



IDMC 15
Meeting 2026

Scientific **PROGRAM**

May 26 to 30, 2026

Hôtel Le Montagnais,
Saguenay, Québec, Canada

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Tuesday, 26 May 2026

09:00 - 13:00 **Pharma Day with Euro-DyMA and MDF**

09:00 - 16:00 **Registration**
Reception Hall

13:00 - 14:00 **Global alliance In-Person Meeting**

16:00 - 22:00 **Welcome Reception in La Baie**
La Baie

Wednesday, 27 May 2026

08:00 - 16:00 **Registration**
Reception Hall

08:15 - 08:30 **Introduction**
La Montagnaise

08:30 - 09:00 **Invited Speakers - Past IDMC organizers**
La Montagnaise

09:00 - 10:00 **Session: Pathogenic Mechanisms I**
La Montagnaise

Single-Molecule Analysis Reveals Asymmetric CpG Methylation at the DM1 Locus Driven by Somatic Instability [Thomas D. Hoekman](#)¹ 80

¹Department of Medical BioSciences, Radboud University Medical Center, Nijmegen, The Netherlands

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¹Institut des cellules souches pour le traitement et l'étude des maladies monogéniques (I-Stem), Corbeil-Essonnes

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¹Laboratory of Gene Therapy, Institute of Molecular Biology and Biotechnology, Adam Mickiewicz University in Poznań, Poland

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¹Massachusetts General Hospital, ²Harvard Medical School

10:00 - 10:30 **Speed Dating**
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¹University of Auckland

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¹University of Rochester

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¹Department of Clinical and Health Psychology and Research Methodology; Faculty of Psychology, University of the Basque Country (EHU), Donostia - San Sebastian, Spain, ²Centro de Investigación Biomédica en Red sobre Enfermedades Neurodegenerativas (CIBERNED), Institute Carlos III, Madrid, Spain

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¹Maastricht University Medical Center

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¹Maastricht University Medical Center

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First Canadian Data from the DM-Scope Registry: Clinical and epidemiological profile of Myotonic dystrophy type 1 Marianne Nury ^{1,2} ¹ Department of Medicine, Faculty of Medicine, Université Laval, Quebec City, Canada, ² Centre intégré universitaire de santé et de services sociaux de la Capitale-Nationale, Québec, QC, Canada	<u>213</u>

Uncovering DMPK-Dependent Mitochondrial Defects in Myotonic Dystrophy Type 1 Muscle Stem Cells [Pauline Garcia](#)^{1,2} ¹CHU Sainte Justine, ²Département de Physiologie et Pharmacologie, Université de Montréal 214

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Integrating proteomics and structure prediction to determine the interactome of proteins involved in muscular dystrophies and myopathies [Yasmine Amina Bachir](#)¹ ¹INRS - Centre Armand-Frappier - Santé Biotechnologie, Unité mixte de recherche (UMR) INRS- UQAC en santé durable 218

Cognitive and social decision-making skills in adult-onset Myotonic Dystrophy type 1 [Maria Carolina Fontana Antunes de Oliveira](#)¹ ¹Université Paris Cité - Memory, Brain and Cognition Laboratory (MC2 Lab) 219

The GRIMN Biobank: A Longitudinal Biobank Coupling Biospecimens with Comprehensive Clinical Phenotyping in DM1 [Melissa Verreault](#)¹ ¹Groupe de recherche interdisciplinaire sur les maladies neuromusculaires (GRIMN), Center for Research and Innovation of the Saguenay-Lac-St-Jean Integrated University Health and Social Services Center, Saguenay, Quebec, Canada 224

12:15 - 13:00

Lunch

Totem Nord-Réserve Centre

13:15 - 13:30

Flash Talk

La Montagnaise

Investigating the basis of sleep dysregulation in myotonic dystrophy type 1 [Belinda Pinto](#)¹ ¹University of Florida 8

A cross-sectional study exploring the alignment between patient-reported and biomechanical dysphagia outcomes in myotonic dystrophy type1 (DM1) [Jodi Allen](#)¹ ¹The National Hospital for Neurology and Neurosurgery, University College NHS Foundations Trust, UK 95

