

The Genetic Testing Registry (GTR): Genetic Tests and More

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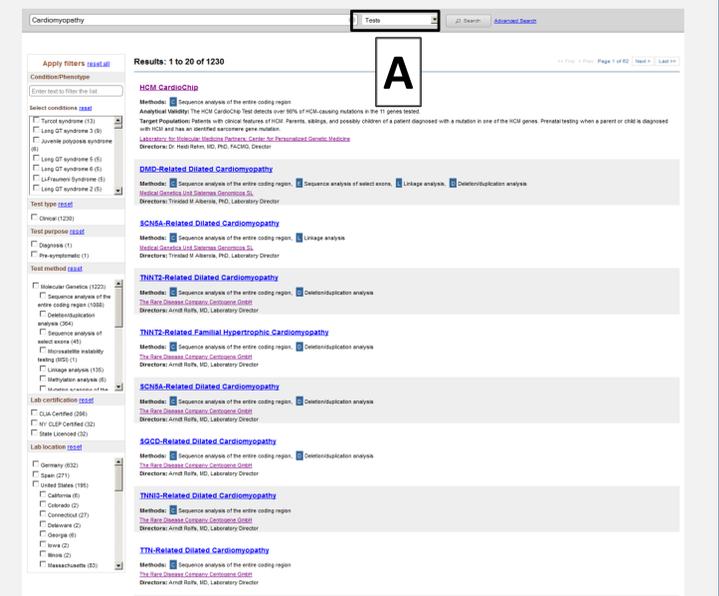
What is the GTR?

The Genetic Testing Registry (GTR) provides a centralized repository of genetic test information populated via voluntary submission from test providers. The scope includes the purpose, methodology, validity, and evidence of utility of each test, as well as laboratory contacts and credentials. The web site also supports ready access to context-specific information about conditions, genes, test standards, practice guidelines, and consumer support sites.

A test registry, not a laboratory directory. In the GTR, you can retrieve a test by name, by test provider name, and by purpose (e.g. name of condition or gene). If you do a non-specific search, as in (A) at the right, you can quickly focus on the result you want by checking the filter boxes.

A portal for medical geneticists. GTR provides a page specific to each condition. This page includes (B):

- The disease characteristics portion of a *GeneReviews* summary, with links to the complete review and each section;
- Summary of the tests available, with links to more details;
- List of associated genes;
- Context-specific links to review articles, practice guidelines, and resources for clinical practice, review of molecular biological details, and consumer resources; and
- Names from SNOMED CT[®] when available.



Representative result of a query for tests related to cardiomyopathy

What is novel to the GTR web site?

