

**PROPOSED ONLINE SUBMISSION FORM FOR THE  
GENETIC TESTING REGISTRY**

This document provides screenshots of the proposed online form for the submission of genetic test information to the Genetic Testing Registry.

The laboratory examples are fictitious.

Public reporting burden for this collection of information is estimated to average 3.0 hours per response, including the time for reviewing instructions, searching existing data sources, gathering and maintaining the data needed, and completing and reviewing the collection of information. An agency may not conduct or sponsor, and a person is not required to respond to, a collection of information unless it displays a currently valid OMB control number. Send comments regarding this burden estimate or any other aspect of this collection of information, including suggestions for reducing this burden, to: NIH, Project Clearance Branch, 6705 Rockledge Drive, MSC 7974, Bethesda, MD 20892-7974, ATTN: PRA (0925-xxxx). Do not return the completed form to this address.

## Your Labs in GTR

[Help](#) | [Contact GTR staff](#) | [FAQ](#)

Add a new lab

Migrate data from GeneTests

Sort by: Modified: newest first

### Generic Laboratory, Inc (Incomplete Information, cannot submit)

Generic Laboratory, Inc  
Generic Institutes  
100 Maple street  
Springfield, OH

Phone: 555-555-5555  
Fax: 555-555-4444  
Email: [info@genericlabs.com](mailto:info@genericlabs.com)

**Lab Director(s)**

John Smith

**Laboratory Affiliations**

Generic Institutes for Disease A

**Laboratory Credentials**

None

**Test**

None

Last modified: 3/15/2011

Edit

View Details

### Generic Laboratory 2, Inc

Generic Laboratory 2 Inc, Generic Institutes  
200 Maple road  
Washington, DC 12345  
United States of America

Phone Number: 1-800-555-5555  
Fax Number: 1-800-555-4444  
Email: [info@genericlabs2.com](mailto:info@genericlabs2.com)  
Website: [www.genericlabs2.com](http://www.genericlabs2.com)

**Lab Director(s)**

Jane Doe, PhD, FACMG

**Laboratory Affiliations**

Generic Clinical Research Services

**Laboratory Credentials**

CLIA Certification Number 00D012345

**Test**

[HCM Cardio Panel](#) for Cardiomyopathy (Incomplete Information)  
[Warfarin Sensitivity Testing](#) for Warfarin sensitivity [\(Submit this test\)](#)

Last modified: 3/14/2011

Submit

Edit

View Details



# Add a New Lab

## Lab Information

## Personnel

## Licensure & Accreditation

## Default Parameters

\*: required input

### Name & Institution

Lab name: \*  Acronyms:

GeneTests ID:

Institution: \*  Acronyms:

Department:

### Address

Country or region: \*

Street & No:

City: \*

State or province: \*

Postal code: \*

Make this mailing address public: \*  Yes  No

Phone: \*

Fax:

Email: \*

Website:

### Types of Service

Service:  Order code:

### Affiliation

Name:  Website:

### Participation in programs

Participation in standardization programs: *(Click to select all that apply)*

- ISCA Consortium (International Standards for Cytogenomic Array)
- CETT Program (Collaboration Education and Test Translation)
- Mutation-specific Databases

Participation in data exchange programs: *(Click to select all that apply)*

- ISCA Consortium (International Standards for Cytogenomic Array)
- CETT Program (Collaboration Education and Test Translation)
- Mutation-specific Databases



## Generic Laboratory, Inc

[Lab Information](#)

**Personnel**

[Licensure & Accreditation](#)

[Default Parameters](#)

### Add a person

First name: \*

Middle Initial:

Last name: \*

Display on GTR site: \*  Yes  No

Primary lab contact: \*  Yes  No

Lab director: \*  Yes  No

Job title:   


- Lab Director
- Genetic Counselor
- Nurse
- Research Nurse

Academic degree(s): *(Click to select all that apply)*

MD   
 PhD   
 MS   
 BS

### Certificates:

#### Add new

Medical board:

Medical specialty:

Medical subspecialty:

Save

Credentials: *(Click to select all that apply)*

FACMG   
 CGC

Show credentials after name:  Yes  No

GTR permissions: \*

View only   
 Edit   
 Add   
 Delete   
 All

### Contact information to be displayed on GTR site

Phone:

Email:

Fax:

Supplementary public contact information:

### Contact information for GTR staff use only

Phone:

Email:

Fax:

(The current input will be saved.)

