Meet Ethan*

At four months of age, Ethan's mother became concerned that he wasn't meeting developmental milestones at the same rate as his siblings had. During a routine appointment, his pediatrician confirmed developmental delays.

Ethan's diagnostic odyssey lasted into adulthood

14 months old

Ethan's pediatrician diagnosed him with global developmental delay because he was not speaking and was not walking. He experienced significant weakness and abnormal tone in his legs. After receiving further neurological assessments, he was diagnosed with spastic diplegic cerebral palsy.

15 months old

Ethan experienced his first grand mal seizure and was admitted to the hospital, where he experienced multiple seizures. Eventually, his seizures were controlled with medication, and he was discharged and referred to the neurology clinic.

2 years - 18 years

Ethan's seizures were managed with a variety of anti-seizure medications over the years, but he continued to experience break through seizures monthly. He never developed the ability to walk, was non-verbal, and had been diagnosed with intellectual disability.

19 years old

Ethan began his transition to adult care. His neurologist noted that he had not received genetic testing and, given Ethan's clinical presentation, referred him to the local genetics clinic for exome sequencing.

Ethan's exome test identified a de novo pathogenic variant in *SLC2A1*, providing him with a diagnosis of *SLC2A1*-related GLUT1 deficiency. Had Ethan received an exome test earlier he could have received disease-specific treatment which could have provided better seizure control and answers for his family years earlier.

Ethan's diagnosis provided answers

- for his symptoms
- for his older siblings and their future family planning
- for his neurologist to offer targeted medical care

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

Photos do not represent actual patients.

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