### NCBI News, May 2014

### BLAST URL domain changes to take effect December 1, 2014

Thursday, May 22, 2014

As of December 1, 2014, BLAST searches sent to the www.ncbi.nlm.nih.gov/blast URL will not function. The officially supported URL domain for BLAST searches at the NCBI is **blast.ncbi.nlm.nih.gov**. Please update your bookmarks, links and any scripts or applications.

## GTR/ClinVar/MedGen webinar on June 18 will explore NCBI resources

Wednesday, May 21, 2014

On June 18, 2014, NCBI will offer a webinar entitled "Introducing 3 NCBI Resources to Navigate Testing for Disease Linked Variants: MedGen, GTR and ClinVar". This webinar will delve into the lifecycle of genetic testing and teach attendees how to navigate the NIH Genetic Testing Registry, ClinVar, and MedGen resources. These resources can be used to prepare for clinical cases, access detailed information about orderable genetic tests, interpret test results, and more.

To register, please go to this link.

#### RefSeq release 65 available on FTP site

Tuesday, May 20, 2014

The full RefSeq release 65 is now available with nearly 52 million records describing 38,633,935 proteins, 7,051,549 RNAs, and sequences from 36,335 different organisms.

As mentioned previously, several changes to the RefSeq release FTP site are now in place with this most recent release, including changes to directory names and file names. For more detail on these changes, please refer to this announcement.

More details about the RefSeq release 65 are included in the release statistics and release notes. In addition, reports indicating the accessions included in the release and the files installed are available.

2 NCBI News

# The NIH Genetic Testing Registry now has information on more than 3,500 cancer tests

Tuesday, May 20, 2014

Thanks to the efforts of testing laboratories that have voluntarily submitted information about their tests to NIH's Genetic Testing Registry (GTR), the database currently has information on more than 17,000 genetic tests, including about 3,500 related to cancer.

In late 2013, GTR expanded to include information about tests that evaluate somatic mutations (alterations in DNA that occur after conception). The response to that expansion was strong: laboratories have already submitted information on over 120 somatic tests for more than 200 conditions. Many of the somatic tests are for use in the area of cancer, including tests for prediction, prognosis, recurrence, and therapeutic management. To explore somatic tests in GTR, see this link.

In addition to somatic tests, GTR includes information on tests for hereditary cancers. GTR has included these types of tests since its inception in 2012, and laboratories have submitted information on more than 3,500 of them to date. For example, GTR has information on 199 tests for *BRCA1* or *BRCA2* from laboratories across the world, including 59 tests offered by 18 US labs. As advanced DNA sequencing techniques permeate clinical testing, complex panels for cancer have grown sharply in GTR: 30 of the tests for *BRCA1* or *BRCA2* are panels that evaluate 5 or more genes.

Pharmacogenetic tests, some of which relate to cancer drug responses, are another category of tests included in GTR. For example, tests have been registered in GTR for responses to tamoxifen, irinotecan, fluorouracil and thioguanine.

For all types of tests and in all medical areas, GTR aims to provide detailed information, such as the purpose of the test, target populations, methods, what it measures, analytical validity, clinical validity, clinical utility, and ordering information. Details about the laboratories include location, contact information, certifications and licenses.

GTR also includes extensive information beyond that relating to individual tests and laboratories. GTR links to context-specific information about medical conditions, genes, sequence variation, test standards, practice guidelines, pharmacogenetic information, clinical trials, molecular resources, and consumer support sites.

The GTR staff is excited about the growing participation in and the use of the database, as well as about recently introduced improvements to its functionality, such as the new advanced search feature for tests that was introduced in March. Searches can quickly find tests based on somatic or germline targets, pharmacogenetic responses, next-generation sequencing methods, number of targets (for complex panels), required specimen types, laboratory location, and more.

To learn more about GTR, visit the Genetic Testing Registry website, or see this article in *Nucleic Acids Research*. If you happen to be attending the American Society of Clinical

Oncology annual meeting at McCormick Place in Chicago, check out the GTR poster on May 31, 1:15 p.m. to 5 p.m. (in the Tumor Biology section of the General Poster Sessions). GTR Director Wendy Rubinstein, M.D., Ph.D. will be at the meeting from May 31 to June 3 and would be happy to talk with you about how to use the GTR website or how your laboratory can participate. You can contact us in advance via gtr@ncbi.nlm.nih.gov to arrange a time to meet.

### SciENcv 2.0 brings major improvements to My NCBI

Friday, May 16, 2014

Recent updates to SciENcv add several new useful features to the service.

#### Users can now:

- create multiple SciENcv profiles;
- download SciENcv profiles in PDF, Word, or XML format;
- add delegates to their SciENcv profiles;
- and add a mini profile in SciENcv to link to PubMed Commons.

The most recent NLM Technical Bulletin describes these changes in more detail, as will an upcoming NCBI Insights blog post.

SciENcv is a feature in My NCBI that helps users create online professional profiles that can be made public to share with others. SciENcv allows you to document education, employment, research activities, ORCID iDs, publications, grants, and other professional contributions. A guide on how to navigate and use SciENcv is on the Bookshelf.

### NCBI Sequence Viewer version 3.2 available

Tuesday, May 06, 2014

NCBI Sequence Viewer has recently been updated and now has support for multi-track upload and improved display of long track titles, as well as improved tooltips for variation features. A full list of new features, improvements and fixes is included in the release notes.

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