

## Insights Report

Understanding the Needs and Priorities of the  
Canadian Mitochondrial Community





# Community Insights Report

## Table of Contents

---

<b>EXECUTIVE SUMMARY</b>	<b>3</b>
Introduction	4
Survey and Interview Summary	6
Survey Composition	6
Geographic Distribution	6
Age Distribution	6
Family History	6
Diagnosis Journey and Timelines	6
Treatment and Symptom Management	7
Quality of Life	8
Sources of Education and Future Priorities	8
Recommendations	8
Conclusion	10
Acknowledgments	11
About MitoCanada	11
<b>GRAPHS AND TABLES</b>	<b>12</b>
Demographics	12
Diagnosis	12
Treatment Landscape	15
Impact on Quality of Life	19



# Understanding the Needs of the Canadian Mitochondrial Community

## Executive Summary

---

In 2021, MitoCanada conducted a community consultation to better understand and identify the needs of the mitochondrial community in Canada. Hosting 1:1 virtual, semi-structured interviews and creating a comprehensive survey, MitoCanada was able to collect and curate valuable community insights to share in our 2022 Canadian Mitochondrial Community Insights Report.

The insights from this consultation revealed much about community composition. From family history, the diagnostic and therapeutic landscape to mitochondrial management and provincial divides, the impacts these complex and chronic mitochondrial disorders have on individuals and families are vast.

The mitochondrial community's geographic distribution was predominately found in large urban centres across Canada. The reported age distribution of community members with a clinical mitochondrial diagnosis was between 6-12 and 45-54 years.

Time to diagnosis varied among age groups. Shorter time to diagnosis was reported among children, and time to diagnosis exceeding three years was reported largely among adults. Tests used to provide a mitochondrial diagnosis varied but typically included either genetic testing, muscle biopsy, biochemical testing or a combination of these methods.

Regarding treatment methods and symptom management, 91% of survey participants reported being prescribed the mito cocktail by a clinician with different combinations and doses of supplements. Differences in prescribed supplements are expected as usage varies based on diagnosed disorder and the unique needs of each mito patient.

Movement therapies such as occupational therapy, physiotherapy, massage therapy, and exercise were well reported as helpful management strategies. Muscle weakness and fatigue were the highest-rated physical symptoms that significantly impacted quality of life. Loss of independence, isolation and helplessness were the highest reported invisible impacts.

The final component of our consultation explored sources of mito education, types of support the community would like to see from MitoCanada and identified key priority areas. With these insights, MitoCanada has crafted recommendations and a step-wise process to aid in their implementation. MitoCanada is deeply committed to improving the physical, social, psychological, and financial outlook of individuals and families affected by mitochondrial disease in Canada.



# Understanding the Needs of the Canadian Mitochondrial Community

## Results, Observations and Recommendations

---

### INTRODUCTION

Since 2010, MitoCanada has worked tirelessly to help support, educate and protect Canadians living with or at risk of developing mitochondrial disease (mito). To reduce the devastating impact of mitochondrial disease and support patients, families and caregivers on their journey, MitoCanada has focused on objectives that drive meaningful change. Through our community consultation, we have respectfully listened and learned from those who have generously shared their lived experiences with mito.

In 2021, MitoCanada conducted a community consultation to better understand and identify the needs of the mitochondrial community in Canada. Hosting 1:1 virtual, semi-structured interviews and creating a comprehensive survey, MitoCanada was able to collect and curate valuable community insights.

We recruited fifteen participants to take part in our virtual interview sessions. Ten participants were adults with a clinical mitochondrial disease diagnosis, five participants were parents or caregivers caring for a child or adult with a clinical mitochondrial disease diagnosis. Interview participants spanned the country; provinces include British Columbia, Alberta, Manitoba, Saskatchewan, Ontario, and Quebec.

The survey was designed in collaboration with individuals, parents and caregivers living with mito, MitoCanada's Patient Family Advisory Council (PFAC) and researchers. Each group provided input and advisement on survey design and content.

The survey was piloted by twelve individuals, divided into two categories "adults living with mitochondrial disease" and "parents-caregivers caring for individuals living with mitochondrial disease."

The online survey was promoted, to recruit Canadian responders, via:

- MitoCanada's social media channels (Facebook and Instagram),
- MitoCanada's website
- MitoCanada's MitoNews eNewsletter
- Allied partner's social media channels
- Emails to physicians, patients and parent-caregivers
- Virtual symposiums
- Podcast

MitoCanada did not incentivize the survey. The survey was launched in January 2021 and closed in September 2021. To improve the statistical power of our analysis, we removed all incomplete and duplicate surveys, bringing our usable survey count to 81.

The objectives of our community consultation were to better understand:

- Demographics of Canada's mitochondrial community
- Diagnostic journey
- Mitochondrial disorder landscape
- Therapeutic landscape
- Clinical trial and research engagement interests
- Quality of life impacts

Through our consultation efforts, our goals were to:

- Identify key priority areas important to the mito community
- Identify community engagement opportunities
- Identify opportunities to co-create education programs and resources, advocacy initiatives and research studies with all mitochondrial disease stakeholders.



