20170504_data_release_notes

Overview of changes in the release of May 4, 2017

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

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

Content

Brief	Explanation
ClinGen Inherited Cardiomyopathy Expert Panel	The ClinGen Inherited Cardiomyopathy Expert Panel is the first ClinGen expert panel to submit data to ClinVar. They provided 101 expert-reviewed interpretations of MYH7 variants.
Invitae	Invitae submitted 32,845 variant interpretations, including both novel and updated interpretations.
GeneDx	GeneDx submitted 9924 novel variant interpretations.
Laboratory for Molecular Medicine, Partners HealthCare Personalized Medicine	Laboratory for Molecular Medicine, Partners HealthCare Personalized Medicine submitted 2501 novel variant interpretations.
CSER-PediSeq	The PediSeq project, part of the CSER Consortium, provided 241 novel variant interpretations.
Ambry Genetics	Ambry provided 168 variant interpretations, including both novel and updated interpretations.
Baylor Genetics	Baylor Genetics provided 146 novel interpretations for variants in BRCA1 and BRCA2.
CSER-NextGen	The NextGen project, part of the CSER Consortium, provided 116 novel variant interpretations.
Department of Medical Genetics,Oslo University Hospital	Oslo University Hospital provided 104 novel interpretations for variants in BRCA1.

new GenotypeSet element in ClinVar XML	The May release uses a new element, GenotypeSet, to indicate that an interpretation was made about two alleles that are on different chromosomes.This case was formerly reported as:
	
	`<MeasureSet Type="CompoundHeterozygote" ID="424841">`
	
	and is now reported as:
	
	`<GenotypeSet Type="CompoundHeterozygote" ID="424841">`
	`<MeasureSet Type="Variant" ID="375249">`
	`<Measure Type="single nucleotide variant" ID="361947">...</Measure>`
	`</MeasureSet>`
	`<MeasureSet Type="Variant" ID="375250">`
	`<Measure Type="single nucleotide variant" ID="361945">...</Measure>`
	`</MeasureSet>`
	`</GenotypeSet>`
	
	Currently all GenotypeSets are of Type "CompoundHeterozygote", indicating a simple allele on each chromosome. In the future we will also include GenotypeSets of Type "Diplotype", indicating a haplotype on one or both chromosomes.
	
	Note that currently the set of variants in each MeasureSet, 375249 and 375250 in the example above, are not searchable on the ClinVar website unless there is a submission with an interpretation for that specific set of variants.

"beta" version of new ClinVar VCF file	We have developed a new version of the ClinVar VCF file: ftp://ftp.ncbi.nlm.nih.gov/pub/clinvar/vcf_GRCh37/vcf_2.0/ ftp://ftp.ncbi.nlm.nih.gov/pub/clinvar/vcf_GRCh38/vcf_2.0/ Details explaining the differences versus the current VCF are in the README. Note that we will continue to provide the current version of the VCF file for a few months. We will announce when the current version will no longer be provided.
Retirement of Variation Reporter	NCBI will retire Variation Reporter at some point in the future: https://www.ncbi.nlm.nih.gov/variation/tools/reporter We have developed a set of stand-alone variation services that replace some of the functions of Variation Reporter, but not all. You can try the new services here: https://api.ncbi.nlm.nih.gov/variation/v0/ If you use Variation Reporter and the services above do not meet your needs, please let us know by emailing tkt-varhd@ncbi.nlm.nih.gov.
Coming soon - ClinVar will adopt the new HGVS standard for variants that are intronic or outside the UTRs	HGVS standard states that "the reference sequence used must contain the residue(s) described to be changed." Therefore "a coding DNA reference sequence does not contain intron or 5' and 3' gene flanking sequences and can therefore **not be used as a reference** to describe variants in introns and up/down-stream of the gene." ClinVar is working to adopt this standard so we encourage our submitters and users to start describing these variants on genomic sequence instead. http://varnomen.hgvs.org/recommendations/general/ http://varnomen.hgvs.org/bg-material/numbering/

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