

20141106_data_release_notes

Overview of changes in the release of November 6, 2014

Overview of submissions: 2014

Date	Total Submissions
Jan 01, 2014	68204
Feb 01, 2014	73492
Mar 01, 2014	83343
Apr 01, 2014	111501
May 01, 2014	112349
Jun 01, 2014	117209
Jul 01, 2014	127132
Aug 01, 2014	127557
Sep 1, 2014	143114
Oct 1, 2014	143601
Nov 1, 2014	144117

Content

Brief	Explanation
Reporting the genomic location of a variant	This month ClinVar added the reporting of genomic locations on scaffolds when there is no location on an assembled chromosome. An example is http://www.ncbi.nlm.nih.gov/clinvar/variation/151445/ , which was defined on assembled chromosomes in GRCh37/hg19 but is localized on HSCHR14_CTG1_UNLOCALIZED in GRCh38/hg38.
GeneDx submission	GeneDx added 143 new records from their clinical testing results. http://www.ncbi.nlm.nih.gov/clinvar/submitters/26957/
Pathway Genomics submission	Pathway Genomics added 166 records based on their clinical testing results. http://www.ncbi.nlm.nih.gov/clinvar/submitters/505209/
Expert panel approval for InSiGHT	InSiGHT, already a submitter to ClinVar, was formally approved by ClinGen as an expert panel. ftp://ftp.ncbi.nlm.nih.gov/pub/clinvar/ClinGen/expert_panels/InSiGHT/

New categories of HGVS expressions	<p>ClinVar is planning to revise how certain types of HGVS expressions are represented:</p> <ul style="list-style-type: none"> • HGVS expressions without an accession.version number, but consistent with the record, will be processed as "HGVS, incomplete" rather than the current mixture of 'nucleotide change' and subcategories of HGVS-like expressions. • protein expressions based on a legacy numbering system will be curated as "HGVS, legacy"
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Overview of Submissions: 2013

Date	Total Submissions
Apr 05, 2013	30333
May 01, 2013	30386
Jun 01, 2013	39047
Jul 01, 2013	39170
Aug 01, 2013	45901
Sep 01, 2013	50263
Oct 01, 2013	52047
Nov 01, 2013	64750
Dec 01, 2013	64881