DETAILED CONFERENCE PROGRAM

Thursday September 11, 2025

3:00-5:00 **Conference Check-In and Poster Board Set Up** Ballroom Foyer and Drawing Room

5:00 **Opening Remarks** Ballroom

Robin J. Parks, PhD
Senior Scientist, Ottawa Hospital Research Institute, CA
Professor, University of Ottawa, CA

Jodi Warman-Chardon, MD/PhD
Associate Professor, University of Ottawa, CA
Neurologist, The Ottawa Hospital/CHEO, CA
Associate Scientist, Ottawa Hospital Research Institute, CA
Scientist, Children's Hospital of Eastern Ontario Research Institute, CA

5:10-6:30 **Keynote Session** Ballroom

Remarks from Session Moderators:
Mireille Khacho, PhD and **Hanns Lochmüller, MD/PhD**
Co-Directors, The Éric Poulin Centre for Neuromuscular Disease, CA

Remarks from: **Homira Osman, PhD,** Vice President, Research and Public Policy, Muscular Dystrophy Canada, CA

Remarks from: **Colleen Doyle,** Director of Canadian Research, ALS Society of Canada, CA

Keynote Speaker Introduction by:
Rashmi Kothary, PhD, Senior Scientist, Ottawa Hospital Research Institute, CA

Keynote Address:
Charlotte Sumner, MD
Professor of Neurology and Neuroscience
Johns Hopkins University School of Medicine, USA

6:30-8:00 **Welcome Reception** Adam Room

DETAILED CONFERENCE PROGRAM

Friday September 12, 2025

7:15-8:00 **Breakfast / Poster Viewing** Drawing Room

8:00-10:00 Ballroom

Session 1 Joint Session: Transformative Technologies in Neuromuscular Disorders

*Moderators**: Bernard Jasmin, PhD, University of Ottawa, CA***

Michael Benatar, MD/PhD Promise & Perils in ALS Therapy Development

Conrad (Chris) Weihl, MD/PhD Comprehensive Variant Analysis: Using Deep Mutational Scans for Clinical Care

Jordi Diaz-Manera, MD/PhD Artificial intelligence in neuromuscular disease diagnosis and care

Elizabeth McNally, MD/PhD Cell - Matrix Interactions in Muscle Degeneration and Regeneration

10:00-10:30 **Coffee Break / Poster Viewing** Drawing Room

10:30-12:00 Laurier Room

Session 2A Clinical: Motor Neuron Disease Pathogenesis and Treatment

*Moderator: **Matti Allen, MD/PhD, The Ottawa Hospital, CA***

Maryam Oskoui, MD Newborn Screening and emerging treatments in Spinal Muscular Atrophy

Agessandro Abrahao, MD Updates on clinical management and trials in ALS

Gordon Jewett, MD Consumer-grade devices for ALS clinical outcome assessment

Panel Discussion

10:30-12:00 Ballroom

Session 2B Basic: Advances in Muscle Stem Cells and Development

*Moderator: **Mary-Ellen Harper, PhD, University of Ottawa, CA***

Nicolas Dumont, PhD Regulation of muscle stem cell myogenic progression by bioactive lipids

Emanuela Gussoni, PhD Modulation of dystrophic severity through a tetraspanin

Fabio Rossi, PhD Fibroblast regulation across species

Short Talk: Rika Maruyama Systemic CRISPR/SaCas9-mediated exon skipping achieves long-term benefit in canine Duchenne muscular dystrophy

12:00-1:30 **Lunch Break / Poster Viewing** Drawing Room

1:00-1:30 **Lunchtime 3 Minute Flash Talks** Ballroom

*Moderator: **Keir Menzies, PhD, University of Ottawa, CA***

1. **Malaichamy Sivasankar** Systematic Reanalysis of NGS Data in 103 NMD Families Enhances Diagnostic Yield, Reveals Deep Intronic Variants, and Identifies ATP2A2 as a Novel Neuromuscular Disease Gene
2. **Ryan Marks** Single AAV vector base editing rescues a nonsense variant in a humanized mouse model of Duchenne muscular dystrophy
3. **Wanda Dupèbe** Skeletal Muscle Transcriptomic Comparison Between Men and Women Individuals with Myotonic Dystrophy Type 1
4. **Pablo Iruzubieta** Presence of symptoms and signs of neuropathy in a cohort of patients with SCA27B

