GTR: Genetic Testing Registry

Central repository of genetic tests voluntarily supplied by test providers

https://www.ncbi.nlm.nih.gov/gtr

National Center for Biotechnology Information • National Library of Medicine • National Institutes of Health • Department of Health and Human Services

Scope

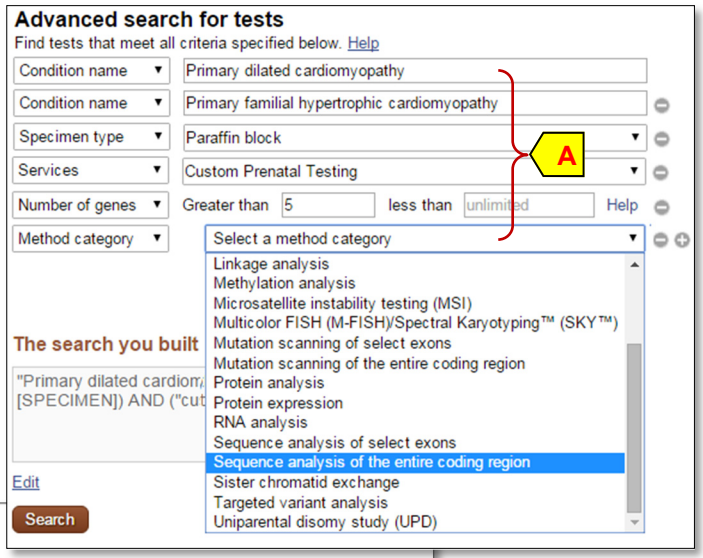
The Genetic Testing Registry (GTR) is a free online resource that provides centralized access to comprehensive genetic test information voluntarily submitted by test providers. The GTR covers clinical and research tests for heritable and somatic mutations, including pharmacogenetic tests as well as tests using complex arrays and multiplex panels. GTR provides a wide range of information, such as the purpose of the test and its limitations, whether it is a clinical or research test, the testing method(s), and what the test measures. Clinical tests have information on analytical validity, as well as evidence of clinical validity and clinical utility. Research tests have information on the study and participation requirements. The name, location, and credentials of laboratories providing the test are displayed. GTR records provide links to context-specific information about conditions, genes, test standards, practice guidelines, and consumer support sites. The primary audience of the initial phase of GTR is the health care community.

