
Thank You FOR BREAKING DOWN BARRIERS

MUSCULAR DYSTROPHY CANADA
DYSTROPHIE MUSCULAIRE CANADA

# Shad's R&R Impact Summary


The 48th edition of Shad's R&R Golf Tournament was an incredible success. Thanks to your amazing dedication, since 1975, you have raised nearly $5.5 million in support of cutting-edge neuromuscular research.

Your support of neuromuscular research will save lives by fueling future discoveries, and ensuring that Canadians living with neuromuscular disorders have access to new life-changing treatments, while also filling the immediate need for advanced treatment and care options.

As a leader in funding ground-breaking research for all neuromuscular disorders, we at Muscular Dystrophy Canada (MDC) know the importance of providing our clients with the most-to-date information they need, while also making advancements to provide more treatments, therapies, and potential cures. Thanks to your dedicated support, you are making sure that people living with a neuromuscular disorder can keep finding hope - in the present and future.

In this report, we want to introduce some of the researchers you helped fund. We hope you find their work inspiring as we continue to break down barriers together.

Thank you for helping thousands of Canadians right now, and for giving hope to future generations.

Sincerely,

**Stacey Lintern**
*CEO*

**David Crawford**
*Chair, Board of Directors*

# Investing in high-impact research

Thanks to your support, MDC can invest in life-changing research. In March, we funded **nine new clinical and translational science research projects** through our research grant competition, a dedicated Canadian source of funding for neuromuscular research. We are honoured to fund these exceptional and bright researchers, clinicians and academics who will lead these projects taking place across Canada.

**Dr Eric Voorn**
*University of Amsterdam*

B-FIT! Taking exercise away from the hospital into the home environment for people with Oculopharyngeal muscular dystrophy

**Dr Tyler Churchward-Venne**
*McGill University*

Dietary protein requirements in adults with muscular dystrophy

**Dr Reshma Amin**
*The Hospital for Sick Children*

Establishing the top ten research priorities for respiratory care of childhood neuromuscular disorders

**Dr Simon Girard**
*Université du Québec à Chicoutimi*

How genealogies (family histories) can help us understand myotonic dystrophy type 1

**Dr Martine Tétreault**
*University of Montreal*

Evaluating the impact of stop variants on MLIP's (Muscular LMNA-Interacting Protein) function in muscle

**Dr James Dowling**
*The Hospital for Sick Children*

Therapy development for centronuclear myopathy caused by DNM2 gene changes

**Dr Pascal Chartrand**
*University of Montreal*

Pre-clinical testing of novel pharmacological inhibitors of toxic DMPK mRNA in models of myotonic dystrophy type 1

**Dr Hernan Gonorazky**
*The Hospital for Sick Children*

Muscle MRI in neuromuscular disorders: The era of artificial intelligence

**Dr Jill Zwicker**
*University of British Columbia*

Reimagining rehabilitation for SMA: Patient and family perspectives

# Investing in high-impact research: researcher spotlight

Dr Eric Voorn
Amsterdam UMC, University of Amsterdam
Recipient of a 2023 clinical science grant

## Taking exercise from the hospital into the home

Evidence shows exercise can have a positive effect on people with neuromuscular disorders (NMDs). However, most of the studies took place in hospitals or rehabilitation centres which are expensive to conduct and can be tiring for participants to attend due to travel. An exercise program in the home environment may be a solution.

Dr. Voorn's team developed such an exercise program, called B-FIT in the Netherlands. B-FIT supports health care professionals to tailor the program to the patient. It can also be used to move exercise away from the hospital setting, and into the home or community. In this project, 20 individuals with oculopharyngeal muscular dystrophy (OPMD) living in Quebec will follow the B-FIT exercise program. They will train in their own home and will be supervised by a trained physiotherapist. At the end of the program, patients and therapists will have their physical fitness evaluated and be asked if they were satisfied with the use of the training guide. It is expected that physical fitness will be improved after the exercise program. **This will help people with OPMD to maintain their independence and to improve their quality of life.**

Dr Reshma Amin
The Hospital for Sick Children
Recipient of a 2023 clinical science grant

## Establishing research priorities for respiratory care of childhood neuromuscular disorders

Children with neuromuscular disorders typically have trouble breathing. Some children use machines to help with this but the machines can be disruptive and hard to use. Most guidelines on the lung health of children with NMDs are based on what experts think, as there is not enough research in this field. And recently, new medicines for some types of neuromuscular disorders have been developed.

