

Consumer Survey on Genetic/Genomic Testing, Designed and Commissioned By PKD Australia.

November 18th –December 2nd, 2019

Introduction

The Medical Services Advisory Council (MSAC) will meet on December 6th and 7th to review Application #1600 for a Medicare Item Number for genetic testing for inherited renal disease (other than Alport Syndrome).

The team at PKD Australia comprises families affected by ARPKD and ADPKD. Community feedback at Consumer Information Seminars held around Australia, in rural and metropolitan areas, has reflected on the historical absence of hope and the future possibilities for improved therapies for patients and their future children and grandchildren.

Aim

In order to capture community feeling about this, PKD Australia developed a survey inviting respondents to reflect on how the possibility of genetic testing could affect themselves or their families.

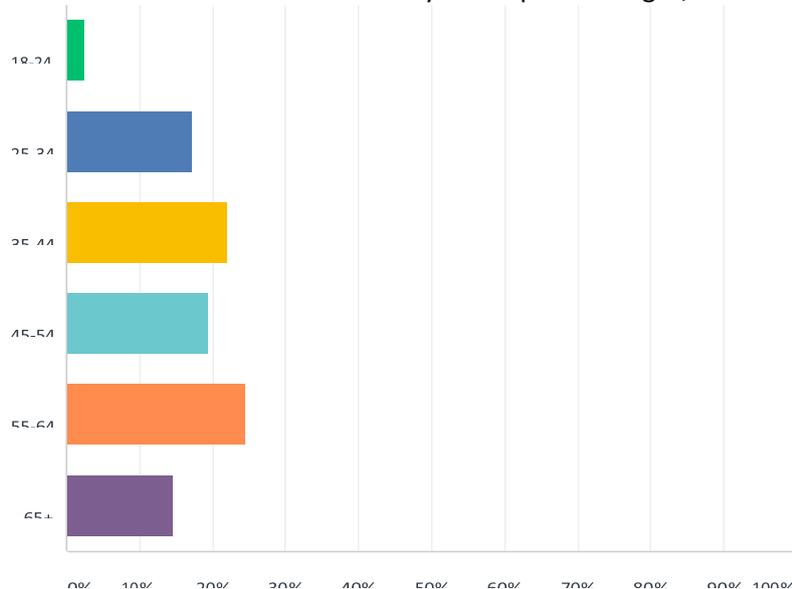
Methods

The survey was developed using Survey Monkey.

Social media was the chief mode of dissemination. An invitation with a link to complete an anonymous survey was sent out on FaceBook, then email and finally in our Newsletter. Spikes in uptake correlated with requests going out. The survey took an average of **3-5 minutes** to complete.

Data analysis

123 people responded to our survey regarding current opinions on genetic testing for PKD in Australia. This survey was developed on Survey Monkey and distributed on social media and via an email newsletter. There was a fairly even spread of ages, as indicated in the graph below:



Most respondents live in metropolitan areas (72.36%) with some respondents living in rural and remote areas.

Of our 123 respondents, 72.36% were people with PKD, 52.85% were relatives of people with PKD, with 1 doctor respondent and several advocates working in the area. Respondents were able to select more than one of these options if appropriate.

Of the 86 patients who responded, they reported the following had been offered to them or a family member:

ANSWER CHOICES	RESPONSES	
Transplant	43.53%	37
Dialysis	52.94%	45
Drug therapy	43.53%	37
Diet and lifestyle strategies	55.29%	47
Genetic testing	21.18%	18

And the following treatments had been used by themselves or a family member

ANSWER CHOICES	RESPONSES	
Transplant	38.82%	33
Dialysis	48.24%	41
Drug therapy	36.47%	31
Diet and lifestyle strategies	61.18%	52
Genetic testing	18.82%	16

There were 27 respondents who were family members of people with PKD and not patients themselves, and they reported that the following management strategies had been offered to their family members:

ANSWER CHOICES	RESPONSES	
Transplant	48.15%	13
Dialysis	40.74%	11
Drug therapy	22.22%	6
Diet and lifestyle strategies	55.56%	15

And the following management strategies had been used:

ANSWER CHOICES	RESPONSES	
Transplant	37.04%	10
Dialysis	37.04%	10
Drug therapy	25.93%	7
Diet and lifestyle strategies	59.26%	16
Genetic testing	33.33%	9

When asked if they would use genetic testing to find a kidney for themselves or a loved one, 94.18% of the patients agreed they would, and 88.89% of family members said they would.

