20200305_data_release_notes

Overview of submissions: 2020

Jan 01, 2020	1,026,969
Feb 01, 2020	1,041,077
Mar 01.2020	1,055,499

Overview of changes in the release of March 5, 2020

Content

Brief	Explanation
American College of Medical Genetics and Genomics (ACMG)	ACMG submitted four novel interpretations from its practice guidelines for various lengths of the triplet repeat in the HTT gene.
Evidence-based Network for the Interpretation of Germline Mutant Alleles (ENIGMA)	The expert panel ENIGMA submitted 189 novel variant interpretations and an update to one record.
ClinGen Hearing Loss Variant Curation Expert Panel	The ClinGen Hearing Loss Variant Curation Expert Panel submitted 23 novel variant interpretations and updates to 14 records.
ARUP Laboratories, Molecular Genetics and Genomics, ARUP Laboratories	ARUP Laboratories, Molecular Genetics and Genomics, ARUP Laboratories provided 4,108 novel variant interpretations and updates for 3,449 records.
Baylor Genetics	Baylor Genetics submitted 984 novel variant interpretations.
Institute of Human Genetics, Klinikum rechts der Isar	Institute of Human Genetics, Klinikum rechts der Isar submitted 662 novel variant interpretations and updates for 17 records.
Sharon lab, Hadassah-Hebrew University Medical Center	Sharon lab, Hadassah-Hebrew University Medical Center submitted 565 variant interpretations from research.
Broad Institute Rare Disease Group, Broad Institute	Broad Institute Rare Disease Group, Broad Institute submitted 257 novel variant interpretations.
Génétique des Maladies du Développement, Hospices Civils de Lyon	Génétique des Maladies du Développement, Hospices Civils de Lyon submitted 142 novel variant interpretations.
use of MANE transcripts in preferred names for variants in ClinVar	We calculate a preferred name for each variant in ClinVar, often using a RefSeq transcript as the reference sequence. We are in the process of preferentially using transcripts from the MANE project in the preferred name. For more information about MANE transcripts, read this NCBI blog post: https://ncbiinsights.ncbi.nlm.nih.gov/2019/03/12/mane-select-v0-5/ and RefSeq's documentation: https://www.ncbi.nlm.nih.gov/refseq/MANE/

coming soon: changes to variant position, reference and alternate alleles in variant_summary.txt	Soon we will update the way that the variant position and reference and alternate alleles are reported in variant_summary.txt. We will add columns for vcf_pos, vcf_ref, and vcf_alt. Variants that are in scope for our VCF file will have their position and ref /alt alleles reported in this way. Variants that are not in scope for our VCF file will have their position reported in the old fields: Start, Stop, ReferenceAllele, AlternateAllele. A variant is expected to reported one way or the other, but not both. This change will make variant_summary.txt more consistent with ClinVar's VCV XML file. Please contact us as clinvar@ncbi.nlm.nih.gov if you have questions or comments about this change.
coming soon: change in how p. expressions are stored in XML	Soon we will change the way we handle p. expressions that are incomplete because they lack an accession.version, e.g. p. Tyr1863Cys. We will store these descriptions in the XML files as alternate names, rather than as HGVS expressions.

Overview of submissions: 2019

Jan 01, 2019	759562
Feb 07, 2019	778673
Mar 01, 2019	782638
Apr 01, 2019	787656
May 01, 2019	795045
Jun 01, 2019	811551
Jul 01, 2019	819827
Aug 01, 2019	825177
Sept 01, 2019	881419
Oct 01, 2019	888298
Nov 01, 2019	889968
Dec 01, 2019	893196

Overview of submissions: 2018

Jan 01, 2018	579543
Feb 01, 2018	582113
Mar 01, 2018	593651
Apr 01, 2018	610005
May 01, 2018	645149
Jun 01, 2018	676018
Jul 01, 2018	676575
Aug 01, 2018	685942
Sep 01, 2018	701880
Oct 01, 2018	708726
Nov 01, 2018	715516
Dec 01, 2018	749203

Overview of submissions: 2017

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

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

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

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

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