FTD DISORDERS REGISTRY®

Penn Frontotemporal Degeneration Center

Understanding the Genetics of FTD

About 40% of people with FTD have FAMILIAL disease, meaning there is more than one affected family member.[1]

The majority of people with a known *genetic cause* have familial FTD.[1]

The other 60% of people with FTD have APPARENT SPORADIC disease, meaning the patient has no affected family members.[1]

The most common genetic causes of FTD are variants in the *C9orf72*, *MAPT*, and *GRN* genes:

C9orf72 Gene (C9)[1]

C9orf72 variants (known as repeat expansions) are the most common cause of genetic FTD, accounting for about 6% of sporadic FTD and up to 30% of familial FTD. C9 expansions can cause FTD, amyotrophic lateral sclerosis (ALS), or a combination of both in an individual or in the same family. C9 expansions can also cause psychiatric and movement problems.

GRN Gene (Progranulin)[1]

GRN variants account for 5% of sporadic FTD and between 5% and 25% of familial FTD. *GRN* is usually associated with behavioral variant FTD (bvFTD), and may also be associated with primary progressive aphasia (PPA), a progressive language and speech disorder.

MAPT Gene (Tau)[1]

MAPT variants account for 2% or less of sporadic FTD and between 5% and 20% of familial FTD. *MAPT* is usually associated with behavioral variant FTD (bvFTD) and may also be associated with progressive supranuclear palsy (PSP) or corticobasal syndrome (CBS).

Other genes more rarely associated include, but are not limited to:

- ***FUS***
- ***CHMP2B***
- ***SQSTM1***
- ***TARDBP***
- ***TBK1***
- ***VCP***

Inheritance Pattern

In the majority of cases, genetic FTD is passed down through **AUTOSOMAL DOMINANT INHERITANCE**

This means that the child of an individual with an FTD-causing gene mutation has a 50% chance of having the same mutation (1 in 2 risk). This also means both men and women can develop and pass on FTD.

Reference: (1) Recent advances in the genetics of frontotemporal dementia

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