Next Steps: Navigating Funding Pathways for Genomic Research Translation

April 2025

Australian Genomics

Contents

Acknowledgement of Country

In the spirit of reconciliation Australian Genomics acknowledges the Traditional Custodians of country throughout Australia and their connections to land, sea, and community.

We pay our respect to their elders past and present and extend that respect to all Aboriginal and Torres Strait Islander peoples today.

Artwork by Yorta Yorta artist, Alkina Edwards, for Australian Genomics.

Abbreviations

ABF	Activity based funding
ACE	Australian Classification Exchange
ADAR	Applicant developed assessment report
AIMD	Active Implantable Medical Device
AR-DRGs	Australian Refined Diagnosis Related Groups
CAC	Clinical Advisory Committee
CAPS	Cancer and Population Screening
CEO	Chief executive officer
CHS	Canberra Health Services
ctDNA	Circulating tumour DNA
CUC	Clinical utility card
DCAR	Department contracted assessment report
DTC	Direct to consumer
ESC	Evaluation Sub-Committee
GHFM	Genomics Health Futures Mission
GP	General practitioner
HCEF	Health Chief Executives Forum
HTA	Health technology assessment
HTAC	Health Technology Advisory Committee
HTGC	Health Technology and Genomics Collaboration
HTI	Health Technology and Innovation
IHACPA	Independent Health and Aged Care Pricing Authority
JAC	Jurisdictional Advisory Committee
LHN	Local health network
MBS	Medicare Benefits Schedule
MCRI	Murdoch Children's Research Institute
MDHTAC	Medical Device and Human Tissue Advisory Committee
MPS	Massively parallel sequencing
MRFF	Medical Research Future Fund
MSAC	Medical Services Advisory Committee
NATA	National Association of Testing Authorities
NDTIAC	New Device Technology and Interventions Approval Committee
NEC	National efficient cost
NHRA	National Health Reform Agreement
NICU	Newborn intensive care unit
NPAAC	National Pathology Accreditation Advisory Council
PASC	PICO Advisory Sub-Committee
PBAC	Pharmaceutical Benefits Advisory Committee
PBS	Pharmaceutical Benefits Scheme
PICO	Population, intervention, comparator and outcomes
PICU	Paediatric intensive care unit
PLAC	Prostheses List Advisory Committee
PSD	Public summary document
QH	Queensland Health
QTFF	Queensland Technology Future Fund
RCH	Royal Children's Hospital
RCPA	Royal College of Pathologists Australia
RDNow	Rare Diseases Now
SAPACT	South Australian Policy Advisory Committee on Technology
SAS	Special Access Scheme
TGA	Therapeutic Goods Administration
VCGS	Victorian Clinical Genetics Service
WAPACT	Western Australia Policy Advisory Committee on Technology
WEA	Whole exome analysis
WES	Whole exome sequencing
WGS	Whole genome sequencing
ZERO	Zero Childhood Cancer Program

Introduction

With the increasing affordability and uptake of genomic sequencing, the direct impacts of human genomics research on healthcare are being realised on a global scale.[1, 2] Genomic technologies, which have been described as disruptive, have the potential to transform the diagnosis, treatment, management, and prevention of health conditions.[3]

Australia is home to internationally renowned genomics researchers, and there have been significant investments in human genomics research. For example, the Australian Government is investing $500.1 million in health genomics research over 10 years through the Genomics Health Futures Mission (GHFM) under the Medical Research Future Fund (MRFF), with a mid-term review completed in 2024 to set the direction of future investment in health genomics research.[4] Research outcomes will have great potential for uptake into the healthcare system, across priority areas such as rare and complex conditions, cancer, pharmacogenomics, governance and technology, and Aboriginal and Torres Strait Islander health.

While genomic technologies are already being integrated into clinical care, standardised pathways and processes for translating genomic technologies remain unclear. Novel technologies, such as long-read sequencing and RNA diagnostics, continue to show further promise. However, the assessment of novel technologies can be challenging and translation into routine healthcare is not always straightforward.

Health technology assessments (HTAs) and funding pathways typically require specific types of evidence to enable streamlined implementation of new technologies into the healthcare system. Therefore, genomic research should be designed to ensure that the evidence collected will meet such requirements. This helps to support a more robust case for ongoing funding and more timely adoption of new practices that are high value and will improve health outcomes for patients.

Purpose and Scope

Identifying and navigating funding pathways for the sustainable implementation of genomic interventions and novel technologies in Australia's complex healthcare system is challenging, yet increasingly necessary.

Depending on the health technology or intervention/service in Australia, access may need to be facilitated by seeking market authorisation from the Therapeutic Goods Administration (TGA), before, or in parallel to a reimbursement application for funding through a subsidy, funding scheme, or state/territory pathway. The TGA is Australia's regulatory authority for therapeutic goods, including medicines, medical devices, and diagnostic tests,[5] and is responsible for assessing the safety, quality and efficacy of new health technologies.[6]

Public funding pathways in Australia that are relevant to genomic and genetic services or interventions include the Medicare Benefits Schedule (MBS), the National Health Reform Agreement (NHRA) and activity based funding (ABF), block funding, and funding arrangements that are specific to state/territory governments. Philanthropic or other private funding pathways can also be pursued, but the application processes for these are often unique and opportunistic.

Key Pathways and Intended Audience

In the following sections, we summarise the characteristics of the key funding pathways (Figure 1) and outline eligible services and evidence requirements to assist researchers with targeted study design. We aim to help researchers with identifying the most suitable pathway for the translation of their research and finding resources with further information.

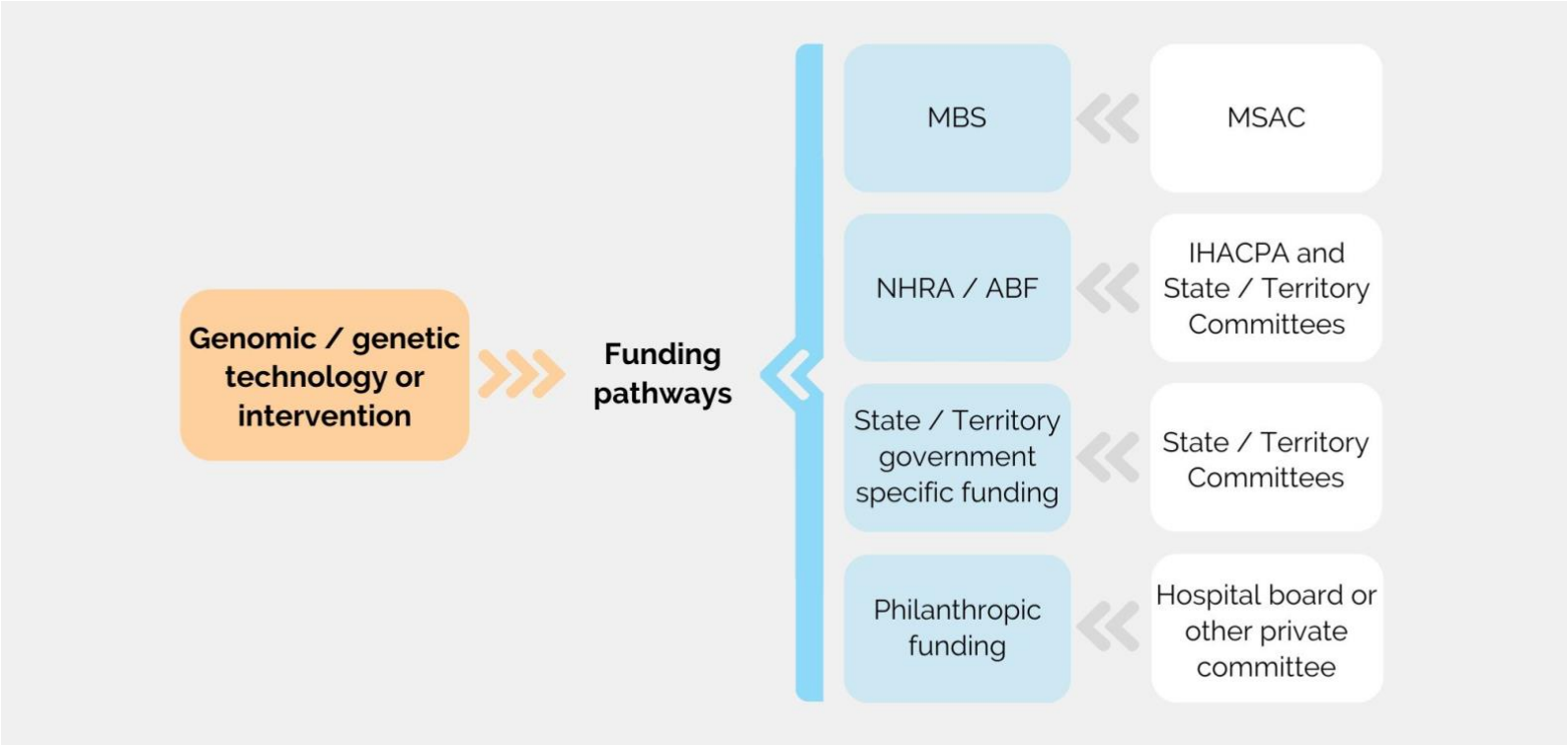


Figure 1: Key funding pathways summarised in this document, with committees including the Medical Services Advisory Committee (MSAC) and Independent Health and Aged Care Pricing Authority (IHACPA).

