20180607_data_release_notes

Overview of changes in the release of June 7, 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
Apr 01, 2018	610005
May 01, 2018	645149
Jun 01, 2018	676018

Content

Brief	Explanation
Invitae	Invitae submitted 57584 records, including both novel and updated records.
GeneDx	GeneDx submitted 48813 records, including both novel and updated records.
Centre for Mendelian Genomics, University Medical Centre Ljubljana	Centre for Mendelian Genomics, University Medical Centre Ljubljana submitted 534 novel interpretations and 2 updated records.
Molecular Diagnostic Laboratory for Inherited Cardiovascular Disease, Montreal Heart Institute	Molecular Diagnostic Laboratory for Inherited Cardiovascular Disease, Montreal Heart Institute submitted 461 novel interpretations.
Genomic Research Center, Shahid Beheshti University of Medical Sciences	Genomic Research Center, Shahid Beheshti University of Medical Sciences submitted 281 novel interpretations.
Department of Psychiatry, Nagoya University	Department of Psychiatry, Nagoya University submitted 219 novel interpretations.
Iberoamerican FH Network	Iberoamerican FH Network submitted 144 novel interpretations.
Undiagnosed Diseases Network, NIH	Undiagnosed Diseases Network, NIH submitted 124 novel interpretations and 5 updated records.
Bioinformatics Core, Luxembourg Center for Systems Biomedicine	Bioinformatics Core, Luxembourg Center for Systems Biomedicine submitted 111 novel interpretations.
Laboratory of Molecular Genetics (Pr. Bezieau's lab), CHU de Nantes	Laboratory of Molecular Genetics (Pr. Bezieau's lab), CHU de Nantes submitted 100 novel interpretations.

change in calculation of aggregate clinical significance for variation records	We have changed the calculation of aggregate clinical significance for variation records so that interpretations from submissions with review status "criteria provided, single submitter" supersede interpretations from submissions with lower review statuses: https://www.ncbi.nlm.nih.gov/clinvar/docs/review_status/#revstat_def Coming soon: We also plan to change the aggregate value for "date last evaluated" so that it is calculated only from submissions that contribute to the aggregate clinical significance.
change in calculation of the aggregate clinical significance for RCV records	 We have changed the calculation for conflicts in interpretation for RC V records to be the same as for Variation records, namely: Conflicts are 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 reports the term 'Pathogenic/Likely pathogenic'' or 'Benign/Likely benign ', respectiv ely, rather than 'conflicting interpretations of pathogenicity'. Formerly, conflicts in interpretation were calculated differently for ClinVar variation (Variation ID) and variation-disease (RCV) records. RCV records had conflicts for any difference in interpretation, including between Benign and Likely Benign, and between Pathogenic and Likely pathogenic. The first archived release to include this change is the June 2018 release.
no longer converting "not provided" as a condition as "not specified"	Formerly, when the interpreted condition was submitted as "not provided" for variants classified as Benign, Likely benign, or Uncertain significance, ClinVar converted "not provided" to "not specified". We are no longer converting "not provided"; we keep the term that was submitted as the condition. Submitters who wish to indicate that they interpreted a variant as Benign, Likely benign, or Uncertain significance for no specific disease should explicitly report "not specified" as the interpreted condition.
"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/

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