

Assertion criteria for CNVs for submission to ClinVar, 2017-01-13 :

Does CNV segregate with disease?	yes	likely pathogenic
	no	likely benign
Large CNV (Mb size)?	yes	likely pathogenic
	no	likely benign
De novo CNV?	yes	likely pathogenic
	no	likely benign
Are similar CNVs reported in other databases/the literature?	yes	This will be taken into account in the assertion
	no	If segregation/de novo status is not yet tested, the CNV will be asserted as variant of unknown significance