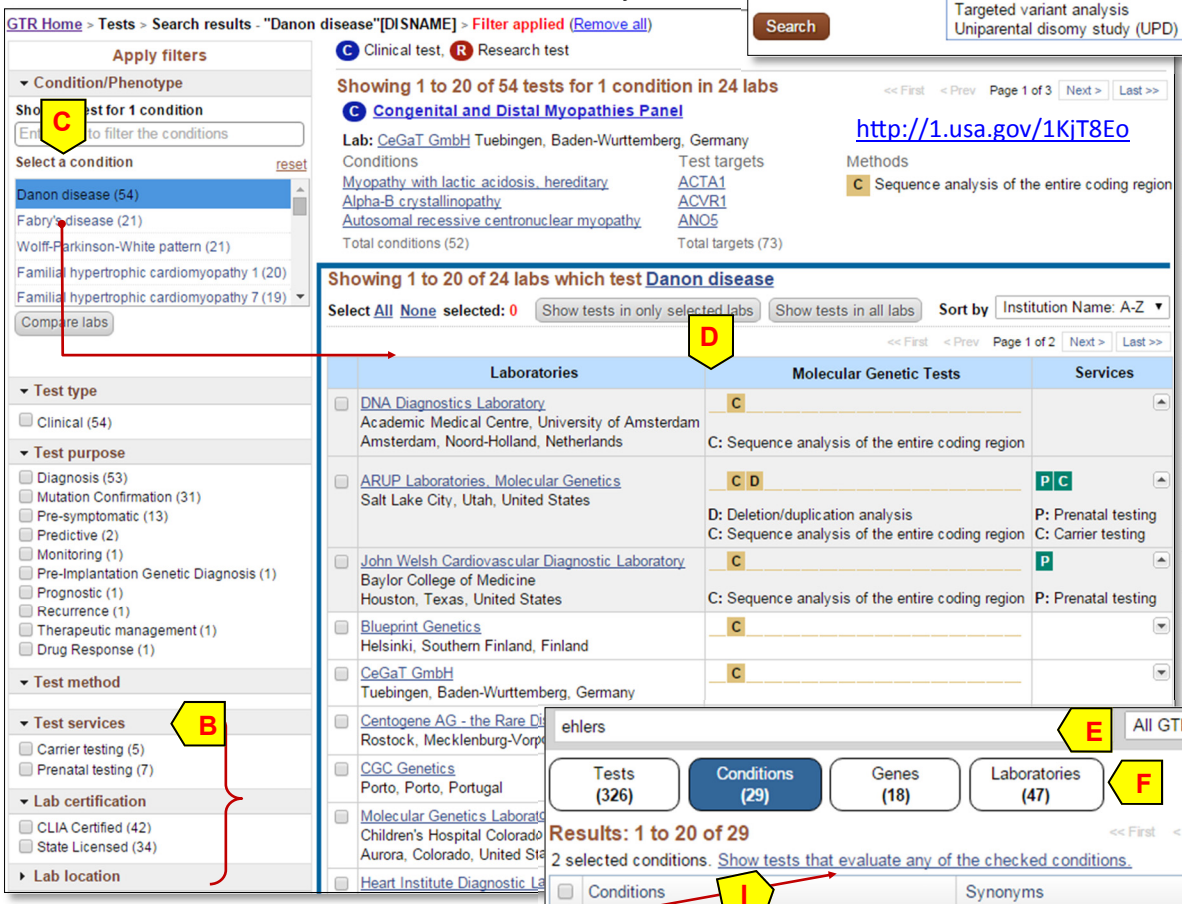
Data Access

The GTR homepage (right) is the gateway for accessing the data stored in the registry. It allows the retrieval of information through several different search strategies using tabs (**A**) above the search box. Users can search by the test name, disease name, trait, drug response, gene symbol, laboratory name, director and staff names, and laboratory location. A tab for *GeneReviews*, containing reviews on over 600 conditions, enables searching of this key resource directly from the GTR homepage. A set of YouTube video tutorials (**B**) provides a quick guide on how to search for and submit information to GTR. The homepage also provides links to online documentation (**C**), the submission portal (**D**), bulk data downloads (**E**), and clinical resources (**F**). The "Locate a Genetics Professional" section (**G**) provides links to external directory services through which genetic professionals can be located for consultation purposes.