(The current input will not be saved.)

(Finish adding people. The current input will be saved.)



# Generic Laboratory, Inc

Lab Information

Personnel

Licensure & Accreditation

Default Parameters

John Smith, MD, Lab Director

Delete Edit

### Adding a person

First name: \*

Middle Initial:

Last name: \*

Display on GTR site: \*  Yes  No

Primary lab contact: \*  Yes  No

Lab director: \*  Yes  No

Job title:

- Lab Director
- Genetic Counselor
- Nurse
- Research Nurse

Academic degree(s): *(Click to select all that apply)*

- MD
- PhD
- MS
- BS

### Certificates:

Add new

Medical board:

Medical specialty:

Medical subspecialty:

Credentials: *(Click to select all that apply)*

- FACMG
- CGC

Show credentials after name:  Yes  No

GTR permissions: \*

- View only
- Edit
- Add
- Delete
- All

### Contact information to be displayed on GTR site

Phone:

Email:

Fax:

Supplementary public contact information:

### Contact information for GTR staff use only

Phone:

Email:

Fax:

(The current input will be saved.)

(The current input will not be saved.)

(Finish adding people. The current input will be saved.)



[GTR Submission](#)

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## Generic Laboratory, Inc

[Lab Information](#)

**Personnel**

[Licensure & Accreditation](#)

[Default Parameters](#)

John Smith, MD, Lab Director

[Edit](#) [Delete](#)

Jane Doe, PhD, Genetic Counselor

[Edit](#) [Delete](#)



# Generic Laboratory, Inc

[Lab Information](#)

[Personnel](#)

[Licensure & Accreditation](#)

[Default Parameters](#)

### CLIA Certification

Certification #:  Exp. Date:

### State License(s)

State:  License #:  Exp. Date:

### Other License(s)

Licensed by:  License #:  Exp. Date:



# Generic Laboratory, Inc

[Lab Information](#)

[Personnel](#)

[Licensure & Accreditation](#)

[Default Parameters](#)

## Optional: Default Parameters (May be overwritten for specific tests)

Test contact policy:

- Pre-test email/phone consultation
- Post-test email/phone consultation
- Laboratory can accept contact only from health care providers

Specimen source:

Peripheral (whole) blood
Buccal swab
Saliva
Amniocytes
Amniotic fluid
Bone marrow
Cell culture
Chorionic villi
Cord blood
Cystic hygroma fluid
Dried blood spot (DBS) card

## Upload Sample Test Reports

Sample negative report:

 

Sample positive report:

 

## Generic Lab: List of Tests

Add a new test

Sort by: Modified: newest first

HCM Cardio Panel **(Incomplete Information)**

**Disease(s):**  
Cardiomyopathy

**Gene(s):**  
HCM

Last modified: 3/21/2011

Edit

View Details



## Generic Lab: Adding a Test

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### Test Information

This test is for: \*  Clinical purposes  Research purposes

Laboratory test name: \*

Short test name:

Manufacturer's test name:

Other test name:  Name type:

Purpose of the test: \* *(Click to select all that apply)*

- Diagnosis
- Screening
- Drug Response
- Risk Assessment
- Presymptomatic
- Mutation Confirmation (family specific or research results, etc)
- Preimplantation genetic diagnosis

Test development:

Lab unique code:

FDA category designation:

- IVD – In Vitro Device
- RUO – Research Use Only
- IVO – Informational/Investigational Use Only
- LDT – Laboratory Developed Test

### FDA Review

#### Add new

FDA review of:

- Test kit(s)
- Assay(s)
- Reagent(s)
- Instrument(s)

FDA regulatory status:

