Australian Government

Department of Health

Application Form

(New and Amended Requests for Public Funding)

(Version 2.5)

This application form is to be completed for new and amended requests for public funding (including but not limited to the Medicare Benefits Schedule (MBS)). It describes the detailed information that the Australian Government Department of Health requires in order to determine whether a proposed medical service is suitable.

Please use this template, along with the associated Application Form Guidelines to prepare your application. Please complete all questions that are applicable to the proposed service, providing relevant information only. Applications not completed in full will not be accepted.

The application form will be disseminated to professional bodies / organisations and consumer organisations that have will be identified in Part 5, and any additional groups that the Department deem should be consulted with. The application form, with relevant material can be redacted if requested by the Applicant.

Should you require any further assistance, departmental staff are available through the contact numbers and email below to discuss the application form, or any other component of the Medical Services Advisory Committee process.

Phone: +61 2 6289 7550

Fax: +61 2 6289 5540

Email: hta@health.gov.au

Website: MSAC Website

PART 1 – APPLICANT DETAILS

1. Applicant details (primary and alternative contacts)

Corporation / partnership details (where relevant): **redacted**
Corporation name: **redacted**
ABN: **redacted**
Business trading name: **redacted**

Primary contact name: redacted

Primary contact numbers
Business: **redacted**
Mobile: **redacted**
Email: **redacted**

Alternative contact name: redacted

Alternative contact numbers
Business: **redacted**
Mobile: **redacted**
Email: **redacted**

2. (a) Are you a consultant acting on behalf of an Applicant?

☐ Yes

☒ No

(b) If yes, what is the Applicant(s) name that you are acting on behalf of?

Insert relevant Applicant(s) name here.

3. (a) Are you a lobbyist acting on behalf of an Applicant?

☐ Yes

☐ No

(b) If yes, are you listed on the Register of Lobbyists?

☐ Yes

☐ No

PART 2 – INFORMATION ABOUT THE PROPOSED MEDICAL SERVICE

4. Application title

Genetic testing for Alport syndrome

5. Provide a succinct description of the medical condition relevant to the proposed service (no more than 150 words – further information will be requested at Part F of the Application Form)

Alport syndrome is one of the commonest causes of inherited renal failure after polycystic kidney disease and reflux nephropathy.

All males with X-linked Alport syndrome develop end-stage renal failure by the age of 40, with 15 – 30% of females developing renal failure by 60 years of age. Those with the less common or recessive form of Alport syndrome (approximately 15%) have renal failure before 40 years of age.

There is no cure for Alport syndrome, however, studies have demonstrated that treatment with angiotensin converting enzyme inhibitors delay the onset of renal failure by up to 13 years if commenced early enough.

6. Provide a succinct description of the proposed medical service (no more than 150 words – further information will be requested at Part 6 of the Application Form)

Genetic testing (targeted whole exome sequencing) to identify one mutation in the *COL4A5* gene, or two mutations in the *COL4A3* or *COL4A4* genes to confirm the diagnosis of X-linked or autosomal recessive Alport syndrome respectively.

7. (a) Is this a request for MBS funding?

☒ Yes

☐ No

(b) If yes, is the medical service(s) proposed to be covered under an existing MBS item number(s) or is a new MBS item(s) being sought altogether?

☐ Amendment to existing MBS item(s)

☒ New MBS item(s)

(c) If an amendment to an existing item(s) is being sought, please list the relevant MBS item number(s) that are to be amended to include the proposed medical service:

Insert relevant MBS item numbers here

(d) If an amendment to an existing item(s) is being sought, what is the nature of the amendment(s)?

i. ☐ An amendment to the way the service is clinically delivered under the existing item(s)
ii. ☐ An amendment to the patient population under the existing item(s)
iii. ☐ An amendment to the schedule fee of the existing item(s)
iv. ☐ An amendment to the time and complexity of an existing item(s)
v. ☐ Access to an existing item(s) by a different health practitioner group
vi. ☐ Minor amendments to the item descriptor that does not affect how the service is delivered
vii. ☐ An amendment to an existing specific single consultation item
viii. ☐ An amendment to an existing global consultation item(s)
ix. ☐ Other (please describe below):

Insert description of 'other' amendment here

(e) If a new item(s) is being requested, what is the nature of the change to the MBS being sought?

i. ☐ A new item which also seeks to allow access to the MBS for a specific health practitioner group
ii. ☐ A new item that is proposing a way of clinically delivering a service that is new to the MBS (in terms of new technology and / or population)

iii. ☐ A new item for a specific single consultation item
iv. ☐ A new item for a global consultation item(s)

(f) Is the proposed service seeking public funding other than the MBS?

