SB212
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My name is Nicholas J. Maragakis, M.D., I am a Professor of Neurology at Johns Hopkins University and serve as the Medical Director of the Johns Hopkins ALS Clinical Trials Unit and the Center for ALS Specialty Care. I also have a laboratory that studies ALS and have been caring, almost exclusively, for patients with ALS for over 20 years.

Amyotrophic Lateral Sclerosis is a neurodegenerative disease with no known cure. It slowly and relentlessly robs individuals of their strength, speech, swallowing, and eventually their ability to breathe—almost uniformly resulting in death within 2-5 years.

ALS was traditionally thought of as a "sporadic" disease—with no known genetic links. Over the last 25 years, however, over 30 genes associated with ALS have been identified. Therefore, we now estimate that 10% of patients with ALS carry these disease-causing genes in their families. One can imagine the tremendous physical and emotional burden carried by individuals with ALS as well as their family members. This is particularly notable because ALS patients have a 50% chance of passing on these ALS disease genes to their children. It is cruel enough that they carry these burdens but for them not to be able to obtain insurance seems particularly cruel.

As a neurologist, we have important reasons to perform genetic testing on patients. First, it tells us something about their particular disease, in this case ALS. ALS patients with some genetic forms of ALS may have a faster or slower course of the disease. Knowing these genetic subtypes can help us advise them on prognosis and medical management. Secondly, genetic testing can tell us about risks to other family members. Understanding those risks can help individuals with family planning. Finally, and this is my hope for the future, we as a community have now developed gene therapies specifically targeting certain ALS causing mutations—providing hope for those individuals affected by ALS. These gene therapies are now being used in patients harboring those specific genes—a powerful reason to know one's genetic background. Excitingly, we are now looking to treating individuals with known ALS mutations before they develop any symptoms—creating a future for them without the physical and emotional burdens of this terrible disease.