- Reviewed
- Approved
- 510(k) Cleared
- PMA Approved

FDA application #:

Upload FDA approval document:

### Test-Specific License(s)

Licensed by:  License #:  Exp. Date:



## Generic Lab: Adding a Test

[Basics](#) **Ordering** [Indication](#) [Methodology](#) [Interpretation](#) [Performance Characteristics](#)

### Ordering Information

URL to order the test:

URL for the test:

Order code:

URL for order code:

Informed consent:  Required  Not required  Based on applicable state law

Pre-test genetic counseling:  Required  Not required

Post-test genetic counseling:  Required  Not required

Test specific lab service:  Test specific lab service code:

Specimen:  Requirement URL:

Who can order this test: *(Click to select all that apply)*

- Health Care Provider
- Public Health Mandate
- Out-of-State Patients
- In-State Patients
- Licensed Physician
- Physician Assistant
- Registered Nurse
- Genetic Counselor

How to order:

Testing strategy:

Citations for testing strategy:

### Contact Information

Contact person:

- Contact policy:
- Pre-test email/phone consultation
  - Post-test email/phone consultation
  - Laboratory can only accept contact from health care providers



# Generic Lab: Adding a Test

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## Disease

Name: \*

- Familial hypertrophic cardiomyopathy 10
- Familial hypertrophic cardiomyopathy 4
- Cardiomyopathy, Hypertrophic, Familial

Disease name to be used for display:

Synonyms: *(Click to select all that apply)*



- Hereditary ventricular hypertrophy
- Asymmetric septal hypertrophy
- Idiopathic hypertrophic subaortic stenosis

Disease synonym to be used for display:

Preferred acronym:

Acronyms: *(Click to select all that apply)*



- 

Disease acronym to be used for display:

Disease type:

- Dysmorphology Syndrome
- Cancer Syndrome
- Neurology

Mode of inheritance:

Disease mechanism:

Similar disorders: *(Click to select all that apply)*

Prevalence:

Citation for Prevalence:



Target population:

Citation for target population:



Save & Continue



# Generic Lab: Adding a Test

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## Disease

Name: \*

Disease name to be used for display:

Synonyms: *(Click to select all that apply)*



- Hereditary ventricular hypertrophy
- Asymmetric septal hypertrophy
- Idiopathic hypertrophic subaortic stenosis

Disease synonym to be used for display:

Preferred acronym:

Acronyms: *(Click to select all that apply)*



- FHC
- ASH
- IHSS

Disease acronym to be used for display:

Disease type:

Mode of inheritance:

Disease mechanism:

Similar disorders: *(Click to select all that apply)*

- Dilated Cardiomyopathy (DCM)
- Sudden Cardiac Death (SCD)

Prevalence:

Citation for Prevalence:

Target population:

Citation for target population:



# Generic Lab: Adding a Test

[Basics](#) | [Ordering](#) | [Indication](#) | **Methodology** | [Interpretation](#) | [Performance Characteristics](#)

## Methodology (page 1 of 2)

### Method

Major method category: \*

- Biochemical Genetics
- Cytogenetics
- Molecular Genetics

Category: \* *(Click to select all that apply)*

- Sequence analysis of the entire coding region Deletion/...
- Sequence analysis of select exons
- Targeted mutation analysis

Primary test methodology:

- PCR-RFLP with Southern hybridization
- RT-PCR with gel analysis
- Trinucleotide repeat by PCR or Southern Blot
- Protein truncation

Platforms: *(Click to select all that apply)*

- Affymetrix GeneChip
- Agilent microarrays
- CodeLink Bioarray
- NimbleGen microarray

Instruments: *(Click to select all that apply)*

- Qiagen AutoPure LS
- Qiagen QIAcube
- Tecan Genesis Robotic Workstation 150
- PerkinElmer Victor3 1420 Multilabel Plate Reader
- Agilent 2100 Bioanalyzer
- Applied Biosystems 7900HT Sequence Detection System
- Applied Biosystems SOLiD v4 System Sequencer

Test procedure:

Confirmation of test results:

Test comment:



# Generic Lab: Adding a Test

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Methodology (page 2 of 2) [Return to page 1](#)

What the test measure: \*

- Nucleotide Mutations
- Haplotypes
- Chromosome Rearrangements
- Full genome

## Enter Test Targets

Add a new target

Target is identified by: Gene Symbol  
Chromosome Location

Gene symbol: \*

Associated Reference Sequences: \* (Click to select all that apply)

- NM\_123456.1
- NM\_123456.3
- NM\_1234577
- Add other reference sequence...