☐ Yes

☒ No

If yes, please advise:

Insert description of other public funding mechanism here

8. What is the type of service:

☐ Therapeutic medical service

☒ Investigative medical service

☐ Single consultation medical service

☐ Global consultation medical service

☐ Allied health service

☐ Co-dependent technology

☐ Hybrid health technology

9. For investigative services, advise the specific purpose of performing the service *(which could be one or more of the following)*:

i. ☐ To be used as a screening tool in asymptomatic populations
ii. ☒ Assists in establishing a diagnosis in symptomatic patients
iii. ☒ Provides information about prognosis
iv. ☒ Identifies a patient as suitable for therapy by predicting a variation in the effect of the therapy
v. ☐ Monitors a patient over time to assess treatment response and guide subsequent treatment decisions
vi. ☒ Is for genetic testing for heritable mutations in clinically affected individuals and, when also appropriate, in family members of those individuals who test positive for one or more relevant mutations (and thus for which the Clinical Utility Card proforma might apply)

10. Does your service rely on another medical product to achieve or to enhance its intended effect?

☐ Pharmaceutical / Biological

☐ Prosthesis or device

☒ No

11. (a) If the proposed service has a pharmaceutical component to it, is it already covered under an existing Pharmaceutical Benefits Scheme (PBS) listing?

☐ Yes

☒ No

(b) If yes, please list the relevant PBS item code(s):

Insert PBS item code(s) here

(c) If no, is an application (submission) in the process of being considered by the Pharmaceutical Benefits Advisory Committee (PBAC)?

☐ Yes (please provide PBAC submission item number below)

☒ No

Insert PBAC submission item number here

(d) If you are seeking both MBS and PBS listing, what is the trade name and generic name of the pharmaceutical?

Trade name: Insert trade name here

Generic name: Insert generic name here

12. (a) If the proposed service is dependent on the use of a prosthesis, is it already included on the Prostheses List?

☐ Yes

☐ No

(b) If yes, please provide the following information (where relevant):

Billing code(s): Insert billing code(s) here

Trade name of prostheses: Insert trade name here

Clinical name of prostheses: Insert clinical name here

Other device components delivered as part of the service: Insert description of device components here

(c) If no, is an application in the process of being considered by a Clinical Advisory Group or the Prostheses List Advisory Committee (PLAC)?

☐ Yes

☐ No

(d) Are there any other sponsor(s) and / or manufacturer(s) that have a similar prosthesis or device component in the Australian market place which this application is relevant to?

☐ Yes

☐ No

(e) If yes, please provide the name(s) of the sponsor(s) and / or manufacturer(s):

Insert sponsor and/or manufacturer name(s) here

13. Please identify any single and / or multi-use consumables delivered as part of the service?

Single use consumables: Insert description of single use consumables here

Multi-use consumables: Insert description of multi use consumables here

PART 3 – INFORMATION ABOUT REGULATORY REQUIREMENTS

14. (a) If the proposed medical service involves the use of a medical device, in-vitro diagnostic test, pharmaceutical product, radioactive tracer or any other type of therapeutic good, please provide the following details:

Type of therapeutic good: In-vitro diagnostic test

Manufacturer's name: Various

Sponsor's name: Not applicable

(b) Is the medical device classified by the TGA as either a Class III or Active Implantable Medical Device (AIMD) against the TGA regulatory scheme for devices?

☒ Class III
☐ AIMD
☐ N/A

15. (a) Is the therapeutic good to be used in the service exempt from the regulatory requirements of the *Therapeutic Goods Act 1989*?

