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Rare perturbations

Management as imagined versus management as done?

Resilient Health Care Network 2022, 14-17 August 2022, Jönköping, Sweden

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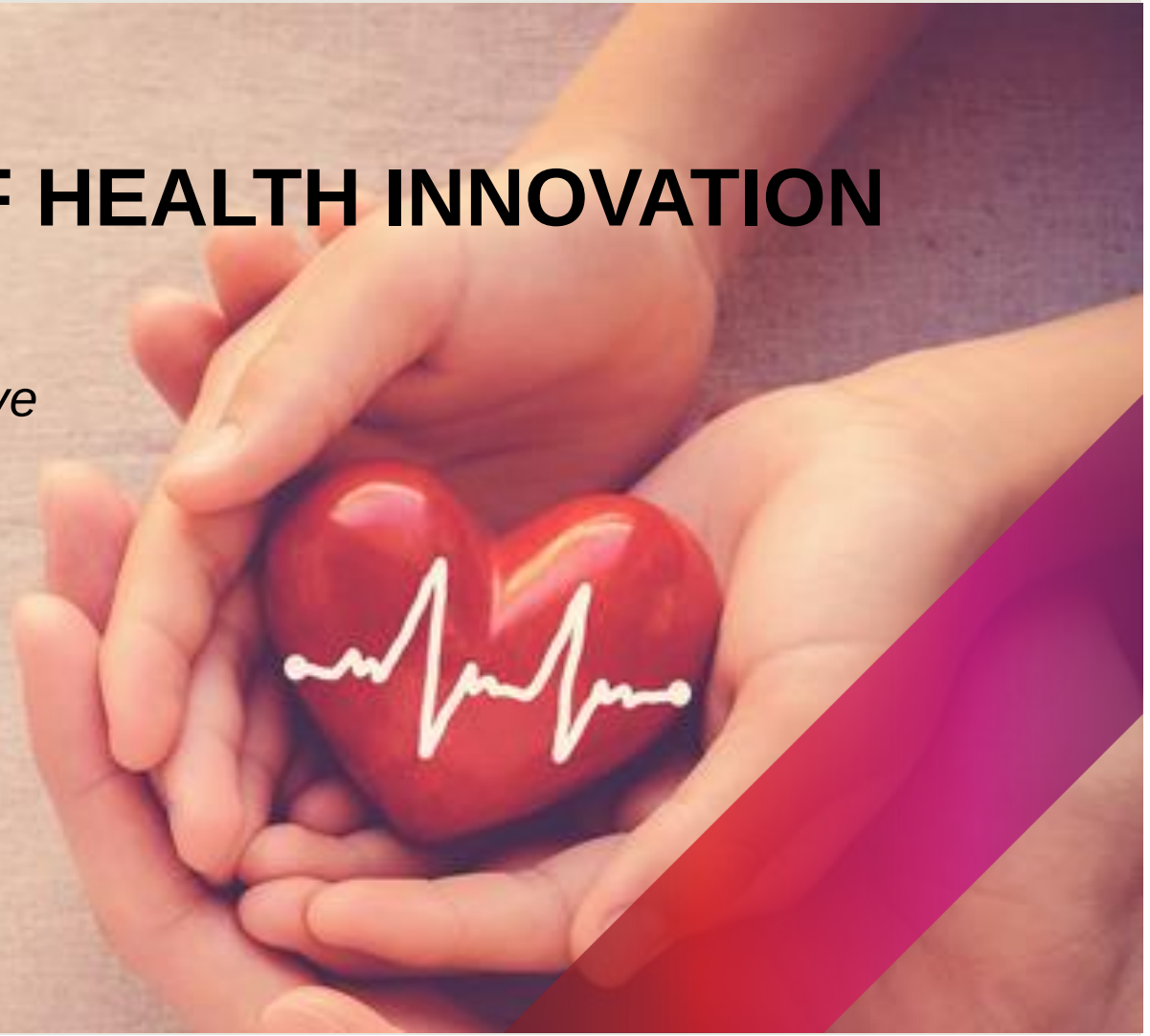

Acknowledgements

Mito Foundation Project Grant 2019-2021

EXPERT RESOURCES PANEL

MITO COMMUNITY ADVISORY PANEL

RESOURCE WORKING PARTY

Abstract

When talking about resilience of health services, we often think about service delivery in response to a large perturbation (e.g., a natural disaster with multiple casualties). What about smaller perturbations? The presentation of a patient with a rare disorder could be considered one of these small, infrequent perturbations. How does the health system respond to patients seeking help for a condition they have never heard of? What does resilience look like in this situation?

