FTD DISORDERS REGISTRY®

Penn Frontotemporal Degeneration Center

Who Should Consider Genetic Testing for FTD, and Why

Finding the cause of a person's frontotemporal degeneration (FTD) diagnosis can be challenging and involve many tests. In some, FTD is caused by a genetic mutation, or variant, that they were born with. Variants are known to increase the risk of developing FTD. Genetic testing can provide this information.

Persons diagnosed with FTD:

FTD-diagnosed persons with a family history of FTD, dementia, amyotrophic lateral sclerosis (ALS), Parkinson's, Alzheimer's, and other neurologic diseases have a higher chance of having a mutation. Those diagnosed with FTD who have no family history of the disease still could have a genetic cause.

Knowing your genetic status can:

- Possibly explain why you have FTD.
- Clarify your diagnosis, and possibly reduce the number of diagnostic tests (and their costs).
- Tell you and your doctor more about your medical needs and the symptoms you might get.
- Let your doctor know what may help you as gene-specific therapies become available.

Knowing your genetic status can help family members:

- If a genetic cause for your FTD is found, family members can choose to have predictive testing to see if they also have the genetic cause of disease.
- If you are tested and a genetic cause is not found, this is also helpful for family members.

Re-testing may be needed over time:

- As new FTD genes are discovered, re-testing may be beneficial. (For example, if you tested prior to 2011 when *C9orf72* repeat expansions were discovered, consider talking with a genetic counselor again about re-testing.)
- If new family members develop neurologic disease or you develop a different neurological condition, re-testing may find a genetic cause.

Knowing your genetic status can help the FTD community:

- You can participate in specific research studies to help scientists learn more about FTD and how to treat it.

DEFINITIONS

Mutations or pathogenic variants: differences in genes that can cause disease

Symptomatic or diagnostic testing: genetic testing of someone with an FTD diagnosis or symptoms

Predictive testing: genetic testing of someone who has no symptoms but due to family history may be at high risk of developing FTD in the future

FTD GENES

Main genes associated with FTD:

- *C9orf72* (C9)
- *MAPT* (tau)
- *GRN* (progranulin)

There are many other less common genetic causes of FTD. A genetic counselor can help you understand your genetic testing options.

Persons who do not have FTD but do have a family history of FTD or other neurologic disorders:

You may not have FTD, but if you have a family history of it or other neurologic disorders, you may be at increased risk of developing FTD. Genetic testing may be able to help clarify your risk for FTD.

Some people choose to learn their genetic status to:

- Inform their children about their risk
- Help with life planning (career goals, reproductive decisions, financial and insurance planning, etc.)
- Establish care with a neurologist who can assist with symptom monitoring
- Relieve some uncertainty
- Participate in research studies

There is no right or wrong decision about learning your genetic status. There are reasons why people choose to test and choose not to test. **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, even if you are certain that you do not want to do genetic testing.

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