# SURVEY and INTERVIEW SUMMARY

## Survey Composition

The survey insights from this consultation revealed much about community composition. From family history, the diagnostic and therapeutic landscape, to mitochondrial management and provincial divides, the impacts these complex and chronic mitochondrial disorders have on individuals and families are vast.

60% of participants were adults living with a clinically diagnosed mitochondrial disorder, and 32% were children, as reported by their parent. 8% reported that had not yet received a diagnosis.

Using Statistics Canada's definition of urban and rural population centres, MitoCanada was able to categorize participants place of residence. "Population centres are classified into three groups, depending on the size of their population :"

- small population centres, with a population between 1,000 and 29,999
- medium population centres, with a population between 30,000 and 99,999
- large urban population centres, with a population of 100,000 or more.

## Geographic Distribution

61% of participants reside in large urban centres, 15% in medium urban centres, 18% in small urban centres and 6% of participants reside in rural areas. The survey did not establish if participants previously resided in smaller or rural centres before diagnosis and moved to larger urban centres to access care. This is a consideration to investigate further.

## Age Distribution

The majority of participants completing the survey fell into two main age categories: 6-12 years (16% of respondents) or 45-54 years (20%)

## Family History

53% of participants reported not having a family history of mitochondrial disease. 26% reported a family history. Of family members affected, females were predominately reported with MELAS being the most prevalent mitochondrial disorder.

## Diagnosis Journey and Timelines

92% of survey respondents reported receiving a clinical mitochondrial disease diagnosis. The highest reported diagnosed disorders were MELAS (22%), Leigh syndrome (21%) and CPEO (21%). 8% of respondents did not have a clinical diagnosis; 44% of respondents without a clinical diagnosis are being treated based on symptoms, 22% await genetic testing and 11% continue to search for a genetic mutation.

The time to diagnosis varied with two clear groupings. 45% reported that it took between one - three years to receive a mitochondrial disease diagnosis, and 32% reported that it took more than seven years.

For those reporting a one - three years diagnosis time frame, 64% were children, and 36% were adults. 20% of those children were diagnosed with Leigh syndrome. In the adult groups, 12% were diagnosed with CPEO and 8% with MELAS. For those reporting a diagnosis taking seven years or more, 94% were adults. Of those adults, 33% reported receiving a MELAS diagnosis, and 17% received a MERFF diagnosis.

Tests used to provide a diagnosis varied, using either genetic testing (52.6%), muscle biopsy (57.8%), biochemical testing (blood, urine) (61.4%) or a combination of these methods.

## Treatment and Symptom Management

In understanding the treatment landscape, participants were asked to identify the treatments and/or interventions used to help manage their mitochondrial disorder.

There is a high adherence rate amongst individuals prescribed the mito cocktail in this survey. 91% reported being prescribed the mito cocktail by a clinician, and 84% reported taking the mito cocktail as prescribed. Average adherence rates for chronic treatments are estimated to be around 50%. Typically, an adherence rate of 80% is needed for optimal therapeutic efficacy.

The combination and dose of supplements used in a mito cocktail are unique to each patient. They are selected based on individual needs, as mitochondrial disorders can present different challenges in different patients. "The use of the mito cocktail is primarily based on the assumption that higher doses of supplements may improve mitochondrial energy generation. This assumption remains largely unproven, and presently the evidence of their effectiveness does not rise to the level required for universal use".<sup>2</sup>

The sentiments related to this therapy and its overall impact on mitochondrial disorder management were varied. Participants shared, "The mito cocktail has helped provide stability and periods of improvement". In contrast, others have shared, "It is impossible to know if the cocktail actually helps without going off of it for a long period of time, which I haven't done."

The reported provincial coverage for the mito cocktail was mixed. 34% reported 100% coverage through provincial plans. Conversely, 43% reported paying 100% out-of-pocket due to little or no provincial coverage. Very few participants shared the out-of-pocket costs incurred in purchasing the mito cocktail. Those who did share expenses also used hospital programs to offset the total cost of this therapy. Therefore, it was difficult to fully capture the direct cost of mito cocktail therapy and the financial burden it possibly places on individuals and families at this time.

Movement therapies such as occupational therapy, physiotherapy and massage therapy were well reported as helpful management strategies.

2. Parikh, roach to the treatment of mitochondrial disease." *Current treatment options in neurology* vol. 11,6 (2009): 414-30. doi:10.1007/s11940-009-0046-0 Sumit et al. "A modern app

Exercise was the highest-rated symptom management strategy used amongst participants. The caveat is that the amount of daily exercise varied due to energy levels and that the time spent exercising was matched with equal rest periods. Exercises used to help manage mito symptoms varied; participants reported: leg stretches, elliptical machine (used to stair climb, walk, or run), swimming, cardio and weights, and walking.

## Quality of Life

Muscle weakness and fatigue were the highest rated physical symptoms having the most significant impact on quality of life. Loss of independence, isolation and helplessness were the highest reported invisible impacts. Participants shared that living with a mitochondrial disorder takes a significant toll on themselves and their family members. Respondents noted family sacrifices ranging from taking on second jobs or taking out loans or second mortgages to help purchase mobility aids or retrofitting homes to improve accessibility.

