NCBI News, December 2016

## New YouTube video: Sequence Viewer: Display Translation Discrepancies

Friday, December 23, 2016

The newest video on the NCBI YouTube channel is a brief introduction to a new set of sequence viewer renderings that better display discrepancies between genomic sequence and annotated features. These discrepancies can occur because RefSeq gene models based on the current genomic sequence can differ from RefSeq transcripts and corresponding proteins that come from our continuous curation efforts. For links to examples used in the video, see the Sequence Viewer tutorial.

Subscribe to the NCBI YouTube channel to receive alerts about new videos ranging from quick tips to full webinar presentations.

Sequence Viewer is a graphical view of sequences and color-coded annotations on regions of sequences stored in the Nucleotide and Protein databases.
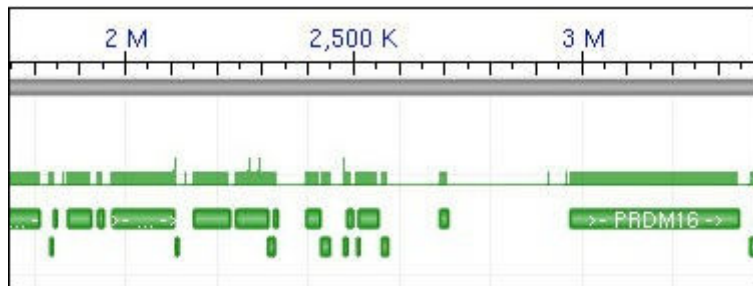
New NCBI Insights post: Converting Lots of GI Numbers to Accession.version

Friday, December 23, 2016

The latest post on the NCBI Insights blog provides a bulk conversion resource for those who those who need to convert more than a few thousand GI numbers to accession.version identifiers.

As you may already know, accession.version identifiers, rather than GI numbers, will be the primary identifiers for sequence records at NCBI.

NCBI Insights is the official NCBI blog, where we share science feature stories, quick tips and what's new at NCBI.

CCDS release 21 for mouse is public in Gene

Thursday, December 22, 2016

The Consensus Coding Sequence (CCDS) update that compares NCBI's Mus musculus annotation release 106 to Ensembl's release 86 is now available in Gene. This update adds 938 new CCDS IDs, and adds 137 genes into the mouse CCDS set. CCDS release 21 includes a total of 25,757 CCDS IDs that correspond to 20,354 GeneIDs.

Also, note that the CCDS survey is still open. NCBI and the CCDS collaboration invite you to take a survey that will help us assess how the human and mouse Consensus CDS data is being accessed and used by the scientific community. We welcome your feedback and suggestions on this data collection. Data gathered from the survey will help us plan the future direction of the CCDS project.

December 21st NCBI Minute: Bulk Conversion of NCBI Sequence GI Identifiers to accession.version

Thursday, December 15, 2016

Next Wednesday, NCBI will demonstrate how to use a downloadable database and Python script to convert GI identifiers to accession.version. The file and service that will be used are suitable for one-time conversion of very large sets of data.

Date and time: Wednesday, December 21, 2016 12:00 PM - 12:15 PM EST

Registration URL: https://attendee.gotowebinar.com/register/6267508028097746946

After registering, you will receive a confirmation email with information about attending the webinar. After the live presentation, the webinar will be uploaded to the NCBI YouTube channel. Any related materials will be accessible on the Webinars and Courses page; you can also learn about future webinars on this page.

Variant interpretations from Illumina double ClinVar data

Monday, December 12, 2016

On Dec 7, 2016, ClinVar released 138,334 variant interpretations submitted by Illumina Clinical Services Laboratory (ICSL) in San Diego, CA. This dataset represents a 57% increase in the number of submitted interpretations and makes ICSL the largest source of data in ClinVar. The contribution from ICSL also provides 78,590 novel variants to the database, an increase of 45% over the previous total of 173,782 variants.

The data were generated from clinical whole genome sequencing performed in the ICSL; variants were interpreted when the associated gene was in a predefined list of genes

associated with Mendelian disorders or when the gene-disease relationship had been manually curated. Information about the criteria that ICSL uses to interpret variants is available on the NCBI website.

ICSL has shared data through other NCBI resources as well, including the Genetic Testing Registry and the GeT-RM browser.

ClinVar is a freely accessible, public archive of reports of the relationships among human variations and phenotypes, with supporting evidence. Interpretations of variants are submitted to ClinVar by clinical testing laboratories, research laboratories, locus-specific databases, genetics clinics, expert panels, and professional societies that establish practice guidelines. The database currently holds 374,018 submitted interpretations representing 263,220 variants. ClinVar provides a public forum for variant interpretations and evidence, so that interpretations may be shared and subjected to peer review.

