20190404_data_release_notes

Overview of changes in the release of April 4, 2019

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

Overview of submissions: 2019

Date	Total Submissions
Jan 01, 2019	759562
Feb 07, 2019	778673
Mar 01, 2019	782638
Apr 01, 2019	787656

Content

Brief	Explanation
ClinGen RASopathy Variant Curation Expert Panel	ClinGen RASopathy Variant Curation Expert Panel submitted 226 novel and updated, expert-reviewed interpretations for variants in nine genes.
ClinGen PAH Variant Curation Expert Panel	ClinGen PAH Variant Curation Expert Panel submitted 158 novel and updated, expert-reviewed interpretations for variants in the PAH gene.
ClinGen Inherited Cardiomyopathy Variant Curation Expert Panel	ClinGen Inherited Cardiomyopathy Variant Curation Expert Panel submitted 102 updated, expert-reviewed interpretations for variants in the MYH7 gene.
ClinGen Hearing Loss Variant Curation Expert Panel	ClinGen Hearing Loss Variant Curation Expert Panel submitted 52 novel and updated, expert-reviewed interpretations for variants in nine genes.
ClinGen PTEN Variant Curation Expert Panel	ClinGen PTEN Variant Curation Expert Panel submitted 19 novel, expert-reviewed interpretations for variants in the PTEN gene.
Praxis fuer Humangenetik Tuebingen	Praxis fuer Humangenetik Tuebingen submitted 1337 novel and 4194 updated interpretations for variants.
Quest Diagnostics Nichols Institute San Juan Capistrano	Quest Diagnostics Nichols Institute San Juan Capistrano submitted 2621 records, including both novel and updated records.
Wong Mito Lab, Molecular and Human Genetics, Baylor College of Medicine	Wong Mito Lab, Molecular and Human Genetics, Baylor College of Medicine submitted 408 novel variant interpretations.
Department of Genetics, Sultan Qaboos University Hospital, Oman	Department of Genetics, Sultan Qaboos University Hospital, Oman submitted 233 novel variant interpretations.
Clinical Genomics Lab, St. Jude Children's Research Hospital	Clinical Genomics Lab, St. Jude Children's Research Hospital submitted 199 novel variant interpretations.
Laboratoire de Cytogenetique, Hospices Civils de Lyon	Laboratoire de Cytogenetique, Hospices Civils de Lyon submitted 135 novel variant interpretations.
NEW: production version of the 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 available in production mode with this release. Read our blog post for more information: https://ncbiinsights.ncbi.nlm.nih.gov/2017/07/25/clinvar-variant-based-xml-summaries/
coming soon: changes to the web display, web search, and E-utilities	The newly designed ClinVar pages will end their "alpha" release soon and move to production on www. This change will be accompanied by some changes to web search and E-utilities. See the web release notes for more information.

coming soon: use of MANE transcripts for 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. Soon we will preferentially use transcripts from the MANE project in the preferred name. Read this NCBI blog post for more information about MANE transcripts: https://ncbiinsights.ncbi.nlm.nih.gov/2019/03/12/mane-select-v0-5/
coming soon - genomic location for variants with defined endpoints will be reported only as "VCF-style"	Genomic location for variants with defined endpoints currently reported in the ClinVar XML in two ways: • start and stop, which are right-shifted similar to the HGVS standard • vcf_start and vcf_stop, which are left-shifted similar to VCF format We plan to simplify reporting of genomic location and only report the location as the left-shifted position, for consistency with VCF format. This change is not yet scheduled. If you have any concerns, please write to us as clinvar@ncbi.nlm.nih.gov.

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
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

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