_refseq_g]!=" and [gendia_v2_desc_g]!=" , concat([gendia_v2_refseq_g], ';', [gendia_v2_desc_g], ''))	
525	[ gendia_v2_hgvs_g_disp ] Show the field ONLY if: [gendia_report_causa_varia nt]=1 AND [gendia_v2_hgvs_ opts(nucleotide)] = 1	Resulting HGVS notation for variant 2 nucleotide (genomic) change:[gendia_v2_hgvs_g]	descriptive	
526	[ gendia_v2_refseq_p ] Show the field ONLY if: [gendia_report_causa_varia nt]=1 AND [gendia_v2_hgvs_ opts(protein)] = 1	Section Header: <i>Variant 2: Protein change</i> Reference sequence for protein change <i>Enter the RefSeq transcript ID with the prefix NP_ , e.g. NP_003997.1 More information and examples see here and here</i>	text, Required	
527	[ gendia_v2_desc_p ] Show the field ONLY if: [gendia_report_causa_varia nt]=1 AND [gendia_v2_hgvs_ opts(protein)] = 1	Description of protein change <i>Enter the protein change in HGVS nomenclature, e.g. p.Ala378fs More information and examples see here</i>	text, Required	
528	[ gendia_v2_hgvs_p ] Show the field ONLY if: [gendia_report_causa_varia	gendia_v2_hgvs_p	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_v2_refseq_p]!=" and	



	nt]=1 AND [gendia_v2_hgvs_opts(protein)] = 1		[gendia_v2_desc_p]!="", concat([gendia_v2_refseq_p], ':', [gendia_v2_desc_p]), '')																
529	[gendia_v2_hgvs_p_disp] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v2_hgvs_opts(protein)] = 1	Resulting HGVS notation for variant 2 protein change: [gendia_v2_hgvs_p]	descriptive																
530	[gendia_v2_refseq_o] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v2_hgvs_opts(other)] = 1	Section Header: <i>Variant 2: Other change</i> Reference sequence for other change <i>Examples for RNA reference sequences: NC_000023.10(NM_004006.2) or LRG_199t1. More information and examples see here and here</i>	text, Required																
531	[gendia_v2_desc_o] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v2_hgvs_opts(other)] = 1	Description of other change <i>Examples for descriptions of RNA changes: r.357_358ins357+1_357+12 or r.11u&gt;g More information and examples see here</i>	text, Required																
532	[gendia_v2_hgvs_o] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v2_hgvs_opts(other)] = 1	gendia_v2_hgvs_o	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_v2_refseq_o]!=" and [gendia_v2_desc_o]!="", concat([gendia_v2_refseq_o], ':', [gendia_v2_desc_o]), ''))																
533	[gendia_v2_hgvs_o_disp] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v2_hgvs_opts(other)] = 1	Resulting HGVS notation for variant 1 other change: [gendia_v2_hgvs_o]	descriptive																
534	[gendia_v2_zygos] Show the field ONLY if: [gendia_known] = 'yes' AND [gendia_report_causa_variant] = 1 AND [gendia_v2_yes] = 1	Section Header: <i>Variant 2: Additional Information</i> Zygoty	dropdown <table border="1"> <tr><td>heterozygous</td><td>Heterozygous</td></tr> <tr><td>compound_heterozygous</td><td>Compound heterozygous</td></tr> <tr><td>homozygous</td><td>Homozygous</td></tr> <tr><td>hemizygous</td><td>Hemizygous</td></tr> <tr><td>mosaic</td><td>Mosaic</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	heterozygous	Heterozygous	compound_heterozygous	Compound heterozygous	homozygous	Homozygous	hemizygous	Hemizygous	mosaic	Mosaic	unknown	Unknown				
heterozygous	Heterozygous																		
compound_heterozygous	Compound heterozygous																		
homozygous	Homozygous																		
hemizygous	Hemizygous																		
mosaic	Mosaic																		
unknown	Unknown																		
535	[gendia_v2_gen_build] Show the field ONLY if: [gendia_known] = 'yes' AND [gendia_report_causa_variant] = 1 AND [gendia_v2_repo] = 'custom_hgvs'	Genomic build	dropdown <table border="1"> <tr><td>GRCh38</td><td>GRCh38 (hg38)</td></tr> <tr><td>GRCh37</td><td>GRCh37 (hg19)</td></tr> </table>	GRCh38	GRCh38 (hg38)	GRCh37	GRCh37 (hg19)												
GRCh38	GRCh38 (hg38)																		
GRCh37	GRCh37 (hg19)																		
536	[gendia_v2_gen_app] Show the field ONLY if: [gendia_known] = 'yes' AND [gendia_report_causa_variant] = 1 AND [gendia_v2_repo] = 'custom_hgvs'	Genetic approach	dropdown <table border="1"> <tr><td>single_gene_screening</td><td>Single Gene Screening</td></tr> <tr><td>acgh</td><td>aCGH</td></tr> <tr><td>snp_array</td><td>SNP array</td></tr> <tr><td>mlpa</td><td>MLPA</td></tr> <tr><td>karyotype</td><td>Karyotype</td></tr> <tr><td>ngs_panel</td><td>NGS panel</td></tr> <tr><td>wes</td><td>WES</td></tr> <tr><td>wgs</td><td>WGS</td></tr> </table>	single_gene_screening	Single Gene Screening	acgh	aCGH	snp_array	SNP array	mlpa	MLPA	karyotype	Karyotype	ngs_panel	NGS panel	wes	WES	wgs	WGS
single_gene_screening	Single Gene Screening																		
acgh	aCGH																		
snp_array	SNP array																		
mlpa	MLPA																		
karyotype	Karyotype																		
ngs_panel	NGS panel																		
wes	WES																		
wgs	WGS																		
537	[gendia_v2_chrom] Show the field ONLY if: [gendia_known] = 'yes' AND [gendia_report_causa_variant] = 1 AND [gendia_v2_repo] = 'custom_hgvs'	Chromosome	dropdown <table border="1"> <tr><td>1</td><td>1</td></tr> <tr><td>2</td><td>2</td></tr> <tr><td>3</td><td>3</td></tr> <tr><td>4</td><td>4</td></tr> </table>	1	1	2	2	3	3	4	4								
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			16	16
			17	17
			18	18
			19	19
			20	20
			21	21
			22	22
			X	X
			Y	Y
			MT	MT

538	[genetic_diagnosis_changes_in_nuclear_dna_complete]	Section Header: Form Status Complete?	dropdown	
			0	Incomplete
			1	Unverified
			2	Complete

**Instrument: Genetic Diagnosis Changes in mitochondrial DNA (genetic\_diagnosis\_changes\_in\_mitochondrial\_dna)**

Active languages: None

539	[mtdna_h1]	Genetic Diagnosis: Changes in mitochondrial DNA	descriptive	
540	[date_of_mtdna]	Date of assessment yyyy-mm-dd	text (date_ymd), Required	
541	[mtdna_desc1]	Please note: this form is only for recording changes in mitochondrial DNA (mtDNA). If you wish to report nuclear DNA variant(s), please use the "Genetic Diagnosis" form.	descriptive	
542	[mtdna_label]	Genetic diagnosis name <i>This is a label to identify this record to the registry user. It will be displayed on the patient's record home page together with the date of assessment. You can name it anything you find helpful.</i>	text, Required	
543	[mtdna_label_disp]	mtdna_label_disp	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(concat([mtdna_label], '(', [date_of_mtdna], ')'))	
544	[mtdna_change_type_known]	Type of mtDNA change known	radio	
			yes	Yes
			no	No
			not_present	Not present
			Field Annotation: @DEFAULT = 'yes'	
545	[mtdna_change_type]	Type of mtDNA change	checkbox, Required	
	Show the field ONLY if: [mtdna_change_type_known] = 'yes'		common_del	mtdna_change_type__common_del
			multiple_del	mtdna_change_type__multiple_del
			depletion	mtdna_change_type__depletion

546	[mtdna_cnv]	Section Header: <i>mtDNA copy number variation</i> Copy number variation in mtDNA	radio <table border="1"> <tr><td>yes</td><td>Yes</td></tr> <tr><td>not_present</td><td>Not present</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	yes	Yes	not_present	Not present	unknown	Unknown			
yes	Yes											
not_present	Not present											
unknown	Unknown											
547	[mtdna_cnv_where] Show the field ONLY if: [mtdna_cnv] = 'yes'	Copy number variation in mtDNA detected	checkbox <table border="1"> <tr><td>muscle</td><td>mtdna_cnv_where__muscle</td><td>in muscle</td></tr> <tr><td>blood</td><td>mtdna_cnv_where__blood</td><td>in blood</td></tr> <tr><td>other</td><td>mtdna_cnv_where__other</td><td>other tissue</td></tr> </table>	muscle	mtdna_cnv_where__muscle	in muscle	blood	mtdna_cnv_where__blood	in blood	other	mtdna_cnv_where__other	other tissue
muscle	mtdna_cnv_where__muscle	in muscle										
blood	mtdna_cnv_where__blood	in blood										
other	mtdna_cnv_where__other	other tissue										
548	[mtdna_cnv_plasmic] Show the field ONLY if: [mtdna_cnv] = 'yes'	State of copy number variation in mtDNA	radio <table border="1"> <tr><td>homoplasmic</td><td>Homoplasmic</td></tr> <tr><td>heteroplasmic</td><td>Heteroplasmic</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	homoplasmic	Homoplasmic	heteroplasmic	Heteroplasmic	unknown	Unknown			
homoplasmic	Homoplasmic											
heteroplasmic	Heteroplasmic											
unknown	Unknown											
549	[mtdna_snp]	Section Header: <i>mtDNA SNP variant</i> mtDNA SNP variant	radio <table border="1"> <tr><td>yes</td><td>Yes</td></tr> <tr><td>not_present</td><td>Not present</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	yes	Yes	not_present	Not present	unknown	Unknown			
yes	Yes											
not_present	Not present											
unknown	Unknown											
550	[mtdna_snp_where] Show the field ONLY if: [mtdna_snp] = 'yes'	mtDNA SNP variant detected	checkbox <table border="1"> <tr><td>muscle</td><td>mtdna_snp_where__muscle</td><td>in muscle</td></tr> <tr><td>blood</td><td>mtdna_snp_where__blood</td><td>in blood</td></tr> <tr><td>other</td><td>mtdna_snp_where__other</td><td>other tissue</td></tr> </table>	muscle	mtdna_snp_where__muscle	in muscle	blood	mtdna_snp_where__blood	in blood	other	mtdna_snp_where__other	other tissue
muscle	mtdna_snp_where__muscle	in muscle										
blood	mtdna_snp_where__blood	in blood										
other	mtdna_snp_where__other	other tissue										
551	[mtdna_snp_plasmic] Show the field ONLY if: [mtdna_snp] = 'yes'	State of mtDNA SNP variant	radio <table border="1"> <tr><td>homoplasmic</td><td>Homoplasmic</td></tr> <tr><td>heteroplasmic</td><td>Heteroplasmic</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	homoplasmic	Homoplasmic	heteroplasmic	Heteroplasmic	unknown	Unknown			
homoplasmic	Homoplasmic											
heteroplasmic	Heteroplasmic											
unknown	Unknown											
552	[mtdna_snp_report] Show the field ONLY if: [mtdna_snp] = 'yes'	How would you like to describe this mtDNA SNP variant? Note: We prefer NCBI ClinVar but if the change in question is not yet described there, you can type in the mtDNA change given in the genetic report.	radio, Required <table border="1"> <tr><td>clinvar</td><td>NCBI ClinVar</td></tr> <tr><td>custom_hgvs</td><td>Custom HGVS-simple notation</td></tr> </table>	clinvar	NCBI ClinVar	custom_hgvs	Custom HGVS-simple notation					
clinvar	NCBI ClinVar											
custom_hgvs	Custom HGVS-simple notation											
553	[mtdna_clinvar_uri] Show the field ONLY if: [mtdna_snp_report] = 'clinvar'	mtDNA variant: NCBI ClinVar URI(please provide website-link) <i>If the change is described in the NCBI ClinVar database, please provide website-link (e.g. <a href="https://www.ncbi.nlm.nih.gov/clinvar/variation/689805/">https://www.ncbi.nlm.nih.gov/clinvar/variation/689805/</a>)</i>	text, Required									
554	[mtdna_variant] Show the field ONLY if: [mtdna_snp_report] = 'custom_hgvs'	Description of mtDNA change based on the Revised Cambridge Reference Sequence (rCRS) of the Human Mitochondrial DNA: NC_012920.1(MT-CYB) <i>Enter the mtDNA change in HGVS nomenclature, e.g. m.578T&gt;C More information and examples see here</i>	text, Required									
555	[mtdna_hgvs] Show the field ONLY if: [mtdna_snp_report] = 'custom_hgvs'	mtdna_hgvs	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([mtdna_variant]!="", concat('NC_012920.1', ':', [mtdna_variant]), ''))									
556	[mtdna_hgvs_disp] Show the field ONLY if: [mtdna_snp_report] = 'custom_hgvs'	Resulting HGVS notation for mtDNA change:[mtdna_hgvs]	descriptive									
557	[genetic_diagnosis_changes_in_mitochondrial_dna_complete]	Section Header: <i>Form Status</i> Complete?	dropdown <table border="1"> <tr><td>0</td><td>Incomplete</td></tr> <tr><td>1</td><td>Unverified</td></tr> <tr><td>2</td><td>Complete</td></tr> </table>	0	Incomplete	1	Unverified	2	Complete			
0	Incomplete											
1	Unverified											
2	Complete											

Instrument: **Negative Genetic Tests (only when no genetic diagnosis)** (negative\_genetic\_tests\_only\_when\_no\_genetic\_diagno)

Active languages: None

558	[ <a href="#">neggentests_h1</a> ]	Negative Genetic Tests (only when no genetic diagnosis)	descriptive						
559	[ <a href="#">gps_done</a> ]	Section Header: <i>Gene Panel Sequencing (GPS)</i> Gene Panel Sequencing (GPS) done	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
560	[ <a href="#">date_of_gps</a> ] Show the field ONLY if: [ <a href="#">gps_done</a> ] = 'yes'	Date of Gene Panel Sequencing (GPS) yyyy-mm-dd <i>If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.</i>	text (date_ymd)						
561	[ <a href="#">gps_panel_name</a> ] Show the field ONLY if: [ <a href="#">gps_done</a> ] = 'yes'	Name of Gene Panel	text						
562	[ <a href="#">gps_n_excluded</a> ] Show the field ONLY if: [ <a href="#">gps_done</a> ] = 'yes'	Number of Genes excluded by GPS	text (integer)						
563	[ <a href="#">wes_done</a> ]	Section Header: <i>Whole Exome Sequencing (WES)</i> Whole Exome Sequencing (WES) done	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
564	[ <a href="#">date_of_wes</a> ] Show the field ONLY if: [ <a href="#">wes_done</a> ] = 'yes'	Date of Whole Exome Sequencing (WES) yyyy-mm-dd <i>If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.</i>	text (date_ymd)						
565	[ <a href="#">wgs_done</a> ]	Section Header: <i>Whole-Genome Sequencing (WGS)</i> Whole-Genome Sequencing (WGS) done	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	yes	Yes	no	No	unknown	Unknown
yes	Yes								
no	No								
unknown	Unknown								
566	[ <a href="#">date_of_wgs</a> ] Show the field ONLY if: [ <a href="#">wgs_done</a> ] = 'yes'	Date of Whole-Genome Sequencing (WGS) yyyy-mm-dd <i>If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.</i>	text (date_ymd)						
567	[ <a href="#">neggentests_comment</a> ]	Section Header: <i>Comments</i> Comments about negative genetic tests	notes						
568	[ <a href="#">gps_label_disp</a> ]	gps_label_disp	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([ <a href="#">gps_done</a> ]), concat('GPS: ', [ <a href="#">gps_done</a> ], if(!isblankormissingcode([ <a href="#">date_of_gps</a> ]), concat(' (', [ <a href="#">date_of_gps</a> ], ')', ')', ')', ')'))						
569	[ <a href="#">wes_label_disp</a> ]	wes_label_disp	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([ <a href="#">wes_done</a> ]), concat('WES: ', [ <a href="#">wes_done</a> ], if(!isblankormissingcode([ <a href="#">date_of_wes</a> ]), concat(' (', [ <a href="#">date_of_wes</a> ], ')', ')', ')', ')'))						
570	[ <a href="#">wgs_label_disp</a> ]	wgs_label_disp	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([ <a href="#">wgs_done</a> ]), concat('WGS: ', [ <a href="#">wgs_done</a> ], if(!isblankormissingcode([ <a href="#">date_of_wgs</a> ]), concat(' (', [ <a href="#">date_of_wgs</a> ], ')', ')', ')', ')'))						
571	[ <a href="#">ngt_label_disp</a> ]	ngt_label_disp	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(concat(if(!isblankormissingcode([ <a href="#">gps_label_disp</a> ], 'GPS: missing'), ';', if(!isblankormissingcode([ <a href="#">wes_label_disp</a> ], [ <a href="#">wes_label_disp</a> ], 'WES: missing'), ';', if(!isblankormissingcode([ <a href="#">wgs_label_disp</a> ], [ <a href="#">wgs_label_disp</a> ], 'WGS: missing'))						
572	[ <a href="#">negative_genetic_tests_only_when_no_genetic_d</a> ]	Section Header: <i>Form Status</i> Complete?	dropdown <table border="1"> <tr> <td>0</td> <td>Incomplete</td> </tr> </table>	0	Incomplete				
0	Incomplete								

