CASE STUDY

Implications to clinical practice

Meet William†

Born prematurely, William was admitted to the NICU. His care team noted his small head circumference, but a cranial ultrasound came back normal. Following an uncomplicated stay, he was discharged at 4 weeks old.

William's journey to a diagnosis took 7 years.

8 months old

Although William met most of his early motor milestones, he still needed support to sit upright. His parents consulted his pediatrician, who advised a wait-and-see approach. By 14 months, William could sit without support.

21 months old

William still wasn't walking, and his muscles seemed tight and stiff. He also had speech delays and was exhibiting behaviors consistent with autism. His pediatrician referred him to both a pediatric neurologist and a developmental-behavioral pediatrician for follow up.

2 years old

The neurologist diagnosed William with spastic diplegic cerebral palsy, and he began being followed in the cerebral palsy clinic. Six months later, the developmental pediatrician diagnosed him with autism spectrum disorder.

3 years old

While following William's progress, the orthopedic surgeon questioned the definitive cause of his cerebral palsy and ordered an MRI, which returned as non-diagnostic. William continued to be monitored in the cerebral palsy clinic.

6 years old

William's parents wondered if there was an underlying cause or explanation for his cerebral palsy and autism. His neurologist made a referral to see a geneticist. While waiting for an appointment, William experienced his first seizure.

7 years old

He was seen by genetics just after his 7th birthday, and a chromosomal microarray was ordered. While awaiting results, he experienced another seizure. The microarray was negative, so an exome was ordered as a trio.

William's exome test results identified a *KIF1A*-related disorder due to a de novo pathogenic variant, explaining all of his initial diagnoses:

- developmental delay
- cerebral palsy
- autism spectrum disorder
- seizures

His journey could have been 7 years shorter.

For up to one third of patients with cerebral palsy, the underlying cause is a genetic condition. Had William's exome test been ordered much earlier, it could have enabled a specific diagnosis, informed family planning, and connected William and his family with gene-related support groups—all in just 4 weeks.

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

GeneDx