

Baby Badger Network ECHO

Problem of Practice Summary & Recommendations

Session Date: Sept. 10, 2026

Question to consider: *To what extent should trio testing be prioritized, and how can we operationalize that decision in real-world clinical workflows?*

Brief Summary:

Genetics was consulted on a 3 week old male with respiratory distress, feeding difficulties, congenital anomalies and dysmorphic features. Microarray was negative. The social situation was complicated resulting in extreme difficulty in communicating with parents

At 8 weeks of life the patient developed malrotation without volvulus and rGS was recommended however multiple attempts to discuss the recommendation, obtain consent and obtain parental samples were unsuccessful. Although mom had consented, sample could not be provided in a timely manner resulting in rGS-singleton.

Among the results were two variants (pathogenic and VUS) in a gene associated with an Autosomal Dominant condition (hearing loss +/- developmental delay) and a more serious Autosomal Recessive condition (severe neurodevelopmental delay, intellectual delay, nonverbal and intestinal rotation).

The AR condition was suspected, however because parental samples weren't analyzed, the phase of the variants (cis or trans) could not be determined. Parents were struggling with the news (denial, fear, guilt etc). G-tube placement was recommended, however parents were reluctant.

Eventually mom provided sample. Results are pending.

Discussion Focus :

- We try our best to make an opportunity to share information about the test with parents and to share how parental samples for trio testing can be very helpful in finding an answer and decreasing time to find an answer. Unfortunately this doesn't always happen.
- Obtaining parental samples in a timely manner is not always possible
- Barriers include:
 - One or both parents are not available (mom is admitted at birthing hospital, one parent is at home with other children, transportation barriers,
 - Parents decline testing
 - Non paternity.or uncertain paternity
 - Parents are overwhelmed with information and overburdened with decisions
 - Some parents may anticipate feeling guilty if the variant was inherited from them
 - Some parents don't want their DNA "out there"
- Suggestions for alleviating Barriers
 - Leaving kits in the room with simple instructions
 - Educating overnight staff so they can answer questions and/or prompt parents
 - Offering genomic testing at a L3 NICU may lower barriers associated with parents not being available due to caring for their other children or having to work.
 - Targeted parental testing to clarify phase is sometimes offered free of charge
 - By the test lab