Meet Sarah*

Near the end of her pregnancy, Sarah's mother was measuring small and the doctor suspected intrauterine growth restriction (IUGR). An ultrasound confirmed IUGR and Sarah's mother was followed closely until delivery.

Birth

Sarah was born full term, her delivery was uneventful, and she was discharged home.

Notably, Sarah was in the 1st percentile for weight and 10th percentile for length.

4 Months Old

During a routine four month visit to the pediatrician, Sarah's doctor noticed she was slow to gain weight and her mother confirmed Sarah was having difficulty eating.

On exam, the pediatrician found a heart murmur and referred Sarah to cardiology for further testing. An echocardiogram identified pulmonary artery stenosis and hypoplasia. Her cardiology team did not recommend surgery since there did not appear to be implications for Sarah's growth and development. However, since these cardiac findings can be associated with some genetic disorders, Sarah was referred to genetics for further evaluation.

16 Months Old

After a 12 month wait, Sarah was seen by a geneticist at 16 months old.

On exam, she was described to have a large forehead, posteriorly rotated ears, and a pointed chin. Combined with her cardiac findings, the geneticist ordered an exome sequencing trio concurrently with a chromosomal microarray.

A few weeks later, the exome sequencing test report was returned and identified a *JAG1 de novo* pathogenic variant, conferring a molecular diagnosis of Alagille syndrome. This diagnosis allowed the care teams to recommend additional evaluations and specialist appointments as well as routine surveillance.

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

Photos do not represent actual patients.

GeneDx