☐ Yes (If yes, please provide supporting documentation as an attachment to this application form)
☒ No

(b) If no, has it been listed or registered or included in the Australian Register of Therapeutic Goods (ARTG) by the Therapeutic Goods Administration (TGA)?

☒ Yes (if yes, please provide details below)
☐ No

ARTG listing, registration or inclusion number: Various

TGA approved indication(s), if applicable: Insert approved indication(s) here

TGA approved purpose(s), if applicable: Insert approved purpose(s) here

16. If the therapeutic good has not been listed, registered or included in the ARTG, is the therapeutic good in the process of being considered for inclusion by the TGA?

☐ Yes (please provide details below)
☐ No

Date of submission to TGA: Insert date of submission here

Estimated date by which TGA approval can be expected: Insert estimated date here

TGA Application ID: Insert TGA Application ID here

TGA approved indication(s), if applicable: If applicable, insert description of TGA approved indication(s) here

TGA approved purpose(s), if applicable: If applicable, insert description of TGA approved purpose(s) here

17. If the therapeutic good is not in the process of being considered for listing, registration or inclusion by the TGA, is an application to the TGA being prepared?

☐ Yes (please provide details below)
☐ No

Estimated date of submission to TGA: Insert date of submission here

Proposed indication(s), if applicable: If applicable, insert description of proposed indication(s)

Proposed purpose(s), if applicable: If applicable, insert description of proposed purpose(s) here

PART 4 – SUMMARY OF EVIDENCE

18. **Provide an overview of all key journal articles or research published in the public domain related to the proposed service that is for your application (limiting these to the English language only).** ***Please do not attach full text articles, this is just intended to be a summary.***

	Type of study design*	**Title of journal article or research project (including any trial identifier or study lead if relevant)**	**Short description of research (max 50 words)****	**Website link to journal article or research (if available)**	**Date of publication*****
1.	Retrospective observational registry	Early angiotensin-converting enzyme inhibition in Alport syndrome delays renal failure and improves life expectancy. 274 people followed for 20 years. ACE inhibitors delayed onset of renal failure	'This supports the need for early diagnosis and early nephroprotective therapy in oligosymptomatic patients'.		Gross et al. Kidney Int 2015; 291:494-501
2.	Retrospective observational registry	Outcomes of male patients with Alport syndrome undergoing renal replacement therapy	456 males with AS in European registries; delayed age at onset of end-stage renal failure		Temme et al. Clin JASN 2012;7: 1969-76
3.	Retrospective observational registry	Incidence of renal failure and nephroprotection by RAAS inhibition in heterozygous carriers of X-chromosomal and autosomal recessive Alport mutations	234 Alport mutation carriers in European registries; figure 5: delayed onset of renal failure with preemptive ACE inhibitors		Temme et al. Kid Int 2012; 81: 779-83
4.	Expert guidelines	Expert guidelines for the management of Alport syndrome and Thin basement membrane nephropathy	Advocates genetic testing, and ACE inhibition		Savige et al. JASN 2013; 24: 364-375
5.	Clinical practice recommendations	Clinical practice recommendations for the treatment of Alport syndrome: a statement of the Alport syndrome research collaborative	Advocates ACE inhibition even before the onset of proteinuria		Kashtan et al. Ped Nephrol 2013: 28: 5 - 11
6.	Animal model	Preemptive Ramipril therapy delays renal failure and reduces renal fibrosis in COL4A3-knockout mice with Alport syndrome	Advocates ACE inhibition even before the onset of proteinuria in mouse model		Gross et al. Kidney Int 2003; 63: 438-446.

	Type of study design*	Title of journal article or research project (including any trial identifier or study lead if relevant)	Short description of research (max 50 words)**	Website link to journal article or research (if available)	Date of publication***
7.	Animal model	Treatment of X-linked hereditary nephritis in Samoyed dogs with ACE inhibition.	ACE inhibition in dogs with Alport syndrome delayed onset of renal failure and glomerular membrane splitting		Grodecki et al. J Com Pathol 1997; 117: 209-225.
8.	Retrospective review of 79 children with ACE I alone or with ARB	Long term treatment by ACE inhibitors and angiotensin receptor blockers in children with Alport syndrome.	Both treatments reduced proteinuria and were well – tolerated.		Zhang et al. Ped Nephrol 2016; 31: 67-72
9.	Registration of study	Safety and efficacy of the ACE inhibitor ramipril in Alport syndrome: double blind , randomised, placebo controlled multicentre phase III EARLY PROTECT Alport trial in pediatric patients	Proposed study in 120 children, randomised to Ramipril or placebo. This study is to obtain the evidence that ACE inhibitors delay renal failure		Gross O et al. ISRN Pediatr 2012: 2012: 436046.

