

7th ERN EURO-NMD ANNUAL MEETING

UPDATE ON EURO NMD WORKING GROUPS

21st February 2024



**European
Reference
Network**

for rare or low prevalence
complex diseases



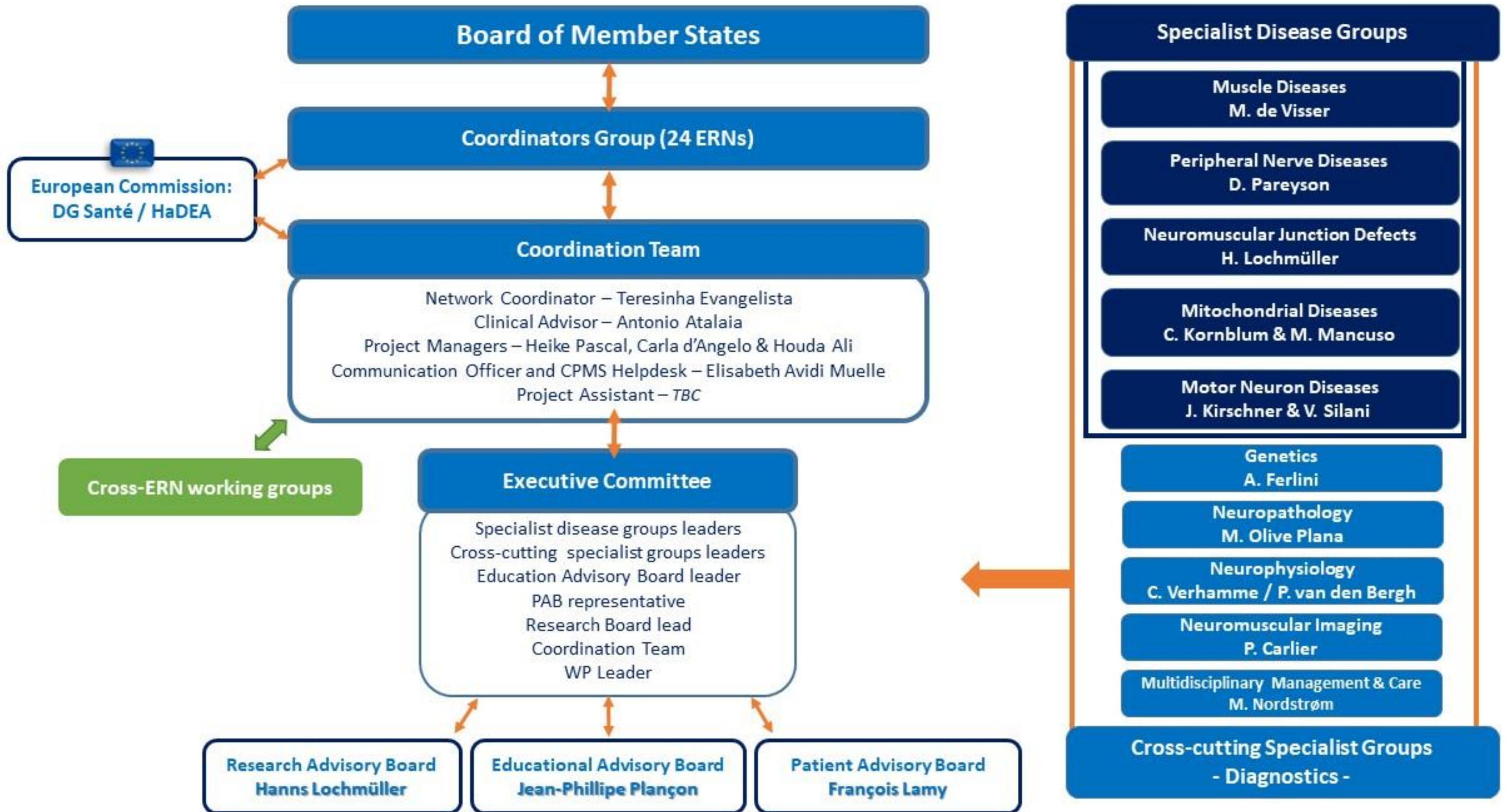
Network
Neuromuscular
Diseases (ERN EURO-NMD)



**Funded by
the European Union**



ERN EURO-NMD Governance



7th ERN EURO-NMD ANNUAL MEETING

21st – 23rd February 2024

UPDATE ON MUSCLE WORKING GROUP ACTIVITIES

Marianne de Visser

Amsterdam University Medical Center

Amsterdam, The Netherlands



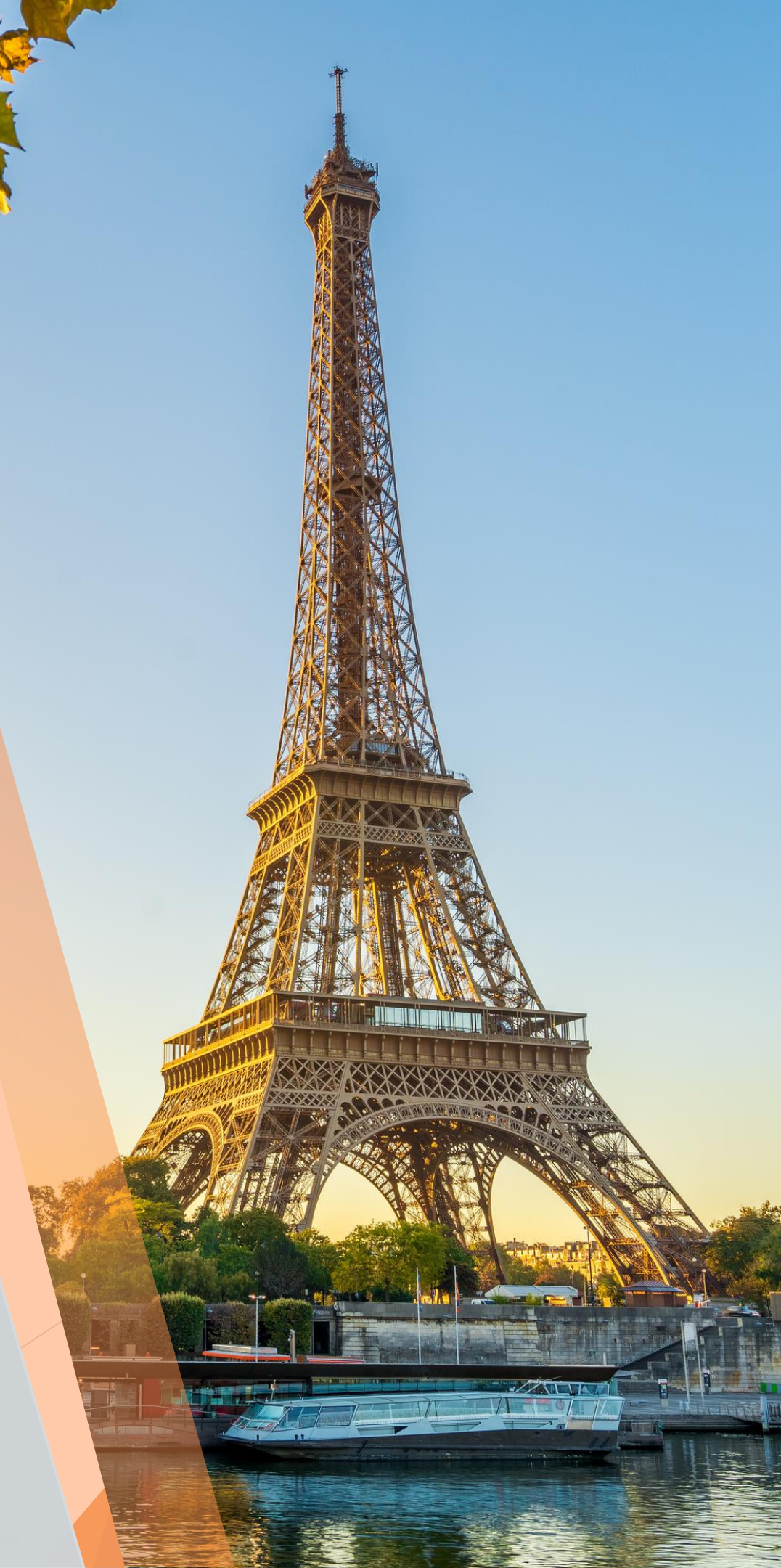
European
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Network

