ABSTRACT REFERENCE GUIDE & POSTER NUMBERS

1. Observational study in adults with spinal muscular atrophy treated with Risdiplam. **Rodrigue X,** Henley K.

2. A Five-Year Update on a Real-World Study of Canadian Adults with Spinal Muscular Atrophy Type 2 and 3 Treated with Nusinersen. **Rodrigue X**, Charron C.

3. Positive predictive value of myositis antibody line blot testing in patients with suspected idiopathic inflammatory myopathy. **Chang Y**, Yang L, Budhram A.

4. NADMED: Targeted REDOX profiling for clinical and research use. **Äänismaa R**, Jansson S, Buzkova J, Euro L, Suomalainen A.

5. Rewiring myogenic cell identity: Comparative analysis of PAX7 and PAX7-FOXO1 in Rhabdomyosarcoma and muscle progenitors. **Ghavidel P** and Sincennes M-C.

6. Thoracic Electric Impedance Tomography Detects Lung Volume Changes in Amyotrophic Lateral Sclerosis. Hansen G, Shaw A, Bolt K, Verity R, Nataraj RT, **Schellenberg KL.**

7. Best practice recommendations for the clinical care of spinal bulbar muscular atrophy. **Schellenberg KL**, Caspar-Bell G, Ellis C, Johnston W, King A, King M, Korngut L, Kushneriuk B, Lavoie AJ, McGonigle R, Newton J, O'Connell C, Shoesmith C, Suchowersky O, Warman-Chardon J, Wunder S, Pfeffer G.

8. Longitudinal analysis of glymphatic function in amyotrophic lateral sclerosis and primary lateral sclerosis. Sharkey RJ, Cortese F, Goodyear BG, Korngut LW, Jacob SM, Sharkey KA, Kalra S, Nguyen MD, Frayne R, Pfeffer G. **Presented by: Soule, T.**

9. Higher than expected incident cases of spinal bulbar muscular atrophy in western Canada. Lamont R, King M, King A, Schellenberg K, Pfeffer G. **Presented by: Soule, T.**

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14. Novel prodrug improves locomotor disfunction in gne deficient zebrafish. **Lau J**, Derkson A, O'Connor E, Morewood J, Spendiff S, Pertusati F, Lochmüller H.

15. Galactose treatment rescues neuromuscular junction transmission in glutamine-fructose-6-phosphate transaminase 1 (Gfpt1) deficient mice. **Holland SH**, Carmona-Martinez R, O'Neil D, Ho K, O'Connor K, Azuma Y, Roos A, Spendiff S, Lochmüller H.

16. Unraveling the Function of MLIP in Myogenic Cells: Toward Understanding Its Role in Rare Myopathy. **Bachir AY,** Sleno L, Tétreault M, Sincennes M-C.

17. Correcting mutations responsible for LGMD R2/2B (dysferlinopathy) with Prime editing. **Bouchard C,** Jiang J, Rousseau J, Tremblay JP.

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20. Maternal transfer of disease modifying therapies for spinal muscular atrophy: a novel, minimally invasive approach to deliver SMN therapy prenatally. **Sutton ER**, Beauvais A, De Repentigny Y, Yaworski R, McMillan H, Schneider B, Mattar P, Kothary R.

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25. 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. **Malaichamy S**, Polavarapu K, Thompson R, Idoux R, Spendiff S, Karcagi V, Herczegfalvi A, Almomen M, Vengalil S, Nashi S, Laurie S, Matalonga L, Beltran S, Voermans N, Roos A, Barić I, Nalini A, Lochmüller H.

26. The Impact of Premature Cellular Senescence in Muscle Regeneration in Myotonic Dystrophy type 1. **Kim T**, Koike T, Conte T, Molina T, Mokhtari I, Duchesne E, Dumont N.

27. Impact of Trained Immunity on Macrophage and Muscle Stem Cell Function in Dystrophic (mdx) Mice. **Deprez A**, Bhattarai S, Liang F, Gusev E, Petrof BJ.

28. Inhibition of translation initiation factor eIF4A in dystrophic muscle stem cells drives cell fate towards differentiation. **Filippelli RL**, Robertson R, Liu Y, Granet JA, Cusmano AA, Gallouzi IE, Pelletier J, Chang NC.

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30. Understanding myotonic dystrophy type 1 using iPSC-derived neurons. **Geertsma H**, Part C, O'Connor K, Baird S, MacKenzie A, Spendiff S, Lochmuller H.

