

Understanding Genetic Testing

Genetic testing involves examining your DNA, the code that tells your body how to grow, develop, and function. Genetic testing can reveal variants, or mutations, in your DNA that may cause or indicate increased risk for conditions such as FTD.

Who should consider genetic testing?



**Individual
Diagnosed
with FTD**



**Biological
Family
Members**

Who can I discuss genetic testing with?



A genetic counselor can work with you to determine the best testing options for your situation

What are my testing options?

- 1 Single Gene vs. Panel Testing**
Genetic testing can be narrow or broad. If there is no known cause for FTD in the family, broad testing is recommended (panel). If there is a known genetic cause, other family members would only need to be tested for that specific variant (single gene or single variant).
- 2 Diagnostic vs. Predictive Testing**
Diagnostic testing can be performed on the affected person in order to determine if there is a genetic cause for their FTD. Predictive testing can be performed on an unaffected person who is at risk for FTD.



How is genetic testing done?

Genetic testing is performed on a blood, cheek swab, or saliva sample.

What are my results options?

If there is already a known genetic cause for FTD in your family, test results are usually a yes/no answer. If you are the first person being tested, possible results are:

- 1 Positive Result**
A positive result indicates that a genetic variant that causes FTD was found. These results are called pathogenic.
- 2 Negative Result**
A negative result means no genetic differences that cause FTD were found in the genes tested.
- 3 Variant of Unknown Significance (VUS)**
VUSs are variants that we need to learn more about before we can say if they can cause FTD. Often, VUSs are normal differences in genes.



How often is a genetic cause found?

Mutations in known FTD genes account for between 5% and 15% of all FTD.¹ The chances are higher when there is a family history.

When possible, it is best if a person with an FTD diagnosis is tested first.



There is no right or wrong decision regarding genetic testing.



If you or a loved one have questions about genetic testing for FTD, schedule an appointment with a genetic counselor (GC). Talk to your neurologist or locate a GC at <https://FindAGeneticCounselor.nsgc.org/> to discuss all options.

Reference: (1) An update on genetic frontotemporal dementia

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When exploring genetic testing, there are many factors to consider:

PROS

Testing may answer the question of why you or your loved one developed FTD.

Testing may help you and your family members better understand risk.

Genetic testing may be covered by insurance and genetic counselors can provide guidance.

Genetic testing information is protected by a law called GINA, which prevents discrimination by your employer and health insurance, in most cases.

CONS

Testing might not be able to answer why you or your loved one developed FTD.

A negative result does not rule out family risk, unless a genetic cause has already been identified.

There is the possibility of learning of uncertain results instead of definitive answers.

GINA does not protect life insurance, disability insurance, or government/military insurance. To learn more, visit www.GINAhelp.org.

It is important to consider what the results might mean for you and your family:

KEEP IN MIND

Finding a genetic cause for FTD may be a step in the right direction in accessing a clinical trial.

Pursuing genetic testing may reduce uncertainty and bring peace of mind to you and your family.

Sometimes testing can lead to more questions than answers, such as if an uncertain result is found.

Test results can help you plan for the future (reproductive, insurance, financial, career, etc.).

While currently there is not a cure for FTD, results may help doctors better understand your diagnosis and manage your care.

Even though testing is your decision, consider discussing genetic testing with family members since your results may impact them, too.

When exploring predictive genetic testing, it is important to consider how the results may impact your life.

Will they increase or decrease your anxiety?

What changes might you make based on the results?



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