Using advanced search to find specific tests

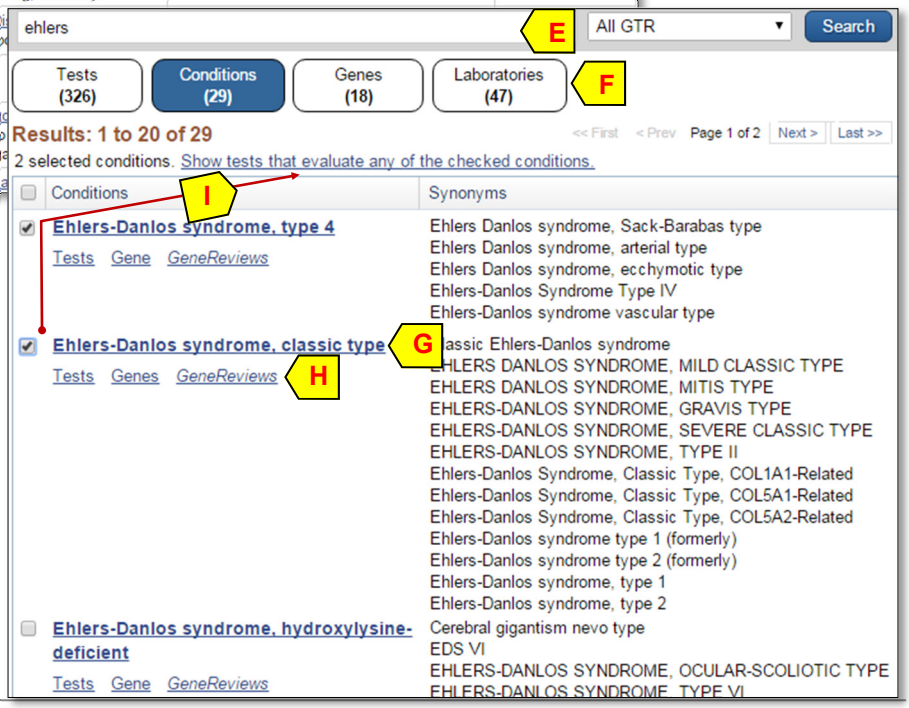
The "Advanced search for tests" allows you to construct custom queries to locate tests specified by the input criteria. The example (**A**) shows a search for panels of more than 5 genes for "Primary dilated cardiomyopathy" and "Primary familial hypertrophic cardiomyopathy" that can be tested on a paraffin block specimen, from a laboratory that provides custom prenatal testing. The list of tests returned by a search (using "Danon disease" as an example search here), can be further narrowed down by selecting items from the preset filters, such as "Test method" and "Lab location" (**B**). Selecting one condition enables the "Compare Labs" button (**C**) and allows the creation of a summary table, listing methodologies (**D**) of available tests for that condition from each laboratory.





