2013-08-30 Web release

Tabular display

- The gene symbol now has a tooltip indicating that the symbol links to the Gene database.
- For the clinical significance filter, the option “Conflicting interpretations” was added.
- For the molecular consequence filter, the options “Near gene” and “UTR” were added.
- A new filter for gene symbol was added.

Full display

- The number of observations now includes only variants reported from clinical testing, not variants reported from literature curation.
- The variant type was added to the Allele description.
- The genomic location, in the format Chr1:1-100000 (on Assembly GRCh37.p10), was added to the Allele description.
- A link to the 1000 Genomes browser was added to the Allele description.
- Comments provided by NCBI staff during curation are now displayed in the Allele description.
- For compound variants, a distinct molecular consequence report per variant is provided in the Allele description.
- In the allele description, multiple values of protein change and other names are now separated by a semi-colon.
- The Clinical Assertions tab now includes links to websites that are provided as citations supporting the clinical significance.
- The Clinical Assertions tab now indicates when a single submitter has provided data from more than one method, e.g. both curation and clinical testing.
- Pages for previous versions of RCV records now include a message indicating that the record has been updated with a link to the current version.

Additional pages

Submission summary

A overview section was added to the page that summarizes data submissions (http://www.ncbi.nlm.nih.gov/clinvar/submitters/). Total numbers of accessioned submissions, genes represented, variations represented, and submitted are available.

Individual submitter pages

On individual submitter pages, such as :

www.ncbi.nlm.nih.gov/clinvar/submitters/21766/

the phenotype table now has links in the Submissions column to retrieve all ClinVar records from the submitter for the listed phenotype.

FTP site

A preliminary export of key data elements from ClinVar is being tested. It is provided as a tab-delimited file, organized around specific reports of variation, even if the variants have not yet been mapped to sequence coordinates.