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.

OMB control number: (0925-xxxx) Expiration date: (xx-xx-xxxx)

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

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
Website: 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

OMB control number: (0925-xxxx) Expiration date: (xx-xx-xxxx)

GTR Submission

Help | Contact GTR staff | FAQ

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:* United States

Street & No:

City:*

State or province:* Alabama

Postal code:*

Make this mailing address public:* Yes No

Phone:*

Fax:

Email:*

Website:

Types of Service

Service: Order code:

Genetic counseling Add Another

Affiliation

Name: Website:

Add Another

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

Save & Continue

OMB control number: (0925-xxxx) Expiration date: (xx-xx-xxxx)

GTR Submission

Help | Contact GTR staff | FAQ

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: American Board of Obstetrics and Gynecology

Medical specialty: Please select a medical board first

Medical subspecialty: Please select a medical board first

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:

Add Another (The current input will be saved.)

Cancel (The current input will not be saved.)

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

OMB control number: (0925-xxxx) Expiration date: (xx-xx-xxxx)

GTR Submission

Help | Contact GTR staff | FAQ

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: American Board of Obstetrics and Gynecology

Medical specialty: Please select a medical board first

Medical subspecialty: Please select a medical board first

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:

Add Another (The current input will be saved.)

Cancel (The current input will not be saved.)

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

OMB control number: (0925-xxxx) Expiration date: (xx-xx-xxxx)

GTR Submission

Help | Contact GTR staff | FAQ

Generic Laboratory, Inc

Lab Information | Personnel | Licensure & Accreditation | Default Parameters

John Smith, MD, Lab Director	Edit	Delete
Jane Doe, PhD, Genetic Counselor	Edit	Delete

Continue Add Another

OMB control number: (0925-xxxx) Expiration date: (xx-xx-xxxx)

GTR Submission

Help | Contact GTR staff | FAQ

Generic Laboratory, Inc

Lab Information | Personnel | Licensure & Accreditation | Default Parameters

CLIA Certification

Certification #: Exp. Date: Add Another

State License(s)

State: Alabama License #: Exp. Date: Add Another

Other License(s)

Licensed by: ISO License #: Exp. Date: Add Another

Continue

OMB control number: (0925-xxxx) Expiration date: (xx-xx-xxxx)

GTR Submission

Help | Contact GTR staff | FAQ

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:

Browse

Sample positive report:

Browse

Finish Adding This Lab

OMB control number: (0925-xxxx) Expiration date: (xx-xx-xxxx)

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

OMB control number: (0925-xxxx) Expiration date: (xx-xx-xxxx)

Generic Lab: Adding a Test

Basics | Ordering | Indication | Methodology | Interpretation | Performance Characteristics

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: Keyword [Add Another]

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: FDA-reviewed

Lab unique code:

FDA category designation:

- IVD – In Vitro Device
- RUO – Research Use Only
- IUO – 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: [Browse]

[Save]

Test-Specific License(s)

Licensed by:	License #:	Exp. Date:	
NYSCLEP			Add Another

[Save & Continue]

OMB control number: (0925-xxxx) Expiration date: (xx-xx-xxxx)

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: Add Another

Specimen: Whole blood Requirement URL: Add Another

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: Add Another

Contact Information

Contact person: John Smith Add New

Contact policy:

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

Save & Continue

OMB control number: (0925-xxxx) Expiration date: (xx-xx-xxxx)

Generic Lab: Adding a Test

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)* Add

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)* Add

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: Add Another

Target population:

Citation for target population: Add Another

Save & Continue

OMB control number: (0925-xxxx) Expiration date: (xx-xx-xxxx)

Generic Lab: Adding a Test

Disease

Name: * Cardiomyopathy, Hypertrophic, Familial

Disease name to be used for display:
Hypertrophic cardiomyopathy

Synonyms: *(Click to select all that apply)*
Add
- Hereditary ventricular hypertrophy
- Asymmetric septal hypertrophy
- Idiopathic hypertrophic subaortic stenosis

Disease synonym to be used for display:

Preferred acronym:
HCM

Acronyms: *(Click to select all that apply)*
Add
- FHC
- ASH
- IHSS

Disease acronym to be used for display:
HCM

Disease type:
- Dysmorphology Syndrome
- Cancer Syndrome
- Neurology

Mode of inheritance:
- Autosomal Dominant
- Autosomal Recessive
- X-linked

Disease mechanism:
Production of altered protein with changed function

Similar disorders: *(Click to select all that apply)*
- Dilated Cardiomyopathy (DCM)
- Sudden Cardiac Death (SCD)

Prevalence:

Citation for Prevalence:
PMID: 18403758
Add Another

Target population:

Citation for target population:
Add Another

Save & Continue

OMB control number: (0925-xxxx) Expiration date: (xx-xx-xxxx)

Generic Lab: Adding a Test

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:

Save & Continue

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Generic Lab: Adding a Test

Methodology (page 2 of 2) Return to page 1

What the test measure: *

- Nucleotide Mutations
- Haplotypes
- Chromosome Rearragements
- 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	1-5 (c.1 - c.1000)
NM_1234577	1 (c.1 - c.100)
	2 (c.201 - c.300)
	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:

Add Another

Save this target

Save & Continue

OMB control number: (0925-xxxx) Expiration date: (xx-xx-xxxx)

Generic Lab: Adding a Test

Upload Sample Reports

Sample negative report:

Browse

Sample positive report:

Browse

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:

Browse

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:

Save & Continue

OMB control number: (0925-xxxx) Expiration date: (xx-xx-xxxx)

Generic Lab: Adding a Test

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 suport 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 suport the above statement:

Add Another

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

Citations to suport the above statement:

Add Another

Save & Continue

NCBI Resources How To Smith,John My NCBI Signout

GENETIC TESTING REGISTRY

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Generic Lab: Adding a Test

Basics Ordering Indication Methodology (Page 1 2) Interpretation Performance Characteristics

Performance Characteristics (page 2 of 2) Return to page 1

Clinical Validity

Statement of clinical validity:

Citations to suport 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 suport the clinical utility:

Citations category:

Clinical Trials
Review

Add Another

Save this clinical utility

Save