	[iagno_complete]		<table border="1"> <tr> <td>1</td> <td>Unverified</td> </tr> <tr> <td>2</td> <td>Complete</td> </tr> </table>	1	Unverified	2	Complete				
1	Unverified										
2	Complete										
<b>Instrument: Undiagnosed (undiagnosed)</b>											
Active languages: None											
573	[undiag_h1]	UNDIAGNOSED	descriptive								
574	[undiag_desc1]	For undiagnosed patients: Please enter a combination of a phenotype and a genetic variant that cannot be definitively diagnosed.	descriptive								
575	[date_of_undiag]	Date of assessment yyyy-mm-dd	text (date_ymd), Required								
576	[date_of_undiag_2]	helper-field for generation of triples. contains same value as date_of_undiag but will only be filled if a second variant is reported.	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([undiag_v2_yes]='yes', [date_of_undiag], ''))								
577	[undiag_label]	Label for findings <i>This is a label to identify this record to the registry user. It will be displayed on the patient's record home page together with the date of assessment. You can name it anything you find helpful.</i>	text, Required								
578	[undiag_label_disp]	undiag_label_disp	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(concat([undiag_label], '(', [date_of_undiag], ''))								
579	[undiag_pheno_hpo]	Section Header: PHENOTYPE Phenotype: HPO-code <i>Element 6.3. in the Set of common data elements for Rare Diseases Registration (link)</i>	text, Required <table border="1"> <tr> <td>BIOPORTAL:HP</td> <td>BIOPORTAL:HP</td> </tr> </table>	BIOPORTAL:HP	BIOPORTAL:HP						
BIOPORTAL:HP	BIOPORTAL:HP										
580	[undiag_pheno_hpo_2]	helper-field for generation of triples. contains same value as undiag_pheno_hpo but will only be filled if a second variant is reported.	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([undiag_v2_yes]='yes', [undiag_pheno_hpo], ''))								
581	[undiag_desc2]	Section Header: GENOTYPE This section is intended to record the change(s) in DNA, RNA or protein sequences according to the standards of the Sequence Variant Nomenclature "HGVS simple" (accessed: 18.13.22). HGVS-codes can be checked with Mutalyzer (accessed: 18.03.22). Please enter all the details that you have available from the report. For autosomal recessive cases, you must enter both variant 1 and variant 2. If the variant is homocytous, enter the identical details twice. For autosomal dominant and X-linked, only one is required.	descriptive								
582	[undiag_report_variant]	Report genetic variant(s)	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> <tr> <td>not_applicable</td> <td>Not applicable</td> </tr> <tr> <td>not_present</td> <td>Not present</td> </tr> </table>	yes	Yes	unknown	Unknown	not_applicable	Not applicable	not_present	Not present
yes	Yes										
unknown	Unknown										
not_applicable	Not applicable										
not_present	Not present										
583	[undiag_listpref] Show the field ONLY if: [undiag_report_variant] = 'yes'	Gene list preference <i>Element 6.3. in the Set of common data elements for Rare Diseases Registration (link)</i>	radio <table border="1"> <tr> <td>all_nmd</td> <td>Search gene in full list for neuromuscular diseases</td> </tr> <tr> <td>ontology</td> <td>If gene not in provided lists: Search gene in biomedical ontologies</td> </tr> </table>	all_nmd	Search gene in full list for neuromuscular diseases	ontology	If gene not in provided lists: Search gene in biomedical ontologies				
all_nmd	Search gene in full list for neuromuscular diseases										
ontology	If gene not in provided lists: Search gene in biomedical ontologies										
584	[undiag_omim_armx] Show the field ONLY if: [undiag_report_variant] = 'yes' AND [undiag_listpref]='all_nmd'	Gene Symbol (HGNC) for all neuromuscular diseases <i>Element 6.3. in the Set of common data elements for Rare Diseases Registration (link)</i>	dropdown (autocomplete) <table border="1"> <tr> <td>601065</td> <td>AARS1 / HGNC:20 / OMIM:601065 (alanyl-tRNA synthetase 1)</td> </tr> <tr> <td>612035</td> <td>AARS2 / HGNC:21022 / OMIM:612035 (alanyl-tRNA synthetase 2, mitochondrial)</td> </tr> <tr> <td>601439</td> <td>ABCC9 / HGNC:60 / OMIM:601439 (ATP binding cassette subfamily C member 9)</td> </tr> <tr> <td>604780</td> <td>ABHD5 / HGNC:21396 / OMIM:604780 (abhydrolase domain containing 5,</td> </tr> </table>	601065	AARS1 / HGNC:20 / OMIM:601065 (alanyl-tRNA synthetase 1)	612035	AARS2 / HGNC:21022 / OMIM:612035 (alanyl-tRNA synthetase 2, mitochondrial)	601439	ABCC9 / HGNC:60 / OMIM:601439 (ATP binding cassette subfamily C member 9)	604780	ABHD5 / HGNC:21396 / OMIM:604780 (abhydrolase domain containing 5,
601065	AARS1 / HGNC:20 / OMIM:601065 (alanyl-tRNA synthetase 1)										
612035	AARS2 / HGNC:21022 / OMIM:612035 (alanyl-tRNA synthetase 2, mitochondrial)										
601439	ABCC9 / HGNC:60 / OMIM:601439 (ATP binding cassette subfamily C member 9)										
604780	ABHD5 / HGNC:21396 / OMIM:604780 (abhydrolase domain containing 5,										

	lysophosphatidic acid acyltransferase)
611103	ACAD9 / HGNC:21497 / OMIM:611103 (acyl-CoA dehydrogenase family member 9)
609575	ACADVL / HGNC:92 / OMIM:609575 (acyl-CoA dehydrogenase very long chain)
617036	ACER3 / HGNC:16066 / OMIM:617036 (alkaline ceramidase 3)
102610	ACTA1 / HGNC:129 / OMIM:102610 (actin alpha 1, skeletal muscle)
102540	ACTC1 / HGNC:143 / OMIM:102540 (actin alpha cardiac muscle 1)
102573	ACTN2 / HGNC:164 / OMIM:102573 (actinin alpha 2)
102576	ACVR1 / HGNC:171 / OMIM:102576 (activin A receptor type 1)
606980	COQ8A / HGNC:16812 / OMIM:606980 (coenzyme Q8A)
600294	ADCY6 / HGNC:237 / OMIM:600294 (adenylate cyclase 6)
612243	ADGRG6 / HGNC:13841 / OMIM:612243 (adhesion G protein-coupled receptor G6)
612498	ADSS1 / HGNC:20093 / OMIM:612498 (adenylosuccinate synthase 1)
604581	AFG3L2 / HGNC:315 / OMIM:604581 (AFG3 like matrix AAA peptidase subunit 2)
610860	AGL / HGNC:321 / OMIM:610860 (amylo-alpha-1, 6-glucosidase, 4-alpha-glucanotransferase)
103320	AGRN / HGNC:329 / OMIM:103320 (agrin)
606830	AGTPBP1 / HGNC:17258 / OMIM:606830 (ATP/GTP binding carboxypeptidase 1)
608570	AHNAK2 / HGNC:20125 / OMIM:608570 (AHNAK nucleoprotein 2)
300169	AIFM1 / HGNC:8768 / OMIM:300169 (apoptosis inducing factor mitochondria associated 1)
604001	AKAP9 / HGNC:379 / OMIM:604001 (A-kinase anchoring protein 9)
138250	ALDH18A1 / HGNC:9722 / OMIM:138250 (aldehyde dehydrogenase 18 family member A1)
609523	ALDH3A2 / HGNC:403 / OMIM:609523 (aldehyde dehydrogenase 3 family member A2)
300776	ALG13 / HGNC:30881 / OMIM:300776 (ALG13 UDP-N-acetylglucosaminyltransferase subunit)
612866	ALG14 / HGNC:28287 / OMIM:612866 (ALG14 UDP-N-acetylglucosaminyltransferase subunit)
607905	ALG2 / HGNC:23159 / OMIM:607905 (ALG2 alpha-1,3/1,6-mannosyltransferase)
617608	ALPK3 / HGNC:17574 / OMIM:617608 (alpha kinase 3)
606352	ALS2 / HGNC:443 / OMIM:606352 (alsin

	Rho guanine nucleotide exchange factor ALS2)
102771	AMPD2 / HGNC:469 / OMIM:102771 (adenosine monophosphate deaminase 2)
105850	ANG / HGNC:483 / OMIM:105850 (angiogenin)
106410	ANK2 / HGNC:493 / OMIM:106410 (ankyrin 2)
609599	ANKRD1 / HGNC:15819 / OMIM:609599 (ankyrin repeat domain 1)
613726	ANO10 / HGNC:25519 / OMIM:613726 (anoctamin 10)
608662	ANO5 / HGNC:27337 / OMIM:608662 (anoctamin 5)
602572	ANXA11 / HGNC:535 / OMIM:602572 (annexin A11)
607245	AP4B1 / HGNC:572 / OMIM:607245 (adaptor related protein complex 4 subunit beta 1)
607244	AP4E1 / HGNC:573 / OMIM:607244 (adaptor related protein complex 4 subunit epsilon 1)
602296	AP4M1 / HGNC:574 / OMIM:602296 (adaptor related protein complex 4 subunit mu 1)
607243	AP4S1 / HGNC:575 / OMIM:607243 (adaptor related protein complex 4 subunit sigma 1)
613653	AP5Z1 / HGNC:22197 / OMIM:613653 (adaptor related protein complex 5 subunit zeta 1)
606350	APTX / HGNC:15984 / OMIM:606350 (aprataxin)
313700	AR / HGNC:644 / OMIM:313700 (androgen receptor)
608136	ARHGEF10 / HGNC:14103 / OMIM:608136 (Rho guanine nucleotide exchange factor 10)
607669	ARL6IP1 / HGNC:697 / OMIM:607669 (ADP ribosylation factor like GTPase 6 interacting protein 1)
613468	ASAH1 / HGNC:735 / OMIM:613468 (N-acylsphingosine amidohydrolase 1)
614215	ASCC1 / HGNC:24268 / OMIM:614215 (activating signal cointegrator 1 complex subunit 1)
604261	ATG5 / HGNC:589 / OMIM:604261 (autophagy related 5)
606439	ATL1 / HGNC:11231 / OMIM:606439 (atlastin GTPase 1)
609369	ATL3 / HGNC:24526 / OMIM:609369 (atlastin GTPase 3)
607585	ATM / HGNC:795 / OMIM:607585 (ATM serine/threonine kinase)
610513	ATP13A2 / HGNC:30213 / OMIM:610513 (ATPase cation transporting 13A2)
182310	ATP1A1 / HGNC:799 / OMIM:182310 (ATPase Na <sup>+</sup> /K <sup>+</sup> transporting subunit alpha 1)
182340	ATP1A2 / HGNC:800 / OMIM:182340

	(ATPase Na <sup>+</sup> /K <sup>+</sup> transporting subunit alpha 2)
108730	ATP2A1 / HGNC:811 / OMIM:108730 (ATPase sarcoplasmic/endoplasmic reticulum Ca <sup>2+</sup> transporting 1)
300011	ATP7A / HGNC:869 / OMIM:300011 (ATPase copper transporting alpha)
601556	ATXN1 / HGNC:10548 / OMIM:601556 (ataxin 1)
611150	ATXN10 / HGNC:10549 / OMIM:611150 (ataxin 10)
601517	ATXN2 / HGNC:10555 / OMIM:601517 (ataxin 2)
607047	ATXN3 / HGNC:7106 / OMIM:607047 (ataxin 3)
607640	ATXN7 / HGNC:10560 / OMIM:607640 (ataxin 7)
603680	ATXN8OS / HGNC:10561 / OMIM:603680 (ATXN8 opposite strand lncRNA)
610194	B3GALNT2 / HGNC:28596 / OMIM:610194 (beta-1,3-N- acetylgalactosaminyltransferase 2)
605517	B4GAT1 / HGNC:15685 / OMIM:605517 (beta-1,4-glucuronyltransferase 1)
601873	B4GALNT1 / HGNC:4117 / OMIM:601873 (beta-1,4-N-acetyl- galactosaminyltransferase 1)
603883	BAG3 / HGNC:939 / OMIM:603883 (BAG cochaperone 3)
612051	BEAN1 / HGNC:24160 / OMIM:612051 (brain expressed associated with NEDD4 1)
609797	BICD2 / HGNC:17208 / OMIM:609797 (BICD cargo adaptor 2)
601248	BIN1 / HGNC:1052 / OMIM:601248 (bridging integrator 1)
606158	BSCL2 / HGNC:15832 / OMIM:606158 (BSCL2 lipid droplet biogenesis associated, seipin)
604577	BVES / HGNC:1152 / OMIM:604577 (blood vessel epicardial substance)
613541	MTRFR / HGNC:26784 / OMIM:613541 (mitochondrial translation release factor in rescue)
614297	C19orf12 / HGNC:25443 / OMIM:614297 (chromosome 19 open reading frame 12)
618682	C1orf194 / HGNC:32331 / OMIM:618682 (chromosome 1 open reading frame 194)
601269	C1QBP / HGNC:1243 / OMIM:601269 (complement C1q binding protein)
614260	C9orf72 / HGNC:28337 / OMIM:614260 (C9orf72-SMCR8 complex subunit)
601011	CACNA1A / HGNC:1388 / OMIM:601011 (calcium voltage-gated channel subunit alpha1 A)
114205	CACNA1C / HGNC:1390 / OMIM:114205 (calcium voltage-gated channel subunit alpha1 C)
604065	CACNA1G / HGNC:1394 / OMIM:604065



	(calcium voltage-gated channel subunit alpha1 G)
607904	CACNA1H / HGNC:1395 / OMIM:607904 (calcium voltage-gated channel subunit alpha1 H)
114208	CACNA1S / HGNC:1397 / OMIM:114208 (calcium voltage-gated channel subunit alpha1 S)
600003	CACNB2 / HGNC:1402 / OMIM:600003 (calcium voltage-gated channel auxiliary subunit beta 2)
601949	CACNB4 / HGNC:1404 / OMIM:601949 (calcium voltage-gated channel auxiliary subunit beta 4)
114180	CALM1 / HGNC:1442 / OMIM:114180 (calmodulin 1)
114182	CALM2 / HGNC:1445 / OMIM:114182 (calmodulin 2)
611414	CALR3 / HGNC:20407 / OMIM:611414 (calreticulin 3)
114220	CAPN1 / HGNC:1476 / OMIM:114220 (calpain 1)
114240	CAPN3 / HGNC:1480 / OMIM:114240 (calpain 3)
114250	CASQ1 / HGNC:1512 / OMIM:114250 (calsequestrin 1)
114251	CASQ2 / HGNC:1513 / OMIM:114251 (calsequestrin 2)
601253	CAV3 / HGNC:1529 / OMIM:601253 (caveolin 3)
603198	CAVIN1 / HGNC:9688 / OMIM:603198 (caveolae associated protein 1)
617714	CAVIN4 / HGNC:33742 / OMIM:617714 (caveolae associated protein 4)
614666	CCDC78 / HGNC:14153 / OMIM:614666 (coiled-coil domain containing 78)
611204	CCDC88C / HGNC:19967 / OMIM:611204 (coiled-coil domain containing 88C)
610150	CCT5 / HGNC:1618 / OMIM:610150 (chaperonin containing TCP1 subunit 5)
601443	CFL2 / HGNC:1875 / OMIM:601443 (cofilin 2)
118490	CHAT / HGNC:1912 / OMIM:118490 (choline O-acetyltransferase)
615903	CHCHD10 / HGNC:15559 / OMIM:615903 (coiled-coil-helix-coiled-coil-helix domain containing 10)
612395	CHKB / HGNC:1938 / OMIM:612395 (choline kinase beta)
609512	CHMP2B / HGNC:24537 / OMIM:609512 (charged multivesicular body protein 2B)
606988	CHP1 / HGNC:17433 / OMIM:606988 (calcineurin like EF-hand protein 1)
100690	CHRNA1 / HGNC:1955 / OMIM:100690 (cholinergic receptor nicotinic alpha 1 subunit)
100710	CHRN1 / HGNC:1961 / OMIM:100710 (cholinergic receptor nicotinic beta 1 subunit)

100720	CHRND / HGNC:1965 / OMIM:100720 (cholinergic receptor nicotinic delta subunit)
100725	CHRNE / HGNC:1966 / OMIM:100725 (cholinergic receptor nicotinic epsilon subunit)
100730	CHRNA3 / HGNC:1967 / OMIM:100730 (cholinergic receptor nicotinic gamma subunit)
118425	CLCN1 / HGNC:2019 / OMIM:118425 (chloride voltage-gated channel 1)
607042	CLN3 / HGNC:2074 / OMIM:607042 (CLN3 lysosomal/endosomal transmembrane protein, battenin)
601273	CLTCL1 / HGNC:2093 / OMIM:601273 (clathrin heavy chain like 1)
116955	CNBP / HGNC:13164 / OMIM:116955 (CCHC-type zinc finger nucleic acid binding protein)
600016	CNTN1 / HGNC:2171 / OMIM:600016 (contactin 1)
602346	CNTNAP1 / HGNC:8011 / OMIM:602346 (contactin associated protein 1)
615623	COA7 / HGNC:25716 / OMIM:615623 (cytochrome c oxidase assembly factor 7)
120320	COL12A1 / HGNC:2188 / OMIM:120320 (collagen type XII alpha 1 chain)
120350	COL13A1 / HGNC:2190 / OMIM:120350 (collagen type XIII alpha 1 chain)
610004	COL25A1 / HGNC:18603 / OMIM:610004 (collagen type XXV alpha 1 chain)
120215	COL5A1 / HGNC:2209 / OMIM:120215 (collagen type V alpha 1 chain)
120220	COL6A1 / HGNC:2211 / OMIM:120220 (collagen type VI alpha 1 chain)
120240	COL6A2 / HGNC:2212 / OMIM:120240 (collagen type VI alpha 2 chain)
120250	COL6A3 / HGNC:2213 / OMIM:120250 (collagen type VI alpha 3 chain)
603033	COLQ / HGNC:2226 / OMIM:603033 (collagen like tail subunit of asymmetric acetylcholinesterase)
609825	COQ2 / HGNC:25223 / OMIM:609825 (coenzyme Q2, polyprenyltransferase)
612898	COQ4 / HGNC:19693 / OMIM:612898 (coenzyme Q4)
614647	COQ6 / HGNC:20233 / OMIM:614647 (coenzyme Q6, monooxygenase)
601683	COQ7 / HGNC:2244 / OMIM:601683 (coenzyme Q7, hydroxylase)
612837	COQ9 / HGNC:25302 / OMIM:612837 (coenzyme Q9)
603646	COX15 / HGNC:2263 / OMIM:603646 (cytochrome c oxidase assembly homolog COX15)
602072	COX6A1 / HGNC:2277 / OMIM:602072 (cytochrome c oxidase subunit 6A1)
602009	COX6A2 / HGNC:2279 / OMIM:602009 (cytochrome c oxidase subunit 6A2)