In Australia, public genomic healthcare services are provided by a public hospital in a non-admitted or outpatient setting.[7] Some funding programs, mainly the MBS, can be accessed as part of eligible private hospital services and private genomic testing services provided outside of the hospital system.

Funding for population-based screening programs is provided through specialised pathways that are not addressed here. The Cancer and Population Screening (CAPS) Committee has been established by the Australian Government Department of Health and Aged Care (the department), and is comprised of deputy Chief Executive Officers (CEOs)/Chief Health/Medical Officers from the Australian Government and each state and territory government health department. Roles of the CAPS Committee include providing direction on emerging evidence and proposals for new tests, technologies, and treatments in current and new screening programs, including genomic population screening programs. Any new technologies related to a proposed genomic population screening program should be considered by the CAPS Committee. Where a HTA is required, the Medical Services Advisory Committee (MSAC) receives and assesses applications for screening programs, for example for newborn bloodspot screening programs. The principles of implementing and managing population-based screening programs, including sufficient funding, are outlined in the Population Based Screening Framework..

The Health Technology and Genomics Collaboration (HTGC) includes Australian Government and state and territory government health department representatives and is responsible for the oversight of a nationally coordinated approach for the assessment, implementation, monitoring and evaluation of new health technology. The HTGC operates in accordance with the National Health Reform Act 2011 and the recently published Framework for the assessment, funding and implementation of high cost, highly specialised therapies and services. We have not covered funding for highly specialised therapies in this document; however, the NHRA Addendum defines highly specialised therapies and describes relevant funding arrangements.

The HTGC is also overseeing implementation of the National Health Genomics Policy Framework to integrate genomics into the Australian health system. The HTGC reports to the Health Chief Executive Forum (HCEF), which is a forum comprised of the health department CEO from each state and territory and the Australian Government for shared decision-making regarding health services in Australia.[8]

Funding Snapshot

Medicare is Australia's universal healthcare system that subsidises health services and products.[9] Medicare covers services delivered in public and private hospitals, medical services (i.e. general practitioner (GP) consultations), diagnostic tests (as delivered by pathology services), imaging, and scans. Medicare is comprised of the:

1. MBS – subsidises medical services and services for private inpatients and private outpatients.
2. Pharmaceutical Benefits Scheme (PBS) – subsidises medicines.
3. NHRA – funds public hospitals treatments, i.e. ABF through an agreement between the Australian Government and the state and territory governments.

Funding details for the MBS and NHRA/ABF are summarised in Table 1. Services for hospital patients are funded by the Australian Government and state, and territory governments, and private health insurance (if applicable), with potential out-of-pocket costs for private patients (Figure 2).

Table 1: Characteristics of ABF and MBS

Program	NHRA / ABF	MBS
Assessment Committee	IHACPA State and territory committees	MSAC
Funded by	State and territory governments Australian Government	Australian Government
Funding source	National Health Funding Pool	Australian Government*
Funding mechanism	Activity-based, with pricing set by IHACPA	Itemised fee for service provision, with subsidies set by Australian Government
Limits	Capped at 6.5% growth	Unlimited, demand-driven
Patients covered	Public patients (inpatients or outpatients)	Non-hospital service patients e.g. primary care, optical, mental health services Private patients (inpatients or outpatients)
Benefits	Widely accessible Provide access to more services, especially for complex conditions/cases	Patients can choose their doctor Shorter wait time

**The Australian Government sets an MBS fee, and out-of-pocket costs or private health insurance rebates may be required to cover actual cost of the service, or "gap".*

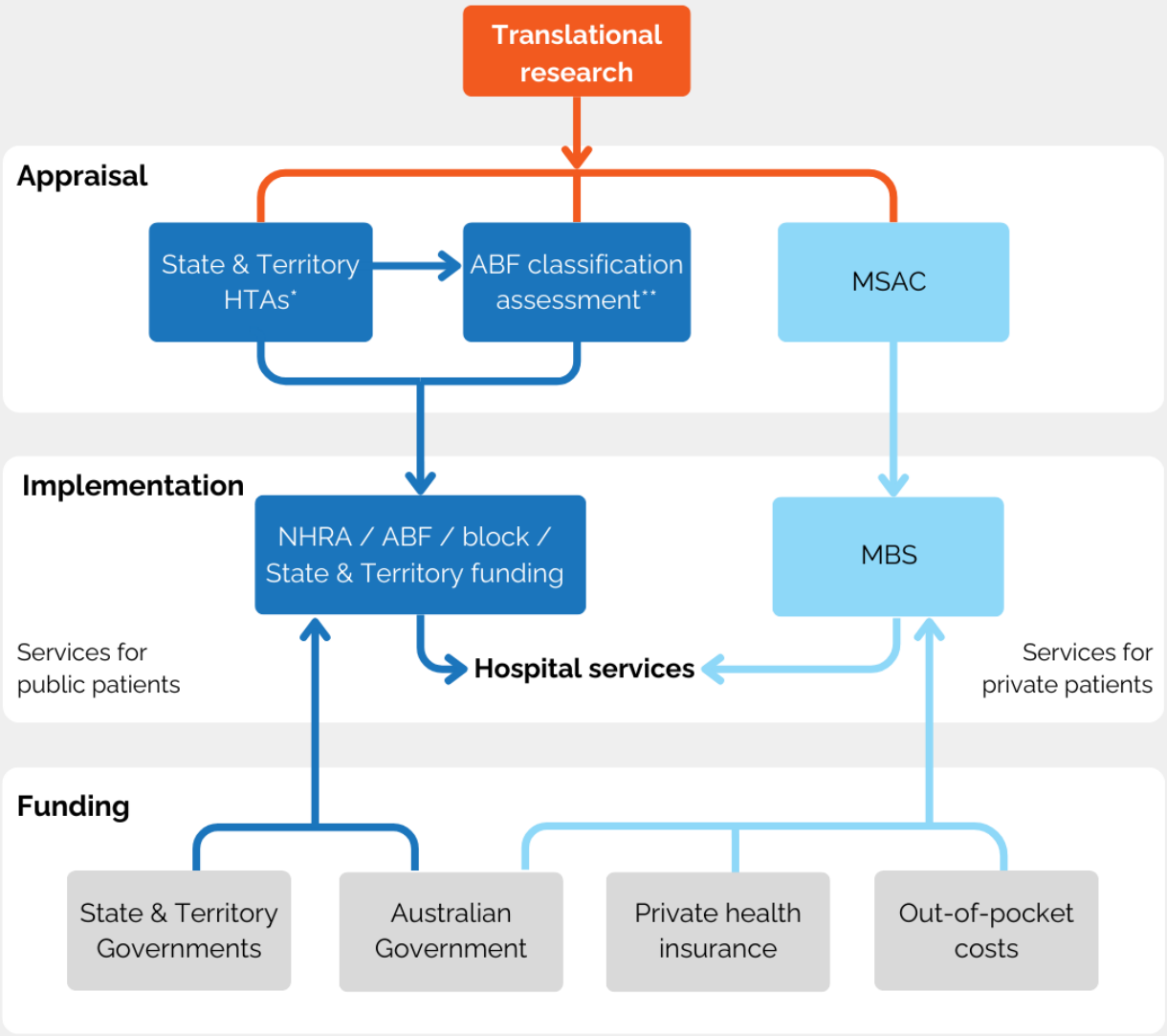


Figure 2: Funding mechanisms for hospital services.

*Decisions regarding the use of a genomic technology as part of in-scope services are made by state and territory governments as system managers. Consideration by IHACPA is not required for funding of new health technologies under the NHRA if a suitable classification already exists. In this scenario, states/territories may use or conduct HTAs to determine what services are provided by their hospitals.

**For new health technologies that are not adequately accounted for in existing classifications. Assessment carried out under IHACPA's New Health Technology Policy.

MBS – The MBS is a list of medical and hospital services that are subsidised by the Australian Government with a unique item number, descriptor, and fee. Funding for the MBS is demand driven. Consultations with GPs and mental health services (out of hospital services) are both categorised as medical services, whereas hospital services are those provided to private patients. MSAC make recommendations about MBS items and the associated fee.

NHRA – The NHRA outlines funding arrangements for public patients (activity based and block funding) agreed upon by the state, territory, and Australian governments.[10] ABF is shared between the Australian Government and state and territory governments, with Australian Government funding capped at 6.5% growth per year.[11] "Activity" includes hospital admissions and outpatient activity (seeing a specialist in an outpatient clinic, for example). The NHRA specifies that funding arrangements will be financially neutral with respect to all patients, regardless of whether they elect to be private or public. In a public hospital, patients are public patients unless they consent to be treated as a private patient.[12] Funding for public and private hospital patients is summarised in Table 2.