The aim of this presentation is to provide food for thought around resilience in the face of rare conditions. We examine the gap between management of a rare disease as it is thought to happen (by specialist services) and how it really happens (as reported by people with a rare disease). How can health services provide high quality, safe care to extremely low prevalence conditions?

We consider qualitative data from 6 focus groups of 20 people genetically diagnosed with a rare condition - Mitochondrial Respiratory Chain Disorders, (hereafter “mito”) or caring for a child diagnosed with mito. Severe mito affects around 1:5,000 births. Childhood presentations tend to be more severe and acute while adult cases often have a milder chronic course. Mito can affect any organ, with highly variable clinical presentations, usually involving multiple systems. Mito has no cure and limited evidence for treatments. We asked participants about their experience accessing health or social care from health providers and disability services. We also collected survey and interview data from 8 mito specialists from across Australia about their management practices after diagnosis and identified several areas for improvement.

Consumer participants revealed that after they left the mito specialist service, many health service providers either ignored the condition or chose not to engage with them. While consumer empowerment and advocacy for one’s own care is a popular approach in the rare disease community (e.g., as advocated by the European Rare Disease Network), our work revealed it was (a) exhausting for people already struggling with fatigue and multiple health issues, setting up inequities of access to care, and (b) difficult due to systemic barriers to care – easy to trigger supportive services for high incidence conditions but a fight for low incidence conditions. We discuss various rare disorder models of care that are emerging in Australia in terms of service resilience.

Perturbations

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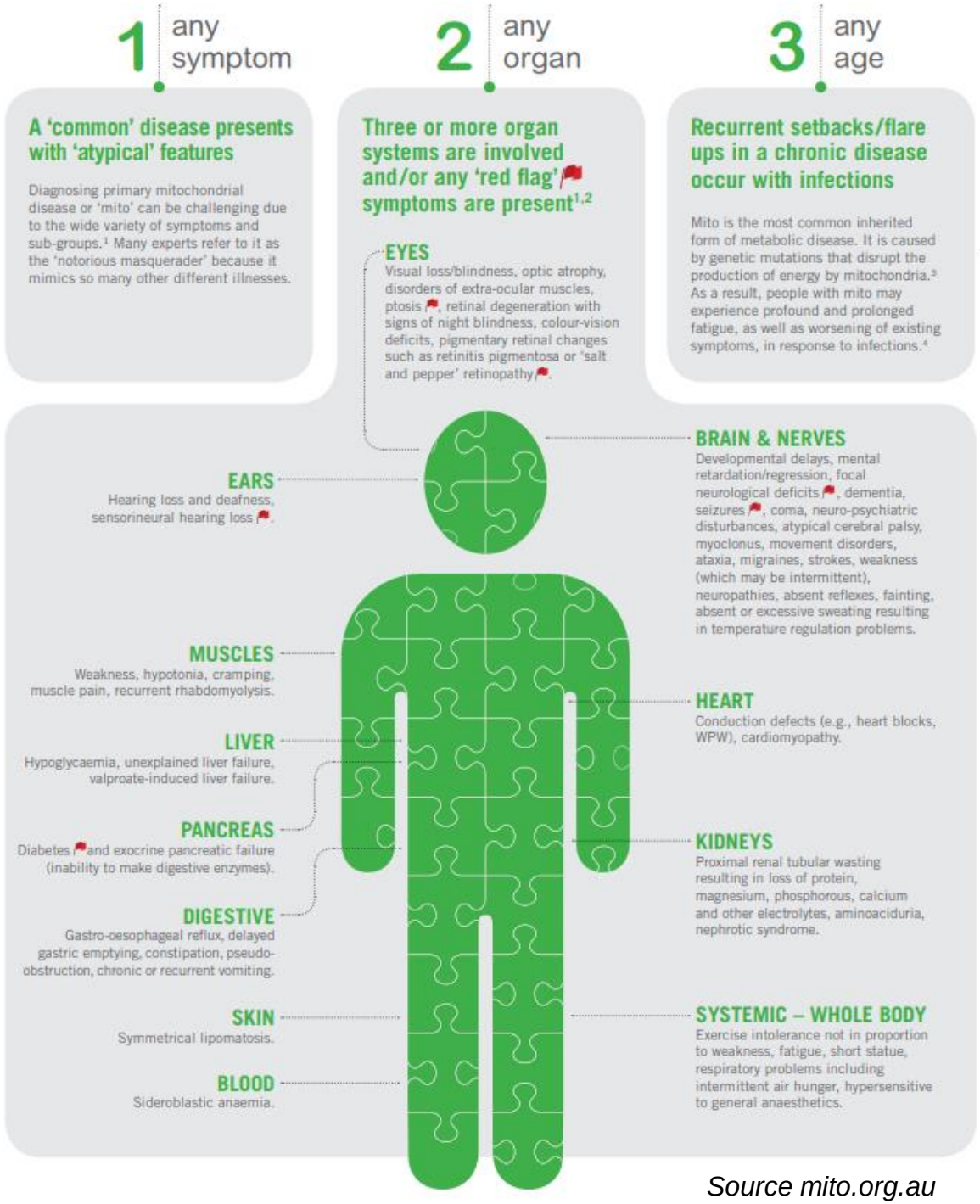
The aim of this presentation is to provide food for thought around resilience in the face of rare conditions.



