Meet Mateo*

Mateo was a healthy baby boy with no major medical concerns or conditions. He was considered small for gestational age at birth and intrauterine growth restriction was noted on ultrasounds during his mom's pregnancy.

Mateo's journey to a genetic diagnosis took almost 3 years.

1 year old

At Mateo's one year check-up his pediatrician noted he appeared short in stature and had a larger head circumference. He recommended that Mateo be seen by a geneticist. The genetics clinic was able to see Mateo a month later and during the exam, noted he had a broad forehead, short and broad fingers, a history of GERD and congenital laryngomalacia. In addition, the macrocephaly appeared to be familial as both his sister, father and uncle have similar characteristics. Genetics ordered a CMA, which was negative, and referred Mateo to endocrinology for further testing related to his short stature.

2 years old

At two years old, Mateo was evaluated by an endocrinologist. While his labs returned normal results, the skeletal survey suggested disproportionate short stature. Based on their clinical exam, the endocrinology clinic ordered a skeletal dysplasia panel.

The skeletal dysplasia panel identified two variants in *NPR2*: one pathogenic and one VUS. The genetic testing laboratory was unable to determine if the variants were in *cis* or in *trans*. The endocrinologist referred the family back to genetics and parental testing was ordered. This additional testing was able to determine the variants were in trans but ultimately led to the same result: one PATH and one VUS variant in *NPR2*. From start to finish, this panel testing and its follow-up took four months.

3 years old

Mateo was seen at genetics one month later for follow-up and an exome test was ordered through GeneDx. Based on Mateo's clinical information and variant data within the GeneDx Infinity™ database, the VUS was classified as LPATH on the GeneDx report. In combination with a PATH variant in *trans*, GeneDx was able to provide a conclusive molecular diagnosis for Mateo.

Mateo's journey could have been two years *shorter*

At three years old Mateo received a genetic diagnosis of *NPR2*-related acromesomelic dysplasia, explaining his disproportionate short stature, short and broad fingers, and prominent forehead. Patients with this genetic variant tend to stop growing in childhood and their final adult height is less than four feet.

GeneDx®

*Case study is based on GeneDx patient testing, with all identifying information removed. Photo does not represent actual patient