20180503_data_release_notes

Overview of changes in the release of May 3, 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

Content

Brief	Explanation
International Society for Gastrointestinal Hereditary Tumours (InSiGHT)	The expert panel International Society for Gastrointestinal Hereditary Tumours (InSiGHT) submitted 7 novel records and updates to 16 records.
GeneDx	GeneDx submitted 49241 records, including novel and updated records.
Laboratory for Molecular Medicine, Partners HealthCare Personalized Medicine	The LMM submitted 3075 novel records and updates to 16068 records.
Ambry Genetics	Ambry Genetics submitted 7457 novel records and and updates to 3 393 records.
Diagnostic Laboratory, Department of Genetics, University Medical Center Groningen	Diagnostic Laboratory, Department of Genetics, University Medical Center Groningen submitted 1897 novel records.
DNA and Cytogenetics Diagnostics Unit, Erasmus Medical Center	DNA and Cytogenetics Diagnostics Unit, Erasmus Medical Center submitted 1473 novel records.
Genome Diagnostics Laboratory, University Medical Center Utrecht	Genome Diagnostics Laboratory, University Medical Center Utrecht submitted 1006 novel records.
Genomic Research Center, Shahid Beheshti University of Medical Sciences	Genomic Research Center, Shahid Beheshti University of Medical Sciences submitted 559 novel records.
Genome Diagnostics Laboratory, VU University Medical Center Amsterdam	Genome Diagnostics Laboratory, VU University Medical Center Amsterdam submitted 433 novel records.
Molecular Diagnostic Laboratory for Inherited Cardiovascular Disease, Montreal Heart Institute	Molecular Diagnostic Laboratory for Inherited Cardiovascular Disease, Montreal Heart Institute submitted 350 novel records.
Bioscientia Institut fuer Medizinische Diagnostik GmbH, Sonic Healthcare	Bioscientia Institut fuer Medizinische Diagnostik GmbH, Sonic Healthcare submitted 175 novel records.
Undiagnosed Diseases Network, NIH	Undiagnosed Diseases Network, NIH submitted 5 updated records and 127 novel records.

Variation ID added to variant_summary.txt	We added a column to the end of the variant_summary.txt file to report the single VariationID that corresponds to the AlleleID in the first column. This column does NOT contain all the AlleleID /VariationID relationships; they continue to be provided by variation_allele.txt.gz. The new column does, however, make it easier to map from variant_summary.txt to our new variant-centric XML.
Explanation of interpretation added to submission_summary.txt	We added a column to the end of the submission_summary.txt file to represent ExplanationOfInterpretation. This is a value submitted when the clinical significance is 'drug response' or 'other', to provide more information about what type of drug response or what the "other" value of clinical significance is.
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'. This change is not found in the May 2018 release, but is expected to be included in the next weekly release on May 10. The first archived release to include this change will be the June 2018 release.
"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