Neuromuscular
Diseases (ERN EURO-NMD)



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MEET THE MUSCLE WORKING GROUP

Nadine van der Beek



Metabolic disorders

Carmen Paradas



Muscular dystrophies, cong. myopathies

Volker Straub



Hereditary neuromuscular diseases

Sabrina Sacconi



Muscular dystrophies

Jens Schmidt



Myositis

Benedikt Schoser



(Non) Myotonic dystrophies & MFMs

Antonio Toscano



Metabolic myopathies

Kristl Claeys



MFMs, cong myopathies, BMD, LGMDs

Marianne de Visser



Myositis, LGMDs

PAB representatives:



Madelon Kroneman

Michela Onali

Silke Schlüter

EDUCATION

Webinars

THEME 'Presenting signs/symptoms in NMDs'

- 2 in 2022
- 5 in 2023
- 4 in 2024

Rhabdomyolysis

Click here to register! [🔗](#)...

TIMESLOT: 29 fev. 2024 16:00

Cognitive Symptoms in Myopathies

...TIMESLOT: 4 jan. 2024 16:00 CET...THEME: Cognitive Symptoms in Myopathies

Respiratory Insufficiency in Neuromuscular Diseases

1:02:30

Distal Myopathies

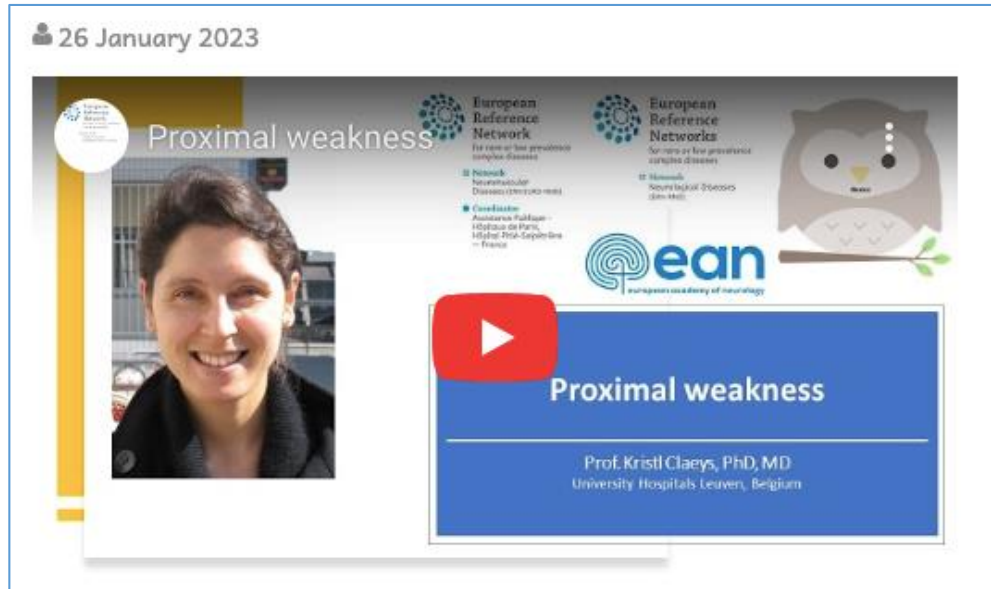
25 January 2024

Prof Bjarne Udd
Director of the Tampere Neuromuscular Center,
Tampere University Hospital, Finland

WEBINARS 2022 and 2023

26 January 2023


Proximal weakness



European Reference Network
For rare or low prevalence complex diseases

Network: Neuromuscular Diseases (ERN-NMD)

Coordinator: Assistance Publique - Hôpitaux de Paris, Hôpital Pitié-Salpêtrière - France



Proximal weakness

Prof. Kristl Claeys, PhD, MD
University Hospitals Leuven, Belgium

17 February 2023

Muscle hyperexcitability and cramps



European Reference Network
For rare or low prevalence complex diseases

Network: Neuromuscular Diseases (ERN-NMD)

Coordinator: Assistance Publique - Hôpitaux de Paris, Hôpital Pitié-Salpêtrière - France



Muscle hyperexcitability and cramps

Dr. Federica Montagnese, MD
Friedrich Baur Institute, Ludwig Maximilians University, Munchen, Germany

24 November 2022

External Ophthalmoplegia



European Reference Network
For rare or low prevalence complex diseases

Network: Neuromuscular Diseases (ERN-NMD)

Coordinator: Assistance Publique - Hôpitaux de Paris, Hôpital Pitié-Salpêtrière - France



External Ophthalmoplegia

Carmen Paradas Lopez, PhD, MD
Hospital Universitario Virgen del Rocío and Instituto de Biomedicina de Sevilla (IIBVS-IBS)

Floppy child

4 May 2023



...PRESENTER: Dr Ulrike Schara-Schmidt

27 April 2023

Fatigue in Neuromuscular Disorders



European Reference Network
For rare or low prevalence complex diseases

Network: Neuromuscular Diseases (ERN-NMD)

Coordinator: Assistance Publique - Hôpitaux de Paris, Hôpital Pitié-Salpêtrière - France



Fatigue in Neuromuscular Disorders

Dr. Nicole B.M. Voet
Radboud University Medical Center, the Netherlands

6 July 2023

Cardiac involvement in Neuromuscular Dis...



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For rare or low prevalence complex diseases

Network: Neuromuscular Diseases (ERN-NMD)

Coordinator: Assistance Publique - Hôpitaux de Paris, Hôpital Pitié-Salpêtrière - France



Cardiac Involvement in Neuromuscular Diseases

Prof. Karim Wahbi, PhD, MD
APHP Hospital Cochin, Paris, France

22 December 2022

Dysphagia in Neuromuscular Diseases



European Reference Network
For rare or low prevalence complex diseases

Network: Neuromuscular Diseases (ERN-NMD)

Coordinator: Assistance Publique - Hôpitaux de Paris, Hôpital Pitié-Salpêtrière - France