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34. DG9-PMO Enhances Cardiac and Skeletal Muscle Delivery for Exon 44 Skipping Therapy in Duchenne Muscular Dystrophy. **Shah MNA**, Wilton-Clark H, Haque F, Powell B, Sutanto LE, Maradiya R, Zhabyeyev P, Roshmi RR, Anwar S, Aslesh T, Lim KRQ, Maruyama R, Bigot A, Young CS, Bittner S, Spencer MJ, Moulton HM, Oudit GY, Yokota T.

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36. A novel therapeutic effect of lauric acid: preventing loss of ambulation in severe mouse models of muscular dystrophy. **Sun Z**, White Z, Kang Y, Lyu Y, Bernatchez P.

37. Chronic loss of the mitochondrial fusion protein OPA1 is detrimental to muscle stem cell maintenance and activity. **Sommers O**, Triolo M, Wade S, Dort J, Khacho M.

38. Transition to home infusions with a low risk of infusion-associated reactions and functional stability: real-world outcomes from the UK Early Access to Medicines Scheme registry for cipaglucosidase alfa plus miglustat in late-onset Pompe disease. Roberts M, **McPherson A**, Jain V, Moffat E, Clarke S, Sitaraman Das S, Zhang J.

39. Senotherapeutics and Exercise-Based Strategies to Restore Defective Muscle Stem Cells in Myotonic Dystrophy Type 1. **Koike T**, Kim T, Mokhtari I, Conte T, Duchesne E, Dumont N.

40. The Impact of DEPP1 Overexpression on Skeletal Muscle Function. **Delisle P**, Hevey A, Capobianco A, Pouliot C, Leduc-Gaudet JP.

41. Identification and Characterization of Serglycin as a Potential Biomarker in GNE Myopathy. **Derksen A**, Spendiff S, Pogoryelova O, Hentschel A, Dobelmann V, Roos A, Lochmüller H.

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52. Proximal Femoral Screw Hemiepiphysiodesis in Spinal Muscular Atrophy: A Case Report in the Gene Therapy Era. **Nauleau-Javaudin A**, Rioux-Trottier E, Fiscaletti M, Martineau-Beaulieu E, Pilon-Cadieux C, Nguyen C-TE.

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55. Investigating a potential muscle-driven mechanism of neuromuscular denervation in ALS. **Boyer M**, Berthod F, Pozzi S.

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73. Autophagy maintains the regenerative capacity of muscle stem cells. Cusmano AA, Granet JA, VanGenderen CA, Robertson R, **Chang NC.**

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86. Limb-Girdle Muscular Dystrophies in a Single-Center Adult Population: Clinical Spectrum and Genetic Findings. **Aldecoa M**, Iruzubieta P, Brais B, O'Ferrall E.

87. Telemedicine and remote monitoring in neuromuscular disease care: Effectiveness and challenges in follow-up care and multidisciplinary coordination. **Gabr M.**

88. Train(e)d: Training Platform for Building National Clinical Outcome Assessment Capacity in Neuromuscular Disease. **Masnata M**, Adamji Z, Osman H, Beattie E, O'Connell C, Gagnon C, Warman-Chardon J.

89. Experience as evidence: mapping the patient journey of Friedreich ataxia in Canada. **Adamji Z**, Osman H, Lintern S.

90. Characterizing spinal cord pathology in the cuprizone autoimmune encephalomyelitis model: evaluating its potential as a comprehensive model for multiple sclerosis research. **Al-Aarg M**, Alves de Almeida MM, De Repentigny Y, Kothary R.

91. A dosage study for aav9-mediated gene therapy and the development of combinatorial therapy for the treatment of spinal muscular atrophy. **Yaworski R**, Beauvais A, Sutton E, Lorson C, Schneider B, Kothary R.

92. ClC-1 chloride channel inhibitor NMD712 improves motor function in mouse models of congenital myasthenic syndromes **Ho K**, Carmona-Martinez R, Skov M, Spendiff S, & Lochmüller H.

93. Regulation of mitophagy by the NIX pathway during MuSC state transition. **Dixon M**, Racine J, Larionov N, Cairns G, Khacho M, Burelle Y.

94. Overview of the Canadian Neuromuscular Disease Registry (CNDR): Applying Real-World Evidence to Improve Disease Research and Outcomes in Canada. **Langevin LM**, Crone M, Hodgkinson V, Jewett G, Lochmüller H, McMillan H, Korngut L, on behalf of the CNDR Investigator Network.

95. Development of an in-silico workflow to characterize the spliceogenicity and protein effect of splice-altering variants. **Zeldin J**, Polavarapu K, Thompson R, Lochmüller H.

96. The Glucocorticoid Receptor Regulates Nuclear Positioning During Skeletal Muscle Differentiation. **Alharbi S**, Wiper-Bergeron N.