** Categorise study design, for example meta-analysis, randomised trials, non-randomised trial or observational study, study of diagnostic accuracy, etc.*

***Provide high level information including population numbers and whether patients are being recruited or in post-recruitment, including providing the trial registration number to allow for tracking purposes.*

**** If the publication is a follow-up to an initial publication, please advise.*

19. Identify yet to be published research that may have results available in the near future that could be relevant in the consideration of your application by MSAC (limiting these to the English language only). *Please do not attach full text articles, this is just intended to be a summary.*

	Type of study design*	Title of research (including any trial identifier if relevant)	Short description of research (max 50 words)**	Website link to research (if available)	Date***
1.	Insert study design	Insert title of research	Insert description	Insert website link	Insert date
2.	Insert study design	Insert title of research	Insert description	Insert website link	Insert date
3.	Insert study design	Insert title of research	Insert description	Insert website link	Insert date
4.	Insert study design	Insert title of research	Insert description	Insert website link	Insert date
5.	Insert study design	Insert title of research	Insert description	Insert website link	Insert date
6.	Insert study design	Insert title of research	Insert description	Insert website link	Insert date
7.	Insert study design	Insert title of research	Insert description	Insert website link	Insert date
8.	Insert study design	Insert title of research	Insert description	Insert website link	Insert date
9.	Insert study design	Insert title of research	Insert description	Insert website link	Insert date
10.	Insert study design	Insert title of research	Insert description	Insert website link	Insert date
11.	Insert study design	Insert title of research	Insert description	Insert website link	Insert date
12.	Insert study design	Insert title of research	Insert description	Insert website link	Insert date
13.	Insert study design	Insert title of research	Insert description	Insert website link	Insert date
14.	Insert study design	Insert title of research	Insert description	Insert website link	Insert date
15.	Insert study design	Insert title of research	Insert description	Insert website link	Insert date

** Categorise study design, for example meta-analysis, randomised trials, non-randomised trial or observational study, study of diagnostic accuracy, etc.*

***Provide high level information including population numbers and whether patients are being recruited or in post-recruitment.*

****Date of when results will be made available (to the best of your knowledge).*

PART 5 – CLINICAL ENDORSEMENT AND CONSUMER INFORMATION

20. List all appropriate professional bodies / organisations representing the group(s) of health professionals who provide the service (please attach a statement of clinical relevance from each group nominated):

Kidney Health Australia
Australian and New Zealand Society of Nephrology

Royal Australasian College of Physicians
Royal College of Pathologists of Australasia
Human Genetics Society of Australia

ANZ Transplant Society

21. List any professional bodies / organisations that may be impacted by this medical service (i.e. those who provide the comparator service):

Not applicable

22. List the relevant consumer organisations relevant to the proposed medical service (please attach a letter of support for each consumer organisation nominated):

Alport Foundation of Australia
Renal transplants groups
Kidney clubs

23. List the relevant sponsor(s) and / or manufacturer(s) who produce similar products relevant to the proposed medical service:

Not applicable

24. Nominate two experts who could be approached about the proposed medical service and the current clinical management of the service(s):

Name of expert 1: **redacted**

Telephone number(s): **redacted**

Email address: **redacted**

Justification of expertise: **redacted**

Name of expert 2: **redacted**

Telephone number(s): **redacted**

Email address: **redacted**

Justification of expertise: **redacted**

Please note that the Department may also consult with other referrers, proceduralists and disease specialists to obtain their insight.

