Supporting Evidence

- Multiple lines of computational evidence supporting a deleterious impact on gene product (e.g., in silico tools: PolyPhen2, SIFT, MutationTaster; splice site: MaxEntScan, NNSplice)
- Conservation data (nucleotide: phyloP; amino acid: Orthologs)
- Additional data
  - In tumor DNA, loss of the allele (co-segregation with disease; moderate evidence)
- Segregation data
  - Non-segregation with disease
- Sample data
  - Multiple affected family members
- Publication data
  - Published in the same variant

Pathogenic Evidence

- Multiple lines of computational evidence supporting a deleterious impact on gene product (e.g., in silico tools: PolyPhen2, SIFT, MutationTaster; splice site: MaxEntScan, NNSplice)
- Conservation data (nucleotide: phyloP; amino acid: Orthologs)
- Additional data
  - In tumor DNA, loss of the allele (co-segregation with disease; strong evidence)
- Segregation data
  - Segregation with disease in multiple affected family members
- Sample data
  - Multiple affected family members
- Publication data
  - Published in the same variant

Likely Pathogenic Evidence

- Multiple lines of computational evidence supporting a deleterious impact on gene product (e.g., in silico tools: PolyPhen2, SIFT, MutationTaster; splice site: MaxEntScan, NNSplice)
- Conservation data (nucleotide: phyloP; amino acid: Orthologs)
- Additional data
  - In tumor DNA, loss of the allele (co-segregation with disease; moderate evidence)
- Segregation data
  - Segregation with disease in multiple affected family members
- Sample data
  - Multiple affected family members
- Publication data
  - Published in the same variant


Supplemental material

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