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99. What are the Costs of Living with FacioScapuloHumeral Dystrophy? The Socioeconomic Burden of FSHD. **Lessard LER**, Grant A, Smith IC, Osman H, Lochmüller Ha, McMillan H, Pfeffer G, Korngut L, Gagnon C, Lintern S, Selby K, Ari B, Thavorn K, Warman-Chardon J.

100. Dystrophic satellite cells undergo mitotic catastrophe and senescence. **Pan W**, Rudnicki M.

101. C9orf72 loss of function leads to cerebellar degeneration in the zebrafish ALS model. **Singh J**, Lescouzères L, Patten K.

102. Characterizing the role of the RNA-binding protein HuR in regulating the structure of neuromuscular junctions in Duchenne muscular dystrophy. **Bhaura G**, Gobin J, Ravel-Chapuis A, Jasmin B.

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104. Identifying cancer cachexia humoral factors affecting muscle stem cell regeneration capacity. **LoFaro M**, Hall D, Rudnicki M.

105. The comparative efficacy and safety of risdiplam versus high-dose nusinersen in children with Type 1 SMA. Kokaliaris C, Hawkins N, Mahajan A, Scott DA, Sideris CE. Non-author local presenter: **Inskip JA.**

106. Deciphering the mechanisms involved in the AMPK-induced therapeutic benefits in Myotonic Dystrophy type 1 skeletal muscle. **Fahmi C**, Lessard L, Mounier R, Ravel-Chapuis A, Jasmin BJ.

107. Characterization of Bitter Melon Natural Compounds that Activate AMPK Signaling As Novel Therapeutics For Myotonic Dystrophy Type 1 (DM1). **Atieh S**, Liu J, Ye Y, Tang C, Ravel-Chapuis A, Jasmin BJ.

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110. Longitudinally tracking disease progression and correlation in oculopharyngeal muscular dystrophy patients using whole-body quantitative MRI. **Satarkar T,** Smith IC, Khan S, Esttaifo F, Meier-Ross K, Melkus G, Sampaio M, Warman-Chardon J.

111. Polarity dysregulation in dystrophin-deficient satellite cells. **Catenacci M**, Rudnicki MA.

112. A muscular dystrophy associated with bi-allelic LEMD2 variants: Expanding the genotype of nuclear envelopathies. Pauper M, Kölbel H, Karakesisoglou I, Schänzer A, Böhm J, Thompson R, Kohlschmidt N, Ringel B, Bonne G, Neuhoff K, Gangfuß A, Aksel Kilicarslan O, Beltran Agullo S, Hentschel A, Schara-Schmidt U, Lochmüller H, Polavarapu K, **Roos A**.

113. Factors that influence adherence to home-based rehabilitation in children and youth with neuromuscular disorders: a scoping review. **Grangé A**, Boulais J, Levac D.

114. Diagnostic Delay Leading to Inpatient ALS Diagnoses: A Case Series. Donaghy R, Seth A, Zhang l, Harris G, Sufit R, Kalb R, **Ajroud-Driss S**.

115. Mitochondrial redox signaling regulates muscle stem cell function through glutathione-s-transferase pi1. **Triolo M**, Faraji D, Wade S, Larionov N, Khacho M.

116. Dystrophic muscle phenotypes can be horizontally transferred via fecal microbiome transplantations. **Butcher J**, Gosse JT, Ravel-Chapuis A, Jasmin BJ, Stintzi A.

117. Retrospective assessment of feeding and nutrition after 2 years of risdiplam treatment in younger children with SMA using the CEDAS. Gorni K, Baranello G, Conway E, Li Y, **Kalache N**.

118. Drug repurposing of olodaterol and forskolin for congenital myasthenic syndromes. **Choueiri C**, Ho K, Spendiff S, Meyer S, Grey M, Lochmüller H.

119. AAV-mediated gene editing strategies to treat Collagen VI-related Myopathies. **Lin KH,** Resner E**,** Maier M, Wagers A.

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121. Characterization of FDA-approved drugs that target the RNA-binding protein Staufen1 and mitigate the atrophy of skeletal muscle fibers following denervation. **Charron M**, Patel D, Almasi S, Chen H, Ravel-Chapuis A, Jasmin B.

122. The involvement of PARylation in muscle satellite cell fate and regeneration in a mouse model of muscular dystrophy. **Della Giustina A**, Saraci K, Menzies KJ**.**

123. Parylation peril: how poly-ADP-ribosylation affects satellite cell fate and regeneration. **Saraci K**, Green A, Niyonambaza S, Giustina AD, Menzies KJ.

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