MITOCHONDRIAL DISEASE &

MENTAL HEALTH

A CALL TO ACTION TO INCORPORATE MENTAL HEALTH INTO MITOCHONDRIAL DISEASE PATIENT CARE

PROPOSED BY

ABOUT OUR CONTIRBUTORS



MITO2i stands as a global nexus for groundbreaking mitochondrial research and medical advancement. This project benefited from MITO2i's research grants, expert content review, and the development of a strategic outreach and knowledge dissemination plan

mit@canada

MitoCanada supports, educates and helps protect Canadians living with or at risk of developing mitochondrial disease. To reduce the devastating impact of mitochondrial disease, MitoCanada focuses on four key priority areas that drive meaningful change, research, education, support and awareness. We support those living with mitochondrial disease in developing education and awareness programs, advocate for those living with, or at risk of developing, mitochondrial disease and fund research that is patient-focused and transformational.





The Population Health Analytics Lab at the Dalla Lana School of Public Health, University of Toronto, led the research, data analysis and interpretation of the papers for mitochondrial disease in Ontario. The Population Health Analytics Lab also led the patient and caregiver focus group to share our research findings and hear how this resonated with those with lived experiences and discuss the potential impact for patient care.

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INTRODUCTION

According to a study in 2021 by Buajitti et al., [<u>1</u>], 1 in 4,000 Ontarians has mitochondrial disease and 18% of patients also have a mental health disorder. Living with mitochondrial disease is really like living with a single battery on a whole house.

"

With rare diseases affecting fewer than 1 in 12 Canadians [2], there is a significant gap in support and treatment for patients, including rare disease patients with Mitochondrial disease.

MITOCHONDRIAL DISEASE

- A Neurometabolic disease comprising of mutations in the genes that encode mitochondrial proteins and their function [3]
- The mitochondria acts as the centres for energy production in all human cells, alterations in this organelle's function can have devastating effects on multiple body organs.
- Drastic effects on the brain , which is highly energy dependent to provide essential life support and higher-level functioning.
- With 350 genes already identified [4], the clinical presentations of mitochondrial disease vary drastically with different degrees of impairment and disability, depending on which mutation is present.



PREVIOUS WORK

In 2021, Buajitti and colleagues performed a population-based cohort study to determine the prevalence and healthcare costs of Mitochondrial disease in Ontario, Canada. Beyond assessing the prevalence of Mitochondrial disease at 1 in 4,000 Ontarians, their findings also highlighted that 18% of mitochondrial patients experienced a mental health problem [1]. Following the cohort study results, Mitochondrial Innovative Initiative (MITO2i), in partnership with the University of Toronto and MitoCanada, performed a knowledge translation and engagement focus group study to determine patients' thoughts and emotions regarding the previous study findings. The focus group was fundamental to understanding how this research and future work can benefit mitochondrial disease patients, caregivers, and the community. This paper aims to promote the focus group members' voices, respond to their concerns, and provide future policy changes and research recommendations. We have also engaged further feedback from physicians treating mitochondrial disease patients regarding current standard care practices and changes to be made. Ultimately the goal is to highlight the importance of mental health and wellbeing of mitochondrial disease patients.

Age of first hospitalizations for patients with mitochondrial disease in Ontario



Almost 1 in 5 patients in Ontario with mitochondrial disease also have a mental health condition



Healthcare costs of patients with mitochondrial disease

BEFORE HOSPITALIZATION

\$6,900

AFTER HOSPITALIZATION

\$21,300

Healthcare costs of patients with mitochondrial disease & mental health condition

BEFORE HOSPITALIZATION

\$10,100

AFTER HOSPITALIZATION

\$33,400

ABOUT THE PATIENTS

The focus group consisted of five participants: four patients and one caregiver. A few patients have also offered their personal experiences as a person living with mitochondrial disease in Ontario. Stories and Quotes have also been taken from MITO2i MITOvoice's. For confidentiality, we have changed patient names within this paper, except for Thomas who requested we use his first name.

RECOMMENDATIONS

Increased awareness and education surrounding mitochondrial disease and its relationship with mental health disorders





Integrated care for all mitochondrial disease patients

MENTAL HEALTH

The focus group identified that while the relationship between mitochondrial disease and mental health disorders is highly apparent, there is a significant gap in the education surrounding the increased susceptibility of patients. They also discussed the importance of physicians actively and routinely checking patients' mental health.

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Typically, self-management is considered in the context of biomechanical/biological issues only, but people living with mitochondrial disease are not solely a collection of their physical symptoms; we are human beings with emotions, stressors, fears, and goals.