This study will ask patients, their families and clinicians about the questions they have around lung health for NMD affected children. The study involves three stages of surveys to determine the most important research questions, with the goal of reaching a consensus on the top ten. **MDC believes strongly in asking the neuromuscular community to share their expertise, guide discussions and inform decisions regarding health care, research, and therapy development. This project focuses on the community expressing their needs, to help prioritize future studies in this field.**

Dr Simon Girard
Université du Québec à Chicoutimi
Recipient of a 2023 translational science grant

## How family histories can help understand myotonic dystrophy type 1

The Saguenay-Lac-Saint-Jean region in Quebec has the highest incidence of Myotonic dystrophy type 1 (DM1) worldwide. This disorder is caused by the expansion of a repeated genetic sequence in the DMPK gene. However, the link between this gene and the severity of the disorder is unclear. Other genetic changes, such as modifier genes (which affect the expression of other (main) genes) have been proposed to better explain the disorder but few human studies have confirmed the presence of such genes in patients with DM1. Using statistical methods, Dr. Girard's study will look for modifier genes in DM1 patients from the Saguenay-Lac-Saint-Jean region.

This could be the first step in beginning therapies or treatments for patients with this disorder and, finding modifier genes associated with earlier DM1 onset could lead to early preventive therapies. **These new findings will have an impact on DM1 research, and for many other rare neuromuscular disorders.**

Dr Martine Tétreault
Centre Hospitalier de l'Université de Montréal (CHUM)
Recipient of a 2023 translational science grant

## How genetic changes cause muscular disorders

Rare neuromuscular disorders are caused by changes in the spelling of genes that are important for keeping muscles healthy. With the advance of technology, we are able to diagnosis patients by identifying differences in their genetic code. Genetic code differences in the gene Muscular LMNA-Interacting Protein (MLIP) is thought to be linked to muscle related disorders, although little is known yet about MLIP's function.

This project will study the effect of MLIP mutations in a laboratory. More specifically, genetic changes found in existing patient's genes will be reproduced to see how muscles are formed, maintained and affected by MLIP. **The study of this mutation will greatly improve the understanding of disorders as well as patient diagnosis.**

## Leveraging the best partnerships

Thanks to donors like you, MDC is also partnering with organizations like the Neuromuscular Disease Network for Canada (NMD4C) to leverage our research investments.

Recently, through our partnership with the NMD4C and the Canadian Society of Clinical Neurophysiologists, MDC funded **three clinical fellowships in neuromuscular medicine and electromyography, and three post-doctoral fellowships in neuromuscular research** ensuring top-ranked researchers receive funding, and early-career scientists can conduct studies focused on neuromuscular research.

*"This initiative helps train and educate the next generation of neuromuscular researchers and clinicians. The post-doctoral fellowships help strengthen neuromuscular research capacity, and the clinical fellowships will strengthen the specialist care available to the neuromuscular community,"* said Stacey Lintern, CEO, Muscular Dystrophy Canada. *"For our clients, that means we are investing in the sustainability of neuromuscular research, which brings us one step closer to finding cures, and in skilled clinicians that will provide clinical care to individuals affected by neuromuscular disorders and help raise standards of care."*

# Congratulations

## to the Neuromuscular Clinical Fellowship recipients!

**Dr Beatrice Soucy**
UNIVERSITY OF CALGARY

**Dr Gloria Mak**
UNIVERSITY OF ALBERTA

**Dr Neha Patel**
UNIVERSITY OF TORONTO

## to the Post-doctoral Fellowship recipients!

**Dr Yuguo Liu**
UNIVERSITÉ DE SHERBROOKE

**Dr Matthew Triolo**
UNIVERSITY OF OTTAWA

**Sean Ng, PhD candidate**
MCMASTER UNIVERSITY

NMD4C

CSCN SCNC Canadian Society of Clinical Neurophysiologists
Société canadienne de neurophysiologie clinique

# Leveraging the best partnerships: researcher spotlight

Dr Matthew Triolo
University of Ottawa
Recipient of a 2023 post-doctoral fellowship grant

## Understanding the biological causes of neuromuscular disorders

Dr. Matthew Triolo is one of the recipients of the three national post-doctoral fellowships in neuromuscular research. His work looks at Duchene Muscular Dystrophy (DMD), a neuromuscular disorder that has high rates of mortality and leads to a decline in mobility, and voluntary activity.