Dysphagia in Neuromuscular Diseases

Prof. Marianne de Visser
Academic Medical Centre, the Netherlands

WEBINARS 2024 AND BEYOND

TOPIC

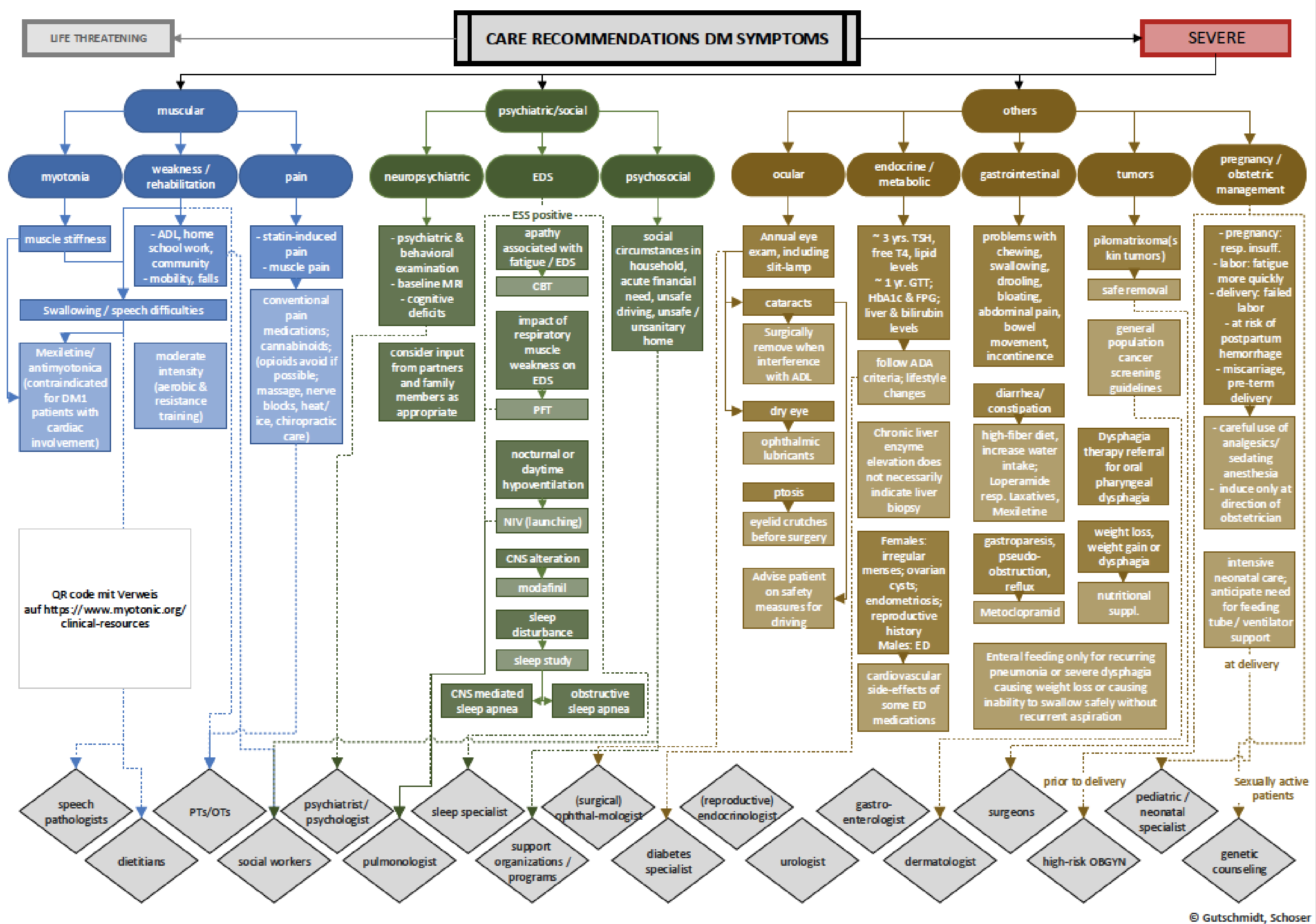
- ▶ TREATABLE DISEASES
- ▶ CLINICALLY ACTIONABLE DISEASES

ANY (OTHER) SUGGESTIONS PLEASE CONTACT

m.devisser@amsterdamumc.nl OR Kristl.Claeys@uzleuven.be

PATIENT CARE

- ▶ **CARE STANDARDS/FLOWCHARTS (DM, PD, FSHD, LGMD, MFM, MYOSITIS)**
- ▶ **ENMC WORKSHOPS**
- ▶ **GUIDELINES**
- ▶ **PATIENT JOURNEY**



© Gutschmidt, Schoser

ENMC WORKSHOPS

Neuromuscular Disorders 33 (2023) 800–816

Downloaden



ELSEVIER

Contents lists available at ScienceDirect

Neuromuscular Disorders

journal homepage: www.elsevier.com/locate/nmd

255th ENMC workshop: Muscle imaging in idiopathic inflammatory myopathies. 15th January, 16th January and 22nd January 2021 – virtual meeting and hybrid meeting on 9th and 19th September 2022 in Hoofddorp, The Netherlands

Marianne de Visser^{a,*}, Pierre Carlier^{b,*}, Jiří Vencovský^c, Kateřina Kubínová^c, Corinna Preusse^d, on behalf of the ENMC Muscle Imaging in Idiopathic Inflammatory Myopathies workshop study group[#]

Inclusion Body Myositis: 10 years of progress – revision of the 'ENMC 2013 diagnostic criteria for IBM' and trial readiness

Number 272

Date 16 JUNE 2023

ENMC recommendations for optimizing bone strength in neuromuscular disorders

Number 274

Date 19 JANUARY 2024



PATIENT CARE

- ▶ **CARE STANDARDS/FLOWCHARTS**
- ▶ **ENMC WORKSHOPS**
- ▶ **GUIDELINES (HYPERCKAEMIA)**
- ▶ **PATIENT JOURNEY (MFM, MYOSITIS)**



EURO-NMD ERN – NEUROPATHY WORKING GROUP: NEW MANAGEMENT GROUP



Davide Pareyson, Chair

Shahram Attarian, EDUCATION

Maike Dohrn, EDUCATION - VGR

Jana Haberlova, CHILDREN

Janneke Hoeijmakers, PAINFUL NEUROPATHIES

Stephan Goedee (+ Davide), GUIDELINES

Marina Grandis, CPMS

Kleopas Kleopa, GENE THERAPY (INTER-ERN GROUP)

Isabella Moroni, TRANSITION

Stefano Previtali, REGISTRY

Teresa Sevilla (Rafael Sivera) (help in CPMS / REGISTRY)

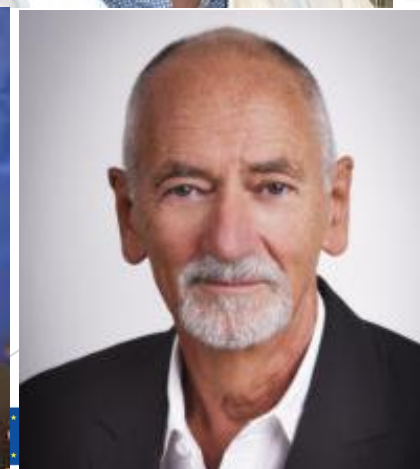
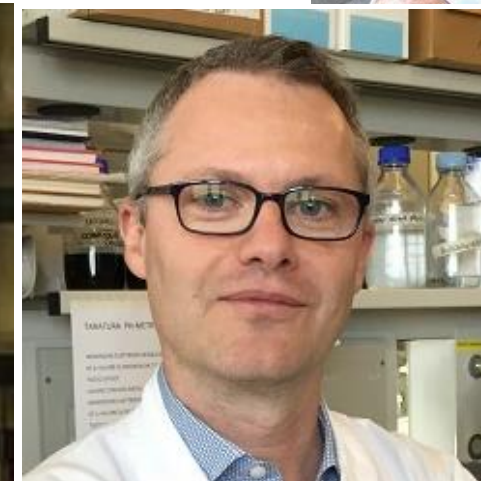
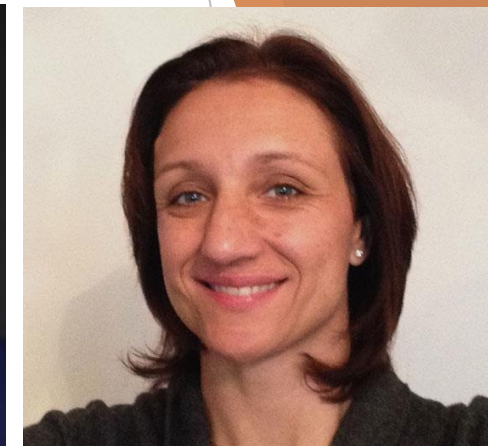
Pieter van Doorn, Advisor