PART 6 – POPULATION (AND PRIOR TESTS), INDICATION, COMPARATOR, OUTCOME (PICO)

PART 6a – INFORMATION ABOUT THE PROPOSED POPULATION

25. Define the medical condition, including providing information on the natural history of the condition and a high level summary of associated burden of disease in terms of both morbidity and mortality:

Alport syndrome is one of the commonest causes of inherited renal failure after polycystic kidney disease and reflux nephropathy. It affects between one in 5 and 10,000 Australians, so that nationally we have a population of 2,000 - 5,000 people with Alport syndrome. However most of these are undiagnosed and are not aware that they are affected.

All males with X-linked Alport syndrome develop renal failure by the age of 40. Fifteen – 30% of females develop renal failure by 60 years of age (Savige et al, 2013). Everyone with the less common or recessive form (15%) develops renal failure before 40 years of age (Storey, JASN 2013) .

Everyone who develops end-stage kidney failure requires dialysis or a kidney transplant. They also have an increased risk of death from cardiac disease. Dialysis costs at least $50,000 a year. Transplantation is cheaper but most patients have to wait an average of 3 years on dialysis.

There is no cure for Alport syndrome but recent studies have demonstrated that treatment with ACE (angiotensin converting enzyme) inhibitors delays the onset of renal failure by up to 13 years if commenced early enough, even before the onset of proteinuria (see previous table). ACE inhibitors not only help control high blood pressure, but also minimise proteinuria, and thus decrease kidney fibrosis or scarring. They are widely used for their blood pressure effects alone, and are safe and inexpensive. In particular, ramipril has been used for years in children .

There is now evidence that early diagnosis and treatment with ACE inhibitors in people with a mutation causing Alport disease delays the onset of renal failure by up to 13 years (Gross, 2015; Temme 2012). Early and accurate diagnosis using genetic testing enables earlier treatment and a longer delay before the development of renal failure.

Fast access to accurate genetic testing for Alport syndrome is critical to instituting ACE inhibitor treatment in Alport syndrome. Currently, genetics tests are requested through referral to a genetic service but there is a long waiting period for patients to be seen, and the genetic services' budget for gene testing is restricted and often focused more on hereditary cancer. This not only delays obtaining the diagnosis in our patients but acts as a disincentive to testing them for genetic mutations. Nationally, nephrologists face the same problem.