When asked if they were or had been concerned about the possibility of passing PKD to their children or grandchildren, 92.94% of patients and 88.89% of family members said they were.

82.15% of the patients with PKD stated that they or a family member would use genetic testing for family planning, with 88.89% of family members agreeing that they would.

When patients were asked if they agreed with the statement "I have sometimes felt that I am or will be a burden on my family" more than three quarters of the patients said they did, with more than half strongly agreeing with the statement.

When asked if people with PKD should have access to Medicare-funded genetic testing to determine their particular PKD mutation, (which may help doctors to provide the best advice and treatment for people with PKD) 96.52% of respondents agreed that they should, with 92.17% strongly agreeing.

The final question about hopes for research into PKD and how it may change the future path for people with PKD invited a written response and was answered by 103 people. Their responses are attached in Appendix A.

Theme analysis of comments:

A theme analysis of the comments made in response to the question 'what are your hopes for research into PKD and how it may change the future path for people with PKD' revealed that by far the most common hope for respondents was a cure to PKD.

A number of comments rationalised this with the significant impact on quality of life and mental health that this disease has on a patient and their family. Other rationales for a cure included the emphasis on hope for children and grandchildren, and a feeling of guilt for passing on the disease.

Many respondents articulated that the current treatments for PKD, negatively impact on quality of life, including dialysis, transplantation and the side effects of Tolvaptan-the only drug treatment currently available for patients.

The other significant hope referred to by respondents were genetic testing for making life decisions and for medical treatment, genetic testing for family planning, to delay end stage kidney failure and to reduce the financial burden of the disease.

Appendix A has all the responses as they were written.

The following table is a full list of themes and the frequency they were present in responses.

Prevent kidney failure/slow progression:	25
Genetic testing for organ donor:	5
Cure:	41
Genetic testing for family planning:	19
Genetic testing for medical treatment/planning:	29
Improved quality of life:	14
Mental health:	2
Identify cause:	1
New treatments (other than dialysis and transplant):	24
Financial burden reduction (including insurance):	8

Summary

Theme Analysis conveys the extensive burden felt by people with PKD and their families.

The rapid responses to the survey, the respondent's willingness to explore and express family experiences of PKD and the frank revelation of hopes for research indicate a robust consensus in support of the application for a Medicare Item Number for genetic testing for inherited renal diseases, such as PKD.

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APPENDIX A

Responses to final question:

What are your hopes for research into PKD and how it may change the future path for people with PKD?

I hope they find an effective solution to kidney failure and a way to minimize those who reach that stage. My daughter and I both have it and I am almost at transplant stage.

We are a 4 generation ADPKD family and are continuing to grow. All advancement in this area will benefit the 5 of us with the disease, plus those that are yet to come. I will need a transplant at some point and it would be great if genetic testing to find an organ donor was more accessible/affordable. My family members feel the same.

I hope they find a cure.

To find a cure

Genetic testing to be available & integrated into health care in Australia. When contemplating children, we initially paid \$4k to send samples for testing in the US to identify our particular PKD gene. This took close to 6 months and proved inconclusive. We then co-opted extended family and went for linkage testing which took another 6 months or more. All up, we went through around 10 years and half a dozen rounds of IVF to eventually have one child (who is free from PKD). IVF was medically necessary because cysts had made natural conception impossible. Fortunately we were able to fund the treatment ourselves (I stopped counting once it hit \$50k but total cost was somewhere north of there). However, I know much of my extended family would not be in a position to do so. Anything that could be done to avoid others having to go through the same experience would be much appreciated.

