|  |  |
| --- | --- |
|  **David’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)?** |  |