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| Patient | Variant | ACGM evidence |
| 1 | NF1 c.5488C>T | PS1 - Same amino acid change as a previously established pathogenic variant regardless of nucleotide changePM2 - Absent from controls (or at extremely low frequency if recessive) (Table 6) in Exome Sequencing Project,1000 Genomes Project, or Exome Aggregation Consortium |
| 2&3 | EEF1A2 c.505G>A | PM2 - Absent from controls (or at extremely low frequency if recessive) (Table 6) in Exome Sequencing Project,1000 Genomes Project, or Exome Aggregation ConsortiumPP1 - Cosegregation with disease in multiple affected family members in a gene definitively known to cause thediseasePP2 - Missense variant in a gene that has a low rate of benign missense variation and in which missense variantsare a common mechanism of diseasePP3 - Multiple lines of computational evidence support a deleterious effect on the gene or gene product(conservation, evolutionary, splicing impact, etc.)PP4 - Patient’s phenotype or family history is highly specific for a disease with a single genetic etiology |
| 4 | NEWXMIF c.2096C>A | PVS1 - null variant (nonsense, frameshift, canonical ±1 or 2 splice sites, initiation codon, single or multiexondeletion) in a gene where LOF is a known mechanism of diseasePM2 - Absent from controls (or at extremely low frequency if recessive) (Table 6) in Exome Sequencing Project,1000 Genomes Project, or Exome Aggregation ConsortiumPM3 - For recessive disorders, detected in trans with a pathogenic variant (null allele in hemizygous male) |
| 5 | DCX c.910G>A | PS1 - Same amino acid change as a previously established pathogenic variant regardless of nucleotide changePM2 - Absent from controls (or at extremely low frequency if recessive) (Table 6) in Exome Sequencing Project,1000 Genomes Project, or Exome Aggregation Consortium |
| 6 | EHMT1 c.3310G>A | PS2 - De novo (both maternity and paternity confirmed) in a patient with the disease and no family historyPP3 - Multiple lines of computational evidence support a deleterious effect on the gene or gene product(conservation, evolutionary, splicing impact, etc.)PP4 - Patient’s phenotype or family history is highly specific for a disease with a single genetic etiology |
| 7 | 1q21.1 microduplication | Contiguous gene duplication syndrome |