Table 2: Funding for public and private hospital patients.

	Public patient	Private patient
Public hospital	Medicare via ABF (free of charge to patient).	MBS, private health insurance, some out-of-pocket costs.
Private hospital	Under certain contracting arrangements private hospitals may deliver public hospital services to public patients.	MBS, private health insurance, some out-of-pocket costs.

Finding Funding

Medicare Benefits Schedule (MBS)

The **MBS** is a list of professional health services that are subsidised by the Australian Government. MBS items provide patient benefits for a wide range of health services including consultations, diagnostic tests, and operations.

Medicare is a substantial funder of genetic and genomic tests, with approximately $76.8M in Medicare benefits for testing subsidised in 2022-23.[13]

MSAC is an independent non-statutory committee that provides expert advice to the Australian Government on the evidence relating to the comparative safety, clinical effectiveness, cost-effectiveness, and total cost of proposed new medical technologies and procedures.[14] Advice from MSAC usually relates to new services proposed for funding under the MBS but can also relate to amendments or reviews of existing services funded by the MBS or other programs. MSAC is also involved in the assessment of codependent technologies, where the combined use of different health services/technologies leads to or enhances the intended clinical effect.[15] The different types of codependent applications include:

- Investigative service/technology + therapeutic service/technology – both requiring support through MSAC.
- Investigative (support through MSAC) + therapeutic pharmaceutical (support through PBAC)
- Consultative (support through MSAC) + therapeutic pharmaceutical (support through PBAC)

Codependent applications can be integrated (combined assessment report lodged via PBAC) or streamlined (separate assessment reports lodged via PBAC and MSAC). More information on codependent technologies is available from MSAC here and PBAC here.

MSAC also makes recommendations in response to requests for non-MBS funding from other bodies where relevant, such as through the NHRA Addendum (e.g. CAR-T cell therapies), or National Blood Agreement (blood/blood-related products). MSAC provides advice to inform funding decisions, but is not responsible for making funding decisions or implementing accepted advice.[16]

Applications to MSAC can be submitted by anyone, including medical professionals, the medical industry, professional organisations, and others seeking Australian Government funding for a new medical service or amending an existing service.[17] MSAC meets three times each year (dates available here), however applications can be submitted (or withdrawn) at any time. MSAC can only assess new health services or technologies if they receive an application or referral.[14]

MSAC has the following subcommittees:[18]

- **PICO Advisory Subcommittee (PASC)** focuses on confirming the population, intervention, comparator, and outcomes (PICO) of the proposed medical service or technology that forms the basis for the assessment report.

- **Evaluation Subcommittee (ESC)** provides advice to MSAC regarding the quality, validity, and relevance of the clinical evidence and economic assessment provided in the assessment report.

Application timelines can vary depending on many factors, including the timing of application submission with respect to MSAC meeting dates and deadlines. Applicants should be aware of these dates to help plan for each stage of an application.

Depending on the complexity of an application and the supporting evidence to be assessed by MSAC or its subcommittees, consideration at more than one meeting may be required and this will impact the overall timeline for the application.

The MSAC application process is summarised below:

Pre-application – Applicants can seek advice for an application before applying to MSAC. Meetings are optional and provide an opportunity to find out more about the application process and the proposed application. Anyone planning to apply can request a meeting. To organise a meeting, submit a request at least 6 weeks before you plan to apply to MSAC by contacting the HTA inbox, or via this form.[19]

Lodge application form – The applicant lodges an online application form through the Health Products Portal (HPP) to provide preliminary information to the HTA team in the department.[20]

Pre-assessment – This phase includes 3 stages; suitability, PICO confirmation development and PASC consideration.[21]

1. **Suitability** – All new applications undergo a check to assess completeness, suitability for the MSAC process and appropriateness of the nominated funding source by the relevant policy area(s) of the department.[22]
    - If the application is **not suitable**, the applicant is notified of the outcome and options, which may include referral to another pathway or committee.
    - If the application **is suitable**, the applicant will be advised of the most appropriate pathway. There are 3 main pathways for assessing applications:[23]

      Standard pathway – the application must go through all stages of the MSAC process, being considered by PASC and ESC before progressing to MSAC (the most common pathway, outlined in Figure 3 on page 13).

      Expedited pathway – applications with a clear PICO can be considered by ESC and then MSAC (bypassing PASC).

      Direct pathway – for less complex applications that can progress directly to MSAC (bypassing both PASC and ESC).

    - **Pre-PASC consultation** (standard pathway only) starts when the secretariat publishes the redacted application summary and PICO set/s, and relevant PASC meeting agenda. Consultation helps MSAC assess health services and technologies for public funding. Applications involve public consultation, where input can be provided by everyone including individuals, organisations, consumers, carers and health professionals, as well as targeted consultation.[24]

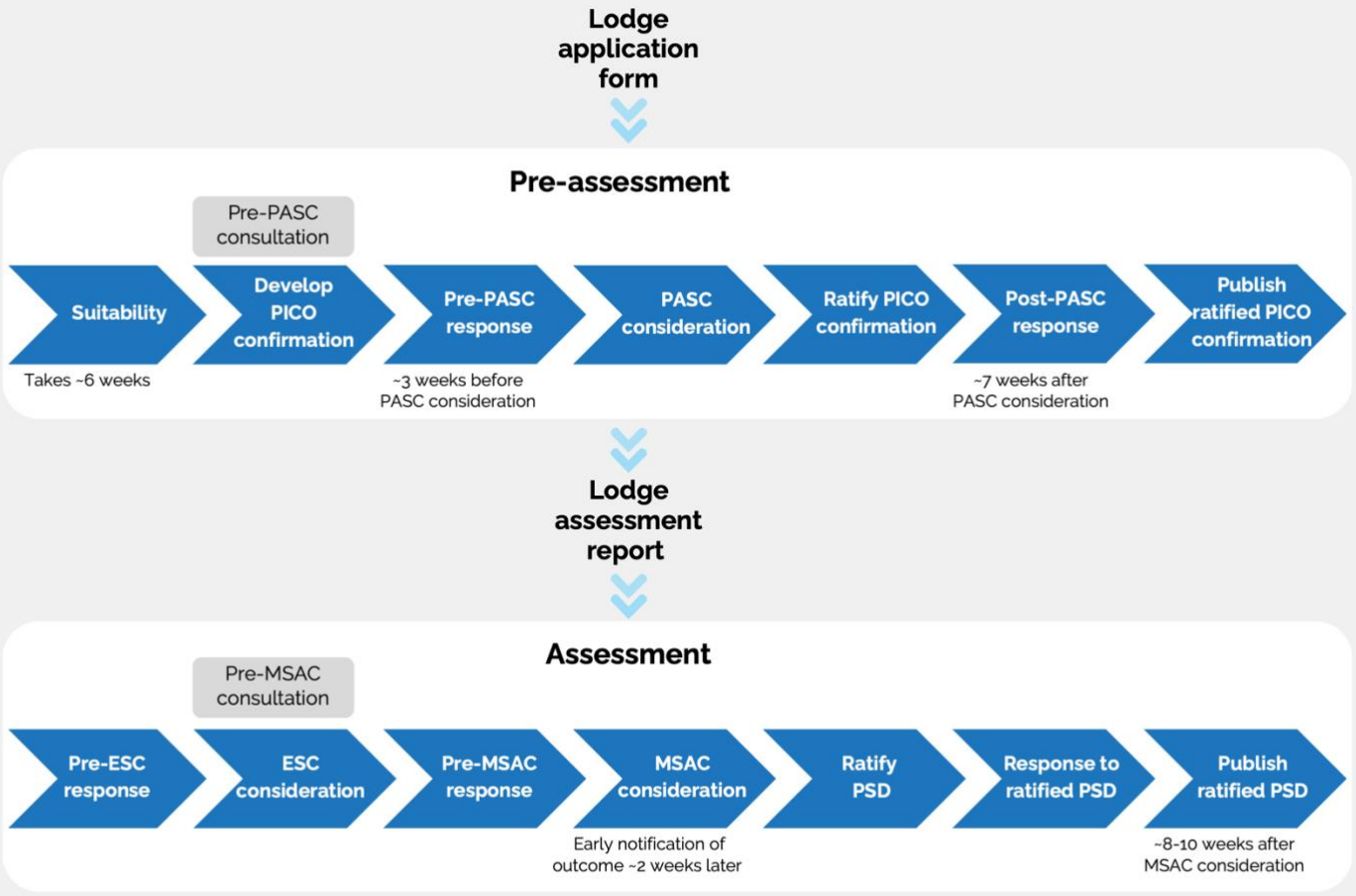


Figure 3: High-level MSAC process for the standard pathway, based on information and approximate timeframes from the MSAC website and MSAC Guidelines. MSAC advice is published in a Public Summary Document (PSD).

