

GENETIC TESTING FOR POLYCYSTIC KIDNEY DISEASE

What is polycystic kidney disease?

Polycystic kidney disease (PKD) is a rare inherited condition in which many fluid-filled sacs (cysts) form in the kidneys. As the cysts grow, the kidneys get bigger and don't work as well. Sometimes, this can cause kidney failure.

Although there is currently no cure for PKD, early detection and treatment can reduce some of the symptoms and help maintain kidney function. There are two main types of PKD: autosomal recessive polycystic kidney disease (ARPKD) and the most common form, autosomal dominant polycystic kidney disease (ADPKD).

Autosomal dominant refers to the way the genetic variant is inherited in families. It means that children who have a parent with ADPKD will each have a 1 in 2 (50%) chance of inheriting the genetic variant causing ADPKD. When someone is found to have a genetic variant causing ADPKD, it has usually been inherited from a parent. However, in some rare cases, the genetic variant may occur for the first time in that person. In around 10% of people with PKD, no known genetic cause can be found.

Why has my specialist recommended this test?

Your symptoms, medical test results, and possibly your family history indicate that you might have PKD. PKD is mostly caused by changes (variants) in genes that are important for kidney function and development.

The Genome.One PKD genetic test will analyse a group of genes associated with large kidney cysts. Over 90% of people with ADPKD will have variants in the genes PKD1 or PKD2. An additional seven genes (HNF1B, GANAB, PKHD1, TSC1, TSC2, OFD1, UMOD) are also analysed. These genes are associated with other conditions that can cause kidney cysts. Knowing which genetic variant is causing PKD will help your doctor to confirm the diagnosis.

What are the possible outcomes?

Genetic testing involves looking at many variants in several genes to help identify the ones that might cause PKD.

There are three potential outcomes to consider:

- **VARIANT FOUND**

A variant is found in a gene that causes PKD. We recommend you talk to your specialist about how this may impact you, your treatment and your family. Other family members may now be referred to a genetic specialist and offered genetic testing to determine if they have or have not inherited the same variant.

- **NO VARIANT FOUND**

No variants were found in any of the genes tested. This does not change your clinical diagnosis or ongoing management. There may still be a variant in a gene not analysed in this test or as yet unknown to science. Your specialist can request to have your genome reanalysed in the future to see if additional variants have been discovered that might be relevant to you.*

- **VARIANT OF UNCERTAIN SIGNIFICANCE (VUS)**

A genetic variant is identified in a gene, but there is uncertainty about whether it actually causes PKD, either in you or in others. This does not change any clinical diagnosis you may have. Your specialist can request that the VUS be reviewed in the future to see if more information about its meaning has been identified.*

What are the benefits of having genetic testing for PKD?

PKD can be passed on through your genes, meaning that there may be implications for both yourself and close family members to consider.

- **Possible benefits for me**
 - Confirmation of your PKD diagnosis, or discovery of the exact genetic cause
 - Information that you and your specialist can use to make decisions about management and treatment options for PKD
 - Information that can be used to help predict the severity of your PKD. Genetic variants in some genes are associated with more severe types of PKD than others
- **Possible benefits for my family**
 - Predictive testing in family members to determine if they have or have not inherited the same variant
 - The option for earlier clinical screening in relatives who may have inherited the same variant, as well as earlier diagnosis and treatment if relevant
 - The avoidance of unnecessary treatment in those who have not inherited the same variant
 - The option for screening of family members as potential kidney donors
 - Guidance for family planning (Genome.One does not offer preimplantation genetic testing)

How do I arrange a test?



1. Your specialist will refer you for testing by completing the Genome.One request form.



2. Your specialist will arrange for a genetic health professional to talk to you about the practicalities and implications of genetic testing. If you decide to proceed with the test you will need to sign a patient consent form.



3. Next you need to provide a blood sample at any pathology collection center that will be sent to Genome.One for testing.



4. Your results will be summarised in a detailed medical report that will be sent to your specialist within 8-12 weeks. Your specialist will then discuss the report with you.

How much does the test cost?

Please contact Genome.One for current pricing. Contact details are available on the back of this sheet.

Tests offered by Genome.One are not currently eligible for Medicare benefits and are not covered by private health insurance.

What happens next if a variant is identified?

Your specialist is the best person to talk to about your genetic testing results and how they influence your ongoing management.

There may also be implications for your family members, and genetic counselling is recommended for you and your family. Genetic counselling is helpful when talking to your family about your genetic test results and the implications for them.

Genetic counselling for PKD

The Human Genetics Society of Australasia recommends genetic counselling when you undergo genetic testing for PKD. Genetic counselling aims to provide you and your family with information and support as you work through the decision to have genetic testing, and can help you understand and adjust to the genetic results. Your specialist will be able to refer you to a genetic counselling service.

Genome.One can offer genetic counselling to individuals both before and after undergoing genetic testing with us at no additional cost. Please contact us if you would like to access this service.

Additional information about genetic counselling and genetic testing can be found in Fact Sheet 6 on the Centre for Genetics Education website www.genetics.edu.au

How is my genetic information stored?

Your blood or saliva sample is held securely in the Genome.One laboratory in accordance with applicable law and standard laboratory practice.

Your personal information is securely stored in paper-based and electronic records in the Genome.One laboratory in accordance with the Genome.One privacy policy and applicable law. Your genomic data is held on servers in the United States of America for the purposes of storage and highly specialised data processing and analysis.

However, before any data is transmitted to the USA, information identifying you is removed and replaced with a secure code. Only staff at Genome.One have the ability to link the code with your personal information.

Information from your test will not be shared with other people without your permission, except where this is required or permitted by law. Information from genetic testing is important for medical research about the genetic causes of disease. When providing consent, you have a choice whether your deidentified data can also be shared with researchers or included in research data banks.

The full Genome.One privacy policy is available at www.genome.one/privacy-policy

How will genetic testing affect my insurance?

Under current Australian law, your private health insurance will not be affected by your decision to have genetic testing, or by any result of genetic testing.

However, genetic testing can have implications for your ability to obtain risk-rated insurance policies, including life insurance, disability insurance and income protection insurance. This could include denial of cover, exclusion from cover or increased premiums.

More information on how genetic testing affects your insurance can be found in Fact Sheet 20 on the Centre for Genetics Education website

www.genetics.edu.au

Patients outside of Australia should obtain advice about the implications of genetic testing for insurance in their home country.

Contact details

Genome.One

370 Victoria St Darlinghurst NSW 2010 Australia

P 1300 G-NOME-1 (1300 466631)

E enquiries@genome.one

Further information and support

PKD Foundation Australia

www.pkdaustralia.org

Kidney Health Australia

www.kidney.org.au

PKD Foundation

www.pkdcure.org

This fact sheet is intended as a general introduction to the topic and is not meant to substitute your health professional's advice. It should be noted that each person's experience is individual and that variations do occur in treatment and management due to personal circumstances, the health professional and one's place of residence. Always consult your doctor if you require further information.

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