Instructions for ClinVar submission spreadsheets

This page provide general information about filling in ClinVar's submission spreadsheet. The divisions in this page primarily correspond to the tabs on the worksheet, but we also give special attention to submitting information about disease/phenotype and your interpretation of clinical significance.

Not every column is described here; instructions are also included in each column in the spreadsheet itself.

Checklist for faster processing

Help your ClinVar curator process your submission faster!

- Provide data in all required fields
- Validate your HGVS expressions with VariantValidator or Mutalyzer
- Do not modify the column headers
- Do not modify the cell validation; use allowed values when there is a list
- Check the instructions for each column on the spreadsheet for correct format and separators for a list

SubmissionInfo tab

ClinVar is asking all submitters to register in the Submission Portal once rather than provide submitter information in the

SubmissionInfo tab with every submission. If you send your submission to ClinVar in the Submission Portal, you do not need to provide submitter/organization/study information on the SubmissionInfo tab.

Submitter information

ClinVar asks for information about both the submitting organization and the people associated with that organization.

- On the spreadsheet, 'submitter' refers to the people from the submitting organization.
    - People are associated with the organization, but their names are not directly on the submission.
- On the website and in downloadable files, 'submitter' refers to the submitting laboratory or other organization.
- The names and other information for people associated with the submitting organization are only displayed on the submitter page, depending on the submitter type .
    - See an example of the submitter page.

Submitter type and submitter role

- Submitter type indicates whether you are the person for ClinVar to contact about the submission, and whether you would like your name, email, etc to be public on the ClinVar submitter page.
- If your submitter type is "Contact" or "Private", do not provide a Submitter role.
- If your submitter type is Contact; Public" or "Public", also provide your Submitter role. This is a label that is used on the page for the submitting organization.

Submitter type	Submitter role	Result
Contact	N/A	ClinVar will contact you with questions about the submission. Your name, email etc will not be publicly available on the ClinVar website.
Contact; Public	any role	ClinVar will contact you with questions about the submission. Your name, email etc will be publicly available on the ClinVar website.
Private	N/A	Your name, email etc will not be publicly available on the ClinVar website.
Public	any role	Your name, email etc will be publicly available on the ClinVar website.

Institution and organization

For institution, please provide the larger group that you are part of:

- Your university
- Your hospital
- NIH

For organization, please provide the specific group that you are part of:

- a name for your lab, e.g. Smith laboratory or Molecular Genetics Diagnostic Laboratory
- if there are multiple parts to describe your specific group, include them as the organization, not the institution.
    - Smith Lab, Dept of Molecular and Human Genetics
    - Molecular Genetics Diagnostic Laboratory, Department of Pathology
- your organization may also be a clinic, an LSDB, or other group

Study name

Provide a study name only if

- the variants were interpreted as part of a named study, such as ClinSeq
- the study name helps to define the submitting organization

Submission name

The submission name is a name for a batch of variant interpretations.

- Any submission may have a submission name, but it is required if you provide either a study name or a study description.
- A submission name is particularly useful if you are submitting a large set of records to ClinVar to be referenced in a publication.
    - You can reference both the submission name and your ClinVar accession numbers in your paper.

    - We recommend use of a submission name, because providing a range of accessions may not make each of your records as discoverable in ClinVar.
- The submission name is not displayed on the website, but you can search for it as [Submitter Batch]
    - e.g. search for EGL003[Submitter Batch]

Variants

ClinVar welcomes submissions of variants interpreted as homozygotes, haplotypes and compound heterozygotes, and we offer the following guidance:

- if each variant has been interpreted independently, please submit the variants separately to ClinVar. Distinct interpretations for each variant may be more useful to those using data in ClinVar.
    - For example, if you observed two variants in compound heterozygosity and you determined that they are pathogenic for an autosomal recessive disease, submit each variant on a separate row as a pathogenic variant for that disease.
    - The submission for each variant can note that it was observed with the other variant and the mode of inheritance.
- if multiple variants have been interpreted together because you cannot interpret them independently, submit them on one row as the appropriate combination, either as a haplotype or as a compound heterozygote.
- if you are submitting a variant that was observed in a homozygote, do not submit the variant as an HGVS expression for the homozygote, e.g. c.[105C>A]; [105C>A]
    - submit the single variant c.105C>A
    - provide the clinical significance based on the single variant's contribution to disease
    - fill in the column "mode of inheritance" if possible
    - if you provide aggregate evidence on the Variant tab, fill in the column "Number of homozygotes"
    - if you provide individual evidence on the CaseData tab, enter "homozygote" in the column "Zygosity"

Sequence variants

HGVS expressions

- Check that your HGVS expressions are valid with VariantValidator or Mutalyzer.
- On the lite spreadsheet template, enter the HGVS expression in the 'HGVS' name column.
- On the full spreadsheet template, enter the accession.version number in the 'Reference sequence' column and the c./g. portion of the HGVS expression in the 'HGVS' column.
    - We only accept NCBI RefSeq accession numbers as the reference sequence due to technical constraints (namely, that we do not have alignment datasets for GenBank accessions).
- Do not include the p. HGVS expression in these columns. It may be provided in the 'Alternate designations' column instead.
    - If you have information on multiple nucleotide changes that result in the same protein change, submit each nucleotide change on a separate row.
- Spreadsheets with examples of valid HGVS expressions that ClinVar accepts and invalid HGVS expressions and corresponding error messages are available.