My hope would be that we can find a cure for PKD

Genetic testing provides people and families with information and CHOICE. Prevention is better than cure and I want genetic testing to stop the transmission of PKD at my generation.

As a mother of a son who has PKD of course the hope is for early detection and intervention for others which did not happen in his case. Genetic testing is a very important part of the early detection and I hope a simple way of testing would be a great outcome. Obviously more government funding for research into this condition would help and if detected this would give people with the disease and their families information to plan for their future outcomes.

I want a cure

That research and genetic testing may help to prevent anyone suffering from this disease as our family has because of the impact on your everyday living, mental health and ongoing medical treatment for life!

Ideally I want a cure, but that seems an unrealistic hope. I hope to use genetic testing to ensure I don't pass on my genes to my children, if I am able to safely carry children with PKD. I would like to slow the decline of my disease, and live a longer and freer life than my father, who died at 58 after years of dialysis, a transplant, diet and lifestyle restrictions and the burden of knowing at any one time that his health was the best it would be, that it would only get worse from there. Having a degenerative disease has a psychological impact; it having no cure and very few treatments (the effectiveness which is still being determined) makes it hard to have hope. Having a treatment that could slow my disease would give me some hope.

I hope research focuses both on cure and lifestyle to better provide both a solution long term and considerable improvement for sufferers already with the disease.

I want the mutated gene eradicated from future generations. It is a ticking time bomb in our lives and can be eradicated in the next generation to a major extent.

If it can stop the diesel I am for it

My son is a spontaneous victim of PKD. There is no evidence of this disease in either my or my husband's families. He is the father of a beautiful daughter aged 5 & really wants to see her reach adulthood. He has had 2 heart attacks in the last 10 months. I really want to know what we can do to prolong his life. He is the most beautiful person and I would sincerely hope that research can assist him. My GP who has PKD in her family, tells me that our son's sisters would not be suitable as donors, should he need a transplant, as they may develop PKD later in life. I passionately hope research can help our son.

A cure

Realistically the best hope is probably around management and treatments such as artificial kidneys or wearable dialysis machines

Find a cure!

Identify how I got the illness as none of my family have it, and how to prevent from spreading further in my body. Also how to ensure my kids don't have it or prevent the illness

Better treatment becomes available to tackle disease progression, genetic testing is an option for people with family history so children can have these treatment options available before kidneys become enlarged and problematic, eventually there will be a treatment available for DNA in embryo's to stop the disease forming.

Research into a future cure.

I hope that we can find a way to find out early if my daughter has PKD so that she if she does have the disease she can be monitored with her blood pressure and perhaps take a drug to slow the disease. I pray there will be a cure for my daughter.

I desperately hope my children will experience a better quality of life, early intervention, improved treatment to slow the disease more so than my husband has experienced.

To remove the mystery around PKD and provide better access to future care and genetic testing

To find the cure for PKD and PLD as well.

Genetic testing be freely available to permit informed choices to be made. I was diagnosed at age 58 and commenced peritoneal dialysis 15 months later. I did dialysis for 6 months and am blessed to have received a live donor transplant from my cousin and am now 4 years post transplant and doing really well. My donor is also in great health and being monitored by his GP in Canada. I have passed this PKD to my twins who are now 35 years and are being followed up by our GP on an annual basis. Since my diagnosis it has also been revealed that both my brothers have been diagnosed with PKD and that their children have also been diagnosed. More funding should be made available to enable further research into PKD.

Hope research will find a cure

to find a cure

I hope that more viable treatments can be identified and an alternative to transplantation and dialysis can be found.

Being able to prevent cyst growth and associated side effects

More options and more covered

May it find a cure or reversal Of the condition

Eliminating the disease by providing the ability to have children without passing on the mutation.

obviously we would like a cure, or atleast a way to slow the progress

We need such a variety of answers -- from pain relief to cyst prevention to treatment for PLD to the appropriate age at which children should be tested.\\ So international cooperation on research priorities seems sensible.