Source mito.org.au

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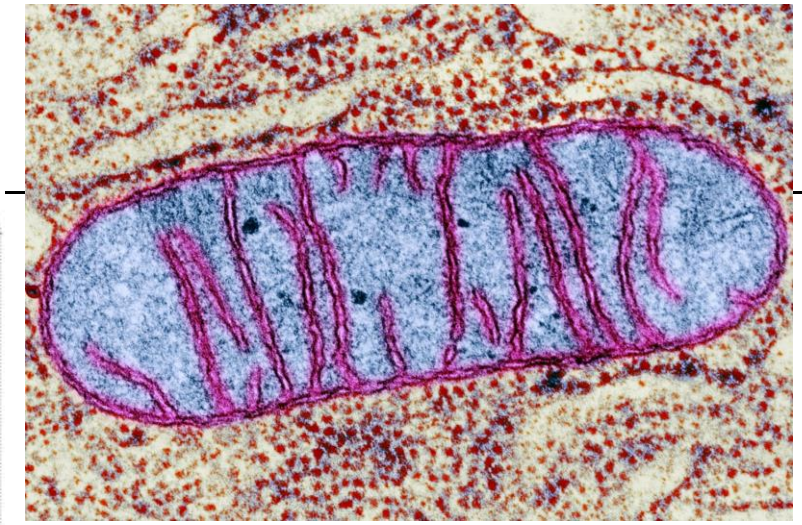

- Mitochondrial Respiratory Chain Disorders
- “Mito”
- 350 different disorders
- Incidence 1 in 5,000 births
- Infant cases severe - survival rate of 12 years
- Adult cases milder chronic phenotypes
- Can affect any organ, with highly variable clinical presentations, usually involving multiple systems

After diagnosis

ACTIVE IMPLEMENTATION OF NEW AUSTRALIAN CONSENSUS GUIDELINES FOR EFFECTIVE DELIVERY OF CLINICAL SERVICES FOR PATIENTS WITH MITOCHONDRIAL DISORDERS

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REVIEW

Genetics

Patient care standards for primary mitocho
disease: a consensus statement from th
Mitochondrial Medicine Society

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Ramona Salvarinova, MD, FRCPC[26], Russell P. Saneto, DO, PhD[27], Fernando S
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The purpose of this statement is to provide consensus-based recommendations for optimal management and care for patients with primary mitochondrial disease. This statement is intended for physicians who are engaged in the diagnosis and management of these patients. Working group members were appointed by the Mitochondrial Medicine Society. The panel included members with several different areas of expertise. The panel members utilized surveys and the Delphi method to reach consensus. We anticipate evolve. Consensus-based recommendatio routine care and management of patien mitochondrial disease.

