

ALS & GENETICS

A Les Turner ALS Foundation Guide for People Living with ALS

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Disclaimer Statement: The information in this guide is not medical advice. Talk to your ALS care team before making any decisions about your health or treatment. Together, you and your care team can find a treatment plan that works for you.

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All people living with ALS, regardless of clinical presentation or family history, should have the option of genetic counseling and genetic testing.

What is a genetic counselor?

A genetic counselor is a health care provider who specializes in genes and how they affect our health. They are here to help you before, during, and after the genetic testing process. Your doctor and/or genetic counselor will provide you with the education and support you need to make an informed decision about genetic testing. If you choose to undergo genetic testing, your doctor or counselor will help you understand the implications and determine next steps.

To schedule an appointment with a genetic counselor at Northwestern Medicine, please call 312.695.7950.

If you are looking to find a genetic counselor in your area, talk to your ALS Care Team or contact [The National Society of Genetic Counselors \(aboutgeneticcounselors.com\)](https://www.aboutgeneticcounselors.com).

For people living with ALS



The **My ALS Decision Tool™**, a first-of-its-kind in the US, online, interactive guide, can walk you through the genetic testing process, the benefits and downsides of genetic testing, and help you make an informed decision. The next time you meet with your ALS care team you can provide them with your decision and ask about next steps.

lesturnerals.org/genetic/about-this-decision-tool

When using The National Society of Genetic Counselors search feature, first choose if you're interested in in-person counseling or telehealth. Then choose the state that you live in, click the specialization of "adult (including complex disease)" and "neurogenic," then hit search for a list of counselors.

Evidence-Based Consensus Guidelines on Genetic Counseling and Testing in ALS

There are over 40 different known genes that are associated with ALS, and researchers are still discovering more. The practice of genetic testing in ALS has been impacted by high patient demand for testing, sponsored testing programs, and a newly approved FDA gene-targeted therapy in ALS. However, many challenges exist, including few genetic counselors in ALS clinics, and differences with laboratory methods and reporting.



To learn more **watch** Jennifer Roggenbuck, MS, CGC, from The Ohio State University, Wexner Medical Center, discuss new evidence-based, consensus guidelines for genetic counseling and testing in ALS.

alslearningseries.org

How does genetic testing work?

If you choose to get genetic testing, you'll meet with a doctor or a genetic counselor (a health care provider who specializes in genes and how they affect our health). After you discuss the possible pros and cons of genetic testing with your doctor or genetic counselor, they'll order the tests you need.

Here's how the genetic testing process works:



Step 1

Your doctor or counselor will ask you to provide a DNA sample. This could be a sample of your blood or saliva.



Step 2

Your doctor or counselor will send your sample to a lab. The genetic testing lab will "read" your DNA and look for pathogenic variants (differences in specific genes that may cause ALS). This step usually takes 4-6 weeks.



Step 3

Your doctor or counselor will explain your testing results. They may also tell you about treatment options or offer guidance to help you and your family plan for the future. You can ask your doctor or counselor for a copy of your testing results. **It's a good idea to save your results in a safe place, so you or your loved ones can look back at them later.**

What are the results of genetic testing?

When you receive your genetic testing results, the report will say any of the following:



Positive result

Means that a pathogenic variant related to ALS was found in your DNA.



Negative result

Means that no pathogenic variant related to ALS was found in your DNA.



Uncertain result

Referred to as a Variant of Uncertain Significance (VUS). This means that a variant was found, but it is not currently known to be related to ALS.

What can I learn from genetic testing?

With genetic testing you may be able to learn:



If the type of ALS you have is linked to a pathogenic variant



If you can participate in certain clinical trials (research studies where participants try new medical treatments) or other research studies



If there are more effective ways to manage your ALS symptoms



A pathogenic variant is a difference in specific genes that can cause people to develop ALS. Genetic testing may tell you if your specific type of ALS was linked to a pathogenic variant.

In 2023, the FDA granted accelerated approval of Biogen's Qalsody (**qalsody.com**) for treatment for SOD1-ALS. Qalsody is the first FDA-approved treatment to target a genetic cause of ALS. Talk to your ALS care team to learn more about Qalsody.





If you're likely to experience other symptoms in the future, like cognitive changes (changes in how you think and behave)



If your children or other family members are more likely to develop ALS and related conditions

There are over 40 different known genes that are associated with ALS, and researchers are still discovering more. The most common are:

SOD1
TARDBP

FUS
C9ORF72



Frontotemporal degeneration or FTD is a term used to describe significant cognitive changes related to ALS and other neurological conditions. FTD can affect the way people think, feel, and interact with others. To learn more about FTD please visit theaftd.org.

How much does genetic testing cost?

