|  |  |
| --- | --- |
| **Jonathan’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* |  |
|  |
|  |
|  |
| **Proposed Molecular Mechanism of Variant Impact** |  |
| **How does this relate back to the phenotype (symptoms/clinical features & diagnosis)?** |  |