Genet Med advance online publication 27

Key Words: care guidelines; consensus

INTERNAL MEDICINE JOURNAL

RACP Specialists. Together

doi:10.1111/imj.15505

POSITION PAPER

Patient care standards for primary mitochondrial disease in Australia: an Australian adaptation of the Mitochondrial Medicine Society recommendations

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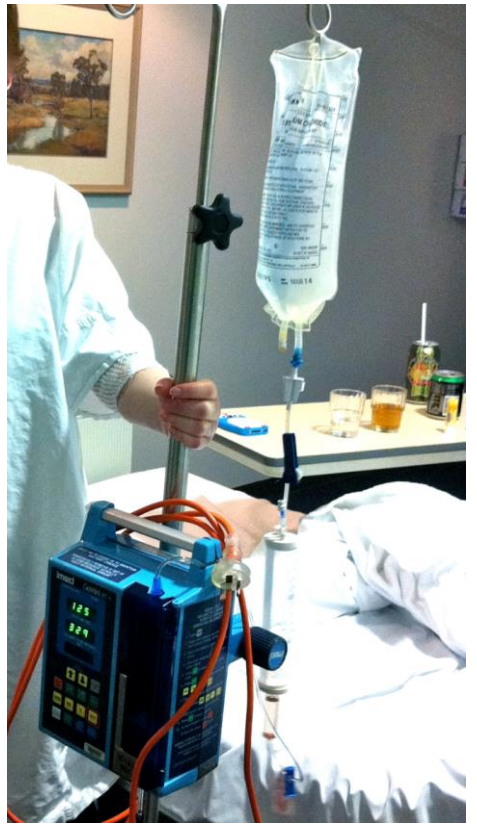
Project Aims

1. Understand the experience of management from the perspective of people living with a mito condition or parents of children with a mito condition.

SMH 2009

2. Understand current clinical contexts and practices from the mito specialists' perspective on diagnosis of a mito condition



Methods summary

UNDERSTANDING CURRENT PRACTICE AFTER DIAGNOSIS

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Consumers

- June–September 2020
- Two short surveys
- Online focus groups
- 20 participants
- 5 states
- 14 adults with mito (3 also had children with mito)
- 6 parents of children with mito
- 5 participants from regional areas
- Experience of the health system

Mito specialist clinicians

- 10 specialist mito services covering all of Australia
- Mapping current practice
- Interviews with clinical leads and teams
- Surveys

Take home messages

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CONSUMERS

"The most striking theme to emerge from the focus groups was the ongoing need to negotiate for every aspect of care."

I've actually struggled to find a GP that is willing to take on mito. I've had two doctors turn around and say we're actually not interested in that. We'll just manage Johnny's other issues. [Parent]

You have to do so much ringing around to find [a physio] that's even got an inkling of what the problem is. [Adult]

Long et al. Orphanet J Rare Dis (2021) 16:310
https://doi.org/10.1186/s13023-021-01939-6

Orphanet Journal of Rare Diseases

RESEARCH

Open Access

The long and winding road: perspectives of people and parents of children with mitochondrial conditions negotiating management after diagnosis

Janet C. Long[1*], Stephanie Best[1,2], Sarah Hatem[1], Tahlia Theodorou[1], Toni Catton[3], Sean Murray[3], Jeffrey Braithwaite[1] and John Christodoulou[4]

Abstract

Background: The diagnostic odyssey for people with a rare disease is well known, but difficulties do not stop at diagnosis. Here we investigate the experience of people, or parents of children with a diagnosed mitochondrial respiratory chain disorder (MRCD) in the management of their disease. The work complements ongoing projects around implementation of consensus recommendations for management of people with MRCD. People with or caring for a child with a formally diagnosed MRCD were invited to take part in an hour-long focus group held via videoconference. Questions elicited experiences of receiving management advice or information specific to their MRCD in four areas drawn from the consensus recommendations: diet and supplements, exercise, access to social services, and mental health. Sessions were audio-recorded, transcribed and analysed using a combination of inductive and deductive coding.

Results: Focus groups were conducted with 20 participants from five Australian states in June–September 2020.