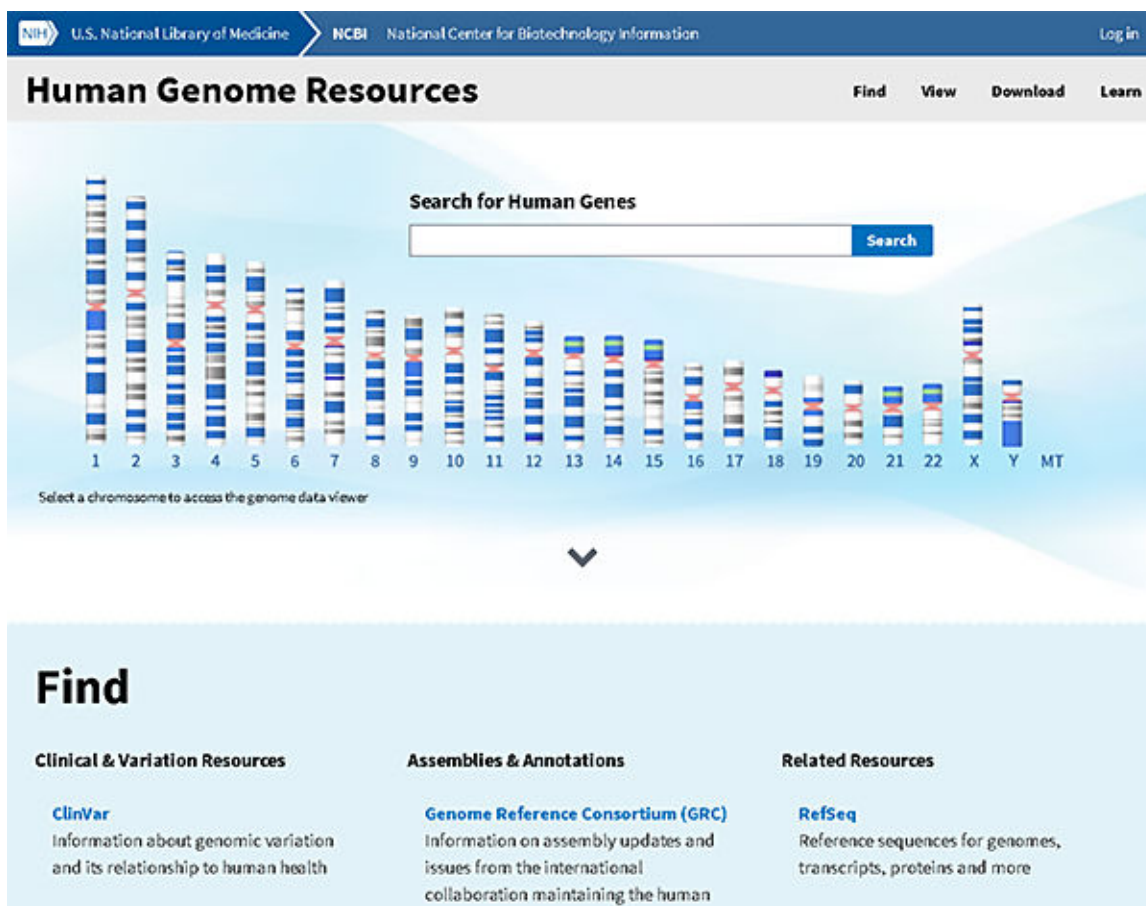## The new Human Genome Resources site: a portal for exploration of the human genome

Monday, December 12, 2016

The new Human Genome Resources site offers access to visualization and analysis tools available for the human genome, as well as other relevant tools like BLAST, the NCBI remapping service and databases that provide human molecular data. The resources are sorted into categories like Find, View, Download and Learn, making it easier to find what you need.

Some specific goals that you can accomplish through the site's guidance are:

- Finding information on individual genes that NCBI RefSeq staff annotate on the human genome assemblies and are archived in the Gene database.
- Visualizing and analyzing the genome by accessing individual chromosomes in the Genome Data Viewer and other available viewers.
- Comparing your sequences with the sequences of the human genome assemblies (BLAST).
- Navigating to the clinical and variation data through the complete listing of NCBI's clinical and variation resources.
- Accessing details about the human genome assemblies and annotation.
- Accessing various large datasets for download on the NCBI FTP site.
- Remapping annotation data between different assemblies (NCBI Genome Remapping Service).

In addition, the portal includes an extensive listing of learning resources that may help you have a better understanding of the wealth of information associated with the human genome.

Figure 1. The new NCBI Human Genome Resources webpage.

Sequence Viewer 3.18 is now available

Thursday, December 08, 2016

Sequence Viewer 3.18 has several new features, improvements and bug fixes, including improved handling of translation discrepancies, a new option for "Left-to-right translations" for the six frame translation track, and improved code generation. For a full list of changes, see the Sequence Viewer release notes.

Sequence Viewer is a graphical view of sequences and color-coded annotations on regions of sequences stored in the Nucleotide and Protein databases.

New on NCBI Insights: Converting GI Numbers to Accession.version

Tuesday, December 06, 2016

The latest blog post on NCBI Insights shows users how to convert GI numbers to accession.version with EFetch.

NCBI Insights is the official NCBI blog, where we share science feature stories, quick tips and what's new at NCBI.

NCBI Tech Talk and Booth at the American Society for Cell Biology 2016 National Meeting

Friday, December 02, 2016

NCBI staff will be participating in the ASCB 2016 National Meeting from Sunday, December 4 to Tuesday, December 6. We will be at booth #939 from 9AM -4PM PDT Sunday to Tuesday, and will present a Tech Talk on Sunday, December 4 at 5:30PM PDT in Theater 2.

The Tech Talk, "Five Useful Teaching Examples Using NCBI BLAST", will present demonstrations that highlight features of BLAST. These readily adaptable examples are useful for teaching biology principles and techniques including evolution, gene expression analysis and more.