

Baby Badger Network ECHO

Problem of Practice Summary & Recommendations

Session Date: June 11, 2026

Question to consider: In a newborn with dysmorphic features and no major organ dysfunction what factors guide the decision to pursue genetic testing during the birth hospitalization?

Brief Summary: The 5-day old infant initially presented with transient tachypnea of newborn (which resolved) and was working on feeding. RN noted a preauricular ear pit which prompted a detailed dysmorphology physical exam. Multiple dysmorphic features were noted. Echo revealed a small VSD.

The team was unsure whether a diagnosis was needed while infant was inpatient or if testing could wait until an outpatient visit. Following a consult with genetics, rapid genome testing was thought to be reasonable. While testing was in progress, infant developed episodes concerning for seizures. Infant was transferred and a genetic diagnosis returned while infant was enroute

Discussion Focus :

- If an infant had one major anomaly and multiple minor anomalies, pursuing genomic testing would be more straightforward. Infant had distinct dysmorphic facial features, but they did not strongly suggest a genetic condition.
- A genetics consult was very helpful.
- Both negative and positive results can be helpful and impact management.
 - A negative result can be reassuring; helping to rule out many genetic conditions. It can also impact management by knowing a “wait and watch” approach is appropriate.
 - A positive results was very helpful in this case. Knowing feeding difficulty is a part of the syndrome, a G tube was placed sooner, likely shortening the hospital stay.
- The sample was drawn and shipped on a weekend. Having good communication with the lab in advance was very helpful, especially when expedited processing was needed
- Communicating the result with the transfer hospital was very helpful in coordinating care for baby's arrival. The transfer hospital got a head start alerting the right teams. The family was given results during transfer and informed they would meet with Genetics at the transfer hospital.
- Sometimes, the logistics of sending genomic testing seems insurmountable and there may be a tendency to defer testing to the outpatient setting. Sending genomic testing in the NICU saves time vs deferring to the outpatient setting.
- Kids go home sooner when they have genomic testing.
- The value of genomic testing is diminished if we only test kids we are sure will get a diagnosis. The full phenotype may not manifest until later and often occurs as a spectrum