20140403_data_release_notes

Overview of changes in the release of April 3, 2014

Number of submissions

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
Jan 01, 2014	68204
Feb 01, 2014	73492
Mar 01, 2014	83343
April 01, 2014	111501

Content

Brief	Explanation
Our first anniversary!	This release marks the first year of ClinVar's public presence. We thank our submitters for sharing their data and our advisors for their very useful suggestions. We still have many items on our list of improvements to make, and look forward to working with you all so that ClinVar will meet more of your needs.
Addition of data from BIC	The Breast Cancer Information Core (BIC) and ClinVar achieved a major milestone in the integration of data about variants in BRCA1 and BRCA2. Data in the BIC were submitted to ClinVar with the interpretations as in the BIC. We expect reviewed interpretations from ENIGMA in the coming months.
improved representation of local duplications	Staff reviewed submissions at the same location that had been described as either an insert or a duplication in HGVS. These were merged as appropriate into one record, and represented according to the HGVS standard as dup. Please let us know if you identify some that our review missed.

Representation of locations on GRCh38	This release marks the first time ClinVar is reporting locations on both GRCh37 (hg19) and GRCh38. ClinVar will continue to show the locations on the web site as GRCh37 fcoordinates or a few more months, and will report locations on both coordinates for the indefinite future. Coordinate changed on all chromosomes, but not the mitochondrion.
curated OMIM records for RECLASSIFIED - VARIANT OF UNKNOWN SIGNIFICANCE	OMIM reclassifies variants when new information is available suggesting that the variant is not pathogenic. In these cases, the phenotype name on the allelic variant is updated to RECLASSIFIED - VARIANT OF UNKNOWN SIGNIFICANCE. ClinVar staff curated records based on these reclassified allelic variants to represent the phenotype as the originally asserted phenotype and the clinical significance as "uncertain significance". Review of cases with a phenotype of 'variant of unknown significance' is continuing.