20180201_data_release_notes

Overview of changes in the release of February 1, 2018

Please also see our RSS feed for information about accessing the data.

Overview of submissions: 2018

Date	Total Submissions
Feb 01, 2018	582113

Content

Brief	Explanation
CFTR2	The CFTR2 database, an expert panel, submitted 290 interpretations, including both novel interpretations and updated records.
Ambry Genetics	Ambry submitted 15419 novel variant interpretations and updates for 17183 records.
Counsyl	Counsyl submitted 570 novel variant interpretation and updates for 2 records.
Submissions owned by ClinVar Staff	A large number of submissions based on the literature were recently removed at the request of the submitter. For 920 variants with no other submissions, ClinVar staff created submissions based on the reported publication, so that these variants were not removed from the database.
Mitochondrial variants in VCF files	Mitochondrial variants are now reported in VCF files for both the GRCh37 and GRCh38 assemblies. Previously, these variants were only reported in the files for GRCh38.
Coming soon - change to variant_summary.txt	ClinVar will add a column to the end of the file reporting the single VariationID that corresponds to the AlleleID in the first column. This column will NOT contain all the AlleleID/VariationID relationships, that continues to be provided by variation_allele.txt.gz. The new column will, however, make it easier to map from variant_summary.txt to our new variant-centric XML.
Coming soon - change in calculation of the aggregate clinical significance for RCV records	Currently, conflicts in interpretation are calculated differently for ClinVar variation (Variation ID) and variation-disease (RCV) records: https://www.ncbi.nlm.nih.gov/clinvar/docs/clinsig/#conflicts Coming soon, we will change the calculation for RCV records to be the same as for Variation records, namely: • Conflicts will be reported between three levels of pathogenicity, i.e. Benign or Likely benign vs Uncertain significance vs Pathogenic or Likely pathogenic. • If the conflict is between 'Pathogenic' and 'Likely pathogenic' or between 'Benign' and 'Likely benign', ClinVar will report the term 'Pathogenic/Likely pathogenic'' or 'Benign/Likely benign', respectively, rather than 'conflicting interpretations of pathogenicity'.

"beta" version of a new variant-centric XML file	We have developed a new variant-centric XML file that aggregates all submitted disease/phenotype information by variant (or set of variants). The new product, called **ClinVarVariationRelease**, is currently in beta release and will move to full release early in 2018. Starting in November, the beta release will be updated bi-weekly; the release notes will note updates to the XSD and known issues. Read our blog post for more information: https://ncbiinsights.ncbi.nlm.nih.gov/2017/07/25/clinvar-variant-based-xml-summaries/ To help us improve the product, we would appreciate your feedback during the beta release. Please send questions, comments, and error reports to clinvar@ncbi.nlm.nih.gov.
ClinVar's VCF directories	We updated the directory structure for the ClinVar VCF files for the October 2017 release. There are still directories for GRCh37 and GRCh38. Within each of these directories: • the main directory includes the VCF files for the current release. • files in the new format (2.0) are archived in the archive_2.0 subdirectory. • files from the old format are archived in the archive_1.0 subdirectory. We stopped providing files in this format in Sept 2017.

Overview of submissions: 2017

Date	Total Submissions
Jan 01, 2017	396005
Feb 01, 2017	405182
Mar 01, 2017	406220
Apr 01, 2017	446265
May 01, 2017	482941
Jun 01, 2017	486420
Jul 01, 2017	488658
Aug 01, 2017	492592
Sep 01, 2017	504299
Oct 01, 2017	512373
Nov 01, 2017	517157
Dec 01, 2017	519359

Overview of submissions: 2016

Date	Total Submissions
Jan 01, 2016	172867
Feb 01, 2016	176710
Mar 01, 2016	178032
Apr 01, 2016	180549
May 01, 2016	181155

Jun 01, 2016	192617
Jul 01, 2016	204415
Aug 01, 2016	209842
Sep 01, 2016	210200
Oct 01, 2016	213499
Nov 01, 2016	236420
Dec 01, 2016	240042

Overview of submissions: 2015

Date	Total Submissions
Jan 01, 2015	149013
Feb 01, 2015	156999
Mar 01, 2015	162455
Apr 01, 2015	171408
May 01, 2015	172044
Jun 01, 2015	173236
Jul 01, 2015	184506
Aug 01, 2015	154686
Sep 01, 2015	158580
Oct 01, 2015	160538
Nov 01, 2015	170931
Dec 01, 2015	172006

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
Dec 1, 2014	148008

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