Normally, specialized muscle stem cells allow muscle to heal and repair itself. In patients with DMD, these cells do not work properly which leads to muscle breakdown. Recent studies have shown that mitochondria, which make most of the energy for the cell, are required for proper muscle stem cell function. Unfortunately, mitochondria in muscle stem cell do not act well in DMD. The goal of this study is to assess whether mitochondria can act as a therapeutic target to treat DMD. **Findings from this work will allow researchers to target muscle stem cell as an intervention for DMD and for other neuromuscular disorders.**

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Dr Neha Patel
University of Toronto
Recipient of a 2023 clinical fellowship grant

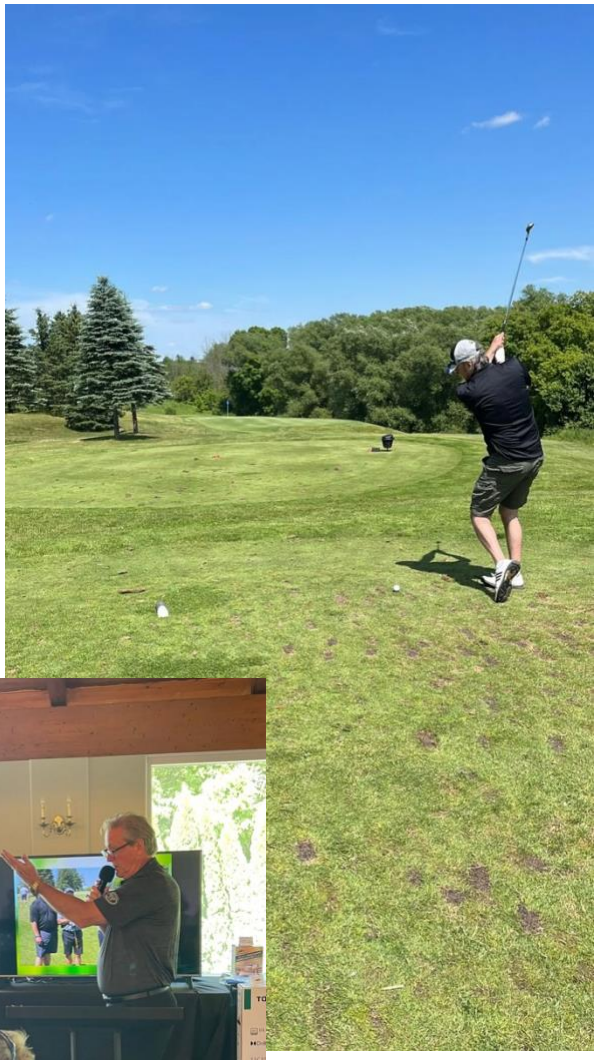
## Providing more neuromuscular specialists for our community

Finding a neuromuscular specialist is an important step in a person's healthcare journey, and we know there are few specialists across the country. That's why we are investing in top-ranked clinicians who will provide clinical care to individuals affected by neuromuscular disorders and help raise standards of care.

Dr. Neha Patel is one of the recipients of the clinical fellowship grant. Dr. Patel is in her final year of Neurology residency training at the University of Toronto and started her Neuromuscular Medicine Fellowship in July 2023 at Sunnybrook Health Sciences Centre. **Individuals with neuromuscular disorders who don't have a diagnosis are doubly disadvantaged. Not only do they not know what is happening to them, but they don't know how it could be potentially treated. By funding the next generation of neuromuscular clinicians, you and MDC are improving the specialist care available to the neuromuscular community so they can get the diagnosis and the treatment they need.**

In 1973, when you first came together to support MDC, the neuromuscular community did not have many options for treatments. With you, we are breaking down barriers and improving the lives of those affected by neuromuscular disorders and their families.

**On behalf of MDC and the neuromuscular community, thank you again.**



We are strongest when we *work together.*

# *Thank You!*

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