## Sources of Education and Future Priorities

The final component of our consultation explored sources used to educate and inform those living with mitochondrial disorders and the types of support the community would like to see MitoCanada provide.

43% of participants shared that they rely on MitoCanada for educational information, support and to keep current on clinical trials. It is interesting to note that only 23% shared that they rely on their specialist for disorder-specific information and clinical trial opportunities.

When asked what types of support they would like MitoCanada to provide, survey and interview participants identified three key priority areas: community support, advocacy, and education.

## RECOMMENDATIONS

Based on insights obtained from this pan-Canadian community consultation, coupled with ongoing dialogue with researchers, clinicians, and other stakeholders, MitoCanada recommends prioritizing the following seven objectives:

- **Increasing public awareness:** If you ask a “person on the street” if they know about mitochondrial disease, the answer is likely to be no. But one day they could give birth to an affected child—or meet someone with a mitochondrial disorder—and knowledge is power. It is time to put mitochondrial disease on the public awareness map.
- **Educating physicians:** Primary care physicians may have a general idea of mitochondrial disease, but lack the knowledge and skills to suspect or detect it or make a referral to a specialist. This lack of knowledge leads to delays in specialist referrals and delays in diagnosis. As the gatekeepers within the Canadian medical system, primary care physicians must be educated to better understand and identify mitochondrial disease.



- **Providing management tools:** Overall, individuals with mitochondrial disease are an uncommonly motivated group, with substantially higher adherence to treatment than individuals with other chronic diseases. At the same time, the mitochondrial disease community does not have the full range of tools they need to manage their disease. Affected families have expressed a need for patient-friendly information on disease management, from nutrition and complementary therapies to pain and digestive issues.
- **Strengthening the community:** The mitochondrial disease community seeks deeper connections. Affected families need each other's support to navigate provincial health systems and to deal with the daily challenges of living with mitochondrial disease. There is an opportunity to strengthen the community through patient conferences, online discussion and support groups, and partnerships with mitochondrial communities from other parts of the world that include all key stakeholders.
- **Raising the evidence bar:** Evidence for mitochondrial disease interventions, from the mito cocktail to exercise modalities, is inconsistent and imprecise, leaving many individuals with doubts about the value of these treatments and the best protocols to follow. It will be important to gather the best evidence from around the world and make it available to affected people and their healthcare providers.
- **Facilitating patient-centred mito research:** Individuals and their families need a simple, accessible mechanism for enrolling in clinical trials, with minimal administrative burden. They also need more opportunities, as patient-partners, to communicate their research priorities to MitoCanada, research institutions, and pharmaceutical manufacturers.
- **Ensuring equitable access:** Some treatments and devices are covered, others are not. Coverage also varies across jurisdictions within Canada. Such inequities make it challenging for individuals and families to predict and budget their disease-related expenditures. Obtaining more granular information about cost burdens and presenting this information to policymakers and private payers will help reduce inequities and establish reasonable coverage standards.

A stepwise process can help ensure these recommendations move forward. The process may include:

Step 1: validating and fine-tuning the recommendations with the community

Step 2: designating working groups

Step 3: providing periodic progress reports for the wider community.

Implementing these recommendations will bolster the aims shared by all stakeholders: improving the physical, social, psychological, and financial outlook for individuals and families affected by mitochondrial disease.

## CONCLUSION

Mitochondrial disease is not only a health challenge, but a journey. MitoCanada is proud to share this journey with affected individuals and their families and, we hope, to make the road a little easier to travel, to manage the symptoms of today while being hopeful of new treatments in the future. We view people living with mitochondrial disease and caregivers as the true experts and are committed to acting on their feedback. This community consultation reflects this commitment.

Clinical studies yield important data that help move treatment forward, but do not always capture the human dimension of living with mitochondrial disease. MitoCanada launched this consultation with the intent of filling this gap, and we have not been disappointed. Participants expressed strong and specific needs that helped point the way to our recommendations. They helped us understand how mitochondrial disease has impacted them. They raised our awareness of the benefits and limitations of treatment. They told us about what's working and what's missing in their lives.

Above all, they confirmed the value of supporting and connecting members of this unique community.

Let the dialogue continue!



## ACKNOWLEDGMENTS

The completion of our Community Insights Report was a true community collaboration. We are proud of what we achieved together and are pleased to share these insights with you. MitoCanada continues to be inspired by the agility and determination of our community partners to transform the outlook of mitochondrial disease in Canada.

Thank you to every individual and caregiver who shared their lived experiences, challenges and barriers and feedback with us through our survey and interviews. Your partnership was instrumental in crafting this inaugural report. We are incredibly grateful for your time and valuable contributions.

Thank you to MitoCanada's Patient-Family Advisory Council (PFAC), patient-partners and researcher-clinicians who assisted with survey development and reviewing our community report. Your diligent reviews and feedback enhanced the report in an immeasurable way.

Thank you to our allied partners for aiding our survey recruitment efforts; as always, we appreciate your continued support.