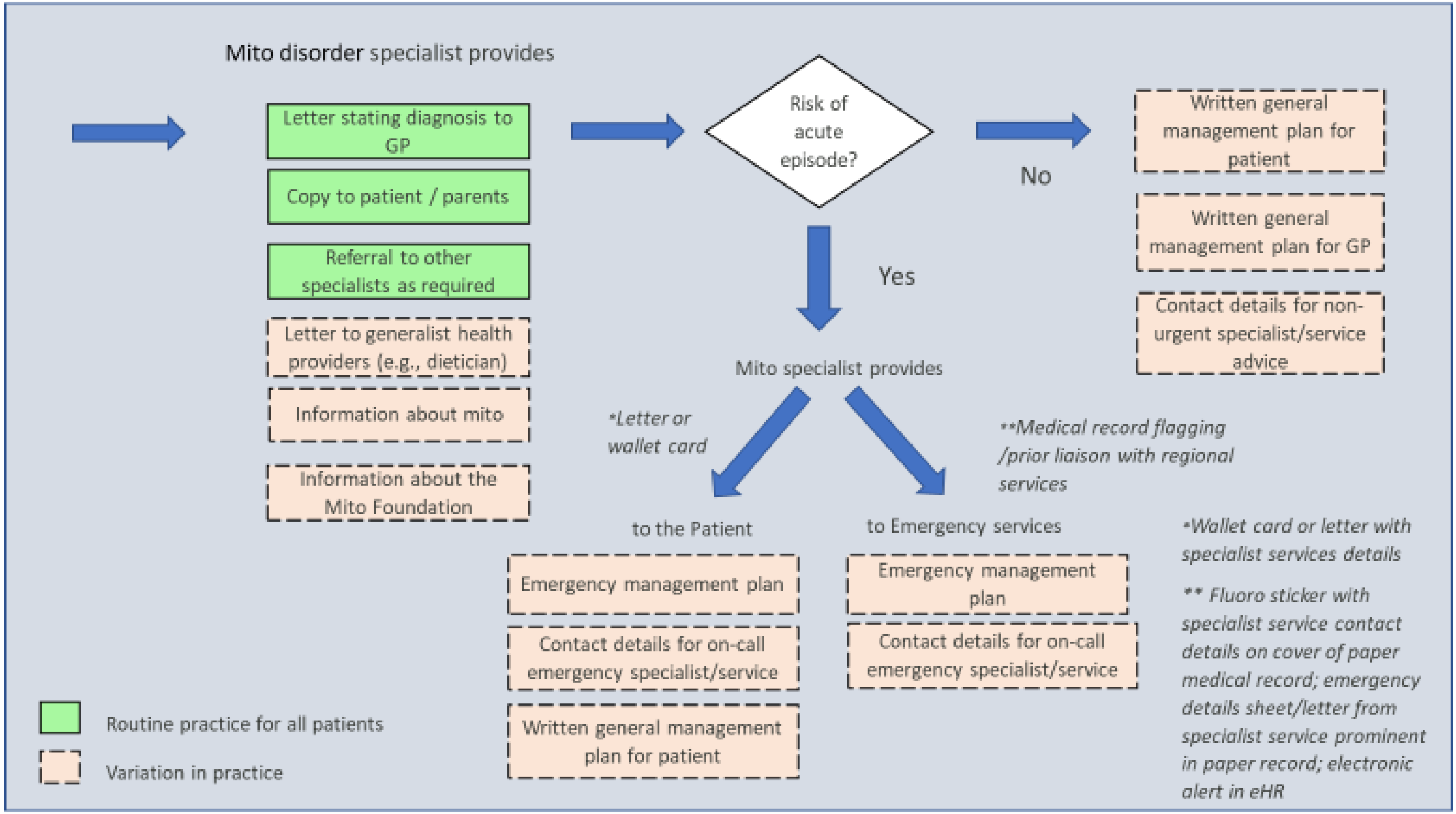
Take home messages

MITO SPECIALIST CLINICIANS

"While high quality management was described within the specialist services, elements that were described by patients as being most helpful for ongoing care were often missing."