2. **PICO confirmation development (standard pathway only)** – An independent HTA group develops the PICO confirmation (with input from the applicant) to be considered by PASC.[25] The applicant can provide a **pre-PASC response** to the draft PICO confirmation and pre-PASC consultation input**.**

3. **PASC consideration** – PASC considers the PICO confirmation, pre-PASC consultation input and the applicant's pre-PASC response. Applicant representatives and clinical/other experts may be invited to attend the PASC meeting, for consideration of the relevant application.[26]

   The outcome of PASC consideration may be:

   - **Application may proceed** – if the PICO confirmation is found to be accurate, the HTA group update it as needed to include the PASC advice. PASC then ratify the PICO confirmation.
   - **More information required** (uncommon) – if the PICO confirmation needs more information, further PASC consideration may be advised before the PICO confirmation can be finalised.
   - **Application cannot proceed** – only occurs if PASC identifies serious concerns that are unable to be resolved, during the PASC meeting or post-meeting process.

The applicant receives the ratified PICO confirmation around 7 weeks after the PASC meeting and has the opportunity to provide a **post-PASC response**, including any redactions to be made. The secretariat then publishes the ratified PICO confirmation with any agreed redactions on the application webpage.

Where relevant, a submission to the TGA for listing on the Australian Register of Therapeutic Goods needs to be submitted prior to commencement of the assessment report.

Lodge assessment report – An **assessment report** is either developed by the applicant (application-developed assessment report or ADAR) or by a contracted HTA group (department-contracted assessment report or DCAR).[27]

- The report presents supporting comparative evidence, including clinical literature and economic evaluation, and should be prepared according to the MSAC Guidelines.

Assessment – This phase begins once the assessment report has been lodged, and includes consideration by ESC and/or MSAC, with the following stages.[28]

1. **Critical review of ADAR** – For applications on the standard pathway or expedited pathway with an ADAR, the secretariat contracts an HTA group to develop a commentary that critically reviews the ADAR.[29]

2. **Pre-ESC response** – The applicant needs to provide a pre-ESC response to the ADAR commentary or to the DCAR.[30, 31] The HTA group may address the pre-ESC response for a DCAR in a rejoinder.
   Note: For applications on the direct pathway, an ADAR is critically reviewed in an overview paper (usually prepared by the secretariat).[32] Since this pathway involves bypassing PASC and ESC, the applicant needs to provide a **pre-MSAC response** to the overview paper.

3. **ESC consideration** – ESC considers the assessment report and other applicable documents during the ESC meeting and provides a written ESC report.[33]

4. **MSAC consideration** – MSAC appraises the evidence, including that provided by PASC and ESC (if applicable), before providing advice to the Australian Government.[34]
   - All applications have a **pre-MSAC consultation** period, which starts when the ESC meeting agenda is published.
   - Around 5 weeks before the MSAC meeting, the applicant needs to provide a **pre-MSAC response** to the ESC report or overview paper and consultation input.[35] A summary of consultation input and copies of consultation feedback from organisations are also provided to the applicant for comment.
   - MSAC or the applicant may request a '**hearing**' during the MSAC meeting if the application fulfills specific criteria.
   - If the applicant agrees to keep the outcome confidential, they can receive early notification of the outcome 10 business days after the MSAC meeting.[36] Otherwise, the applicant must wait 6-8 weeks to receive the outcome when the PSD is shared with the applicant, before it is published. The PSD is published around 8-10 weeks after the MSAC meeting.

- MSAC's advice may be to *support or partially support funding*, *not support funding*, *defer advice,* or *provide other advice*. More information on each outcome and the relevant next steps is available here.
- If funding was not supported or advice was deferred, the applicant can request a post-MSAC debrief meeting.

Evidence required for an application to MSAC

The **application form** requires:

- **Application details**, including succinct descriptions of the health service/technology, medical condition(s), and any relevant MBS item(s).
- **PICO sets** – there can be multiple PICO sets for one health service/technology, and each set requires a purpose and rationale.
- **Proposed MBS items** for MBS-related applications, with a description of the current funding mechanism(s), and attachment of a cost break down.
- **Change in health outcomes**, with a rationale for whether the service/technology is better or no worse than the specified comparator(s), with separate consideration of safety and effectiveness.
- **Estimated utilisation,** with percentage uptake for the first three years and supporting references.
- **Consultation,** including lists of all appropriate professional bodies/organisations, patient and consumer advocacy organisations/individuals, sponsor(s)/manufacturer(s)**.**
- **Statement of Clinical Relevance** for MBS-related applications from the most relevant professional medical college or society.
- **Regulatory information,** including TGA classification.
- **Codependent details** if relevant, including whether a submission will be made to the Pharmaceutical Benefits Advisory Committee (PBAC) or Medical Device and Human Tissue Advisory Committee (MDHTAC, previously known as the Prostheses List Advisory Committee or PLAC).[37]
- **Referral from other committee** where relevant for applications requesting non-MBS funding

More information on each section of an MSAC application is available here, and the MSAC website has application record examples for MBS funding or non-MBS funding.

The **PICO set** requires:

- Definition of the target population, intervention, comparator(s), and outcomes (PICO) to help inform assessment of the proposed technology/service.
  - **Prior tests** – (where relevant) to refine the relevant population.
  - **Population** – characterise the targeted patient population, disease, or condition.
  - **Intervention** – describe the proposed health service/technology, including whether it is investigative or therapeutic (or both), and relevant settings in which it will be delivered. Molecular diagnostic tests are classified as investigative technologies.[38]
  - **Comparator** – nominate the appropriate comparator(s) with a rationale and include whether they will be wholly/partially replaced, displaced, or used in combination.
  - **Outcomes** – list the key health outcomes that need to be measured to assess the clinical claim for the proposed health technology; these include

health benefits, harms, resources, and the value of knowing. Information about resulting changes in patient management, or prognosis is also required. In particular for genomic test applications, the multiplicity of test purpose and outcomes needs to be considered in the development of the PICO and assessment report.

- **Proposed MBS items** for MBS-related applications – describe current funding for the technology and provide details for at least one potential MBS item.
- **Algorithms** – describe the clinical management algorithm before, during, and after the use of the proposed health technology and explain any differences to the algorithm(s) for comparator(s). Diagrams of the clinical management algorithm with and without the proposed health technology are also required.
- **Claims** – describe why the proposed technology is claimed to be superior, non-inferior, or inferior to the comparator(s) in terms of health outcomes (with separate claims for safety and effectiveness), and whether immediate costs and related consequences are more/equivalent/less than those for the comparator.
- **Summary of evidence** – summarise recent high quality clinical studies that support use of the proposed service/technology and relevant research that is yet to be published.

Multiple PICO sets might be needed if the health service/technology can be used for different purposes or across multiple populations. For example, cascade testing should be described in a PICO set that is separate to testing of the proband. Refer to the PICO set template (MBS funding) or PICO set template (non-MBS funding) for detailed guidance.

The **assessment report** requires synthesis and evaluation of the highest quality evidence regarding safety, effectiveness, and cost-effectiveness in the population and indication from the PICO confirmation. The templates for the main text of an ADAR and DCAR are identical, but the executive summary differs:

- **ADAR** – executive summary should provide a brief overview of the clinical claim and value, along with rationale for seeking public funding and expected benefits for patients.
- **DCAR** – executive summary should summarise key findings of the assessment report in a format that reflects the structure of the advice from ESC.

The ADAR and DCAR templates include 5 main sections and 12 appendices. Each section includes Technical Guidance subsections (labelled as TG 1, TG 2, etc.), some of which may not be relevant to every assessment report. These sections and subsections are outlined in Table 3, which is adapted from the MSAC Guidelines.

Table 3: Main sections and technical guidance for the assessment report.

Section 1: Context

TG 1-5	• Purpose of the application • PICO (including clinical management algorithms) • Proposed funding arrangements • History of MSAC submissions • Approach to assessment (including full HTA, and exemplar/facilitated assessment which aims to simplify assessment of gene-related investigative technologies)

Section 2: Clinical evidence

Section 2A: Therapeutic technologies		**or**	**Section 2B: Investigative technologies**	

TG 6-8	• Effectiveness
	• Safety
	• Interpretation of evidence
			TG 9-16	• Assessment framework
				• Direct from test to health outcomes evidence
				• Linked evidence
				  ◦ Test accuracy
				  ◦ Change in management
				  ◦ Health outcomes
				• Safety
				• Special cases
				• Interpretation of evidence

Section 3: Economic evaluation

Section 3A: Cost-effectiveness analysis (for a clinical claim of superiority or inferiority)		**or**	**Section 3B: Cost minimisation** (for a clinical claim of noninferiority)	
TG 17-25	• Overview and rationale of the economic evaluation • Model development process • Population and setting • Model transition probabilities, variables and extrapolation, health outcomes • Health care resource use and costs • Model validation • Results of the base-case economic evaluation • Uncertainty analysis: model inputs, structure, and assumptions		**TG 26**	• Cost minimisation approach

Section 4: Use of the health technology in practice

TG 27	• Use of the health technology in practice

Section 5: Additional relevant information

TG 28-29	• Value of knowing • Other relevant considerations (including ethical, legal, and social aspects)

MSAC may also hold a stakeholder forum before or after an MSAC meeting to explore key questions for an application or emerging technology. MSAC may invite individuals with relevant knowledge and representatives from relevant consumer and professional organisations. A record of the discussion will be published on the application webpage.