DETAILED CONFERENCE PROGRAM

Friday September 12, 2025 continued

1:30-3:00 Laurier Room

Session 3A Clinical: Muscular Dystrophies – Treatment and Pathogenesis

Moderator: ***Hanns Lochmüller, MD/PhD, The Ottawa Hospital, CA***

Cam-Tu Nguyen, MD Clinical Updates in Dystrophinopathies

Duygu Selcen, MD Updates in Myofibrillar myopathies

Carsten Bönnemann, MD Novel disease identification in Muscle Disease

Panel Discussion

1:30-3:00 Ballroom

Session 3B Basic: Challenges and Opportunities in Amyotrophic Lateral Sclerosis

Moderator: ***Michael De Lisio, PhD, The Ottawa Hospital, CA***

Maxime Rousseaux, PhD TDP-43 SUMOylation safeguards from neuronal demise

Aaron Gitler, PhD An emergent disease-associated motor neuron state precedes cell death in a mouse model of ALS

Clotilde Lagier-Tourenne, PhD Disruption of RNA metabolism in ALS/FTD and emerging therapeutic strategies

Short Talk: Tyler Soule The oral microbiome in ALS shows differentially abundant organisms in limb versus bulbar onset disease: a binational study

3:00-4:00 **Poster Session (Odd Numbers) - Coffee/Snacks** Drawing Room

4:00-5:30 Laurier Room

Session 4A Clinical: Myasthenia Gravis Neuromuscular Junction Disorders

Moderator: ***Benjamin Beland, MD, The Ottawa Hospital, CA***

Neelam Goyal, MD Diagnostics in Myasthenia and Guidelines

Pushpa Narayanaswami, MD Myasthenia Gravis - Clinical Trials and Therapies

Hanns Lochmüller, MD/PhD Inherited neuromuscular junction disorders with genetic overlaps

Panel Discussion

4:00-5:30 Ballroom

Session 4B Basic: Muscle Disease Pathogenesis and Treatment I

Moderator: ***Mireille Khacho, PhD, University of Ottawa, CA***

Maurice Swanson, PhD RNA pathomechanisms in neuromuscular disease

Laurent Schaeffer, PhD Combinatorial therapy to improve SMA treatment

Michael Rudnicki, PhD Developing innovative regenerative therapies for neuromuscular disease

Short Talk: Amy Lam Single-AAV-mediated adenine base editing efficiently restores dystrophin expression and muscle function in mdx 4cv mice

5:30-7:00 **Informal Poster Viewing and Cocktail / Networking Reception** Drawing Room

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Saturday September 13, 2025

7:15-8:00 **Breakfast / Poster Viewing** Drawing Room

8:00-9:30 Laurier Room

Session 5A Clinical: Imaging and Genomics in Neuromuscular Disease

Moderator: ***Hugh McMillan, PhD, Children's Hospital of Eastern Ontario, CA***

Marcos Sampaio, MD and Gerd Melkus, PhD Muscle MRI Key Techniques and Clinical Implementation **Giorgio Tasca, MD/PhD** Imaging and natural history studies in FSHD

John Vissing, MD/PhD Quantitative MRI in RYR1 and ANO5

Panel Discussion

8:00-9:30 Ballroom

Session 5B Basic: Translational Research in Spinal Muscular Atrophy

Moderator: ***Rashmi Kothary, PhD, Ottawa Hospital Research Institute, CA***

Umrao Monani, PhD A chaperone modifier of the spinal muscular atrophy disease phenotype

Heidi Fuller, PhD Biomolecular insights into the potential for a precision medicine approach for spinal muscular atrophy

Ewout Groen, PhD Moving translational SMA research forward in the therapeutic era

Short Talk: Megan Baird Identification of early symptomatic gene expression changes in motor neurons in a large animal model of motor neuron disease

