2019-04-04 Web Release

Release notes, April 4, 2019

Coming soon: the new design for ClinVar pages will become the default view! We anticipate this change to happen mid-late May. Thank you to all of our users who provided feedback on the new design while it was in its alpha release. Once it is in production, we will continue to improve the display and usability of the website. Use the feedback button on each Variation page or send us your comments at clinvar@ncbi.nlm.nih.gov.

When the new design becomes the default view, URLs on the preview domain (e.g. https://preview.ncbi.nlm.nih.gov/clinvar/variation/9/) will no longer work. Only the new design will be accessible on www; the old page design will not be accessible.

The redesigned pages have several new features which were described in a blog post [https://ncbiinsights.ncbi.nlm.nih.gov/2017/12/18/clinvar-unveils-new-more-intuitive-variation-display/]. One notable feature is the VCV accession number. This accession number represents the variant or set of variants represented by the ClinVar record. This new accession number means that the web pages will be accompanied by some changes to searching ClinVar and to using E-utilities.

Changes to searching ClinVar

There will be two new ways to search the web pages:

- Query with a VCV accession number to view the latest record for that variation
- Query with a VCV accession.version number to view the latest record for that version of the variation record
    - Note that query by accession.version does not work the same way on NCBI's general search page: https://www.ncbi.nlm.nih.gov/search/?term=. On that page, query by accession.version returns the latest record for the latest version, regardless of the version number in your query.

Querying for a Variation ID will remain the same, returning the latest record for that variation.

Searching for anything else, like a gene symbol or a disease name, will also work the same way.

Changes to E-utilities

The new web pages use ClinVar's new variation-centric XML as the source of data and new accession numbers, beginning with VCV. Thus E-utilities for ClinVar are being updated to support queries for VCV accessions and to return the new XML format.

E-search

- Query with a VCV accession number (without the version) or a Variation ID to retrieve the Variation ID, which can be used with E-fetch to retrieve the latest VCV record.
- Query with a VCV accession and version is not supported. If you know the accession and version number of the record you want, use that as your query for E-fetch.
- Query with any other search term (such as a gene symbol or a disease name) to retrieve all Variation IDs that satisfy the query. Those IDs can be used with E-fetch to retrieve the latest VCV records.

E-fetch

- Query by VCV accession number (without the version) or a Variation ID to retrieve the latest VCV record.
- Query by VCV accession and version to retrieve the latest XML for that accession.version.

The valid types will be:

- VCV record (ret_type = vcv) [Note: this ret_type is new]
- RCV record (ret_type = clinvarset)
- variation record (ret_type = variation).
    - Note: the variation records (ret_type = variation) will continue to be available for approximately six months BUT they will not be updated. Please update your scripts to use ret_type vcv and to use our new VCV records for variant-centric data.

Note:

For ret_type = vcv, you will also need to provide an id_type, e.g.

https://www.ncbi.nlm.nih.gov/entrez/eutils/efetch.fcgi?db=clinvar&rettype=vcv&id=9,10&id_type=variationid

E-summary

- Query with a Variation ID to retrieve the document summary for a VCV record.

Release notes, March 15, 2018

- A link to ClinVar's Advanced search function was added under the search bar.

- Records for structural variants that affect >50 genes now load without issue. For example, see https://preview.ncbi.nlm.nih.gov/clinvar/variation/442998/#id_third
- In the "Submitted interpretations" table, only standard values for clinical significance are displayed by default. If a non-standard value was provided in the submission, that non-standard value is displayed as a pop-up. For example, see SCV000057251.3 on the following page: https://preview.ncbi.nlm.nih.gov/clinvar/variation/40399/
- In the "Submitted interpretations" table, the comment on clinical significance was moved to the "Supporting information" column. For example, see SCV000061612.4 on the following page: https://preview.ncbi.nlm.nih.gov/clinvar/variation/40399/
- On the evidence page, the scrollbar is now attached to the page window, so it should always be visible.

Known issues

General

- new records that are displayed in the "classic" pages may not be available yet in the new display
- 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

Evidence page

- 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

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)

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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

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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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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