HUDSONALPHA
INSTITUTE FOR BIOTECHNOLOGY

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Family study ID#: 000XX-C

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This page summarizes your genetic test results from research performed at the HudsonAlpha Institute for Biotechnology through the Genomic Diagnosis in Children with Developmental Delay project. Please see the accompanying result report for more detailed information.

- We identified a genetic variant that we believe is the cause for your child's neurological symptoms.
- The genetic variant is in the SHOC2 gene known to be associated with *Noonan-like syndrome with loose anagen hair,* characterized by cardiac anomalies, cognitive delays, growth deficiency, characteristic facial features and often sparse slow growing hair.
- Based on information provided by your physician, your child's symptoms are consistent with this diagnosis.
- The likelihood of having another child with SHOC2 related symptoms would be low (<1%) because the gene variant is *de novo* (a new variant found only in the affected child and not present in either parent). Other family members would also not be expected to have an increased chance of having a child with the condition.
- If the affected child were to have children, each child would have a 50% chance of inheriting the variant and the condition.
- Our understanding of genetic variation and how it contributes to developmental delay and neurologic symptoms will continue to change and improve over time. We encourage you to continue follow-up with your healthcare providers to keep up to date on new information.

Thank you for your participation in this research study. It has been a pleasure working with your family.

Sincerely,

Kelly East, MS, CGC
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