608846	CPT1C / HGNC:18540 / OMIM:608846 (carnitine palmitoyltransferase 1C)
600650	CPT2 / HGNC:2330 / OMIM:600650 (carnitine palmitoyltransferase 2)
614631	CRPPA / HGNC:37276 / OMIM:614631 (CDP-L-ribitol pyrophosphorylase A)
123590	CRYAB / HGNC:2389 / OMIM:123590 (crystallin alpha B)
600824	CSRP3 / HGNC:2472 / OMIM:600824 (cysteine and glycine rich protein 3)
604927	CTDP1 / HGNC:2498 / OMIM:604927 (CTD phosphatase subunit 1)
607667	CTNNA3 / HGNC:2511 / OMIM:607667 (catenin alpha 3)
616120	CWF19L1 / HGNC:25613 / OMIM:616120 (CWF19 like cell cycle control factor 1)
610670	CYP2U1 / HGNC:20582 / OMIM:610670 (cytochrome P450 family 2 subfamily U member 1)
603711	CYP7B1 / HGNC:2652 / OMIM:603711 (cytochrome P450 family 7 subfamily B member 1)
128239	DAG1 / HGNC:2666 / OMIM:128239 (dystroglycan 1)
615820	DCAF8 / HGNC:24891 / OMIM:615820 (DDB1 and CUL4 associated factor 8)
601143	DCTN1 / HGNC:2711 / OMIM:601143 (dynactin subunit 1)
614603	DDHD1 / HGNC:19714 / OMIM:614603 (DDHD domain containing 1)
615003	DDHD2 / HGNC:29106 / OMIM:615003 (DDHD domain containing 2)
125660	DES / HGNC:2770 / OMIM:125660 (desmin)
606983	DGAT2 / HGNC:16940 / OMIM:606983 (diacylglycerol O-acyltransferase 2)
601465	DGUOK / HGNC:2858 / OMIM:601465 (deoxyguanosine kinase)
614984	DHTKD1 / HGNC:23537 / OMIM:614984 (dehydrogenase E1 and transketolase domain containing 1)
300377	DMD / HGNC:2928 / OMIM:300377 (dystrophin)
605377	DMPK / HGNC:2933 / OMIM:605377 (DM1 protein kinase)
601810	DNA2 / HGNC:2939 / OMIM:601810 (DNA replication helicase/nuclease 2)
604139	DNAJB2 / HGNC:5228 / OMIM:604139 (Dnaj heat shock protein family (Hsp40) member B2)
611332	DNAJB6 / HGNC:14888 / OMIM:611332 (Dnaj heat shock protein family (Hsp40) member B6)
602378	DNM2 / HGNC:2974 / OMIM:602378 (dynamin 2)
126375	DNMT1 / HGNC:2976 / OMIM:126375 (DNA methyltransferase 1)
610285	DOK7 / HGNC:26594 / OMIM:610285 (docking protein 7)
610746	DOLK / HGNC:23406 / OMIM:610746

	(dolichol kinase)
191350	DPAGT1 / HGNC:2995 / OMIM:191350 (dolichyl-phosphate N-acetylglucosaminophosphotransferase 1)
603503	DPM1 / HGNC:3005 / OMIM:603503 (dolichyl-phosphate mannosyltransferase subunit 1, catalytic)
603564	DPM2 / HGNC:3006 / OMIM:603564 (dolichyl-phosphate mannosyltransferase subunit 2, regulatory)
605951	DPM3 / HGNC:3007 / OMIM:605951 (dolichyl-phosphate mannosyltransferase subunit 3, regulatory)
125645	DSC2 / HGNC:3036 / OMIM:125645 (desmocollin 2)
125671	DSG2 / HGNC:3049 / OMIM:125671 (desmoglein 2)
125647	DSP / HGNC:3052 / OMIM:125647 (desmoplakin)
113810	DST / HGNC:1090 / OMIM:113810 (dystonin)
601239	DTNA / HGNC:3057 / OMIM:601239 (dystrobrevin alpha)
606009	DUX4L1 / HGNC:3082 / OMIM:606009 (double homeobox 4 like 1 (pseudogene))
600112	DYNC1H1 / HGNC:2961 / OMIM:600112 (dynein cytoplasmic 1 heavy chain 1)
603009	DYSF / HGNC:3097 / OMIM:603009 (dysferlin)
130610	EEF2 / HGNC:3214 / OMIM:130610 (eukaryotic translation elongation factor 2)
129010	EGR2 / HGNC:3239 / OMIM:129010 (early growth response 2)
605512	ELOVL4 / HGNC:14415 / OMIM:605512 (ELOVL fatty acid elongase 4)
611805	ELOVL5 / HGNC:21308 / OMIM:611805 (ELOVL fatty acid elongase 5)
603722	ELP1 / HGNC:5959 / OMIM:603722 (elongator acetyltransferase complex subunit 1)
300384	EMD / HGNC:3331 / OMIM:300384 (emerin)
131370	ENO3 / HGNC:3354 / OMIM:131370 (enolase 3)
601752	ENTPD1 / HGNC:3363 / OMIM:601752 (ectonucleoside triphosphate diphosphohydrolase 1)
190151	ERBB3 / HGNC:3431 / OMIM:190151 (erb-b2 receptor tyrosine kinase 3)
600543	ERBB4 / HGNC:3432 / OMIM:600543 (erb-b2 receptor tyrosine kinase 4)
611604	ERLIN1 / HGNC:16947 / OMIM:611604 (ER lipid raft associated 1)
611605	ERLIN2 / HGNC:1356 / OMIM:611605 (ER lipid raft associated 2)

608053	ETFA / HGNC:3481 / OMIM:608053 (electron transfer flavoprotein subunit alpha)
231675	ETFDH / HGNC:3483 / OMIM:231675 (electron transfer flavoprotein dehydrogenase)
606489	EXOSC3 / HGNC:17944 / OMIM:606489 (exosome component 3)
606019	EXOSC8 / HGNC:17035 / OMIM:606019 (exosome component 8)
603550	EYA4 / HGNC:3522 / OMIM:603550 (EYA transcriptional coactivator and phosphatase 4)
611026	FA2H / HGNC:21197 / OMIM:611026 (fatty acid 2-hydroxylase)
615584	FAM111B / HGNC:24200 / OMIM:615584 (FAM111 trypsin like peptidase B)
611592	FARS2 / HGNC:21062 / OMIM:611592 (phenylalanyl-tRNA synthetase 2, mitochondrial)
612322	FASTKD2 / HGNC:29160 / OMIM:612322 (FAST kinase domains 2)
604269	FAT2 / HGNC:3596 / OMIM:604269 (FAT atypical cadherin 2)
604580	FBLN5 / HGNC:3602 / OMIM:604580 (fibulin 5)
605654	FBXL4 / HGNC:13601 / OMIM:605654 (F-box and leucine rich repeat protein 4)
608533	FBXO38 / HGNC:28844 / OMIM:608533 (F-box protein 38)
614585	FDX2 / HGNC:30546 / OMIM:614585 (ferredoxin 2)
611104	FGD4 / HGNC:19125 / OMIM:611104 (FYVE, RhoGEF and PH domain containing 4)
601515	FGF14 / HGNC:3671 / OMIM:601515 (fibroblast growth factor 14)
300163	FHL1 / HGNC:3702 / OMIM:300163 (four and a half LIM domains 1)
609390	FIG4 / HGNC:16873 / OMIM:609390 (FIG4 phosphoinositide 5-phosphatase)
606596	FKRP / HGNC:17997 / OMIM:606596 (fukutin related protein)
607440	FKTN / HGNC:3622 / OMIM:607440 (fukutin)
610595	FLAD1 / HGNC:24671 / OMIM:610595 (flavin adenine dinucleotide synthetase 1)
300017	FLNA / HGNC:3754 / OMIM:300017 (filamin A)
102565	FLNC / HGNC:3756 / OMIM:102565 (filamin C)
609144	FLVCR1 / HGNC:24682 / OMIM:609144 (FLVCR heme transporter 1)
601278	FRG1 / HGNC:3954 / OMIM:601278 (FSHD region gene 1)
137070	FUS / HGNC:4010 / OMIM:137070 (FUS RNA binding protein)
606829	FXN / HGNC:3951 / OMIM:606829 (frataxin)

600819	FXR1 / HGNC:4023 / OMIM:600819 (FMR1 autosomal homolog 1)
606800	GAA / HGNC:4065 / OMIM:606800 (alpha glucosidase)
605379	GAN / HGNC:4137 / OMIM:605379 (gigaxonin)
600287	GARS1 / HGNC:4162 / OMIM:600287 (glycyl-tRNA synthetase 1)
614518	GATAD1 / HGNC:29941 / OMIM:614518 (GATA zinc finger domain containing 1)
606463	GBA / HGNC:4177 / OMIM:606463 (glucosylceramidase beta)
609471	GBA2 / HGNC:18986 / OMIM:609471 (glucosylceramidase beta 2)
607839	GBE1 / HGNC:4180 / OMIM:607839 (1,4- alpha-glucan branching enzyme 1)
603698	GBF1 / HGNC:4181 / OMIM:603698 (golgi brefeldin A resistant guanine nucleotide exchange factor 1)
606598	GDAP1 / HGNC:15968 / OMIM:606598 (ganglioside induced differentiation associated protein 1)
618128	GDAP2 / HGNC:18010 / OMIM:618128 (ganglioside induced differentiation associated protein 2)
138292	GFPT1 / HGNC:4241 / OMIM:138292 (glutamine--fructose-6-phosphate transaminase 1)
605072	GIPC1 / HGNC:1226 / OMIM:605072 (GIPC PDZ domain containing family member 1)
121013	GJA5 / HGNC:4279 / OMIM:121013 (gap junction protein alpha 5)
304040	GJB1 / HGNC:4283 / OMIM:304040 (gap junction protein beta 1)
603324	GJB3 / HGNC:4285 / OMIM:603324 (gap junction protein beta 3)
608803	GJC2 / HGNC:17494 / OMIM:608803 (gap junction protein gamma 2)
608603	GLDN / HGNC:29514 / OMIM:608603 (gliomedin)
603371	GLE1 / HGNC:4315 / OMIM:603371 (GLE1 RNA export mediator)
615320	GMPPB / HGNC:22932 / OMIM:615320 (GDP-mannose pyrophosphorylase B)
610863	GNB4 / HGNC:20731 / OMIM:610863 (G protein subunit beta 4)
603824	GNE / HGNC:23657 / OMIM:603824 (glucosamine (UDP-N-acetyl)-2- epimerase/N-acetylmannosamine kinase)
602580	GOLGA2 / HGNC:4425 / OMIM:602580 (golgin A2)
604027	GOSR2 / HGNC:4431 / OMIM:604027 (golgi SNAP receptor complex member 2)
611778	GPD1L / HGNC:28956 / OMIM:611778 (glycerol-3-phosphate dehydrogenase 1 like)
602368	GRID2 / HGNC:4576 / OMIM:602368 (glutamate ionotropic receptor delta)

	type subunit 2)
604473	GRM1 / HGNC:4593 / OMIM:604473 (glutamate metabotropic receptor 1)
603942	GYG1 / HGNC:4699 / OMIM:603942 (glycogenin 1)
138570	GYS1 / HGNC:4706 / OMIM:138570 (glycogen synthase 1)
610467	HACD1 / HGNC:9639 / OMIM:610467 (3- hydroxyacyl-CoA dehydratase 1)
610876	HACE1 / HGNC:21033 / OMIM:610876 (HECT domain and ankyrin repeat containing E3 ubiquitin protein ligase 1)
142810	HARS1 / HGNC:4816 / OMIM:142810 (histidyl-tRNA synthetase 1)
605206	HCN4 / HGNC:16882 / OMIM:605206 (hyperpolarization activated cyclic nucleotide gated potassium channel 4)
606873	HEXB / HGNC:4879 / OMIM:606873 (hexosaminidase subunit beta)
601314	HINT1 / HGNC:4912 / OMIM:601314 (histidine triad nucleotide binding protein 1)
142600	HK1 / HGNC:4922 / OMIM:142600 (hexokinase 1)
164017	HNRNPA1 / HGNC:5031 / OMIM:164017 (heterogeneous nuclear ribonucleoprotein A1)
600124	HNRNPA2B1 / HGNC:5033 / OMIM:600124 (heterogeneous nuclear ribonucleoprotein A2/B1)
607137	HNRNPDL / HGNC:5037 / OMIM:607137 (heterogeneous nuclear ribonucleoprotein D like)
142984	HOXD10 / HGNC:5133 / OMIM:142984 (homeobox D10)
190020	HRAS / HGNC:5173 / OMIM:190020 (HRas proto-oncogene, GTPase)
602195	HSPB1 / HGNC:5246 / OMIM:602195 (heat shock protein family B (small) member 1)
604624	HSPB3 / HGNC:5248 / OMIM:604624 (heat shock protein family B (small) member 3)
608014	HSPB8 / HGNC:30171 / OMIM:608014 (heat shock protein family B (small) member 8)
118190	HSPD1 / HGNC:5261 / OMIM:118190 (heat shock protein family D (Hsp60) member 1)
142461	HSPG2 / HGNC:5273 / OMIM:142461 (heparan sulfate proteoglycan 2)
615316	IBA57 / HGNC:27302 / OMIM:615316 (iron-sulfur cluster assembly factor IBA57)
603502	IFRD1 / HGNC:5456 / OMIM:603502 (interferon related developmental regulator 1)
600502	IGHMBP2 / HGNC:5542 / OMIM:600502 (immunoglobulin mu DNA binding protein 2)
602366	ILK / HGNC:6040 / OMIM:602366

	(integrin linked kinase)
610982	INF2 / HGNC:23791 / OMIM:610982 (inverted formin 2)
607875	INPP5K / HGNC:33882 / OMIM:607875 (inositol polyphosphate-5-phosphatase K)
611911	ISCU / HGNC:29882 / OMIM:611911 (iron-sulfur cluster assembly enzyme)
600536	ITGA7 / HGNC:6143 / OMIM:600536 (integrin subunit alpha 7)
147265	ITPR1 / HGNC:6180 / OMIM:147265 (inositol 1,4,5-trisphosphate receptor type 1)
147267	ITPR3 / HGNC:6182 / OMIM:147267 (inositol 1,4,5-trisphosphate receptor type 3)
601920	JAG1 / HGNC:6188 / OMIM:601920 (jagged canonical Notch ligand 1)
605267	JPH2 / HGNC:14202 / OMIM:605267 (junctophilin 2)
173325	JUP / HGNC:6207 / OMIM:173325 (junction plakoglobin)
601421	KARS1 / HGNC:6215 / OMIM:601421 (lysyl-tRNA synthetase 1)
613727	KBTBD13 / HGNC:37227 / OMIM:613727 (kelch repeat and BTB domain containing 13)
176260	KCNA1 / HGNC:6218 / OMIM:176260 (potassium voltage-gated channel subfamily A member 1)
176267	KCNA5 / HGNC:6224 / OMIM:176267 (potassium voltage-gated channel subfamily A member 5)
176264	KCNC3 / HGNC:6235 / OMIM:176264 (potassium voltage-gated channel subfamily C member 3)
605411	KCND3 / HGNC:6239 / OMIM:605411 (potassium voltage-gated channel subfamily D member 3)
176261	KCNE1 / HGNC:6240 / OMIM:176261 (potassium voltage-gated channel subfamily E regulatory subunit 1)
603796	KCNE2 / HGNC:6242 / OMIM:603796 (potassium voltage-gated channel subfamily E regulatory subunit 2)
604433	KCNE3 / HGNC:6243 / OMIM:604433 (potassium voltage-gated channel subfamily E regulatory subunit 3)
152427	KCNH2 / HGNC:6251 / OMIM:152427 (potassium voltage-gated channel subfamily H member 2)
602323	KCNJ12 / HGNC:6258 / OMIM:602323 (potassium inwardly rectifying channel subfamily J member 12)
600681	KCNJ2 / HGNC:6263 / OMIM:600681 (potassium inwardly rectifying channel subfamily J member 2)
600734	KCNJ5 / HGNC:6266 / OMIM:600734 (potassium inwardly rectifying channel subfamily J member 5)
607542	KCNQ1 / HGNC:6294 / OMIM:607542 (potassium voltage-gated channel



	subfamily Q member 1)
610657	WASHC5 / HGNC:28984 / OMIM:610657 (WASH complex subunit 5)
615759	KIDINS220 / HGNC:29508 / OMIM:615759 (kinase D interacting substrate 220)
601255	KIF1A / HGNC:888 / OMIM:601255 (kinesin family member 1A)
605995	KIF1B / HGNC:16636 / OMIM:605995 (kinesin family member 1B)
603060	KIF1C / HGNC:6317 / OMIM:603060 (kinesin family member 1C)
608283	KIF21A / HGNC:19349 / OMIM:608283 (kinesin family member 21A)
614026	KIF26B / HGNC:25484 / OMIM:614026 (kinesin family member 26B)
602821	KIF5A / HGNC:6323 / OMIM:602821 (kinesin family member 5A)
611729	KLC2 / HGNC:20716 / OMIM:611729 (kinesin light chain 2)
615340	KLHL40 / HGNC:30372 / OMIM:615340 (kelch like family member 40)
607701	KLHL41 / HGNC:16905 / OMIM:607701 (kelch like family member 41)
611201	KLHL9 / HGNC:18732 / OMIM:611201 (kelch like family member 9)
605739	KY / HGNC:26576 / OMIM:605739 (kyphoscoliosis peptidase)
308840	L1CAM / HGNC:6470 / OMIM:308840 (L1 cell adhesion molecule)
156225	LAMA2 / HGNC:6482 / OMIM:156225 (laminin subunit alpha 2)
600133	LAMA4 / HGNC:6484 / OMIM:600133 (laminin subunit alpha 4)
601033	LAMA5 / HGNC:6485 / OMIM:601033 (laminin subunit alpha 5)
150325	LAMB2 / HGNC:6487 / OMIM:150325 (laminin subunit beta 2)
309060	LAMP2 / HGNC:6501 / OMIM:309060 (lysosomal associated membrane protein 2)
603590	LARGE1 / HGNC:6511 / OMIM:603590 (LARGE xylosyl- and glucuronyltransferase 1)
605906	LDB3 / HGNC:15710 / OMIM:605906 (LIM domain binding 3)
150000	LDHA / HGNC:6535 / OMIM:150000 (lactate dehydrogenase A)
607908	LIMS2 / HGNC:16084 / OMIM:607908 (LIM zinc finger domain containing 2)
603795	LITAF / HGNC:16841 / OMIM:603795 (lipopolysaccharide induced TNF factor)
150330	LMNA / HGNC:6636 / OMIM:150330 (lamin A/C)
616112	LMOD3 / HGNC:6649 / OMIM:616112 (leiomodin 3)
605518	LPIN1 / HGNC:13345 / OMIM:605518 (lipin 1)
618299	LRP12 / HGNC:31708 / OMIM:618299

