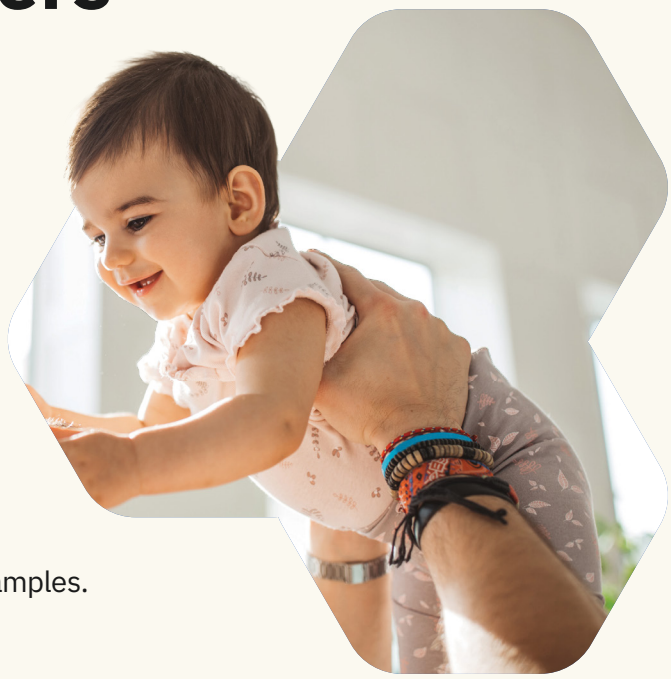
Why *trio* testing matters

Trio testing can provide critical inheritance information that changes how variants are classified, enabling faster diagnoses, access to disease-specific management, targeted therapies, research opportunities, and support resources.



Trio testing can increase diagnostic yield by 7-15%[1-4] compared to testing that does not include biological relative samples.

Investigating the cause of infantile seizures

1 month

At 1 month, Lola's seizures began. EEG confirmed epileptic encephalopathy, while brain imaging and metabolic testing were normal. An epilepsy gene panel identified 2 variants of uncertain significance (VUS). Lola was put on antiseizure medication.

2.5 months

At her Genetic's appointment, Lola was not identified to have any dysmorphic features. It was noted that she had not fully developed head control yet. Upon review of the panel results, a chromosomal microarray (CMA) was offered and, if negative, exome testing as a trio was discussed as a possible next step.

4 months

Lola and her parents attended a follow-up appointment in the genetics clinic. Her CMA was negative and exome testing was again discussed. Lola's parents opted to proceed with exome testing for Lola and agreed to each provide a sample so the testing could be performed as a trio. Kits were sent to the family home for sample collection.

GeneDx®

The impact of trio testing on *diagnosis*

Report without trio test

- Proband exome identified the same two VUS, including one in *CDKL5*, without inheritance context
- No molecular diagnosis

Medical management *without* trio

- Limited disease-specific prognosis
- Limited treatment recommendations
- Uncertainty for management

Report with trio test

- Trio exome confirmed the variant in *CDKL5* was de novo, providing the evidence needed to classify it as pathogenic
- The second VUS was observed in an unaffected parent and considered unlikely to be clinically relevant, so it was not reported
- Molecular diagnosis established: CDKL5 Deficiency Disorder (CDD)

Medical management *after* trio

- Management under guidelines for CDD
- Referral to multidisciplinary specialists
- Disease-specific therapies, such as ganaxolone
- Clinical trials
- Research studies
- Patient support organizations
- More informed prognosis
- Family recurrence-risk counseling

The impact of trio testing

Lola's diagnosis was only possible because trio exome testing provided inheritance information needed to classify a disease-causing variant.

This directly impacted clinical management, treatment options, and family support resources.

Start with trio testing to maximize diagnostic yield and help families get answers sooner.

Learn more about convenient options to order trio testing.
genedx.co/how-to-order