



# GTR: Genetic Testing Registry

Central repository of genetic tests voluntarily supplied by test providers

<http://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 (<http://www.ncbi.nlm.nih.gov/gtr>, shown below) 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.

**GTR: GENETIC TESTING REGISTRY**

**All GTR** Tests Conditions/Phenotypes Genes Labs GeneReviews **A** [Advanced search for tests](#)

Find all types of GTR records, including tests, conditions/phenotypes, genes, and labs.

[YouTube GTR Tutorials](#) **B** [Search All GTR](#)

**IMPORTANT NOTE:** NIH does not independently verify information submitted to the GTR; it relies on submitters to provide information that is accurate and not misleading. NIH makes no endorsements of tests or laboratories listed in the GTR. GTR is not a substitute for medical advice. **Patients and consumers** with specific questions about a genetic test should contact a health care provider or a genetics professional.

**NIH thanks labs for registering over 25,000 tests for 5,000 conditions and 3,700 genes!**  
[YouTube basic search video](#)

**Molecular Resources**

- [ClinVar](#) Information about sequence variation and its relationship to human health. NIH.
- [Genetics & Medicine](#) NCBI's molecular medicine databases and tools. NIH. [See tips.](#)

**Clinical Resources** **F**

- [GeneReviews](#) Clinical information on genetic conditions, University of Washington. See NCBI's [Advanced Search](#).
- [MedGen](#) Medical genetics conditions, clinical features, practice guidelines, hierarchies and more. NIH.
- [OMIM](#) Online Mendelian Inheritance in Man, Johns Hopkins University.
- [Orphanet](#) Rare diseases and orphan drugs, European consortium.
- [NHGRI Talking Glossary](#) Genetic terms, images and animation. NIH.

**About GTR®**

The Genetic Testing Registry (GTR®) provides a central location for voluntary submission of genetic test information by providers. The scope includes the test's purpose, methodology, validity, evidence of the test's usefulness, and laboratory contacts and credentials. The overarching goal of the GTR is to advance the public health and research into the genetic basis of health and disease.

- [How to use GTR](#) **C**
- [Frequently asked questions](#)
- [GTR News](#)
- [GTR Information at NIH Office of the Director](#)
- [GTR in the community](#)
- [Contact us and provide feedback](#)

**Submitting Information to GTR**

- [How to submit data](#) **D**
- [Code of Conduct](#)
- [Access the Submission user interface](#)

**Files Available for Download** **E**

- [List of files](#)
- [Field Definitions pdf | Word](#)

**Locate a Genetics Professional** **G**

- [ACMG Genetics Clinics Database](#) American College of Medical Genetics and Genomics database, with map-based views.
- [NSGC Directory](#) National Society of Genetic Counselors directory.
- [NCL Cancer Genetics Services Directory](#) National Cancer Institute directory of professionals who provide cancer genetics services.
- [ABMGG Directory](#) American Board of Medical Genetics and Genomics directory of board-certified geneticists.
- [ABGC Directory](#) American Board of Genetic Counselors directory of board-certified genetic counselors.

**Consumer Resources**

- [Genetics Home Reference](#) Consumer-friendly information about genetic variation and human health. NIH.

## Using advanced search to find specific tests

The advanced search allows users 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 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.

**Advanced search for tests**  
Find tests that meet all criteria specified below. [Help](#)

Condition name: Primary dilated cardiomyopathy  
Condition name: Primary familial hypertrophic cardiomyopathy  
Specimen type: Paraffin block  
Services: Custom Prenatal Testing  
Number of genes: Greater than 5 less than unlimited [Help](#)  
Specimen type: Paraffin block  
Select a specimen type: Frozen tissue, Isolated DNA, Paraffin block, Peripheral (whole) blood, Plasma

**The search you built**  
"Primary dilated cardiomyopathy" AND "Primary familial hypertrophic cardiomyopathy" AND "Paraffin block" [SPECIMEN TYPE]

**Apply filters**

- Condition/Phenotype: Showing test for 1 condition. Enter text to filter the conditions. Select a condition: Danon disease (6)
- Test type
- Test purpose
- Test method: Molecular Genetics (6), Sequence analysis of the entire coding region (5), Deletion/duplication analysis (2)
- Test services
- Lab certification
- Lab location: United States (4), California (1), Utah (3), Finland (2)

**Showing 1 to 20 of 27 labs**  
Select All None selected: 0

Laboratories	Molecular Genetic Tests	Services
<input type="checkbox"/> DNA Diagnostics Laboratory Academic Medical Centre, University of Amsterdam Amsterdam, Noord-Holland, Netherlands	C: Sequence analysis of the entire coding region	
<input type="checkbox"/> ARUP Laboratories, Molecular Genetics Salt Lake City, Utah, United States	D: Deletion/duplication analysis C: Sequence analysis of the entire coding region	P: Prenatal testing C: Carrier testing
<input type="checkbox"/> John Welsh Cardiovascular Diagnostic Laboratory Baylor College of Medicine Houston, Texas, United States	C: Sequence analysis of the entire coding region	P: Prenatal testing
<input type="checkbox"/> Blueprint Genetics Helsinki, Southern Finland, Finland	C	
<input type="checkbox"/> Centogene AG - the Rare Disease Company Rostock, Mecklenburg-Vorpommern, Germany	C D	P C
<input type="checkbox"/> CGC Genetics	C	

## 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 the "See all results" (F), the system will search all 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 (G). On the Conditions page, the name of each condition (H) 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 (I). Checking the boxes for any conditions activates a link (J) 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.

