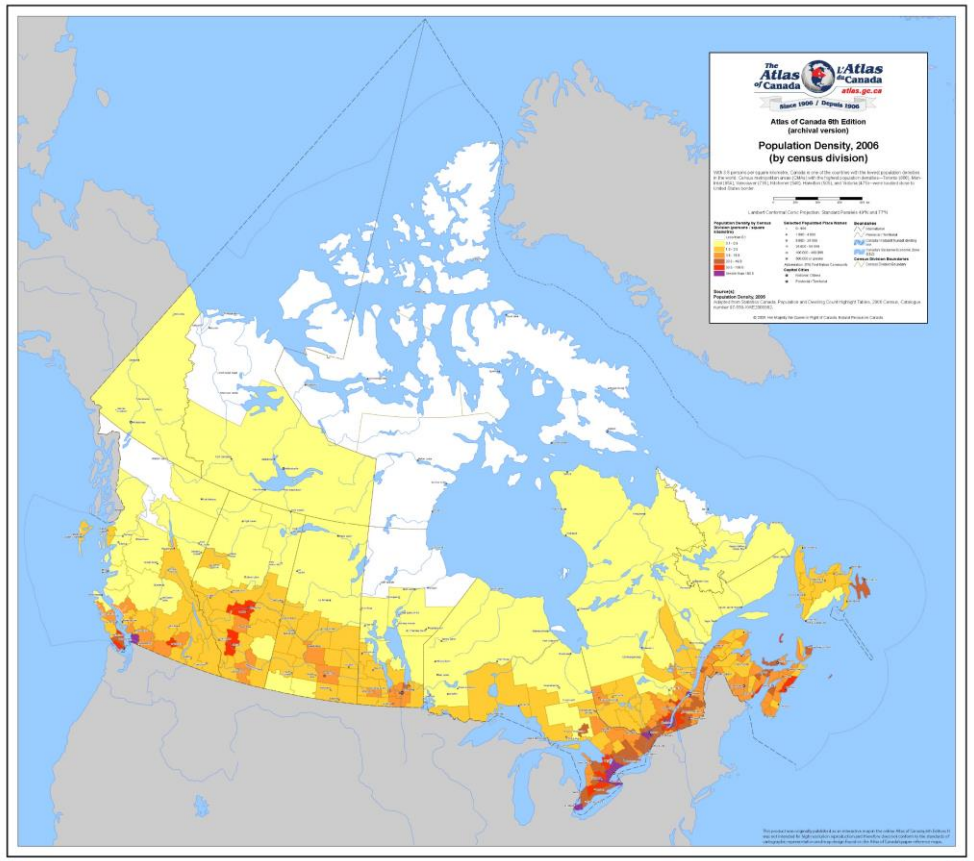
Canada is located in North America but is not part of the U.S.A.

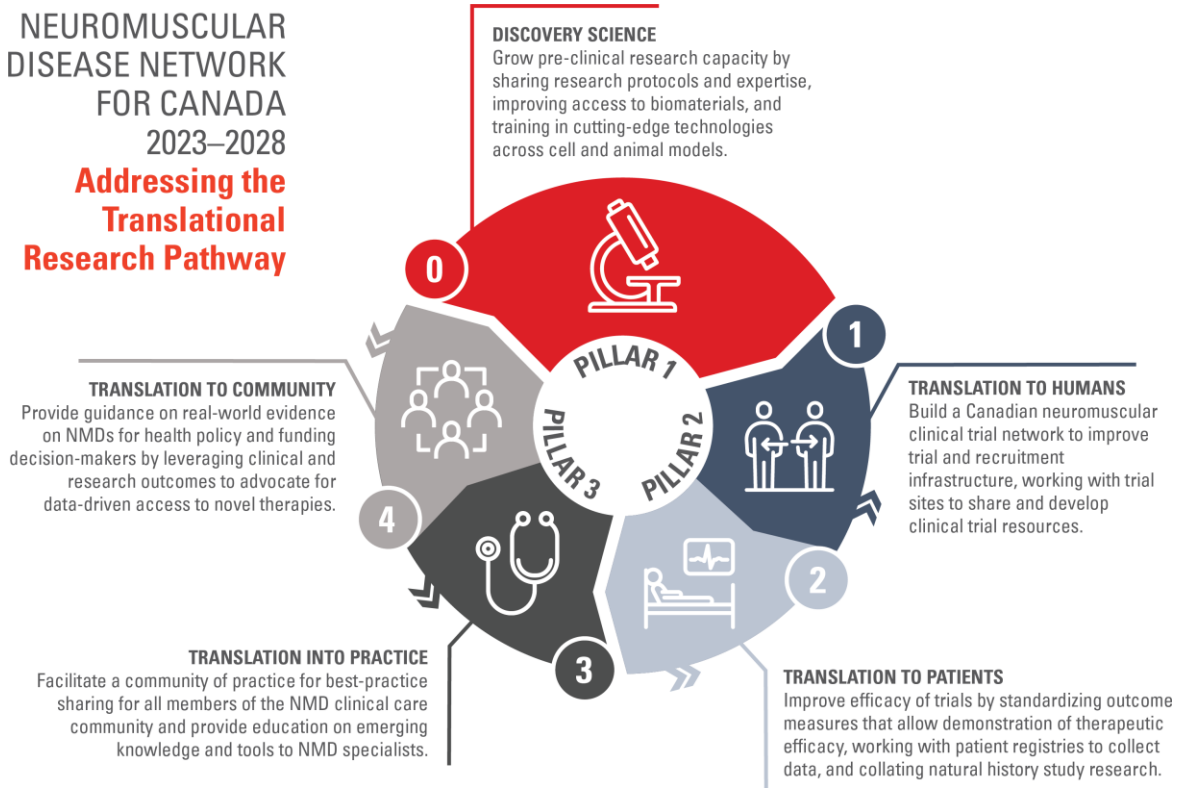


41.6 MILLION

- Similar population size to Poland, **30x larger landmass**
- Canada's GDP is $3,070 trillion CAD or **€2,019 trillion Euros:** similar to Italy, **half** of Germany
- CIHR: funding health research
- Health Canada: regulatory agency
- High prevalence: DM1, OPMD, SBMA

European leaders meet in London to discuss Russia-Ukraine strategy

Neuromuscular Disease Network for Canada (NMD4C)



The pan-Canadian network that brings together the country's leading clinical, scientific, technical, and patient expertise to improve care, research, and collaboration in neuromuscular disease

Steering Committee


Dr Hanns Lochmüller


Dr Homira Osman


Dr Natasha Chang


Dr James Dowling


Dr Jodi Warman Chardon


Dr Lawrence Korngut


Dr Cynthia Gagnon


Dr Rashmi Kothary

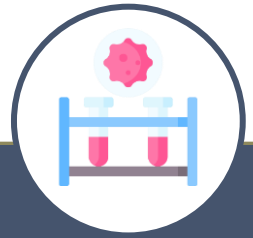
802 MEMBERS

409 clinicians & researchers
138 allied health
127 students
103 patients & family
45 industry

MUSCULAR DYSTROPHY CANADA
DYSTROPHIE MUSCULAIRE CANADA

CIHR IRSC
Canadian Institutes of Health Research | Instituts de recherche en santé du Canada

NMD4C



Pre-Clinical Science

Supporting preclinical researchers and strengthen cutting-edge basic research.

Tasks:

- Cell-based NMD Research
- Animal-Based NMD Research
- Human Samples and Biobanking

Clinical Research

Increasing neuromuscular clinical research capacity in Canada.

Tasks:

- Clinical Trial Network
- Participant Recruitment and Evidence Collection
- Outcome Measures and Natural History

Clinical Research & Education

Improving neuromuscular clinical education, care and access to treatments.

