20150507_data_release_notes

Overview of changes in the release of May 7, 2015

Overview of submissions: 2015

Date	Total Submissions
Jan 01, 2015	149013
Feb 01, 2015	156999
March 1, 2015	162455
Apr 01, 2015	171408
May 1, 2015	172044

Content

Brief	Explanation
RettBASE submission	RettBASE submitted 973 variants in genes related to Rett Syndrome. Some of these were updates to the submission RettBASE had made to dbSNP several years ago.
Counsyl submission	Counsyl submitted 1024 variants.
Children's Mercy Hospitals and Clinics submission	Children's Mercy Hospitals and Clinics submitted 34 variants in ZEB2 related to Mowat-Wilson syndrome.
Children's Hospital of Eastern Ontario submission	Children's Hospital of Eastern Ontario submitted 957 variants.
University Children's Hospital Zurich submission	University Children's Hospital Zurich submitted 42 variants related to MTHFR deficiency.
GeneDx submission	GeneDX submitted 186 variants, both novel and updates.
MedSeq submission	As part of the CSER Consortium (https://cser-consortium.org/), the MedSeq Project submitted 52 variants.
Invitae submission	Invitae submitted more than 1250 variants, both novel and updates, with about half being novel and the rest updates.
University of Washington Cytogenetics and Genomics Laboratory submission	The University of Washington Cytogenetics and Genomics Laboratory submitted seven structural variants.

rs#	Because of the release processing for build 144 of dbSNP, not all the variants in scope for dbSNP were assigned rs# in this release. Thus these data will also be missing from ClinVar's VCF file.
ftp site: variant_summary	Several columns were added to ftp://ftp.ncbi.nlm.nih.gov/pub/clinvar/tab_delimited/variant_summary.txt.gz, namely ReferenceAllele, AlternateAllele, and SubmitterCategories. For complete documentation, please refer to our README file (ftp://ftp.ncbi.nlm.nih.gov/pub/clinvar/README.txt)
ftp site: conflict report	Two columns were added to ftp://ftp.ncbi.nlm.nih.gov/pub/clinvar/tab_delimited/summary_of_conflicting_data.txt, namely Conflict_Reported and Variant_Type. For complete documentation, please refer to our README file (ftp://ftp.ncbi.nlm.nih.gov/pub/clinvar/README.txt)
Coming soon - changes to review status	ClinVar's definitions of review statuses will change in the upcoming months. Submitters will be asked to provide their assertion criteria used to classify variants. Submissions without assertion criteria will have the review status "single submitter - no criteria provided" and will not contribute to the calculation of gold stars on aggregate records. Submissions with assertion criteria will have the review status "single submitter - criteria provided" and will contribute to the number of stars. Documentation is available here: http://www.ncbi.nlm.nih.gov/clinvar/docs/assertion_criteria/
Clinical significance	ClinVar now only reports conflicts in clinical significance within the terms for pathogenicity. In other words, Uncertain significance and Pathogenic result in a conflict; risk factor and Pathogenic do not. Clinical significance for the variant now uses a 7-tier system - the five tiers of pathogenicity recommended by ACMG, along with Pathogenic/likely pathogenic, and Benign/likely benign if those are the only values. In other words, if all submitted interpretations for a variant are Pathogenic, the variant is reported as Pathogenic, not Pathogenic/likely pathogenic as had been done previously. Coming soon - ClinVar will change reporting of clinical significance to provide a clinical significance for the variant in the germline context, and a separate value for clinical significance in the somatic context.

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