Growth, development, and social participation in congenital and childhood-onset myotonic dystrophy type 1: A nationwide survey in Japan [Yuzuha Ichimura](#)¹ ¹Clinical Neurophysiology, The University of Osaka Graduate School of Medicine 119

DM1 RAN Proteins Accumulate in Autopsy Brain Regions with Degenerative and Neuroinflammatory Changes and Promote Tau Aggregation in Neuronal Cells [Monica Banez-Coronel](#)¹ ¹University of Florida 190

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Patient Engagement in Research

La Montagnaise

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Session: Children with DM

La Montagnaise

Natural history of oropharyngeal dysphagia in children with congenital and childhood myotonic dystrophy type 1: a 3-year longitudinal cohort study [Saskia Scholten](#)¹ ¹Department of Rehabilitation, Donders Institute for Brain, Cognition and Behaviour, Radboud university medical center, Amalia Children's Hospital, Nijmegen, The Netherlands 40

On the Move: Longitudinal Insights Advancing the Understanding of Motor Function in Paediatric Myotonic Dystrophy Type 1 [Lynn Orriëns](#)¹
¹Department of Pediatric Neurology, Amalia Children's Hospital, Radboud University Medical Centre, Nijmegen, the Netherlands

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Cognitive and Behavioral Impairment in Congenital and Childhood-Onset Myotonic Dystrophy Type 1: A Longitudinal Analysis [Anna Falco](#)¹ ¹The NeMO Center in Rome, U.O.C. Neuropsichiatria Infantile, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, Rome, Italy.

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Step Back in Time: The Story of Val-Jalbert and Saguenay–Lac-Saint-Jean
Val-Jalbert

Thursday, 28 May 2026

08:00 - 16:00

Registration

Reception Hall

08:15 - 08:30

Introduction

La Montagnaise

08:30 - 09:00

Invited Speaker - Roman H. Khonsari

La Montagnaise

09:00 - 10:15

Session: Development of Biomarkers and Clinical Outcome Assessments I

La Montagnaise

Clinical Validation of Socially Assistive Robot-Administered Physical Tests in Myotonic Dystrophy Type 1 [Killian LACHAUX](#)¹ [31](#)

¹Département d'Informatique et Mathématique, Université du Québec à Chicoutimi, Chicoutimi, Québec, Canada.

Multi-omic profiling stratified by the Splicing Index enables circulating biomarker discovery in myotonic dystrophy type 1 [Melissa Hale](#)¹ [118](#)

¹Center for Inherited Myology Research, Department of Neurology, Virginia Commonwealth University, USA

DM1-Hub registry: building a nationwide infrastructure to define the natural history of myotonic dystrophy type 1 [Gisela Nogales-Gadea](#)¹ [156](#)

¹Badalona Neuromuscular Research Group (GRENBA), Germans Trias i Pujol Research Institute (IGTP), Badalona, Catalonia, Spain

Cerebrospinal fluid multi-omic biomarkers of myotonic dystrophy type 1 [Preeti Kumari](#)¹ [187](#)

¹Massachusetts General Hospital, Harvard Medical School

Disease progression in Myotonic Dystrophy Type 2 [Johanna Hamel](#)¹ [207](#)

¹University of Rochester Medical Center

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Coffee Break

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Session: Clinical Manifestations, Activity and Participation I

La Montagnaise

Cognitive and Social cognition functioning in adults with childhood myotonic dystrophy type 1 [Simon-Pierre Gagnon](#)^{1,2} [56](#)

¹Groupe de Recherche Interdisciplinaire sur les Maladies Neuromusculaires (GRIMN), ²Faculté de médecine et des sciences de la santé, Université de Sherbrooke (UdS)

A Multidimensional Profile of Dysphagia in Myotonic Dystrophy Type 1 (SwallowDM1) [Jodi Allen](#)¹ [100](#)

¹The National Hospital for Neurology & Neurosurgery, University College London NHS Foundation Trust, UK

Preimplantation Genetic Testing in Myotonic Dystrophy Type 1: Clinical Outcomes and Insights [Johanna Bruijines](#)¹ [126](#)

¹Department of Neurology and MHeNS School for Mental Health and Neuroscience, Maastricht University Medical Centre+, Maastricht, The Netherlands

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Session: Cell and Animal Models for DM