## About MitoCanada

### MitoCanada is energizing lives!

We use our collective energy to reduce the devastating impact of mitochondrial disease. All of our efforts are dedicated to creating a world where all lives are powered by healthy mitochondria.

Every day, we support those living with mitochondrial disease with information, initiatives and opportunities to inspire, empower and connect while enhancing quality of life. We develop education and awareness programs.

We advocate for those living with, or at risk of developing, mitochondrial disease. And, we fund research that is patient-focused and transformational.

To learn more about MitoCanada's, please visit: [www.mitocanada.org](http://www.mitocanada.org)

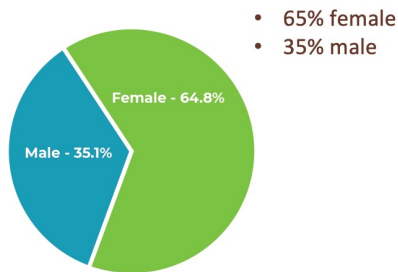
To support MitoCanada by making a donation, please visit: [www.mitocanada.org/donate](http://www.mitocanada.org/donate)



# Community Survey Graphs and Tables

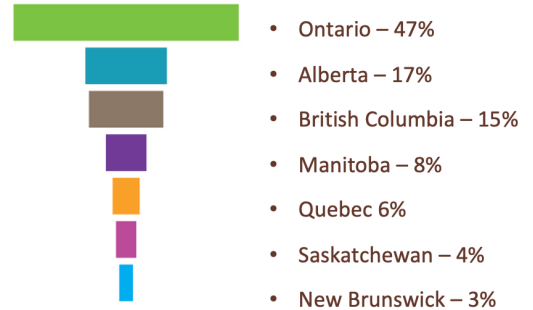
## DEMOGRAPHICS

There were 81 participants of which:

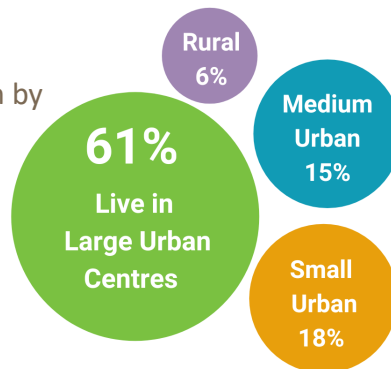


- 65% female
- 35% male

Province of Residence



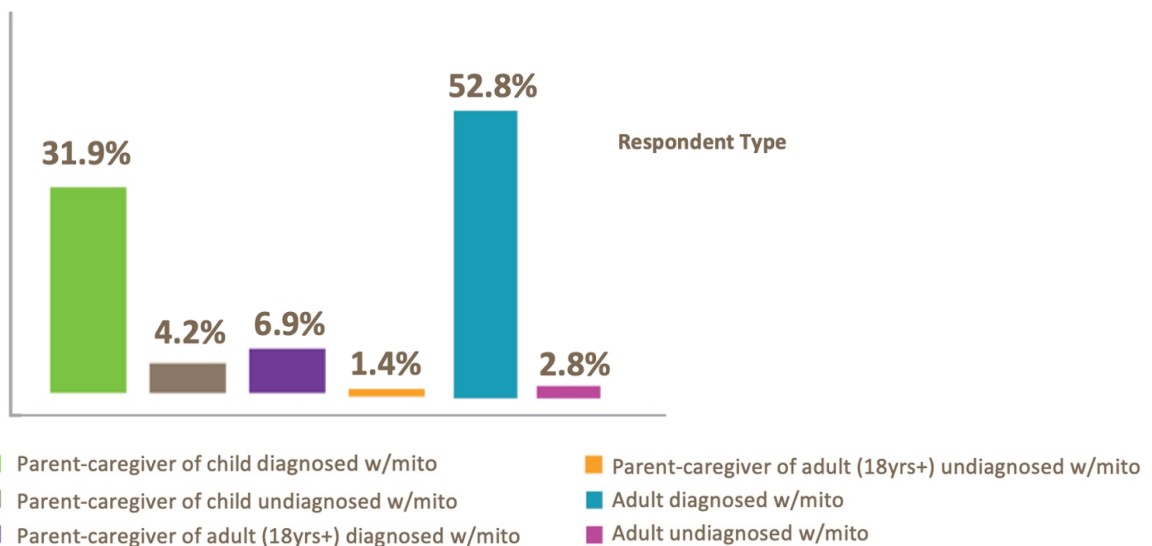
Geographic Distribution by Population Centres



## DIAGNOSIS

Respondent Type

- 92% of participants have received a mitochondrial disease diagnosis
- 8% of participants were undiagnosed

