20140807_data_release_notes

Overview of changes in the release of August 7, 2014

Overview of submissions: 2014

Date	Total Submissions
Jan 01, 2014	68204
Feb 01, 2014	73492
Mar 01, 2014	83343
April 01, 2014	111501
May 01, 2014	112349
June 01, 2014	117209
Jul 01, 2014	127132
August 01, 2014	127557

Content

Brief	Explanation
Preferred name	The method of calculating the preferred name of the variant was changed based on the type of variation. For those reported as copy number gain or copy number loss, the name is now constructed byt combining values for assembly, cytogenetic band, chromosome location and copy number, e.g. GRCh37/hg19 16p11.2(chr16:154974667-155226096)x3. When both outer and inner boundaries of the variant are reported, the inner boundary is chosen for the display. For other variants, the use of HGVS expressions was modified to include the accession.version, and the gene symbol in parentheses, *e.g*. NM_004006.1(DMD):c.3G>T.
SCRP notes	Case-level notes associated with the Sharing Clinical Reports Project have been removed from the aggregate data display at the request of the submitter.
HGVS	The method of reporting no sequence change was modified. Rather than repeating the sequence that is not modified followed by '=', only '=' is appended: For example: **now**: NP_000760.1:p.Pro227= **previously**: NP_000760.1:p.Pro227Pro=
Submissions	Major submissions include an update from Ambry Genetics that added more than 4,100 additional variants. These are not included in the statistics for August 1.

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