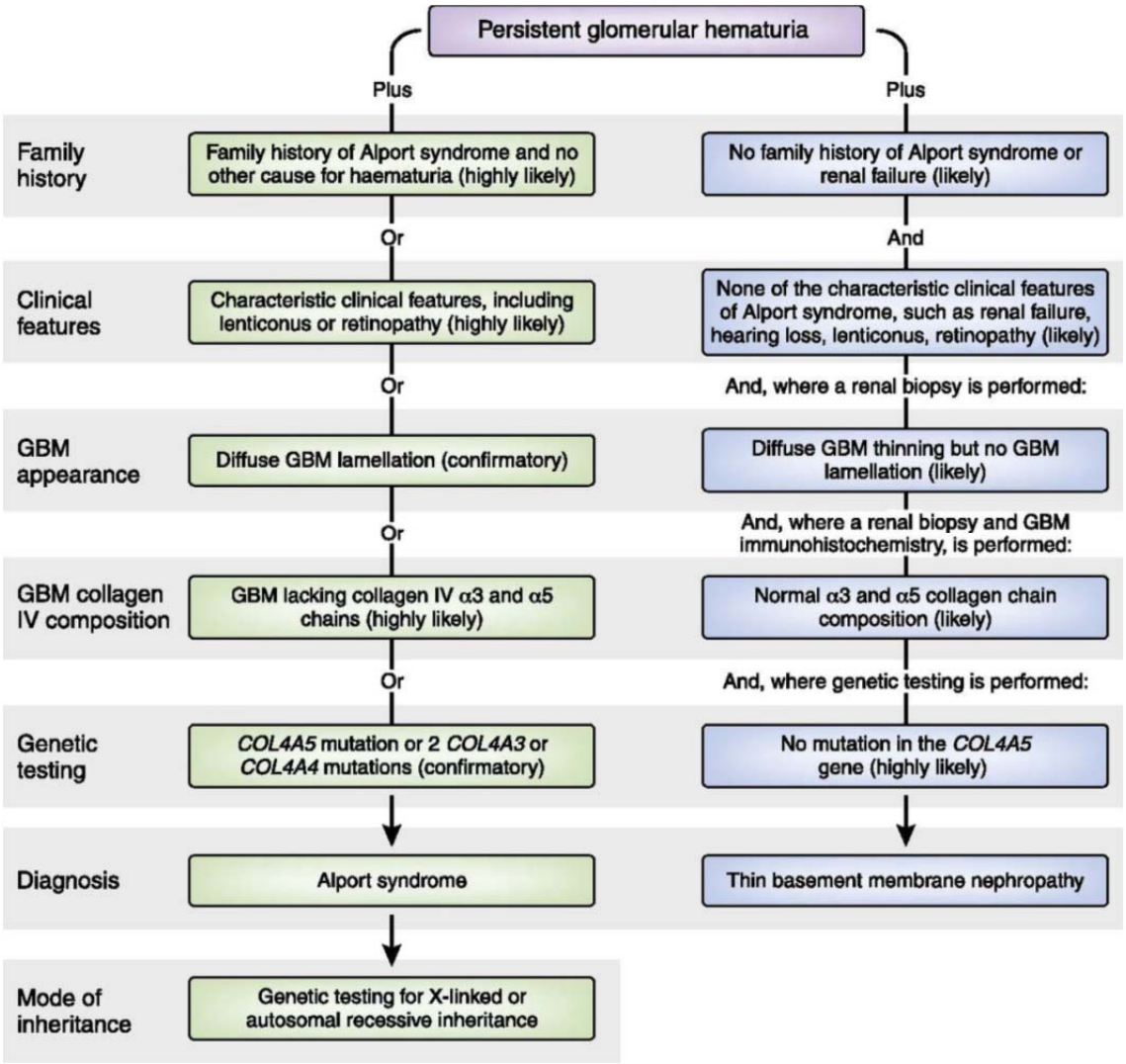
Reimbursement of genetic testing for Alport syndrome through the MBS would enable us to test those individuals suspected of having Alport syndrome, decrease the time taken for testing, and decrease the overall cost to the health system of the extra years on dialysis that were saved.

The following indicates that gene testing is the diagnostic test for choice for Alport syndrome for the reasons provided above (Expert Guidelines for the Management of Alport Syndrome and Thin Basement Membrane Nephropathy. link to journal article)

26. Specify any characteristics of patients with the medical condition, or suspected of, who are proposed to be eligible for the proposed medical service, including any details of how a patient would be investigated, managed and referred within the Australian health care system in the lead up to being considered eligible for the service:

Individuals who have persistent glomerular haematuria (bleeding from the kidney filter). The algorithm below indicates who should undergo genetic testing in order to confirm the diagnosis of Alport syndrome (as distinct from Thin basement membrane nephropathy) so that ACE inhibitor treatment can be instituted. In addition knowing the mode of inheritance (X-linked or autosomal recessive) will enable appropriate reproductive counselling for the individual and indicate who else in the family should be tested.

Even where a renal biopsy is performed genetic testing has further advantages. It is more accurate (both more sensitive and more specific) than a renal biopsy, indicates the mode of inheritance which has implications for other family members, and also indicates whether renal failure occurs (relatively) early or late.



27. Define and summarise the current clinical management pathway *before* patients would be eligible for the proposed medical service (supplement this summary with an easy to follow flowchart [as an attachment to the Application Form] depicting the current clinical management pathway up to this point):

Persistent lifelong dysmorphic or glomerular haematuria +/- proteinuria. Further evidence is a family history of haematuria, hearing loss, retinopathy or renal failure. No evidence for another cause such as SLE or post-streptococcal glomerulonephritis.

PART 6b – INFORMATION ABOUT THE INTERVENTION

28. Describe the key components and clinical steps involved in delivering the proposed medical service:

Referral to a speciality renal clinic.

1st visit: Assessment by nephrologist and geneticist – confirm hematuria, proteinuria, BP and renal function, construct family tree, check for heating loss and retinopathy, exclude other causes

Undertake consent and genetic testing

2nd visit: Explain diagnosis and consequences, determine who else in family needs testing

Start ACE inhibitors, include mutation on national registry (so that distant family members can undergo cascade testing)

Discharge

29. Does the proposed medical service include a registered trademark component with characteristics that distinguishes it from other similar health components?

