

# 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](https://als.org/GeneticTesting)