20170406_data_release_notes

Overview of changes in the release of Apr 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

Content

Brief	Explanation
Color Genomics	Color Genomics submitted 374 novel interpretations for variants in several genes associated with breast cancer.
Ambry Genetics	Ambry Genetics submitted 169 interpretations for variants in PALB2, including both novel and updates.
Claritas Genomics	Claritas Genomics submitted 30 novel interpretations.
Merging of duplicate accessions	ClinVar continues to work with submitters to remove duplicate submissions when the duplicates are identified after accessions are assigned. We appreciate the cooperation of the submitters in this process. ClinVar is improving its data validation steps so potential duplicates are caught earlier in the processing of submissions. This month we merged accessions for the Breast Cancer Information Core (BIC), Emory, and Invitae.
rs numbers missing from the ClinVar VCF files	A few rs numbers are missing from the ClinVar VCF files, including rs113993960 for deltaF508. We will update the files to add back the rs numbers shortly.
"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	NCBI is considering whether to retire Variation Reporter: https://www.ncbi.nlm.nih.gov/variation/tools/reporter 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/ 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 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