Poster Abstracts from NMD4C at Ottawa NMD 2023

A full list of all abstracts is available on the Ottawa NMD 2023 website here.

Compliance to Duchenne Muscular Dystrophy Care Considerations in Canada

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Background: Duchenne muscular dystrophy (DMD) is a neuromuscular disease resulting in progressive muscle degeneration. Without disease-modifying therapies and a cure, the implementation of multidisciplinary care has significantly impacted the progression of DMD. This study examined existing care practices for patients in Canada, evaluated the extent to which these practices adhere to international care guidelines, and sought to pinpoint aspects of care where current standards are not met.

Methods: A 175-question cross-sectional survey was sent to clinical professionals actively involved in the care of DMD patients in Canada. The survey focused on clinicians' familiarity with the guidelines and their experiences with evaluations, techniques, and interventions across ten domains of management: neuromuscular, respiratory, cardiac, endocrinology/corticosteroid usage, gastrointestinal/nutritional, bone health/osteoporosis, orthopedic/surgical, rehabilitation, psychosocial support, and transitions.

Results: A total of 94 expert clinicians completed the survey. 71% were highly familiar with the care considerations. Suboptimal adherence to care guidelines was noted in endocrine, gastrointestinal/nutritional, and psychosocial management for ambulatory-pediatric patients. Psychosocial as well as gastrointestinal/nutritional management remained problematic for non-ambulatory patients. Whereas, all domains, except for cardiac care and endocrinology, were sub-optimal for adult patients. This aligns with the gaps in the process of transitioning from pediatric to adult healthcare.

Conclusion: While specialized and multi-disciplinary care has positively contributed to outcomes for Canadians with DMD, disease experts felt that enhancing care coordination and refining the approach in certain areas can lead to better outcomes and support patients to live fulfilling lives well into adulthood.

Expert Patient Capacity Building in Neuromuscular Disease Research

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Background: The Strategy for Patient-Oriented Research (POR) emphasizes incorporating patient-partners on research teams to reorient and reprioritize research questions to match those of patients and to bring more effective and meaningful improvements in health outcomes and systems. This approach has gained traction in recent years and is promoted by scientific funding organizations. However, many patients and researchers struggle with how to operationalize research partnerships, both realistically and effectively. Training and resources on POR exist, but none are specific to theoretical and practical considerations related to neuromuscular disease (NMD), and lack content in inclusion, diversity, equity, and accessibility (IDEA). This project examined existing training resources, evaluated the unique needs of patient-partners and sought to develop e-learning modules from a NMD perspective.

Methods: A literature search and landscaping of existing POR resources was conducted. Following formal assessment of resources, through participatory methods, patient-partners were involved in developing competencies, learning objectives, original content and activities on readiness, IDEA and relationship building. A backwards educational design process was employed.

Results: Available high-quality Canadian and international-POR resources and best practices were identified and published in a repository/resource catalogue, which has received 256 views-to-date. Three easy-to-use and accessible NMD e-training modules were developed in English and French in spring 2023, with 40 patient-partners and researchers successfully completing the training to date.

Conclusion: Incorporating patient engagement in the research process has been embraced, but adoption has been low. The 'importND' resources now equip NMD researchers and patients in Canada to meaningfully collaborate in the research process.

The Neuromuscular Disease Network for Canada (NMD4C): An interdisciplinary national collaboration to tackle unmet needs for NMD research and care

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In 2020, the CIHR and MDC-funded NMD4C was launched with a mission to improve the care, research, and treatment of neuromuscular diseases (NMDs) for all Canadians. NMD4C has united >530 Canadian NMD stakeholders to share best practices, resources and expertise, build clinical and research capacity, and facilitate access to novel therapies.

To build capacity for researchers and physicians, NMD4C has held monthly continuing professional development (CPD)-accredited webinars, acquired Royal College approval of neuromuscular medicine as an Area of Focused Competency, launched a 41-week CPD-accredited online neuromuscular fellowship curriculum , and invited outstanding early-career (EC) researchers and physicians to present/chair webinars and integrate into working groups, published >40 EC blogs and profiles, and established annual EC awards and postdoctoral and clinical fellowship awards. We co-created online NMD patient-oriented research training modules. We have published a Canadian guidance on gene replacement therapy in spinal muscular atrophy, provided guidance on NMD respiratory care and vaccination during the COVID pandemic, developed infographics to disseminate Canadian clinical practice guidelines, identified gaps in multidisciplinary Duchenne muscular dystrophy standards of care in Canada, and submitted clinician group input on CADTH's draft reimbursement recommendations and treatments under review. To strengthen research resources and infrastructure, we developed a virtual NMD biobank catalogue, launched two new registry disease datasets, and established dedicated clinical trial support for bringing more NMD clinical trials to Canada.

With renewed funding until 2028, we will expand our collaborative community of NMD stakeholders to address emerging challenges for advancing care and research in Canada.