Patients' Representatives:

Jean-Philippe Plançon – EPODIN

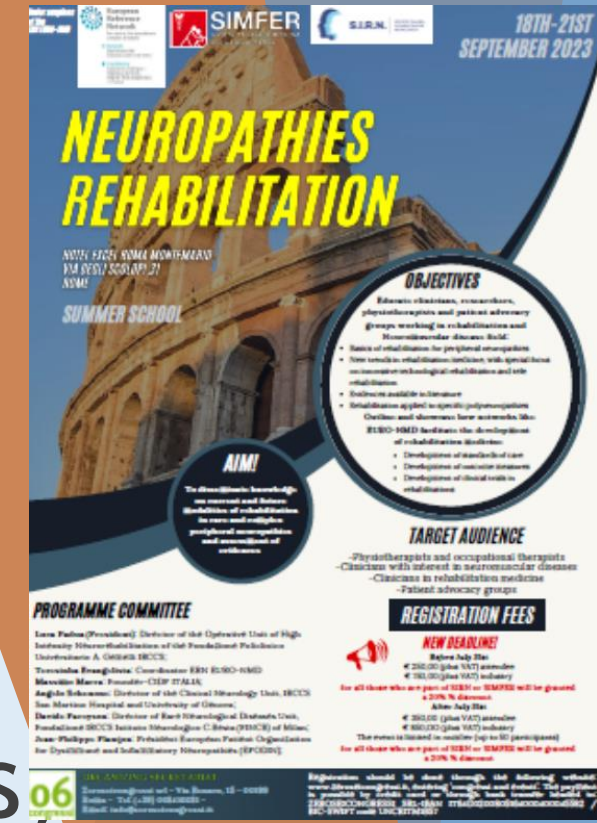
Daniel Tanesse/Arabela Acalinei – European CMT Federation

Jean-Christophe Fidalgo – Amyloidosis Alliance



Neuropathy WG update

- ▶ Course on Rehabilitation in Neuropathies, Rome 18-21 Sept 2023
- ▶ Guidelines on diagnosis and treatment TTR-related amyloid neuropathy:
 - ▶ PNS/EAN (ERN)
 - ▶ Isabel Conceição, Michael Polydefkis - Davide Pareyson (chairs)
 - ▶ Final composition of the task force; first kick-off call Oct 27; 2nd call Dec 21; in-person meeting Feb 16-17, 2024 in Vienna: selected 14 PICOs
- ▶ Other guidelines to endorse (GBS) + planned (PNS GL committee)
- ▶ October 2023: 4 webinars for the CMT month awareness (Previtali, Pareyson, Kleopa, Attarian)
- ▶ Transition (WP9): review of the literature, existing protocols for transition



Plans for the future

- ✓ Neuropathy WG Meeting this evening
- ✓ Webinars and VGRs (use of CPMS)
- ✓ Registry for ultrarare diseases
- ✓ Guidelines - making decision tools (develop / endorse formally)
- ✓ Transition for peripheral neuropathies (joint with other WG and Teresinha)
- ✓ Periodic meetings of the Neuropathy Working Group
- ✓ Inter-ERN groups: Gene Therapy – Postgraduate Curriculum on neurological and related disorders
- ✓ Interactions and collaboration with PAGs (patients' journey, etc.)

7th ERN EURO-NMD ANNUAL MEETING

NMJ working group (MG/CMS) – report

21st – 23rd February 2024

Hanns Lochmüller
Freiburg/Ottawa



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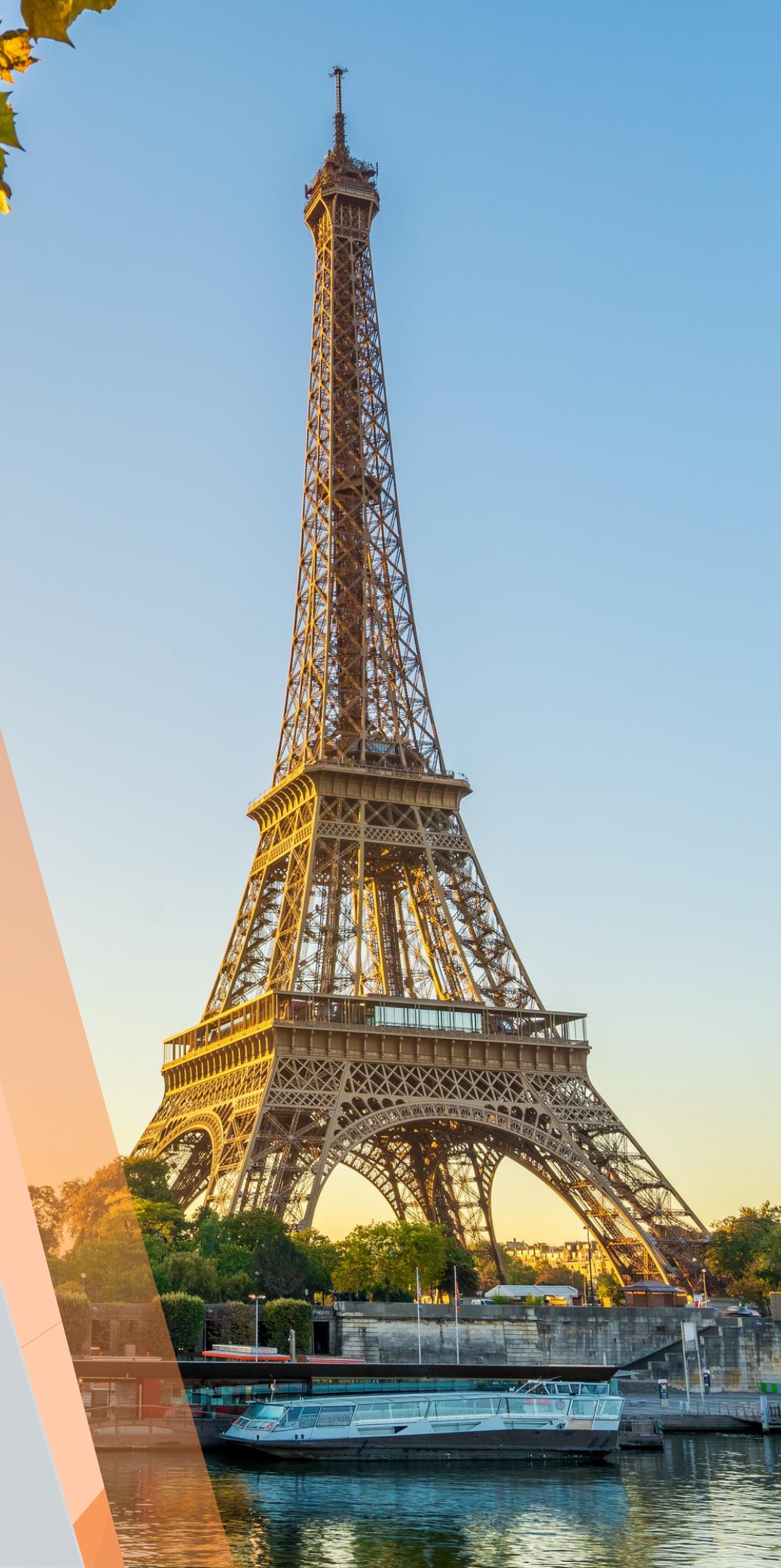


