

Baby Badger Network ECHO

Problem of Practice Summary & Recommendations

Session Date: October 8 2026

Brief Summary

Infant was transferred to Level III from OSH on DOL1. Discussion of exome occurred on DOL 9. Samples collected on DOL 10 The infant quickly improved and was discharged on DOL 13. Results returned on DOL 16 (Problem of Practice 1).

A heterozygous Likely Pathogenic variant was identified in a gene associated with two Autosomal Recessive conditions. There is preliminary (insufficient) evidence of causality with an Autosomal Dominant condition. The lab report states, "This result is consistent with this proband being at least a carrier for an autosomal recessive disorder but does not establish a molecular diagnosis as a second variant was not identified in this gene." (Problem of Practice 2)

Problem of Practice 1

What to do when results come back after the baby has been discharged? The inpatient team does not "see" discharged patients on their dashboard; therefore, they do not know when results become available. Additionally, the ordering physician may not be on-service or on-call when results are reported. The discharge summary stated, "results pending".

At the time of this case, there was no established process for receiving and disclosing results after discharge. There was no clear hand-off as to who and how a provider would be notified of results. It was not clear what the follow up plan would be if the results were positive or negative.

Questions from Presenter

1. Do you have a tracking system for rGS results? •Who owns the process? How do results that return after discharge get conveyed to parents and who discusses those results with them?

Recommendations from Group:

- When entering order in the EMR, cc everyone on the team to receive notification when results return AND/OR when completing TRF (or online portal order) request results are shared with the NICU team (one institution has a group email with all team members for this purpose).
- Identify one or two individuals on the NICU team to watch for results.
- Perform a quarterly review all patients who had genomic testing to ensure results were delivered.

2. Should all patients who have rGS be seen by outpatient Genetics? When?

Recommendations from Group

Baby Badger Network ECHO

Problem of Practice Summary & Recommendations

Session Date: October 8 2026

- Preferably, all infants who receive genomic testing (regardless of the result) should be referred to outpatient genetics.
- Add language to the discharge summary which informs the PCP to refer to Genetics for follow up

Problem of Practice 2

What to do when interpretation of the findings isn't straightforward? The patient's phenotype had some overlap with the phenotypes of all conditions, specifically polydactyly. The likely pathogenic variant was paternally inherited, and dad had a h/o polydactyly.

Recommendations from the Group

- It happens. It is not uncommon for genomic testing to reveal the gaps in our understanding of the genetic causes of disease and a specific case. These gaps in knowledge can result in an uncertain interpretation.