20170706_data_release_notes

Overview of changes in the release of July 6, 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
Jun 01, 2017	486420
Jul 01, 2017	488658

Content

Brief	Explanation
InSiGHT	The expert panel InSiGHT submitted 50 variant interpretations, including both novel and updates.
Ambry Genetics	Ambry submitted more than 2000 novel variant interpretations and updated interpretations for more than 1200 variants.
GeneDx	GeneDx submitted 1713 novel variant interpretations.
Department of Medical Sciences, Uppsala University	The Department of Medical Sciences at Uppsala University submitted 102 novel variant interpretations for osteogenesis imperfecta.
Invitae	Invitae submitted 86 variant interpretations, including both novel and updates.
variant_summary.txt.gz	The variant_summary.txt file in the directory ftp://ftp.ncbi.nlm.nih.gov/pub/clinvar/tab_delimited/ has been modified to restore the reporting of all conditions, in the order of the reporting of the RCV accession. Before this fix, conditions without a 'CUI' in MedGen were skipped.
variation_allele.txt.gz	The variation_allele.txt file in the directory ftp://ftp.ncbi.nlm.nih.gov/pub/clinvar/tab_delimited/ has been corrected to report relationships for compound heterozygotes correctly. The release of June, 2017, had not updated the content to reflect new VariationIDs assigned to what is now being reported in the release XML (ClinVarFullRelease_00-latest.xml.gz) as a GenotypeSet.

change to reporting of method type	The method that was used to collect the data for the interpretation is reported in the XML as MethodType. A MethodResult may also be included in the XML. Reporting of Method Result has changed from:
	
	`<Method>`
	`...`
	`<MethodType>clinical testing</MethodType>`
	`<MethodResult>Pass</MethodResult>`
	`...`
	`<\Method>`
	
	to:
	
	`<Method>`
	`...`
	`<MethodType>clinical testing</MethodType>`
	`<ObsMethodAttribute>`
	`<Attribute Type="MethodResult">Pass</Attribute>`
	`</ObsMethodAttribute>`
	`...`
	`</Method>`

submission and reporting of the testing laboratory's intepretation	ClinVar is expanding to allow submissions from clinical providers. These submissions may include the interpretation provided by the laboratory that performed the clinical genetic testing and the date that the variant was reported to the submitter. This interpretation is archived in the submitted section of the XML but it is not included in the calculation of aggregate clinical significance. The interpretation from the testing laboratory is represented in the XML as:
	
	`<ClinVarAssertion>`
	`...`
	`<ObservedIn>`
	`<Method>`
	`<ObsMethodAttribute>`
	`<Attribute Type=TestingLaboratory dateValue="2017-06-23">Name of testing laboratory</Attribute>`
	`<Comment>Pathogenic</Comment>`
	`</ObsMethodAttribute>`
	`</Method>`
	`</ObservedIn>`
	`...`
	`</ClinVarAssertion>`
	
	or, if the organization ID is provided instead of the name of the testing laboratory:
	
	`<ClinVarAssertion>`
	`...`
	`<ObservedIn>`
	`<Method>`
	`<ObsMethodAttribute>`
	`<Attribute Type=TestingLaboratory dateValue="2017-06-23" integerValue="123456"/>`
	`<Comment>Pathogenic</Comment>`
	`</ObsMethodAttribute>`
	`</Method>`
	`</ObservedIn>`
	`...`
	`</ClinVarAssertion>`

"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. Please send any feedback to clinvar@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