	(LDL receptor related protein 12)
604270	LRP4 / HGNC:6696 / OMIM:604270 (LDL receptor related protein 4)
610933	LRSAM1 / HGNC:25135 / OMIM:610933 (leucine rich repeat and sterile alpha motif containing 1)
159460	MAG / HGNC:6783 / OMIM:159460 (myelin associated glycoprotein)
609479	MAP3K20 / HGNC:17797 / OMIM:609479 (mitogen-activated protein kinase kinase kinase 20)
157140	MAPT / HGNC:6893 / OMIM:157140 (microtubule associated protein tau)
156560	MARS1 / HGNC:6898 / OMIM:156560 (methionyl-tRNA synthetase 1)
609728	MARS2 / HGNC:25133 / OMIM:609728 (methionyl-tRNA synthetase 2, mitochondrial)
164015	MATR3 / HGNC:6912 / OMIM:164015 (matrin 3)
160000	MB / HGNC:6915 / OMIM:160000 (myoglobin)
603294	MCM3AP / HGNC:6946 / OMIM:603294 (minichromosome maintenance complex component 3 associated protein)
612453	MEGF10 / HGNC:29634 / OMIM:612453 (multiple EGF like domains 10)
608507	MFN2 / HGNC:16877 / OMIM:608507 (mitofusin 2)
615076	MGME1 / HGNC:16205 / OMIM:615076 (mitochondrial genome maintenance exonuclease 1)
608677	MIB1 / HGNC:21086 / OMIM:608677 (MIB E3 ubiquitin protein ligase 1)
120520	MME / HGNC:7154 / OMIM:120520 (membrane metalloendopeptidase)
616661	MORC2 / HGNC:23573 / OMIM:616661 (MORC family CW-type zinc finger 2)
604041	MPDU1 / HGNC:7207 / OMIM:604041 (mannose-P-dolichol utilization defect 1)
137960	MPV17 / HGNC:7224 / OMIM:137960 (mitochondrial inner membrane protein MPV17)
159440	MPZ / HGNC:7225 / OMIM:159440 (myelin protein zero)
600814	MRE11 / HGNC:7230 / OMIM:600814 (MRE11 homolog, double strand break repair nuclease)
607118	MRPL3 / HGNC:10379 / OMIM:607118 (mitochondrial ribosomal protein L3)
611849	MRPL44 / HGNC:16650 / OMIM:611849 (mitochondrial ribosomal protein L44)
611987	MRPS25 / HGNC:14511 / OMIM:611987 (mitochondrial ribosomal protein S25)
601788	MSTN / HGNC:4223 / OMIM:601788 (myostatin)
617619	MSTO1 / HGNC:29678 / OMIM:617619 (misato mitochondrial distribution and morphology regulator 1)

590065	MT-TM / HGNC:7492 / OMIM:590065 (mitochondrially encoded tRNA-Met (AUA/G))
590090	MT-TT / HGNC:7499 / OMIM:590090 (mitochondrially encoded tRNA-Thr (ACN))
300415	MTM1 / HGNC:7448 / OMIM:300415 (myotubularin 1)
603557	MTMR2 / HGNC:7450 / OMIM:603557 (myotubularin related protein 2)
614667	MTO1 / HGNC:19261 / OMIM:614667 (mitochondrial tRNA translation optimization 1)
613669	MTPAP / HGNC:25532 / OMIM:613669 (mitochondrial poly(A) polymerase)
601296	MUSK / HGNC:7525 / OMIM:601296 (muscle associated receptor tyrosine kinase)
160794	MYBPC1 / HGNC:7549 / OMIM:160794 (myosin binding protein C1)
600958	MYBPC3 / HGNC:7551 / OMIM:600958 (myosin binding protein C3)
608568	MYH14 / HGNC:23212 / OMIM:608568 (myosin heavy chain 14)
160740	MYH2 / HGNC:7572 / OMIM:160740 (myosin heavy chain 2)
160720	MYH3 / HGNC:7573 / OMIM:160720 (myosin heavy chain 3)
160710	MYH6 / HGNC:7576 / OMIM:160710 (myosin heavy chain 6)
160760	MYH7 / HGNC:7577 / OMIM:160760 (myosin heavy chain 7)
160741	MYH8 / HGNC:7578 / OMIM:160741 (myosin heavy chain 8)
160780	MYL1 / HGNC:7582 / OMIM:160780 (myosin light chain 1)
160781	MYL2 / HGNC:7583 / OMIM:160781 (myosin light chain 2)
160790	MYL3 / HGNC:7584 / OMIM:160790 (myosin light chain 3)
160770	MYL4 / HGNC:7585 / OMIM:160770 (myosin light chain 4)
606566	MYLK2 / HGNC:16243 / OMIM:606566 (myosin light chain kinase 2)
615345	MYMK / HGNC:33778 / OMIM:615345 (myomaker, myoblast fusion factor)
607295	MYO18B / HGNC:18150 / OMIM:607295 (myosin XVIIIIB)
604875	MYO9A / HGNC:7608 / OMIM:604875 (myosin IXA)
604103	MYOT / HGNC:12399 / OMIM:604103 (myotilin)
605602	MYOZ2 / HGNC:1330 / OMIM:605602 (myozenin 2)
608517	MYPN / HGNC:23246 / OMIM:608517 (myopalladin)
609701	NAGLU / HGNC:7632 / OMIM:609701 (N- acetyl-alpha-glucosaminidase)
612803	NARS2 / HGNC:26274 / OMIM:612803 (asparaginyl-tRNA synthetase 2,

	mitochondrial)
605262	NDRG1 / HGNC:7679 / OMIM:605262 (N-myc downstream regulated 1)
606934	NDUFAB1 / HGNC:18828 / OMIM:606934 (NADH:ubiquinone oxidoreductase complex assembly factor 1)
161650	NEB / HGNC:7720 / OMIM:161650 (nebulin)
162230	NEFH / HGNC:7737 / OMIM:162230 (neurofilament heavy chain)
162280	NEFL / HGNC:7739 / OMIM:162280 (neurofilament light chain)
604588	NEK1 / HGNC:7744 / OMIM:604588 (NIMA related kinase 1)
609798	NEK9 / HGNC:18591 / OMIM:609798 (NIMA related kinase 9)
613121	NEXN / HGNC:29557 / OMIM:613121 (nexilin F-actin binding protein)
162030	NGF / HGNC:7808 / OMIM:162030 (nerve growth factor)
608145	NIPA1 / HGNC:17043 / OMIM:608145 (NIPA magnesium transporter 1)
605955	NKX6-2 / HGNC:19321 / OMIM:605955 (NK6 homeobox 2)
608701	NMNAT2 / HGNC:16789 / OMIM:608701 (nicotinamide nucleotide adenylyltransferase 2)
614154	NOP56 / HGNC:15911 / OMIM:614154 (NOP56 ribonucleoprotein)
108780	NPPA / HGNC:7939 / OMIM:108780 (natriuretic peptide A)
600417	NT5C2 / HGNC:8022 / OMIM:600417 (5'- nucleotidase, cytosolic II)
191315	NTRK1 / HGNC:8031 / OMIM:191315 (neurotrophic receptor tyrosine kinase 1)
606694	NUP155 / HGNC:8063 / OMIM:606694 (nucleoporin 155)
602552	NUP88 / HGNC:8067 / OMIM:602552 (nucleoporin 88)
605290	OPA1 / HGNC:8140 / OMIM:605290 (OPA1 mitochondrial dynamin like GTPase)
602432	OPTN / HGNC:17142 / OMIM:602432 (optineurin)
610277	ORAI1 / HGNC:25896 / OMIM:610277 (ORAI calcium release-activated calcium modulator 1)
602279	PABPN1 / HGNC:8565 / OMIM:602279 (poly(A) binding protein nuclear 1)
167410	PAX7 / HGNC:8621 / OMIM:167410 (paired box 7)
176740	PCNA / HGNC:8729 / OMIM:176740 (proliferating cell nuclear antigen)
300906	PDK3 / HGNC:8811 / OMIM:300906 (pyruvate dehydrogenase kinase 3)
131340	PDYN / HGNC:8820 / OMIM:131340 (prodynorphin)
601757	PEX7 / HGNC:8860 / OMIM:601757

	(peroxisomal biogenesis factor 7)
610681	PFKM / HGNC:8877 / OMIM:610681 (phosphofructokinase, muscle)
176610	PFN1 / HGNC:8881 / OMIM:176610 (profilin 1)
612931	PGAM2 / HGNC:8889 / OMIM:612931 (phosphoglycerate mutase 2)
311800	PGK1 / HGNC:8896 / OMIM:311800 (phosphoglycerate kinase 1)
171900	PGM1 / HGNC:8905 / OMIM:171900 (phosphoglucomutase 1)
311870	PHKA1 / HGNC:8925 / OMIM:311870 (phosphorylase kinase regulatory subunit alpha 1)
602753	PHOX2A / HGNC:691 / OMIM:602753 (paired like homeobox 2A)
602026	PHYH / HGNC:8940 / OMIM:602026 (phytanoyl-CoA 2-hydroxylase)
613629	PIEZO2 / HGNC:26270 / OMIM:613629 (piezo type mechanosensitive ion channel component 2)
606102	PIP5K1C / HGNC:8996 / OMIM:606102 (phosphatidylinositol-4-phosphate 5-kinase type 1 gamma)
602861	PKP2 / HGNC:9024 / OMIM:602861 (plakophilin 2)
615698	PLD3 / HGNC:17158 / OMIM:615698 (phospholipase D family member 3)
601282	PLEC / HGNC:9069 / OMIM:601282 (plectin)
611101	PLEKHG5 / HGNC:29105 / OMIM:611101 (pleckstrin homology and RhoGEF domain containing G5)
172405	PLN / HGNC:9080 / OMIM:172405 (phospholamban)
300401	PLP1 / HGNC:9086 / OMIM:300401 (proteolipid protein 1)
170715	PMP2 / HGNC:9117 / OMIM:170715 (peripheral myelin protein 2)
601097	PMP22 / HGNC:9118 / OMIM:601097 (peripheral myelin protein 22)
605610	PNKP / HGNC:9154 / OMIM:605610 (polynucleotide kinase 3'-phosphatase)
609059	PNPLA2 / HGNC:30802 / OMIM:609059 (patatin like phospholipase domain containing 2)
603197	PNPLA6 / HGNC:16268 / OMIM:603197 (patatin like phospholipase domain containing 6)
612123	PNPLA8 / HGNC:28900 / OMIM:612123 (patatin like phospholipase domain containing 8)
615618	POGLUT1 / HGNC:22954 / OMIM:615618 (protein O-glucosyltransferase 1)
174763	POLG / HGNC:9179 / OMIM:174763 (DNA polymerase gamma, catalytic subunit)
604983	POLG2 / HGNC:9180 / OMIM:604983 (DNA polymerase gamma 2, accessory)

	subunit)
606822	POMGNT1 / HGNC:19139 / OMIM:606822 (protein O-linked mannose N-acetylglucosaminyltransferase 1 (beta 1,2-))
614828	POMGNT2 / HGNC:25902 / OMIM:614828 (protein O-linked mannose N-acetylglucosaminyltransferase 2 (beta 1,4-))
615247	POMK / HGNC:26267 / OMIM:615247 (protein O-mannose kinase)
607423	POMT1 / HGNC:9202 / OMIM:607423 (protein O-mannosyltransferase 1)
607439	POMT2 / HGNC:19743 / OMIM:607439 (protein O-mannosyltransferase 2)
605824	POPDC3 / HGNC:17649 / OMIM:605824 (popeye domain containing 3)
604325	PPP2R2B / HGNC:9305 / OMIM:604325 (protein phosphatase 2 regulatory subunit Bbeta)
616458	PRDM12 / HGNC:13997 / OMIM:616458 (PR/SET domain 12)
605557	PRDM16 / HGNC:14000 / OMIM:605557 (PR/SET domain 16)
609557	PREPL / HGNC:30228 / OMIM:609557 (prolyl endopeptidase like)
602743	PRKAG2 / HGNC:9386 / OMIM:602743 (protein kinase AMP-activated non-catalytic subunit gamma 2)
176980	PRKCG / HGNC:9402 / OMIM:176980 (protein kinase C gamma)
170710	PRPH / HGNC:9461 / OMIM:170710 (peripherin)
311850	PRPS1 / HGNC:9462 / OMIM:311850 (phosphoribosyl pyrophosphate synthetase 1)
617413	PRUNE1 / HGNC:13420 / OMIM:617413 (prune exopolyphosphatase 1)
605725	PRX / HGNC:13797 / OMIM:605725 (periaxin)
104311	PSEN1 / HGNC:9508 / OMIM:104311 (presenilin 1)
600759	PSEN2 / HGNC:9509 / OMIM:600759 (presenilin 2)
608625	PTRH2 / HGNC:24265 / OMIM:608625 (peptidyl-tRNA hydrolase 2)
607204	PUM1 / HGNC:14957 / OMIM:607204 (pumilio RNA binding family member 1)
600473	PURA / HGNC:9701 / OMIM:600473 (purine rich element binding protein A)
608109	PUS1 / HGNC:15508 / OMIM:608109 (pseudouridine synthase 1)
608455	PYGM / HGNC:9726 / OMIM:608455 (glycogen phosphorylase, muscle associated)
617220	PYROXD1 / HGNC:26162 / OMIM:617220 (pyridine nucleotide-disulphide oxidoreductase domain 1)
602298	RAB7A / HGNC:9788 / OMIM:602298

	(RAB7A, member RAS oncogene family)
164760	RAF1 / HGNC:9829 / OMIM:164760 (Raf-1 proto-oncogene, serine/threonine kinase)
601592	RAPSN / HGNC:9863 / OMIM:601592 (receptor associated protein of the synapse)
610924	RBCK1 / HGNC:15864 / OMIM:610924 (RANBP2-type and C3HC4-type zinc finger containing 1)
613171	RBM20 / HGNC:27424 / OMIM:613171 (RNA binding motif protein 20)
612413	RBM7 / HGNC:9904 / OMIM:612413 (RNA binding motif protein 7)
609139	REEP1 / HGNC:25786 / OMIM:609139 (receptor accessory protein 1)
609347	REEP2 / HGNC:17975 / OMIM:609347 (receptor accessory protein 2)
613114	RETREG1 / HGNC:25964 / OMIM:613114 (reticulophagy regulator 1)
102579	RFC1 / HGNC:9969 / OMIM:102579 (replication factor C subunit 1)
604123	RNASEH1 / HGNC:18466 / OMIM:604123 (ribonuclease H1)
609948	RNF216 / HGNC:21698 / OMIM:609948 (ring finger protein 216)
612159	RPH3A / HGNC:17056 / OMIM:612159 (rabphilin 3A)
604712	RRM2B / HGNC:17296 / OMIM:604712 (ribonucleotide reductase regulatory TP53 inducible subunit M2B)
603183	RTN2 / HGNC:10468 / OMIM:603183 (reticulon 2)
613516	RUBCN / HGNC:28991 / OMIM:613516 (rubicon autophagy regulator)
605862	RXYLT1 / HGNC:13530 / OMIM:605862 (ribitol xylosyltransferase 1)
180901	RYR1 / HGNC:10483 / OMIM:180901 (ryanodine receptor 1)
180902	RYR2 / HGNC:10484 / OMIM:180902 (ryanodine receptor 2)
180903	RYR3 / HGNC:10485 / OMIM:180903 (ryanodine receptor 3)
604490	SACS / HGNC:10519 / OMIM:604490 (saccin molecular chaperone)
603560	SBF1 / HGNC:10542 / OMIM:603560 (SET binding factor 1)
607697	SBF2 / HGNC:2135 / OMIM:607697 (SET binding factor 2)
604385	SCN11A / HGNC:10583 / OMIM:604385 (sodium voltage-gated channel alpha subunit 11)
600235	SCN1B / HGNC:10586 / OMIM:600235 (sodium voltage-gated channel beta subunit 1)
601327	SCN2B / HGNC:10589 / OMIM:601327 (sodium voltage-gated channel beta subunit 2)
608214	SCN3B / HGNC:20665 / OMIM:608214 (sodium voltage-gated channel beta

	subunit 3)
603967	SCN4A / HGNC:10591 / OMIM:603967 (sodium voltage-gated channel alpha subunit 4)
608256	SCN4B / HGNC:10592 / OMIM:608256 (sodium voltage-gated channel beta subunit 4)
600163	SCN5A / HGNC:10593 / OMIM:600163 (sodium voltage-gated channel alpha subunit 5)
603415	SCN9A / HGNC:10597 / OMIM:603415 (sodium voltage-gated channel alpha subunit 9)
604272	SCO2 / HGNC:10604 / OMIM:604272 (synthesis of cytochrome C oxidase 2)
607982	SCYL1 / HGNC:14372 / OMIM:607982 (SCY1 like pseudokinase 1)
600857	SDHA / HGNC:10680 / OMIM:600857 (succinate dehydrogenase complex flavoprotein subunit A)
606210	SELENON / HGNC:15999 / OMIM:606210 (selenoprotein N)
604061	SEPTIN9 / HGNC:7323 / OMIM:604061 (septin 9)
608465	SETX / HGNC:445 / OMIM:608465 (senataxin)
600119	SGCA / HGNC:10805 / OMIM:600119 (sarcoglycan alpha)
600900	SGCB / HGNC:10806 / OMIM:600900 (sarcoglycan beta)
601411	SGCD / HGNC:10807 / OMIM:601411 (sarcoglycan delta)
608896	SGCG / HGNC:10809 / OMIM:608896 (sarcoglycan gamma)
603729	SGPL1 / HGNC:10817 / OMIM:603729 (sphingosine-1-phosphate lyase 1)
608206	SH3TC2 / HGNC:29427 / OMIM:608206 (SH3 domain and tetratricopeptide repeats 2)
601978	SIGMAR1 / HGNC:8157 / OMIM:601978 (sigma non-opioid intracellular receptor 1)
608005	SIL1 / HGNC:24624 / OMIM:608005 (SIL1 nucleotide exchange factor)
604878	SLC12A6 / HGNC:10914 / OMIM:604878 (solute carrier family 12 member 6)
600682	SLC16A1 / HGNC:10922 / OMIM:600682 (solute carrier family 16 member 1)
600336	SLC18A3 / HGNC:10936 / OMIM:600336 (solute carrier family 18 member A3)
600111	SLC1A3 / HGNC:10941 / OMIM:600111 (solute carrier family 1 member 3)
603377	SLC22A5 / HGNC:10969 / OMIM:603377 (solute carrier family 22 member 5)
190315	SLC25A1 / HGNC:10979 / OMIM:190315 (solute carrier family 25 member 1)
606521	SLC25A19 / HGNC:14409 / OMIM:606521 (solute carrier family 25 member 19)
613698	SLC25A20 / HGNC:1421 / OMIM:613698