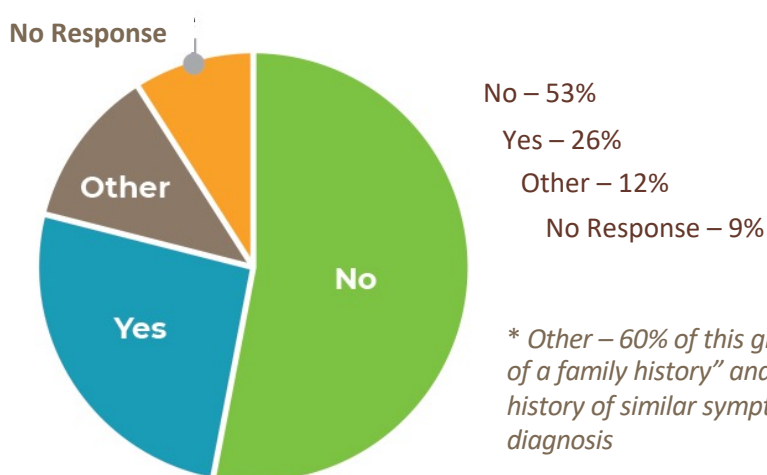


## Age range for participants:

12.5%	0-5 years old	7.5%	25-34 years old
16.3%	6-12 years old	12.5%	35-44 years old
3.8%	13-17 years old	20%	45-54 years old
2.5%	18-24 years old	16.3%	>55 years old

\*7% did not disclose their age

## Reported Family History of Mitochondrial Disease:

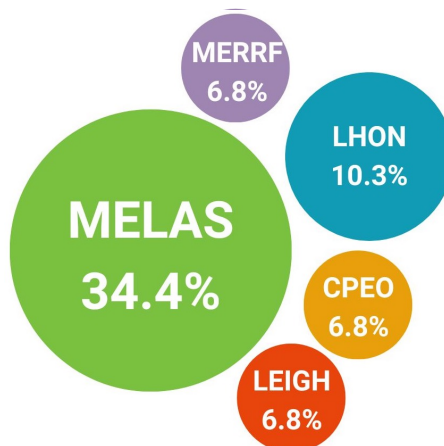


\* Other – 60% of this group reported “not being sure of a family history” and 40% reported a family history of similar symptoms but not receiving a diagnosis

## Those reporting a family members diagnosed with a mitochondrial disease

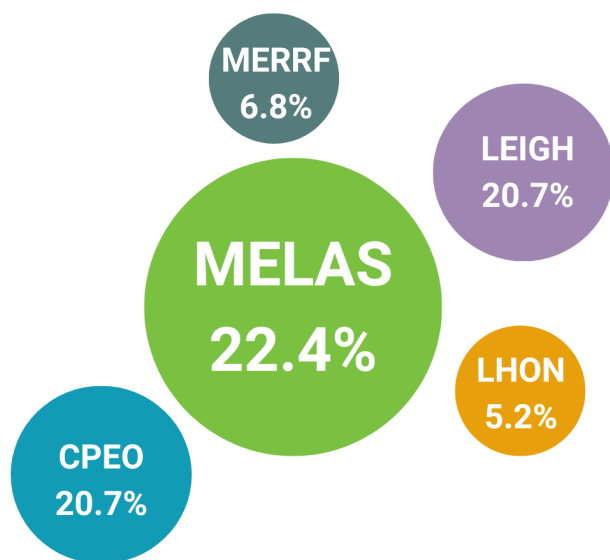
- 19% had a grandmother diagnosed
- 0% had a grandfather diagnosed
- 55% had a mother diagnosed
- 0% had a father diagnosed
- 42% had a sister diagnosed
- 13% had a brother diagnosed
- 16% had a daughter diagnosed
- 13% had a son diagnosed

## Top 5 mitochondrial disorders reported among family members:

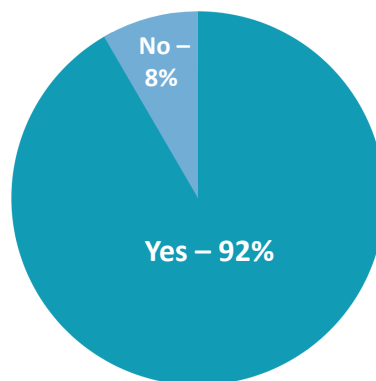




## Top 5 reported mitochondrial disease diagnosis



Participants reported receiving a clinical mitochondrial disease diagnosis



The time it took to receive a mitochondrial disease diagnosis after first seeking medical care for symptoms, fell into four distinct ranges:

- 9% reported it took less than a year
- 45% reported it took 1-3 years
- 13% reported it took 4-6 years
- 32% reported it took >7 years

46% reported receiving a diagnosis between the ages of 0-17 yrs.

54% reported receiving a diagnosis as an adult, aged 18 years and older.



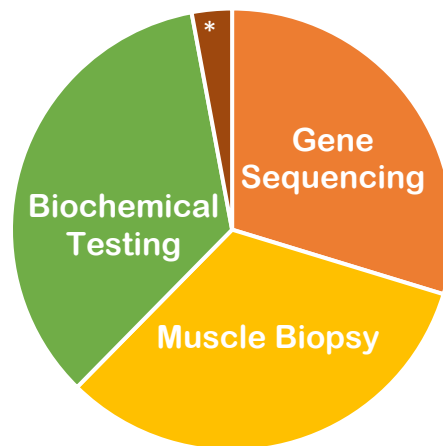
The peak age of diagnosis among youth was between the ages of 0-5, with 32% reporting a diagnosis in this age range.



The peak age of diagnosis among adult was between the ages of 45-55, with 21% reporting a diagnosis in this age range.