**GTR: GENETIC TESTING REGISTRY**

All GTR Tests Conditions/Phenotypes Genes Labs GeneReviews [Advanced search for tests](#)

ehlers  [Search All GTR](#)

ATCS: Ehlers-Danlos syndrome, musculocontractural type Condition and labs.  
EDS10: Ehlers-Danlos syndrome dysfibronectinemic type Condition tests or laboratories listed in the GTR. GTR relies on submitters to provide information about a genetic test should contact a  
EDS11: Ehlers-Danlos syndrome, familial joint laxity type Condition  
EDS1: Ehlers-Danlos Syndrome, Classic Type, COL1A1-Related Condition  
EDS1: Ehlers-Danlos Syndrome, Classic Type, COL5A2-Related Condition  
[See all results](#)

Tests (277) Conditions (30) Genes (18) Laboratories (44)

**Results: 1 to 20 of 30**  
2 selected conditions. [Show tests that evaluate any of the checked conditions](#)

Condition	Synonyms
<input checked="" type="checkbox"/> Ehlers-Danlos syndrome, type 4	Ehlers Danlos syndrome, Sack-Barabas type Ehlers Danlos syndrome, arterial type Ehlers Danlos syndrome, ecchymotic type Ehlers-Danlos Syndrome Type IV Ehlers-Danlos syndrome vascular type
<input checked="" type="checkbox"/> Ehlers-Danlos syndrome, type 1	EDS I EHLERS-DANLOS SYNDROME, GRAVIS TYPE EHLERS-DANLOS SYNDROME, SEVERE CLASSIC TYPE EHLERS-DANLOS SYNDROME, TYPE I Ehlers-Danlos Syndrome, Classic Type, COL1A1-Related Ehlers-Danlos Syndrome, Classic Type, COL5A1-Related Ehlers-Danlos Syndrome, Classic Type, COL5A2-Related
<input type="checkbox"/> Ehlers-Danlos syndrome, hydroxylysine-deficient	Cerebral gigantism nevo type EDS VI EHLERS-DANLOS SYNDROME, OCULAR-SCOLIOTIC TYPE EHLERS-DANLOS SYNDROME, TYPE VI EHLERS-DANLOS SYNDROME, TYPE VIA Ehlers-Danlos Syndrome, Kyphoscoliotic Form Ehlers-Danlos syndrome kyphoscoliotic type Ehlers-Danlos syndrome type 6 (formerly) Nevo syndrome

## 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.

[GTR Home](#) > [Tests](#) > Warfarin Sensitivity (CYP2C9 & VKORC1) 3 Mutations

### Warfarin Sensitivity (CYP2C9 & VKORC1) 3 Mutations

Clinical test [for Warfarin response](#)  
Offered by [ARUP Laboratories, Molecular Genetics](#)

GTR Test ID: GTR000500237.4  
Last updated: 2014-11-04  
Test version history

Overview | **How To Order** | Indication | Methodology | Performance Characteristics | Interpretation | Laboratory Contact

Test order code: 0051370

**Test name**  
Warfarin Sensitivity (CYP2C9 & VKORC1) 3 Mutations (WARF GENO)

**Purpose of the test**  
This is a clinical test intended for Drug Response

**Condition**  
1 condition tested. Click [Indication tab](#) for more information.  
[Warfarin response](#) \*

**Methodology**  
Molecular Genetics  
Targeted variant analysis PCR/DNA  
Hybridization/Electrochemical Detection

**Clinical validity**  
About 40% of dose variance could be explained taking into consideration both VKORC1 and CYP2C9 genetic polymorphisms. Warfarin concentrations reach steady state in 3-5 days if a patient does not carry CYP2C9\*2 or CYP2C9\*3, 6-9 days if a patient carries one CYP2C9\*2 or one CYP2C9\*3 variant, and >10 days if the patient is either homozygote or a compound heterozygote for CYP2C9\*2 or/and CYP2C9\*3. However, the actual clinical performance of warfarin will depend on other genetic and non-genetic factors.