La Montagnaise

A repressible CUG repeat RNA mouse model to study the neurological manifestations and their reversibility in myotonic dystrophy type 1 [Larissa Nitschke](#)¹ [11](#)

¹Baylor College of Medicine

Bioengineered 3D Muscle Tissues Identify an MBNL1-Independent Mechanism of Calcitriol-Mediated Myotonia Rescue [Juan M. Fernández-Costa](#)^{1,2} [24](#)

¹Institute for Bioengineering of Catalonia (IBEC), ²Barcelona Institute of Science and Technology (BIST)

Transcriptomic and molecular characterization of a neuronal mouse model for myotonic dystrophy type 1 (DM1) [Juan D. Arboleda](#)¹ [108](#)

¹Department

of Molecular Genetics & Microbiology, Center for NeuroGenetics, University of Florida, Gainesville FL 32610

Consequences of congenital myotonic dystrophy during neuromuscular development [Caroline Hermitte](#)¹ ¹Institut des cellules souches pour le traitement et l'étude des maladies monogéniques (I-Stem), Corbeil-Essonnes 122

RNA and RAN Protein Gain-of-Function Effects in a Novel DM2 BAC Transgenic Mouse Model [Hannah Gollhofer](#)^{1,2} ¹Center for NeuroGenetics, University of Florida, ²Department of Molecular Genetics and Microbiology, University of Florida 179

12:45 - 13:45 **Lunch**
Totem Nord-Réserve Centre

13:45 - 15:00 **Session: Pathogenic Mechanisms II**
La Montagnaise

Evidence of nuclear DMPK transcript degradation and cytoplasmic export in DM1 cells using dSTORM Super Resolution Microscopy [Petter Hamilton-Stanley](#)¹ ¹University of Nottingham 90

Axon initial segment disruption and impaired vesicle transport reveal neuron-intrinsic mechanisms of brain dysfunction in DM1 [Louison Daussy](#)¹ ¹Sorbonne University, Inserm, Centre de Recherche en Myologie, Paris, France 128

Toxic CUG RNA repeats disrupt developmentally regulated splicing in oligodendrocytes causing transient hypomyelination in a mouse model of myotonic dystrophy [Gabriele Ordazzo](#)¹ ¹Sorbonne Université, Inserm, Institut de Myologie, Centre de Recherche en Myologie, Paris, France 134

Converging mechanisms of DMPK and TCF4 CTG repeat expansions underpin Fuchs endothelial corneal dystrophy [Christina Zarouchlioti](#)¹ ¹UCL Institute of Ophthalmology, London, UK 162

Autism-related traits in myotonic dystrophy type 1 [Łukasz Sznajder](#)¹ 165
¹University of Nevada, Las Vegas, NV, USA

15:00 - 15:15 **Flash Talk**
La Montagnaise

Evaluating drug potential of macrolide azithromycin in Myotonic Dystrophy 1 (DM1) [Tushar Ghosh](#)¹ ¹University of Nottingham 85

Methylation of CCG variant repeats is associated with heterogeneous methylation of CpG sites surrounding DMPK expansion in DM1 patients [Jovan Pesovic](#)¹ ¹Center for Human Molecular Genetics, University of Belgrade-Faculty of Biology, Belgrade, Serbia 173

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Apathy as a Distinct Executive Phenotype in Adult Myotonic Dystrophy Type 1 [Melissa M. Dixon](#)¹ ¹University of Utah 206

Proteogenomic Discovery of Splice-Junction Peptides as Novel Biomarkers in Cerebrospinal Fluid of Myotonic Dystrophy Type 1 (DM1) [Marwa Zafarullah](#)¹ ¹Department of Neurology and Neurological Sciences, Stanford School of Medicine, Stanford, CA 210

15:15 - 16:15 **Late-Breaking session**
La Montagnaise

Long-Read Sequencing Reveals Elevated Mitochondrial DNA Structural Variations Associated with Disease Severity in Myotonic Dystrophy Type 1 [Wenhan Zhang](#)^{1,2} ¹Fudan University, ²Huashan Hospital, Fudan University 220