The MSAC application pathways are designed to be a flexible approach to HTA and can be navigated for a range of different genomic and genetic applications.

Genetic testing for heritable kidney disease (other than Alport syndrome)

Heritable genetic kidney diseases reduce kidney function which, over time, progresses to kidney failure requiring dialysis and a kidney transplant in some cases. Early identification of kidney diseases is essential so that treatments can be started early to delay disease progression. About 30% of kidney diseases are caused by an inherited genetic variant in a single gene. Variants arising for the first time in the affected person (de novo variants) account for another 10%.[39]

While there were existing MBS items funding genetic testing for Alport syndrome (introduced in 2019 after MSAC application 1449), many other non-Alport genetic kidney conditions were not eligible for testing via the MBS. The same academic applicant submitted an application to MSAC for the funding of genetic testing for heritable kidney diseases other than Alport syndrome in 2019.

While MSAC and its sub-committees acknowledged the clinical need, they considered the quality of evidence low. Their assessment noted the lack of clinical trial data and that the complex economic evaluation was largely uninformative. ESC noted issues related to lack of evidence, particularly in ascertaining clinical validity and utility, and for policy considerations.

However, the reported diagnostic rate of at least 20% was considered acceptable (>10% was considered acceptable). Other areas of strength identified by MSAC included that genetic testing reduces the need for invasive procedures, that negative results in cascade testing release relatives from observation, and that affected family members will not inadvertently be used as kidney donors. The value for reproductive decision making was also recognised. MSAC decided that the 'cost per measure of diagnostic yield' was acceptable, and the test would cost the MBS relatively little per year.

Public consultation feedback raised the importance of the "value of knowing" – empowering people to take control of their lives. Patient advocacy and consumer group feedback to the public consultation was considered. Overall, the consultation feedback strongly supported MBS funding for the application.

Research data independently provided by the KidGen consortium during the assessment was considered highly valuable. KidGen also corresponded with MSAC about the reimbursement fee and offered solutions. MSAC ultimately rejected proposals to increase the fee, but instead included an allowance for whole exome sequencing (WES) rather than just whole genome sequencing (WGS). This test agnostic approach was taken to keep costs down and because few labs can offer National Association of Testing Authorities (NATA) accredited WGS in Australia. Other adjustments made by MSAC through the process included the simplification of populations from the seven originally proposed in the application down to two, plus the cascade testing population. The item numbers do not name a minimum set of genes for analysis and MSAC removed the need for consultation with a clinical geneticist for test ordering. MSAC foreshadowed a move toward generalised item numbers for cascade testing and reanalysis.

MSAC acknowledged the need for this test to be on the MBS to increase equitable access to testing for everyone, recommending to the Minister for Health and Aged Care that six separate items be funded for adults and children, reanalysis, and reproductive testing in July 2021. The Minister announced the listing of the six item numbers for funding on the MBS from July 2022.[40]

Genetic testing for childhood syndromes

Monogenic syndromes are a group of heterogeneous disorders that typically present during infancy or early childhood.[41] The molecular diagnosis of childhood syndromes is complex, with over 1000 genes implicated. Clinical features of individual syndromes depend on the genetic basis and may include but are not limited to single or multi-organ functional anomalies, facial dysmorphism, congenital malformations, and/or varying degrees of intellectual disability. Early diagnosis of these syndromes can enable timely access to pathways for treatment and symptom management and can inform reproductive planning.

Early genomic testing for paediatric patients has high clinical and diagnostic utility and is cost effective in comparison to current standard diagnostic pathways.[42] Genome-wide copy number assessment by microarray only achieves a diagnosis in 10-20% of children presenting with clinical features of monogenic syndromes.[41] Whole exome sequencing (WES) has been shown to achieve a diagnosis rate of 58% in a childhood syndromes infant cohort, compared with a diagnosis rate of 13% for single or multi-gene sequencing.[43]

Whole exome analysis (WEA) was previously unavailable to children in Australia through the MBS. In 2017, an application was submitted to MSAC to request an MBS listing for WEA in patients <18 years old with undiagnosed suspected monogenic syndromic genetic disorder, with cascade testing of relatives.[44] The application proposed that the WEA be restricted to genes known to cause these syndromes, with analysis of a phenotype-driven list of candidate genes first, followed by a broader analysis of all other known genes with clinical evidence to indicate potential involvement in the patient's condition. Genes that are not currently associated with syndromic disorders or that are unrelated to the patient's clinical features were to be excluded from the analysis. Cascade testing was proposed for relative of affected individuals for whom a diagnosis was made via WEA, with investigation limited to the causative gene variant(s) identified in the affected individual. Applicants were invited to use the clinical utility card (CUC) Proforma.

Following consideration in July 2018, MSAC did not support this application but acknowledged the unmet clinical need, and potential benefits such as avoiding further diagnostic testing and improving reproductive confidence for the patient's parents.[45] Concerns raised by MSAC included the breadth and heterogeneity of the syndromes in the application, the limited data available for evidence of changes in clinical management and overall improvements in health outcomes, the best type of technology for performing the test, difficulties in limiting the test to the proposed target population, along with issues related to equity of access, ethics of consent and workforce capacity. MSAC also queried the age range of 0-18 years for the proposed patient population, as the oldest patient in the evidence presented was only 10 years old.

MSAC held a stakeholder meeting in October 2018 to facilitate further discussion of these issues with consumers, providers, and requesters, and inform its reconsideration of the application. A key outcome of this meeting was the broadening of testing criteria for this application. The original testing criteria required two or more of the following clinical features: [46]

- intellectual disability
- single or multiple congenital anomalies
- dysmorphic facial features

Continued ...

During the stakeholder meeting, the following three sets of criteria were agreed upon:

- at least moderate intellectual disability confirmed by the results of a credentialed psychometric test in a child aged 2 years or older; or
- at least severe developmental delay in a child aged younger than 2 years; or
- dysmorphic facial features AND one or more major structural congenital anomalies.

The new criteria were associated with an expected expansion in the volume of genetic testing, and the applicant was therefore required to provide additional supporting evidence and revised utilisation estimates in the reapplication.

At a second MSAC meeting in November 2018, MSAC 'supported MBS listing of next-generation WEA for childhood syndromes in affected individuals, with limited reanalysis, and targeted cascade testing of relatives of patients with a genetic diagnosis.' MSAC excluded trio testing from this item, with the rationale that trio testing should be requested by a sub-specialised clinical geneticist in consultation with the laboratory, which are both typically located in public hospitals. Potential cost-offsets, such as avoiding a diagnostic odyssey and unnecessary treatments, were noted. However, MSAC advised that the utilisation estimates and proposed item descriptors needed further revision, and should be provided for MSAC Executive review.

Following multiple revisions, the proposed utilisation estimates and item descriptors were supported by MSAC at a third meeting in August 2019. This meeting predominantly focused on the issue of trio testing, which had been raised during the stakeholder meeting. Several advantages of trio testing were noted by MSAC, but the greater cost of reagents compared to singleton testing and increased effort from the requesting clinician were also highlighted. MSAC subsequently supported the listing of a second item specifically for trio testing of affected individuals with an upper limit of $2,900 for the item fee.

MSAC recommended the listing of six separate MBS items, including specific items for trio testing, singleton testing, re-analysis, cascade testing for biological siblings, first-degree biological relatives, and biological parents or other biological relatives who have similarities in phenotype or a suspected monogenic condition. The items for trio testing and singleton testing specified a maximum age limit of 10 years old for the patient. The Minister for Health and Aged Care subsequently announced that six item numbers would be listed for funding on the MBS from May 2020.[47] Approval from a clinical geneticist is required on a case-by-case basis, and two cycles of re-analysis are supported.

National Health Reform Agreement

The 2020-25 National Health Reform Agreement (NHRA) is an agreement between all state and territory governments and the Australian Government. The NHRA is aimed at improving health outcomes for all Australians and outlines transparency, governance and financing of Australia's public hospital system.

The Australian Government contributes towards healthcare funding through ABF (under the NHRA), the MBS, and the PBS. These funding models are financially neutral with respect to all patients, meaning there is no gain or loss based on the funding model used, nor by the patient's choice to be treated as a private or a public patient. This ensures there are no funding incentives for hospitals to treat public or private patients differently.

Public hospitals are funded on an activity basis (see ABF section below) and funding is shared between the Australian and state and territory governments. The NHRA recognises that states and territories are responsible for managing public hospitals.

Activity Based Funding (ABF)

ABF is a way of funding public hospitals, where they get paid for the services (or activity) they provide. The level of activity is determined based on the number and type of patients they treat. State and territory public hospital systems in Australia are largely funded through ABF.

ABF uses national classifications for service types and price weights that are independently determined by the Independent Health and Aged Care Pricing Authority (IHACPA).