Tasks:

- Training and Curriculum
- Community of Practice
- From Trials into Practice

Themes:

- Early-Career Support
- Equity, Diversity, Inclusion and Indigeneity (EDII)
- Expert Patient Capacity Building
- Knowledge Translation & Impact Assessment
- Open Science

MUSCULAR DYSTROPHY CANADA
DYSTROPHIE MUSCULAIRE CANADA

CIHR IRSC
Canadian Institutes of Health Research | Instituts de recherche en santé du Canada

Strong National and International Collaborations

CHEO RESEARCH INSTITUTE INSTITUT DE RECHERCHE

CNDR.ORG Canadian Neuromuscular Disease Registry

RareKids CAN

neuro

MICYRN better health for mothers and children

Can-GARD Canadian Gene Cure Advanced Therapies for Rare Disease

Gene Cure Canada Thérapies innovantes pour les maladies rares

Stem Cell Network

CARE forRARE

RARE DISEASES MODELS and MECHANISMS NETWORK

Pewaseskwan

CSCN SCNC

Canadian Association of Pompe Association canadienne de Pompe

RÉSEAU CANADIEN DES MALADIES RARES

CANADIAN RARE DISEASE NETWORK

DEFEAT DUCHENNE CANADA

Myotonic Dystrophy FOUNDATION

cure SMA

cnag

SCPOR

CTSR

TREAT-NMD Neuromuscular Network

EURORDIS RARE DISEASES EUROPE

INSTITUT DE MYOLOGIE INNOVER POUR GUÉRIR

European Reference Network for rare or low prevalence complex diseases

EUROPEAN NEUROMUSCULAR CENTRE

FSHD CTRN FSHD CLINICAL TRIAL RESEARCH NETWORK

MDCC MUSCULAR DYSTROPHY CANADA DYSTROPHIE MUSCULAIRE CANADA

CIHR IRSC Canadian Institutes of Health Research Instituts de recherche en santé du Canada

Prioritize Patient Engagement

- Partner at all levels with patient organization Muscular Dystrophy Canada
- Collaboration with patient advocacy organizations
- Creation of patient-oriented research training for NMDs (ImPORTND)
- Patient partners woven into all working groups and activities
- Events connecting emerging basic science researchers with patient partners

Support the next generation of researchers

- Annual early-career awards & postdoctoral fellowship funding competitions
- Annual research trainee summer school
- Basic science trainee committee
- Online resource for research protocols
- Integrating ECRs into working groups, steering committee, chairing and presenting webinars
- Sponsor trainee visits between national collaborators beginning fall 2025

Support the next generation of clinicians

- Gained recognition of Neuromuscular Medicine as an Area of Focused Competency with Canada's Royal College of Medicine
- Annual early-career awards & clinical fellowship funding competitions
- Standardized national neuromuscular fellowship curriculum
- Multi-disciplinary care lecture series
- Annual clinical summer school beginning fall 2025

Clinical Capacity Building

Clinical Trial Network

- **Objectives:** Increase the number of neuromuscular clinical trials conducted in Canada, Enhance trial capacity and Support a cohesive network of clinician researchers and their teams.
- **Database:** Landscape of Canadian NM trials.

Communities of Practice

1. Clinical Research Coordinators & Trial Personnel
2. Physiotherapists and Occupational Therapists
3. Neuromuscular Physiatrists

A centralized hub to foster collaboration, knowledge exchange, and skill enhancement to elevate NMD research and care across Canada.

Canadian Neuromuscular Community of Practice
Communauté de pratique canadienne sur les maladies neuromusculaires

MUSCULAR DYSTROPHY CANADA
DYSTROPHIE MUSCULAIRE CANADA

CIHR IRSC
Canadian Institutes of Health Research
Instituts de recherche en santé du Canada

Capacity Building/Research Resources

LEARNMD Virtual Training Platform

Educational content and webinars covering neuromuscular disorders, translational and clinical research, therapies, and management strategies.

Royal College accredited webinar series

Virtual Canadian Neuromuscular Biobank

Development of virtual Canadian biobank, a central catalogue which will contain the neuromuscular samples available for research at the different biobanking sites.

- Virtual catalogue built on open-science-focused LORIS software from leading group at The Neuro.
- Minimum data for inclusion agreed on by 8 Canadian biobanks.

Work in progress: Agreements for data and sample exchange in progress with legal teams.

CNDR: a national collaborative pan-neuromuscular registry (Dr. Victoria Hodgkinson, Dr. Lawrence Korngut)

- Clinic-based recruitment & prospective clinical data collection
- Launched in 2010, sustainably operating and continuing to grow with industry, not-for-profit, and government funding

Year	Clinical Datasets	# Registered
2010	DM	602
2010	DMD	828
2012	SMA	505
2012	ALS	2586
2016	LGMD	193
2022	CMS	37
2025	FSHD	296
2025	OPMD	*76
2025	SBMA	*27

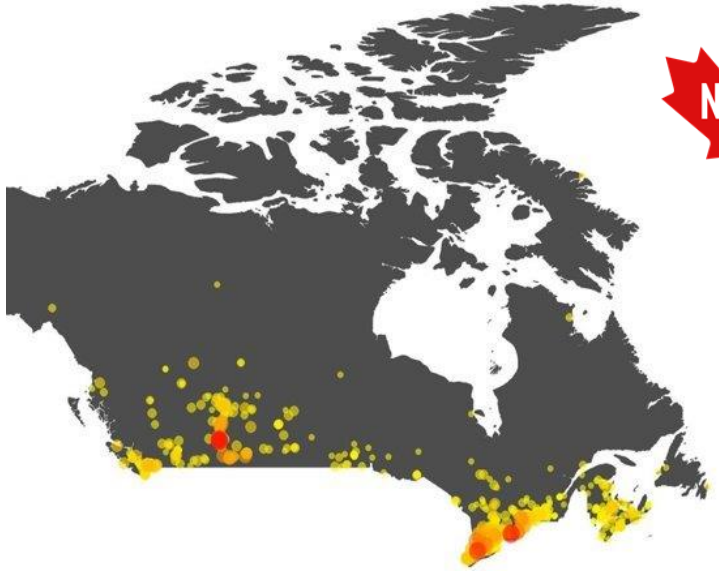
*no clinical data, pending dataset launch

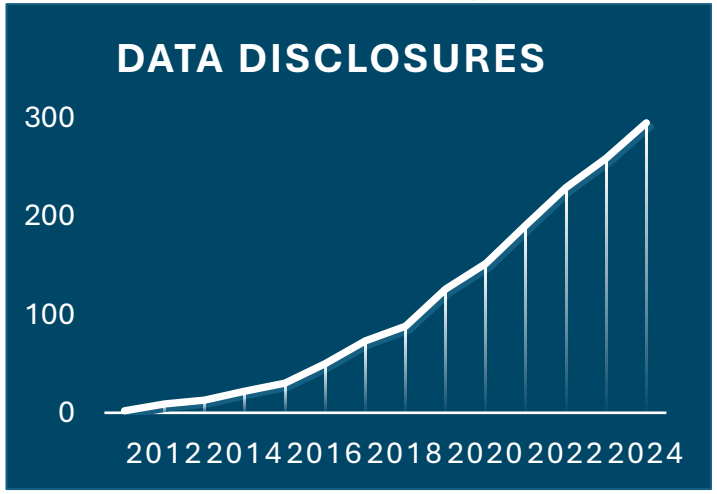
Over 7000 patients in every province & territory

