20171005_data_release_notes

Overview of changes in the release of October 5, 2017

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

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

Content

Brief	Explanation
ClinGen Inherited Cardiomyopathy Expert Panel	The ClinGen Inherited Cardiomyopathy Expert Panel submitted 102 updated interpretations.
ARUP Laboratories	ARUP Laboratories submitted 3309 interpreted variants, including both novel and updated interpretations.
Quest Diagnostics Nichols Institute San Juan Capistrano	Quest Diagnostics Nichols Institute San Juan Capistrano submitted 2 478 interpreted variants, including both novel and updated interpretations.
Laboratory for Molecular Medicine, Partners HealthCare Personalized Medicine	The LMM submitted 129 interpreted variants, including both updated and novel interpretations.
CSER-BASIC3	The CSER-BASIC3 project submitted 79 novel variant interpretations.

Changes to ClinVarFullRelease	• ClinVar now distinguishes between mode of inheritance (MOI) and penetrance values that were submitted versus values that come from authoritative sources such as OMIM and Orphanet.
	  • Submitted MOI and penetrance are reported in ClinVarFullRelease as ClinVarAssertion/AttributeSet/Attribute.
	  • MOI and penetrance from authoritative sources are reported in MedGen
	  • This change was effective with the weekly release of September 11, 2017
	• The MalaCard link name was removed. This value was for internal processing only, and links to MalaCards can be found in MedGen.
	• Some early submissions to ClinVar submitted terms for clinical significance other than the terms used by ClinVar. These values are converted to the standard values used in ClinVar. Some but not all records had a comment explaining that the value of clinical significance had been converted to standard values in submission processing. With this release, all records with a converted value of clinical significance now include the comment.

"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 later in 2017. 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 have updated the directory structure for the ClinVar VCF files. 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 subd irectory. • files from the old format are archived in the archive_1.0 subdire ctory. We stopped providing files in this format in Sept 2017.
Coming soon - ClinVar will adopt the new HGVS standard for variants that are intronic or outside the UTRs	HGVS standard states that "the reference sequence used must contain the residue(s) described to be changed." Therefore "a coding DNA reference sequence does not contain intron or 5' and 3' gene flanking sequences and can therefore **not be used as a reference** t o describe variants in introns and up/down-stream of the gene." ClinVar is working to adopt this standard so we encourage our submitters and users to start describing these variants on genomic sequence instead. http://varnomen.hgvs.org/recommendations/general/ http://varnomen.hgvs.org/bg-material/numbering/

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