

Importance and Expectations of Genetic Testing

Genes provide instructions that tell our bodies how to develop and function. Although the genetic instructions between us are similar, we all carry genetic differences (or variants) that are unique!

Sometimes, these genetic variants cause genes to stop working or work differently than expected. In some cases, this can result in a genetic (inherited) disease.



Disease management was changed in

40%

of patients with a primary immunodeficiency after reaching a genetic diagnosis^{1*}

*Reported from a study of 187 patients and family members that underwent genetic testing

What genetic testing may tell me:



Assist my doctor in determining a diagnosis



Assist me in making informed decisions about my health and medical management



Help understand the chance that a current or future pregnancy may have a genetic disorder



Inform me if other family members should be genetically tested

Getting a genetic test is easy

1

Talk to your healthcare provider

- Your doctor will order the genetic test
- Genetic counseling can be ordered

2

Provide a sample

- Samples can be saliva, blood, or a swab of your gum, depending on the test you are completing
- Either you or your physician's office staff will send the sample to the genetic lab for testing

3

Speak with a genetic counselor

- If your physician's office has ordered genetic counseling, you should receive a call from them
- If your physician did **not** order genetic counseling, reach back out to your physician's office and ask them to refer you to a genetic counselor
- A genetic counselor can help answer your questions before the genetic test as well as help you understand your results

4

Receive a summary

- A summary of your results will be provided to you and your physician

What are my next steps?

If there is a genetic diagnosis, it is important to review this information with your provider. You can learn more about your genetic diagnosis and determine whether other blood relatives may be at risk for the disease. Genetic testing may be considered for additional blood relatives including aunts, uncles, grandparents, cousins, etc.



Connect with an APDS Clinical Educator (ACE)

- Help you navigate questions that you or your family members have related to genetic testing
- Can assist with clinical resources such as Family Tree Mapping
- Note, ACEs do not offer medical or treatment advice or replace discussions with a doctor



SCAN ME

Reference

1. Quinn J, et al. *Immunol Res.* 2020;68(3):126-134.