

# Patient information for the *APOL1* Genotyping Program\*

## 1. What is the *APOL1* gene?

**A:** The *APOL1* gene is important to many different organs in the body, including the kidneys. Everyone has two copies of the *APOL1* gene. However, when there are changes or variations in both copies of the *APOL1* gene, there is an increased chance for an individual to develop a genetic form of kidney disease called *APOL1*-mediated kidney disease (AMKD).

## 2. What is AMKD?

**A:** Over the past 3,000-10,000 years, the *APOL1* gene evolved in specific ways in people who lived in sub-Saharan Africa to protect them from resistant forms of an infectious parasite. This evolution resulted in the emergence of “genetic variants” or “risk variants” of the *APOL1* gene.

In 2010, scientists found that people with two *APOL1* risk variants face increased risk of kidney disease. AMKD is a progressive form of chronic kidney disease (CKD) that leads to rapidly declining kidney function. AMKD is a genetic form of kidney disease and is not your fault.

## 3. Why should I consider testing for *APOL1*?

**A:** While anyone can have a change in their *APOL1* gene, the G1 and G2 variants appear more frequently in people of African descent. Approximately 10%-15% of Black Americans have the G1 or G2 variants in both copies of their genes, putting them at a higher risk for developing AMKD. The purpose of *APOL1* genetic testing is to identify people who may be at increased risk for AMKD to help inform disease management.

## 4. Will there be a cost to me for *APOL1* testing?

**A:** The *APOL1* Genotyping test is currently available to eligible patients at no cost through a partnership between Vertex Pharmaceuticals and Labcorp. Insurance will not be billed and you will not be responsible for the cost of testing. You are eligible for the program if you:

- Have African ancestry
- Have been diagnosed with CKD but do not have diabetes
- Have not had a kidney transplant
- Have not undergone dialysis

Additional terms and conditions apply.

## 5. What changes could be found on my results?

Possible results—variants	Result summary	Impact on kidney function
G0/G0	Negative	No increased risk for CKD
G0/G1 or G0/G2	Negative	No increased risk for CKD
G1/G1 or G2/G2 or G1/G2	Positive	Increased lifetime risk for CKD

## 6. What do the results mean for my health and my family?

**A: Positive results**—Results with any combination of G1 or G2 variants in both genes are considered positive. Positive results in patients who are already diagnosed with CKD may indicate that you have AMKD. If you have AMKD, your kidney disease may get worse faster than other types of kidney disease. Based on these results, you should speak with your healthcare provider about available options for monitoring and treatment, including relevant clinical trial options. Your family members may also be at increased risk and can be informed about the testing options.

**Negative results**—If you are found to have only one G1 or G2 variant and have at least one typical copy of your gene (called G0), your risk for AMKD has not been found to be higher than anyone else in the general population with your same ancestry. If your results are negative (both genes are G0), your risk for AMKD is not increased based on the results. However, there are other possible genetic and environmental causes for CKD, so you should continue to speak with your healthcare provider.

## 7. What should I expect after testing is complete?

**A:** Results will be delivered to your ordering healthcare provider and the Labcorp Patient™ portal within approximately 5-7 days from the time the sample is received at the lab. You should consult your healthcare provider for any questions or follow-up.

## 8. Is genetic counseling available?

**A: If your results are positive**, you will receive a phone call from a Labcorp genetic counselor. The incoming call will be from (215) 873-2078. On the initial call, the genetic counselor will schedule a time to meet virtually via phone or video with you and any family members or other individuals you would like to include.

**If your results are negative**, you will not automatically be contacted by a genetic counselor; however, genetic counseling is available to you if you would like additional information. Please contact Labcorp at (215) 873-2078 to schedule time with a genetic counselor.

\*Vertex Pharmaceuticals is sponsoring this *APOL1* Genotyping Program in collaboration with Labcorp, who will perform *APOL1* genotyping and genetic counseling for eligible patients at no cost. Program is subject to change or discontinuation without notice. Additional terms and conditions apply. Participating patients, as well as their treating health care providers, are not required to order, purchase, prescribe and/or obtain any other product or service from Vertex Pharmaceuticals, Labcorp or any of their affiliates.



# Patient information for apolipoprotein L1 (*APOL1*) genetic testing

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## Common concerns about genetic testing

### 1. Who will have access to my genetic results?

**A:** *APOL1* test results are confidential and will be sent only to you and your healthcare provider unless otherwise authorized by you. Test results and protected health information (PHI) are protected by numerous state and federal laws including but not limited to the Health Insurance Portability and Accountability Act (HIPAA). HIPAA is a federal law that gives you rights over your health information and sets rules regarding who can access your PHI and how it can be used. Labcorp will only share aggregated, deidentified metrics with Vertex pharmaceuticals

### 2. How will my genetic material be used?

**A:** Your lab sample will be tested at Labcorp with the single-gene *APOL1* test. After testing, we will store your sample for a minimum of 30 days and a maximum of 60 days. After that time, samples are destroyed.

### 3. Can the results of my genetic testing be used against me by my employer or health insurance provider?

**A:** No, they cannot. In 2008, the Genetic Information Nondiscrimination Act (GINA) was signed into law. GINA protects against discrimination based on genetic information. The law prevents employers and health insurers from asking about genetic information or from using genetic information to make work or insurance decisions. Because of GINA, covered health insurers in the group market cannot use genetic information to determine eligibility, cost, coverage, or benefits of a health insurance policy based on your genetic test results. GINA does not cover life, disability, or long-term care insurance. There are also exceptions for employers with less than 15 employees and individuals receiving insurance through federal government or military.

### 4. How can my genetic test results impact my family?

**A:** GINA protections extend to all genetic information from an individual and their family members. Safeguards are in place around genetic tests and family history. GINA was enacted to encourage Americans to take advantage of genetic testing for their medical care. Your genetic results may prompt your family members to be screened. For example, if you have a positive *APOL1* result, your family members may also be at an increased risk and can be informed about testing options.

For more information on GINA,  
scan the QR code or visit  
<https://www.ashg.org/advocacy/gina/>

