Australian Government

Department of Health

Targeted Consultation Survey on MSAC Application 1476- Genetic testing for childhood syndromes

(Version 0.1)

This feedback survey relates to the application form for new and amended requests for public funding (including but not limited to the Medicare Benefits Schedule (MBS)).

Please use this template, to prepare your feedback on the application form. You are welcome to provide feedback from either a personal or group perspective for consideration by the Department of Health when the application form is being reviewed.

The data collected will be used to inform the Medical Services Advisory Committee (MSAC) process to ensure that when proposed healthcare interventions are assessed for public funding in Australia, they are patient focused and seek to achieve best value.

This feedback survey should take approximately 15 minutes to complete.

You may also wish to supplement your responses with further documentation or diagrams or other information to assist the Department in considering your feedback.

Thank you for taking the time to provide valuable feedback.

Responses may be provided to the MSAC, its subcommittees, a health technology assessment group and the applicant. Should you require de-identification please contact the HTA team (details below).

While stakeholder feedback is used to inform the application process, you should be aware that your feedback may be used more broadly by the applicant.

Please reply to the HTA Team:
Email: HTA@health.gov.au
Postal: MDP 959 GPO 9848 ACT 2601
Phone: 02 6289 7550

PART 1 – PERSONAL AND ORGANISATIONAL INFORMATION

1. Respondent details

Name:	Sean Murray
Email:	Sean.Murray@amdf.org.au
Phone No:	(02) 8033 4113

2. (a) Is the feedback being provided on an individual basis or by a collective group? (please select)

Individual	
Collective Group	X

(b) If individual, specify the name of the organisation you work for

(c) If collective group, specify the name of the group

Australian Mitochondrial Disease Foundation (AMDF)

3. How would you best identify yourself?

General practitioner	
Specialist	
Researcher	
Consumer	
Care giver	
Other	X

(a) If other, please specify

AMDF is the patient representative organisation for Australians affected by mitochondrial disease (mito), providing support to patients whilst seeking a cure.

PART 2 – CLINICAL NEED AND PUBLIC HEALTH SIGNIFICANCE

4. Describe your experience with the medical condition (disease) and/or proposed intervention and/or service relating to the application form

AMDF provides support to patients and families affected by mito. This includes speaking extensively to families who are on the diagnostic odyssey. This is a time of uncertainty, anxiety and isolation for families as they are left in the dark about prognosis and what the future may hold for them.
AMDF has sought input from its Scientific and Medical Advisory Panel and Mito Community Advisory Panel in completing this consultation survey.

5. What do you see as the benefit(s) of the proposed medical service, in particular for the person involved and/or their family and carers?

The key benefit of the service is the reduction in time for families between clinical presentation and receiving a formal genetic diagnosis. Securing a timely diagnosis can ensure patients receive a coordinated healthcare approach and provides families a sense of relief.

A formal genetic diagnosis ensures children and their families:

- Have timely access to treatments and symptoms management pathways to alter the progression of the disease, and avoid harmful treatments

- Can access financial and other relevant support, and assists families in planning for the future

- Can make informed decisions about reproduction and accessing reproductive technologies, and has the potential to restore reproductive confidence in families

- Have the opportunity to participate in research and access treatments through clinical trials

6. What do you see as the disadvantage(s) of the proposed medical service, in particular for the person involved and/or their family and carers?

AMDF recognises that approval of this application will reduce the time from clinical presentation to a formal diagnosis for patients compared to the current diagnostic pathway. It is essential that the service is adequately funded to prevent backlogs of patients and thus long waiting periods. AMDF acknowledges that the applicants suggest that the current wait times of 8-12 weeks will be reduced through provision of adequate funding.
Even in the absence of a diagnosis, the proposed testing is unlikely to cause harm to patients and families in comparison to standard care.

7. What other benefits can you see from having this intervention publically funded on the Medicare Benefits Schedule (MBS)?

8. What other services do you believe need to be delivered before or after this intervention, eg Dietician, Pathology etc?

It is essential that patients and their families receive access to genetic counselling prior to testing and once diagnosis has been confirmed. There will need to be genetic counselling provided for extended family members who may themselves be at risk or at risk of passing on mito to their future offspring. As per the application, paediatric sub-specialists and clinical geneticists would be required to discuss eligible patients with a multi-disciplinary review team to ensure patient suitability for WEA.

PART 3 – INDICATION(S) FOR THE PROPOSED MEDICAL SERVICE AND CLINICAL CLAIM

9. Do you agree or disagree with the proposed population(s) for the proposed medical service as specified in Part 6a of the application form?

Strongly Agree	X
Agree	
Disagree	
Strongly Disagree	

(a) Specify why or why not:

AMDF strongly supports genetic testing for all patients with a clinical presentation of mitochondrial disease for a confirmed genetic diagnosis. AMDF acknowledges that at this point in time, there is only evidence available for the cost effectiveness of WEA testing in children, as per the application.

10. Have all the associated interventions been adequately captured in Part 6b of the application form?

Yes	X
No	

(b) Please explain:

11. Do you agree or disagree that the comparator(s) to the proposed medical service as specified in Part 6c of the application form?

Strongly Agree	X
Agree	
Disagree	
Strongly Disagree	

(a) Specify why or why not:

As per the application, "There is no direct comparator to the whole exome sequencing diagnostic test proposed". Therefore, AMDF strongly agrees with the application using current standard care as the comparator in the absence of a diagnosis.

12. Do you agree or disagree with the clinical claim made for the proposed medical service as specified in Part 6d of the application form?

Strongly Agree	X
Agree	
Disagree	
Strongly Disagree	

(b) Specify why or why not:

AMDF strongly agrees with the clinical process outlined in part 6b of the application, as identified by medical experts and researchers in the area.

PART 4 – COST INFORMATION FOR THE PROPOSED MEDICAL SERVICE

13. Do you agree with the proposed MBS item descriptor, as specified in Question 53 of the application form?

Strongly Agree	X
Agree	
Disagree	
Strongly Disagree	

(c) Specify why or why not:

AMDF agrees with information around costs and time required for services as outlined in Question 53 by medical experts and researchers.

14. Do you agree or disagree with the proposed MBS fee, as specified in Question 53 of the application form?

Strongly Agree	X
Agree	
Disagree	
Strongly Disagree	

(d) Specify why or why not:

AMDF agrees with the proposed MBS fee as specified on the application. AMDF supports minimising out-of-pocket costs for patients in accessing WEA testing as is necessary for obtaining a genetic diagnosis and minimising the diagnostic odyssey for patients.

PART 5 – ADDITIONAL COMMENTS

15. **Do you have any additional comments on the proposed intervention and/or medical condition (disease) relating to the proposed medical service?**

Please see attached letter of support from AMDF's CEO, on behalf of AMDF.

16. **Do you have any comments on this feedback survey? Please provide comments or suggestions on how this process could be improved.**

Again, thank you for taking the time to provide valuable feedback.