	(solute carrier family 25 member 20)
103220	SLC25A4 / HGNC:10990 / OMIM:103220 (solute carrier family 25 member 4)
610823	SLC25A42 / HGNC:28380 / OMIM:610823 (solute carrier family 25 member 42)
610826	SLC25A46 / HGNC:25198 / OMIM:610826 (solute carrier family 25 member 46)
603690	SLC33A1 / HGNC:95 / OMIM:603690 (solute carrier family 33 member 1)
607882	SLC52A2 / HGNC:30224 / OMIM:607882 (solute carrier family 52 member 2)
613350	SLC52A3 / HGNC:16187 / OMIM:613350 (solute carrier family 52 member 3)
608761	SLC5A7 / HGNC:14025 / OMIM:608761 (solute carrier family 5 member 7)
107310	SLC9A1 / HGNC:11071 / OMIM:107310 (solute carrier family 9 member A1)
604990	SLC9A3R1 / HGNC:11075 / OMIM:604990 (SLC9A3 regulator 1)
614982	SMCHD1 / HGNC:29090 / OMIM:614982 (structural maintenance of chromosomes flexible hinge domain containing 1)
600354	SMN1 / HGNC:11117 / OMIM:600354 (survival of motor neuron 1, telomeric)
601627	SMN2 / HGNC:11118 / OMIM:601627 (survival of motor neuron 2, centromeric)
610457	SMPD4 / HGNC:32949 / OMIM:610457 (sphingomyelin phosphodiesterase 4)
600322	SNAP25 / HGNC:11132 / OMIM:600322 (synaptosome associated protein 25)
601017	SNTA1 / HGNC:11167 / OMIM:601017 (syntrophin alpha 1)
616105	SNX14 / HGNC:14977 / OMIM:616105 (sorting nexin 14)
147450	SOD1 / HGNC:11179 / OMIM:147450 (superoxide dismutase 1)
182500	SORD / HGNC:11184 / OMIM:182500 (sorbitol dehydrogenase)
607111	SPART / HGNC:18514 / OMIM:607111 (spartin)
604277	SPAST / HGNC:11233 / OMIM:604277 (spastin)
615950	SPEG / HGNC:16901 / OMIM:615950 (striated muscle enriched protein kinase)
610844	SPG11 / HGNC:11226 / OMIM:610844 (SPG11 vesicle trafficking associated, spatacsin)
608181	SPG21 / HGNC:20373 / OMIM:608181 (SPG21 abhydrolase domain containing, maspardin)
602783	SPG7 / HGNC:11237 / OMIM:602783 (SPG7 matrix AAA peptidase subunit, paraplegin)
182810	SPTAN1 / HGNC:11273 / OMIM:182810 (spectrin alpha, non-erythrocytic 1)

604985	SPTBN2 / HGNC:11276 / OMIM:604985 (spectrin beta, non-erythrocytic 2)
606214	SPTBN4 / HGNC:14896 / OMIM:606214 (spectrin beta, non-erythrocytic 4)
605712	SPTLC1 / HGNC:11277 / OMIM:605712 (serine palmitoyltransferase long chain base subunit 1)
605713	SPTLC2 / HGNC:11278 / OMIM:605713 (serine palmitoyltransferase long chain base subunit 2)
601530	SQSTM1 / HGNC:11280 / OMIM:601530 (sequestosome 1)
615521	STAC3 / HGNC:28423 / OMIM:615521 (SH3 and cysteine rich domain 3)
605921	STIM1 / HGNC:11386 / OMIM:605921 (stromal interaction molecule 1)
607207	STUB1 / HGNC:11427 / OMIM:607207 (STIP1 homology and U-box containing protein 1)
603921	SUCLA2 / HGNC:11448 / OMIM:603921 (succinate-CoA ligase ADP-forming subunit beta)
611224	SUCLG1 / HGNC:11449 / OMIM:611224 (succinate-CoA ligase GDP/ADP-forming subunit alpha)
185620	SURF1 / HGNC:11474 / OMIM:185620 (SURF1 cytochrome c oxidase assembly factor)
604126	SVIL / HGNC:11480 / OMIM:604126 (supervillin)
608441	SYNE1 / HGNC:17089 / OMIM:608441 (spectrin repeat containing nuclear envelope protein 1)
608442	SYNE2 / HGNC:17084 / OMIM:608442 (spectrin repeat containing nuclear envelope protein 2)
610949	SYT14 / HGNC:23143 / OMIM:610949 (synaptotagmin 14)
600104	SYT2 / HGNC:11510 / OMIM:600104 (synaptotagmin 2)
605078	TARDBP / HGNC:11571 / OMIM:605078 (TAR DNA binding protein)
300394	TFAFAZZIN / HGNC:11577 / OMIM:300394 (tafazzin, phospholipid-lysophospholipid transacylase)
604649	TBCD / HGNC:11581 / OMIM:604649 (tubulin folding cofactor D)
604834	TBK1 / HGNC:11584 / OMIM:604834 (TANK binding kinase 1)
600075	TBP / HGNC:11588 / OMIM:600075 (TATA-box binding protein)
604488	TCAP / HGNC:11610 / OMIM:604488 (titin-cap)
607198	TDP1 / HGNC:18884 / OMIM:607198 (tyrosyl-DNA phosphodiesterase 1)
605764	TDP2 / HGNC:17768 / OMIM:605764 (tyrosyl-DNA phosphodiesterase 2)
615000	TECPR2 / HGNC:19957 / OMIM:615000 (tectonin beta-propeller repeat containing 2)
617242	TECRL / HGNC:27365 / OMIM:617242

	(trans-2,3-enoyl-CoA reductase like)
602498	TFG / HGNC:11758 / OMIM:602498 (trafficking from ER to golgi regulator)
190230	TGFB3 / HGNC:11769 / OMIM:190230 (transforming growth factor beta 3)
613900	TGM6 / HGNC:16255 / OMIM:613900 (transglutaminase 6)
603518	TIA1 / HGNC:11802 / OMIM:603518 (TIA1 cytotoxic granule associated RNA binding protein)
607251	TIMM22 / HGNC:17317 / OMIM:607251 (translocase of inner mitochondrial membrane 22)
188250	TK2 / HGNC:11831 / OMIM:188250 (thymidine kinase 2)
616101	TMEM240 / HGNC:25186 / OMIM:616101 (transmembrane protein 240)
612048	TMEM43 / HGNC:28472 / OMIM:612048 (transmembrane protein 43)
616609	TMEM65 / HGNC:25203 / OMIM:616609 (transmembrane protein 65)
188380	TMPO / HGNC:11875 / OMIM:188380 (thymopoietin)
191040	TNNC1 / HGNC:11943 / OMIM:191040 (troponin C1, slow skeletal and cardiac type)
191043	TNNI2 / HGNC:11946 / OMIM:191043 (troponin I2, fast skeletal type)
191044	TNNI3 / HGNC:11947 / OMIM:191044 (troponin I3, cardiac type)
191041	TNNT1 / HGNC:11948 / OMIM:191041 (troponin T1, slow skeletal type)
191045	TNNT2 / HGNC:11949 / OMIM:191045 (troponin T2, cardiac type)
600692	TNNT3 / HGNC:11950 / OMIM:600692 (troponin T3, fast skeletal type)
610032	TNPO3 / HGNC:17103 / OMIM:610032 (transportin 3)
601243	TOP3A / HGNC:11992 / OMIM:601243 (DNA topoisomerase III alpha)
605204	TOR1A / HGNC:3098 / OMIM:605204 (torsin family 1 member A)
614512	TOR1AIP1 / HGNC:29456 / OMIM:614512 (torsin 1A interacting protein 1)
191010	TPM1 / HGNC:12010 / OMIM:191010 (tropomyosin 1)
190990	TPM2 / HGNC:12011 / OMIM:190990 (tropomyosin 2)
191030	TPM3 / HGNC:12012 / OMIM:191030 (tropomyosin 3)
614138	TRAPPC11 / HGNC:25751 / OMIM:614138 (trafficking protein particle complex subunit 11)
603283	TRDN / HGNC:12261 / OMIM:603283 (triadin)
614141	TRIM2 / HGNC:15974 / OMIM:614141 (tripartite motif containing 2)
602290	TRIM32 / HGNC:16380 / OMIM:602290

	(tripartite motif containing 32)
606474	TRIM54 / HGNC:16008 / OMIM:606474 (tripartite motif containing 54)
606131	TRIM63 / HGNC:16007 / OMIM:606131 (tripartite motif containing 63)
604501	TRIP4 / HGNC:12310 / OMIM:604501 (thyroid hormone receptor interactor 4)
602345	TRPC3 / HGNC:12335 / OMIM:602345 (transient receptor potential cation channel subfamily C member 3)
605427	TRPV4 / HGNC:18083 / OMIM:605427 (transient receptor potential cation channel subfamily V member 4)
604723	TSFM / HGNC:12367 / OMIM:604723 (Ts translation elongation factor, mitochondrial)
611695	TTBK2 / HGNC:19141 / OMIM:611695 (tau tubulin kinase 2)
188840	TTN / HGNC:12403 / OMIM:188840 (titin)
600415	TTPA / HGNC:12404 / OMIM:600415 (alpha tocopherol transfer protein)
176300	TTR / HGNC:12405 / OMIM:176300 (transthyretin)
191110	TUBA4A / HGNC:12407 / OMIM:191110 (tubulin alpha 4a)
602661	TUBB3 / HGNC:20772 / OMIM:602661 (tubulin beta 3 class III)
606075	TWINK / HGNC:1160 / OMIM:606075 (twinkle mtDNA helicase)
131222	TYMP / HGNC:3148 / OMIM:131222 (thymidine phosphorylase)
314370	UBA1 / HGNC:12469 / OMIM:314370 (ubiquitin like modifier activating enzyme 1)
609787	UBAP1 / HGNC:12461 / OMIM:609787 (ubiquitin associated protein 1)
300264	UBQLN2 / HGNC:12509 / OMIM:300264 (ubiquilin 2)
191342	UCHL1 / HGNC:12513 / OMIM:191342 (ubiquitin C-terminal hydrolase L1)
605836	UNC13B / HGNC:12566 / OMIM:605836 (unc-13 homolog B)
611220	UNC45B / HGNC:14304 / OMIM:611220 (unc-45 myosin chaperone B)
185880	VAMP1 / HGNC:12642 / OMIM:185880 (vesicle associated membrane protein 1)
605704	VAPB / HGNC:12649 / OMIM:605704 (VAMP associated protein B and C)
193065	VCL / HGNC:12665 / OMIM:193065 (vinculin)
601023	VCP / HGNC:12666 / OMIM:601023 (valosin containing protein)
300913	VMA21 / HGNC:22082 / OMIM:300913 (vacuolar ATPase assembly factor VMA21)
608877	VPS13D / HGNC:23595 / OMIM:608877 (vacuolar protein sorting 13 homolog D)
609927	VPS37A / HGNC:24928 / OMIM:609927

			<table border="1"> <tr> <td></td> <td>(VPS37A subunit of ESCRT-I)</td> </tr> <tr> <td>602168</td> <td>VRK1 / HGNC:12718 / OMIM:602168 (VRK serine/threonine kinase 1)</td> </tr> <tr> <td>614884</td> <td>VWA3B / HGNC:28385 / OMIM:614884 (von Willebrand factor A domain containing 3B)</td> </tr> <tr> <td>191050</td> <td>WARS1 / HGNC:12729 / OMIM:191050 (tryptophanyl-tRNA synthetase 1)</td> </tr> <tr> <td>605232</td> <td>WNK1 / HGNC:14540 / OMIM:605232 (WNK lysine deficient protein kinase 1)</td> </tr> <tr> <td>605131</td> <td>WWOX / HGNC:12799 / OMIM:605131 (WW domain containing oxidoreductase)</td> </tr> <tr> <td>194360</td> <td>XRCC1 / HGNC:12828 / OMIM:194360 (X-ray repair cross complementing 1)</td> </tr> <tr> <td>603623</td> <td>YARS1 / HGNC:12840 / OMIM:603623 (tyrosyl-tRNA synthetase 1)</td> </tr> <tr> <td>610957</td> <td>YARS2 / HGNC:24249 / OMIM:610957 (tyrosyl-tRNA synthetase 2)</td> </tr> <tr> <td>613915</td> <td>ZBTB42 / HGNC:32550 / OMIM:613915 (zinc finger and BTB domain containing 42)</td> </tr> <tr> <td>617828</td> <td>ZFH2 / HGNC:20152 / OMIM:617828 (zinc finger homeobox 2)</td> </tr> <tr> <td>612012</td> <td>ZFYVE26 / HGNC:20761 / OMIM:612012 (zinc finger FYVE-type containing 26)</td> </tr> <tr> <td>610243</td> <td>ZFYVE27 / HGNC:26559 / OMIM:610243 (zinc finger FYVE-type containing 27)</td> </tr> </table>		(VPS37A subunit of ESCRT-I)	602168	VRK1 / HGNC:12718 / OMIM:602168 (VRK serine/threonine kinase 1)	614884	VWA3B / HGNC:28385 / OMIM:614884 (von Willebrand factor A domain containing 3B)	191050	WARS1 / HGNC:12729 / OMIM:191050 (tryptophanyl-tRNA synthetase 1)	605232	WNK1 / HGNC:14540 / OMIM:605232 (WNK lysine deficient protein kinase 1)	605131	WWOX / HGNC:12799 / OMIM:605131 (WW domain containing oxidoreductase)	194360	XRCC1 / HGNC:12828 / OMIM:194360 (X-ray repair cross complementing 1)	603623	YARS1 / HGNC:12840 / OMIM:603623 (tyrosyl-tRNA synthetase 1)	610957	YARS2 / HGNC:24249 / OMIM:610957 (tyrosyl-tRNA synthetase 2)	613915	ZBTB42 / HGNC:32550 / OMIM:613915 (zinc finger and BTB domain containing 42)	617828	ZFH2 / HGNC:20152 / OMIM:617828 (zinc finger homeobox 2)	612012	ZFYVE26 / HGNC:20761 / OMIM:612012 (zinc finger FYVE-type containing 26)	610243	ZFYVE27 / HGNC:26559 / OMIM:610243 (zinc finger FYVE-type containing 27)
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585	<p>[ <a href="#">undiag_hgnc_ont</a> ]</p> <p>Show the field ONLY if: [undiag_report_variant] = 'yes' AND [undiag_listpref]='ontology'</p>	<p>Gene HGNC-code (browse ontology)If the affected gene is not present in the neuromuscular gene list above (e.g. a new gene not previously associated with neuromuscular disease) please search for the gene at <a href="https://www.genenames.org/tools/search/">https://www.genenames.org/tools/search/</a> and enter the numerical HGNC ID here (e.g. for DMD HGNC:2928 you would enter HGNC_2928)</p> <p><i>Element 6.3. in the Set of common data elements for Rare Diseases Registration (link)</i></p>	<p>text, Required</p> <table border="1"> <tr> <td>BIOPORTAL:HGNC</td> <td>BIOPORTAL:HGNC</td> </tr> </table>	BIOPORTAL:HGNC	BIOPORTAL:HGNC																								
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586	<p>[ <a href="#">undiag_omim_ont</a> ]</p> <p>Show the field ONLY if: [undiag_report_variant] = 'yes' AND [undiag_listpref]='ontology'</p>	<p>Gene OMIM-code (browse ontology)If the affected gene is not present in the neuromuscular gene list above (e.g. a new gene not previously associated with neuromuscular disease) please search for the gene at <a href="https://www.omim.org/">https://www.omim.org/</a> and enter the numerical OMIM ID here (e.g. for DMD you would enter 300377; you also find the code for this example if you type 'Dystrophin' to the box on the right)</p> <p><i>Element 6.3. in the Set of common data elements for Rare Diseases Registration (link)</i></p>	<p>text, Required</p> <table border="1"> <tr> <td>BIOPORTAL:OMIM</td> <td>BIOPORTAL:OMIM</td> </tr> </table>	BIOPORTAL:OMIM	BIOPORTAL:OMIM																								
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587	<p>[ <a href="#">undiag_omim</a> ]</p> <p>Show the field ONLY if: [undiag_report_variant] = 'yes'</p>	<p>Gene OMIM code calculated field</p>	<p>calc</p> <p>Calculation: if([undiag_report_variant] = 'yes', if([undiag_listpref]='ontology', [undiag_omim_ont], if([undiag_listpref]='all_nmd', [undiag_omim_armx], "")), "")</p> <p>Field Annotation: @READONLY @HIDDEN @HIDDEN-PDF</p>																										
588	<p>[ <a href="#">undiag_v1_report</a> ]</p> <p>Show the field ONLY if: [undiag_report_variant] = 'yes'</p>	<p>Section Header: <i>Variant 1</i></p> <p>How would you like to describe variant 1?Note: We prefer NCBI ClinVar but if the change in question is not yet described there, you can give a dbSNP ID if available, or you can type in the protein or cDNA change given in the genetic report.</p>	<p>radio, Required</p> <table border="1"> <tr> <td>clinvar</td> <td>NCBI ClinVar</td> </tr> <tr> <td>dbSNP</td> <td>dbSNP</td> </tr> <tr> <td>custom_hgvs</td> <td>Custom HGVS-simple notation</td> </tr> </table>	clinvar	NCBI ClinVar	dbSNP	dbSNP	custom_hgvs	Custom HGVS-simple notation																				
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custom_hgvs	Custom HGVS-simple notation																												
589	<p>[ <a href="#">undiag_v1_clinvar_uri</a> ]</p> <p>Show the field ONLY if: [undiag_v1_report] = 'clinvar'</p>	<p>NCBI ClinVar URI (please provide website-link)</p> <p><i>If the change is described in the NCBI ClinVar database, please provide website-link (e.g. <a href="https://www.ncbi.nlm.nih.gov/clinvar/variation/4886/">https://www.ncbi.nlm.nih.gov/clinvar/variation/4886/</a>)</i></p>	<p>text</p>																										

