Evidence Summary

Australian Genomics Health Alliance

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Federal funding for clinical genomic tests

A key focus of Australian Genomics is to determine in what clinical situations genomic testing has demonstrable benefit, and then direct this evidence into the established systems for medical services review.

Our goal is to ensure Australians benefit from subsidised and effective diagnostic testing.

Background

Complex rare genetic disorders often present with overlapping clinical features and are genetically heterogeneous, making timely and accurate diagnosis challenging.

Broader genomic tests such as whole exome and whole genome sequencing have been shown to be both clinically effective and cost saving for certain genetic conditions [1, 2]. The clinical indications for which there is evidence to support the use of these tests will also inevitably increase.

In Australia, the independent non-statutory Medical Services Advisory Committee (MSAC) is one pathway for the assessment and recommendation for public funding of medical services.

Project aims

The aims of this project were two-fold:

- Secure an MSAC recommendation for subsidised access to genomic testing for the diagnosis of **childhood syndromes** and **intellectual disability** using genomic analysis.
- Use this initial case to inform the establishment of an Australian Genomics pipeline of funding applications to MSAC to enable greater access to genomic testing in clinical practice.

Our approach

The Australian Genomics application to MSAC was the first of its kind, seeking genomic analysis for childhood syndromes and intellectual disability, with a flexible clinically-driven gene list.

The application was supported by a growing body of international research indicating the clinical utility and cost effectiveness of genomic testing for these conditions, including the Melbourne Genomics Childhood Syndromes Cohort[2].

Once it was established that there was strong evidence from these studies to form an application to MSAC, a working group was established with national representation from the fields of clinical genetics, genetic counselling, diagnostics and pathologists.
This working group provided input and guidance into the initial application, which covered a recommended population, intervention, comparators and outcomes for this service.

While the Health Technology Assessment process was externally contracted by MSAC, there were key points where the Australian Genomics working group were able to review and provide further input to the 'Protocol, Intervention, Comparator, Outcome' (PICO) development and the assessment report development. See the process illustrated at Figure 1.

About MSAC

MSAC is an independently appointed committee, which provides advice to the Australian Government on whether a new service should be publicly funded, through the Medicare Benefits Scheme (MBS) or alternative mechanism.

Recommendation by MSAC follows an assessment of the medical services' comparative safety, clinical effectiveness, cost-effectiveness, and total cost, using the best available evidence.

This is a rigorous process that ensures Australians have access to medical services that have been shown to be safe and clinically effective, as well as representing value-for-money for both patients and taxpayers.

msac.gov.au

Members of the working group also participated in a stakeholder meeting organised by MSAC which was highly valuable in refining the application.

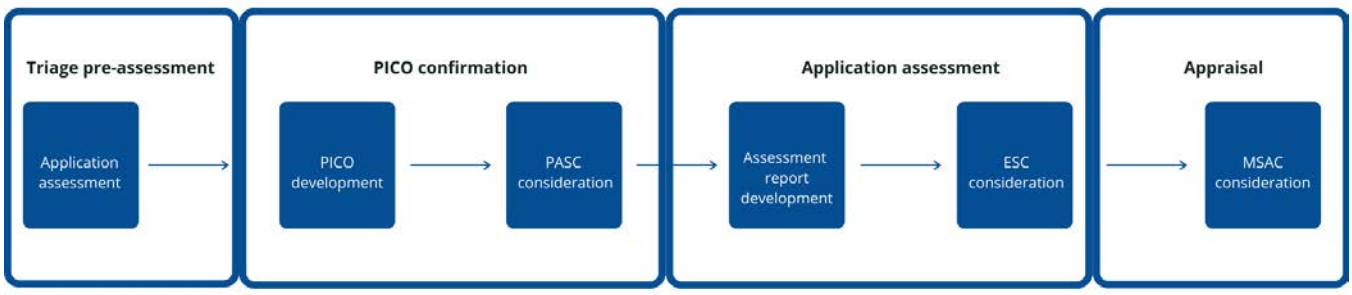

Figure 1. Adapted from MSAC Reform Implementation: Process Framework (March 2016)
PICO: Protocol, Intervention, Comparator, Outcomes; **PASC**: PICO Advisory Sub-committee;
ESC: Evaluation Sub-committee; MSAC Medical Services Advisory Committee.

Key Outcomes

In August 2019, MSAC recommended funding a modified version of the application led by Australian Genomics.