When asked what tests or procedures were used to receive a clinical mitochondrial disease diagnosis, participants reported:

- Biochemical testing 61.4%
- Gene sequencing 52.6%
- Muscle biopsy 57.8%
- Other 5.2%



\* Other

Participants reported the following health care professionals gave their mitochondrial disease diagnosis:



Neurologist – 29%



Neuromuscular Specialist – 33%



Geneticist – 33%



Other – 13%

*\*includes ophthalmologists, metabolic specialists*

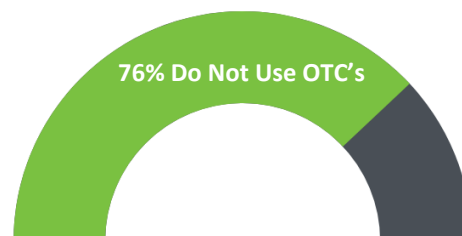
## TREATMENT LANDSCAPE

In this survey, participants were asked a series of questions about treatments, including prescribed medications, supplements and therapies, and asked to share their attitudes towards these interventions.

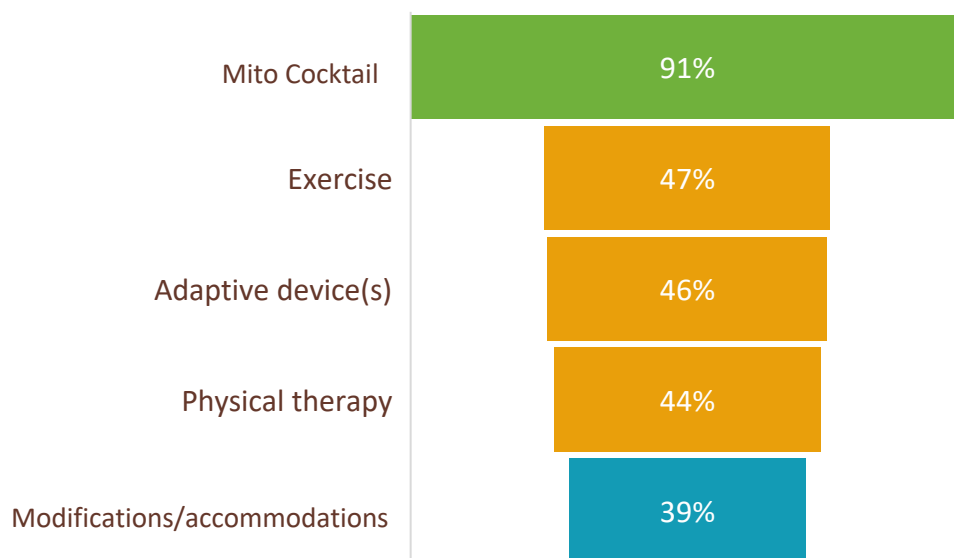
When asked if over-the-counter (OTCs) medications are used to manage mitochondrial disease symptoms, 76% of participants shared not using OTCs, while 24% disclosed using OTCs.

The types of OTCs used include:

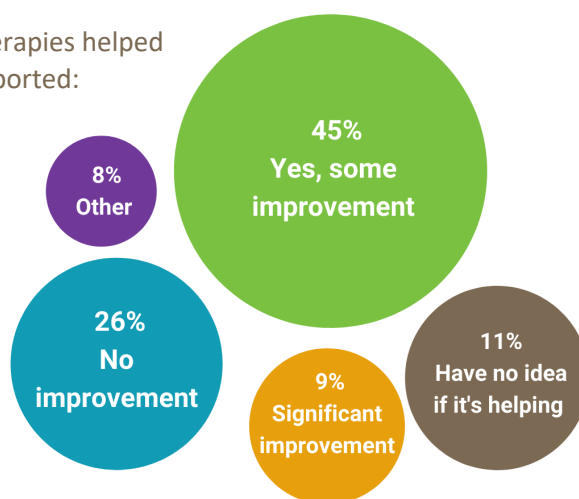
- Eye drops
- Loperamide
- Analgesics (acetaminophen, ibuprofen)
- Vitamin D



When asked to share what treatment(s) or therapy have been prescribed to manage their mitochondrial disease, participants shared:



When asked if prescribed treatment(s) or therapies helped improve quality of life (QoL), participants reported:



Participants also shared:

“Appears to have stabilized our son metabolically and reduced progression of dysfunction.”

