

20200507_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
Apr 02, 2020	1,084,731
May 01, 2020	1,136,163

Overview of changes in the release of May 7, 2020

Content

Brief	Explanation
ClinGen RASopathy Variant Curation Expert Panel	ClinGen RASopathy Variant Curation Expert Panel submitted 28 novel variant interpretations and updates to 3 records.
ClinGen CDH1 Variant Curation Expert Panel	ClinGen CDH1 Variant Curation Expert Panel submitted 24 novel variant interpretations and an update to 1 record.
ClinGen PTEN Variant Curation Expert Panel	ClinGen PTEN Variant Curation Expert Panel submitted 20 novel variant interpretations.
ClinGen Myeloid Malignancy Variant Curation Expert Panel	ClinGen Myeloid Malignancy Variant Curation Expert Panel submitted 19 novel variant interpretations.
ClinGen Hearing Loss Variant Curation Expert Panel	ClinGen Hearing Loss Variant Curation Expert Panel submitted 15 novel variant interpretations.
ClinGen CDH1 Variant Curation Expert Panel	ClinGen CDH1 Variant Curation Expert Panel submitted 1 novel variant interpretation.
Invitae	Invitae submitted 42,985 novel variant interpretations and updates to 134,565 records.
Blueprint Genetics	Blueprint Genetics submitted 2,734 novel variant interpretations.
Brotman Baty Institute, University of Washington	Brotman Baty Institute, University of Washington submitted functional evidence for 2,639 variants in BRCA1.
Leiden Open Variation Database	Leiden Open Variation Database submitted 547 variants from the PALB2 LOVD database, curated by Marc Tischkowitz and Arleen D. Auerbach.
GeneKor MSA	GeneKor MSA submitted 593 novel variant interpretations.
Myriad Women's Health, Inc.	Myriad Women's Health, Inc. submitted 476 variant interpretations from carrier testing.
Cancer Genomics Group, Japanese Foundation For Cancer Research	Cancer Genomics Group, Japanese Foundation For Cancer Research submitted 298 novel variant interpretations.
The ITHANET community portal, The Cyprus Institute of Neurology and Genetics	The ITHANET community portal, The Cyprus Institute of Neurology and Genetics submitted 288 novel variant interpretations.

Victorian Clinical Genetics Services, Murdoch Childrens Research Institute	Victorian Clinical Genetics Services, Murdoch Childrens Research Institute submitted 149 novel variant interpretations.
coming soon: changes to variant position, reference and alternate alleles in variant_summary.txt	<p>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 be reported one way or the other, but not both.</p> <p>This change will make variant_summary.txt more consistent with ClinVar's VCV XML file.</p> <p>Please contact us as clinvar@ncbi.nlm.nih.gov if you have questions or comments about this change.</p>
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
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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