9:30-10:30 **Poster Session (Even Numbers) - Coffee/Snacks** Drawing Room

10:30-12:00 Laurier Room

Session 6A Clinical: Advances in Autoimmune and Genetic Myopathies

Moderator: **Jodi Warman-Chardon, MD/PhD**

Elie Naddaf, MD Advances in Inclusion Body Myositis

Erin O'Ferrall, MD Challenging Cases: How useful is muscle biopsy in clinical myopathies and muscular dystrophies

Jodi Warman-Chardon MD/PhD Genetic Testing in Limb Girdle Muscular Dystrophy

Panel Discussion

10:30-12:00 Ballroom

Session 6B Basic: Muscle Disease Pathogenesis and Treatment II

Moderator: ***Aymeric Ravel-Chapuis, PhD, University of Ottawa, CA***

Marco Sandri, PhD Dissecting the gene networks that control muscle wasting

Mireille Khacho, PhD Decoding Mitochondrial Signals Driving Muscle Stem Cell Function and Dysfunction

Denis Guttridge, PhD Local inflammation in the muscle microenvironment in cancer cachexia

Short Talk: Emma Sutton Maternal transfer of disease modifying therapies for spinal muscular atrophy: a novel, minimally invasive approach to deliver SMN therapy prenatally

12:00-1:30 **Lunch Break / Poster Viewing** Drawing Room

DETAILED CONFERENCE PROGRAM

Saturday September 13, 2025 continued

1:00-1:30 **Lunchtime 3 Minute Flash Talks** Ballroom

Moderator: ***Yan Burelle, PhD., University of Ottawa, CA***

1. **Shatha Atieh** Characterization of Bitter Melon Natural Compounds that Activate AMPK Signaling As Novel Therapeutics For Myotonic Dystrophy Type 1 (DM1)
2. **James Butcher** Dystrophic muscle phenotypes can be horizontally transferred via fecal microbiome transplantations
3. **Kelly Ho** ClC-1 chloride channel inhibitor NMD712 improves motor function in mouse models of congenital myasthenic syndromes
4. **Utkarsh Dang** A comprehensive and queryable database with thousands of findings on biomarkers from >20 serum and tissue datasets for DMD

1:30-3:00 Laurier Room

Session 7A Clinical: Advances in Clinical Autoimmune and Genetic Neuropathies

Moderator: ***Alberto Aleman, MD, The Ottawa Hospital, CA***

Christopher Klein, MD Paraprotein Associated Peripheral Neuropathies: Evolving Concepts

Jasper Morrow MD/PhD Diagnosis and Monitoring in Charcot-Marie-Tooth disease

Mario Saporta, MD/PhD Treatment Advances for Charcot-Marie-Tooth disease

Panel Discussion

1:30-3:00 Ballroom

Session 7B Basic: Neuromuscular Disease and Therapy

Moderator: ***Nadine Wiper-Bergeron, PhD, University of Ottawa, CA***

Christian Lorson, PhD Insight into IGHMBP2 function: leveraging novel animal models of SMARD1 and CMT2S

Richard Robitaille, PhD Glia regulation of neuromuscular junction in health and disease

Dwi Kemaladewi, PhD Therapeutic genetics and disease modeling in LAMA-deficient congenital muscular dystrophy

Short Talk: Aymeric Ravel-Chapuis Dysregulation of the RNA-binding protein HuR impairs neuromuscular junction integrity in muscular dystrophies

3:00-3:30 **Coffee Break / Poster Viewing** Drawing Room

3:30-5:00 Ballroom

Session 8 Joint Session: Future Directions in Neuromuscular Research

Moderators: ***Robin Parks, PhD and Jodi Warman-Chardon, MD/PhD***

Lyndsay Murray, PhD Axon degeneration and regeneration in mouse models of SMA

Kym Boycott, MD/PhD Impact and challenges of emerging diagnostic genetic testing techniques in NMD

Hanns Lochmüller, MD/PhD Phase 1b Study of the Safety, Tolerability, Pharmacokinetics, Immunogenicity, and Efficacy of ARGX-119 in Participants With DOK7 Congenital Myasthenic Syndromes

5:00 **Closing Remarks** Ballroom

Robin Parks, PhD and **Jodi Warman-Chardon, MD/PhD**