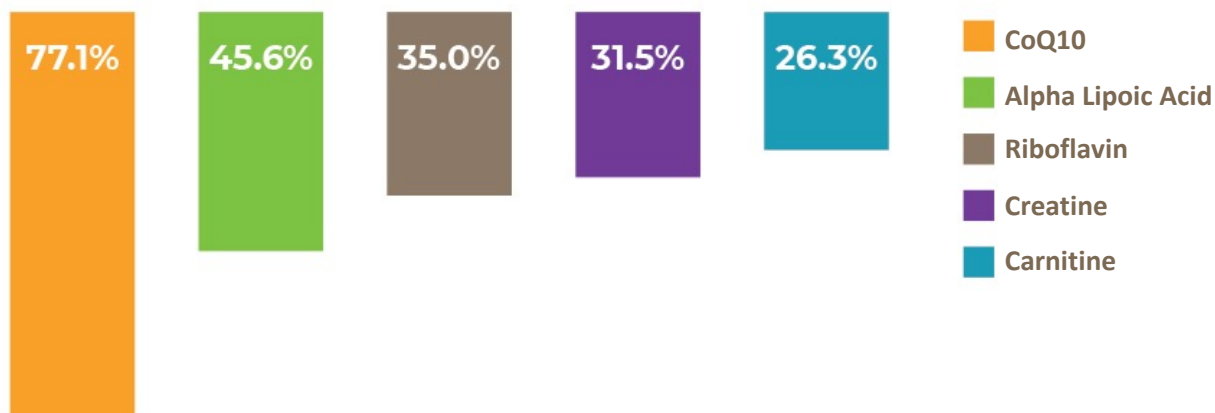
“Hard to say, until you stop, there is no way of knowing if they are helping.”

“Believe the mito cocktail did help with symptoms at the onset”

“It has stopped me from plummeting at times and kept me stable at others, and then have periods of improvement”

## Using and Accessing the Mito Cocktail

Participants were asked to list supplements that comprise their mito cocktail. The following are the most common supplements reported:



When asked if participants take their mito cocktail as prescribed by their clinician:

- 84% reported taking the mito cocktail as prescribed, also sharing:

*"Prescribed by our functional medicine doctors, not our Mito doctors because they have basically said mito cocktail has no proven evidence that it helps and basically no treatment or cure.*

*Just deal with symptoms as they arise."*

*"it's expensive, and some elements do not seem to help me, though others definitely do.*

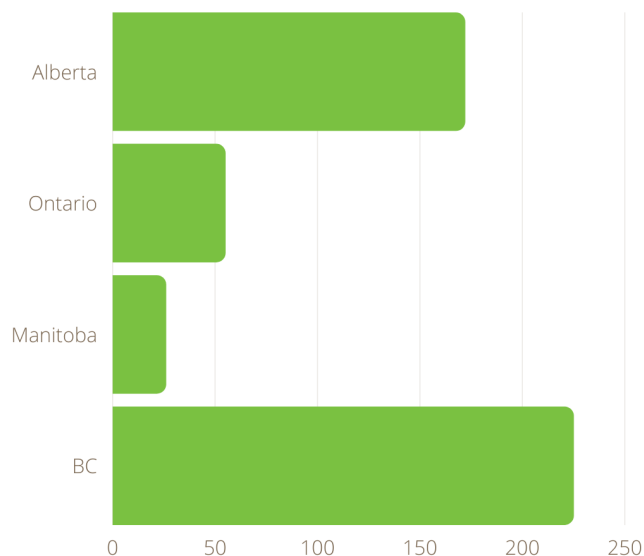
*Still I take it religiously, as directed."*

- 16% reported not taking the mito cocktail as prescribed, falling into one of the four following categories:
  - 6% reported taking the mito cocktail sometimes
  - 4% reported taking it partially
  - 2% reported not taking the mito cocktail as prescribed
  - 4% reported other reasons for not taking the cocktail

## Mito Cocktail Coverage Insights

When asked about coverage for the mito cocktail:

- 34% reported receiving 100% provincial-public coverage.
- 11% reported partial provincial-public coverage
- 0% reported 100% coverage from private insurance providers
- 11% reported partial coverage from private insurance providers
- 43% reported paying 100% out-of-pocket



The average monthly, out-of-pocket cost, by province, for the mito cocktail:

- Alberta \$172/person
- Ontario \$55/person
- Manitoba \$26/person
- British Columbia \$225/person

*\*few participants provided financial details on the cost of their mito cocktail therapy*

## Coverage Insights by Province

### Ontario

Adult respondent shared receiving coverage through hospital program and the Inherited Metabolic Diseases (IMD) program.

### Alberta

Parent-caregiver respondents reported receiving mito cocktail coverage through the Metabolic Clinic at Alberta Children's Hospital. Some received additional coverage through the Family Support for Children with Disabilities (FSCD) program.

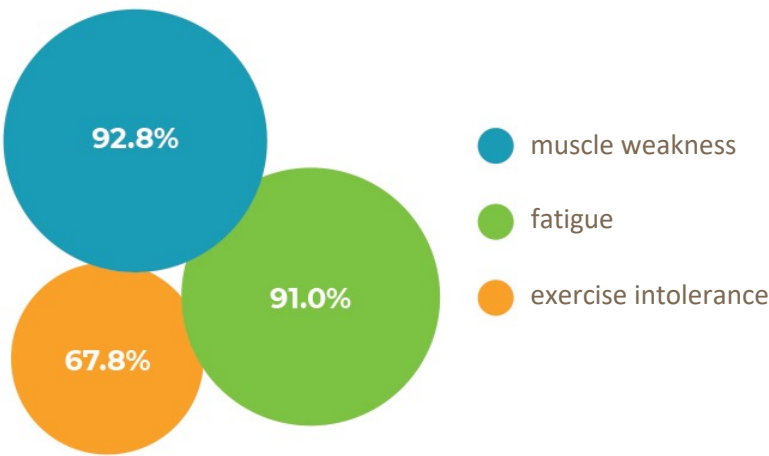
### British Columbia

Taurine and arginine covered through the BC Metabolic Nutrition Program, with carnitine partially covered by British Columbia Medical (BCM).



