Australian Government

Department of Health

Consultation Survey on MSAC Application 1600

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

This feedback survey relates to the application form and Population, Intervention, Comparator and Outcome (PICO) Confirmation for new and amended requests for public funding (including but not limited to the Medicare Benefits Schedule (MBS)).

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

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

This feedback survey should take approximately 15 minutes to complete.

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

Thank you for taking the time to provide valuable feedback.

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

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

Please reply to the HTA Team:

Email: HTA@health.gov.au

Postal: MDP 959 GPO 9848 ACT 2601

PART 1 – PERSONAL AND ORGANISATIONAL INFORMATION

1. Respondent details

Name: Charmaine Green

Email: admin@pkdaustralia.org

Phone No: 0414130088

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

☐ Individual

✓ ☐ Collective Group

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

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

Polycystic Kidney Disease (PKD) Australia

3. How would you best identify yourself?

☐ General Practitioner

☐ Specialist

☐ Researcher

✓ ☐ Consumer

☐ Care giver

☐ Other

(a) If other, please specify

PART 2 – CLINICAL NEED AND PUBLIC HEALTH SIGNIFICANCE

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

The team at PKD Australia is made up of people affected by the genetic condition PKD, their families, friends, health professionals including nephrologists, geneticists and researchers (as part of PKDA's Scientific Advisory Board). PKD Australia provide support and education to the PKD community and are in regular contact with hundreds of individuals and families with the disease.

PKD is a lifelong, genetic kidney disease, representing a significant burden on the community and on the health care system. PKD affects more than 25,000 Australians and accounts for 6% of new patients requiring renal replacement therapies.

Polycystic Kidney Disease (PKD) is relentlessly progressive. As the disease develops, cysts appear on both kidneys and the kidneys themselves enlarge, often up to five times their normal size, leading to kidney failure.

There are two types of PKD: **A**utosomal **D**ominant **PKD** (ADPKD) and **A**utosomal **R**ecessive **PKD** (ARPKD)

If you have ADPKD there is a 1 in 2 (50 %) chance of passing the faulty gene onto each child. In some cases, ADPKD occurs when there is no family history, the gene abnormality having arisen from a new genetic mutation. Many of those affected may experience regular pain and high blood pressure while over half may develop kidney failure by the time they are 60 years old.

ARPKD is the rarer and more severe form of the disease that is usually diagnosed in utero or at birth and affects approximately 1 in 20,000 individuals. It usually presents as enlarged kidneys with no family history of kidney disease. Both parents need to carry the gene defect for the child to inherit the disease.

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

- help to confirm whether or not a person has PKD. This may be helpful for people who have an atypical presentation, do not quite fulfil the ultrasound diagnostic criteria or those who have a family history of ADPKD but do not yet have signs of ADPKD (This requires prior identification of the genetic variant in a family by testing an affected person in the family with clear diagnosis of ADPKD).
- better understand the condition in the family and may sometimes help predict how the disorder might affect the person in the future.
- may be used to help with predicting the clinical outcome and help to guide clinical management (e.g Tolvaptan for ADPKD)
- inform family planning decisions.

- enable identification of appropriate living kidney donors for transplantation (for patients considering donating a kidney to a family member with ADPKD or if you have ADPKD and a family member is considering donating a kidney to you). This requires prior identification of the genetic variant in your family by testing an affected person in the family with clear diagnosis of ADPKD.

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

Genetic Testing is a very safe and minimally invasive method (requiring only a blood test) of determining a diagnosis which has great accuracy. There are some cases where diagnosis cannot be made e.g findings of variants of uncertain significance or no variant being identified as the cause of the condition. Therefore other methods of clinical diagnosis may still be required. As with any diagnosis of a genetic condition, appropriate support e.g through a genetic counsellor should be offered.

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

Genetic testing is currently paid for by state health systems, but which tests are paid for is totally dependent on the individual hospital. Some hospitals will pay for any genetic testing, some hospitals pay for genetic testing only if it is very likely to change the patient's management, and other hospitals don't pay for any testing. This variability is the reason we are strongly advocating for Medicare-rebatable genetic testing - so all PKD patients in Australia can have the same access to genetic testing, which will become part of the best practice care for the patient.

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

Genetic testing will provide an excellent means of diagnosing PKD and therefore is a service of utmost importance in a clinical care pathway and should be prioritised. Medicare funded Pre-implantation Genetic Diagnosis (PGD) is a logical future step and valuable tool in family planning for those who choose to undergo. This had been previously reviewed by MSAC (Application 1165 - Consultation protocol to guide the assessment of Pre-implantation Genetic Diagnosis). PKD Australia believe decisions on how or whether to have children is a very personal one. For many the decision is down to their lived experience of having the disease and the options available to them. There is no right or wrong choice.

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

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

- [x] Strongly Agree
- [ ] Agree
- [ ] Disagree
- [ ] Strongly Disagree

(a) Specify why or why not:

PKD Australia are in favour for genetic testing for PKD and see the benefits of genetic testing in other forms of genetic kidney disease that are listed in the application. Genetic testing may also help differentiate the diagnosis between different genetic renal diseases.

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

- [x] Yes
- [ ] No

(b) Please explain:

The diagnosis pathway has been well described.

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

- [ ] Strongly Agree
- [x] Agree
- [ ] Disagree
- [ ] Strongly Disagree

We agree that medical imaging is currently the main diagnostic tool (Renal ultrasound and CT scan). We would also like to comment on the use of MRI as a diagnostic and management tool (prognosis). We believe Genetic testing will form part of the best practice care for patients with PKD.

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

✓ ☐ Strongly Agree

☐ Agree

☐ Disagree

☐ Strongly Disagree

(a) Specify why or why not:

We again emphasise the best practice care that genetic testing will bring to those with inherited kidney diseases. In particular for PKD, diagnosis will be improved. This is particularly important for the increase in adult diagnosis of ARPKD, in which genetic testing is invaluable. In ADPKD diagnosis often relies on an age to number of cyst ratio as detected by renal imaging which can be inaccurate and also means diagnosis can only be made in adulthood. The specific genetic cause of the disease can predict outcomes in ADPKD. Genetic variants in PKD1 are associated with a more severe, earlier-onset type of ADPKD, compared to variants in PKD2. This forms part of the PRO-PKD score which is used to help determine clinical management. (please note; a genetic test is not required for treatment with Tolvaptan in Australia under the approved PBS criteria, but it may be used as part of therapeutic decision making in some other countries overseas).

PART 4 – COST INFORMATION FOR THE PROPOSED MEDICAL SERVICE

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

- ☐ Strongly Agree
- ✓ ☐ Agree
- ☐ Disagree
- ☐ Strongly Disagree

(b) Specify why or why not:

We can see an item descriptor in Q 51 and agree with this description

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

- ☐ Strongly Agree
- ✓ ☐ Agree
- ☐ Disagree
- ☐ Strongly Disagree

(c) Specify why or why not:

We can see a fee in Q 51 and agree with this description.

PART 5 – ADDITIONAL COMMENTS

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

There is much research on how genetic testing can improve the treatment and outcomes for renal patients and we strongly support this application

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

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