52 Clinics

136+ Investigators

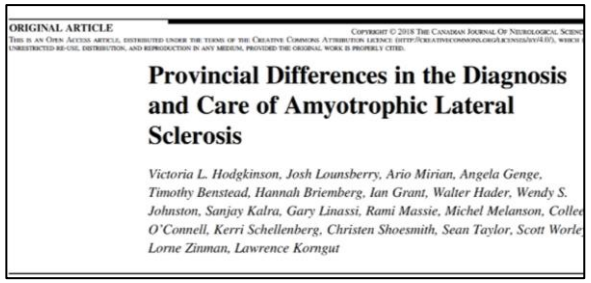
Over 300 Data Usages





CNDR:
Data and Network Impacts

➢ Data Analyses



ORIGINAL ARTICLE

Provincial Differences in the Diagnosis and Care of Amyotrophic Lateral Sclerosis

Victoria L. Hodgkinson, Josh Lounsberry, Ario Mirian, Angela Genge, Timothy Benstead, Hannah Briemberg, Ian Grant, Walter Hader, Wendy S. Johnston, Sanjay Kalra, Gary Linassi, Rami Massie, Michel Melanson, Collee O'Connell, Kerri Schellenberg, Christen Shoesmith, Sean Taylor, Scott Worle Lorne Zinman, Lawrence Korngut

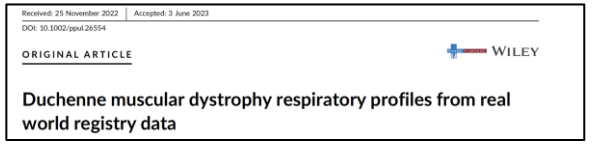
The Canadian Journal of Neurological Sciences (2024), 1–12
doi:10.1017/cjn.2024.1

Canadian Journal of Neurological Sciences / Journal Canadien des Sciences Neurologiques

Original Article

A Study on the Incidence and Prevalence of 5q Spinal Muscular Atrophy in Canada Using Multiple Data Sources

Tiffany R. Price[1], Victoria Hodgkinson[2], Grace Westbury[2], Lawrence Korngut[2], Micheil A. Innes[3], Christian R. Marshall[4,5], Tanya N. Nelson[6,7], Lijia Huang[8], Jillian Parboosingh[9] and Jean K. Mah[1]



Received: 25 November 2022 | Accepted: 3 June 2023
DOI: 10.1002/ppul.26554

ORIGINAL ARTICLE

WILEY

Duchenne muscular dystrophy respiratory profiles from real world registry data

The Canadian Journal of Neurological Sciences (2025), 52, 119–128
doi:10.1017/cjn.2024.49

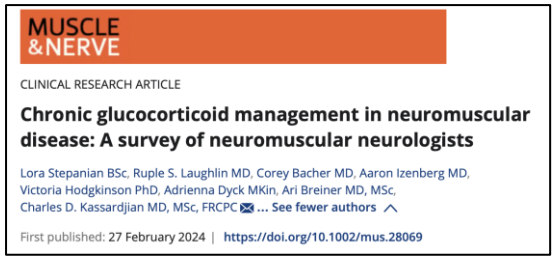
Canadian Journal of Neurological Sciences / Journal Canadien des Sciences Neurologiques

Original Article

A Real-World Study of Nusinersen Effects in Adults with Spinal Muscular Atrophy Type 2 and 3

Isabelle Côté[1], Victoria Hodgkinson[2], Marianne Nury[3], Louis Bastenier-Boutin[3] and Xavier Rodrigue[3,4]

➢ Network Consensus/ Practice Review



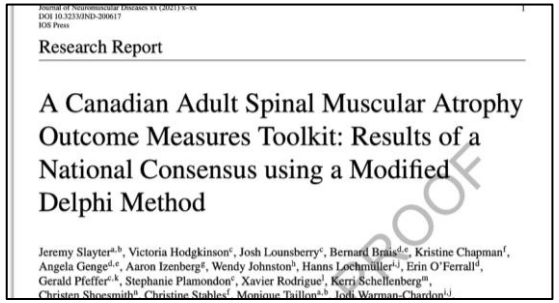
MUSCLE & NERVE

CLINICAL RESEARCH ARTICLE

Chronic glucocorticoid management in neuromuscular disease: A survey of neuromuscular neurologists

Lora Stepanian BSc, Ruple S. Laughlin MD, Corey Bacher MD, Aaron Izenberg MD, Victoria Hodgkinson PhD, Adrienna Dyck MKin, Ari Breiner MD, MSc, Charles D. Kassardjian MD, MSc, FRCPC ... See fewer authors

First published: 27 February 2024 | https://doi.org/10.1002/mus.28069



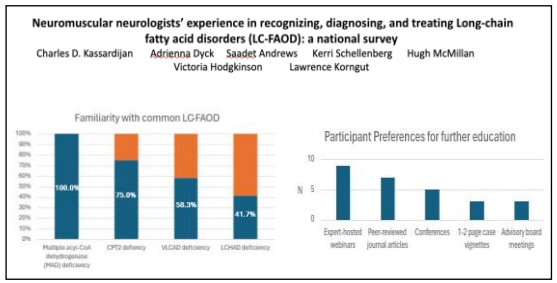
Journal of Neuromuscular Diseases xx (2021) x–xx
DOI 10.3233/JND-200617
IOS Press

Research Report

A Canadian Adult Spinal Muscular Atrophy Outcome Measures Toolkit: Results of a National Consensus using a Modified Delphi Method

PROOF

Jeremy Slayter[a,b], Victoria Hodgkinson[c], Josh Lounsberry[c], Bernard Brais[d,e], Kristine Chapman[f], Angela Genge[d,e], Aaron Izenberg[g], Wendy Johnston[h], Hanns Lochmüller[i,j], Erin O'Ferrall[d], Gerald Pfeffer[c,k], Stephanie Plamondon[c], Xavier Rodrigue[l], Kerri Schellenberg[m], Christen Shoesmith[h], Christine Stables[f], Monique Taillon[a,b], Jodi Warman-Chardon[i,j]



Neuromuscular neurologists' experience in recognizing, diagnosing, and treating Long-chain fatty acid disorders (LC-FAOD): a national survey

Charles D. Kassardjian | Adrienna Dyck | Saadet Andrews | Kerri Schellenberg | Hugh McMillan | Victoria Hodgkinson | Lawrence Korngut

Familiarity with common LC-FAOD

Participant Preferences for further education

➢ Patient Recruitment/ surveys

Factors Associated With Health-Related Quality of Life in Children With Duchenne Muscular Dystrophy

Journal of Child Neurology 2016, Vol. 31(7) 879-886
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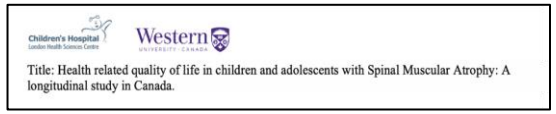
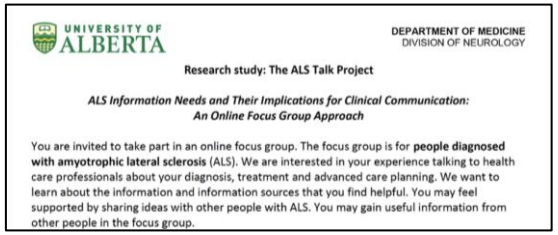
DOI: 10.1177/0883073815627879
jcn.sagepub.com

SAGE



Children's Hospital London Health Sciences Centre | Western University Canada

Title: Health related quality of life in children and adolescents with Spinal Muscular Atrophy: A longitudinal study in Canada.



