



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.

GTR: GENETIC TESTING REGISTRY

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

<https://www.ncbi.nlm.nih.gov/gtr> Search All GTR

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

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

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 you!
Over **32,000** tests for **5,800** conditions and **3,900** genes!
Watch GTR on YouTube (1/5) << || >>

Quick Links

- Labs that offer genomic testing services
- Panels with 5 or more genes including *BRCA1* and *BRCA2*
- Cancer / somatic tests
- Single-gene tests (NOT panels)
- Pharmacogenetic responses and links to those tests **F**
- Comparative Genomic Hybridization tests
- All GTR content

[Tell us what other quick links you need!](#)

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 conditions 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

Molecular Resources

- ClinVar Information about sequence variation and its relationship to human health, NIH.
- MedGen Information about human disorders and phenotypes having a genetic component, NIH.
- Genetics & Medicine NCBI's molecular medicine databases and tools, NIH. [See tips.](#)

Clinical Resources

- 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.

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.
- NCI 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 Counseling directory of board-certified genetic counselors.

Consumer Resources

- Genetics Home Reference Consumer-friendly information about genetic variation and human health, NIH.
- Office of Rare Diseases Research Rare diseases information for patients, families, healthcare providers, researchers, educators and students, NIH.

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.

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

Condition name

Condition name

Specimen type

Services

Number of genes less than

Method category

The search you built

"Primary dilated cardiomyopathy (SPECIMEN)" AND ("Primary familial hypertrophic cardiomyopathy (SPECIMEN)) AND ("Custom Prenatal Testing")

[Edit](#)

Select a method category

- Linkage analysis
- Methylation analysis
- Microsatellite instability testing (MSI)
- Multicolor FISH (M-FISH)/Spectral Karyotyping™ (SKY™)
- Mutation scanning of select exons
- Mutation scanning of the entire coding region
- Protein analysis
- Protein expression
- RNA analysis
- Sequence analysis of select exons
- Sequence analysis of the entire coding region
- Sister chromatid exchange
- Targeted variant analysis
- Uniparental disomy study (UPD)

GTR Home > Tests > Search results - "Danon disease"[DISNAME] > Filter applied (Remove all)

Apply filters

Condition/Phenotype

Show tests for 1 condition

Enter text to filter the conditions

Select a condition

Danon disease (54)

Fabry disease (21)

Wolff-Parkinson-White pattern (21)

Familial hypertrophic cardiomyopathy 1 (20)

Familial hypertrophic cardiomyopathy 7 (19)

Test type

Clinical (54)

Test purpose

Diagnosis (53)

Mutation Confirmation (31)

Pre-symptomatic (13)

Predictive (2)

Monitoring (1)

Pre-Implantation Genetic Diagnosis (1)

Prognostic (1)

Recurrence (1)

Therapeutic management (1)

Drug Response (1)

Test method

Test services

Carrier testing (5)

Prenatal testing (7)

Lab certification

CLIA Certified (42)

State Licensed (34)

Lab location

Clinical test, Research test

Showing 1 to 20 of 54 tests for 1 condition in 24 labs

Congenital and Distal Myopathies Panel

Lab: [CeGaT GmbH](#) Tuebingen, Baden-Wuerttemberg, Germany

Conditions

Myopathy with lactic acidosis, hereditary

Alpha-B crystallinopathy

Autosomal recessive centronuclear myopathy

Total conditions (52)

Test targets

ACTA1

ACVR1

ANO5

Total targets (73)

<http://1.usa.gov/1KjT8Eo>

Methods

Sequence analysis of the entire coding region

Showing 1 to 20 of 24 labs which test Danon disease

Select All None selected: 0 Show tests in only selected labs Show tests in all labs Sort by Institution Name: A-Z

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

ehlers

Tests (326) Conditions (29) Genes (18) Laboratories (47)

Results: 1 to 20 of 29

2 selected conditions. Show tests that evaluate any of the checked conditions.

Conditions	Synonyms
<input checked="" type="checkbox"/> Ehlers-Danlos syndrome, type 4 Tests Gene GeneReviews	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, classic type Tests Genes GeneReviews	Classic Ehlers-Danlos syndrome EHLERS DANLOS SYNDROME, MILD CLASSIC TYPE EHLERS DANLOS SYNDROME, MITIS TYPE EHLERS-DANLOS SYNDROME, GRAVIS TYPE EHLERS-DANLOS SYNDROME, SEVERE CLASSIC TYPE EHLERS-DANLOS SYNDROME, TYPE II Ehlers-Danlos Syndrome, Classic Type, COL1A1-Related Ehlers-Danlos Syndrome, Classic Type, COL5A1-Related Ehlers-Danlos Syndrome, Classic Type, COL5A2-Related Ehlers-Danlos syndrome type 1 (formerly) Ehlers-Danlos syndrome type 2 (formerly) Ehlers-Danlos syndrome, type 1 Ehlers-Danlos syndrome, type 2
<input type="checkbox"/> Ehlers-Danlos syndrome, hydroxylysine-deficient Tests Gene GeneReviews	Cerebral gigantism nevo type EDS VI EHLERS-DANLOS SYNDROME, OCULAR-SCOLIOTIC TYPE EHLERS-DANLOS SYNDROME, TYPE VI

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.

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,

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

Test order code [: 0051370](#)

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

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](#)*

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

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 and CYP2C9*3. However, the actual clinical performance of warfarin will depend on other genetic and non-genetic factors.

Citations

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](http://www.aruplab.com/guides/ug/tests/0051370.jsp)

Test services
Custom mutation-specific/Carrier testing

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
Cornell 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.

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
CYP2C9 (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	rs1799853	Warfarin response
CYP2C9 (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	rs1057910	Warfarin response
VKORC1 (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		Warfarin response

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

[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
 Website: <http://www.partners.org/personalizedmedicine/lmm>

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

<https://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
 Phone: 617-768-8576
 Fax: 617-768-8513
 Email: hrehm@partners.org

Director: Scott Weiss, MD, Lab Director
 Phone: 617-768-8500
 Email: scott.weiss@channing.harvard.edu

Clinical Testing Assistant, Laboratory Contact
 Phone: 617-768-8500
 Fax: 617-768-8513
 Email: lmm@partners.org

Sami Amr, PhD, FACMG, Lab Associate Director
 Phone: 617-768-8500
 Fax: 617-768-8513
 Email: samr@partners.org

Mitchell Dillon, MS, CGC, Genetic Counselor
 Phone: 617-768-8555
 Fax: 617-768-8513
 Email: mwdillon@partners.org

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