CASE STUDY

Implications to clinical practice

Meet Oliver*

Shortly after his birth, routine newborn assessment identified that Oliver exhibited mild hypotonia. Without additional findings, Oliver was discharged. But by 15 months of age, Oliver's parents started noticing he wasn't meeting the normal milestones and wasn't speaking or walking. They brought their concerns to Oliver's pediatrician.

Oliver's journey to a genetic diagnosis took almost 7 years.

15 months old

Oliver's pediatrician referred him for a hearing test and a consultation with a speech and language pathologist based on his speech delay. His hearing test was normal, but he was identified to have expressive language delay.

2–3 years old

Oliver's pediatrician referred him for a hearing test and a consultation with a speech and language pathologist based on his speech delay. His hearing test was normal, but he was identified to have expressive language delay.

6 years old

Oliver's pediatrician referred him for a hearing test and a consultation with a speech and language pathologist based on his speech delay. His hearing test was normal, but he was identified to have expressive language delay.

7 years old

After 9 months on the waitlist, the geneticist evaluated Oliver and ordered exome sequencing as a trio including the parental samples. Exome sequencing provided results in 3 weeks, and identified a de novo pathogenic variant in *MED13,* finally explaining the findings of:

- autism spectrum disorder
- intellectual disability and developmental delay
- hypotonia
- expressive language delay
- Duane's syndrome
- scoliosis

His journey could have been 4 years shorter.

Following society guidelines, Oliver's doctor could have ordered exome after his initial presentation of developmental delay. The exome would have delivered results in 3 weeks, enabling a specific diagnosis, informing family planning, and connecting Oliver and his family with gene-related support groups and resources when he was 3 years old.

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

GeneDx