# 2014-05-01\_Web\_Release

# Release notes, May 1, 2014

You will find that ClinVar made several changes to the Web site recently.filter\_display.PNGsend\_to.PNG

# Functionality

You can now download your query results to a speadsheet. Let's say you wanted to download a brief report of data in ClinVar related to Lynch syndrome and that had been reviewed by a panel of experts. These are the steps you could follow:

- go to ClinVar: http://www.ncbi.nlm.nih.gov/clinvar/
- Enter lynch as your query term: http://www.ncbi.nlm.nih.gov/clinvar?term=lynch
- Select Expert panel from the choice of filters under Review status at the left:
- Open the Send to dialog

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Clinical significance		Gene	Variation	Freq	Pheno	ntyne	Clinical Significance	Review Status	Chr	Location (GRCh37.p10)
Probably not pathogenic (86) Variant of unknown	□ <u>See detail</u> 1.	s MSH2	NM_000251.1:c433	IT>G	Lynci syndr		Benign	reviewed by expert panel	2	47629898
significance (766) Pathogenic (1156)	□ <u>See detail</u> 2.	s MSH2	NM_000251.1:c118	IT>C	Lynci syndr		Benign	reviewed by expert panel	2	47630213
Review status ✓ Expert panel (2362)	ar <mark>□ <u>See detail</u> 3.</mark>	<u>s</u> MSH6	MSH6:c.3163G>C (p.Ala1055Pro)		Lynci syndr		Uncertain significance	reviewed by expert panel	2	48028285

• In that menu, select File and Tabular (text)

	<u>Send to:</u> ⊘					
Choose Destination						
© File ⊂ Collections	C Clipboard					
Download 2362 Format	items.					
Tabular (text)	•					
Sort by Default order	·					
Create File						

• You can then display the results in a spreadsheet.

#### **Documentation**

ClinGen and ClinVar continue to collaborate on improving the coverage in ClinVar, the clarity of representation of the data, and the level of review

of current submissions. You will now notice a link to ClinGen on ClinVar's home page, as well as a new option under the How to submit menu, namely Expert panel. This takes you to a new page in our documentation set, http://www.ncbi.nlm.nih.gov/clinvar/docs/expert\_panel/, which provides instructions for earning attribution as an expert panel.

You will find new content in on our FAQ pages, and other documents are revised as our users request clarification. Please do not hesitate to contact us if you cannot find an answer to your question.

## **FTP site**

The ftp site was reorganized slightly to add more subdirectories README files:

- new subdirectories for vcf files based on GRCh37 and GRCh38
- moved \*xsd files to subdirectories specific to submission and data export
- moved the community folder to document\_archives

## Did you know?

You may be interested to know that the tool to view variation in sequence coordinates was recently updated.

http://www.ncbi.nlm.nih.gov/variation/view/

The tool supports both GRCh37 and GRCh38, and has multiple cool features such as:

- easy exon by exon navigation,
- support for uploading your data and viewing it in the context the public data set,
- filtering results in many ways
- downloading your result set
- and many more

For a quick start, please review the help documentation.

http://www.ncbi.nlm.nih.gov/variation/view/help/