# IMPACT ON QUALITY OF LIFE

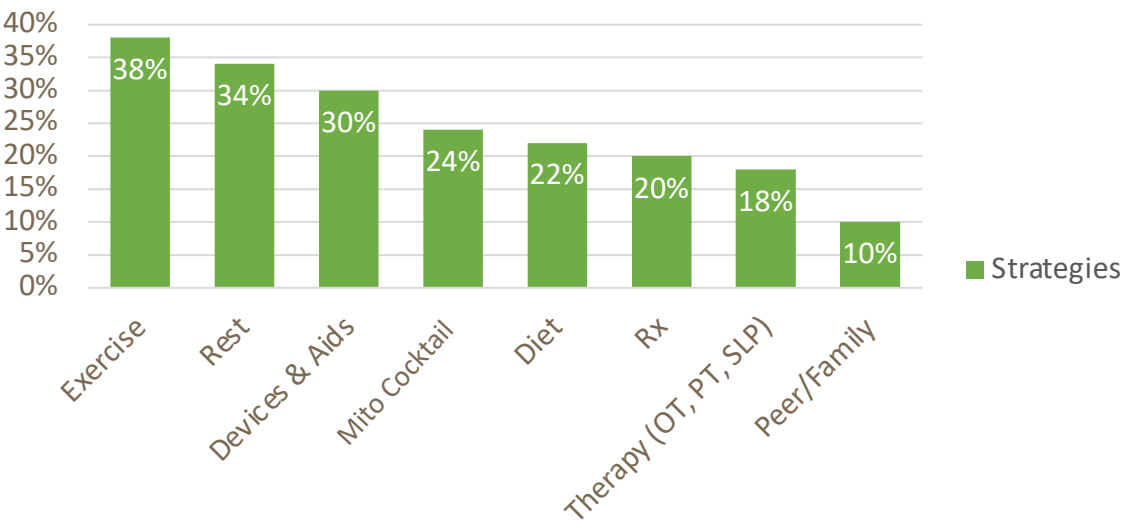
Participants were asked to identify what mitochondrial symptoms had the greatest impact on their quality of life.



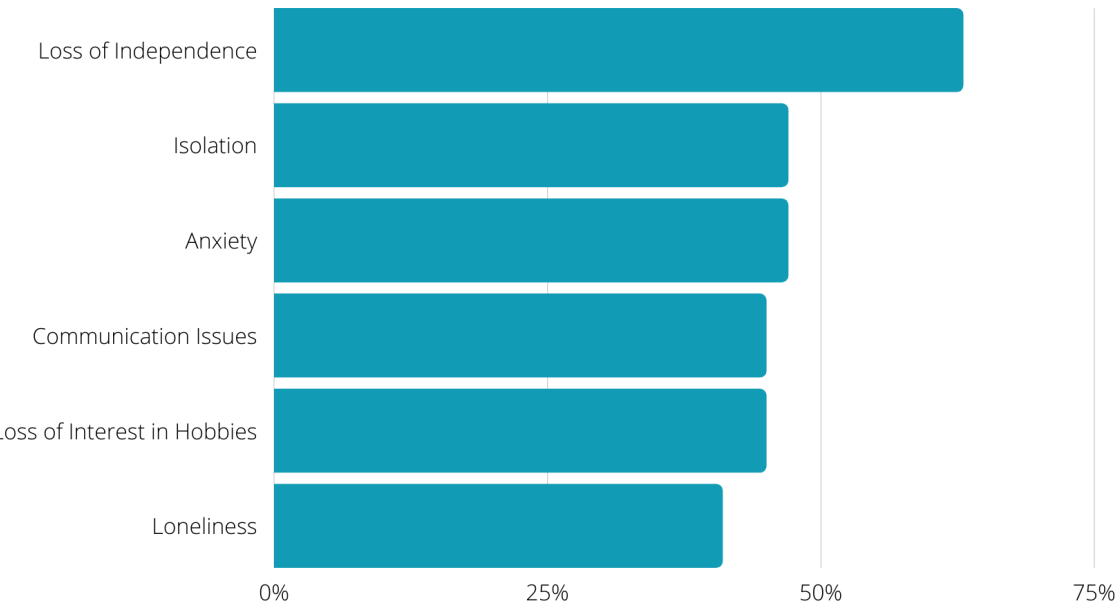
While muscle weakness, fatigue and exercise intolerance were reported as the top three symptoms (as shown above), the following were also ranked "high" in impacting quality of life:

- gastrointestinal problems (57%)
- pain (54%), and
- vision problems (52%)

Participants reported using the following strategies to help manage their mitochondrial disease symptoms:



Participants were asked to share how mitochondrial disease has impacted their life:



Participants were asked to share sources they use to obtain information about mitochondrial disease, to improve their quality of life:

