

Knowing is strength.

IMPORTANT FACTS ABOUT GENETIC AMYOTROPHIC LATERAL SCLEROSIS (ALS)

Patient portrayal

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. 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 on to my children? How quickly will my disease progress? Genetic testing may help you understand your 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>).

To learn more about genetic ALS, talk to your doctor or visit insideALS.eu.



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

References:

1. Kenna KP, McLaughlin RL, Byrne S, et al. Delineating the genetic heterogeneity of ALS using targeted high-throughput sequencing. *J Med Genet.* 2013;50(11):776–783. 2. Zou ZY, Zhou ZR, Che CH, et al. Genetic epidemiology of amyotrophic lateral sclerosis: a systematic review and meta-analysis. *J Neurol Neurosurg Psychiatry.* 2017;88(7):540–549. 3. Hardiman O, Al-Chalabi A, Chio A, et al. Amyotrophic lateral sclerosis. *Nat Rev Dis Primers.* 2017;3:17071. 4. Roggenbuck J, Quick A, Kolb SJ. Genetic testing and genetic counseling for amyotrophic lateral sclerosis: an update for clinicians. *Genet Med.* 2017;19(3):267–274. 5. Pansarasa O, Bordoni M, Diamanti L, et al. SOD1 in Amyotrophic lateral sclerosis: “Ambivalent” behavior connected to the disease. *Int J Mol Sci.* 2018;19(5):1345.