2016-02-04_Web_Release

Release notes, February 4, 2016

Query hint

Did you know you can query ClinVar by identifiers from the HUGO Gene Nomenclature Committee (HGNC)?

ClinVar supports retrieval of data by searching on HGNC identifiers for genes, based on the new field HGNC (*e.g.* https://www.ncbi.nlm.nih.gov/clinvar/?term=1884[hgnc]). For queries to the web site, you can use the numeric value with the field name, e.g. 1884[hgnc], or the concatenation of HGNC with the id, *e.g.* HGNC:1884. Please remember if you enter only a number in ClinVar's web site (i.e. without a field name), it tries to process that number as a VariationID.

Testing Laboratory

Submissions to ClinVar may be provided by a group other than the laboratory which did the genetic testing, such as a genetics clinic. For these submissions, we ask that they indicate who the testing laboratory is. This is helpful to users in the cases where the testing laboratory has also submitted to ClinVar. The variation pages now display the name of the testing laboratory in the "Clinical assertions" tab, in the column with the Submitter and Study name, e.g.:

http://www.ncbi.nlm.nih.gov/clinvar/variation/3121/

Indication for testing

Genetics testing laboratories often do not know the actual phenotypes that were observed in a patient, but they may know the indication from testing that was provided on a test requisition form. The indication for testing can be provided in a ClinVar submission. This information is now displayed on the variation pages, on the "Supporting observations" tab, in the column with Phenotypes, e.g.:

http://www.ncbi.nlm.nih.gov/clinvar/variation/219033/#supporting-observations