Data from the initial cohort of the FREEDOM2-DM1 multiple-ascending 222

dose trial of PGN-EDODM1. [James McArthur](#)¹ ¹PepGen Inc., Boston, MA

Multi-Omics CSF Profiling Reveals CNS Dysfunction Candidate Biomarkers in Myotonic Dystrophy Type 1 (DM1) [Marwa Zafarullah](#)¹

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¹Department of Neurology and Neurological Sciences, Stanford School of Medicine, Stanford, CA

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Poster and Networking Session

Tipi-tente-wigwam-Réserve Sud/Nord

Balance and coordination in myotonic dystrophy type 1 [Sarah Mollet](#)¹

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¹University of Auckland

The FREEDOM-DM1 clinical trial demonstrated unprecedented splicing correction with single doses of PGN-EDODM1, with an acceptable safety profile. [Johanna Hamel](#)¹ ¹University of Rochester

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Differentiated Cognitive Profiles in Myotonic Dystrophy Type 1: A Cluster Analysis [Irati Larrañaga](#)^{1,2}

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¹Department of Clinical and Health Psychology and Research Methodology; Faculty of Psychology, University of the Basque Country (EHU), Donostia - San Sebastian, Spain, ²Centro de Investigación Biomédica en Red sobre Enfermedades Neurodegenerativas (CIBERNED), Institute Carlos III, Madrid, Spain

Respiratory function in Myotonic dystrophy type 1- a prospective single center study [Olöf Eliasdóttir](#)¹

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¹Department of Clinical Neuroscience, Institute of Neuroscience and Physiology, Sahlgrenska Academy, University of Gothenburg, Blåa stråket 7, 41345, Gothenburg, Sweden

Investigating the basis of sleep dysregulation in myotonic dystrophy type 1 [Belinda Pinto](#)¹

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¹University of Florida

Harmonization and federated analysis of myotonic dystrophy registries to model heterogeneous disease trajectories. Results from the 287th ENMC International Workshop [Leandre Fontaine](#)¹

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¹Maastricht University Medical Center

Responsiveness and the minimal clinically important difference of the DM1-ActivC in Myotonic Dystrophy type 1 [Leandre la Fontaine](#)¹

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¹Maastricht University Medical Center

Natural history of PR interval and QRS duration in Myotonic Dystrophy Type 1 [David Bovenkerk](#)¹

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¹1. Department of Cardiology, Cardiovascular Research Institute Maastricht (CARIM), Maastricht University Medical Centre, Maastricht, The Netherlands

Addressing gastrointestinal symptoms in myotonic dystrophy: consensus-based recommendations for clinical practice and research

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[Saskia Scholten](#)¹ ¹Department of Rehabilitation, Donders Institute for Brain, Cognition and Behaviour, Radboud university medical center, Amalia Children's Hospital, Nijmegen, The Netherlands

Investigating the role of the transcriptional kinase, CDK13, in mutant DMPK transcription and RNA foci formation. [Jessie Brown](#)¹

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¹University of Nottingham

Hepatic insulin resistance in myotonic dystrophy type 1 is highly associated with acquired factors [HIROTO TAKADA](#)¹

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¹NHO Aomori National Hospital

Intensive speech therapy training in people with myotonic dystrophy type 1. [Simone Knuijt](#)¹

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¹Donders institute for brain, cognition and behavior, Radboud university medical center, Department of rehabilitation, Nijmegen, the Netherlands.

Measurement properties and clinical relevance of outcome measures for myotonia in myotonic dystrophy type 1: protocol of the MyoMeasure study [Elise Taken](#)¹

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¹Radboud University Medical Center, the Netherlands

Assessment of sleep-wake patterns in adults with the infantile-onset form of myotonic dystrophy type 1: an actigraphy study [Luc Laberge](#)¹

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¹ÉCOBES - Recherche et transfert, Cégep de Jonquière