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EURO-NMD ERN – Neuromuscular junction WG (MG/CMS)

- Guidelines on juvenile MG (in progress, led by J. Palace)
- ENMC workshop on seronegative MG (February 2024)
- ENMC workshop on MG and registries (T. Evangelista participating)
- Planning of MG and CMS research projects based on EURO-NMD registry (D. Natera, A. Nascimento, H. Lochmuller, L. Maggi)
- Review on PROMs across all NMDs completed, manuscript in preparation (N. Voet); potential utility for EURO-NMD registry
- CMS natural history study (led by Oxford): ongoing
- Contribution to curriculum (led by M. de Visser, A. Evoli)
- Survey mapping the use of innovative MG therapies across Europe

New Co-Chair – Neuromuscular junction WG (MG/CMS)

Dr Lorenzo Maggi (Besta Institute, Milan)



Dr. Maggi is a consultant neurologist for muscle and neuromuscular junction diseases at Fondazione IRCCS Istituto Neurologico Carlo Besta, leading a clinical and laboratory team contributing to clinical, diagnostic and research activities, including clinical trials. He is the Coordinator of the Italian Network of the Congenital Myasthenic Syndromes (with Dr. Rodolico) and Myasthenia Gravis (with Dr. Liguori). He is member of the board of the Italian Association of Myology.

7th ERN EURO-NMD ANNUAL MEETING

EURO-NMD Imaging Update

21st February 2024

Pierre G. CARLIER

St Luc University Hospital, Brussels, Belgium

Liege State University, Liege, Belgium

Erasme University Hospital, Brussels, Belgium



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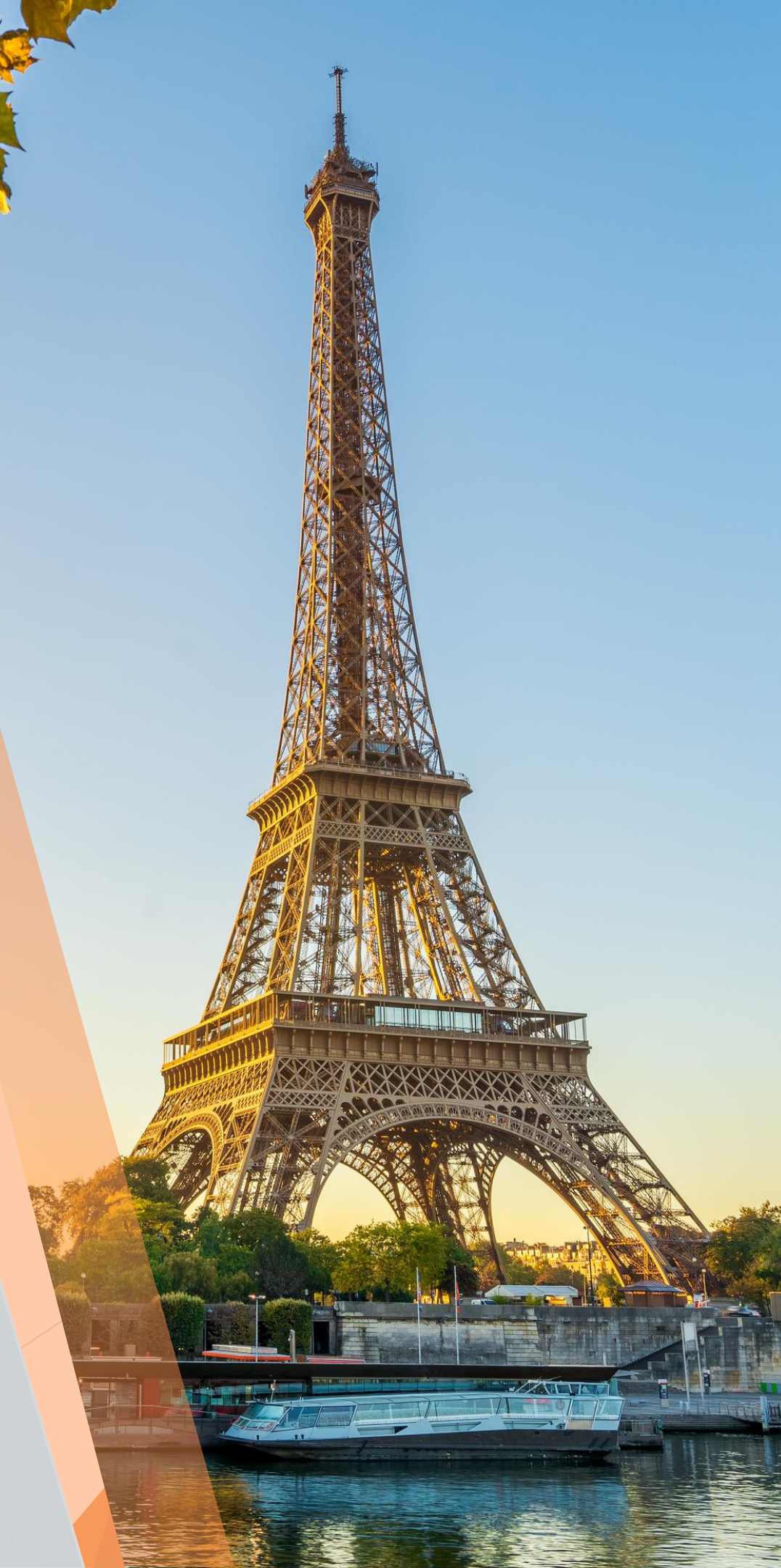


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Diseases (ERN EURO-NMD)