Not applicable

30. If the proposed medical service has a prosthesis or device component to it, does it involve a new approach towards managing a particular sub-group of the population with the specific medical condition?

Not applicable

31. If applicable, are there any limitations on the provision of the proposed medical service delivered to the patient (i.e. accessibility, dosage, quantity, duration or frequency):

Once in a lifetime only

32. If applicable, identify any healthcare resources or other medical services that would need to be delivered <u>at the same time</u> as the proposed medical service:

Genetic counselling

33. If applicable, advise which health professionals will primarily deliver the proposed service:

Approved pathologists in accredited pathology testing laboratories

34. If applicable, advise whether the proposed medical service could be delegated or referred to another professional for delivery:

Not applicable

35. If applicable, specify any proposed limitations on who might deliver the proposed medical service, or who might provide a referral for it:

Referred by a nephrologist/geneticist to approved Pathologists in accredited pathology testing laboratories

36. If applicable, advise what type of training or qualifications would be required to perform the proposed service as well as any accreditation requirements to support service delivery:

Approved Pathologists in accredited pathology testing laboratories who whole exome sequencing and variant interpretation

37. (a) Indicate the proposed setting(s) in which the proposed medical service will be delivered (select all relevant settings):

- ☐ Inpatient private hospital
- ☒ Inpatient public hospital
- ☒ Outpatient clinic
- ☐ Emergency Department
- ☒ Consulting rooms
- ☐ Day surgery centre
- ☐ Residential aged care facility
- ☐ Patient's home
- ☒ Laboratory
- ☐ Other – please specify below

Specify further details here

(b) Where the proposed medical service is provided in more than one setting, please describe the rationale related to each:

Patients may be recruited in various settings, usually as part of a multidisciplinary review, that includes a counsellor. However, the genetic testing and analysis will only be performed in a few nation-wide NATA-accredited laboratories.

38. Is the proposed medical service intended to be entirely rendered in Australia?

- ☒ Yes
- ☐ No – please specify below

Specify further details here

PART 6c – INFORMATION ABOUT THE COMPARATOR(S)

39. Nominate the appropriate comparator(s) for the proposed medical service, i.e. how is the proposed population currently managed in the absence of the proposed medical service being available in the Australian health care system (including identifying health care resources that are needed to be delivered at the same time as the comparator service):

Renal biopsy that includes microscopy, immunofluorescence and electron microscopy.

40. Does the medical service that has been nominated as the comparator have an existing MBS item number(s)?

☒ Yes (please provide all relevant MBS item numbers below)

☐ No

Item number 36561

41. Define and summarise the current clinical management pathways that patients may follow *after* they receive the medical service that has been nominated as the comparator (supplement this summary with an easy to follow flowchart [as an attachment to the Application Form] depicting the current clinical management pathway that patients may follow from the point of receiving the comparator onwards including health care resources):

Patients will return to their referring practitioner for ongoing lifelong care that includes ACE inhibition.

42. (a) Will the proposed medical service be used in addition to, or instead of, the nominated comparator(s)?

☒ Yes

☐ No

(b) If yes, please outline the extent of which the current service/comparator is expected to be substituted:

This could replace a renal biopsy (item number 36561) that includes light microscopy, immunofluorescence and electron microscopy. The current cost is $172.50. Renal biopsy has risks with a 4% chance of a major bleed and is insensitive for children and for women with X-linked Alport syndrome, and in anyone with endstage renal failure when the kidney is too scarred to demonstrate the distinctive features. Kidney biopsy is thus less sensitive and less specific than gene testing.

Genetic testing also has the other advantages outlined above (indicates mode of inheritance and other at-risk family members, can be used for cascade testing, indicates likely age at onset of renal failure, can be combined with preimplantation genetic diagnosis).

43. Define and summarise how current clinical management pathways (from the point of service delivery onwards) are expected to change as a consequence of introducing the proposed medical service including variation in health care resources (Refer to Question 39 as baseline):

This is an added specialist service that could be offered at a few centres nationally in combination with a genetic counsellor. The patient would then return to their own nephrologist for ongoing care.