IHACPA is an independent government agency established under the NHRA to improve health outcomes for all Australians. Prices calculated by IHACPA enable implementation of ABF and are applied to activity estimates to determine in-year funding flows for public hospitals, however, funding is ultimately reconciled based on actual activity. States and territories are required to report service and cost data to IHACPA, which is used to inform price calculations and funding levels in a cyclical manner.

Prices are also used to indicate how much the Australian Government will contribute towards public hospital funding each year and to set a benchmark for states and territories about the efficient or average cost of providing public hospital services across Australia. Block funding, described in more detail below, is used to fund smaller or unique services such as rural or regional hospitals.[48]

IHACPA has different classifications for different settings. The Australian Refined Diagnosis Related Groups (AR-DRGs) classification is used for admitted acute patient care.[49] Genomics services for admitted acute patients are priced and ultimately funded as part of episodes of care classified using AR-DRGs. Meanwhile, the Tier 2 Non-Admitted Services Classification (Tier 2) is used for non-admitted patient care.[50]

Within Tier 2, there are two main classes that exist for genomics, which are described below. Clinical genetics services in Australia often provide outpatient services in public hospitals, which can record activity against these classifications. Classes are designed to cover pathology test costs and associated healthcare professionals' time, as well as a range of other public hospital costs (e.g. administration). ABF prices are developed to reflect the average service cost, including all the associated costs (such as for pathology

tests) relating to providing that service. Sometimes the actual cost of services delivered will be lower than the average ABF price for that service, and other times it will be higher.

Tier 2 classes used for genomics services provided in the non-admitted setting in public hospitals:

- *20.08 Genetics* (20 series - medical consultation). This covers the diagnosis, management, and treatment of hereditary and/or genetic disorders, where the usual provider is a clinical geneticist.
- *40.66 Genetic counselling* (40 series - allied health and/or clinical nurse specialist interventions) was recently introduced and covers the provision of genetic counselling for hereditary and genetic disorders in a specialist clinic/unit where the usual provider is a genetic counsellor.

Tier 2 also has classes for multidisciplinary case conferences (*20.56 Multidisciplinary case conference - patient not present, and 40.62 Multidisciplinary case conference – patient not present*). A multidisciplinary case conference must involve three or more health professionals from different specialties, arranged in advance to discuss a patient in detail and to coordinate care (which can include genetic health professionals). More detail about the classifications is available here.

Currently, many public clinical genetics services in Australia are funded by or transitioning to ABF.[51] The number of service events for genetics in Australia's public hospital system that were recorded against the *20.08 Genetics* classification) has increased over recent years, from 65,457 events in 2019-20 to 80,321 events in 2021-22.[52, 53]

The 2025-26 price weight for the Tier 2 class 20.08 results in a price of $1,098 per service event, and for the class 40.66 produces a price of $398 per service event. If the definition of a multidisciplinary clinic (an interaction between three or more healthcare providers with one non-admitted patient, containing therapeutic/clinical content and result in a date entry in the patient's medical record) is met for either of these Tier 2 classes, a 50% weighting is added to the usual price.

In theory, all genetic or genomic tests (including the more costly WES and WGS), and indeed any other pathology rest required to deliver a service event should be covered by the non-admitted Tier 2 price weight. Accurate reporting of genomic activity, including pathology costs, will ensure price weights are appropriately adjusted over time. However, there is a three-year time lag and associated funding gap between incurring costs and updated pricing which has been acknowledged in the recent Mid-Term Review of the National Health Reform Agreement Addendum 2020-2025.[54]

Unlike the MBS, there is a not an approved list of pathology tests (including genetic and genomic tests) that can be accessed for ABF-funded services. As states and territories manage hospitals, they may have jurisdictional processes (see *State and Territory Government Funding and HTA Pathways* section below) that might involve conducting HTAs to determine which tests and services can be provided. In some jurisdictions, the departments or hospitals delivering the service in question may have a large amount of autonomy as to which tests they deem appropriate to utilise for their service delivery. Where a suitable Tier 2 class for a service exists, for example *20.08 Genetics*, consideration by IHACPA is not required before introducing a new health technology (e.g. a

genetic/genomic test that is delivered as part of this non-admitted service event. Please note, IHACPA does have a policy that outlines a process for considering new health technologies and any associated classification refinements, described in more detail below.

New Health Technology Policy

IHACPA has a New Health Technology Policy process to assess new health technologies for inclusion into the ABF classifications.

A new health technology is defined by IHACPA as '*an intervention developed to prevent, diagnose or treat medical conditions; promote health; or provide rehabilitation*'.[39] New submissions are accepted on an ongoing basis and can be submitted through the Australian Classification Exchange (ACE) portal.

Before making a submission, IHACPA recommends reviewing the current classifications available on their website to determine if the new health technology is already accounted for. Submissions should represent major advances in the quality of patient care and new capabilities that are not already captured in existing classifications.

The submission form available through the ACE portal includes sections requesting information about the new health technology, the volume or anticipated volume of delivery, associated costs, benefits, alternatives, and an implementation schedule as outlined below.

- **New health technology:**
  - Submitter details
  - Date of submission
  - New health technology
  - Description of the technology, including details of the intervention, service delivery setting, treatment cohort information, and confirmation that the technology is not already captured in the classifications. If a technology can be captured/accounted for in existing classifications or code, it may not need a new unique classification/code. Instead, it may be delivered in the public hospital system by claiming activity for an existing classification/code. IHACPA will provide advice based on the technology/intervention.

- **Submission details:**
  - Details/status of MSAC application, if applicable
  - Details of TGA approval, if applicable
  - Anticipated uptake in Australia, in terms of patient and service delivery
  - Existing research, grant, or other source of funding if applicable
  - Costs associated with the technology, such as cost per episode of care
  - Alternatives to the technology currently in use, and cost of alternatives
  - Benefits associated with the technology, including impacts on service delivery/patient care, risk assessment, cost effectiveness and cost-benefit analysis, and any relevant studies
  - Any international experience
  - Implementation schedule

IHACPA, in consultation with classification working groups and the Clinical and Jurisdictional Advisory Committees (CAC and JAC) will review submissions, assess the impact on the national classification systems, and refer new health technologies for classification development where required.[55]

Acute Care Genomics

The Acute Care Genomics research program developed a model for the delivery of ultra-rapid whole genome testing for critically ill infants/children. The research shortened the time to diagnosis to less than three days, resulting in a change in management and care for 76% of the patients who received a diagnosis, with clearly evidenced clinical utility and demonstrated cost effectiveness.[56]

IHACPA determined that this test was not suitable for classification refinement in the admitted acute setting, as diagnostic tests are not routinely classified. Critically ill infants and children are likely to be classified to complex AR-DRGs as part of their stay in a newborn/paediatric intensive care unit (NICU/PICU), which will have relatively high price weights. These may account for the costs of rapid genetic testing. Furthermore, where these tests are being delivered and costed to paediatric patients, the costs of genetic services will inform the AR-DRG prices over time. IHACPA advises such testing may be delivered as part of admitted and non-admitted services that are in scope for the NHRA.

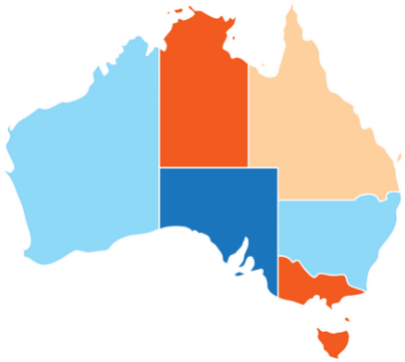
However, decisions regarding the use of rapid genomic testing as part of in scope services are made by state and territory governments as system managers. Reporting of accurate service data (including cost) by hospitals is used to determine and incorporate adjustments to future pricing, upon which ABF is based.

Block Funding

Block funding, determined by the national efficient cost (NEC) is used for public hospital services that are not currently suitable for ABF.[57] IHACPA, in consultation with jurisdictions, maintains block funding criteria and identifies whether hospital services and functions are eligible for block funding only or mixed ABF and block funding. The eligibility of a public hospital service for block funding is determined by using a low volume threshold.[58] For example, smaller services in remote areas or other low volume, specialist services may be funded in this way.

Block funding supports teaching, training, and research in public hospitals and public health programs. For some public hospital services, such as smaller rural and regional hospitals, block funding is more appropriate.[59] Categories of block funding in 2024-25 included: teaching, training and research, small rural hospitals, non-admitted mental health, non-admitted home ventilation services, and highly specialised therapies (e.g. CAR-T cell therapies).

State and Territory Government Funding and HTA Pathways



States and territories are responsible for the management of hospitals and contribute funding via the national health funding arrangements outlined in the NHRA. Additionally, state- and territory-based funding programs exist that may provide options for funding genomic research/technologies in the absence of established national funding (i.e. via MBS or ABF). The location of some genomic research or testing hubs across Australia contributes to variation across jurisdictional funding pathways. However, there are examples where a specialised service provided by one laboratory may be accessed by more than one jurisdiction. For example, clinical testing services for the Northern Territory and Tasmania that are provided by Victorian Clinical Genetics Services (VCGS). This can provide flow-on effects to implement services across Australia. There are also state and territory-specific funding opportunities and centres dedicated to facilitating research translation, some of which are included in the Resources section (see pages I-II).