590	[ <a href="#">undiag_v1_snp_id</a> ] Show the field ONLY if: [undiag_v1_report] = 'dbsnp'	dbSNP RS ID <i>If the change has a dbSNP ID, enter it here, e.g. 'rs606231128'</i>	text												
591	[ <a href="#">undiag_v1_hgvs_opts</a> ] Show the field ONLY if: [undiag_v1_report] = 'custom_hgvs'	Variant 1: What kind of change(s) would you like to report in "HGVS simple"-notation? Please enter all the details that you have available from the report – e.g. if the report lists both the cDNA and protein changes, please provide both.	checkbox, Required <table border="1"> <tr> <td>cdna</td> <td>undiag_v1_hgvs_opts__cdna</td> <td>Report cDNA char (c.)</td> </tr> <tr> <td>nucleotide</td> <td>undiag_v1_hgvs_opts__nucleotide</td> <td>Report nucleic acid char (g.)</td> </tr> <tr> <td>protein</td> <td>undiag_v1_hgvs_opts__protein</td> <td>Report protein char (p.)</td> </tr> <tr> <td>other</td> <td>undiag_v1_hgvs_opts__other</td> <td>Report other of change (e.g. (r.) c non-coding)</td> </tr> </table>	cdna	undiag_v1_hgvs_opts__cdna	Report cDNA char (c.)	nucleotide	undiag_v1_hgvs_opts__nucleotide	Report nucleic acid char (g.)	protein	undiag_v1_hgvs_opts__protein	Report protein char (p.)	other	undiag_v1_hgvs_opts__other	Report other of change (e.g. (r.) c non-coding)
cdna	undiag_v1_hgvs_opts__cdna	Report cDNA char (c.)													
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592	[ <a href="#">undiag_v1_refseq_c</a> ] Show the field ONLY if: [undiag_v1_hgvs_opts(cdna)] = 1	Section Header: <i>Variant 1: cDNA change</i> Reference sequence for cDNA change <i>Enter the RefSeq transcript ID with the prefix NM_ or NR_ etc., e.g. 'NM_173660.5'. More information and examples see here and here</i>	text, Required												
593	[ <a href="#">undiag_v1_desc_c</a> ] Show the field ONLY if: [undiag_v1_hgvs_opts(cdna)] = 1	Description of cDNA change <i>Enter the cDNA change in HGVS nomenclature, e.g. c.1124_1127dupTGCC or c.346G&gt;A More information and examples see here</i>	text, Required												
594	[ <a href="#">undiag_v1_hgvs_c</a> ] Show the field ONLY if: [undiag_v1_hgvs_opts(cdna)] = 1	undiag_v1_hgvs_c	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([undiag_v1_refseq_c]!=" and [undiag_v1_desc_c]!=", concat([undiag_v1_refseq_c], ':', [undiag_v1_desc_c]), ''))												
595	[ <a href="#">undiag_v1_hgvs_c_disp</a> ] Show the field ONLY if: [undiag_v1_hgvs_opts(cdna)] = 1	Resulting HGVS notation for variant 1 cDNA change: [undiag_v1_hgvs_c]	descriptive												
596	[ <a href="#">undiag_v1_refseq_g</a> ] Show the field ONLY if: [undiag_v1_hgvs_opts(nucleotide)] = 1	Section Header: <i>Variant 1: Nucleotide (genomic) change</i> Reference sequence for nucleotide (genomic) change <i>Enter the RefSeq transcript ID with the prefix NC_ or NG_ etc., e.g. 'NC_000023.10'. More information and examples see here and here</i>	text, Required												
597	[ <a href="#">undiag_v1_desc_g</a> ] Show the field ONLY if: [undiag_v1_hgvs_opts(nucleotide)] = 1	Description of nucleotide (genomic) change <i>Enter the nucleotide change in HGVS nomenclature, e.g. g.34805_34808dup More information and examples see here</i>	text, Required												
598	[ <a href="#">undiag_v1_hgvs_g</a> ] Show the field ONLY if: [undiag_v1_hgvs_opts(nucleotide)] = 1	undiag_v1_hgvs_g	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([undiag_v1_refseq_g]!=" and [undiag_v1_desc_g]!=", concat([undiag_v1_refseq_g], ':', [undiag_v1_desc_g]), ''))												
599	[ <a href="#">undiag_v1_hgvs_g_disp</a> ] Show the field ONLY if: [undiag_v1_hgvs_opts(nucleotide)] = 1	Resulting HGVS notation for variant 1 nucleotide (genomic) change:[undiag_v1_hgvs_g]	descriptive												
600	[ <a href="#">undiag_v1_refseq_p</a> ]	Section Header: <i>Variant 1: Protein change</i> Reference sequence for protein change	text, Required												

	Show the field ONLY if: [undiag_v1_hgvs_opts(protein)] = 1	Enter the RefSeq transcript ID with the prefix NP_, e.g. NP_003997.1 More information and examples see here and here																	
601	[undiag_v1_desc_p]  Show the field ONLY if: [undiag_v1_hgvs_opts(protein)] = 1	Description of protein change <i>Enter the protein change in HGVS nomenclature, e.g. p.Ala378fs More information and examples see here</i>	text, Required																
602	[undiag_v1_hgvs_p]  Show the field ONLY if: [undiag_v1_hgvs_opts(protein)] = 1	undiag_v1_hgvs_p	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([undiag_v1_refseq_p]!=" and [undiag_v1_desc_p]!=", concat([undiag_v1_refseq_p], ':', [undiag_v1_desc_p]), ''))																
603	[undiag_v1_hgvs_p_disp]  Show the field ONLY if: [undiag_v1_hgvs_opts(protein)] = 1	Resulting HGVS notation for variant 1 protein change: [undiag_v1_hgvs_p]	descriptive																
604	[undiag_v1_refseq_o]  Show the field ONLY if: [undiag_v1_hgvs_opts(other)] = 1	Section Header: <i>Variant 1: Other change</i> Reference sequence for other change <i>More information and examples see here and here</i>	text, Required																
605	[undiag_v1_desc_o]  Show the field ONLY if: [undiag_v1_hgvs_opts(other)] = 1	Description of other change <i>Examples for descriptions of RNA changes: r.357_358ins357+1_357+12 or r.11u&gt;g More information and examples see here</i>	text, Required																
606	[undiag_v1_hgvs_o]  Show the field ONLY if: [undiag_v1_hgvs_opts(other)] = 1	undiag_v1_hgvs_o	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([undiag_v1_refseq_o]!=" and [undiag_v1_desc_o]!=", concat([undiag_v1_refseq_o], ':', [undiag_v1_desc_o]), ''))																
607	[undiag_v1_hgvs_o_disp]  Show the field ONLY if: [undiag_v1_hgvs_opts(other)] = 1	Resulting HGVS notation for variant 1 other change: [undiag_v1_hgvs_o]	descriptive																
608	[undiag_v1_zygos]  Show the field ONLY if: [undiag_report_variant] = 'yes'	Section Header: <i>Variant 1: Additional Information</i> Zygoty	dropdown <table border="1"> <tr> <td>heterozygous</td> <td>Heterozygous</td> </tr> <tr> <td>compound_heterozygous</td> <td>Compound heterozygous</td> </tr> <tr> <td>homozygous</td> <td>Homozygous</td> </tr> <tr> <td>hemizygous</td> <td>Hemizygous</td> </tr> <tr> <td>mosaic</td> <td>Mosaic</td> </tr> <tr> <td>unknown</td> <td>Unknown</td> </tr> </table>	heterozygous	Heterozygous	compound_heterozygous	Compound heterozygous	homozygous	Homozygous	hemizygous	Hemizygous	mosaic	Mosaic	unknown	Unknown				
heterozygous	Heterozygous																		
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unknown	Unknown																		
609	[undiag_v1_gen_build]  Show the field ONLY if: [undiag_report_variant] = 'yes' AND [undiag_v1_report] = 'custom_hgvs'	Genomic build	dropdown <table border="1"> <tr> <td>GRCh38</td> <td>GRCh38 (hg38)</td> </tr> <tr> <td>GRCh37</td> <td>GRCh37 (hg19)</td> </tr> </table>	GRCh38	GRCh38 (hg38)	GRCh37	GRCh37 (hg19)												
GRCh38	GRCh38 (hg38)																		
GRCh37	GRCh37 (hg19)																		
610	[undiag_v1_gen_app]  Show the field ONLY if: [undiag_report_variant] = 'yes' AND [undiag_v1_report] = 'custom_hgvs'	Genetic approach	dropdown <table border="1"> <tr> <td>single_gene_screening</td> <td>Single Gene Screening</td> </tr> <tr> <td>acgh</td> <td>aCGH</td> </tr> <tr> <td>snp_array</td> <td>SNP array</td> </tr> <tr> <td>mlpa</td> <td>MLPA</td> </tr> <tr> <td>karyotype</td> <td>Karyotype</td> </tr> <tr> <td>ngs_panel</td> <td>NGS panel</td> </tr> <tr> <td>wes</td> <td>WES</td> </tr> <tr> <td>wgs</td> <td>WGS</td> </tr> </table>	single_gene_screening	Single Gene Screening	acgh	aCGH	snp_array	SNP array	mlpa	MLPA	karyotype	Karyotype	ngs_panel	NGS panel	wes	WES	wgs	WGS
single_gene_screening	Single Gene Screening																		
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611	<p>[ <b>undiag_v1_chrom</b> ]</p> <p>Show the field ONLY if: [undiag_report_variant] = 'yes' AND [undiag_v1_report] = 'custom_hgvs'</p>	Chromosome	<p>dropdown</p> <table border="1"> <tr><td>1</td><td>1</td></tr> <tr><td>2</td><td>2</td></tr> <tr><td>3</td><td>3</td></tr> <tr><td>4</td><td>4</td></tr> <tr><td>5</td><td>5</td></tr> <tr><td>6</td><td>6</td></tr> <tr><td>7</td><td>7</td></tr> <tr><td>8</td><td>8</td></tr> <tr><td>9</td><td>9</td></tr> <tr><td>10</td><td>10</td></tr> <tr><td>11</td><td>11</td></tr> <tr><td>12</td><td>12</td></tr> <tr><td>13</td><td>13</td></tr> <tr><td>14</td><td>14</td></tr> <tr><td>15</td><td>15</td></tr> <tr><td>16</td><td>16</td></tr> <tr><td>17</td><td>17</td></tr> <tr><td>18</td><td>18</td></tr> <tr><td>19</td><td>19</td></tr> <tr><td>20</td><td>20</td></tr> <tr><td>21</td><td>21</td></tr> <tr><td>22</td><td>22</td></tr> <tr><td>X</td><td>X</td></tr> <tr><td>Y</td><td>Y</td></tr> <tr><td>MT</td><td>MT</td></tr> </table>	1	1	2	2	3	3	4	4	5	5	6	6	7	7	8	8	9	9	10	10	11	11	12	12	13	13	14	14	15	15	16	16	17	17	18	18	19	19	20	20	21	21	22	22	X	X	Y	Y	MT	MT
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612	<p>[ <b>undiag_desc3</b> ]</p> <p>Show the field ONLY if: [undiag_report_variant] = 'yes'</p>	<p>Section Header: <i>Variant 2 (if applicable)</i></p> <p>Please report only an additional variant for the gene selected above (selected gene-OMIM: [undiag_omim]). If you wish to report a variant for another gene, please report that in a separate 'Undiagnosed'-form for that gene.</p>	descriptive																																																		
613	<p>[ <b>undiag_v2_yes</b> ]</p> <p>Show the field ONLY if: [undiag_report_variant] = 'yes'</p>	Report a second variant for gene-OMIM: [undiag_omim]	<p>radio</p> <table border="1"> <tr><td>yes</td><td>Yes</td></tr> <tr><td>not_present</td><td>Not present</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	yes	Yes	not_present	Not present	unknown	Unknown																																												
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not_present	Not present																																																				
unknown	Unknown																																																				
614	<p>[ <b>undiag_v2_report</b> ]</p> <p>Show the field ONLY if: [undiag_v2_yes] = 'yes'</p>	<p>How would you like to describe variant 2? Note: We prefer NCBI ClinVar but if the change in question is not yet described there, you can give a dbSNP ID if available, or you can type in the protein or cDNA change given in the genetic report.</p>	<p>radio, Required</p> <table border="1"> <tr><td>clinvar</td><td>NCBI ClinVar</td></tr> <tr><td>dbSNP</td><td>dbSNP</td></tr> <tr><td>custom_hgvs</td><td>Custom HGVS-simple notation</td></tr> </table>	clinvar	NCBI ClinVar	dbSNP	dbSNP	custom_hgvs	Custom HGVS-simple notation																																												
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615	<p>[ <b>undiag_v2_clinvar_uri</b> ]</p> <p>Show the field ONLY if: [undiag_v2_report] = 'clinvar'</p>	<p>NCBI ClinVar URI (please provide website-link) <i>If the change is described in the NCBI ClinVar database, please provide website-link (e.g. <a href="https://www.ncbi.nlm.nih.gov/clinvar/variation/4886/">https://www.ncbi.nlm.nih.gov/clinvar/variation/4886/</a>)</i></p>	text																																																		
616	<p>[ <b>undiag_v2_snp_id</b> ]</p> <p>Show the field ONLY if: [undiag_v2_report] = 'dbSNP'</p>	<p>dbSNP RS ID <i>If the change has a dbSNP ID, enter it here, e.g. 'rs606231128'</i></p>	text																																																		
617	<p>[ <b>undiag_v2_hgvs_opts</b> ]</p> <p>Show the field ONLY if: [undiag_v2_report] = 'custom_hgvs'</p>	<p>Variant 2: What kind of change(s) would you like to report in "HGVS simple"-notation? Please enter all the details that you have available from the report – e.g. if the report lists both the cDNA and protein changes, please provide both.</p>	<p>checkbox, Required</p> <table border="1"> <tr> <td>cdna</td> <td>undiag_v2_hgvs_opts__cdna</td> <td>Report cDNA char (c.)</td> </tr> <tr> <td>nucleotide</td> <td>undiag_v2_hgvs_opts__nucleotide</td> <td>Report</td> </tr> </table>	cdna	undiag_v2_hgvs_opts__cdna	Report cDNA char (c.)	nucleotide	undiag_v2_hgvs_opts__nucleotide	Report																																												
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				nucl (gen char (g.)
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618	[undiag_v2_refseq_c] Show the field ONLY if: [undiag_v2_hgvs_opts(cdna)] = 1	Section Header: <i>Variant 2: cDNA change</i> Reference sequence for cDNA change <i>Enter the RefSeq transcript ID with the prefix NM_ or NR_ etc., e.g. 'NM_173660.5'. More information and examples see here and here</i>	text, Required	
619	[undiag_v2_desc_c] Show the field ONLY if: [undiag_v2_hgvs_opts(cdna)] = 1	Description of cDNA change <i>Enter the cDNA change in HGVS nomenclature, e.g. c.1124_1127dupTGCC or c.346G&gt;A More information and examples see here</i>	text, Required	
620	[undiag_v2_hgvs_c] Show the field ONLY if: [undiag_v2_hgvs_opts(cdna)] = 1	undiag_v2_hgvs_c	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([undiag_v2_refseq_c]!=" and [undiag_v2_desc_c]!=", concat([undiag_v2_refseq_c], ':', [undiag_v2_desc_c]), ''))	
621	[undiag_v2_hgvs_c_disp] Show the field ONLY if: [undiag_v2_hgvs_opts(cdna)] = 1	Resulting HGVS notation for variant 1 cDNA change: [undiag_v2_hgvs_c]	descriptive	
622	[undiag_v2_refseq_g] Show the field ONLY if: [undiag_v2_hgvs_opts(nucleotide)] = 1	Section Header: <i>Variant 2: Nucleotide (genomic) change</i> Reference sequence for nucleotide (genomic) change <i>Enter the RefSeq transcript ID with the prefix NC_ or NG_ etc., e.g. 'NC_000023.10'. More information and examples see here and here</i>	text, Required	
623	[undiag_v2_desc_g] Show the field ONLY if: [undiag_v2_hgvs_opts(nucleotide)] = 1	Description of nucleotide (genomic) change <i>Enter the nucleotide change in HGVS nomenclature, e.g. g.34805_34808dup More information and examples see here</i>	text, Required	
624	[undiag_v2_hgvs_g] Show the field ONLY if: [undiag_v2_hgvs_opts(nucleotide)] = 1	undiag_v2_hgvs_g	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([undiag_v2_refseq_g]!=" and [undiag_v2_desc_g]!=", concat([undiag_v2_refseq_g], ':', [undiag_v2_desc_g]), ''))	
625	[undiag_v2_hgvs_g_disp] Show the field ONLY if: [undiag_v2_hgvs_opts(nucleotide)] = 1	Resulting HGVS notation for variant 1 nucleotide (genomic) change:[undiag_v2_hgvs_g]	descriptive	
626	[undiag_v2_refseq_p] Show the field ONLY if: [undiag_v2_hgvs_opts(protein)] = 1	Section Header: <i>Variant 2: Protein change</i> Reference sequence for protein change <i>Enter the RefSeq transcript ID with the prefix NP_, e.g. NP_003997.1 More information and examples see here and here</i>	text, Required	
627	[undiag_v2_desc_p] Show the field ONLY if: [undiag_v2_hgvs_opts(protein)] = 1	Description of protein change <i>Enter the protein change in HGVS nomenclature, e.g. p.Ala378fs More information and examples see here</i>	text, Required	
628	[undiag_v2_hgvs_p] Show the field ONLY if: [undiag_v2_hgvs_opts(protein)] = 1	undiag_v2_hgvs_p	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([undiag_v2_refseq_p]!=" and	