You can put an emergency warning within the emergency department so you can have a flag come up when a person presents to triage. *[Mito Specialist 3]*

[My child] had multiple strokes before she died, and it was only after a dozen strokes, they put a red flag on her name at the hospital ED and triage nurse. Previously, it took hours to try and get action. *[Parent/GP]*


Mito disorder specialist provides
Letter stating diagnosis to GP
Copy to patient / parents
Referral to other specialists as required
Letter to generalist health providers (e.g., dietician)
Information about mito
Information about the Mito Foundation
Risk of acute episode?
No
Written general management plan for patient
Written general management plan for GP
Contact details for non-urgent specialist/service advice
Yes
Mito specialist provides
*Letter or wallet card
**Medical record flagging /prior liaison with regional services
to the Patient
Emergency management plan
Contact details for on-call emergency specialist/service
Written general management plan for patient
to Emergency services
Emergency management plan
Contact details for on-call emergency specialist/service
Routine practice for all patients
Variation in practice
*Wallet card or letter with specialist services details
** Fluoro sticker with specialist service contact details on cover of paper medical record; emergency details sheet/letter from specialist service prominent in paper record; electronic alert in eHR

Barriers to resilience

COMMUNICATION ISSUES: SYSTEMS ARE THERE … BUT NOT USED

I personally drilled my GP to make sure everything we do she uploads onto the master database that doctors and everybody can access. She does her bit, but then I go to the next specialist, or the next mito meeting and they ask me the questions which have already been answered had they taken the time to read this stuff. *[Adult]*

…with the GP and everything I've had to tell them read the last letter [from Mito specialist] ... and then they say yeah, I need to read that. Like it's not an instant click that there's something else they need to look at. Same with the specialist. You have to constantly remind them about it. Don't forget about that, don't forget about that. *[Adult]*

Barriers to resilience

COMMUNICATION ISSUES: SYSTEMS ARE THERE … BUT NOT USED

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The [regional] hospital locally didn't know what to do with my daughter so that was a stress, because they knew nothing about it so trying to get them to communicate… I would say "please call Sydney for advice" and they would say to me "no we can see what's coming on; we can manage this" to the point where I would actually pick her up and take her home out of the hospital because I was afraid they would kill her. [Parent, doctor]

Patients have an emergency wallet card that has info saying they have a rare disease, "I'm a patient at St Elsewhere's Hospital, the hospital number, and depending on their condition they might have 5 line summary of the emergency plan on the back e.g., investigate for stroke, treat like stroke as normal, also give arginine and please call us. [Mito specialist 1]

[My child] had to go in for tonsils and I said to them "please contact the [mito specialist] team regarding anaesthesia and everything else." "Nah, nah" they said. "Don't worry—he'll be fine." Well, he wasn't fine. [Parent]

Barriers to resilience

ACCESS TO INFORMATION AND ADVICE

We didn't get any guidance or anything. Just researched a lot through the Mito Foundation and other support groups that came up about exercise.

The GPs, I'm going to say, are almost useless. We've been turned away several times. We'll go to the GP for little things like - I know metabolically he's pretty stable, I've had him for 7 years, so I know what unstable looks like. I need to know whether he's got an ear infection or a UTI. And they'll be like "no you have to go to the emergency."
[Parent]

Barriers to resilience

THE VALUE OF THAT DIAGNOSIS LETTER…

I actually have a folder that I literally carry around to every appointment that I have with me, which is why I was able to convince the gastroenterologist last week that I didn't have myasthenia gravis. [Adult]

The hardest thing is getting your GP to understand that it's mito and not something else… [Adult]

Yes, so now whenever [we] go to any other specialist I just give them that [letter of diagnosis]. It's not coming from a neurotic person... you know that is from the [endocrinologist]... and it has the diagnosis, and this is the thing - it helps so much having that letter. [Parent]

Questions

HOW CAN WE IMPROVE SYSTEM RESILIENCE FOR PEOPLE WITH RARE DISEASES?

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- Are there systems and processes in place to support communication and management of people with rare diseases in your setting?
- Is it a bad thing to compartmentalise the rare disease – specialist can look after the Mito and GP look after the rest?
- Patient empowerment …. Should a patient have to advocate for their care?
- Is over-reliance on guidelines (for high prevalence conditions) detrimental here? What is the alternative?

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