To find a way to stop this from happening or to be able to replace damaged kidneys without having to donor transplants.

Allow people to know where they stand and what they are dealing with. Help those hoping for children to make informed decisions.

Eventually to be able to remove/significantly lower chance of ADPKD being passed on. Improve transplant success or creation of artificial kidney usable by all blood types.

I hope for a cure or at the least early detection so treatment can start early

A cure would be ideal, better understanding on how the disease works and more encouraging treatment options rather than waiting for the kidneys to die

I hope genetic testing will enable the gene to be eradicated so that the quality of people's lives improves. Alternatively, I hope that organs can be grown synthetically without relying on human donation. I also hope that it becomes illegal to discriminate in the insurance industry based on genetics. I have already been discriminated against with salary continuance insurance no longer offered to me even though I am in the very early stages of polycystic kidney disease.

I hope that one day we can eradicate PKD all together. This may be able to be achieved through genetic testing. My grandchildren will benefit from this.

Hopefully they find a cure one day or something that doesn't require the person to have dialysis or transplant. Less evasive.

a cure

That researchers will find a way to stop PKD and prevent this awful disease ruining the lives of my children and grandchildren, that my siblings and I will be able to access a transplant when needed.

I chose not to have children as I didn't want to pass PKD on. Unfortunately other members of my family didn't and I worry about those kids future.

I'm hoping I can manage my PKD and not require a transplant a hoping scientific research and our public healthcare system can help make that happen!

That the research will find a cure but will provide the information necessary to allow informed choices to be made

My hopes are that PKD can be become a treatable condition in my lifetime.

Hope genetic testing can be used to eliminate the disease and assist with transplants for those of us who already have it

I hope research will find a cure for PKD so that future generations will not have the pain and debilitation caused by this horrid disease

research into: Genetic testing is a must, to understand the disease and also the management of it. integrative health, diet, exerices, water consumption. webiste specliast knowledge looking at the current treatment, side effect etc. A live site. I could actually run this but I'm not medially trained so need to do this alongside with a medic. I spent hours trying to understand this condition myself. I'm sure so many other people have too. law to be changed: that by law doctors have to tell immediate relatives if their patient is diagnosed with PKD or PLD.

Earlier detection means things can be put in place to slow progression

It would be good to gene map people in such a way that people can decide whether to have children knowing the risks of passing PKD on to them

Treat PKD in genetic level.

To stop the disease from progressing into danger zone

More early intervention to stop advanced disease

Finding a cure would be awesome

To find a cure

Research to find a drug without horrendous side effects that could 'switch off' the disease process. Plus research into an implantable artificial kidney

I hope that they can find a cure and until then at least have a way to prevent the decline in our kidney function. I have tried the medication Tolvaptan and it seemed to be working but I had a reaction to the medication and had to stop.

Early genetic testing, at at 18 will allow patients to maximize good lifestyle and nutrition choices to hopefully slow down the progression. And hopefully a drug will be approved that a patient could start taking early in life to slow progression

My husband and I and are at the stage where we are ready for kids but I don't want to pass PKD onto them so we have met with a geneticist and fertility doctor to see what our options are. This path feels like a long and daunting one. My hope is if we go down this path our kids will never have to go through the same process or worry about getting PKD at all.

Research into effective medication and lifestyle management that is accessible and affordable to all. The hope to eliminate dialysis and transplant.

I hope that a cure will be available in the future

I hope a cure will be found so i have long life to see my children become parents and manage pkd effectively if my children have acquired it.

More research on lifestyle changes that can be made to slow/halt progress.

My hope would be that a cure would be found for PKD, so it could be eliminated. Until that happens genetic testing is the only way to eliminate this awful disease. The cost of genetic testing is way less than the long term cost of medication/ dialysis and inability to work and pay taxes. We have already assisted family members to have genetic testing to ensure their children did not have the PKD gene. This cost us \$16000 out of pocket, as it is currently only partially covered by Medicare . This is currently the only way to eliminate PKD.

