IMPORTANT FACTS ABOUT GENETIC AMYOTROPHIC LATERAL SCLEROSIS (ALS)

1. **Genetics plays an important role in ALS.[1]**

   The risk of developing ALS is different across continents and ethnicities, with some of these differences determined by underlying genetic variation.[5] Different mutations in the same ALS-associated gene can result in different rates of disease progression.[1]

2. **Approximately 5–10% of all ALS cases are associated with a genetic mutation.[2]**

   Although most cases of ALS are not associated with known genetic mutations, 5-10% of patients with ALS have a family history of the disease.[2] Even patients without a family history of ALS can have a genetic mutation, and at least 25 genes are known to play a role in the development of ALS.[5] The first ALS-linked gene identified was the *SOD1* gene in 1993.[6] Mutations in another gene, the *C9orf72* gene are most common among European patients, accounting for approximately 34% of cases with a known family history of ALS and approximately 5% of cases without a family history of ALS.[2]

3. **Patients with seemingly "sporadic" ALS (i.e., unknown cause(s) of ALS) may have a genetic form of ALS.[3]**

   Genetic forms of ALS have been identified in those with a family history of ALS as well as in those who do not have a known family history of ALS.[3]

4. **There are many reasons to undergo genetic testing after an ALS diagnosis.[4]**

   People living with ALS often have a lot of questions, including: Why did I develop this disease? What is the likelihood I will pass it to my children? How quickly will my disease progress? Genetic testing may help an individual understand a diagnosis, discover if there are opportunities to participate in clinical research trials targeting genetic forms of ALS, and help inform long-term life choices.[4]

5. **Ongoing clinical trials may be options for people with genetic forms of ALS.**

   Clinical trials focusing on genetic causes of ALS are ongoing (https://www.clinicaltrials.gov).

ALS, amyotrophic lateral sclerosis; SOD1, superoxide dismutase 1; C9orf72, chromosome 9 open reading frame 72.

References:

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