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EURO-NMD

Building bridges and breaking barriers
in rare neuromuscular diseases

Imaging group

G. Tasca



R.-Y. Carlier



J. Salama



J.A. Urtizberea



O. Scheidegger



P. Carlier



S. Gerevini



W. Stenzel





EURO-NMD

Building bridges and breaking barriers
in rare neuromuscular diseases

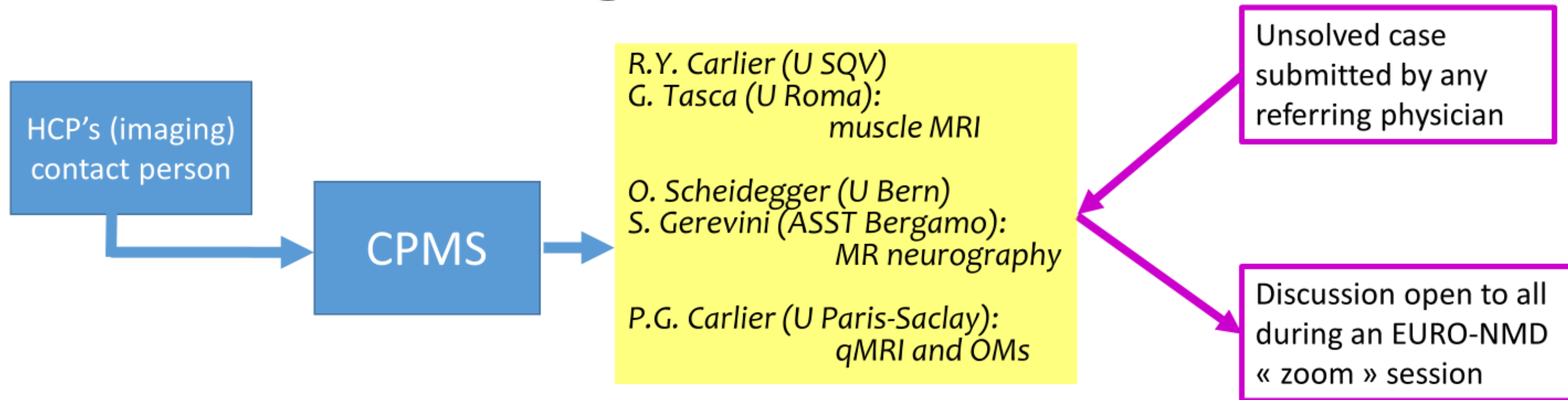
Imaging group objectives

- *To assist HCPs with diagnostic imaging*
- *To promote better NMD imaging protocols*
- *To help developing more advanced NMD imaging protocols*
- *To build an imaging atlas of NMDs*
- *To support an imaging data base*
- *To develop an AI based diagnostic imaging software*

*collaboration with
MYO-SHARE (MYO-MRI)
and EU IT department*



Imaging group: diagnostic activities



- In practice: # ask to be registered in the mailing list to EURO-NMD coordination
send anonymized cases to pghrmgc@gmail.com
look for case discussion dates in the EURO-NMD newsletter
next session: Tuesday February 8th 5PM CET

Feb 19 online meeting with HCPs imaging representatives

« Grand Rounds » favored to specific imaging discussion

CPMS is a deterrent to case submission

more involvement of radiologists needed

AND YOU? WHAT IS YOUR OPINION?



Imaging group: educational activities

- Hands-on training and workshop

✓ June 2018 and , 2019: Paris, Institute of Myology,
immediately after the Myology Summer School

Update on diagnostic imaging
Hands-on session on diagnostic and quantitative MR imaging
Hands-on session on muscle strength and activity measurements

✓ New format in 2022: training
exported to HCPs sites,
✓ 4 on-site trainings: Kaunas, Riga,
Göttingen, Tartu

2023: 2 failed attempts
2024: ?

- Webinars

✓ 4 in 2019 and 2020

2024? 2025?

- MYO-MRI conference

✓ On line in 2021

✓ Hybrid in Berlin in Nov 2023

again in Berlin in 2025

EURO-NMD

Building bridges and breaking barriers
in rare neuromuscular diseases



Imaging group: educational activities

- SOPs

MYO-MRI *Journal of Neuromuscular Diseases* 3 (2016) 1–28

Skeletal Muscle Quantitative Nuclear
Magnetic Resonance Imaging
and Spectroscopy as an Outcome Measure
for Clinical Trials

Pierre G. Carlier^{a,b,c,*}, Benjamin Marty^{a,b}, Olivier Scheidegger^{a,d}, Paulo Loureiro de Sousa^e,
Pierre-Yves Baudin^f, Eduard Snezhko^e and Dmitry Vlodavets^g

J Neurol (2017) 264:2053–2067

MYO-MRI **Quantifying fat replacement of muscle by quantitative MRI
in muscular dystrophy**

Jedrzej Burakiewicz¹ · Christopher D. J. Sinclair^{2,3} · Dirk Fischer^{4,5} ·
Glenn A. Walter⁶ · Hermien E. Kan¹ · Kieren G. Hollingsworth⁷

EURO-NMD | Building bridges and breaking barriers
in rare neuromuscular diseases

To come:
Imaging protocols
update
OM update
MRNeurography

Qs for the audience

HCPs interested in on-site training?

volunteers for webinars?

volunteers for writing SOPs, reviews, ...?

what are your needs that are currently unmet?



Imaging group future

- Welcome more imaging specialists in the core group. Applications opened
- More cases discussion from more HCPs and open to all
- MRI on-site training.
Imaging protocols installation on your scanner. Applications opened.
- New webinars. Update SOPs. If you have particular needs, contact us
- Adding momentum to e-health initiatives (MYO-SHARE, EU IT depts)
- Better meet your needs. Suggestions and requests to **pghrmgc@gmail.com**

EURO-NMD

Building bridges and breaking barriers
in rare neuromuscular diseases

7th ERN EURO-NMD ANNUAL MEETING

ERN-MND Working Group ALS and other motor neuron diseases

21st February 2024

Vincenzo Silani

IRCCS Istituto Auxologico Italiano

Milano, Italy

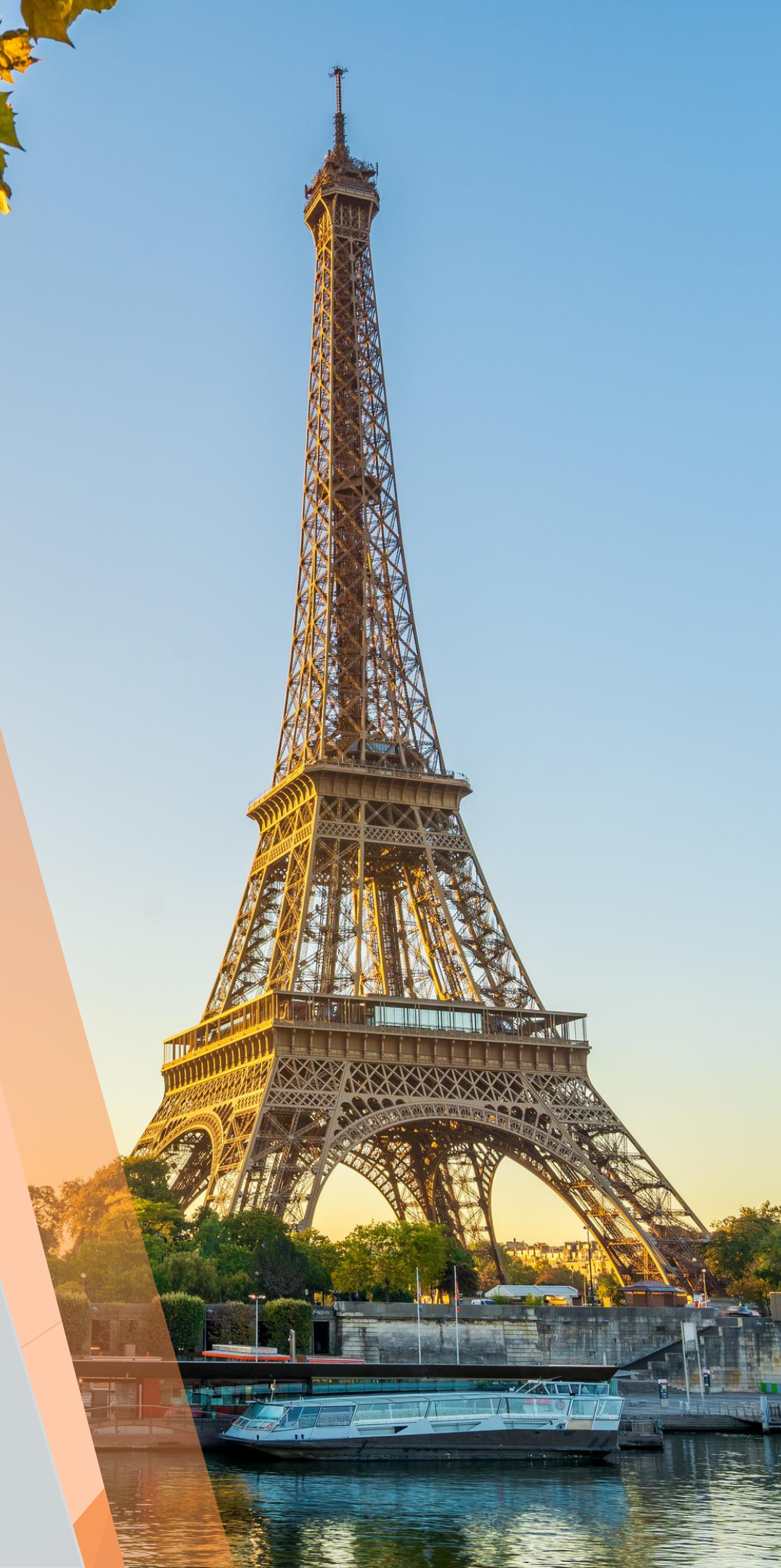


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Activities of the ERN EURO-MND Working Group ALS and other motor neuron diseases