To lessen the impact on the person & family. For family planning so the disease can be eradicated. For better treatments.

At this current time patients are not told a lot about the different types of PKD nor how this may impact on their future. I would personally be interested in finding out which type of PKD myself and my son have (I suspect it would be the same as my brother and father) but it has never been offered to me. I would only be able to partake in this testing if it was bulk billed. Kidney disease is increasing quickly in Australia and a great financial burden on our healthcare system. Any future treatment that can be genetically directed individually is going to be money well spent, more successful and lessen the burden on the system in the long term.

I don't have hopes for research. My diagnosis in 1991 was purely accidental, so I hope that doctors aka general practitoners in future, learn about pkd.

Discover treatment to prevent PKD progressing to ESKD, as well as something like gene editing to prevent disease being inherited. These will allow people to lead normal productive lives.

Eliminate renal failure

I hope that research will be fully funded and a solution evolves that cures pkd.

I'm hoping for a cure. I have pkd have had a transplant but my live is going to be a big problem for me in the future and both my children have pkd so hope there will be a cure for them

An easily accessible, affordable treatment to increase quality and length of life for people with pkd. A cure would be amazing but, I believe, not possible, at least in my lifetime

Diagnosis early. Research grants from Government Medicare rebates for sufferers Find cures

Treatment to slow or fix the disease

Genetic testing to provide pt with an option of kidney transplants sooner than when waiting when the pt renal function is so low. PKD sufferers can have multiple co morbidities and it can be too late for a transplant if had to wait for because of kidney function levels and the trial of dialysis. I have a 4 year old that I would love to not have a worry for when she is older. If I new there were options prior to conceiving as just told by RCH melb I would have done all I can prior. Although now I need to work on the now and consider every option and genetic testing would help change everything

More research to better understand the disease and earlier diagnosis

I hope most strongly this will be available soon.

In the near future a means of shrinking cysts in existing PKD patients. Long term a way of preventing this disease from being passed on to future generations.

More research into treatment for ARPKD and identifying disease causing genes for family planning.

Hopefully we can eradicate PKD so that it is not passed on to future generations

My hopes are that we can find a way provide care and treatment for people living with PKD that allows them to live their lives how they wish.

Drugs to slow cyst growth with less side effects than tolvaptan

A cure would be wonderful but realistically slowing progression and better management of symptoms eg pain, hypertension, depression etc.

I hope research will find a way to reduce the size and number of cysts, or that doctors will eventually be able to stop the growth of cysts at a time when kidney function is normal.

My hopes are to find a cure so that transplants and dialysis will no longer be needed

It would help understand where the disease began as well as inform personalised treatment and drug options

Avoiding transplant and lengthening the life span.

People need as much information as they can to make better decision

Understanding the disease. Better treatment.

Hopefully they will find a cure so this disease will stop

I hope research will either limit progression of PKD without significant secondary health concerns, or eliminate PKD altogether

I hope that there will one be a cure or a preventive measure that will dramatically slow down the progression of PKD

I donated to my husband but my daughter has inherited this horrid disease. What sort of a future does her future hold. I hope for a miracle, treatment and a cure

I do hope that genetics will be the answer to this incurable disease that has afflicted the four generations of my family, (that I am aware of); my grandfather, my mother (both passed at 48 and 49 years of age respectively), myself (at 56 with only 32% renal function) and my only two children (both diagnosed but with NO available treatment for them). It is critical that genetic testing be part of an encompassing array of supporting health & medical testing for those afflicted.

A cure for this! I have PKD, which I got off my dad. He has just had a port in place getting ready for dialysis as his kidney function has dropped down to 14%, dropping 7% in 3 months. I hate seeing dad how he is now. And it makes me worry about what I am going to be like at his age due to my own PKD. I have also decided against having children of my own as I do not want to pass this down.