Serum Proteomic Profiling Reveals Acute Inflammatory Response as a Key Pathogenic Mechanism in Myotonic Dystrophy Type 1 Giovanni Meola ^{1,2} ¹ Department of Biomedical Sciences for Health, Università degli Studi di Milano, Milan, Italy., ² Department of Neurorehabilitation Sciences, Casa Di Cura Igea, Milan, Italy	19
Splicing modulation of DMPK exon 15 as a therapeutic approach for myotonic dystrophy type 1 Mireia Gromaz Iborra ^{1,2} ¹ Department of Gene Expression, Adam Mickiewicz University, Poznan, Poland, ² Institute of Molecular Biology and Biotechnology	20
Gut microbiota profiles in children with myotonic dystrophy type 1 are marked by reduced Faecalibacterium abundance Anne M. van Uden ^{1,2} ¹ Department of Paediatric Neurology, Donders Institute for Brain, Cognition and Behaviour, Radboud university medical center, Amalia Children's Hospital, Nijmegen, the Netherlands, ² Department of Medical BioSciences, Radboud university medical center, Nijmegen, the Netherlands	21
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Feasibility, Acceptability, and Reproducibility of Rehabilitation Programs for Individuals with Myotonic Dystrophy Type 1: A Scoping Review Anne-Gabrielle Brassard ¹ ¹ 1. School of Rehabilitation Sciences, Faculty of Medicine, Université Laval, Quebec City, Quebec, Canada. 2. Center for Interdisciplinary Research in Rehabilitation and Social Integration (Cirris), Capitale-National Integrated University Health and Social Services Center, Quebec City, Quebec, Canada	30
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18:30 - 23:00

Bites, Sips & Saguenay Vibes

Downtown Chicoutimi

18:30 - 23:00

Special Activity for Trainees

CORAMH

Friday, 29 May 2026

08:15 - 08:30

Introduction

La Montagnaise

08:30 - 09:00

Invited Speaker - Jason Robert Guertin

La Montagnaise

09:00 - 10:00

Session: Development of Biomarkers and Clinical Outcome Assessments II

La Montagnaise

Developing digital endpoints to assess ambulation in DM1: analytical validation and feasibility of using a wearable sensor in daily living [61](#)
Stéphane Motola¹ ¹Sysnav, Vernon, France

Beyond balance: exploring cerebellar cognition in Myotonic Dystrophy Type 1 [132](#)
Carola Rita Ferrari Aggradi¹ ¹NeMO Clinical Center, Milan, Italy

Cross-sectional and 3-year longitudinal analysis of RNA mis-splicing in vastus lateralis muscle in a DM1 cohort [141](#)
Cécilia Légaré^{1, 2, 3, 4} ¹Centre de Recherche du Centre Hospitalier de l'Université Laval (CR-CHUL), Axe Neurosciences, Université Laval, Québec City, Canada, ²École des sciences de la réadaptation, Faculté de médecine, Université Laval, Québec, Canada, ³Groupe de Recherche Interdisciplinaire sur les Maladies Neuromusculaires (GRIMN), Centre intégré universitaire de santé et de services sociaux du Saguenay-Lac-Saint-Jean, Saguenay, Québec, Canada, ⁴RNA Institute, College of Arts and Sciences, University at Albany-SUNY, Albany, NY 12222, USA

Multicenter Multimodal Imaging Reveals System-Level CNS Disruption in Pediatric Myotonic Dystrophy Type 1 [203](#)
Tahereh Kamali¹ ¹Stanford University

10:00 - 10:30

Coffee Break

10:30 - 11:30

Session: Clinical Manifestations, Activity and Participation II

La Montagnaise

Frailty in DM1: Prevalence and Associations with Disease-Specific Factors [3](#)
Irati Larrañaga^{1, 2} ¹Department of Clinical and Healthy Psychology and Research Methodology Psychology Faculty, University of the Basque Country, ²Centro de Investigación Biomédica en Red sobre Enfermedades Neurodegenerativas (CIBERNED), Institute Carlos III, Madrid, Spain.

Everyday Cognitive Failures in Myotonic Dystrophy Type 1 (DM1): A Longitudinal Study [86](#)
STEFAN WINBLAD¹ ¹Department of Psychology, University of Gothenburg, Sweden

Vitamin D Deficiency and Respiratory Muscle Dysfunction in Myotonic Dystrophy Type 1 [114](#)
Daniel Jaraj¹ ¹Department of Neurology, Sahlgrenska University Hospital, Gothenburg university, Sweden

Identifying an Appropriate Patient-Reported Outcome Measure for Oropharyngeal Dysphagia in Myotonic Dystrophy Type 1 [148](#)
Claudia Côté^{1, 2}
¹Université Laval, ²GRIMN

