Medicare Benefits Schedule (MBS) Review Advisory Committee (MRAC).
c/- Australian Government Department of Health
MDP 960
GPO Box 9848
Canberra ACT 2601

7 October 2022

Dear Committee members,

Re: Medicare Benefits Schedule (MBS) Review Advisory Committee (MRAC) - Genetic Counselling Working Group Draft Final Report

Thank you for inviting public comment on this report.

The aim of this submission is to provide input into the decision making process on behalf of the Australian mitochondrial disease (mito) community. Mito Foundation supports policies that improve access for the mito community to genetic counsellors. We believe that creating MBS items for genetic counselling can play an important role in improving access, particularly by making genetic counsellors available in a wider variety of settings.

Background

Mito Foundation supports patients with mito and their families, funds essential research into the prevention, diagnosis, treatment and cures of mito, and increases awareness and education about this devastating disease. Our work is informed by our Mito Community Advisory Panel, regular engagement with the wider mito community through support services and through research projects.

Although the need for expert genetic counselling is significant, the mito community can struggle to access services

Mito is a relatively complex genetic condition, caused by mutations in at least 338 genes in both nuclear and mitochondrial DNA(1). Testing is often inconclusive. Almost any type of inheritance can be involved, including de novo mutations. People can have different symptoms and severity of symptoms even with the same gene mutation. Multiple gene mutations can contribute to symptoms. All of these factors make test results particularly difficult to interpret and the expertise of a genetic counsellor particularly valuable.

The diagnostic odyssey for mito, like many rare genetic diseases, is often long and complex(2). Genetic counsellors can: support informed decisions on whether genetic testing will be beneficial; help people understand the type of genetic testing being considered; and support people to adjust to a diagnosis of mito.

Even though there is significant need for expert genetic counselling, mito community members report to Mito Foundation several challenges with accessing services:

- Reluctance from their managing clinicians to refer to hospital based clinical genomics services
- Referrals being rejected as outside the scope of the service
- Lack of genetic counsellors within mito specialist teams
- Long waiting lists for hospital based services

The report finds that waiting times are up to nine months, with variation between services. These waiting periods and even significantly longer are regularly reported to us by the mito community. We also expect that long wait times discourage referrals and contribute to tightening of service eligibility criteria.

Genomics-led diagnosis of mito and emerging reproductive technologies will drive increased need for genetic counselling services

Mito Foundation agrees that demand for genetic counselling is likely to increase. There are several key drivers that will increase the need for genetic counselling by the Australian mito community:

- There is a move towards a genomics first approach to diagnosing mito(3). Medicare funding of genomic testing (if MSAC application *1675 – Whole Genome Sequencing for the diagnosis of mitochondrial disease* is supported) will accelerate the implementation of this in Australia.
- The *Mitochondrial Donation Law Reform (Maeve's Law) Bill 2021* was passed in the Australian Senate in March 2022 and it is expected that the MRFF funded pilot stage will commence in 2023. Having a confirmed mitochondrial DNA genetic cause of mito is a key eligibility criterion for mitochondrial donation.
- The increasing awareness of the value of pre-implantation genetic diagnosis for women with mitochondrial DNA changes.
- The increasing use of reproductive carrier screening in families with and without a family history of mito.

We support collaborative approaches between the states and the Commonwealth to ensure genetic counselling services are available to meet these needs.

Service delivery in multiple settings will benefit the mito community

Creating MBS items for genetic counselling will incentivise genetic counsellors to provide services outside of public clinical genomics services. The introduction of genetic medicine into other medical specialities, including neurology, cardiology, ophthalmology, infertility, and many other fields, has been made possible in large part by genetic counsellors (3). Physicians with inadequate genetics knowledge in each of these scenarios gain from working with genetics experts.

A neurologist is often the key specialist for a person with mito, with one US based study finding more than half of diagnoses were delivered by a neurologist(2), with less than 20% by a clinical geneticist. We know that many Australian adults with mito see their neurologist as a private patient; 38% of respondents to Mito Foundation's 2022 community survey told us they mainly see their health professionals in a private hospital

or practice (unpublished data). Ophthalmology is another key speciality frequently involved in making a mito diagnosis with known genetic causes. The possibility of pathways that integrate care between a genetic counsellor and privately practicing neurologists, ophthalmologists and other specialists would be welcomed by many in the mito community.

Mito Foundation would also welcome policies that increase access to expert genetic counselling in the context of using assisted reproductive technologies. While specialist nurses often provide genetic counselling within fertility clinics, better integration with genetic counsellors would ensure that services are confident to support families with complex conditions like mito.

We support a multi-pronged response to the challenge of providing expert genetic counselling to the mito community

We see a need for cooperative work between the states and Commonwealth to improve access. Mainstreaming of genetic counselling skills is an important part of this solution, with training for general practitioners, nurses and medical specialists playing a key role in their ability to support and guide families, such as through the reproductive genetic screening process.

Creating MBS items for genetic counselling will enhance existing services. Work to increase the services offered in public hospitals through block and/or activity-based funding is also required to meet demand and patient preferences, but should not be an isolated solution.

While we appreciate the workforce challenges in this reform, we agree with HGSA's position that existing training programs are able to meet the projected demand over time. We see this as an implementation challenge and not a reason to reject the proposal.

Further information

Please do not hesitate to contact us for further discussion regarding this submission.

Yours sincerely,

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References

1. Stenton SL, Prokisch H. Genetics of mitochondrial diseases: Identifying mutations to help diagnosis. EBioMedicine. 2020 Jun;56:102784.

2. Grier J, Hirano M, Karaa A, Shepard E, Thompson JLP. Diagnostic odyssey of patients with mitochondrial disease: Results of a survey. Neurol Genet. 2018 Apr;4(2):e230.

3. Watson E, Davis R, Sue CM. New diagnostic pathways for mitochondrial disease. J Transl Genet Genomics [Internet]. 2020 [cited 2022 Oct 7]; Available from: https://jtggjournal.com/article/view/3509