Meet Hannah*

Hannah is a 25 year old living with intractable epilepsy.

Hannah's diagnostic odyssey lasted into adulthood

7 months old

As an infant, Hannah experienced two seizures. She was referred to a pediatric neurologist but was not immediately prescribed anti-seizure medications (ASMs). She was eventually diagnosed with epilepsy with focal onset seizures and started her first ASM.

8 months - 6 years

It took time to find a combination of ASM's that kept Hannah's seizures under control and medication adjustments were continually made. She began to experience tonic-clonic seizures, which required further ASM adjustments from her neurologist. She was also diagnosed with developmental delay and eventually mild intellectual disability.

13 years old

Hannah's seizures worsened. While she previously could go months without a seizure, she was now experiencing frequent breakthrough seizures. Multiple ASM adjustments occurred, and she had a vagus nerve stimulation device implanted.

25 years old

As an adult, Hannah continued to experience monthly tonic-clonic seizures and adjustments to her numerous ASMs. Her neurologist referred her to a genetics clinic for exome sequencing.

Hannah's exome test results identified a de novo pathogenic loss-of-function variant in *SCN1A*. An earlier diagnosis could have resulted in earlier seizure control since many of Hannah's previous medications were contraindicated with her specific *SCN1A* variant.

Hannah's diagnosis brought clarity to:

- her symptoms
- future family planning and reproductive counseling for her and her siblings
- medical management for her neurologist

*Case study is based on GeneDx patient testing, with all identifying information removed.

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