20180301_data_release_notes

Overview of changes in the release of March 1, 2018

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

Overview of submissions: 2018

Date	Total Submissions
Jan 01, 2018	579543
Feb 01, 2018	582113
Mar 01, 2018	593651

Content

Brief	Explanation
Color Genomics, Inc.	Color Genomics, Inc. submitted updates for 161 records and 10,250 novel variant interpretations.
GeneDx	GeneDx submitted 896 novel variant interpretations.
Mayo Clinic Genetic Testing Laboratories	Mayo Clinic Genetic Testing Laboratories submitted 500 variant interpretations, including novel and updated interpretations.
Clinical Molecular Genetics Laboratory, Johns Hopkins All Children's Hospital	Clinical Molecular Genetics Laboratory, Johns Hopkins All Children's Hospital submitted updates for 2 records and 333 new variant interpretations.
Department of Pathology and Laboratory Medicine, Sinai Health System	Department of Pathology and Laboratory Medicine, Sinai Health System provided updates for 182 variant interpretations.
Update to variant_summary.txt	The ClinSigSimple column in the variant_summary.txt file was updated to: ■ correct some rows that reported 1 but with no submissions that were pathogenic or likely pathogenic ■ report -1 for "included variants"; these are variants that are only in ClinVar when included in a haplotype or genotype that was interpreted, and there is no direct interpretation for the variant itself.
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 to submission_summary.txt	A column will be added at the end to represent the ExplanationOfInterpretation submitted to add value to interpretations of 'drug response' or 'other'.

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; however, further development is on hold. If your work depends on moving this new file into production, please let us know by emailing clinvar@ncbi.nlm.nih.gov. The beta release is updated bi-weekly; the release notes 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/
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