- ▶ New guidance from 9 Feb 2023 - 47 members/10 Countries
- ▶ Proposed general topics of the Working Group:
 - homogeneous genetic testing and counselling
 - NFL homogenous testing in CSF/plasma
 - cognitive/behavioral homogenous testing
- ▶ Updates guidelines on the management of amyotrophic lateral sclerosis - European Journal of Neurology DOI: [10.1111/ene.16264](https://doi.org/10.1111/ene.16264), in press
- ▶ Lay version of the guidelines promoted

Activities of the ERN EURO-MND Working Group

ALS and other motor neuron diseases

► Homogenous genetic testing: *C9orf72*

frontiers | Frontiers in Neurology

TYPE Brief Research Report
PUBLISHED 31 January 2024
DOI 10.3389/fneur.2024.1284459

Check for updates

OPEN ACCESS

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University of São Paulo, Brazil

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PUBLISHED 31 January 2024

CITATION
Giardina E, Mandich P, Ghidoni R, Ticozzi N,
Rossi G, Fenoglio C, Tiziano FD,
Esposito F, Capellari S, Nacmias B,
Mineri R, Campopiano R, Di Pilla L,
Sammarone F, Zampatti S, Peconi C, De
Angelis F, Palmieri I, Galandra C, Nicodemo E,
Origone P, Gotta F, Ponti C, Nicsanu R,
Benussi L, Peverelli S, Ratti A, Ricci M, Di
Fede G, Magri S, Serpente M, Lattante S,
Domi T, Carrera P, Saltimbanco E, Bagnoli S,
Ingannato A, Albanese A, Tagliavini F, Lodi R,
Caltagirone C, Gambardella S, Valente EM and
Silani V (2024) Distribution of the *C9orf72*
hexanucleotide repeat expansion in healthy
subjects: a multicenter study promoted by
the Italian IRCCS network of neuroscience
and neurorehabilitation.
Front. Neurol. 15:1284459.
DOI 10.3389/fneur.2024.1284459

Distribution of the *C9orf72* hexanucleotide repeat expansion in healthy subjects: a multicenter study promoted by the Italian IRCCS network of neuroscience and neurorehabilitation

Emiliano Giardina^{1,2*}, Paola Mandich^{3,4}, Roberta Ghidoni⁵,
Nicola Ticozzi^{6,7}, Giacomina Rossi⁸, Chiara Fenoglio^{9,10},
Francesco Danilo Tiziano^{11,12}, Federica Esposito^{13,14},
Sabina Capellari^{15,16}, Benedetta Nacmias^{17,18}, Rossana Mineri¹⁹,
Rosa Campopiano²⁰, Luana Di Pilla²⁰, Federica Sammarone²⁰,
Stefania Zampatti¹, Cristina Peconi¹, Flavio De Angelis^{21,22},
Ilaria Palmieri²³, Caterina Galandra²³, Eleonora Nicodemo²⁴,
Paola Origone^{3,4}, Fabio Gotta³, Clarissa Ponti^{3,4},
Roland Nicsanu⁵, Luisa Benussi⁵, Silvia Peverelli⁶,
Antonia Ratti^{6,25}, Martina Ricci⁸, Giuseppe Di Fede⁸,
Stefania Magri²⁶, Maria Serpente⁹, Serena Lattante^{11,27},
Teuta Domi²⁸, Paola Carrera²⁹, Elisa Saltimbanco¹⁵,
Silvia Bagnoli^{17,18}, Assunta Ingannato^{17,18}, Alberto Albanese³⁰,
Fabrizio Tagliavini³¹, Raffaele Lodi³², Carlo Caltagirone³³,
Stefano Gambardella^{20,34}, Enza Maria Valente^{23,24} and
Vincenzo Silani^{6,7}

967 subjects - 13 centers

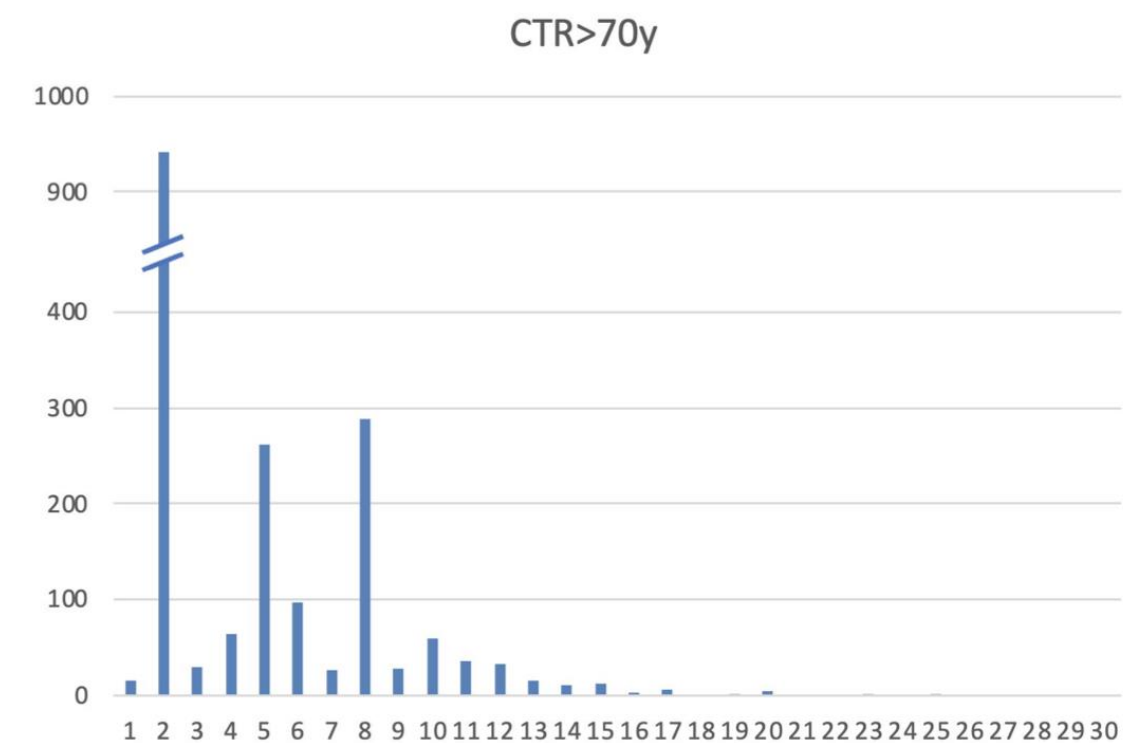


FIGURE 2
Allelic distribution of *C9orf72* alleles in our cohort.