PART 6d – INFORMATION ABOUT THE CLINICAL OUTCOME

44. Summarise the clinical claims for the proposed medical service against the appropriate comparator(s), in terms of consequences for health outcomes (comparative benefits and harms):

The accurate diagnosis of Alport syndrome has many advantages for both the affected individual and the health service. It means that practitioners can begin treatment to delay the onset of renal failure, without the need for a renal biopsy, since ACE inhibitor treatment delays the onset of end-stage renal failure by up to 13 years; it enables accurate reproductive advice for the individual and other family members; family members can be screened; we can predict females who have a higher risk of end stage renal failure from the nature of their mutations; and known mutations can be used in preimplantation genetic diagnosis.

45. Please advise if the overall clinical claim is for:

☒ Superiority

☐ Non-inferiority

46. Below, list the key health outcomes (major and minor – prioritising major key health outcomes first) that will need to be specifically measured in assessing the clinical claim of the proposed medical service versus the comparator:

Safety Outcomes:

With genetic testing, renal biopsies are not needed to make the diagnosis of Alport syndrome. This obviates the risk of bleeding (6%), and the rare likelihood of losing the kidney or death.

Renal biopsies are often equivocal for the diagnosis of Alport syndrome, so the diagnosis may be overlooked.

Clinical Effectiveness Outcomes:

Enables the diagnosis of Alport syndrome to be made with certainty

This means that ACE inhibitor treatment can be started, and delay the onset of end-stage renal failure by up to 13 years.

The nature of the mutations can be used to predict the likelihood of early onset renal failure that necessitates even more aggressive treatment. The nature of the mutations also indicates the type of treatment that should be used in the future, eg chaperones for missense mutations.

Other affected family members can then be found from cascade testing. They also may need ACE inhibitor treatment.

The nature of the mutations will enable reproductive planning.

PART 7 – INFORMATION ABOUT ESTIMATED UTILISATION

47. Estimate the prevalence and/or incidence of the proposed population:

Approximately 5000 patients in Australia

48. Estimate the number of times the proposed medical service(s) would be delivered to a patient per year:

Once in a lifetime

49. How many years would the proposed medical service(s) be required for the patient?

Once in a lifetime. Ongoing treatment with ACE inhibitor treatment.

50. Estimate the projected number of patients who will utilise the proposed medical service(s) for the first full year:

There are currently about 500 Alport patients on the ANZ data registry but we estimate there are 5000 altogether. We expect that we might diagnose an extra 200 Alport families in the first year with genetic testing.

51. Estimate the anticipated uptake of the proposed medical service over the next three years factoring in any constraints in the health system in meeting the needs of the proposed population (such as supply and demand factors) as well as provide commentary on risk of 'leakage' to populations not targeted by the service:

May be 200- 300 families a year.

PART 8 – COST INFORMATION

52. Indicate the likely cost of providing the proposed medical service. Where possible, please provide overall cost and breakdown:

Genetic test cost - $1500 (testing, lab equipment, analysis and reporting, etc)

Cost of venesection and transport to testing lab - $30

Cost of consultation and counselling before and after testing - $300

53. Specify how long the proposed medical service typically takes to perform:

Three months

54. If public funding is sought through the MBS, please draft a proposed MBS item descriptor to define the population and medical service usage characteristics that would define eligibility for MBS funding.

Category (insert proposed category number here) – (insert proposed category description here)
Proposed item descriptor: insert proposed item descriptor here Fee: $1800

PART 9 – FEEDBACK

The Department is interested in your feedback.

55. How long did it take to complete the Application Form?

Eight hours

56. (a) Was the Application Form clear and easy to complete?

☒ Yes

☐ No

(b) If no, provide areas of concern:

Describe areas of concern here

57. (a) Are the associated Guidelines to the Application Form useful?

☐ Yes

☐ No

(b) If no, what areas did you find not to be useful?

Insert feedback here

58. (a) Is there any information that the Department should consider in the future relating to the questions within the Application Form that is not contained in the Application Form?

☐ Yes

☐ No

(b) If yes, please advise:

Insert feedback here