

Baby Badger Network ECHO

Case Recommendation Summary

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Brief Case Summary:

Newborn with prenatally identified dilated bowel loop and polyhydramnios. Conceived by IVF. Maternal history of Hirschsprung's. Admitted for respiratory insufficiency. Genetics consult noted macrocephaly, retrognathia, high arched palate, hypotonia and unexplained hyperventilation. rWGS was ordered on DOL 2. On DOL 7, genomic testing resulted and was consistent with a diagnosis of congenital myopathy. Infant was transferred to a Level IV NICU with a known genetic diagnosis.

Discussion Focus

- Having genomic test result and report available to recipient hospital at time of transfer was critically important in managing care and in assessing parent understanding and concerns.
 - The rapid diagnosis prevented the need for additional workup. For example, PHOX2B was on the differential, but not able to be detected on the genomic test that was ordered. Had genomic testing returned neg, PHOX2B analysis would have been recommended.
 - Recipient hospital genetics team was able to assess parents' understanding of results and what they had read from "Dr. Google" in the interim.
 - In most cases, this information is not available / accessible in the recipient hospital EMR. Docs scanned into Media tab are not viewable in CareEverywhere.
 - The genomic test report allows for confirmation of the gene name, c., p. and classification. The test report may also have important modeling information and cited literature for the variant.
 - Delays in accessing prior genetic testing (pending or resulted) may delay the recipient hospital's clinical recommendations.

Recommendations

- It is extremely helpful when the birthing/outside hospital provides patient information (pertinent HPI, pending and resulted genetic tests, genetic test reports) to recipient hospital in advance of transfer.
 - Proactively obtain names / contact information of recipient hospital NICU providers.
 - Identify an individual on birthing hospital / outside NICU team for this role.

Commented [AH1]: I didn't know what OSH stood for. Is birthing hospital the best alternative here?