11:30 - 12:30

Session: Clinical Management, Rehabilitation and Quality of Life Improvement

La Montagnaise

Energy Expenditure and the Accuracy of Predictive Equations in Myotonic Dystrophy Type 1 [53](#)
Isis B.T. Joosten¹ ¹Department of Neurology and School for Mental Health and Neuroscience, Maastricht University Medical Centre+, Maastricht, The Netherlands

Feasibility, acceptability and effects of a telerehabilitation-based Respiratory Training Program in Myotonic Dystrophy Type 1 [71](#)
Elie Fiogbé^{1, 2, 3} ¹Groupe de recherche interdisciplinaire sur les maladies neuromusculaires (GRIMN), Saguenay-Lac-St-Jean Integrated University Health and Social Services Center, Saguenay, Quebec, Canada, ²School of Rehabilitation, Faculty of Medicine and Health Sciences, Université de Sherbrooke, Sherbrooke, Quebec, Canada,

³Centre de recherche du Centre hospitalier universitaire de Sherbrooke (CRCHUS),
Université de Sherbrooke, Québec, Canada

The Current Landscape of Perinatal Information Provision and Genetic Counseling in Congenital Myotonic Dystrophy Type 1 in Japan [Ayumi Yonei](#)¹ [1](#)Department of Genetic Counseling, The University of Osaka Hospital 74

Impact of assisted reproductive technologies on reproductive outcomes in women with myotonic dystrophy: a retrospective study [Patricia Garay-Albizuri](#)^{1, 2, 3} [1](#)Department of Neurology, Donostialdea Integrated Health Organisation, Osakidetza, San Sebastian, Spain, [2](#)Neuromuscular Diseases Group, Neurosciences Area, Biogipuzkoa Health Research Institute, San Sebastian, Spain, [3](#)CIBERNED, CIBER, Spanish Ministry of Science & Innovation, Carlos III Health Institute, Madrid, Spain 183

12:30 - 13:30

Lunch

Totem Nord-Réserve Centre

13:30 - 14:45

Session 5: Preclinical and Clinical Drug Development

La Montagnaise

Multiscale imaging uncovers xenogeneic regenerative capacity of human pericytes as a cell therapeutic vehicle [Renée H.L. Raaijmakers](#)^{1, 2} [1](#)Radboud university medical center, Radboud Institute for Medical Innovation and Donders Institute for Brain Cognition and Behaviour, Department of Human Genetics, 6500 HB, Nijmegen, The Netherlands, [2](#)Radboud university medical center, Radboud Institute for Medical Innovation, Department of Medical BioSciences, 6500 HB, Nijmegen, The Netherlands 42

Scaling therapeutic discovery in DM1: A validated high-throughput platform combining in vitro screening and machine learning [Virginia Arechavala-Gomez](#)^{1, 2} [1](#)Nucleic Acid Therapeutics for Rare Diseases (NAT-RD), Biobizkaia Health Research Institute, Barakaldo, Spain, [2](#)Ikerbasque, Basque Foundation for Science, Bilbao, Spain 82

Tricyclo-DNA antisense oligonucleotide compounds to tackle toxic CUGexp-RNA in a mouse model of Myotonic Dystrophy type 1 [Julie FAGIOLI](#)¹ [1](#)Sorbonne Université, Inserm, Institut de Myologie, Centre de Recherche en Myologie, Paris, France 83

Sensor-Regulated Decoy Gene Therapy for Myotonic Dystrophy Type 1 [Ludovic Arandel](#)¹ [1](#)Sorbonne Université, Inserm, Institut de Myologie, Centre de Recherche en Myologie, Paris, France 135

Interventionally Contracting Somatic CTG Repeat Expansions as a Disease-Modifying Strategy for Myotonic Dystrophy Type 1 [Shugian Tang](#)^{1, 2} [1](#)Program of Genetics & Genome Biology, The Hospital for Sick Children., [2](#)Department of Molecular Genetics, University of Toronto. 193

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Remembering the Journey: A DM1 Movies and Stories Session

Tipi-tente-wigwam-Réserve Sud/Nord

17:30 - 23:59

Award and Gala Night: Dine, Dance & Celebrate!

La Montagnaise

Saturday, 30 May 2026

08:30 - 18:30

Family Day

La Montagnaise
