Case Study

MyOme Rare Disease
Real Answers for Real Families

1

Meet 8-year old Sam*

Diagnosed with congenital hypotonia, autism spectrum disorder, global developmental delay, and gait abnormalities

2

Diagnostic Odyssey

Whole-exome sequencing by another lab did not detect any pathogenic variants associated with the clinical phenotype

3

MyOme test ordered

Rare Disease Exome Analysis identified a previously undetected single-exon deletion in the SOX5 gene, which can cause Lamb-Shaffer syndrome

4

Answers that matter

A genetic diagnosis enabled more personalized care and opened the door to specialized support networks and research opportunities

Our Genome First Approach Detects More

MyOme's Rare Disease Exome Analysis is run on a comprehensive, **whole-genome** backbone that enables deeper coverage and detection of complex variants.

Starting With More Means Finding More

Backed by whole-genome sequencing, we offer improved detection and faster results, so every family can get the answers and care they need.

Learn more at www.MyOme.com/our-tests/diagnostic

*Name is fictionalized

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