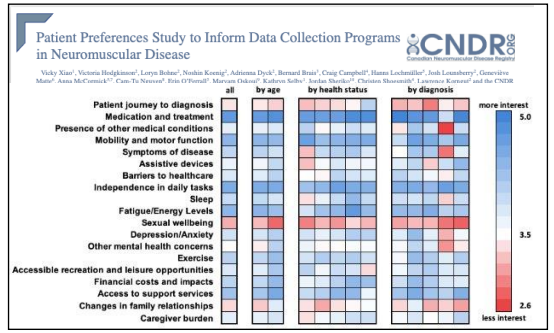
UNIVERSITY OF ALBERTA

DEPARTMENT OF MEDICINE
DIVISION OF NEUROLOGY

Research study: The ALS Talk Project

ALS Information Needs and Their Implications for Clinical Communication: An Online Focus Group Approach

You are invited to take part in an online focus group. The focus group is for **people diagnosed with amyotrophic lateral sclerosis** (ALS). We are interested in your experience talking to health care professionals about your diagnosis, treatment and advanced care planning. We want to learn about the information and information sources that you find helpful. You may feel supported by sharing ideas with other people with ALS. You may gain useful information from other people in the focus group.



Patient Preferences Study to Inform Data Collection Programs in Neuromuscular Disease

CNDR.org

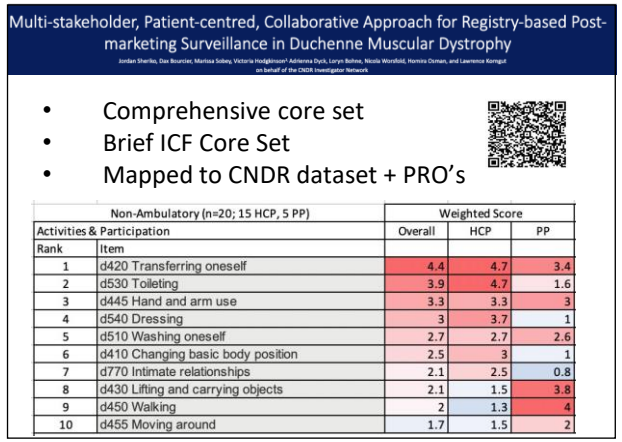
Canadian Neuromuscular Disease Registry

Patient journey to diagnosis
Medication and treatment
Presence of other medical conditions
Mobility and motor function
Symptoms of disease
Assistive devices
Barriers to healthcare
Independence in daily tasks
Sleep
Fatigue/Energy Levels
Sexual wellbeing
Depression/Anxiety
Other mental health concerns
Exercise
Accessible recreation and leisure opportunities
Financial costs and impacts
Access to support services
Changes in family relationships
Caregiver burden

all | by age | by health status | by diagnosis

more interest 5.0 — 3.5 — 2.6 less interest

➢ Upcoming Studies



Multi-stakeholder, Patient-centred, Collaborative Approach for Registry-based Post-marketing Surveillance in Duchenne Muscular Dystrophy

- Comprehensive core set
- Brief ICF Core Set
- Mapped to CNDR dataset + PRO's

Non-Ambulatory (n=20; 15 HCP, 5 PP)		Weighted Score		
Activities & Participation		Overall	HCP	PP
Rank	Item			
1	d420 Transferring oneself	4.4	4.7	3.4
2	d530 Toileting	3.9	4.7	1.6
3	d445 Hand and arm use	3.3	3.3	3
4	d540 Dressing	3	3.7	1
5	d510 Washing oneself	2.7	2.7	2.6
6	d410 Changing basic body position	2.5	3	1
7	d770 Intimate relationships	2.1	2.5	0.8
8	d430 Lifting and carrying objects	2.1	1.5	3.8
9	d450 Walking	2	1.3	4
10	d455 Moving around	1.7	1.5	2

THE CANADIAN NEUROMUSCULAR DISEASE REGISTRY: ACCESS to ALS DISEASE-MODIFYING THERAPY in CANADA

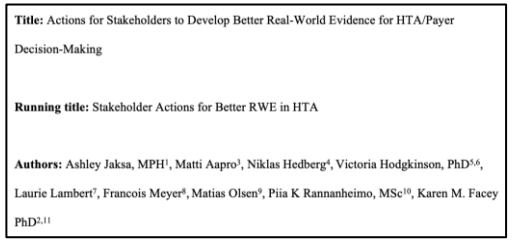
Real-world Survival Effectiveness of Edaravone in Amyotrophic Lateral Sclerosis: A Propensity Score Weighted, Registry-based, Canada-wide Cohort Study

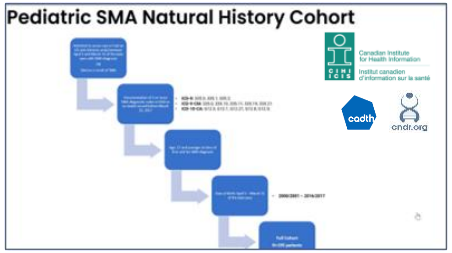
Characterizing SOD1 and c9ORF72 positive ALS: Findings from the Canadian Neuromuscular Disease Registry

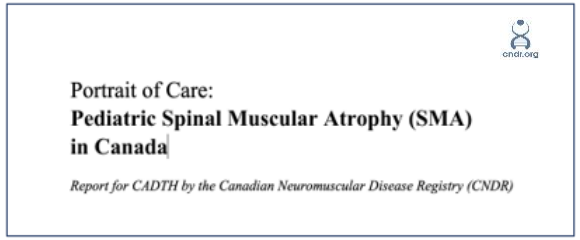
Title: Discontinuation of sodium phenylbutyrate/ursodoxicoltaurine (Albrioza) in Canadians with ALS in the Canadian Neuromuscular Disease Registry

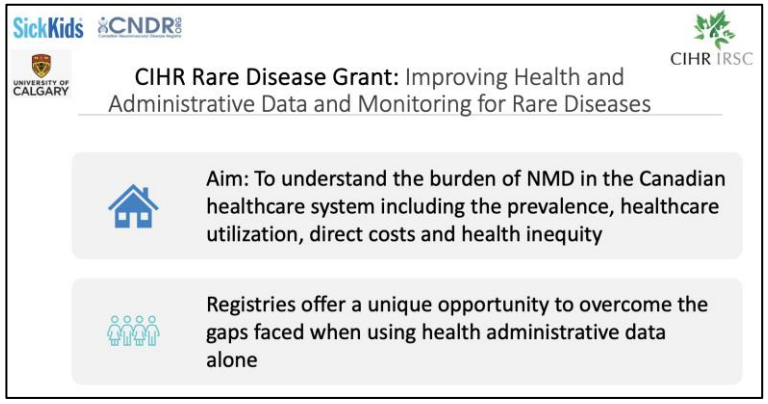
CNDR:
Leadership in RWD for Decision-Making

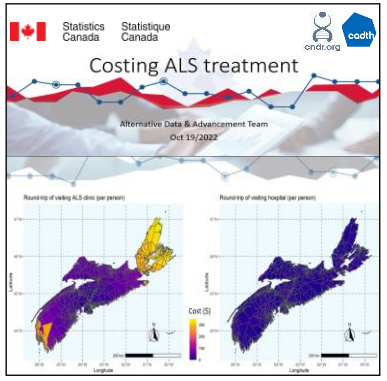
- Registry leadership in Canada with HTA
- Engagement & alignment internationally
- Improving use of robust data to support an understanding of high-cost therapies

