20150402_data_release_notes

Overview of changes in the release of April 2, 2015

Overview of submissions: 2015

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

Content

Brief	Explanation
VCF and CLINSIG	Modified the reporting of clinical significance in the VCF files when the variant was of somatic origin. Previously those were represented as type 'other' (255); now they are reported according to their specific value.
Allowed values for clinical significance	The clinical significance of 137 records was modified from either Pathogenic/Likely pathogenic to Likely pathogenic and Benign/Likely benign to Likely benign. The option of submitting Pathogenic/Likely pathogenic or Benign/Likely benign as Clinical significance was disabled.
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 critiera 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 (still being updated) will be 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, Pathogenic/likely pathogenic, and Benign/likely benign. In other words, if all submitted interpretations for a variant are Pathogenic, the variant is reported as Pathogenic, not Pathogenic/likely pathogenic. 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 the clincial significance in the somatic context.
Ambry submission	Ambry Genetics submitted an update of cancer-predisposing variants bringing their total to over 9800 variants. This submission included 5800 novel Ambry variants.
GeneDx submission	GeneDx submitted 1664 cancer-related variants.
University of Chicago	University of Chicago Genetic Services Laboratory submitted 3442 interpretations.
Exon deletions and seq_loc	For exon deletions that are not clearly defined at the genomic level,, ClinVar no longer calculates the location on the genome, due to the uncertainty of the actual genomic change.
New submission spreadsheet templates	Updated submission templates are available on the ftp site: ftp://ftp.ncbi.nlm.nih.gov/pub/clinvar/submission_templates/ including a new version of the full template, version 3.1, and of the lite template, L1.4. ClinVar will continue to accept submissions on the older spreadsheets until May 31, 2015.

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