

2018-01-31 Web Release

Release notes, January 31, 2018

- In the "Submitted interpretations" table, links to "Evidence details" were fixed.
- In the "Submitted interpretations" table, redundant citations were removed.
- A bug that caused an "Error 404" in the Evidence table was fixed, e.g. preview.ncbi.nlm.nih.gov/clinvar/variation/437909/evidence/
- A bug that results in no data shown for some citations in the table for "Citations for this variant" was fixed.
- The "Submitted interpretations" table now shows the count of PubMed IDs, instead of a list.
- On the Evidence table, the "Choose columns" button is now visible when many observations are in the table, e.g. preview.ncbi.nlm.nih.gov/clinvar/variation/9/evidence/
- Search results now display data for protein change and accession (note that the VCV accession numbers are not versioned at this time, so we do not recommend citing them yet)

Known issues

General

- new records that are displayed in the "classic" pages may not be available yet in the new display
- a link to the Advanced search page is not available
- options to download and print the page are not available

Top section in blue

- Links to "See interpretations for this variant in combination with other variants" are incorrect (note: this link is only found on some records, e.g. a simple variant that has been interpreted as part of a haplotype or genotype)
- Variation ID is not displayed; however, the Variation ID is part of the VCV accession number, e.g. for VCV000137919, the Variation ID is 137919

Variant details tab

- Variation IDs for each component haplotype and variation are not displayed on genotype and haplotype records
- Molecular consequence is not displayed for all HGVS expressions on transcripts
- For haplotype and genotype records, links in the section "This haplotype includes the following variants:" are incorrect.
- All allele frequencies are displayed as "Minor allele frequency"
- Allele frequencies are not displayed from all sources (1000 Genomes, GO-ESP, and ExAC)

Conditions tab

- the section for "Clinical features observed in individuals with this variant" displays the HPO identifier, rather than the HPO term
- the term "not provided" links to MedGen with no result

Genes tab

- the table of genes is not sortable; currently genes are listed in alphabetical order
- links to the ClinGen Dosage Sensitivity Map are not provided

Submitted interpretations table

- some "Comments on clinical significance" (displayed in the Submitter column) are not displayed; in the "classic" view, this is the comment displayed in the "Summary evidence" tab, under "Description"

Evidence page

- the table on this page is wide and the scrollbar is attached to the bottom of the table, not to the page window. If it's not visible, please scroll down to find the scrollbar.
- data for several columns is not displayed, including number of families, clinical features, zygosity, result, and sex

Citations table

- The table includes some citations specific to the condition, but not to the variant

December 13, 2017

Alpha release of ClinVar Web 2.0

The alpha release represents a redesign of the ClinVar Variation pages.

Notes on data

This release is for users to review the new design only. The data underlying the page has not gone through all content checks yet so please continue to use the data that is displayed on the "classic" ClinVar Variation pages.

The data in this alpha release is synchronized with the data on the ftp site for the beta release of our new variant-centric XML:

ftp://ftp.ncbi.nlm.nih.gov/pub/clinvar/xml/clinvar_variation/beta/

It is not necessarily synchronized with the data displayed on the "classic" ClinVar variation pages. If you have concerns about data discrepancies between the two displays, please contact us at clinvar@ncbi.nlm.nih.gov.

Features

Improvements to this display include:

- Updated design
- Introduction of VCV accession numbers, with versioning
- Clearer representation of interpretations per condition
- Improved display of information about the gene(s) for the variation
- Improved display of evidence; more evidence types are included and data is not represented redundantly in multiple tabs
- Easier access to all citations reported to ClinVar for a variation
- Explicit representation of "included" variations; in other words, variations that do not have a direct interpretation in ClinVar but that are part of a haplotype or genotype that does have an interpretation in ClinVar
- Feedback button for your questions, comments, and suggestions

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Top section in blue

- Links to "See interpretations for this variant in combination with other variants" are incorrect (note: this link is only found on some records, e.g. a simple variant that has been interpreted as part of a haplotype or genotype)

Variant details tab

- Variation IDs for each component haplotype and variation are not displayed on genotype and haplotype records
- Molecular consequence is not displayed for all HGVS expressions on transcripts
- For haplotype and genotype records, links in the section "This haplotype includes the following variants:" are incorrect.

Conditions tab

- the section for "Clinical features observed in individuals with this variant" displays the HPO identifier, rather than the HPO term
- the term "not provided" links to MedGen with no result

Genes tab

- the table of genes is not sortable; currently genes are listed in alphabetical order

Submitted interpretations table

- links to "Evidence details" are broken. Please use the link at the top of the column to "See all" supporting information.

Citations table

- some citations are listed more than once in the same table

Evidence page

- the "Choose columns" button is not visible when more than 8 observations are in the table
- the table on this page is wide and the scrollbar is attached to the bottom of the table, not to the page window. If it's not visible, please scroll down to find the scrollbar.

Search results

- search results do not have data for protein change or accession