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| --- | --- |
| A picture containing text, person, indoor, child  Description automatically generated **Alexis & Noah’s Case** | **NOTES** |
|  **Phenotype** |   |
| **Preliminary Diagnosis** |   |
| **Genetic Variation(s)** |   |
| **Laboratory Assertion(s)** |   |
| **Variant Information:*** Asserted interpretation listed in **ClinVar**
* HGVS names from **ClinVar**
* Is population data available in **dbSNP**?
 |   |
|   |
|   |
| **Gene Information in****NCBI Gene:*** Symbol and Name
* Gene Summary
* Tissue Expression information
* Gene Ontology information
 |   |
|   |
|   |
|   |
| **Ultimate Impacted Biomolecule based on:*** **GDV**to view the chromosome and gene region
* **RefSeqGene Graphics** view of gene region and transcript(s)
* **RefSeq Protein Graphics** view of protein and domains
* **CDD or iCn3D** to view a structure*, as needed*
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|  |
| **Proposed Molecular Mechanism of Variant Impact** |   |
| **How does this relate back to the phenotype (symptoms/clinical features & diagnosis)?** |  |