

Genetic Testing



Everyone diagnosed with ALS has the option to have genetic counseling and genetic testing.

This is a very personal decision, and understanding the potential benefits and drawbacks is important.

What is genetic testing?

Genetic testing looks for harmful changes in your DNA, the information inside your cells that makes you who you are. Genetic tests are usually performed on a saliva sample, cheek swab or blood sample.

What are the potential benefits of testing?

Your results may help you better understand why you got ALS, how it might progress, your family members' potential risk and options for family planning. Knowing that you have a specific ALS-linked gene mutation could make you eligible for clinical trials or an FDA-approved gene-targeted therapy.

What are some potential drawbacks?

The results of genetic testing may lead people to feel complex emotions, including anger, sadness, anxiousness, guilt and uncertainty. Individual and family reactions to the results could also strain relationships and may affect family planning.

What genes should I be tested for?

According to guidelines developed by a nationwide group of 40 experts, people with ALS should be tested for:

- *C9orf72*
- *SOD1*
- *FUS*
- *TARDBP*
- Any other gene considered to be strongly or definitely associated with ALS by ClinGen (examples: *ALS2*, *CHMP2B*, *KIF5A*, *NEK1*, *UBQLN2*)
- Any gene targeted by an FDA-approved therapy

Source: Evidence-Based Consensus Guidelines for ALS Genetic Testing and Counseling. *Annals of Clinical and Translational Neurology*. <https://doi.org/10.1002/actn3.51895>

If you are interested in genetic testing, talk with your doctor or a genetic counselor.

For more information, visit als.org/GeneticTesting

Genetic Counseling



Everyone with ALS should have the opportunity to meet with a genetic counselor before and after testing.

While strongly encouraged, genetic counseling is not required, and people who don't have access to a genetic counselor can still get tested.

What is a genetic counselor?

Genetic counselors are trained to help people understand their family medical history, the genetics of ALS and the potential impacts of genetic testing.

What should I expect during a pre-testing counseling session?

- The genetic counselor will ask about your family's history of ALS, frontotemporal demetia and other types of motor neuron diseases, movement disorders and demetias.
- Your genetic counselor will assess your risk of having an ALS-linked genetic mutation.
- You will talk about the potential personal, psychological and economic impacts of testing on both you and your family members.
- Your genetic counselor will explain the possible results and current limitations of testing.

What will we discuss after I get my results?

- What these results may mean for both you and your family members.
- Possible next steps, including relevant clinical trials and FDA-approved therapies, if the results show a mutation is present.

What should I ask about?

- Would genetic testing be helpful for me? Why or why not?
- What test is recommended and why?
- What will I learn from having this test?
- How will the results potentially impact me and my family?
- What won't I learn from having this test?
- Will testing and/or counseling be covered by my insurance?

If you are interested in genetic counseling, talk with your doctor.

For more information, visit [als.org/GeneticCounseling](https://www.als.org/GeneticCounseling)