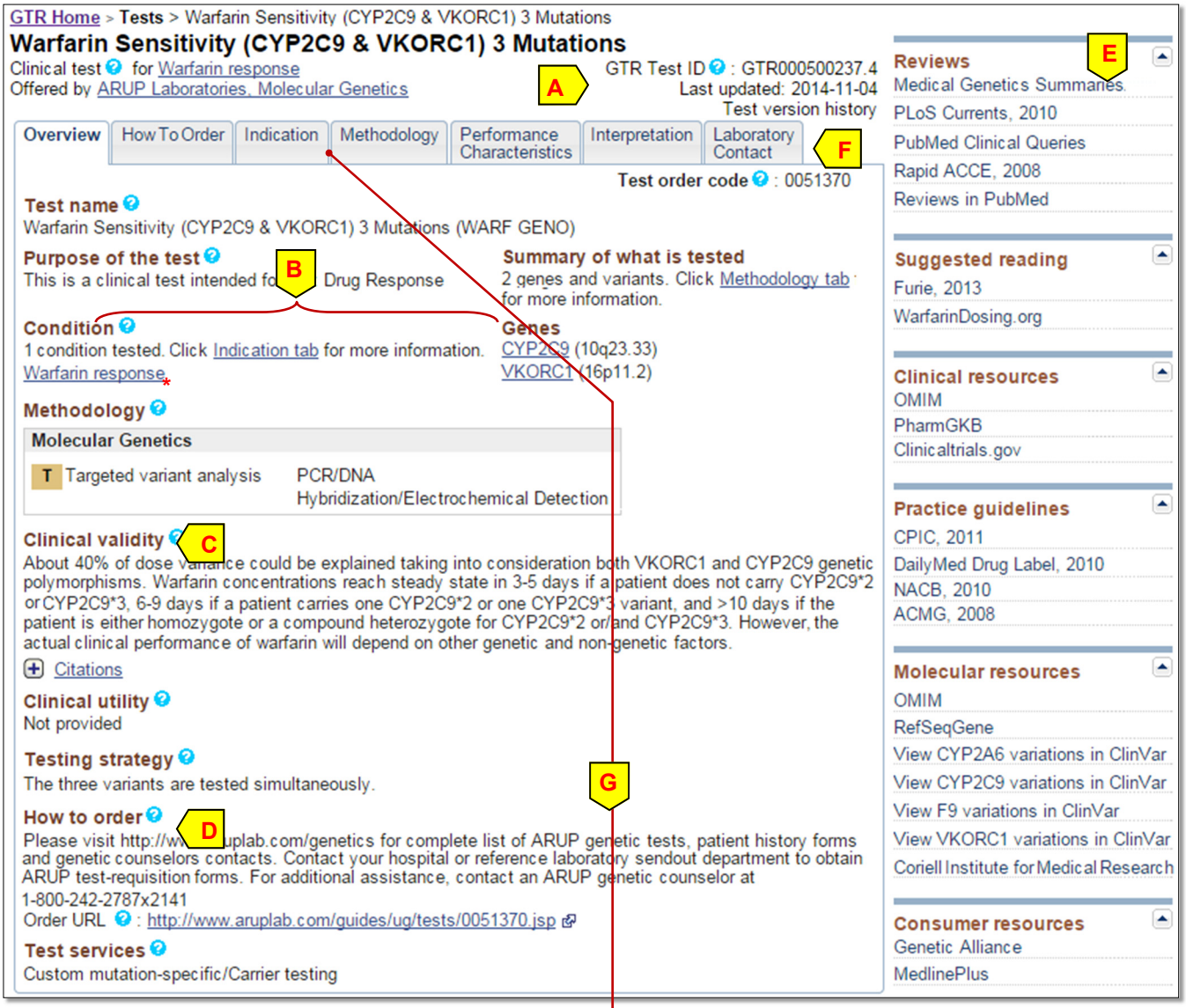
Using the All GTR tab

The main search box in the GTR homepage defaults to the "All GTR" tab. Entering a term (such as "Ehlers") and clicking the "Search All GTR" button (**E**) without selecting from the suggested list (or clicking "See all results"), the system will search all of GTR and display the result from the most appropriate category. Results from Tests, Conditions, Genes, and Laboratories are readily accessible by clicking the corresponding buttons (**F**). On the Conditions page, the name of each condition (**G**) links to the condition-specific page, providing information on clinical features, related conditions, and practice guidelines. Relevant links below a condition name provide information by linking to associated tests, genes, and article(s) from *GeneReviews*, respectively (**H**). Checking the boxes for any conditions activates a link (**I**) at the top, which can be used to retrieve a subset of tests for any of the selected conditions. The search box at the top (not shown) can be used to edit an existing search or start a new one.

