Gregory W. Fulton ALS Center

Genetic Testing in ALS

Barrow Neurological Institute

Familial ALS

Approximately 10% of patients with ALS have a familial ALS. This means that they have inherited a gene mutation that we know causes ALS and has been confirmed by genetic testing. If someone has familial ALS, they might not have a family member who has had ALS. Some gene mutations can cause dementia or parkinsonism in other family members. Most genes ultimately cause ALS at some point of the individual's life. Some mutations are associated with very slow or very rapid disease progression, and some may cause dementia. Examples of the most common genetic mutations associated with ALS are SOD1, C9orf72, TARDBP, and FUS.

Benefits of Genetic Testing:

- Can aid in supporting current ALS diagnosis
- Could provide eligibility for current research studies
- Availability to future gene-targeted treatment options
- Family awareness, which could lead to early diagnosis and utilization of disease-modifying medications
- Fairly simple lab test, which is done by blood draw or saliva sample

Risks and Concerns:

- Negative testing results do not rule out the diagnosis
- Family implications, including the psychological stress of communicating positive results
- Future family planning considerations
- Genetic discrimination in medical, disability, and life insurance policies
- Results may take a couple weeks, which can cause stress/anxiety

What does it mean if my test is negative?

Negative DNA testing results greatly reduce the likelihood of you having a genetic form of the disease but do not rule it out completely. This is because we do not know all of the genes that cause ALS or may not have the ability to test for them.

90% of patients with ALS have sporadic ALS which means they do not have a gene that was passed to them and do not have a risk of passing a gene for ALS to family.

What are variant(s) of uncertain significance?

These results show a genetic variant in your DNA testing, but it is unclear if it causes ALS or is simply a benign, unrelated finding. If further research finds an association between the gene variant and ALS, you will be notified of these changes.

What does it mean if my test is positive?

Positive results mean that you have a gene that causes ALS. This means your biological siblings and children are at risk of carrying the gene as well. The most common cause of inheritance is autosomal dominant, which means you have a 50% chance of passing this down to your children. You should discuss this further with your provider and/or genetic counselor. This could provide you with access to gene-based research studies.

- Fulton.Research@CommonSpirit.org
- ClinicalTrials.gov

Genetic Counseling

If you have completed genetic testing through our clinic, genetic counseling services are available to you. The appointment is an hour long, and translation services are available.