

20170302_data_release_notes

Overview of changes in the release of Mar 2, 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

Content

Brief	Explanation
PharmGKB submission of expert-reviewed variants	Expert panel PharmGKB provided updates to 275 variant interpretations and submitted 50 novel interpretations.
Counsyl submission	Counsyl provided 5013 novel variant interpretations.
CIMBA submission	The Consortium of Investigators of Modifiers of BRCA1/2 (CIMBA) provided their first submission to ClinVar, comprising 3330 novel variant interpretations.
CHOP submission	The Children's Hospital of Philadelphia provided 56 novel variant interpretations.
new MeasureRelationship/@Types	New types for MeasureRelationship were introduced to the ClinVar XML in the December release: <ul style="list-style-type: none">• within single gene - the variant is contained within a single gene• within multiple genes by overlap - the variant is within multiple genes because the genes overlap on the genome; the genes may overlap on the same or on opposite strands• genes overlapped by variant - one or more genes overlap the variant, e.g. large structural variants• near gene, upstream - the variant is within 2 kb upstream of the gene• near gene, downstream - the variant is within 5 kb downstream of the gene• asserted, but not computed - the variant-gene relationship was asserted in a submission to ClinVar, but we cannot compute the relationship based on the location of the variant and the genomic annotation of the gene
Updated VCF file posted April 13, 2017	An updated VCF file was posted to the FTP site on April 13, 2017: ftp://ftp.ncbi.nlm.nih.gov/pub/clinvar/vcf_GRCh37/archive/2017/clinvar_20170228_updated.vcf.gz 58 rs numbers were corrected. They represent variants that may be expressed with left- or right-shifted positions. For each variant the rs numbers for the two representations had been merged but the location had not been updated to use the left-most position. These locations have been corrected in the updated file.
"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.

Possible retirement of Variation Reporter	<p>NCBI is considering whether to retire Variation Reporter: https://www.ncbi.nlm.nih.gov/variation/tools/reporter</p> <p>We have developed a set of stand-alone variation services that would 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/</p> <p>To help us in guiding the decision, if you are a user of Variation Reporter, let us know for what specific purpose you access it by emailing tk-varhd@ncbi.nlm.nih.gov.</p>
Coming soon - ClinVar will adopt the new HGVS standard for variants that are intronic or outside the UTRs	<p>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.</p> <p>http://varnomen.hgvs.org/recommendations/general/</p> <p>http://varnomen.hgvs.org/bg-material/numbering/</p>

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