20131107_data_release_notes

Summary of changes in this data release

Number of submissions

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

Content

Brief	Explanation
variant_summary.txt	The file ftp://ftp.ncbi.nlm.nih.gov/pub/clinvar/tab_delimited/variant_summary.txt.gz was modified to pad null numeric fields with -1, and null character fields with a dash (hyphen). This extraction is our tab delimited report of a subset of variation data.
representation of LRG	Representation of variation in genomic and c. coordinates based on LRG sequences has been added to the detailed display. The LRG expressions are searchable as well (e.g. http://www.ncbi.nlm.nih.gov/clinvar/?term=LRG_292t1%3Ac.68_69delAG).
CFTR2	132 clinical assertions have been submitted to ClinVar from the CFTR2 database. These records represent single variations with a clinical assertion of "pathogenic".
integration from dbSNP	Data from several submitters that originally reported their medically related variation to dbSNP have been incorporated into ClinVar. These include UniProtKB/Swiss-Prot, the Samuels laboratory, Cardiovascular Biomedical Research Unit, and Epithelial Biology.

Expected modifications in the next month

Brief	Explanation
clinicial significance	Terms to report clinical significance will be modified to conform to the new ACMG guidelines.

methods	The terms used to categorize methods of data capture are being reviewed by ClinGen advisors. Terms like 'curation' are likely to be replaced. Definitions of the new terms will be provided in the data dictionary and the supporting documents on the web.
SCRP	The SCRP data set will be updated in the next release. The number of variants is expected to about double. http://www.sharingclinicalreports.org/