ACMG RECOMMENDATIONS FOR CNV CURATION

Riggs ER et al. Technical standards for the interpretation and reporting of constitutional copy-number variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics (ACMG) and the Clinical Genome Resource (ClinGen). Genet Med. 2020 Feb;22(2):245-257. PMID: 31690835.

- This paper establishes a semi-quantitative point-based scoring method for CNV pathogenicity
- Supplementary Material 1 has a helpful walk-through of CNV curation concepts
- Supplementary Material 3 has valuable case examples with demonstrations of curation tools
- The scoring calculator is applicable to rare CNVs suspected to be autosomal dominant with complete penetrance
- The scoring calculator is not intended for relatively common variants that may have incomplete penetrance – such variants require large case-control studies that may be found on PubMed or ClinGen
- While the scoring calculator is not applicable to all CNVs, the concepts presented here are very helpful for the curation of most CNVs

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