To standardize and validate analytical methods, each participating institution was provided with the same lot of the commercially available AmplitudeXPCR/CE *C9orf72* Kit (Asuragen, Inc.).

Activities of the ERN EURO-MND Working Group

ALS and other motor neuron diseases

▶ Webinars organized

Speakers	Topic of the webinar session
Philip Van Damme	Presentation of the new guidelines on ALS management
Andrea Calvo	Update on ALS approved therapies
Rachael Marsden	Management of relationships and sexuality in ALS patients
Juan Vazquez	Genetic therapies and therapy developments for ALS

- ▶ Discussion of Clinical Cases organized
- ▶ Postgraduate Curriculum on neurological and related disorders: Prof. Philippe Corcia
- ▶ Merging with the Multidisciplinary management group

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WG Motor neuron diseases
Subgroup SMA

21st February 2024

Jan Kirschner
University of Freiburg



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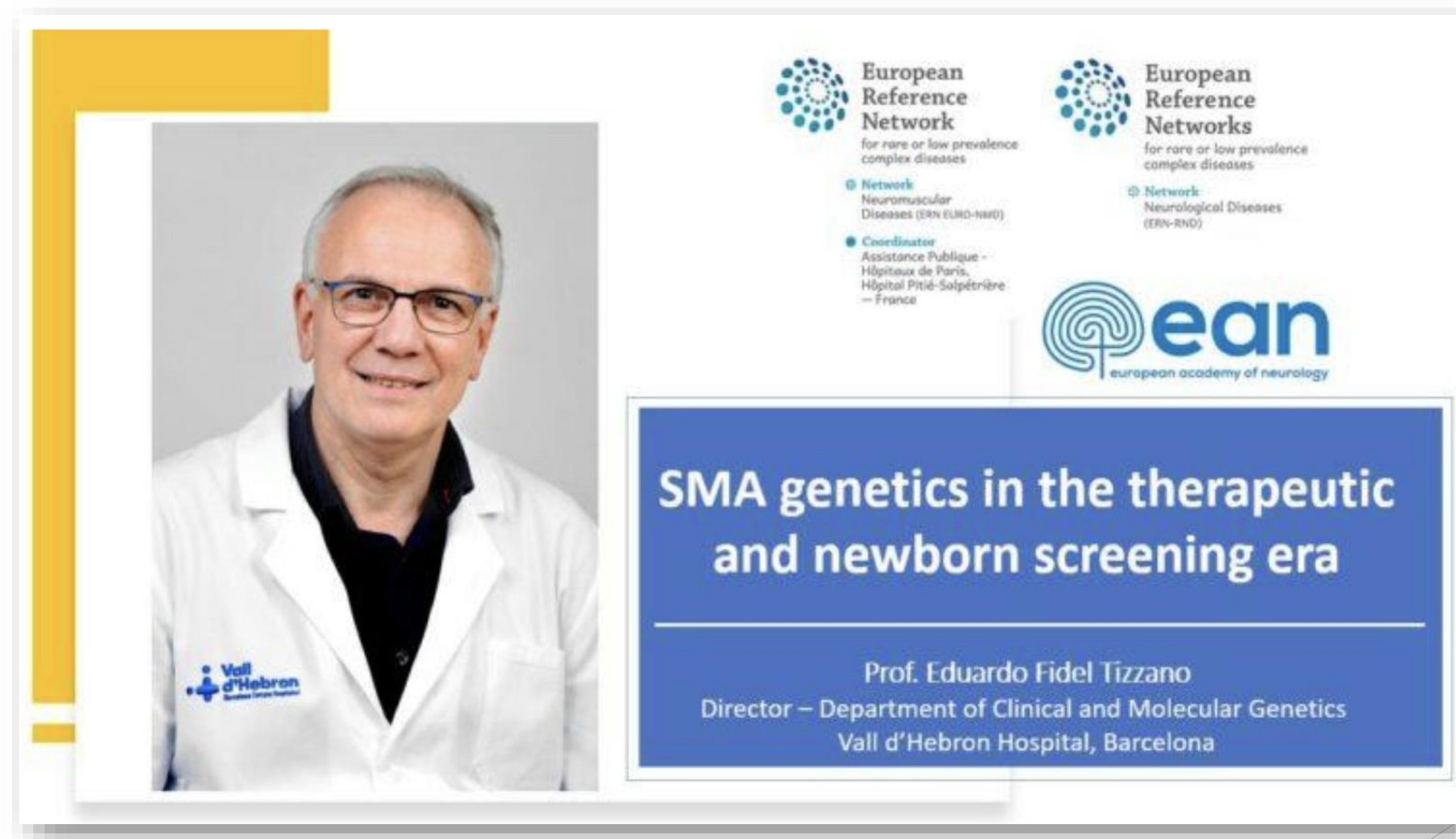
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Motor Neuron Diseases

Subgroup Spinal Muscular Atrophy



The slide features a portrait of Prof. Eduardo Fidel Tizzano on the left, wearing a white lab coat with the Vall d'Hebron logo. To the right, there are logos for the European Reference Network (ERN) Neurological Diseases (ERN-RND) and the European Academy of Neurology (EAN). The main title of the webinar is "SMA genetics in the therapeutic and newborn screening era". Below the title, the speaker's name and affiliation are listed: Prof. Eduardo Fidel Tizzano, Director – Department of Clinical and Molecular Genetics, Vall d'Hebron Hospital, Barcelona.

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Network: Neurological Diseases (ERN-RND)

Coordinator: Assistance Publique – Hôpitaux de Paris, Hôpital Pitié-Salpêtrière – France

European Reference Networks
for rare or low prevalence complex diseases

Network: Neurological Diseases (ERN-RND)

ean
european academy of neurology

SMA genetics in the therapeutic and newborn screening era

Prof. Eduardo Fidel Tizzano
Director – Department of Clinical and Molecular Genetics
Vall d'Hebron Hospital, Barcelona


Webinar on 15 February 2024 - THANK YOU!

Motor Neuron Diseases


Subgroup Spinal Muscular Atrophy

European Journal of Paediatric Neurology 28 (2020) 38–43


<https://doi.org/10.1016/j.ejpn.2020.07.001>
Contents lists available at ScienceDirect



European Journal of Paediatric Neurology



European ad-hoc consensus statement on gene replacement therapy for spinal muscular atrophy



Janbernd Kirschner ^{a,*}, Nina Butoianu ^b, Nathalie Goemans ^c, Jana Haberlova ^d, Anna Kostera-Pruszczyk ^e, Eugenio Mercuri ^{g,h}, W. Ludo van der Pol ^k, Susana Quijano-Roy ⁱ, Thomas Sejersen ^j, Eduardo F. Tizzano ^f, Andreas Ziegler ^l, Laurent Servais ^{m,n,1}, Francesco Muntoni ^{o,1}

2024 Update of European consensus statement on gene therapy

Motor Neuron Diseases

Subgroup Spinal Muscular Atrophy

- ▶ Challenges with changing/new phenotypes
- ▶ Survivors with SMA type 1 (e.g. 2 copies of *SMN2*)
- ▶ Early scoliosis
- ▶ Bulbar function
- ▶ Cognitive function
- ▶ Transition into adulthood

- ▶ Joint Meeting with the Multidisciplinary Management Working Group on Friday, February 23rd
 - ▶ Discuss specific needs for the field and next steps