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Appendix: Interpretation of Sequence Analysis Results

Created: April 5, 2007; Revised: May 19, 2016.

Types of sequence variants that may be detected [1]

- Pathogenic sequence variants reported in the literature
- Sequence variants predicted to be pathogenic but not reported in the literature
- Variants of unknown clinical significance [2]
- Sequence variants predicted to be likely benign but not reported in the literature
- Benign sequence variants reported in the literature or in population databases

Possibilities when a sequence variant is not detected

- Patient does not have a pathogenic variant in the tested gene (e.g., a sequence variant exists in another gene at another locus).
- Patient has a sequence variant that cannot be detected by sequence analysis (e.g., a large deletion).
- Patient has a sequence variant in a region of the gene (e.g., an intron or regulatory region) not covered by the laboratory's test.

1. Adapted from Standards and Guidelines for the Interpretation of Sequence Variants: A Joint Consensus Recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology [2015]

2. Family studies may be used to determine if a sequence variant segregates with the phenotype or occurred *de novo*.

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