GENETIC TESTING MAY PLAY AN IMPORTANT ROLE IN MANAGING AMYOTROPHIC LATERAL SCLEROSIS (ALS)[1]

PEOPLE LIVING WITH ALS—SPORADIC OR FAMILIAL—MAY HAVE A CAUSATIVE GENETIC MUTATION... AND ONLY GENETIC TESTING CAN UNCOVER IT.[1,2]

More than 25 genes have been associated with an increased risk of developing ALS[1,3]

- ALS has historically been divided into 2 categories:[4,5]
  - —**Familial ALS** is defined by a known family history of the disease; in many of these patients, ALS is due to an inherited and identifiable genetic mutation
  - —**Sporadic ALS** is defined by a lack of family history of the disease; in a smaller proportion of these patients, ALS may also be due to a genetic mutation

- Although only a small proportion of patients with sporadic ALS have a known genetic mutation, sporadic ALS represents the majority of ALS diagnoses—therefore many genetic ALS cases can be found among seemingly sporadic ALS cases[1,2]
- Genetic ALS describes any ALS that has been caused by a genetic mutation, regardless of family history of disease[1,3]
- The first gene mutation linked to ALS was reported in 1993, in a gene called superoxide dismutase 1 (*SOD1*). Mutations in *SOD1* are thought to cause ALS via toxic gain of function[5,6]
  - —*SOD1* ALS is the best understood genetic form of ALS; more than 180 different mutations in *SOD1* have been identified[7,8]
- *C9orf72* is the most frequent ALS-associated gene; in addition to *SOD1*, other common mutations include *FUS* and *TARDBP*[2,5]

THE IMPORTANCE OF GENETIC TESTING IN ALS

Individuals with a clinical diagnosis of ALS and their family members may benefit from genetic testing[1]

Genetic testing can help people living with ALS understand why they developed the disease, assess their odds of passing it on to their children, and help determine how quickly their disease will progress if they harbor a known mutation[1,9]

Family members may also benefit from knowing whether they are at increased risk of developing ALS[1]

Genetic testing can also help patients discover opportunities to participate in relevant clinical research trials. These trials are further evaluating the role of genetic mutations in ALS and may assist in the discovery of genetically targeted treatments in the future[1,3,10]

Pre- and post-test genetic counseling may be considered for all patients with ALS and asymptomatic individuals. Important topics may include the limitations of genetic testing, interpretation of results, and implications for family members and/or family planning.[1,10]

GENETIC TESTING TODAY MAY HELP PROVIDE A BETTER TOMORROW FOR PEOPLE IMPACTED BY ALS[1,10]

CONSIDER THE BENEFITS OF KNOWING MORE ABOUT ALS

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