Australian Government

Department of Health

Consultation Survey on MSAC Application 1680

Genetic testing for childhood hearing impairment

MSAC welcomes feedback on MSAC applications for public funding from individuals, organisations representing health professionals or consumers and/or carers, and from other stakeholders. Please use this template to prepare your feedback. You may also attach additional information if you consider it may be useful in informing MSAC and its sub-committees.

Sharing consultation feedback

Submitted consultation feedback will be shared with the Applicant and with MSAC and its sub-committees.

- The applicant will receive a summary of comments from individuals, with the individual's name and other identifying information removed.
- MSAC and its sub-committees will receive both the summary and copies of the comments, with the name of the individual and other identifying information removed.
- Consultation feedback from groups or organisations will be provided in a complete form to both the Applicant and to MSAC and its sub-committees.

Please do not include information in your feedback that you do not want shared as outlined above. In addition, to protect privacy, do not include identifying personal (e.g. name) or sensitive (e.g. medical history) information about third parties, such as medical professionals or friends/relatives.

How consultation feedback is used

MSAC and its sub-committees consider consultation feedback when appraising an application, including to better understand the potential impact of the proposed medical technology/service on consumers, carers, and health professionals. A summary of consultation feedback will be included in the Public Summary Document (PSD) published on the MSAC website once MSAC has completed its appraisal. The PSD may also cite feedback from groups/organisations, including the name of the organisation. As such, organisations should not include information or opinions in their feedback that they would not wish to see in the public domain.

Consultation deadlines. Please ensure that feedback is submitted by the pre-PASC or pre-MSAC consultation deadline for this application. Consultation deadlines for each PASC and MSAC meeting are listed in the PASC and MSAC and ESC calendars available on the MSAC website. They are also published in the MSAC Bulletin. Feedback received after the respective deadlines may not be considered.

For further information on the MSAC consultation process please refer to the MSAC Website or contact the Consumer Evidence and Engagement Unit on email: commentsMSAC@health.gov.au.
Thank you for taking the time to provide your feedback. Please return your completed survey to:

Email: commentsMSAC@health.gov.au

Mail: MSAC Secretariat,
MDP 960, GPO Box 9848,
ACT 2601

PART 1 – PERSONAL AND ORGANISATIONAL INFORMATION

1. Respondent details

Name: Matilda Haas

Email: matilda.haas@mcri.edu.au

Phone No: 0403287727

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

☐ Individual
☒ Collective Group

If an individual, specify the name of the organisation you work for

If a collective group, specify the name of the group

Australian Genomics

3. How would you best identify yourself?

☐ General Practitioner
☐ Specialist
☐ Researcher
☐ Consumer
☐ Care giver
☒ Other

If other, please specify

Research Projects and Partnership Manager submitting a response on behalf of Australian Genomics in consultation with the network of clinicians, researchers, and diagnosticians.

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

Australian Genomics is an Australian Government initiative supporting genomic research and its translation into clinical practice. Through broad engagement and a national collaborative approach, it achieves two key objectives: to improve efficiency, reach and timeliness of genomic research projects, and to support Commonwealth State and Territory health departments in the implementation of genomics research outcomes by refining and communicating evidence to inform policy development.

Australian Genomics engages with current and emerging government policy and priorities to identify gaps and opportunities, to support policy and action for integrating genomic technologies into the health system. By interfacing with consumers, governments, industry and global genomics initiatives, Australian Genomics drives change and growth in the sector.

Australian Genomics maintains an informed position in relation to current translational genomic research programs currently underway in Australia, and their progression toward implementation. Hearing loss or impairment in infants has been an area under investigation and has been considered for incorporation under different pathways: as part of the newborn hearing test, as discussed in this application, as part of a broader newborn genomic testing-based screening program, and as part of a reproductive carrier screening program.

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 medical service outlined in the application is a diagnostic test that the applicants indicate will increase the rate of genetic diagnosis in hearing impairment in young people from 20% to 60%. An early diagnosis of the cause of hearing impairment would, in some cases, lead to avoidance of other tests, surveillance strategies, alter use of services, streamline care and interventions, and would be predictive of future health problems. The information could also be used to maximise a child's developmental potential. The applicants also suggest that such a diagnosis may inform appropriate access to precision therapies that are in development. A diagnosis may also inform reproductive decision-making.

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?

It is interesting to note than in a cohort research study, 60% families took up the opportunity to undergo sequencing to find a genetic diagnosis for hearing impairment. This figure may be lower in a research setting than in a standard of care setting, but it would seem important to understand consumer motivations toward such a test, and factor this into service design, as well as calculations about uptake of the test.

7. What other benefits can you see from having this intervention publically funded?

One of the main benefits is that this test is an additional intervention that can be combined with the newborn hearing screening test, which already reaches up to 98% of newborns in Australia. Thus, if it is integrated as standard of care as a follow-on test, it could further improve outcomes of the hearing test across the full cohort for which hearing loss is detected, in an equitable way.

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

Genetic counselling services would be needed for pre-test counselling and post-test result return. If test uptake rates are low (in a research setting) then this could indicate need for improved understanding of the potential outcomes, risks and benefits of the test, all of which can be covered by a genetic counsellor at the time of providing consent.