Patients with rare diseases face psychological implications beyond physical symptoms. Symptom management, medical exams, and unpredictable future increase anxiety and stress. Mitochondrial disease patients experience daily stressors that require physicians to treat them holistically, focusing on both body and mind for overall wellbeing.

One of the focus group members put it best, referencing Canadian Physician Sir William Osler (1849-1919);

"

The good physician treats the disease; the great physician treats the patient who has the disease [5]



MENTAL HEALTH AWARENESS

THE PATIENT EXPERIENCE

After many tests over seven years, 13-year-old Lucy, now 20, was finally diagnosed with mitochondrial disease. Not once in the seven years had they been asked about their mental health. Mental health support must be available for children who are particularly vulnerable and unable to articulate their feelings.

Beth has shared that mental health care has been lacking in the broader health care system, with no psychological support provided after a near-fatal car accident. This gap in mental health services has become even more evident as they have not received any mental health support for their mitochondrial disease.

For Thomas, who had his first seizure at twelve, the next six years consisted of visiting doctor to doctor until diagnosis of Myoclonic Epilepsy and Ragged Red Fibres (MERRF) at 18. He was happy to receive a diagnosis finally but had to learn what mitochondrial disease is and navigate his disability on his own. Now Thomas shares his story to bring awareness to the disease and his disability. He advocates for patients to seek support from counsellors to be emotionally in a better head space to make the best decisions for themselves and their health

"

The right headspace can be life-changing.



MENTAL HEALTH AWARENESS

GAPS

1.Education available to mitochondrial disease patients about mental health and wellbeing

2.Health Care practitioner awareness

3. The prevalence of mental Health disorders associated with mitochondrial disease in Ontario

RECCOMENDATIONS

1. Easily accessible online modules and information packages for families regarding mental health and mitochondrial disease

Target: Mitochondrial initiatives and patient focused groups such as:

- <u>MitoCanada</u>
- <u>MITO2i</u>
- Lily Foundation
- My Mito Mission
- <u>Welcome Centre for Mitochondrial Research</u>
- <u>Mitochondrial Medicine Society</u>
- United Mitochondrial Disease Foundation (UMDF)
- <u>Mitochondrial Care Network</u>
- <u>MitoAction</u>
- <u>MitoFoundation</u>
- <u>Mitocon</u>

2. Education to healthcare providers not only about mitochondrial disease itself but also the mental health component of the disease.

Target: Target: Information to clinicians at research centres and clinical sites via brochures, workshops, and trainings.

3. Further research into mental health disorders and mitochondrial disease

Target: academic institutions, hospitals, and mitochondrial research initiatives

MENTAL HEALTH AWARENESS

ASSETS

- Continuing medical education (CME) credits can be leveraged for education on mitochondrial disease and mental health.
- <u>UMDF</u> offers Courses about mitochondrial disease; found at <u>MitoU</u>
- <u>MitoAction</u> includes a programs and support section with resources, programs, education, and planning. They have also included a day-to-day section on their website that provides advice, including living with mitochondrial disease, managing your energy, travelling and care management. MitoAction also hosts podcasts for patients to discuss mental health and mitochondrial disease.
- The <u>Lily Foundation</u> offers emotional and practical support, advice, and connection to medical specialists, for patients and families. The foundation also has coping tip pages for receiving a diagnosis and support for families, young people, and adults.
- The <u>Welcome Centre for Mitochondrial Research</u> at Newcastle University provided readings and updates to patients and families during the covid-19 pandemic regarding mental health and psychological advice. They also host annual patient days, inviting over 100 patients to interact informally with one another and their scientists.
- <u>My Mito Mission</u> offers peer support groups and helplines. They also have UK and global Facebook groups to build a community of mitochondrial disease patients to connect.
- The <u>Mitochondrial Care Network</u> provides <u>A guide to the Newly Diagnosed</u>, which includes where to find support, such as rare disease support networks.
- The <u>MitoFoundation</u> based in Australia offers helplines to patients and patient meetups. They also have a video on the <u>Emotional Challenges of Physical Symptoms</u> and <u>Mindfulness</u> to Manage Your Health and Life Challenges.
- <u>Mito21</u> presents MITOvoices, a podcast that engages patient advocates, community members and researchers to share their voices and raise awareness and education about mitochondrial disease.
- <u>MitoCanada</u>'s community insights report in 2021 aimed to identify and address the needs of mitochondrial patients across Canada.



STANDARD CARE

Mental health care is vital when the disease impacts the brain resulting in neurological symptoms.

Mitochondrial patients in Ontario are followed by pediatric or adult specialists from different hospitals, including SickKids and McMaster and others. Appointments are generally every six months to one year. Focus group members identified the following main themes regarding their standard care.

