CASE STUDY

Meet Anne†

a newborn presenting with colonic atresia

Ordering rapid genome sequencing as a first-tier genetic test can:

Prevent

- Multiple genetic tests
- Extended stays in the NICU
- Unnecessary suffering

Provide

- A timely and accurate diagnosis
- Improved treatment management
- Access to early interventions

Anne received surgery to repair the colonic atresia on day two of life and was transferred to the NICU following the procedure for post-operative monitoring and management. **While in the NICU, Anne experienced feeding intolerance, repeated infections, and was not recovering as expected.** Her doctors ordered a complete blood count (CBC) and flow cytometry as part of her workup.

Results of that testing showed hypogammaglobulinemia and decreased T-cell function but **didn't fully explain her symptoms.**

With no obvious answers, **her medical team ordered rapid genome sequencing, which revealed compound heterozygous variants in the *TTC7A* gene** associated with autosomal recessive *TTC7A* deficiency, also known as gastrointestinal defects and immunodeficiency syndrome 1 (*GIDID1*).

As a result of this testing, Anne's team made important and informed medical management changes, including:

- IgG replacement
- Pentamidine for PJP prophylaxis
- Palivizumab to prevent respiratory syncytial virus (RSV)
- **Feeding intolerance was determined to be due to her genetic condition vs. post-operative complications**

The rapid genome test delivered actionable results within 5 days.

The results helped determine the correct treatment plan for Anne and the path forward for her family and care team.

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

GeneDx