State and territory-specific processes are nuanced. The information in the following sections should be referred to as a starting point only, and we strongly encourage researchers to engage with local representatives as early as possible.

Australian Capital Territory

The Canberra Health Services (CHS) Health Technology Advisory Committee (HTAC) assesses applications for the introduction of new health technologies into CHS, which have already been recommended by MSAC or are subject to the in-principal approval of MSAC and have been approved by the TGA.

The CHS Introduction of New Health Technology Policy has further details on the roles and responsibilities of the HTAC, and the process for implementing new health technologies in a CHS facility. This policy is for clinical staff who are proposing that a new health technology be used in a CHS facility, and to any staff involved in the implementation process.

The policy only applies specifically to new technologies that:

- Are approved by the TGA and proposed for clinical implementation in a CHS facility.
- Have modification(s) and/or upgrades that will significantly change an existing clinical procedure, treatment, or technology already in use within a CHS facility, leading to potential adverse or unknown impacts on safety and efficacy that require assessment.
- Are approved by the TGA under the Special Access Scheme (SAS).

Refer to the policy for detailed eligibility criteria. The application process requires the submission of an application package to the HTAC Secretariat. This should include a completed *Introduction of New Health Technology Application Form,* which can be found in Attachment B of the Introduction of New Health Technology Policy.

Requirements of the Application Form include:

- Applicant details
- Description of the Department/Service/Location, including explanation of why the health technology is being proposed for introduction
- Detailed description of the technology
- Processes, including whether the technology will replace or be used in conjunction with an existing procedure/treatment/technology and details of advantages if it will replace current procedures
- Expected benefits and potential risks for patients and staff
- Quality and safety, including plans for monitoring and evaluating the technology
- Impacts for staffing and resources, with plans for the development of required skills/credentials
- Declaration of conflicts of interest, and signature from the applicant, Unit Director and Executive Director.

Following HTAC review the applicant, Unit Director, and Executive Director are notified of whether the application has or has not been approved. Applications and enquiries should be directed to the HTAC Secretariat.

New South Wales

Researchers should engage directly with NSW Health to develop partnership from the design phase of research projects to increase opportunities for translation.

Northern Territory

A genomics service has recently been established with funding from the Northern Territory Government's Department of Health. The service is exclusive to diagnostic operations and does not have an associated formal policy or guideline at the time of writing. Key drivers for the establishment of this service included shortening turnaround times for results, reducing costs associated with patient transport and treatment, and enhancing scientific and medical expertise within the local workforce.

Queensland

Specified grants are provided to Hospital and Health Services in Queensland for costs incurred by ABF facilities for services which cannot be appropriately funded through the ABF model. These include high-cost patient outliers, the Limited Indication Medication Scheme, and endorsed statewide services such as Clinical Genetics. Refer to the Queensland Health (QH) Hospital and Health Services Funding and Purchasing Guidelines for more information.

Innovative health technologies are introduced into QH through two main mechanisms; horizon scanning of the global landscape to identify new developments in health technologies for consideration, and the Queensland Technology Future Fund (QTFF) to support the pilot evaluation of proven technologies within QH. These two mechanisms are managed by the Health Technology and Innovation (HTI) team, Office of Research and Innovation, Clinical Planning and Service Strategy division within the Queensland Department of Health. An overview of the end-to-end processes for technology implementation are detailed in the QH Health Technology Innovation Framework. This internal document provides a structured and consistent approach to identifying, assessing, implementing, and monitoring and evaluating novel health technologies.

The QTFF funds health technology pilots led by QH employees and may garner external expertise through collaboration. To be eligible for funding, the technology must be TGA approved and ready to pilot. Genomic technologies are broadly classified as diagnostic devices or testing, patient treatment or therapies, patient monitoring or technology driven models of care, and digital solutions including artificial intelligence. HTAs are performed on QTFF applications, followed by a review from the QTFF executive committee, which consists of divisional Deputy Director-Generals and clinical experts. The HTI team identifies opportunities for the future active scale and spread of successful technology pilots across the Queensland Health system.

Requirements of the QTFF application form include:

- description of the technology and the proposed pilot implementation
- clinical benefits
- safety considerations
- evidence/data to support technology readiness
- pilot project evaluation metrics and methodology
- funds sought for the technology and supporting staff, hardware, software, licences, consumables.

South Australia

The South Australian Policy Advisory Committee on Technology (SAPACT) is responsible for conducting HTAs to inform SAPACT Advisory Recommendations, promoting equity of access for health technologies in SA Health and advising the SA Health Department on potential impacts of new health technologies.[60] The Committee works closely with the Local Health Network (LHN) New Technology Committees, and with SA Health Procurement.

SAPACT review diagnostic and treatment interventions that are:

- high cost, with predicted expenditure per year of ≥$100,000 for a LHN or ≥$300,000 within the South Australian Public Health System; or
- high-risk, with TGA classification as class III or Active Implantable Medical Device (AIMD) regardless of predicted expenditure.

High-volume, low-cost health technologies are generally not considered by SAPACT, as they are currently covered by other assessment processes. Applications for SAPACT assessment can be initiated by clinicians, clinical networks, and other state-wide groups.[61] Potential applications should be discussed with the SAPACT HTA Program manager or with the LHN New Technology Committee to determine eligibility for assessment by the LHN or SAPACT.

If the technology meets SAPACT criteria, the SAPACT Application Form should be submitted to SAPACT with relevant evidence attached. The application form requires information regarding:

- Stage of technology development
- Parameters for consideration, description of the technology, clinical comparator/existing treatment options, and outcomes assessment by addressing PICO criteria
- Clinical safety and effectiveness
- Social, ethical, and equity of access considerations
- Training requirements
- Relevant clinical guidelines

SAPACT also accepts completed LHN New Technology Committees Applications Forms. The Committee meets up to five times per year and makes recommendations based on the SAPACT HTA Decision-Making Criteria, which include the following potential outcomes:

- *"Recommended for clinical use with no further need for assessment.*
- *Restricted recommendation for clinical use subject to implementation under audit conditions.*
- *Restricted recommendation for clinical use with financial or operational restrictions.*
- *Not recommended for clinical adoption. Re-application may be undertaken in the future.*
- *Not Recommended, subject to implementation in clinical trial with approval from SA Health Human Research and Ethics Committee."*

SAPACT informs the applicant and associated LHN New Technology Committee of the outcome, and publishes a HTA Assessment Decision Summary on the SAPACT webpage.

Tasmania

The Department of Health New Devices, Technology and Interventions Policy outlines processes for the assessment of new technologies, including genomic technologies, in the public health system in Tasmania, and the governance of those processes.

Matters considered during assessment include impacts on clinical services, evidence, ethical considerations, staff training, experience, credentialling, financial, human and material impacts, both immediate and projected, consent, clinical governance oversight, feasibility and sustainability, risks and benefits in relation to patient care, other risks, and impacts on disinvestment.

The process requires an application form from the relevant area (e.g. laboratory, clinical area) and a number of levels of approval are required:

- Line manager, Clinical Stream/Divisional Director and Facility Business Manager before being submitted to the Executive Director Medical Services (or equivalent).
- The Executive Director Medical Services evaluates the application and progresses it to the Chief Executive (or equivalent) and the relevant Secretary with a recommendation to progress or deny the application.
- The Chief Executive may seek further information from the Chief Financial Officer and other relevant stakeholders and will determine whether the new application should be referred to the New Device Technology and Interventions Approval Committee (NDTIAC).
- The application to the NDTIAC requires a business case approved by the facility Chief Executive (or equivalent) and relevant Deputy Secretary.
  The NDTIAC will then make the decision regarding adoption of the technology.

Victoria

The Victorian Health Technology Program provides recommendations and policy guidance for health technologies in the state public healthcare system. The assessment of new and existing health technologies and clinical practices through this program informs advice to the Victorian Department of Health regarding safety, implementation, and clinical/cost effectiveness. Victorian funding and approval decisions for new health technologies, including genomic technologies, follow Australian Government processes.

