



Leslie'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)?