Genetic testing for childhood syndromes was subsequently approved for listing on the Medicare Benefits Schedule effective May 1, 2020. These items include:

- **73358:** Singleton testing of affected individuals with childhood syndromes and/or intellectual disability.
- **73359:** Trio testing of affected individuals, with an upper limit of $2,900 for this fee item.
- **73360:** Re-analysis of previously collected data in certain circumstances.
- **73361-3:** Cascade and segregation testing for certain purposes.

For exact wording and specifics on item numbers please use the MBS Online directory.

Details on item numbers can also be found in the *Health Insurance (Pathology Services Table) Regulations 2020*, made under the *Health Insurance Act 1973.*

Considerations for future applications

The application process for funding of genomic testing for childhood syndromes and/or intellectual disability has provided important insights for both how future genomic test applications may be assessed by MSAC, and for the broader provision of clinical genomics in Australia.

Our observations from this process include:

- In order to mainstream genomics, testing needs to be accessible through a broad range of clinicians, as appropriate for the clinical indication. In this recent recommendation, childhood syndromes and intellectual disability funded testing will be made available through specialist paediatricians as well as genetics services.
- Technology agnostic applications are important to future proof diagnostic tests for genetic conditions: whole exome analysis may be performed on whole exome or whole genome generated sequence.
- While a range of international and local evidence is used for evaluation and determining the advice of MSAC, local multi-site studies are often the strongest evidence to support an application.
- Clear articulation of the current standard of care compared to genomic testing in a particular clinical setting is important. This can be challenging in a fast moving field like genomics.
- Creating a common understanding of the technology and the clinical setting is key to progressing a complex application. Stakeholder meetings can assist in achieving this.

Implementation challenges

Funding for clinical genomics has largely been a State/Territory responsibility and services have primarily resided in public hospitals through genetics clinics. Patients' access to MBS funded testing under certain clinical models needs to be ensured to maintain equity.

Availability of reimbursement for genomic tests may also drive interest more broadly from the pathology sector to expand service offerings, creating benefits through competition where service delivery and quality remain high.

Furthermore, as genomics expands into non-genetic specialities, it is important to consider system capacity and availability of training to ensure appropriate item use and delivery of informed consent.

Review

While a great deal has been learned throughout the process of this initial application, there will also be benefit in the review of the use of the item numbers post implementation into pathology.

MSAC has recommended a review of the fees be carried out two years after listing on the MBS, alongside a broader review of the utilisation of the items overall, particularly looking at:

- Diagnostic yield for laboratories and requesters
- Who requests tests
- Out-of-pocket costs for patients

Impact

There are a number of levels where this project has and will continue to have significant impact:

- One of the main challenge's clinicians have faced in providing genomic testing to patients is the lack of sustainable funding for these tests. With the provision of an MBS item number, eligible patients across Australia will have access to funded testing. It is expected that around 3,000 Australian families will benefit each year.

- Other applications for broad scale genomic testing will also benefit from the increased genomic literacy of the committees involved in the review process (PASC, ESC and MSAC), as well as the public availability of prior application and summary documentation from these processes.

- Previously, genomic testing was the realm of clinical genetic departments. With the expanding relevance of genomic testing into other clinical specialities there will inevitably be an impact on the clinicians in these areas. Initially this may require further training, but ultimately it will mean better access to care for patients through a decentralised model for genomic testing.

Conclusion

Australian Genomics, along with other major stakeholders, will continue to advance research evidence into clinical genomics through the ongoing proposal of funding for genomic testing in areas where there is strong evidence for its use.

While sustainable funding for genomic tests is only one part of the delivery of clinical genomics, it is a necessary part if we are to see the full potential of what genomics can deliver for patients and Australian healthcare.

References

1. Stark Z, Schofield D, Alam K, et al. Prospective comparison of the cost-effectiveness of clinical whole-exome sequencing with that of usual care overwhelmingly supports early use and reimbursement. Genet Med. 2017;19(8):867-874. Available at: https://pubmed.ncbi.nlm.nih.gov/28125081/

2. Stark Z, Tan TY, Chong B, et al. A prospective evaluation of whole-exome sequencing as a first-tier molecular test in infants with suspected monogenic disorders. Genet Med. 2016;18(11):1090-1096. Available at: https://pubmed.ncbi.nlm.nih.gov/26938784/

Acknowledgments

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