Meet Violet*

During pregnancy, anatomy ultrasound revealed brain and heart abnormalities, including incomplete development of the corpus callosum and bilateral superior vena cava (SVC). A fetal echocardiogram confirmed dextrocardia and double inlet left ventricle (DILV) with double outlet right ventricle (DORV), prompting prenatal genetic evaluation.

Prenatal

Violet's parents received a genetics consult because these findings are often associated with genetic disorders. An amniocentesis was performed and a chromosomal microarray (CMA) was ordered, and came back negative.

Birth - 6 Weeks Old

The delivery care plan included immediate NICU admission for Violet.

After birth, Violet experienced respiratory distress requiring positive pressure ventilation and continuous positive airway pressure (CPAP). A postnatal echocardiogram reconfirmed DILV with DORV and additional findings (malposed great arteries, mildly hypoplastic aortic arch, bilateral superior vena cava, and atrial septal defect).

She was then transferred to the CICU for cardiac management. During her CICU admission, Violet underwent cardiac catheterization and MRI, which confirmed partial agenesis of the corpus callosum. Genetics was then consulted, and exam findings included:

- Microcephaly
- Bitemporal narrowing
- Low anterior hairline
- Simple ears
- Mild facial dysmorphisms
- Single transverse palmar crease

The genetics team requested a post-natal analysis of the prenatal CMA, and ordered a multi-gene congenital heart disease panel. Both tests came back negative.

7 Weeks Old

Violet was discharged from the CICU at seven weeks old and referred for multiple specialist appointments in the outpatient setting, including a follow-up with genetics.

6 Months Old

At Genetics follow-up, Violet's parents reported developmental concerns. She had missed early motor milestones such as maintaining head control and rolling. Given her clinical features, congenital heart disease, and prior negative testing, genome sequencing was ordered.

Results identified a pathogenic variant in *ARID1B*, consistent with Coffin–Siris syndrome, which is associated with:

- **Cardiac malformations**
- **Hypotonia**
- **Microcephaly**
- **Facial dysmorphism**
- **Abnormal hair growth**
- **Intellectual disability**

Despite prenatal and postnatal genetic evaluations, Violet did not receive a molecular diagnosis until six months of age. Had a more comprehensive test been ordered initially, Violet's parents and care teams could have started symptom-based care and early intervention sooner.

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

Photos do not represent actual patients.

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