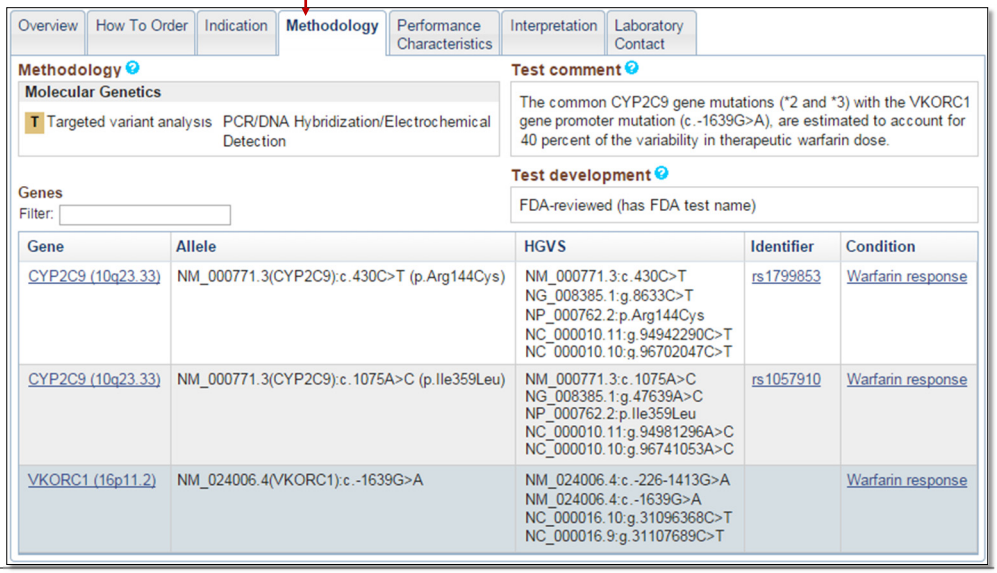


Navigating among different categories

The GTR web site provides a portal for medical genetics information which can be displayed in different formats, such as condition/disease-specific reports, test details, laboratory summaries, and gene-specific reports. This approach makes it easy to display key information and to quickly navigate to a specific category of information. A GTR-registered test for warfarin response (http://www.ncbi.nlm.nih.gov/gtr/tests/500237) is shown below.