	[n]] = 1		[undiag_v2_desc_p]!="", concat([undiag_v2_refseq_p], ':', [undiag_v2_desc_p]), '')																
629	[undiag_v2_hgvs_p_disp] Show the field ONLY if: [undiag_v2_hgvs_opts(protein)] = 1	Resulting HGVS notation for variant 1 protein change: [undiag_v2_hgvs_p]	descriptive																
630	[undiag_v2_refseq_o] Show the field ONLY if: [undiag_v2_hgvs_opts(other)] = 1	Section Header: <i>Variant 2: Other change</i> Reference sequence for other change <i>Examples for RNA reference sequences: NC_000023.10(NM_004006.2) or LRG_199t1. More information and examples see here and here</i>	text, Required																
631	[undiag_v2_desc_o] Show the field ONLY if: [undiag_v2_hgvs_opts(other)] = 1	Description of other change <i>Examples for descriptions of RNA changes: r.357_358ins357+1_357+12 or r.11u&gt;g More information and examples see here</i>	text, Required																
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634	[undiag_v2_zygos] Show the field ONLY if: [undiag_report_variant] = 'yes' AND [undiag_v2_yes] = 'yes'	Section Header: <i>Variant 2: Additional Information</i> Zygoty	dropdown <table border="1"> <tr><td>heterozygous</td><td>Heterozygous</td></tr> <tr><td>compound_heterozygous</td><td>Compound heterozygous</td></tr> <tr><td>homozygous</td><td>Homozygous</td></tr> <tr><td>hemizygous</td><td>Hemizygous</td></tr> <tr><td>mosaic</td><td>Mosaic</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	heterozygous	Heterozygous	compound_heterozygous	Compound heterozygous	homozygous	Homozygous	hemizygous	Hemizygous	mosaic	Mosaic	unknown	Unknown				
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wes	WES																		
wgs	WGS																		
637	[undiag_v2_chrom] Show the field ONLY if: [undiag_report_variant] = 'yes' AND [undiag_v2_report] = 'custom_hgvs'	Chromosome	dropdown <table border="1"> <tr><td>1</td><td>1</td></tr> <tr><td>2</td><td>2</td></tr> <tr><td>3</td><td>3</td></tr> <tr><td>4</td><td>4</td></tr> <tr><td>5</td><td>5</td></tr> <tr><td>6</td><td>6</td></tr> <tr><td>7</td><td>7</td></tr> </table>	1	1	2	2	3	3	4	4	5	5	6	6	7	7		
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2	2																		
3	3																		
4	4																		
5	5																		
6	6																		
7	7																		

			<table border="1"> <tr><td>8</td><td>8</td></tr> <tr><td>9</td><td>9</td></tr> <tr><td>10</td><td>10</td></tr> <tr><td>11</td><td>11</td></tr> <tr><td>12</td><td>12</td></tr> <tr><td>13</td><td>13</td></tr> <tr><td>14</td><td>14</td></tr> <tr><td>15</td><td>15</td></tr> <tr><td>16</td><td>16</td></tr> <tr><td>17</td><td>17</td></tr> <tr><td>18</td><td>18</td></tr> <tr><td>19</td><td>19</td></tr> <tr><td>20</td><td>20</td></tr> <tr><td>21</td><td>21</td></tr> <tr><td>22</td><td>22</td></tr> <tr><td>X</td><td>X</td></tr> <tr><td>Y</td><td>Y</td></tr> <tr><td>MT</td><td>MT</td></tr> </table>	8	8	9	9	10	10	11	11	12	12	13	13	14	14	15	15	16	16	17	17	18	18	19	19	20	20	21	21	22	22	X	X	Y	Y	MT	MT
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X	X																																						
Y	Y																																						
MT	MT																																						
638	[undiagnosed_complete]	Section Header: <i>Form Status</i> Complete?	dropdown <table border="1"> <tr><td>0</td><td>Incomplete</td></tr> <tr><td>1</td><td>Unverified</td></tr> <tr><td>2</td><td>Complete</td></tr> </table>	0	Incomplete	1	Unverified	2	Complete																														
0	Incomplete																																						
1	Unverified																																						
2	Complete																																						

**Instrument: Medication (medication)**

Active languages: None

639	[medication_h1]	MEDICATION	descriptive
640	[medi_desc1]	NOTE: Please fill in this form more than once if the patient has interrupted taking the same medication in the meantime and do not replace start date in the existing form.	descriptive
641	[medi_desc2] Show the field ONLY if: [medi_status] = 'discontinued'	WARNING: You reported that this medication has already been discontinued. The current status of this form is: [medication_complete]. Are you sure you want to edit this form instead of filling an additional medication form?	descriptive
642	[date_of_assess_medi]	Date of assessment yyyy-mm-dd <i>Date of visit when the medication was documented. If this date is not known and cannot be estimated, please use the current date (date you enter this information here).</i>	text (date_ymd), Required
643	[medication_label]	Medication label <i>This is a label to identify this record to the registry user. It will be displayed on the patient's record home page together with the dosage and timeframe. You can name it anything you find helpful - consider something like the medication name.</i>	text
644	[medication_label_display]	medication_label_display	text Field Annotation: @CALCTEXT(concat([medication_label], '(', [medi_dosage], ',', [medi_dosage_unit], ',', [medi_freq], ',', if([medi_freq_timeunit] = 'other', [medi_freq_timeunit_custom], [medi_freq_timeunit]), ':', concat(right([date_of_medi_start],2), ',', mid([date_of_medi_start],6,2), ',', left([date_of_medi_start],4), '- ', if([medi_status]="ongoing", "ongoing", concat(right([date_of_medi_stop],2), ',', mid([date_of_medi_stop],6,2), ',', left([date_of_medi_stop],4)))))) @HIDDEN @HIDDEN-PDF
645	[medication_matrix]	Codesystem Code registered? Medication name Chemical	descriptive

		Entities of Biological Interest (CheBI) {medi_has_chebi_code} {medi_chebi_code} Medical Subject Headings (MeSH) {medi_has_mesh_code} {medi_mesh_code} ?									
646	[medi_has_chebi_code]	CheBI code registered for this medication	yesno, Required <table border="1"> <tr><td>1</td><td>Yes</td></tr> <tr><td>0</td><td>No</td></tr> </table>	1	Yes	0	No				
1	Yes										
0	No										
647	[medi_chebi_code] Show the field ONLY if: [medi_has_chebi_code]=1	CheBI-code	text, Required <table border="1"> <tr><td>BIOPORTAL:CHEBI</td><td>BIOPORTAL:CHEBI</td></tr> </table>	BIOPORTAL:CHEBI	BIOPORTAL:CHEBI						
BIOPORTAL:CHEBI	BIOPORTAL:CHEBI										
648	[medi_has_mesh_code]	MeSH code registered for this medication	yesno, Required <table border="1"> <tr><td>1</td><td>Yes</td></tr> <tr><td>0</td><td>No</td></tr> </table>	1	Yes	0	No				
1	Yes										
0	No										
649	[medi_mesh_code] Show the field ONLY if: [medi_has_mesh_code]=1	MeSH-code	text, Required <table border="1"> <tr><td>BIOPORTAL:MESH</td><td>BIOPORTAL:MESH</td></tr> </table>	BIOPORTAL:MESH	BIOPORTAL:MESH						
BIOPORTAL:MESH	BIOPORTAL:MESH										
650	[is_specific_treatment_nmd]	Is this medication a specific treatment for the neuromuscular disease? <i>If this medication is used to treat other symptoms of the patient, please select "No".</i>	yesno, Required <table border="1"> <tr><td>1</td><td>Yes</td></tr> <tr><td>0</td><td>No</td></tr> </table>	1	Yes	0	No				
1	Yes										
0	No										
651	[treatment_effect] Show the field ONLY if: [is_specific_treatment_nmd]=1	Treatment effect <i>Effect of the specific treatment for neuromuscular disease</i>	radio <table border="1"> <tr><td>positive</td><td>Positive</td></tr> <tr><td>negative</td><td>Negative</td></tr> <tr><td>none</td><td>None</td></tr> <tr><td>unknown</td><td>Unknown</td></tr> </table>	positive	Positive	negative	Negative	none	None	unknown	Unknown
positive	Positive										
negative	Negative										
none	None										
unknown	Unknown										
652	[is_cardiac_medication]	Cardiac medication <i>Is this medication a cardiac medication?</i>	yesno, Required <table border="1"> <tr><td>1</td><td>Yes</td></tr> <tr><td>0</td><td>No</td></tr> </table>	1	Yes	0	No				
1	Yes										
0	No										
653	[date_of_medi_start_i]	Section Header: <i>Timeframe</i> Medication start date imputation	radio, Required <table border="1"> <tr><td>no_imputation</td><td>Exact date of medication start known</td></tr> <tr><td>day</td><td>Day of medication start date unknown</td></tr> <tr><td>day_month</td><td>Day and month of medication start date unknown</td></tr> </table> <p>Field Annotation: @DEFAULT = 'no_imputation'</p>	no_imputation	Exact date of medication start known	day	Day of medication start date unknown	day_month	Day and month of medication start date unknown		
no_imputation	Exact date of medication start known										
day	Day of medication start date unknown										
day_month	Day and month of medication start date unknown										
654	[date_of_medi_start]	Medication start date yyyy-mm-dd <i>On what date did the patient start taking this medication? If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.</i>	text (date_ymd), Required								
655	[medi_status]	Medication status	radio, Required <table border="1"> <tr><td>ongoing</td><td>Medication ongoing</td></tr> <tr><td>discontinued</td><td>Medication discontinued</td></tr> </table> <p>Field Annotation: @DEFAULT = 'ongoing'</p>	ongoing	Medication ongoing	discontinued	Medication discontinued				
ongoing	Medication ongoing										
discontinued	Medication discontinued										
656	[date_of_medi_stop_i] Show the field ONLY if: [medi_status] = 'discontinued'	Medication end date imputation	radio, Required <table border="1"> <tr><td>no_imputation</td><td>Exact date of medication discontinuation known</td></tr> <tr><td>day</td><td>Day of medication end date unknown</td></tr> <tr><td>day_month</td><td>Day and month of medication end date unknown</td></tr> </table>	no_imputation	Exact date of medication discontinuation known	day	Day of medication end date unknown	day_month	Day and month of medication end date unknown		
no_imputation	Exact date of medication discontinuation known										
day	Day of medication end date unknown										
day_month	Day and month of medication end date unknown										
657	[date_of_medi_stop] Show the field ONLY if: [medi_status] = 'discontinued'	Medication end date yyyy-mm-dd <i>On what date did the patient stop taking this medication? If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of</i>	text (date_ymd), Required								


	d'	July of that year.																											
658	[medi_dosage_known]	Section Header: Dosage Dosage known	radio, Required <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>not_applicable</td> <td>Not applicable</td> </tr> </table> Field Annotation: @DEFAULT = 'yes'	yes	Yes	no	No	not_applicable	Not applicable																				
yes	Yes																												
no	No																												
not_applicable	Not applicable																												
659	[medi_dosage] Show the field ONLY if: [medi_dosage_known] = 'yes'	Dosage <i>If the same treatment is administered several times in different doses on the same day, please indicate the cumulative dose.</i>	text (number), Required																										
660	[medi_dosage_unit] Show the field ONLY if: [medi_dosage_known] = 'yes'	Dosage unit	dropdown, Required <table border="1"> <tr> <td>g</td> <td>g (gram)</td> </tr> <tr> <td>mg</td> <td>mg (milligram)</td> </tr> <tr> <td>microg</td> <td>µg (microgramm)</td> </tr> <tr> <td>mmol</td> <td>mmol (millimol)</td> </tr> <tr> <td>micromol</td> <td>µmol (micromol)</td> </tr> <tr> <td>ml</td> <td>ml (millilitre)</td> </tr> <tr> <td>microl</td> <td>µl (microlitre)</td> </tr> <tr> <td>sessions</td> <td>session(s)</td> </tr> </table>	g	g (gram)	mg	mg (milligram)	microg	µg (microgramm)	mmol	mmol (millimol)	micromol	µmol (micromol)	ml	ml (millilitre)	microl	µl (microlitre)	sessions	session(s)										
g	g (gram)																												
mg	mg (milligram)																												
microg	µg (microgramm)																												
mmol	mmol (millimol)																												
micromol	µmol (micromol)																												
ml	ml (millilitre)																												
microl	µl (microlitre)																												
sessions	session(s)																												
661	[medi_admin_freq_known]	Frequency of administration known	radio, Required <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> <tr> <td>not_applicable</td> <td>Not applicable</td> </tr> </table> Field Annotation: @DEFAULT = 'yes'	yes	Yes	no	No	not_applicable	Not applicable																				
yes	Yes																												
no	No																												
not_applicable	Not applicable																												
662	[medi_freq] Show the field ONLY if: [medi_admin_freq_known] = 'yes'	Frequency of administration	text (number), Required																										
663	[medi_freq_timeunit] Show the field ONLY if: [medi_admin_freq_known] = 'yes'	Frequency of administration: time unit	dropdown (autocomplete), Required <table border="1"> <tr> <td>once</td> <td>once</td> </tr> <tr> <td>per_hour</td> <td>per hour</td> </tr> <tr> <td>per_day</td> <td>per day</td> </tr> <tr> <td>per_week</td> <td>per week</td> </tr> <tr> <td>t2weeks</td> <td>every 2 weeks</td> </tr> <tr> <td>t3weeks</td> <td>every 3 weeks</td> </tr> <tr> <td>t4weeks</td> <td>every 4 weeks</td> </tr> <tr> <td>t6weeks</td> <td>every 6 weeks</td> </tr> <tr> <td>t8weeks</td> <td>every 8 weeks</td> </tr> <tr> <td>t3months</td> <td>every 3 months</td> </tr> <tr> <td>t4months</td> <td>every 4 months</td> </tr> <tr> <td>t6months</td> <td>every 6 months</td> </tr> <tr> <td>other</td> <td>other</td> </tr> </table>	once	once	per_hour	per hour	per_day	per day	per_week	per week	t2weeks	every 2 weeks	t3weeks	every 3 weeks	t4weeks	every 4 weeks	t6weeks	every 6 weeks	t8weeks	every 8 weeks	t3months	every 3 months	t4months	every 4 months	t6months	every 6 months	other	other
once	once																												
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t8weeks	every 8 weeks																												
t3months	every 3 months																												
t4months	every 4 months																												
t6months	every 6 months																												
other	other																												
664	[medi_freq_timeunit_custom] Show the field ONLY if: [medi_admin_freq_known] = 'yes' AND [medi_freq_timeunit] = 'other'	Frequency of administration: custom time unit	text, Required																										
665	[medication_complete]	Section Header: Form Status Complete?	dropdown <table border="1"> <tr> <td>0</td> <td>Incomplete</td> </tr> <tr> <td>1</td> <td>Unverified</td> </tr> </table>	0	Incomplete	1	Unverified																						
0	Incomplete																												
1	Unverified																												

Instrument: **WHODAS 2.0 12-item** (whodas\_20\_12item)  Enabled as survey

Active languages - Data Entry: None | Survey: None

666	[whodas_title]	WHODAS 2.0 12-item self-administered	descriptive										
667	[whodas_desc1]	This questionnaire asks about difficulties due to health conditions. Health conditions include diseases or illnesses, other health problems that may be short or long lasting, injuries, mental or emotional problems, and problems with alcohol or drugs. Think back over the past 30 days and answer these questions, thinking about how much difficulty you had doing the following activities.	descriptive										
668	[date_of_whodas]	Current Date yyyy-mm-dd	text (date_ymd), Required										
669	[whodas_answered_by]	Who answers this survey?	radio <table border="1"> <tr> <td>patient</td> <td>Patient</td> </tr> <tr> <td>caregiver</td> <td>Caregiver (on behalf of the patient)</td> </tr> </table>	patient	Patient	caregiver	Caregiver (on behalf of the patient)						
patient	Patient												
caregiver	Caregiver (on behalf of the patient)												
670	[whodas_subscores_known]	Result of the individual data items is known	radio, Required <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> </table> Field Annotation: @HIDDEN-SURVEY @DEFAULT = 'yes'	yes	Yes	no	No						
yes	Yes												
no	No												
671	[whodas_score_manual] Show the field ONLY if: [whodas_subscores_known] = 'no'	Manual Overall WHODAS-Score [%] <i>Please enter a number in the range [0, 100]. Element 8.1. in the Set of common data elements for Rare Diseases Registration (link)</i>	text (number, Min: 0, Max: 100), Required Field Annotation: @HIDDEN-SURVEY										
672	[whodas_desc2] Show the field ONLY if: [whodas_subscores_known] = 'yes'	In the past 30 days, how much difficulty did you have in:	descriptive										
673	[whodas_s1] Show the field ONLY if: [whodas_subscores_known] = 'yes'	Standing for long periods such as 30 minutes?	radio, Required <table border="1"> <tr> <td>0</td> <td>None</td> </tr> <tr> <td>1</td> <td>Mild</td> </tr> <tr> <td>2</td> <td>Moderate</td> </tr> <tr> <td>3</td> <td>Severe</td> </tr> <tr> <td>4</td> <td>Extreme or cannot do</td> </tr> </table> Custom alignment: LH	0	None	1	Mild	2	Moderate	3	Severe	4	Extreme or cannot do
0	None												
1	Mild												
2	Moderate												
3	Severe												
4	Extreme or cannot do												
674	[whodas_s2] Show the field ONLY if: [whodas_subscores_known] = 'yes'	Taking care of your household responsibilities?	radio, Required <table border="1"> <tr> <td>0</td> <td>None</td> </tr> <tr> <td>1</td> <td>Mild</td> </tr> <tr> <td>2</td> <td>Moderate</td> </tr> <tr> <td>3</td> <td>Severe</td> </tr> <tr> <td>4</td> <td>Extreme or cannot do</td> </tr> </table> Custom alignment: LH	0	None	1	Mild	2	Moderate	3	Severe	4	Extreme or cannot do
0	None												
1	Mild												
2	Moderate												
3	Severe												
4	Extreme or cannot do												
675	[whodas_s3] Show the field ONLY if: [whodas_subscores_known] = 'yes'	Learning a new task, for example, learning how to get to a new place?	radio, Required <table border="1"> <tr> <td>0</td> <td>None</td> </tr> <tr> <td>1</td> <td>Mild</td> </tr> <tr> <td>2</td> <td>Moderate</td> </tr> <tr> <td>3</td> <td>Severe</td> </tr> <tr> <td>4</td> <td>Extreme or cannot do</td> </tr> </table> Custom alignment: LH	0	None	1	Mild	2	Moderate	3	Severe	4	Extreme or cannot do
0	None												
1	Mild												
2	Moderate												
3	Severe												
4	Extreme or cannot do												
676	[whodas_s4]	How much of a problem did you have joining in community	radio, Required										

