SUBMISSION DATE:

Enter sequence variants from your cohort into the table below. Please begin each variant on a new line. For compound heterozygous variants, enter a linking ID in the first column. You need only enter details unique to the second variant.

	Gene			
##Local ID / Linking ID	**Gene symbol**	**Genome build**	**Reference sequence**	**HGVS**
Local ID Optional, but highly recommended. The stable unique identifier your organisation uses to identify this variant. Linking ID REQUIRED if submitting compound heterozygous variants.	Required. This must be the preferred symbol from HGNC. (https://www.genenames.org/)	Required. The genome assembly that was used to call variants in this submission. All variants in this file should use the same genome assembly. Allowed values: NCBI36, hg18, GRCh37, hg19, GRCh38, hg38.	Required. The reference sequence for the HGVS expression provided in the next column, e.g. NM_000492.3, NG_016465.3, or NC_000007.13.	Required. Provide the c. or g. portion of the nucleotide HGVS expression for the variant(s) being reported related to condition.
Please start your submission in the next row. For compound heterozygous variants, enter the details of the second variant in a new row and provide a linking ID.				

Variant definition

Chromosome	Start	Stop	Reference allele	Alternate allele	Molecular consequence
Required. Provide the chromosome number.	Required. The start location for the reference allele in chromosome coordinates.	Required. The stop location for the reference allele in chromosome coordinates.	Required. The reference allele for the submitted variant. For an insertion, enter '-'	Required. The alternate allele for the submitted variant. For a deletion, enter '-'	Required. The predicted effect of a variant on a biological sequence as described by Sequence Ontology. Select all that apply.

Interpreted condition - the condition for which you are interpreting the variant			Clinical significance		
Condition ID type	**Condition name**	**Condition comment**	**Variant classification**	**Date last evaluated**	**Assertion method**
Required. Please specify the reference database for the condition name. Allowed values are: OMIM, HPO, MONDO.	Required. Please specify the condition name/ID curated against. Standard ontology term required.	Optional. Free text describing the condition observed in individuals with this variant.	Required. Click cell for drop-down list; allowed values include Pathogenic, Likely pathogenic, Uncertain significance, Likely benign, Benign, Cannot be classified.	Optional. Date the clinical significance of the variant was last evaluated by the submitter. If not available, leave blank. Please use the format yyyy-mm-dd.	Strongly recommended. The method used to for interpretation. (e.g. ACMG guidelines)

			Details of test and individuals tested		
ACMG evidence criteria (e.g. BA1)	**Interpretation summary**	**Mode of inheritance**	**Allele origin**	**Zygosity**	**Affected status**
Required. Provide the ACMG evidence criteria applied. Select all that apply.	Required. Provide a summary of the variant interpretation.	Required. Supported values are from the Human Phenotype Ontology (HPO). Allowed values include: Autosomal dominant inheritance, Autosomal recessive inheritance, Autosomal unknown, Codominant, Genetic anticipation, Mitochondrial inheritance, Oligogenic, Sex-limited autosomal dominant, Somatic mutation, Sporadic, X-linked inheritance, Y-linked inheritance, Other. If you provide Other, please specify, e.g. Other:new mode of inheritance.	Required. The genetic origin of the variant for individuals in each aggregate observation. Allowed values: germline, de novo, somatic, maternal, paternal, inherited, unknown, uniparental, biparental. Note that biparental and uniparental are intended for the context of uniparental disomy. If you'd like to indicate zygosity, please report counts of homozygotes and heterozygotes in columns BV-BY. For *de novo* variants, please indicate "*de novo* ", not the origin of the chromosome.	Required. Indicate zygosity for the variant in the individual. Allowed values: Single heterozygote, compound heterozygote, homozygote, hemizygote.	Required: Indicate whether or not the individuals in each aggregate observation had the interpreted condition. Allowed values: yes, no, unknown.

				Details of testing results	
Clinical features	**Comment on clinical features**	**Tissue**	**Other family members tested**	**Family ID**	**Segregation observed**
Required. Provide a list of clinical features observed in these individuals. HPO terms may be used to indicate the clinical features. Separate multiple terms with a semicolon. If you wish to provide a free text comment on the observed phenotype, please use the "Comment on clinical features" column.	Optional. To provide a free text explanation of clinical features provided in the previous column, e.g. to describe the progression of disease or diagnosis.	Optional. Tissue sampled to assay the variation. Highly recommended for somatic variants.	Required. List other family members tested. If no family members have been tested enter '-'	Strongly recommended. If you would like to indicate which cases (represented on different rows) were part of the same family, please provide an anonymous identifier for the family.	Required. Indicate whether segregation of the variant and reported condition was observed in this individual's family.

	Population data				Methods.
Number of individuals with variant	**Number of homozygotes**	**Number of single heterozygotes**	**Number of compound heterozygotes**	**Number of hemizygotes**	**Test name or type**
Required. The total number of individuals with the variant allele.	Strongly recommended. Number of individuals homozygous for the variant.	Optional. Number of individuals who are single heterozygotes for the variant.	Optional. Number of individuals who are compound heterozygotes, where another pathogenic variant was identified.	Optional. Number of individuals who are hemizygous for the variant.	Strongly recommended. The name or type of test used to identify this variant. If the test is registered with the Genetic Test Registry (GTR), please submit the GTR Test ID, e.g. GTR000074114.1.

			General Comments
Platform type	**Platform name**	**Testing laboratory**	**Comment**
Strongly recommended. Type of platform used for data capture, e.g. next-gen sequencing, microarray.	Optional. Name of platform used for data capture, e.g. HiSeq, MiSeq.	Required. Please indicate the name of the testing lab.	Optional. Free text. You can put important information that isn't included in any of the other fields here.