PATIENT-INITIATED REQUESTS FOR SUPPORT

The focus group members discussed that comprehensive standard practice should include referrals without the patients' responsibility to initiate the request. Joe requested mental health support but was only offered online and group sessions through the Ontario Structured Psychotherapy Program, which they found insufficient to support their psychological well-being.



I believe this is nothing more than making time as part of each appointment/follow-up to ask people about how they are coping with this illness and address the emotional, psychological, social, spiritual, etc. impacts of the disease in addition to the physical. A physician does not need to be a psychologist to have these basic conversations about a person's feelings and emotions.





STANDARD CARE

THE RIGHT SUPPORT

Focus group members identified that obtaining the proper support for mitochondrial disease is challenging due to the scarcity of specialists and psychologists, leaving patients responsible for educating themselves and mental health workers on their condition.

DELAYS IN CARE

Focus group members report extensive delays in care, especially when specific support services require patient-initiated requests.

Other delays in care and increased wait times result from the current state of the Ontario health care system and the lack of available and affordable mental health specialists [<u>6</u>].

Thomas expressed that though he asked his doctor for a referral to counselling services, it took many months before he was able to find support.



When Thomas first spoke to a counsellor, he did not find the sessions helpful due to the counsellor's lack of knowledge and understanding regarding mitochondrial disease. Later he tried another counsellor and found that by presenting information about the condition, he could have better conversations about the impacts the disease and disability have on his mental health.

FUNDING SERVICES

Many psychologists are departing from publicly funded roles, such as hospitals, in favour of more lucrative opportunities in the private sector. This results in numerous vulnerable patients being unable to afford vital mental health care [<u>6</u>]. The focus group stated that although financial assistance is available for various aspects of mitochondrial disease, mental health services are not sufficiently covered, creating a notable deficiency in comprehensive care and a barrier to accessing support.

"

I am followed by a psychiatrist at McMaster however, there needs to be more therapy available as I cannot afford to pay for therapy.

"

STANDARD CARE

GAPS

- 1. Discussions regarding mental health as standard care for mitochondrial patients
- 2. Mental Health counsellors and clinicians that understand mitochondrial disease and the physical/psychological associations of the disease that impact mental health
- 3. Reducing delays in care
- 4. Funding for services

RECCOMENDATIONS

1. Routine check-ins by healthcare providers with patients about their mental wellbeing

Target: healthcare providers

2. Information regarding the right supports for patients, including the specialist's role and scope of expertise

Target: Local mitochondrial disease initiatives

3. Reducing delays in care by increasing mental health staff and specialists

Target: Ontario Healthcare System

4. Increased funding for patients to access mental health services

Target: Government Policies

ASSETS

1. Psychometric tools that screen for mental health disorders are widely available, validated and used. Within SickKids, the HEADSSS tool is used to assess mental health of adolescent patients in the metabolic disease clinic (see pediatric supports)



ADULTS VS PEDIATRICS

The information presented here is based on the experiences of two hospitals, SickKids in Toronto, and McMaster in Hamilton. However, to gain a complete understanding of the current standard practices for patients with mitochondrial disease, it is important to engage with other Ontario centers that treat such patients. This paper serves as a preliminary document to facilitate further discussion on patient care pathways in Ontario.

STANDARD PEDIATRIC CARE

For pediatric patients within a large urban hospital, including Toronto's SickKids, access to care and services is integrated with multidisciplinary teams to support the well-being of patients and their families. Routine visits include questions and screening for the behavioural and emotional functioning of the patient. For patients that require further support, referrals are available to other services such as psychiatry and social work.



ROUTINE QUESTIONS

At each visit, caregivers are asked questions regarding their child's emotional and behavioural functioning. Adolescents' mood and mental health should be asked confidentially in an environment where the young person feels their concerns are heard and addressed.

Caregivers are also asked questions regarding school supports such as IEP (independent educational program) and functioning within the classroom.

Existing support services are addressed, including occupational therapists, physiotherapists, speech and language therapists, access to home nursing if needed, personal support workers, etc.

SCREENING

Screening questions should be age-group specific and depend on the cognitive age of the patient. Screening questions look at symptoms of disorders including anxiety, mood, psychotic, eating, personality, autism spectrum and neurodegenerative.

Adolescent screening:

At SickKids, the HEEADSSS screening assessment tool is a psychosocial interview that addresses the young person's risks and functioning. This tool consists of (1) Home, (2) Education, (3) Eating/exercise, (4) Activities, (5) Drugs/Substances, (6) Sexuality, (7) Suicide/Depression and (8) Safety.

