Meet Zoe*

During pregnancy, Zoe's mom was followed closely for intrauterine growth restriction and left unilateral renal agenesis that were identified on ultrasound. At birth, Zoe was small for gestational age and her kidney anomaly was confirmed. The only additional medical concern noted was that Zoe was born with an additional digit. The digit was removed, and Zoe was discharged home.

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

2 months old

At two months of age, Zoe's parents became concerned about her growth and development. Zoe was hospitalized multiple times for failure to thrive and was seen by multiple pediatric gastroenterologists and speech language therapists. An upper endoscopy and tests for celiac and thyroid were ordered; all test results came back normal. During this time, Zoe was not losing or gaining any weight, leading clinicians to determine that the failure to thrive was largely behavioral. The clinicians recommended adding high calorie foods and supplements to her diet.

6 months old

At six months, Zoe saw a pediatric endocrinologist due to concerns of short stature. Multiple laboratory tests were ordered, and all test results were normal. Zoe's short stature was thought to be secondary to failure to thrive as her overall growth was proportionate, but each parameter (weight, length, head circumference) was below the first percentile for her age.

6-18 months old

Over the next few months, Zoe experienced delayed speech and motor milestones and was seen by various speech and occupational therapists. At a year and half, Zoe's development caught up and these appointments were discontinued. During this time, she was also seen at a genetics clinic and had a CMA, which was normal.

2 years old

At two years old Zoe was hospitalized again due to failure to thrive. During her hospitalization, genetics was consulted and a follow-up in the outpatient clinic was recommended. Four months later, Zoe was seen by genetics and an exome was ordered.

Zoe could have received an answer 2 years *sooner*

The results identified a *GLI2* pathogenic variant, which is associated with Culler-Jones syndrome, explaining the extra digit at birth, delayed developmental milestones and her short stature. This condition is associated with endocrine abnormalities, leading Zoe to be referred to an endocrinologist to optimize her medical care moving forward.

GeneDx®

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