**Clinical utility**  
Not provided

**Testing strategy**  
The three variants are tested simultaneously.

**How to order**  
Please visit <http://www.aruplab.com/genetics> for complete list of ARUP genetic tests, patient history forms and genetic counselors contacts. Contact your hospital or reference laboratory sendout department to obtain ARUP test-requisition forms. For additional assistance, contact an ARUP genetic counselor at 1-800-242-2787x2141  
Order URL: <http://www.aruplab.com/guides/ug/tests/0051370.jsp>

**Test services**  
Custom mutation-specific/Carrier testing

**Genes**  
[CYP2C9](#) (10q23.33)  
[VKORC1](#) (16p11.2)

**Summary of what is tested**  
2 genes and variants. Click [Methodology tab](#) for more information.

**Reviews**  
Medical Genetics Summaries  
PLoS Currents, 2010  
PubMed Clinical Queries  
Rapid ACCE, 2008  
Reviews in PubMed

**Suggested reading**  
Furie, 2013  
[WarfarinDosing.org](#)

**Clinical resources**  
OMIM  
PharmGKB  
[Clinicaltrials.gov](#)

**Practice guidelines**  
CPIC, 2011  
DailyMed Drug Label, 2010  
NACB, 2010  
ACMG, 2008

**Molecular resources**  
OMIM  
RefSeqGene  
[View CYP2A6 variations in ClinVar](#)  
[View CYP2C9 variations in ClinVar](#)  
[View F9 variations in ClinVar](#)  
[View VKORC1 variations in ClinVar](#)  
Coriell Institute for Medical Research

**Consumer resources**  
Genetic Alliance  
MedlinePlus

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.

Overview | How To Order | Indication | **Methodology** | Performance Characteristics | Interpretation | Laboratory Contact

**Methodology**  
Molecular Genetics  
Targeted variant analysis PCR/DNA Hybridization/Electrochemical Detection

**Test comment**  
The common CYP2C9 gene mutations (\*2 and \*3) with the VKORC1 gene promoter mutation (c.-1639G>A), are estimated to account for 40 percent of the variability in therapeutic warfarin dose.

**Test development**  
FDA-reviewed (has FDA test name)

Genes  
Filter:

Gene	Allele	HGVS	Identifier	Condition
<a href="#">CYP2C9</a> (10q23.33)	NM_000771.3(CYP2C9):c.430C>T (p.Arg144Cys)	NM_000771.3:c.430C>T NG_008385.1:g.8633C>T NP_000762.2:p.Arg144Cys NC_000010.11:g.94942290C>T NC_000010.10:g.96702047C>T	<a href="#">rs1799853</a>	<a href="#">Warfarin response</a>
<a href="#">CYP2C9</a> (10q23.33)	NM_000771.3(CYP2C9):c.1075A>C (p.Ile359Leu)	NM_000771.3:c.1075A>C NG_008385.1:g.47639A>C NP_000762.2:p.Ile359Leu NC_000010.11:g.94981296A>C NC_000010.10:g.96741053A>C	<a href="#">rs1057910</a>	<a href="#">Warfarin response</a>
<a href="#">VKORC1</a> (16p11.2)	NM_024006.4(VKORC1):c.-1639G>A	NM_024006.4:c.-226-1413G>A NM_024006.4:c.-1639G>A NC_000016.10:g.31096368C>T NC_000016.9:g.31107689C>T		<a href="#">Warfarin response</a>

## 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). MyNCBI 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/](http://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 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/](http://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 (89) and minimal (31) fields being requested for clinical tests is posted online ([www.ncbi.nlm.nih.gov/gtr/docs/fieldrequirements/](http://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](mailto:gtr@ncbi.nlm.nih.gov)

GTR Home > Laboratories > Laboratory for Molecular Medicine

### Laboratory for Molecular Medicine

Laboratory for Molecular Medicine, LMM  
 Laboratory for Molecular Medicine (Partners HealthCare Personalized Medicine)  
 65 Landsdowne Street  
 Cambridge, Massachusetts, United States 02139-4232  
 Phone: 617-768-8500  
 Fax: 617-768-8513  
 Email: [lmm@partners.org](mailto:lmm@partners.org)  
 Website: <http://www.partners.org/personalizedmedicine/lmm>

[Submissions in ClinVar](#)  
[Add to preferred labs](#)

<http://www.ncbi.nlm.nih.gov/gtr/labs/21766/>

GTR Lab ID: 21766, Last updated: 2015-02-26

#### Personnel

**Director:** Heidi Rehm, PhD, FACMG, Lab Director  
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 Fax: 617-768-8513  
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#### Conditions and tests

386 conditions/phenotypes with 47 tests

Enter text to narrow down the list

Autosomal recessive cutis laxa type 1B	2 tests
Autosomal recessive cutis laxa type IA	2 tests
Baraitser-Winter Syndrome 2	1 test
Barter syndrome type 4	1 test
Becker muscular dystrophy	3 tests
Benign scapulo-peroneal muscular dystrophy	4 tests

#### List of services

- Clinical Testing/Confirmation of Mutations Identified Previously
- Mutation Confirmation
- Result interpretation
- Whole Exome Sequencing
- Whole Genome Sequencing

#### List of certifications/licenses

**Certifications**  
 CLIA, Number: 22D1005307, Expiration date: 2016-07-01

**Licenses**  
 MD - Maryland Department of Health and Mental Hygiene DHMH, Number: 1200, Expiration date: 2015-06-30  
 PA - Pennsylvania Department of Health PADOH, Number: 033982, Expiration date: 2015-08-15

#### Participation in external programs

**Standardization programs**

- Locus-specific Databases

**Data exchange Programs**

- ICCG (International Collaboration for Clinical Genetics) - Previously ISCA
- Locus-specific Databases