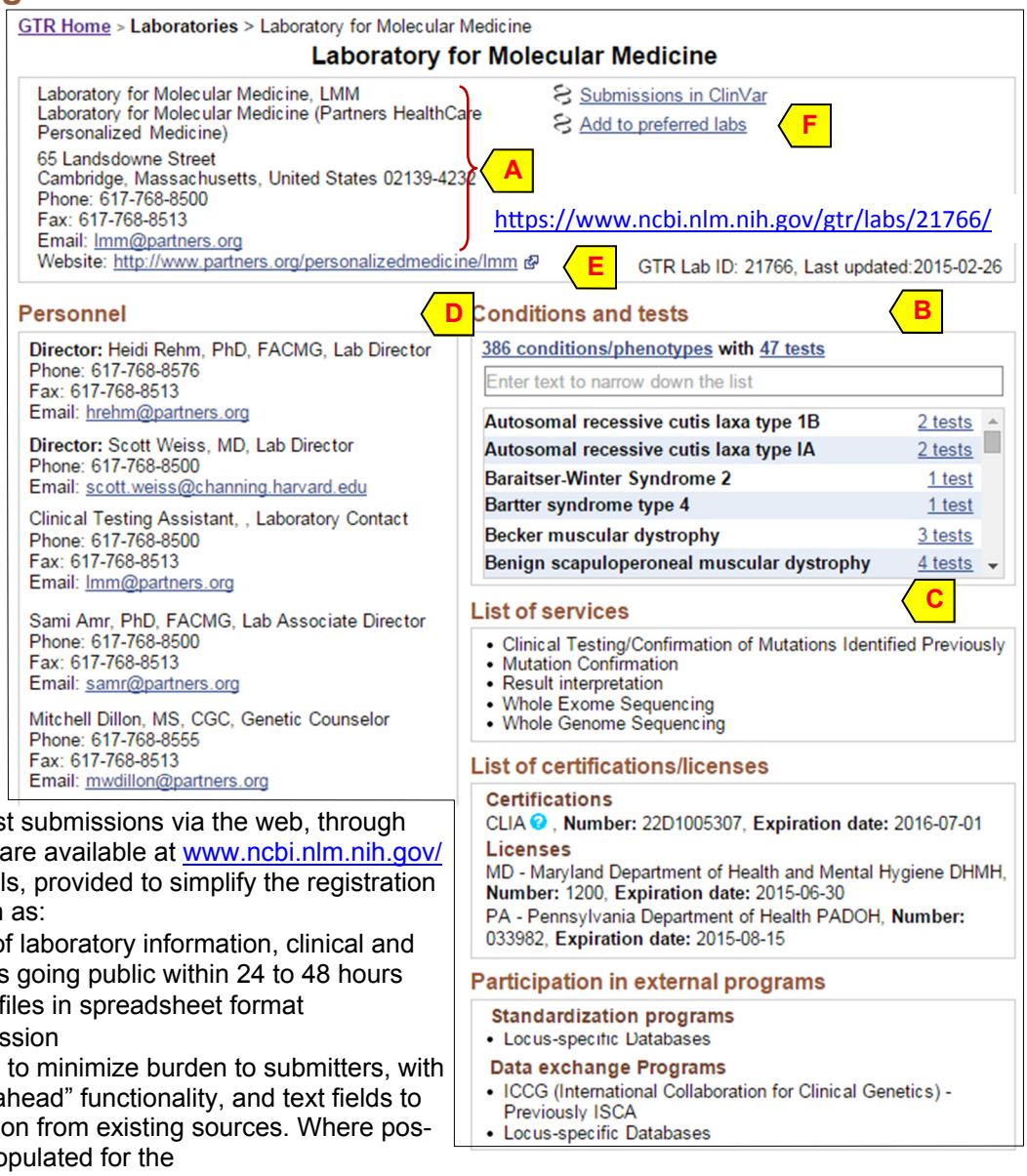


As a GTR-registered entry, this test is assigned an accession and version (**A**), which uniquely identifies it and its subsequent updates. The default display shows the overview of the test, describing the condition and genes involved (**B**), reported clinical validity (**C**), and how to order the test (**D**) from the laboratory. Context-sensitive links (**E**) relevant to this test are grouped into different categories and shown in the right-hand column. Other available information is organized under remaining tabs (**F**). For example, the "Methodology" tab (**G**) provides the details about the methods and targets on which the test is based.



Information on testing laboratories

Laboratory-specific pages in GTR display the contact information (**A**), the tests (**B**), and services (**C**) offered by the laboratory. Laboratories may also choose which staff members they want to show publicly in the GTR (**D**). GTR links to specific laboratories can be bookmarked (**E**). My NCBI enables selection of preferred labs (**F**) to customize the view of preferred labs and their available tests.



Submit test information to GTR

GTR accepts laboratory and test submissions via the web, through My NCBI. Detailed instructions are available at www.ncbi.nlm.nih.gov/gtr/docs/submit/. A range of tools, provided to simplify the registration process, includes features such as:

- Online form-based editing of laboratory information, clinical and research tests, with updates going public within 24 to 48 hours
- Bulk uploading clinical test files in spreadsheet format
- Fully automatic XML submission

The system has been designed to minimize burden to submitters, with extensive use of menus, "type ahead" functionality, and text fields to allow cut-and-paste of information from existing sources. Where possible, fields are automatically populated for the submitter. For example, once a disease condition for which a test is used is completed, disease identifiers, synonyms, acronyms and disease types related to the condition are automatically populated for review. In addition, test data identical for all tests in the lab's menu can be entered in the "Default parameters" section of the lab record to enable the system to pre-populate them for new tests being registered. Bulk submission of data is useful for providing information on multiple genetic tests and/or multiplex panel tests.

Submission templates are available online (ftp.ncbi.nlm.nih.gov/pub/GTR/submission_templates/) and in your lab's submission overview page. GTR provides information about the test provider as well as the availability, accuracy, validity and usefulness of each test, therefore a minimal set of fields are required for all submissions. The complete list of optional and minimal fields being requested for clinical tests is posted online (www.ncbi.nlm.nih.gov/gtr/docs/fieldrequirements/). About half of the minimal fields describe the laboratory, e.g., name, contacts, and are completed once. Research tests, i.e. tests that are performed for the purpose of contributing to generalizable knowledge or for a laboratory to generate data in order to make technical improvements to a test, require information about the condition(s), test target(s) and methodologies as well as information about the study and researchers. Minimal fields are clearly marked for easy entry with an orange asterisk in the submission user interface.

Contact

GTR welcomes feedback from the user community!
Questions and comments specific to GTR, as well as listserv subscription requests should be sent to: gtr@ncbi.nlm.nih.gov