CASE STUDY

Implementing society guidelines into clinical practice

female

6 months old

Meet Claire†

A 6-month-old female presenting with developmental delay and seizures. Claire initially began experiencing frequent seizures at the age of 5 months and her parents also noticed a delay in her development over time. She was referred to a pediatric neurologist.

Claire's journey to a genetic diagnosis took almost 2 years.

6 months old

Initial testing with an EEG showed multifocal epilepsy, while the brain MRI showed no abnormalities. Claire's parents requested genetic testing and her pediatric neurologist ordered a narrow, sponsored NGS seizure panel with ~300 genes. There were **no significant findings.**

19 months old

After being followed for **more than a year**, Claire's family was desperate to have a reason for her symptoms and were planning to have more children. They requested a referral to a geneticist.

23 months old

After waiting **4 months** for an appointment with a geneticist, Claire was offered whole exome sequencing.

2 years old

The exome test delivered results in **6 weeks** that ended the diagnostic odyssey for Claire and her family. Results showed pathogenic variants in *NAPB*, causing autosomal recessive early-onset epileptic encephalopathy.

Her journey could have been 2 months.

Following society guidelines, Claire's doctor ordered exome right away. The exome delivered results in **6 weeks**, enabling a specific diagnosis and effective treatment.

6 months old — 8 months old

Take a guideline-driven approach. Start with exome.

† Fictionalized case study for illustrative purposes only

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