While genetic testing has historically been expensive, the costs have significantly decreased over time. Your genetic counselor can provide you with testing options and help gather information about anticipated costs. Some genetic testing companies may work with your insurance company to cover the cost, too.

You can also ask your counselor about programs that provide genetic testing at no cost.

Emotions associated with genetic testing

The genetic testing process may be overwhelming or confusing at times. You may feel anxious as you wait for your results. Learning about your results may bring up difficult emotions for you and your loved ones, too.

For example, you might feel shocked or relieved to get answers about the cause of your ALS. Or, if you don't get the information you were hoping to learn, you might feel sad or frustrated. If you choose to share genetic testing results with your family members, they may have strong feelings about your results and their own risk of developing ALS. Some people may choose not to get genetic testing for these or other reasons.



No matter what you decide, your ALS care team is here to listen and support your decision.

Tips for sharing your genetic testing results with family

The genetic testing process can be overwhelming or confusing at times. You may feel anxious as you wait for your results. Learning about your results may bring up difficult emotions for you and your loved ones, too.

When sharing genetic testing results with your family members, they may have strong feelings about your results and their own risk of developing ALS.



Remember, you get to choose:

Whether to have genetic testing

What to do with the information you learn from genetic testing

Who to share your results with

**LES
TURNER
ALS
FOUNDATION**

Here are some helpful tips for sharing your genetic testing results with family, if you choose to.



You are not alone. A genetic counselor can help you every step of the way. Genetic counselors are also available for other family members who want to discuss their risks.



You are in control. You can decide whether you want to share your results and with whom.



Practice what you want to say. Write down a list of things you want to cover. You don't need to have all the answers, and you don't need to explain everything in one sitting.



Find the right time. Make sure you and the person you are telling are not distracted or in the middle of something. A quiet space with undivided attention is best.

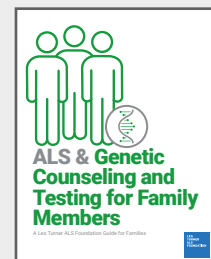


Consider writing a letter or email, if having a conversation may be too difficult. Genetic counselors can also write letters that explain the testing, the results, and their meaning that you can share with your family.



To learn more about genetic counseling and predictive genetic testing for people at risk of developing ALS and other conditions, watch this webinar from our ALS Learning Series by Laynie Dratch, MsC, CGC, genetic counselor at the Penn ALS and FTD clinics.

alslearningseries.org



For more information about genetic counseling, read the **ALS & Genetic Counseling and Testing for Family Members** Guide.

lesturnerals.org/resources

■ Additional resources

For more in-depth information on ALS and genetics, check out these resource guides from the International Alliance of ALS/MND Associations: www.als-mnd.org/support-for-pals-cals/clinical-care/genetic-counselling-and-testing/

The ALS Trial Navigator (ALS.net/ALS-Trial-Navigator) is a comprehensive resource that guides people living with ALS as they learn more about finding and participating in clinical trials for ALS. After answering a few questions, you can learn about research opportunities, set personal priorities, and find trials that meet your criteria. The Navigator also has an interactive map to help you find trials in your area.

Notes

Notes

Learn more

The Les Turner ALS Foundation exists to guide you to answers, support you and your loved ones and advance scientific research. To learn more about living with ALS visit, lesturnerals.org/resources.

My ALS Decision Tool™

If you have ALS, you will need to make some important decisions about your health care. As your disease progresses, your ALS care team may recommend different care options. You can use this tool to learn about some common ALS treatments, answer a few questions to help you think through what is most important to you and get ready to talk with your ALS care team about your options. To learn more, visit: alsdecisions.org.

**MY ALS
DECISION
TOOL**

ALS Learning Series

Our online ALS Learning Series aims to empower the ALS community through the latest information and insights. Educational webinars and interactive Q&A's covering a diverse array of topics, from nutrition to respiratory care, are offered monthly featuring members of the Foundation's Support Services team, our Lois Insolia ALS Clinic at Northwestern Medicine and other national ALS experts. To learn more about ALS care and research, visit: alslearningseries.org.

**ALS
LEARNING
SERIES**

My ALS Communication Passport to Quality Care

My ALS Communication Passport to Quality Care was created to make your life easier. You will be able to share health information and care preferences with caregivers. You have a lot of information to keep track of, and this tool will help you do that. To find out more, visit: lesturnerals.org/passport.

**MY ALS
COMMUNICATION
PASSPORT
TO QUALITY CARE**

Support Groups

We facilitate support groups to provide people living with ALS, their caregivers and family the opportunity to share their experiences, give encouragement and help each other navigate their journey with ALS. To find out more, visit: lesturnerals.org/support-groups.