ADULTS VS PEDIATRICS STANDARD PEDIATRIC CARE

REFERRALS

The SickKids Neurometabolic clinic has a tiered system of referrals and supports based on the severity of the problem assessed at screening.





All families and patients are provided with Neurology Clinic nurse support contact numbers

Mild, situational, or related to accessing resources -> Referral to Neurology

Therapists from CCAC (Community Care Access Centre) can visit and evaluate the home situation

For more significant problems (i.e. bipolar disorder, schizophrenia, autism spectrum disorder, attention deficit hyperactivity disorder) -> Referral to a child psychiatrist at SickKids for diagnosis and management, a community child psychiatrist or a community mental health facility. Further, supports include pharmacotherapies, monitoring and behaviour management.

Further interventions, including multidisciplinary care team meetings between neurometabolic, Psychiatry and social work, may be needed.

TRANSITIONING FROM PEDIATRIC TO ADULT

Once a pediatric patient turns 18, they are no longer a part of the pediatric hospital setting, eliminating their access to treatment from multidisciplinary care services. Beyond receiving a referral to an adult specialist, no transition support is currently part of standard care, leaving vulnerable young patients to self-navigate and advocate for resources and support.

LACK OF ADULT RESOURCES

Transitioning from pediatric to adulthood means losing a community and a place that felt like home. I don't feel part of a community unless I create my own

"

Thomas identified the lack of community support as an adult and the isolation associated with disability. He discussed what it would mean for him to have access to a regular support group with other mitochondrial disease community members and persons with a disability.

Focus group members identified that patients with mitochondrial disease require a holistic approach to treatment, including efficient case management, solid support systems, and improved understanding among specialists.

The integrated care approach prioritizes patients, involves collaboration among specialists and services, and is continuous. This model allows patients to focus on their health rather than the coordination of their care [$\underline{7}$].

Unlike the integrated care available in pediatric hospitals, adult resources are separate and require self-navigation by the patient. At McMaster, approximately 30% of mitochondrial patients have mental health issues discussed at routine appointments. The current state of Ontario's healthcare system has made it even harder for specialty clinics, including McMaster, to refer their patients to psychiatrists. The lack of available psychiatrists, psychologists and overall mental health services appears to be much more significant in adult patients than in pediatric patients.



Many people living with a mitochondrial disease carry the daily burden and weight of the illness, and it is important to have opportunities to have effective and therapeutic conversations about this

ADULTS VS PEDIATRICS

GAPS

- 1. Lack of available mental health services and specialists (i.e. Psychologists, psychiatrists, support staff, etc.) and wait times for both pediatrics and adults
- 2. Support for pediatric patients transitioning from multidisciplinary care to adult services as they turn 18.
- 3. Lack of integrated care for adult patients

ASSETS

 A subgroup for transitioning neurometabolic patients to adult services is part of the working group in child neurology at SickKids.

RECCOMENDATIONS

1. Increase the importance of prioritizing mental health services for adult and pediatric mitochondrial patients.

Target: Health Care Centres, Mitochondrial Disease Initiatives, and foundations.

2. Transition support for pediatric patients once they have turned 18

Target: Pediatric and Community Hospitals

3. Further research is necessary to understand the mental health and support needs of patients with mitochondrial disorders. Developing an effective integrated care model requires qualitative and longitudinal studies.

Target: Research centres, Clinical sites, Public Health agency's



FINAL REMARKS

Through recent research initiatives and collaborations with the focus group and mitochondrial disease community members, we have identified essential points and recommendations to incorporate mental health care for mitochondrial patients into patient care:

- 1. There is a pressing need for increased awareness and education surrounding mitochondrial disease and mental health within the medical, research and mitochondrial disease communities.
- 2. Standard care for mitochondrial disease patients must include comprehensive conversations and support for mental health, acknowledging the relationship between physical and psychological well-being.
- 3. An integrated care model for adult patients to promote a comprehensive approach to addressing complex healthcare needs.

Collaboration and partnership between patients, mitochondrial disease initiatives, hospitals, and policymakers are essential for fostering a more holistic treatment approach and supporting patients. Research activities need to be strengthened through qualitative and longitudinal studies to understand further the disease's progression and its effects on mental health. By engaging advisory groups and international collaborators, greater knowledge exchange will accelerate research advances. Looking ahead, the community aims to build an influential policy paper that can shape healthcare practices and public policies in addressing mitochondrial disease and mental health collectively. This paper hopes to bring forward continued advocacy, fundraising, and collaboration to research and promote the wellbeing of those affected by mitochondrial disease and its associated mental health challenges.

Mitochondria research matters to millions.



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