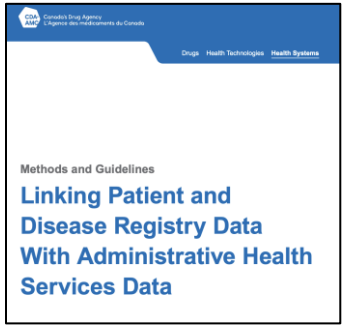


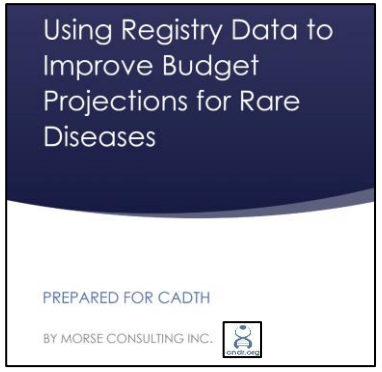












NMD4C

PROMOT

Performing a Rare Disease-Oriented Master Observational Trial

Dr. Cynthia Gagnon

Four (4) flagship diseases:

1. **Oculopharyngeal muscular dystrophy**
2. **Congenital myopathies**
3. **Congenital myasthenias**
4. **Myofibrillar myopathies**

Cynthia Gagnon
Anita Burgun
Hanns Lochmuller
Heidi Howard
Olivier Lambercy
Sébastien Gaboury
Martin Vallières
Santa Slokenberga

Daniel Natera de Benito
Ignacio Escuder Bueno
Louis Bédard
Homira Osman
Elizabeth Angelevski
Félix Camirand-Lemyre
Guillaume Bassez
Capucine Trollet

Horizon Europe

EJP RD

Project Overview

Objective

To develop a precision medicine approach integrating ethical, legal, and social considerations to address challenges in prognosis and clinical trial readiness for rare diseases, through a Learning and Knowledge Mobilization Platform (LEAP FORWARD) based on the adapted Master Observational Trial (MOT).

The proposal will support the identification of biomarkers / predictors for prognosis and documentation of the evolution of a group of four (4) neuromuscular disorders as well as a common standardized protocol for natural history study and clinical trials.

Project Overview

Project Phases

- **Phase 1:** Identify and classify signs and symptoms specific to target diseases, enabling multimodel data collection for optimal disease impact assessment.

- **Phase 2:** Test MOT among four neuromuscular diseases in two countries (Canada, Spain) to validate feasibility in rare diseases.

BIND STUDY: Assessing the Indirect Burden of Inherited & Inflammatory Neuromuscular Disease in Canada (Dr. Jodi Warman-Chardon)

Survey development

Ethics Approval

Recruitment Strategy -> Pilot Study with MDC

Bind 1.0*

5 years
-Mobility
-Vehicle
-Home

Bind 2.0

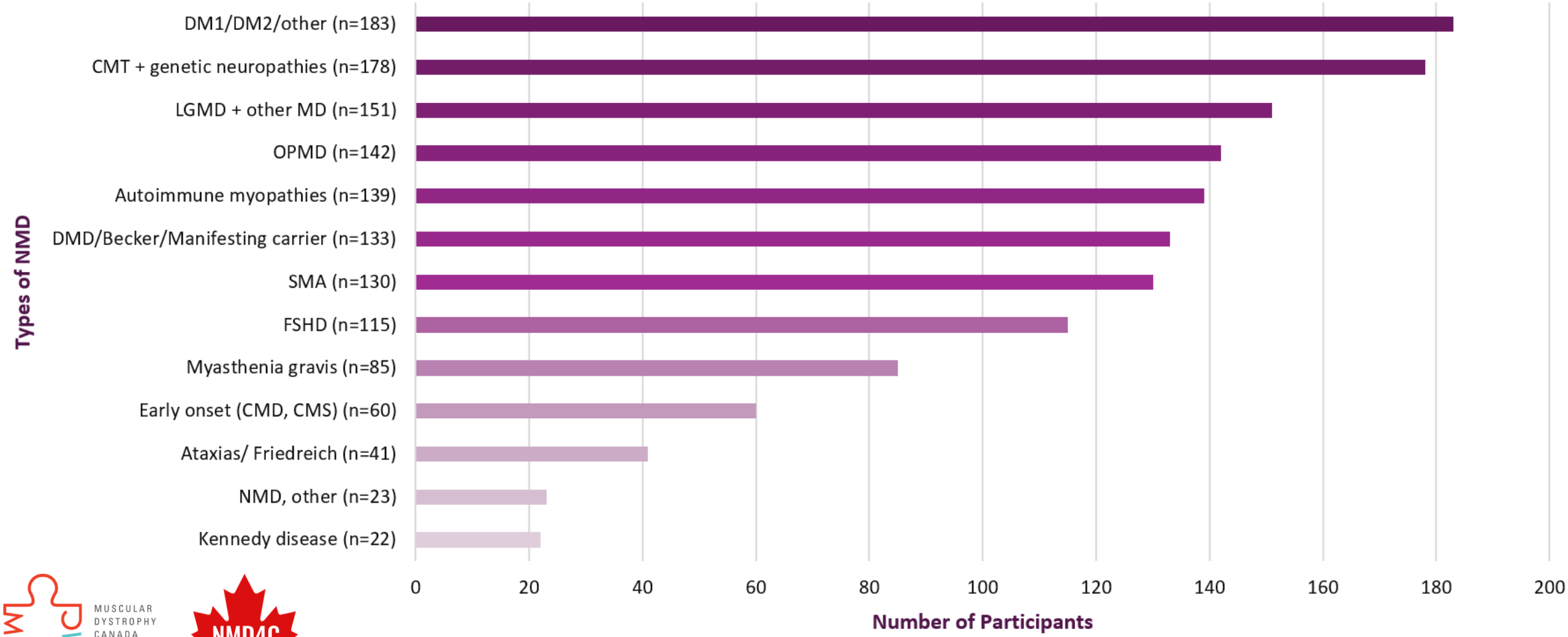
Data Analysis

MUSCULAR DYSTROPHY CANADA
DYSTROPHIE MUSCULAIRE CANADA

NMD4C

BIND : a national, cross-sectional survey of individuals living with a NMD and their caregivers

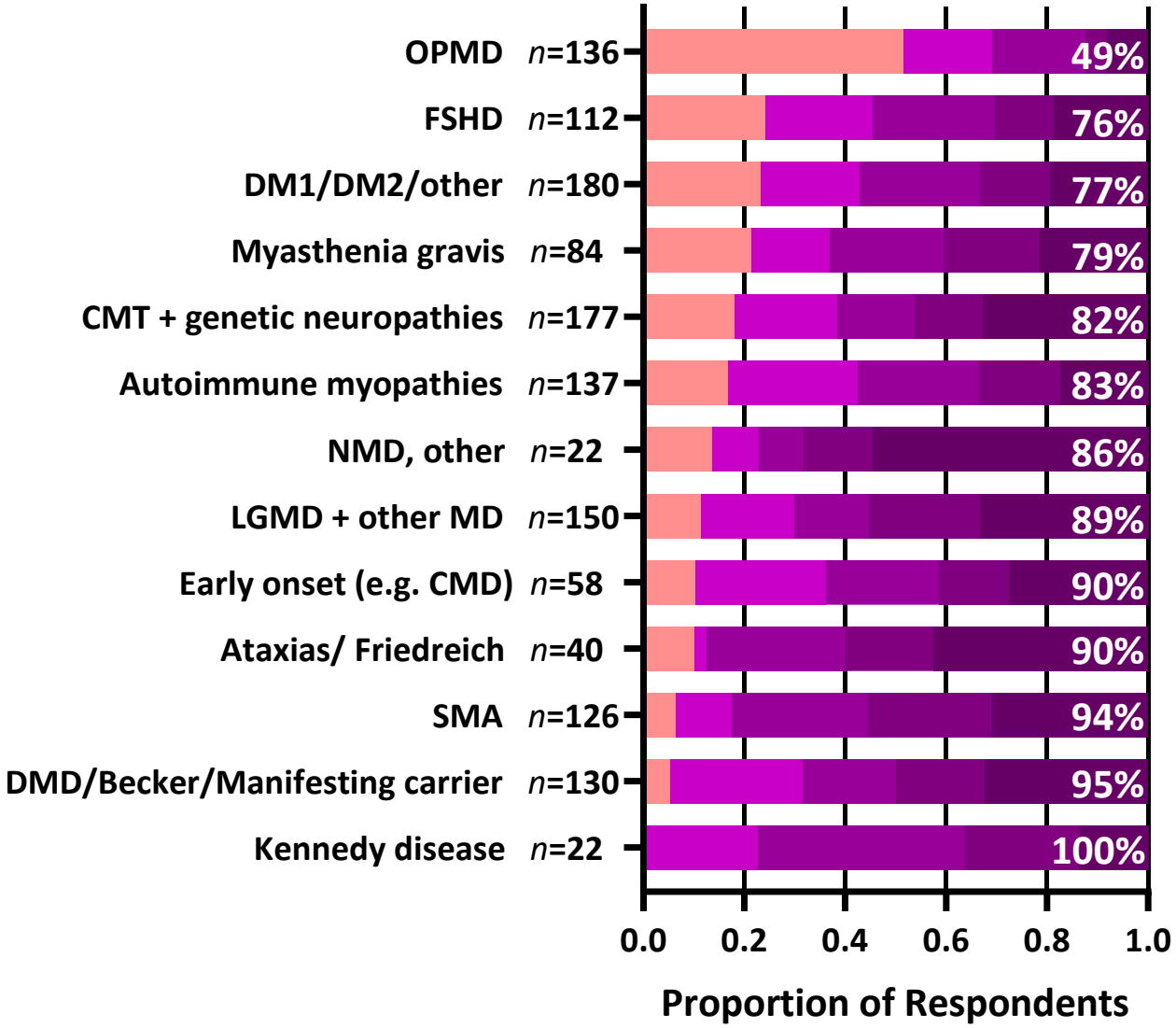
NMD Subtypes: Survey Respondents for BIND



MUSCULAR DYSTROPHY CANADA
DYSTROPHIE MUSCULAIRE CANADA

NMD4C

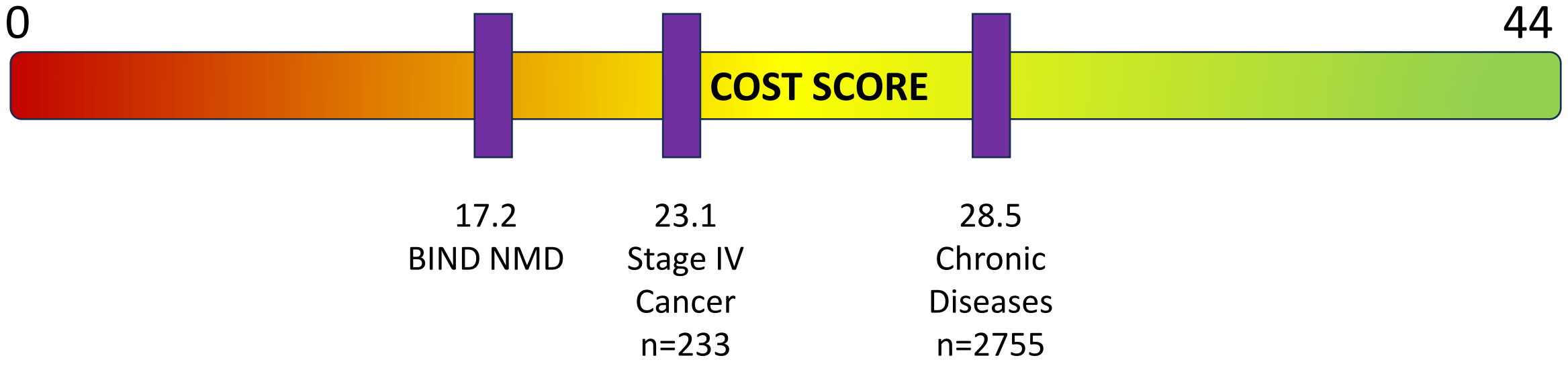
Impact of NMD causes financial hardship



MUSCULAR DYSTROPHY CANADA
DYSTROPHIE MUSCULAIRE CANADA

NMD4C

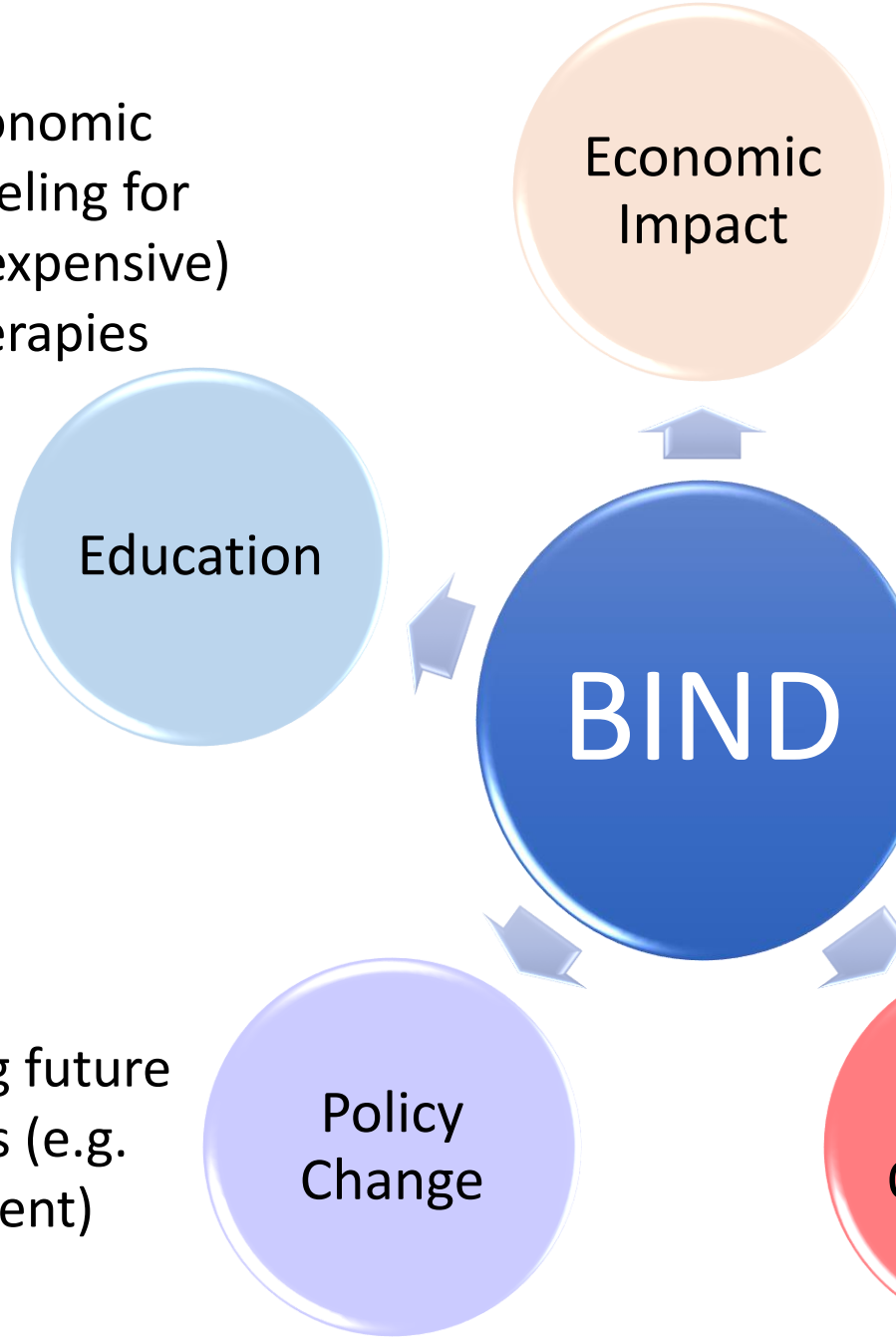
Comprehensive Score for Financial Toxicity (COST): High Financial Toxicity in NMD



Lower COST scores and higher financial toxicity associated with
"some" high school education or completed high school, unable to work, unemployed or a student, lower household incomes, single, widowed, divorced or separated, and no private health insurance

Pavela et al., Health Serv Ins, 2021
De Souza et al., Cancer, 2017

Impact: Informs Care for NMD Stakeholders



Economic modeling for new (expensive) therapies

Assemble one the largest cohort of people with NMD

Enable cross-jurisdictional comparisons

Forecasting future resources (e.g. equipment)

Real-World Evidence for Canadian Neuromuscular Disease (CIHR-Dr. Amin)

NMD4C

MUSCULAR DYSTROPHY CANADA
DYSTROPHIE MUSCULAIRE CANADA

Highly Elevated Prevalence of Spinobulbar Muscular Atrophy in Indigenous Communities in Canada Due to a Founder Effect

Jamie N. Leckie, BSc, Matthew M. Joel, BSc, Kristina Martens, BSc, Alexandra King, MD, Malcolm King, PhD, Lawrence W. Korngut, MD, MSc, A.P. Jason de Koning, PhD, Gerald Pfeffer, MD, PhD,* and Kerri L. Schellenberg, MD, MMedEd*

Correspondence
Dr. Pfeffer
gerald.pfeffer@ucalgary.ca

Neurol Genet 2021;7:e607. doi:10.1212/NXG.0000000000000607

83% of SBMA patients in neuromuscular clinics are Indigenous

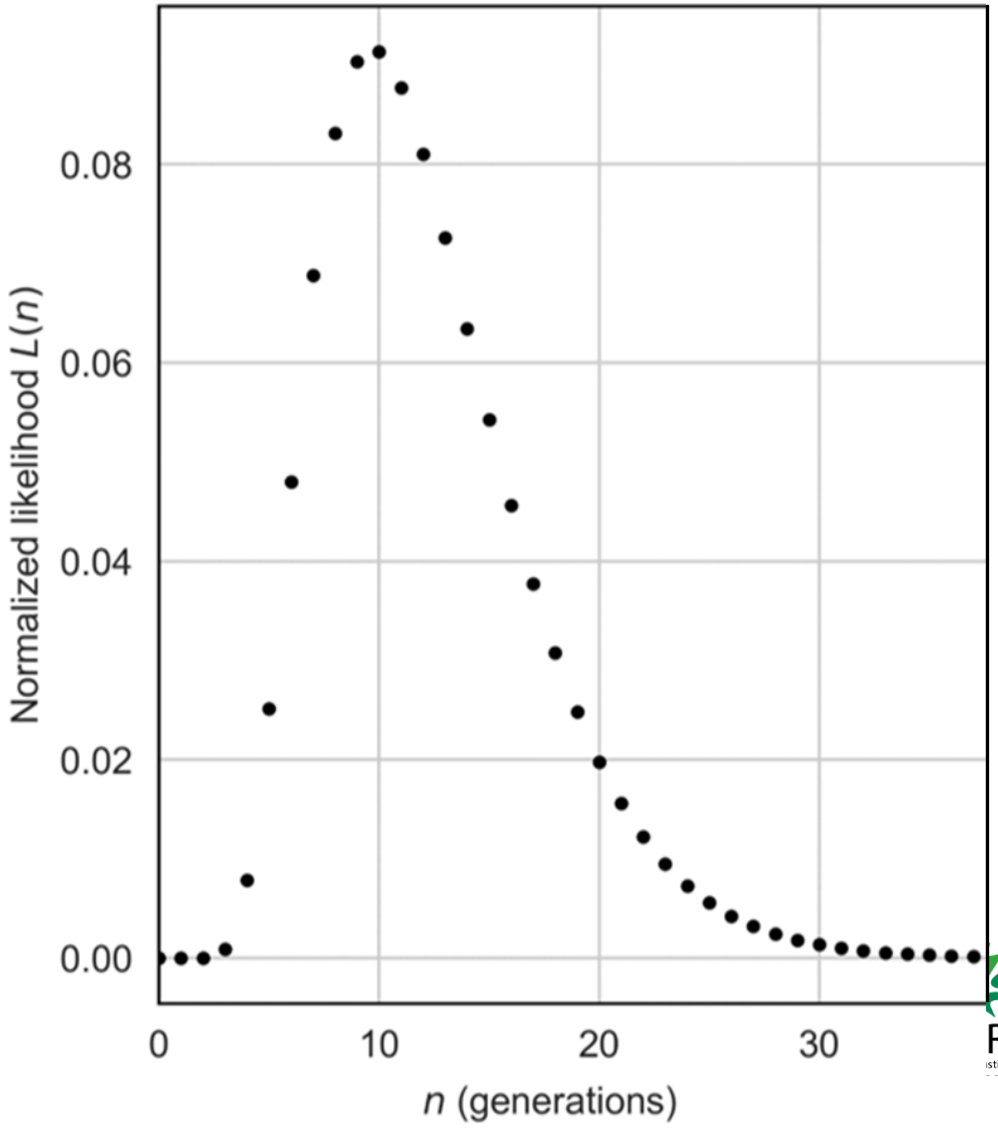
Highest prevalence in world identified in Saulteaux First Nation (186/100,000)

Likely to be underestimated

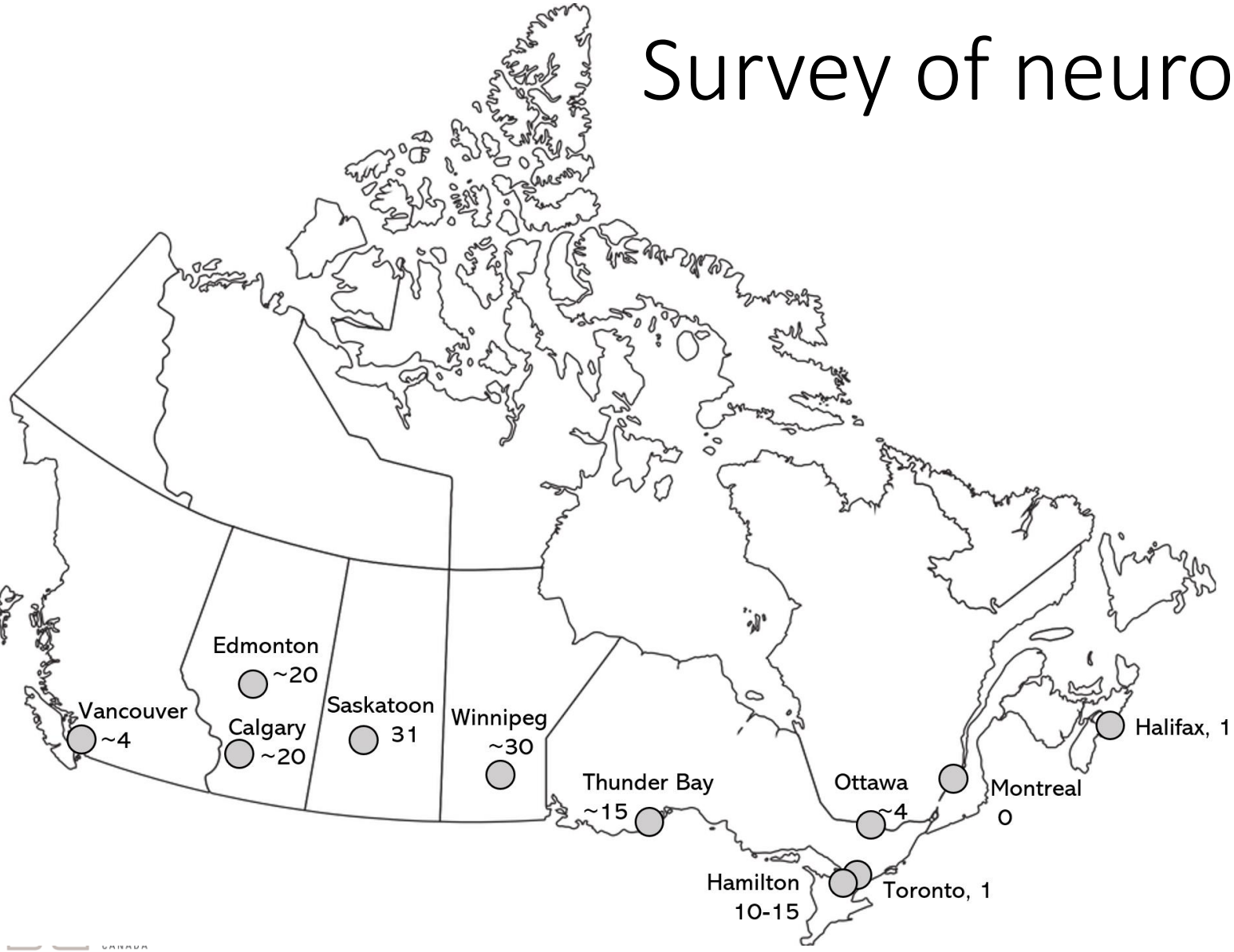
Leckie et al 2021

A shared haplotype is present across most cases of SBMA with Indigenous heritage

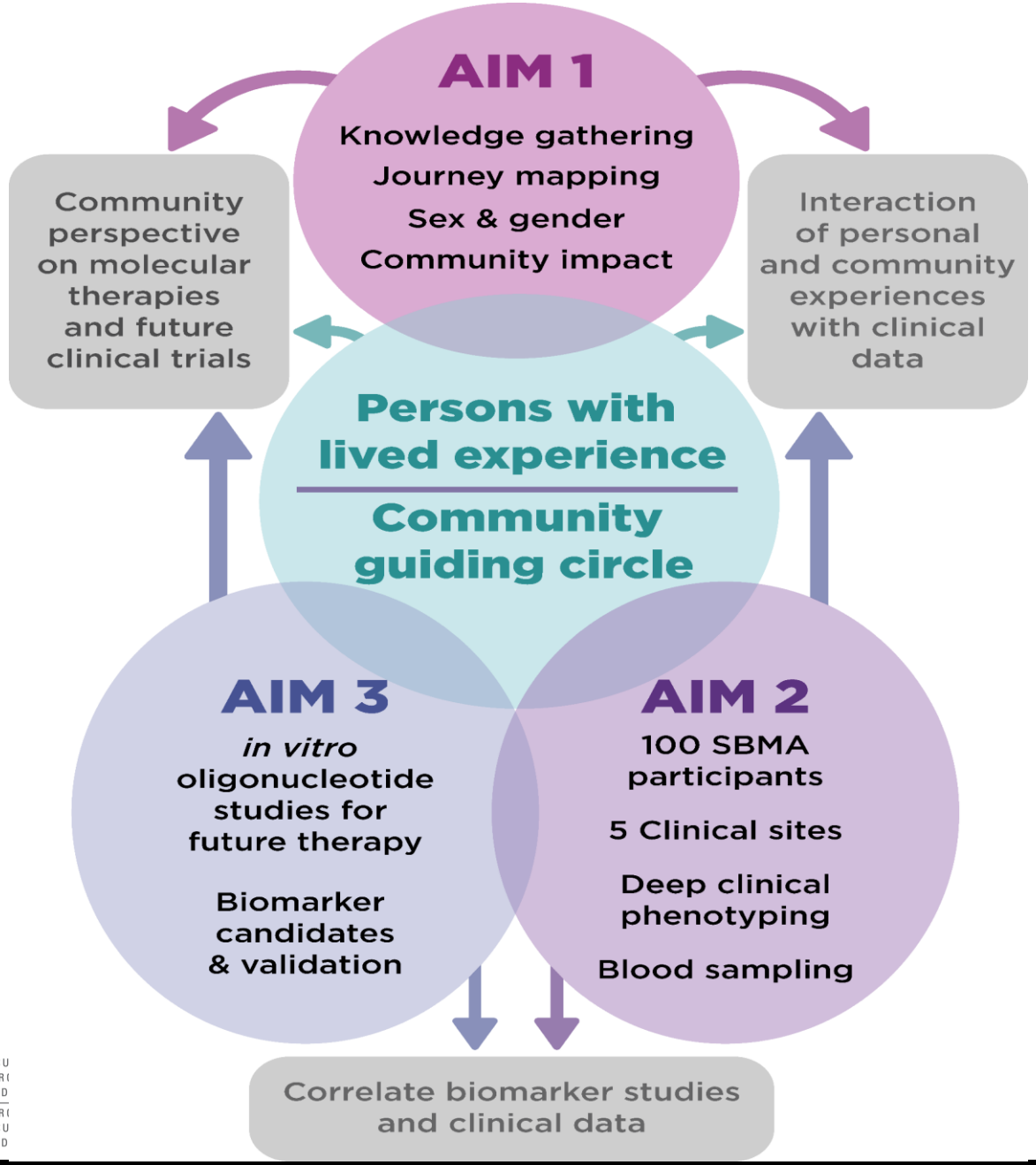
ID	DXS1213	DXS1194	*AR* (CAG)	*AR* (GGC)	DXS1111	DXS135	DXS1125	Ethnicity
1	18.5	13.5	55	23	13	15	30	Saulteaux
2	18.5	13.5	52	23	13	15	30	Cree
3	18.5	13.5	44	23	13	15	30	Cree/Saulteaux
4	18.5	13.5	52	23	13	15	30	Cree
5	18.5	13.5	52	23	13	15	30	Cree
6	18.5	13.5	54	NA	13	15	30	Saulteaux
7	18.5	13.5	44	23	13	15	30	Cree
8	18.5	13.5	40	NA	13	15	30	Indigenous
9	18.5	13.5	49	23	13	15	30	Cree/French
10	18.5	13.5	43	23	13	16	31	Cree
11	18.5	13.5	42	23	14	16	31	Cree
12	18.5	NA	41	23	14	16	31	Cree
13	18.5	13.5	52	23	15	15	30	Cree
14	17.5	18	41	23	13	16	31	Czech
15	17.5	14.5	40	23	16	16	37	Swedish
16	17.5	13.5	40	16	15	16	31	Unknown
17	17.5	13.5	42	26	14	16	31	Northern European
18	17.5	18	41	16	14	16	31	Métis/European
19	17.5	18	44	16	14	16	37	Métis/English
20	17.5	14.5	41	22	15	16	30	Métis/Scottish
21	17.5	15.5	45	16	13	15	28	Indigenous
C1	17.5	19	18	21	14	15	27	
C2	17.5	14.5	19	16	14	16	31	
C3	18.5	15.5	35	23	16	16	31	
C4	17.5	17.5	17	19	14	15	28	
C5	18.5	18.5	20	23	13	15	29	



Survey of neuromuscular clinics



Cree traditional territory.
(courtesy Victor Temprano/Native-Land.ca)



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Thank you!

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