Victorian funding for rare genetic diseases

In the 2017-18 State Budget, the Victorian Government allocated $8.3M in funding over four years for public access to genomic testing for rare genetic diseases in adults and children.[62] This funding also supports access to genetic counselling, clinical genetic consultations, and multi-disciplinary care across genomic sequencing and specialties currently not funded under Medicare. This investment builds upon the $25M funding previously provided by the Victorian Government to build genomic sequencing capability in Victoria through the Melbourne Genomics Health Alliance.[63]

Western Australia

The Western Australian Policy Advisory Committee on Technology (WAPACT) are part of the Government of Western Australia Department of Health. This committee evaluates new high risk, high-cost health technologies that are expected to exceed $250,000 in annual or single acquisition cost.[64]

The introduction of new health technologies into the Western Australian health system is guided by the Health Technology Governance Policy. The policy states that WA Health Service Providers must ensure the safe introduction of health technologies, and the discontinuation of technologies that have a lack of evidence or potential for harm. Each Health Service Provider should have local governance processes with a dedicated health technology committee or equivalent local authority that is responsible for:

- Evaluating the safety, efficacy, and cost effectiveness of new health technologies prior to implementation. Information from national HTA may be used for such purposes, or advice can be requested from WAPACT.
- Monitoring, evaluating and safety reporting for new technologies that are introduced, as per TGA requirements.
- Ensuring staff are qualified to use implemented technologies.
- Considering disinvestment opportunities through regular review.
- Notifying WAPACT about an intention to implement or significantly extend a health technology that presents a high risk to patient safety or has implications for state-wide planning.

Philanthropy

Philanthropy is a non-government funding source for the implementation of findings from medical research that plays an important, sometimes bridging, role amongst the patchwork of Commonwealth, state, and territory funding in Australia.[65]

Philanthropy has been highly relevant to genomics, particularly due to the perceived urgency for implementing genomic technologies and a lack of other fit-for-purpose funding pathways. While there are many different philanthropic funding sources, donations and bequests can be sporadic and usually do not offer long-term support.

Rare Diseases Now program

The Rare Diseases Now (RDNow) program was established in 2019 with funding from The Royal Children's Hospital (RCH) Foundation.[66] This program leverages research and clinical expertise at VCGS and the Murdoch Children's Research Institute (MCRI) to offer access to leading multi-omics technologies and diagnostic pathways for paediatric patients with undiagnosed rare disease.[67] As of September 2023, 110 families had completed testing through RDNow, and 57 patients had received a diagnosis. [66]

The priorities of private, not-for-profit, and philanthropic organisations are often targeted towards improving quality of life for a specific patient group. The process of applying for funding, and the evidence required, varies between organisations.

However, proposals generally require applicants to build a business case, with a focus on highlighting benefits and impacts for patients. Applicants may also be required to outline strategies for ensuring the long-term sustainability of the project beyond the funding period, and to describe future directions for research outcomes.

Wilson Centre for Blood Cancer Genomics

The Wilson Centre for Blood Cancer Genomics was established in 2017 with a $5.5M donation from the Wilson Family.[68] Based at the Peter MacCallum Cancer Centre, the Wilson Centre is translating leading genomic technologies into the clinic and currently offers accredited massively parallel sequencing (MPS) assays for DNA, RNA, and circulating tumour DNA (ctDNA) to patients with blood cancer. The Wilson Family have since donated a further $3.5M to support the Wilson Centre.

Zero Childhood Cancer Program

The Zero Childhood Cancer Program (ZERO) has received private, philanthropic, and government funding. ZERO is a collaborative initiative led by the Children's Cancer Institute and the Kids Cancer Centre at Sydney Children's Hospital, and is the most comprehensive child cancer precision medicine program in Australia.[69] In its first trial phase (2017-20), ZERO was available to children with a less than 30% chance survival, and the molecular basis was identified in almost all cases (94%) for the first 247 patients enrolled.[70] Philanthropic support was received through a partnership with the Lions Kids Cancer Genome Project, and $67M in funding was awarded through the MRFF and the Minderoo Foundation's Collaborate Against Cancer Initiative, to expand eligibility to all children and young people diagnosed with cancer in Australia by the end of 2023.[71, 72]

Emerging Applications

The Genomic Autopsy Study

The Genomic Autopsy study offers genomic testing to investigate genetic aetiologies in cases of pregnancy loss and perinatal death that remain unexplained following standard-of-care autopsy.[73] This study was established in Adelaide in 2015, and through a partnership between SA Pathology, the Women's and Children's Hospital, the University of South Australia, and Australian Genomics, it evolved from a state-based project to a national research program offering diagnostic testing with reduced turnaround times for improved clinical utility.[74]

Funding and support to enable recruitment have included $0.99M in funding from the National Health and Medical Research Council (NHMRC), $3.4M in MRFF funding through the GHFM, and national support from Australian Genomics. Comprehensive genomic investigation identified a likely or candidate cause in over 50% of cases for the first 200 families enrolled in the study.[75] The research funding to support national recruitment into the study has ended, however the Women's and Children's Health Network has committed to fund 15 clinically referred cases per year in perpetuity, with other interstate hospitals also referring as a fee-for-service on an *ad hoc* basis. Funding mechanisms are being pursued to implement this service as part of the clinical standard-of-care pathway for fetal and neonatal loss and avoid inequity of access becoming a significant issue for families across Australia.

Pharmacogenomic testing

Pharmacogenomic testing can guide the optimisation of drug therapies by providing critical information regarding drug toxicity and efficacy.[76] Pharmacogenomics has the potential to transform precision medicine approaches.Broader pharmaco*genomic* testing is not yet subsidised by the MBS, due to a lack of MSAC applications, however there are MBS items for pharmaco*genetic* tests (which analyse only one gene or biomarker).[77]

> DPYD genotyping to predict fluoropyrimidine-induced toxicity (MSAC application 1760) is currently under review. This application is seeking a new MBS item for identifying DPYD gene variants in patients with solid organ tumours who are undergoing standard chemotherapy treatment with fluoropyrimidines.[78]

Patients can pay for direct-to-consumer (DTC) pharmacogenomic testing from international or Australian companies. However, the test results need to be integrated with other aspects of the patient's management. Australia's National Pathology Accreditation Advisory Council (NPAAC) guidelines for The Provision of Direct to Consumer Genetic Tests recommend that providers '*should not offer DTC pharmacogenetic testing without strongly advising the consumer not to initiate or alter the dosage of any existing medication, on the basis of the test results, without first consulting a relevant medical practitioner.*'

A generic template for pharmacogenomic MSAC applications is being developed by the Royal College of Pathologists Australia (RCPA) as part of the Australian Genomics-funded project 'Indications for pharmacogenomic testing in Australia'.[79] This project also involves the development of two pharmacogenomic MSAC applications, which will validate the template and provide a framework for future applications related to testing for drug-gene pairs.

Considerations for an Evolving Landscape

Early engagement with local and national stakeholders is vital to assist with translating research into practice or progressing a funding application. The importance of engaging with consumers, including Aboriginal and Torres Strait Islander peoples, is increasingly in focus and will ensure research and HTA align with the needs and expectations of the community.[80, 81] On the other hand, consulting with policy makers will ensure that research and HTA are prioritised towards addressing healthcare system priorities and gaps.

The Australian Government supported and resourced the HTA Policy and Methods Review (HTA Review) which concluded in 2024. The HTA Review aims to ensure that Australia's HTA processes keep pace with rapid advances in health technology and to minimise barriers to access. The HTA Review was overseen by a Reference Committee, a panel of experts and leaders representing patients, scientific and clinical practice, industry, and health sector public administration. Priority issues considered during the course of the HTA Review included consumer engagement and partnering with First Nations people. The 50 recommendations outline comprehensive reforms to improve HTA processes, addressing inequities, wait times and transparency, including a recommendation to support the development of guidance on the assessment and appraisal of genomic technologies and gene therapies for HTA decisions in Australia.[82]

Next Steps: Navigating Funding Pathways for Genomic Research Translation provides an overview of the pathways and evidence required to support the sustainable implementation of findings from genomics research in Australia. Most of the funding pathways outlined here are broadly applicable to health care specialties outside of genomics and genetics. It should be noted that there are many aspects taken into consideration by decision makers for each funding application and assessment pathway. Meeting evidence requirements does not necessarily guarantee that funding will be granted.

Conclusion

Navigating pathways for research translation is complex, yet critically important for enabling timely access to evidence-based genomics interventions and technologies for patients. Pathways and approaches to implementing genomic testing and technologies will continue to evolve as they are increasingly adopted by healthcare systems.

Resources

General

- Medicare billing in public hospitals – overview
- HTA Policy and Methods Review

NHRA

- 2020–25 Addendum to National Health Reform Agreement (NHRA)

MSAC

- Guidelines for preparing assessments for MSAC
- Guidelines summary for stakeholders
- MSAC Process Framework
- All MSAC Resources
- HPP Guidance for lodging an MSAC application
- Contact details

Activity Based Funding

- Tier 2 Non-Admitted Services Definitions Manual 2024–25
- New Health Technology Policy
- New Health Technology Submission Form

Block Funding

- Understanding the NEP and NEC Determinations

State and Territory Government Funding and HTA Pathways

Australian Capital Territory

- CHS Introduction of New Health Technology Policy

South Australia

- SAPACT Application Form
- SAPACT HTA Decision-Making Criteria
- SA Health HTA Policy
- Health Translation SA HTSA MRFF Catalyst Grant Program

Tasmania

- Long-Term Plan for Healthcare in Tasmania 2040

Victoria

- Victorian Health Technology Program
- Policy and Funding Guidelines
- Victorian Medical Research Acceleration Fund

Western Australia

- Health Technology Governance Policy

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