January 30, 2024

The Honorable Pamela Beidle
Chair, Senate Finance
Room 3 East Wing, Miller Senate Office Building,
11 Bladen Street, Annapolis, MD 21401-1991

The Honorable Chair Beidle, Vice Chair Klausmeier, and Members of the Finance Committee

RE: SB 142 – The Genetic Testing Protection Act of 2024

Position: SUPPORT

My name is Kuldip Dave, Ph.D., and I am Senior Vice President for Research at the ALS Association. I am writing today in support of SB 142: The Genetic Testing Prohibition on Disability, Life, and Long-Term Care Insurance (Genetic Testing Protection Act of 2024), but most importantly, in support of the more than 220 families with ALS we currently serve in the state of Maryland as well as the tens of thousands more affected by the disease across the country.

One of the greatest scientific achievements of our time was mapping the human genome, a project that was led in Maryland. Now, the state and this Committee have the opportunity to lead the way in advancing medicine yet again through The Genetic Testing Protection Act, which if passed, will provide the safeguards needed to realize the full benefits of genetic science.

Genetic Basis of ALS and New Opportunities for Treatment

In 2019, researchers from Harvard University and the University of Queensland in Australia estimated that 40% of diseases have a genetic component.[1] This includes ALS. ALS is a devastating neurodegenerative disease that progressively robs people of their ability to move, speak, eat, and eventually breathe. There is no way to stop or reverse this deterioration once it starts, meaning that most people only live for 2–5 years after being diagnosed.

Although there is still no cure, we are living in an era of unprecedented change in ALS care catalyzed by the knowledge we've gained about the genetic underpinnings of the disease. We estimate that roughly 10–15% of ALS is driven by gene mutations that are either passed down in families or occur randomly during development. So far, researchers have identified more than 40 genes linked to ALS.

One of these genes is *SOD1*, which is the second-most common cause of familial ALS. The most prevalent *SOD1* gene mutations in North America are associated with younger age of onset and shorter survival.

[1] Lakhani, C.M., Tierney, B.T., Manrai, A.K. *et al.* Repurposing large health insurance claims data to estimate genetic and environmental contributions in 560 phenotypes. *Nat Genet* **51**, 327–334 (2019). https://doi.org/10.1038/s41588-018-0313-7

Last year, families devastated by *SOD1*-ALS were given new hope when the Food and Drug Administration approved the first genetically targeted treatment for ALS. Tofersen, now known as Qalsody, was developed to specifically target the RNA produced from mutated *SOD1* genes to stop the production of toxic *SOD1* proteins that cause ALS. Because of this, only people with a *SOD1* mutation can benefit from taking this drug, thereby underscoring the importance of genetic testing for people living with ALS.

Increasing Importance of Genetic Testing for ALS

With the approval of tofersen, at least 10 other therapies targeting ALS-linked genes being tested in clinical trials, and almost a dozen more being developed preclinically, genetic testing is being recognized as a vital part of ALS clinical management. According to new evidence-based consensus guidelines published last year in the *Annals of Clinical and Translational Neurology*, everyone living with ALS should be offered genetic counseling and testing.[2]

Genetic testing results not only have implications for people living with ALS but also their family members. Having a first-degree relative test positive for an ALS-linked mutation significantly increases a family member's risk of developing the disease. It also potentially paves the way for prevention.

In a study being conducted at Johns Hopkins, Dr. Nicholas Maragakis and colleagues are trying to see if tofersen can delay the onset of ALS — or perhaps even prevent the disease from developing all together — in people with a *SOD1* mutation who have no ALS symptoms. Not everyone with a *SOD1* mutation will develop ALS in their lifetime, but what if we could stop the disease before it starts in those individuals who would eventually be affected — like using cholesterol levels in blood and treatment with cholesterol-reducing drugs to prevent heart disease? Imagine the economic, societal, and personal costs that would be saved.

Threat of Genetic Discrimination Holds Back Progress — And Harms Marylanders

Despite the tremendous benefits this research could provide to individuals and the entire ALS community, it is difficult and time-consuming to recruit participants because few people with ALS and their family members know their genetic status. Fear of their genetic information being used against them is one reason why people say they don't get tested.

Thus, the threat of genetic discrimination creates a serious dilemma for Marylanders — risk their physical health because they don't know their genetic status or risk their financial health because they do.

[2] Roggenbuck, J., Eubank, B.H.F., Wright, J. *et al*. Evidence-based consensus guidelines for ALS genetic testing and counseling. *Ann Clin Transl Neurol* **11**, 2074-2091 (2023). https://doi.org/10.1002/acn3.51895

The Genetic Testing Protection Act will help allay this fear by putting protections in place for accessing life and disability insurance by people who have undergone genetic testing, requested genetic testing, or received genetic test results. Such protections will not only benefit those living in this state, but through the amazing science being done in Maryland, bring life-changing new genetic therapies to everyone who needs them faster.

For all these reasons, I respectfully request your support for The Genetic Testing Protection Act. Thank you to Senator Klausmeier for introducing this legislation and to the members of the Senate Finance Committee for your time and consideration.

Sincerely,

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