Select relevant exons for each associate reference sequence:

NM_123456.1	<input type="checkbox"/> 1-5 (c. 1 - c.1000)
NM_1234577	<input type="checkbox"/> 1 (c. 1 - c.100)
	<input type="checkbox"/> 2 (c.201 - c.300)
	<input type="checkbox"/> 3 (c.301 - c.400)

Relevant gene variants: (Click to select all that apply)

- All variants
- A123R
- V133T
- Add an additional variant...

Clinical significance of variant A123R:

- Pathogenic
- Presumed pathogenic

Clinical significance applies to condition:

Citations to support the above clinical significance:



## Generic Lab: Adding a Test

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### Upload Sample Reports

Sample negative report:

 

Sample positive report:

 

### Variants of Unknown Significance (VUS): Policy and Interpretation

What is the protocol for interpreting a variation as a VUS?

What software is used to interpret novel variations?

What is the laboratory's policy on reporting novel variations?

Upload sample VUS report:

 

Are family members with defined clinical status recruited to assess significance of VUS without charge?

Yes  
  No  
  Decline to answer

Will the lab re-contact the ordering physician if variant interpretation changes?

Yes  
  No  
  Decline to answer

Research performed after clinical testing is complete:



# Generic Lab: Adding a Test

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## Performance Characteristics (page 1 of 2)

### Availability

Test performance location(s): \*

- Entire test performed in-house
- Entire test performed externally
- Specimen preparation performed in-house
- Specimen preparation performed externally
- Wet lab work performed in-house
- Wet lab work performed externally
- Interpretation performed in-house
- Interpretation performed externally
- Report generated in-house
- Report generated externally

If parts of the test is performed externally: \*

- I am authorized to enter details of the test:  Yes  No
- This entry has been reviewed by the external collaborator(s) for accuracy:  Yes  No

### Analytical Validity

Analytical validity: \* (Discuss number of specimens, analytical specificity, precision, and accuracy)

Citations to support analytical validity: \*

Add Another

### Assay Limitations

Assay limitations: (Discuss limit of detection and test restrictions)

Citations to support assay limitations:

Add Another

### Quality Control

Is proficiency testing performed for this test?  Yes  No

Method for proficiency testing:

▲
▼
▢
 Formal PT program  
 Alternative assessment  
 Intra-laboratory

Provider for proficiency testing:

▲
▼
▢
 CAP  
 Alternative assessment  
 Intra-laboratory

CAP test list:

▲
▼
▢
 Item1  
 Item2  
 Item3

Description of proficiency testing method: (Discuss testing results, reportable range, testing intervals, and number of specimens per interval)

Citations to support the above statement:

Add Another

Description of Internal test validation method: (Discuss reportable range)

Citations to support the above statement:

Add Another

Save & Continue



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## Performance Characteristics (page 2 of 2) [Return to page 1](#)

### Clinical Validity

Statement of clinical validity:

Citations to support the above statement:

[Add Another](#)

### Clinical Utility

Add a new clinical utility

Select a category of clinical utility or enter a new one:

- Establish or confirm diagnosis
- Guidance for management
- Guidance for selecting a drug therapy and/or dose
- Reproductive decision-making
- Avoidance of invasive testing
- Predictive risk information for patient and/or family members
- Lifestyle planning
- Sufficient research has not been conducted to demonstrate the utility of the test.

Statement to explain the clinical utility:

Citations to support the clinical utility:

Citations category:



[Add Another](#)

[Save this clinical utility](#)

[Save](#)