

Baby Badger Network ECHO

Problem of Practice Summary & Recommendations

Session Date: April 9, 2026

Presenter: Jennifer Stengrevics, PAC Children's Wisconsin, Medical College of Wisconsin

Problem of Practice: How can providers introduce concern for a genetic condition to a family in an honest manner without overwhelming them?

Brief Summary:

Clinical Perspective

Female born SGA at 37w1d. Pregnancy was notable for IUGR. cfDNA and parental carrier screening were un concerning. Infant required CPAP from 3-5 minutes of life for respiratory distress. Parents were told their baby was healthy and beautiful.

At DOL2, infant was transferred from NBN to NICU for respiratory failure, seizure-like activity, apnea, hypertonia, hyperreflexia. Initial workup included: sepsis, TORCH, EEG, head u/s, renal u/s, brain MRI, C-spine MRI and ECHO. Physical exam findings included multiple dysmorphic features. Providers were Initially concerned for large chromosomal abnormality, but as the NICU course continued and the phenotype became more compelling, there were strong concerns for genetic syndrome and a genetics consult was requested.

Parent Perspective

Parents experienced an abrupt shift from a "healthy and beautiful" newborn to one with "dysmorphic features", having significant problems and who may have a genetic condition. Parents were overwhelmed and confused given "normal" prenatal genetic screening. They were hesitant to talk with genetics. They were reluctant to pursue broad genomic testing, but agreed to a karyotype, which was normal. Genetics met with family again when feeding difficulties and bilateral SNHL were identified. By this time, parents had had time to process and ultimately agreed to broader testing.

Diagnosis

A large pathogenic deletion on 8p and a large pathogenic duplication on 16p were identified, suggestive of an unbalanced chromosomal translocation. Reanalysis of the karyotype and FISH studies confirmed der(8)t(8;16)(p23.2;p13.2) with good genotype:phenotype correlation. The size of these CNVs were under the detection limit of a karyotype (5-10 MB).

Unbalanced translocations can result when one parent harbors a balanced translocation. Parental testing was recommended to determine whether either parent harbors a balanced translocation and provide recurrence estimated.

Parent Perspective

Parental testing was declined. Will be revisited in the outpatient setting.

Clinical Take Aways

- Negative karyotype does not rule out chromosomal disorder
- Advanced genomic testing is essential in complex cases
- Reanalysis is critical when suspicion remains high
- Reconsider prior results when new data emerges
- Low risk prenatal screening often provides false reassurance to parents.

Baby Badger Network ECHO

Problem of Practice Summary & Recommendations

Session Date: April 9, 2026

Presenter: Jennifer Stengrevics, PAC Children's Wisconsin, Medical College of Wisconsin

Summary of Discussion

We reminded ourselves that

- Parents may be grieving the loss of what they expected for their baby and the birthing experience the transition is scary and identity shifting
- Parents may feel a loss of control as more and more tests come back "abnormal". It is important to let them know genetic testing is a choice.
- As parents cope over time, they may be more agreeable to genomic testing.
- It may be helpful to talk about genomic testing as a tool to help determine how to care for baby, rather than looking for a genetic condition or label.

How can "dysmorphic features" be discussed with family

- Being "beautiful" or "cute" is not mutually exclusive with being "dysmorphic".
- A dysmorphic feature, by itself, is not concerning. However, certain patterns can be suggestive of a genetic condition.
- "Physical features" may be a softer term
- May be helpful to point out (or even celebrate) the "normal" things baby is doing
- Point out that baby still looks like other family members.

If a parent Googles the term "dysmorphic", the definition will include: having a malformed, misshapen, or abnormal body part

Discussion Focus