	Show the field ONLY if: [whodas_subscores_known] = 'yes'	activities (for example, festivities, religious or other activities) in the same way as anyone else can?	<table border="1"> <tr><td>0</td><td>None</td></tr> <tr><td>1</td><td>Mild</td></tr> <tr><td>2</td><td>Moderate</td></tr> <tr><td>3</td><td>Severe</td></tr> <tr><td>4</td><td>Extreme or cannot do</td></tr> </table> <p>Custom alignment: LH</p>	0	None	1	Mild	2	Moderate	3	Severe	4	Extreme or cannot do
0	None												
1	Mild												
2	Moderate												
3	Severe												
4	Extreme or cannot do												
677	[whodas_s5] Show the field ONLY if: [whodas_subscores_known] = 'yes'	How much have you been emotionally affected by your health problems?	<p>radio, Required</p> <table border="1"> <tr><td>0</td><td>None</td></tr> <tr><td>1</td><td>Mild</td></tr> <tr><td>2</td><td>Moderate</td></tr> <tr><td>3</td><td>Severe</td></tr> <tr><td>4</td><td>Extreme or cannot do</td></tr> </table> <p>Custom alignment: LH</p>	0	None	1	Mild	2	Moderate	3	Severe	4	Extreme or cannot do
0	None												
1	Mild												
2	Moderate												
3	Severe												
4	Extreme or cannot do												
678	[whodas_s6] Show the field ONLY if: [whodas_subscores_known] = 'yes'	Concentrating on doing something for ten minutes?	<p>radio, Required</p> <table border="1"> <tr><td>0</td><td>None</td></tr> <tr><td>1</td><td>Mild</td></tr> <tr><td>2</td><td>Moderate</td></tr> <tr><td>3</td><td>Severe</td></tr> <tr><td>4</td><td>Extreme or cannot do</td></tr> </table> <p>Custom alignment: LH</p>	0	None	1	Mild	2	Moderate	3	Severe	4	Extreme or cannot do
0	None												
1	Mild												
2	Moderate												
3	Severe												
4	Extreme or cannot do												
679	[whodas_s7] Show the field ONLY if: [whodas_subscores_known] = 'yes'	Walking a long distance such as a kilometre [or equivalent]?	<p>radio, Required</p> <table border="1"> <tr><td>0</td><td>None</td></tr> <tr><td>1</td><td>Mild</td></tr> <tr><td>2</td><td>Moderate</td></tr> <tr><td>3</td><td>Severe</td></tr> <tr><td>4</td><td>Extreme or cannot do</td></tr> </table> <p>Custom alignment: LH</p>	0	None	1	Mild	2	Moderate	3	Severe	4	Extreme or cannot do
0	None												
1	Mild												
2	Moderate												
3	Severe												
4	Extreme or cannot do												
680	[whodas_s8] Show the field ONLY if: [whodas_subscores_known] = 'yes'	Washing your whole body?	<p>radio, Required</p> <table border="1"> <tr><td>0</td><td>None</td></tr> <tr><td>1</td><td>Mild</td></tr> <tr><td>2</td><td>Moderate</td></tr> <tr><td>3</td><td>Severe</td></tr> <tr><td>4</td><td>Extreme or cannot do</td></tr> </table> <p>Custom alignment: LH</p>	0	None	1	Mild	2	Moderate	3	Severe	4	Extreme or cannot do
0	None												
1	Mild												
2	Moderate												
3	Severe												
4	Extreme or cannot do												
681	[whodas_s9] Show the field ONLY if: [whodas_subscores_known] = 'yes'	Getting dressed?	<p>radio, Required</p> <table border="1"> <tr><td>0</td><td>None</td></tr> <tr><td>1</td><td>Mild</td></tr> <tr><td>2</td><td>Moderate</td></tr> <tr><td>3</td><td>Severe</td></tr> <tr><td>4</td><td>Extreme or cannot do</td></tr> </table> <p>Custom alignment: LH</p>	0	None	1	Mild	2	Moderate	3	Severe	4	Extreme or cannot do
0	None												
1	Mild												
2	Moderate												
3	Severe												
4	Extreme or cannot do												
682	[whodas_s10] Show the field ONLY if: [whodas_subscores_known] = 'yes'	Dealing with people you do not know?	<p>radio, Required</p> <table border="1"> <tr><td>0</td><td>None</td></tr> <tr><td>1</td><td>Mild</td></tr> <tr><td>2</td><td>Moderate</td></tr> <tr><td>3</td><td>Severe</td></tr> <tr><td>4</td><td>Extreme or cannot do</td></tr> </table>	0	None	1	Mild	2	Moderate	3	Severe	4	Extreme or cannot do
0	None												
1	Mild												
2	Moderate												
3	Severe												
4	Extreme or cannot do												

			Custom alignment: LH										
683	[whodas_s11] Show the field ONLY if: [whodas_subscores_known] = 'yes'	Maintaining a friendship?	radio, Required <table border="1"> <tr><td>0</td><td>None</td></tr> <tr><td>1</td><td>Mild</td></tr> <tr><td>2</td><td>Moderate</td></tr> <tr><td>3</td><td>Severe</td></tr> <tr><td>4</td><td>Extreme or cannot do</td></tr> </table> Custom alignment: LH	0	None	1	Mild	2	Moderate	3	Severe	4	Extreme or cannot do
0	None												
1	Mild												
2	Moderate												
3	Severe												
4	Extreme or cannot do												
684	[whodas_s12] Show the field ONLY if: [whodas_subscores_known] = 'yes'	Your day-to-day work?	radio, Required <table border="1"> <tr><td>0</td><td>None</td></tr> <tr><td>1</td><td>Mild</td></tr> <tr><td>2</td><td>Moderate</td></tr> <tr><td>3</td><td>Severe</td></tr> <tr><td>4</td><td>Extreme or cannot do</td></tr> </table> Custom alignment: LH	0	None	1	Mild	2	Moderate	3	Severe	4	Extreme or cannot do
0	None												
1	Mild												
2	Moderate												
3	Severe												
4	Extreme or cannot do												
685	[whodas_h1] Show the field ONLY if: [whodas_subscores_known] = 'yes'	Overall, in the past 30 days, how many days were these difficulties present? <i>Record number of days</i>	text (number, Min: 0, Max: 30) Custom alignment: LH										
686	[whodas_h2] Show the field ONLY if: [whodas_subscores_known] = 'yes'	In the past 30 days, for how many days were you totally unable to carry out your usual activities or work because of any health condition? <i>Record number of days</i>	text (number, Min: 0, Max: 30) Custom alignment: LH										
687	[whodas_h3] Show the field ONLY if: [whodas_subscores_known] = 'yes'	In the past 30 days, not counting the days that you were totally unable, for how many days did you cut back or reduce your usual activities or work because of any health condition? <i>Record number of days</i>	text (number, Min: 0, Max: 30) Custom alignment: LH										
688	[whodas_sc_simple_calc] Show the field ONLY if: [whodas_subscores_known] = 'yes'	Calculated Overall WHODAS-Score [%] <i>Will only contain values if none of whodas_s1 - whodas_s12 is missing. Element 8.1. in the Set of common data elements for Rare Diseases Registration (link)</i>	calc Calculation: $if(!isblankormissingcode([whodas_s1]) AND !isblankormissingcode([whodas_s2]) AND !isblankormissingcode([whodas_s3]) AND !isblankormissingcode([whodas_s4]) AND !isblankormissingcode([whodas_s5]) AND !isblankormissingcode([whodas_s6]) AND !isblankormissingcode([whodas_s7]) AND !isblankormissingcode([whodas_s8]) AND !isblankormissingcode([whodas_s9]) AND !isblankormissingcode([whodas_s10]) AND !isblankormissingcode([whodas_s11]) AND !isblankormissingcode([whodas_s12]))$ , (round(sum([whodas_s1], [whodas_s2], [whodas_s3], [whodas_s4], [whodas_s5], [whodas_s6], [whodas_s7], [whodas_s8], [whodas_s9], [whodas_s10], [whodas_s11], [whodas_s12]) / 48 * 100, 2)), "") Field Annotation: @HIDDEN-SURVEY										
689	[whodas_pdf]	Auto-uploaded survey PDF	file Field Annotation: @HIDDEN-SURVEY @HIDDEN-PDF										
690	[whodas_20_12item_complete]	Section Header: Form Status Complete?	dropdown <table border="1"> <tr><td>0</td><td>Incomplete</td></tr> <tr><td>1</td><td>Unverified</td></tr> <tr><td>2</td><td>Complete</td></tr> </table>	0	Incomplete	1	Unverified	2	Complete				
0	Incomplete												
1	Unverified												
2	Complete												
Instrument: <b>PGI-C (only available if date of birth filled in Baseline)</b> (pgic_only_available_if_date_of_birth_filled_in_bas) 													
Enabled as survey													
Active languages - Data Entry: DE, EN, FR   Survey: DE, EN, FR													
691	[pgic_h1]	Patient Global Impression scale: Change (PGI-C)	descriptive										



692	[date_of_pgic]	Date of assessment yyyy-mm-dd	text (date_ymd), Required														
693	[age_at_pgic]	Age of patient (years)	calc Calculation: rounddown(datediff([first-event-name] [date_of_birth], [date_of_pgic], "y")) Field Annotation: @READONLY														
694	[pgic_answered_by]	Who answers this survey?	radio <table border="1"> <tr> <td>patient</td> <td>Patient</td> </tr> <tr> <td>caregiver</td> <td>Caregiver (on behalf of the patient)</td> </tr> </table>	patient	Patient	caregiver	Caregiver (on behalf of the patient)										
patient	Patient																
caregiver	Caregiver (on behalf of the patient)																
695	[pgic_1a] Show the field ONLY if: starts_with([event-name], 'e nrolment') AND [age_at_pgic < 18	How has your neuromuscular condition changed over the last 6 months?	radio <table border="1"> <tr> <td>1</td> <td>Very Much Improved</td> </tr> <tr> <td>2</td> <td>Much Improved</td> </tr> <tr> <td>3</td> <td>Minimally Improved</td> </tr> <tr> <td>4</td> <td>No Change</td> </tr> <tr> <td>5</td> <td>Minimally Worse</td> </tr> <tr> <td>6</td> <td>Much Worse</td> </tr> <tr> <td>7</td> <td>Very Much Worse</td> </tr> </table>	1	Very Much Improved	2	Much Improved	3	Minimally Improved	4	No Change	5	Minimally Worse	6	Much Worse	7	Very Much Worse
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7	Very Much Worse																
696	[pgic_1b] Show the field ONLY if: starts_with([event-name], 'e nrolment') AND [age_at_pgic >= 18	How has your neuromuscular condition changed over the last year?	radio <table border="1"> <tr> <td>1</td> <td>Very Much Improved</td> </tr> <tr> <td>2</td> <td>Much Improved</td> </tr> <tr> <td>3</td> <td>Minimally Improved</td> </tr> <tr> <td>4</td> <td>No Change</td> </tr> <tr> <td>5</td> <td>Minimally Worse</td> </tr> <tr> <td>6</td> <td>Much Worse</td> </tr> <tr> <td>7</td> <td>Very Much Worse</td> </tr> </table>	1	Very Much Improved	2	Much Improved	3	Minimally Improved	4	No Change	5	Minimally Worse	6	Much Worse	7	Very Much Worse
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7	Very Much Worse																
697	[pgic_2] Show the field ONLY if: starts_with([event-name], 'vi sit')	How has your neuromuscular condition changed since you were first enrolled in this patient registry on [first-event- name][date_of_consent]	radio <table border="1"> <tr> <td>1</td> <td>Very Much Improved</td> </tr> <tr> <td>2</td> <td>Much Improved</td> </tr> <tr> <td>3</td> <td>Minimally Improved</td> </tr> <tr> <td>4</td> <td>No Change</td> </tr> <tr> <td>5</td> <td>Minimally Worse</td> </tr> <tr> <td>6</td> <td>Much Worse</td> </tr> <tr> <td>7</td> <td>Very Much Worse</td> </tr> </table>	1	Very Much Improved	2	Much Improved	3	Minimally Improved	4	No Change	5	Minimally Worse	6	Much Worse	7	Very Much Worse
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7	Very Much Worse																
698	[pgic_3a] Show the field ONLY if: starts_with([event-name], 'vi sit')	Have you started a new treatment or medicine based on advice from your specialist doctor at this site since you last answered this questionnaire?	radio <table border="1"> <tr> <td>yes</td> <td>Yes</td> </tr> <tr> <td>no</td> <td>No</td> </tr> </table>	yes	Yes	no	No										
yes	Yes																
no	No																
699	[pgic_3b] Show the field ONLY if: [pgic_3a] = 'yes'	What new treatment or medicine are you thinking about?	text														
700	[pgic_3c] Show the field ONLY if: [pgic_3a] = 'yes'	How has your neuromuscular condition changed since you started your new treatment plan?	radio <table border="1"> <tr> <td>1</td> <td>Very Much Improved</td> </tr> <tr> <td>2</td> <td>Much Improved</td> </tr> <tr> <td>3</td> <td>Minimally Improved</td> </tr> <tr> <td>4</td> <td>No Change</td> </tr> <tr> <td>5</td> <td>Minimally Worse</td> </tr> <tr> <td>6</td> <td>Much Worse</td> </tr> <tr> <td>7</td> <td>Very Much Worse</td> </tr> </table>	1	Very Much Improved	2	Much Improved	3	Minimally Improved	4	No Change	5	Minimally Worse	6	Much Worse	7	Very Much Worse
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701	[pgic_3b_prev]	pgic_3b_prev	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT([event-name][pgic_3b][previous-														

702	[pgic_4a] Show the field ONLY if: starts_with([event-name], 'vi sit') AND [pgic_3b_prev] != ""	Last time you answered this questionnaire you said you had started the following new treatment: [event-name][pgic_3b] [previous-instance] Are you still on this treatment?	instance)) radio yes Yes no No
703	[pgic_4b] Show the field ONLY if: [pgic_4a] = 'yes'	How has your neuromuscular condition changed since you started this treatment plan?	radio 1 Very Much Improved 2 Much Improved 3 Minimally Improved 4 No Change 5 Minimally Worse 6 Much Worse 7 Very Much Worse
704	[pgic_only_available_if _date_of_birth_filled_i n_bas_complete]	Section Header: Form Status Complete?	dropdown 0 Incomplete 1 Unverified 2 Complete

**Instrument: End of Data Collection (end\_of\_data\_collection)**

Active languages: None

705	[eodc_h1]	END OF DATA COLLECTION	descriptive
706	[eodc_current_date]	Current date	text (date_ymd), Required
707	[eodc_reason]	Reason for end of data collection <i>Element 3.1. in the Set of common data elements for Rare Diseases Registration (link)</i>	radio, Required death Death of patient lost_fu Patient lost in follow-up opted_out Patient opted-out
708	[eodc_date_of_death_kno wn] Show the field ONLY if: [eodc_reason] = 'death'	Date of death known <i>Element 3.2. in the Set of common data elements for Rare Diseases Registration (link).</i>	radio, Required yes Yes no No
709	[eodc_date_of_death] Show the field ONLY if: [eodc_date_of_death_know n] = 'yes'	Date of death yyyy-mm-dd <i>Element 3.2. in the Set of common data elements for Rare Diseases Registration (link). If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.</i>	text (date_ymd), Required
710	[eodc_date_of_loss_fu_k nown] Show the field ONLY if: [eodc_reason] = 'lost_fu'	Date of loss to follow-up known	radio, Required yes Yes no No
711	[eodc_date_of_loss_fu] Show the field ONLY if: [eodc_date_of_loss_fu_know n] = 'yes'	Date of loss to follow-up yyyy-mm-dd	text (date_ymd), Required
712	[eodc_date_of_opt_out_k nown] Show the field ONLY if: [eodc_reason] = 'opted_out'	Date of opt-out known	radio, Required yes Yes no No
713	[eodc_date_of_opt_out] Show the field ONLY if: [eodc_date_of_opt_out_kno wn] = 'yes'	Date of opt-out yyyy-mm-dd	text (date_ymd), Required
714	[eodc_date]	eodc_date: Hidden calculated field that will always contain the date of end of data collection regardless of the reason for end of data collection. We use this to simplify writing of logic e.g. for Data Quality rules.	text Field Annotation: @CALCTEXT(if([eodc_date_of_death_known] = 'yes', [eodc_date_of_death], if([eodc_date_of_loss_fu_known] = 'yes', [eodc_date_of_loss_fu],

			if([eodc_date_of_opt_out_known] = 'yes', [eodc_date_of_opt_out, "])) @READONLY @HIDDEN @HIDDEN-PDF						
715	[end_of_data_collection _complete]	Section Header: <i>Form Status</i> Complete?	dropdown <table border="1"> <tr> <td>0</td> <td>Incomplete</td> </tr> <tr> <td>1</td> <td>Unverified</td> </tr> <tr> <td>2</td> <td>Complete</td> </tr> </table>	0	Incomplete	1	Unverified	2	Complete
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