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
- ☐ Agree
- ☒ Disagree
- ☐ Strongly Disagree

Specify why or why not:

In response to Q24 the applicants say that "A more complex genetic syndrome is identifiable in 20% children detected through newborn hearing screening" – have the applicants determine what proportion of this 20% would be eligible for testing under MBS item number 73358 (whole exome analysis for a suspected monogenic condition)?

The flow chart and appendix 1 referred to in this section are missing from our version of the application form, making it difficult to consider this question and which is why we have selected "disagree" from the options above.

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

- ☒ Yes
- ☐ No

Please explain:

It would be good for the applicants to clarify in response to Q27 whether this intervention is targeted only toward patients with non-syndromic hearing loss.

It is also unclear about the relevance of the cochlear implant in this cohort: is it an early intervention that could be applied with a positive diagnosis, or is that instead determined by physiological / auditory metrics? Given the impact on neurological /social development, presumably guiding the appropriate identification of recipients of this early intervention would be critical.

The applicant suggests the test could be ordered by "A paediatrician with expertise in managing children with hearing loss" and that paediatricians will be required to do basic training in genetic counselling – who provides that training and accreditation/micro credentialling will need to be determined if the test is recommended for funding.

This section also describes the importance of genetic counselling for post-test result return, but not for pre-test counselling. Not all paediatricians would necessarily be appropriately qualified to engage in genetic counselling with a family. Engaging the services of genetic counsellors is cost-effective and also maximises the efficiency of specialist and multidisciplinary clinics. Genetic counsellors also manage patient pre-consultation preparation.

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
- ☒ Agree
- ☐ Disagree
- ☐ Strongly Disagree

Please explain:

As a note, given that the applicants report that the deafness panel offered commercially outside of Australia has a slightly higher diagnostic rate than WEA at this time, it would be good to have been presented with greater detail about why the proposed approach is the best option.

It is also slightly unclear which investigations (eg MRI, ECG, CMV testing, audiograms) would be able to be avoided and in how many cases, and how that information can be accurately incorporated into the comparator.

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
- ☒ Agree
- ☐ Disagree
- ☐ Strongly Disagree

Specify why or why not:

The clinical claim is that the test will increase genetic diagnoses from 20% to 60%.

Although referred to in response to other questions in the application, clinical effectiveness outcomes in this question response do not list any change in clinical management, or mention access to different treatment including precision therapies. The main clinical claim, therefore, is release from surveillance and other investigations, as well as personal utility for families.

PART 4 – COST INFORMATION FOR THE PROPOSED MEDICAL SERVICE

13. **Do you agree with the proposed service descriptor?** MSAC is transitioning to new application forms so the relevant question in the application form will vary depending on the version used. For medical services on the MBS, see question 51 or 53. For medical services seeking funding from a source other than the MBS, see question 52 (new application forms only—labelled v. 2.5).

- ☐ Strongly Agree
- ☒ Agree
- ☐ Disagree
- ☐ Strongly Disagree

Specify why or why not:

The re-analysis service is proposed for every 1-2 years – perhaps it would be best to revise this time frame to 18 months, in line with the MBS item number 73358 (whole exome analysis for a suspected monogenic disorder). Many upcoming MSAC applications will include a reanalysis item and developing a standard in the field will likely contribute to the reanalysis being performed at regular intervals.

The stated turnaround time for the test is 8 weeks, however, the turnaround time for other currently available panel tests such as a cardiac panel (also around 100 genes) is 12 weeks.

The item descriptor should be broken down further into independent proband, reanalysis and cascade testing of family members items. Also, whether copy number variant analysis is feasible to do from accredited WES should be determined in relation to this test.

14. **Do you agree with the proposed service fee?** MSAC is transitioning to new application forms, so the relevant question in the application form will vary depending on the version used. For medical services on the MBS, see question 51 or 53. For medical services seeking funding from a source other than the MBS, see question 52 (new application forms only—labelled v. 2.5).

- ☐ Strongly Agree
- ☒ Agree
- ☐ Disagree
- ☐ Strongly Disagree

Specify why or why not:

The application states that currently no other Australian laboratories offer specific analysis for this indication – this should be confirmed. Also, to ensure equitable access, a competitive market and good turnaround times, other laboratories should be given opportunity to develop the test and complete accreditation requirements.

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?

Of note is the good support from professional bodies and consumer organisations (response to Qs 19 and 21) for this medical service.

"The suggested diagnostic test is agnostic of technology, and hence it is not prescriptive to the methodologies/equipment and reagents involved" – this raises quality assurance issues in all applications that adopt this stance and MSAC will need to develop a position and strategy for ensuring safety and quality of agnostic medical test/services.

In response to Q8 the applicant selected the box "Identified a patient as suitable for therapy…" but it is unclear from the information in the rest of the application that this is the case? The clinical claim focuses the avoidance of other tests or release from surveillance.

In response to Q9 the applicant selected the box "no" in relation to whether another medical product can achieve or enhance its intended effect. It is unclear throughout the application whether any diagnosis achieved by this test would inform the suitability for a cochlea implant?

In Part 3, question 14 and other associated questions, the medical device or service is incorrectly classified – the applicants answered N/A but all human genetic tests are classed as TGA Class 3 IVDs https://www.tga.gov.au/sme-assist/medical-devices-regulation-introduction

In response to Q50, the applicants state that "Risk of leakage would be considered nil due to targeted testing of a well-defined population, restricted ordering to paediatricians with appropriate training or clinical geneticists". Is there a proposed method of determining appropriate training aligning with this item number if it is supported by MSAC?

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.