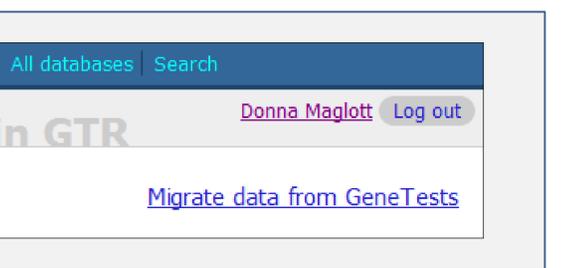
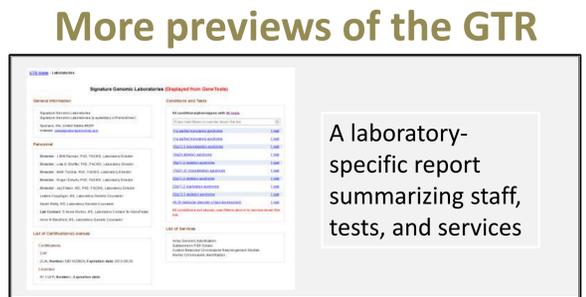
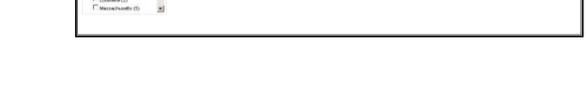
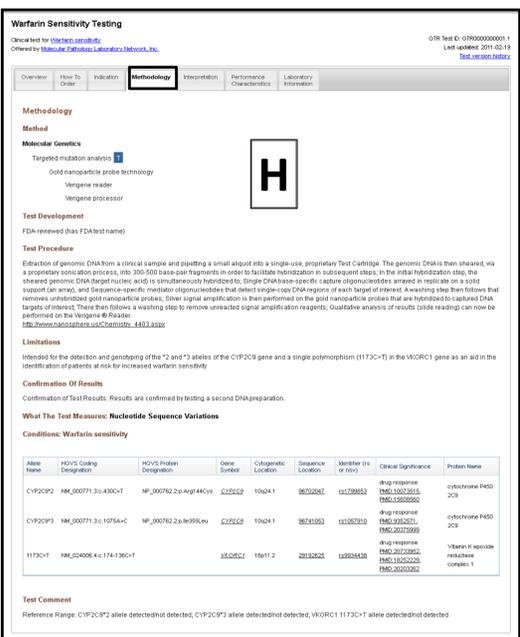
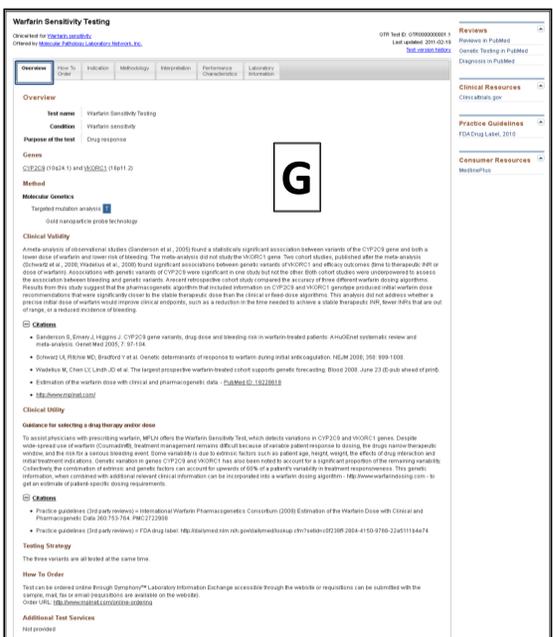
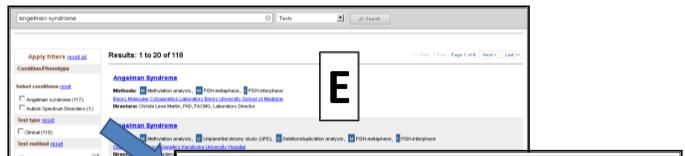
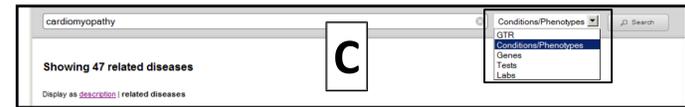
Global search. Search on any term or combination of terms, across all the GTR or a subset. In this example (C), the term *cardiomyopathy* is used to search for diseases with a name containing that word, but the same query could be directed against all of the GTR, only laboratory data, or only test names.

Save preferences for displays. Your preferences for displays and retrieval sets (such as your laboratory) can be established using the MyNCBI interface, via the **NCBI Site Preferences** section (D).

Quickly filter results. If your query results in many answers, the GTR provides filter choices to allow you to find quickly a desired subset. In the example in (E), the tests query for Angelman syndrome resulted in 118 listings, 117 for Angelman syndrome and 1 for Autism Spectrum Disorders. To limit the results to tests for Angelman syndrome only, based on sequence analysis, in labs that are NY CLEP-certified, all you need to do is check the appropriate boxes and review the subset of interest (F). You may consider additional methods offered by the laboratory in anticipation of further testing in the proband or family. Also, in the view showing all tests offered by a laboratory (J), a filter makes it easier to focus on the desired test.

Detailed information about each test to facilitate evaluation. As a registry of genetic tests, the GTR provides many details about each test. The test-specific report has many features, some of which are shown below. For this poster, the right panel, which is provided in each section, is reproduced only on the Overview image (G). **Note:** Pharmacogenetic tests are in scope for the GTR.

- G. Overview:** Provides a summary of the method, clinical validity, clinical utility, and ordering information.
- H. Methodology:** Includes the approach and the molecular details of the variations being assessed. Links are provided to displays of each variation on the genome relative to the exons of the gene, links to PubMed supporting the clinical significance, etc.
- I. Performance Characteristics:** Includes analytical validity, proficiency testing and FDA clearance/approval status.
- J. Stable accession and version history:** The GTR assigns an accession number, with versions, for each test. If a test is updated, a new version is created. The version history allows you to review previous versions.



How do I submit test information?*

Automatically **Migrate data from GeneTests**
 Web interface to facilitate submission of supplemental data
 Bulk upload via spreadsheet or XML

*Available early 2012



Contact us: gtr@ncbi.nlm.nih.gov **Demonstrations and Discussion:** NCBI Booth 217
Home page: http://www.ncbi.nlm.nih.gov/gtr/

