

PanelApp Tag	Tag description
anticipation	Where the age of manifestation reduces and the severity of symptoms of a genetic condition increases from one generation to the next.
nucleotide-repeat-expansion	For this phenotype, nucleotide-repeat-expansions can cause the disorder.
early-onset	This relevant phenotype shows early onset.
adult-onset	This relevant phenotype shows only adult onset.
CNV	There are pathogenic copy number variants reported for this gene.
deletions	There are pathogenic deletion variants reported for this gene
dominant-negative	Gene2Phenotype lists dominant negative as the mutation consequence.
founder-effect	There is only a single variant that has been reported to have a founder effect in a particular population, or a shared haplotype has been observed. Genes with a founder-effect tag are rated Red as there is not currently enough evidence that other variants in the gene are disease causing
gene-duplication	There are pathogenic gene duplication variants reported for this gene.
missense	Only missense variants have been reported for this gene for this phenotype/disorder.
monogenic-polygenic:	Both monogenic associations and polygenic associations have been reported for this gene.
polygenic	Only associations in combination with other variants in other genes have been reported. Genes with a 'polygenic' tag are rated Red as monogenic variants are reported for the 100,000 Genomes interpretation pipeline
multifactorial	The genetic association is in combination with environmental/other factors. Note that these genes are <u>not</u> Green genes.
new-gene-name	The HGNC gene symbol has been updated compared to the gene symbol on PanelApp. These symbols will be updated in-line with Ensembl.
non-coding-known-pathogenic	There are non-coding pathogenic variants reported for this gene
treatable	There is a treatment that targets variants in this gene, or there are treatment options for this disorder based on a diagnosis from variants in this gene, or supplementation may prevent or alleviate symptoms.
mosaicism	Mosaicism has been reported
x-linked over dominance	Gene2Phenotype records dominant negative as the mutation consequence
promoter	Pathogenic variants have been reported in the gene promoter, or variations to promoter length have been associated with the phenotype.
regulatory region	Pathogenic variants have been reported in the gene regulatory region known to influence expression of the corresponding gene.
structural-variant	Pathogenic structural rearrangements have been reported
sva	Pathogenic short interspersed nuclear element, variable number of tandem repeats, and Alu composite have been reported
pathogenic-synonymous	For genes with synonymous variants with proven pathogenicity

pharmacogenetics	Genes in which variants can affect drug response e.g. variants in CYP2C8 are associated with cerivastatin-induced rhabdomyolysis.
y-chromosome	Gene is encoded by the Y-chromosome.