Early detection and a treatment plan before kidney function becomes so low dialysis or transplant are the only option

Artificial kidney perhaps

1) Pre implantation genetic diagnosis to STOP the disease passed to the next generation 2) better treatment options for patients with ADPKD

Knowledge of the affected gene will help my family- sons and daughters, prepare for their future- assisting job-choice, life-style, superannuation, family planning, sport and leisure pursuits.



PKD
Australia

Survey on Genetic Testing for PKD in Australia

Please select your age range:

- 18-24 45-54
 25-34 55-64
 35-44 65+

Please select the best answer to match the area you live in:

- Metropolitan
 Rural
 Remote
 Other (please specify)

How did you hear about this survey?

- Website
 Email
 Doctor
 Word of mouth
 Other (please specify)

Please select however many options fit you:

- I am a person with PKD
- I am a relative of a person with PKD
- I am a doctor or allied health professional working with PKD
- I am an advocate working in the area of PKD
- Other (please specify)



Survey on Genetic Testing for PKD in Australia

What management options have been offered to you or your family members with PKD? (Please choose all that apply)

- | | |
|---|--|
| <input type="checkbox"/> Transplant | <input type="checkbox"/> Diet and lifestyle strategies |
| <input type="checkbox"/> Dialysis | <input type="checkbox"/> Genetic testing |
| <input type="checkbox"/> Drug therapy | |
| <input type="checkbox"/> Other (please specify) | |

What management strategies have been used for you or a family member with PKD? (Please choose all that apply)

- | | |
|---|--|
| <input type="checkbox"/> Transplant | <input type="checkbox"/> Diet and lifestyle strategies |
| <input type="checkbox"/> Dialysis | <input type="checkbox"/> Genetic testing |
| <input type="checkbox"/> Drug therapy | |
| <input type="checkbox"/> Other (please specify) | |

Please rate your level of agreement with the following statement: I would use genetic testing to find a suitable kidney donor for myself or a loved one.

- | | |
|--|---|
| <input type="radio"/> Strongly agree | <input type="radio"/> Disagree |
| <input type="radio"/> Agree | <input type="radio"/> Strongly disagree |
| <input type="radio"/> Neither agree nor disagree | |

Please rate your level of agreement with the following statement: I am concerned/have been concerned about passing PKD on to my children or grandchildren

- Strongly agree
- Disagree
- Agree
- Strongly disagree
- Neither agree nor disagree

Please rate your level of agreement with the following statement: I or a family member would be interested in genetic testing for family planning

- Strongly agree
- Disagree
- Agree
- Strongly disagree
- Neither agree nor disagree

Please rate your level of agreement with the following statement: I have sometimes felt that I am or will be a burden on my family

- Strongly agree
- Agree
- Neither agree nor disagree
- Disagree
- Strongly disagree



Survey on Genetic Testing for PKD in Australia

What management options have been offered to you or your family members with PKD? (Please choose all that apply)

- | | |
|---|--|
| <input type="checkbox"/> Transplant | <input type="checkbox"/> Diet and lifestyle strategies |
| <input type="checkbox"/> Dialysis | <input type="checkbox"/> Genetic testing |
| <input type="checkbox"/> Drug therapy | |
| <input type="checkbox"/> Other (please specify) | |

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- | | |
|---|--|
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- | | |
|--|---|
| <input type="radio"/> Strongly agree | <input type="radio"/> Disagree |
| <input type="radio"/> Agree | <input type="radio"/> Strongly disagree |
| <input type="radio"/> Neither agree nor disagree | |



Survey on Genetic Testing for PKD in Australia

Please rate your level of agreement with the following statement: People with PKD should have access to Medicare-funded genetic testing to determine their particular PKD mutation. (This may help doctors to provide the best advice and treatment for people with PKD)

- Strongly agree
- Disagree
- Agree
- Strongly disagree
- Neither agree nor disagree

What are your hopes for research into PKD and how it may change the future path for people with PKD?