Meet Nora*

Nora was a healthy, full-term baby who showed no signs of growth or developmental issues as a newborn. Around 4 months of age, Nora's parents took her to the pediatrician for crying and ear pulling, and she was diagnosed with her first ear infection. Her pediatrician also noticed that she had not gained much weight since her last visit.

5 months old

Nora continued to eat well, despite not gaining much weight. However, her length had also stagnated and her pediatrician suspected malabsorption. They referred Nora to a gastroenterologist for a more thorough assessment.

7 months old

The gastroenterologist did not identify any initial explanation for Nora's failure to thrive and opted to monitor her through follow-up appointments. Around that time, her parents noticed patches of eczema and asked if a food allergy could be the cause of her poor growth, prompting her pediatrician to refer Nora to an immunologist and allergist. In addition, Nora's ear infections were becoming a recurrent issue so the pediatrician also referred her to an audiologist to check for potential hearing loss.

8 months old

Nora was confirmed to not have any food allergies, but was tested for immunoglobulins, vaccine response, and lymphocyte status. All results were deemed unremarkable, and she was subsequently referred to a geneticist and endocrinologist for further assessment of her poor growth and weight gain. Separately she was diagnosed with mild to moderate conductive hearing loss–most likely due to fluid in her ears from her chronic ear infections–and was referred to an otolaryngologist.

11 months old

Nora's geneticist ordered a chromosomal microarray (CMA) and testing for Russell-Silver syndrome (RSS) on account of her poor growth, low weight, triangular-shaped face and relative macrocephaly.

14 months old

Nora had tubes placed in her ears, which resolved her conductive hearing loss. Her initial bloodwork from the endocrinologist was unremarkable and she was scheduled for a endoscopy to further investigate her poor growth. Her eczema continued to worsen, though, and she returned to the genetics clinic to receive the results of her negative CMA and RSS testing. The geneticist suggested exome sequencing to ensure that she received the most comprehensive assessment.

Nora's exome test results identified a de novo gain-of-function (GOF) variant in *STAT3*. This result provided her family with a diagnosis of STAT3 GOF syndrome, which explained her clinical findings and allowed her health care team to tailor their approach for her ongoing medical care.

Her journey could have been months shorter.

Had Nora received an exome test earlier, she would likely have had fewer tests and reached a diagnosis sooner. Leading with exome testing as the initial genetic test would have shortened her diagnostic journey and started her on a targeted treatment plan sooner.

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

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