


Information Collection Screenshots

Online submission form for the Genetic Testing Registry (GTR)

This document provides updated screenshots of the online form for the submission of genetic test information to the Genetic Testing Registry (GTR).

12/22/2014

SECURE LOGIN SYSTEM





GTR Submission

[Contact GTR Staff](#) [Help Documents](#) [GTR Homepage](#) [Log in](#)

- OMB NO: 0925-0651
- EXPIRATION DATE: 2/28/2015
- [Burden statement](#)


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Last Revision: 438633 2014-05-29 14:08:55

BURDEN STATEMENT

Resources How To kattmanb My NCBI Sign Out

GTR: GENETIC TESTING REGISTRY

[Overview](#) [About](#) [Public Search Site](#) [Submission](#) [Lab Submission](#) [Clinical Test Submission](#) [Research Test Submission](#) [Print this document](#)

OMB NO: 0925-0651
EXPIRATION DATE: 02/28/2015

Burden Statement:

Public reporting burden for this collection of information is estimated to vary from 14 minutes to 30 minutes 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 (0925-0651). Do not return the completed form to this address.

You are here: [NCBI](#) > [GTR](#) > [GTR OMB Approval](#) [Write to the Help Desk](#)

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GTR CODE OF CONDUCT

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GTR Submission

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Code of Conduct

Test submitters providing test information to the Genetic Testing Registry (GTR) agree to abide by a code of conduct. Failure to honor this code of conduct may result in the removal of the submitter's test information from the GTR. Submitters agree to the following terms in the code of conduct:

- To uphold the integrity of the GTR through the submission of information that is accurate and not misleading.
- To assure the accuracy of the data at the time of submission and to review and, if necessary, update the submitted information at least once a year.
- To make no explicit or implicit claims that the National Institutes of Health, the Department of Health and Human Services, or the U.S. Government approves or endorses tests listed in, or any other information submitted to, the GTR.

To reference their participation in the GTR, test submitters may refer to the fact that information about their tests is available in the GTR and provide the relevant URL(s) but make no explicit or implicit claims that their tests listed in the GTR, or other information submitted to the GTR, have been approved or endorsed by the National Institutes of Health (NIH), the Department of Health and Human Services, or the U.S. Government. If this stipulation is not honored, NIH reserves the right to take action, including, in its sole discretion, removing the submitter's tests from the GTR.

In addition, users are encouraged to report any acts of inappropriate endorsement claims or any other breaches of this Code of Conduct on our [Contact GTR](#) page.

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Last Revision: 446721 2014-09-17 12:16:38

AMA CPT CODE LICENSE AGREEMENT

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GTR Submission

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AMA CPT Code License Agreement

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Registrants are defined as genetic test developers who are adding their tests to the National Center for Biotechnology Information's Genetic Testing Registry ("Genetic Testing Registry") as maintained by the National Library of Medicine.

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YOUR LABS IN GTR



GTR Submission

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[Help Documents](#)
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 Acting as [Lisa Catalano](#)
[Switch back to cit|kattmanb](#)
[Log out](#)
[Migrate data from GeneTests at NCBI](#)

 OMB NO: 0925-0651
 EXPIRATION DATE: 2/28/2015
[Burden statement](#)

Emory Genetics Laboratory

Review lab submission

Submission Status

Lab ID: 500060
 Status: processed-ok
 Last modified: 2014-06-13

Click here for the public display of this lab. Please note: It takes 24-48 hours to process a submission. If the lab record was submitted or edited in the last 48 hours, the public site may not display your currently submitted data.

If you want to delete this laboratory from the GTR, please [contact GTR staff](#).

Lab General Information

Lab name: **Emory Genetics Laboratory, Emory University**
 Address: 2165 North Decatur Road
 Decatur Georgia 30033
 Email: eglgc@emory.edu
 Website: <http://genetics.emory.edu/eg/>
 Phone: 855-831-7447
 Fax: 404-778-8559

Submission of Tests

Clinical test: [Add a new clinical test](#)
[Add tests by spreadsheet](#)
for complex clinical tests or many tests
 Research test: [Add a new research test](#)

Annual Review

Last review performed: 2014-06-16
 Next review due: 2015-06-16

[Perform Annual Review](#) [read more](#)

Completion resets the next due date one year forward.

Lab Director(s)

Christin Collins, PhD
 Bradford Coffee, PhD
 Lora Bean, PhD
 Madhuri Hegde, PhD
 Alice Tanner, PhD, MS
 Katie Rudd, PhD
 Yuan Xue, PhD, MD
 Patricia Hall, PhD
 J Sharer, PhD
 John Alexander, PhD
 Zunyan Dai, PhD

Lab Credentials

CLIA: 11D0683478 exp: 2014-03-15
 MD - Maryland Department of Health and Mental Hygiene DHMH: 1346 exp: 2014-06-30
 PA - Pennsylvania Department of Health PADOH: 031676 exp: 2013-08-15
 College of American Pathologists, CAP: 7181693 exp: 2014-08-02

Tests in this lab(875)

Test name	Test type	Submission status	Action	Use to create a new test	Link to public site
1q21.1 Deletion/Duplication Analysis	Clinical test	Processed successfully	Update test	Copy	ID: GTR000508266
3-Hydroxy-3-Methylglutaryl CoA Lyase Deficiency (HMG): HMGCL Full Gene Sequencing	Clinical test	Processed successfully	Update test	Copy	ID: GTR000502886
3-Hydroxy-3-Methylglutaryl CoA Lyase Deficiency (HMG): HMGCL Gene Deletion/Duplication	Clinical test	Processed successfully	Update test	Copy	ID: GTR000502887
3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC): MCCC1/MCCC2 Full Gene Sequencing	Clinical test	Processed successfully	Update test	Copy	ID: GTR000502913
3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC): MCCC1/MCCC2 Gene Deletion/Duplication	Clinical test	Processed successfully	Update test	Copy	ID: GTR000502918
Aarskog-Scott Syndrome: FGD1 Full Gene Sequencing	Clinical test	Processed successfully	Update test	Copy	ID: GTR000501493

ADD A NEW LAB

NCBI


GTR Submission

[Contact GTR Staff](#) [Help Documents](#) [GTR Homepage](#) [Acting as Lisa Catalano](#) [Switch back to cit|kattmanb](#) [Log out](#)

New

Submission ID:SUB180239

[Lab information](#) [Personnel](#) [Licensure and accreditations](#) [Default parameters](#) [Overview](#)

* Required field. * Completed field. Hover over  to display help information.

Laboratory & Institution Name

* **Name of laboratory**  **Acronym of lab name** 

GeneTests at NCBI lab ID, if known 

Name of institution  **Acronym of institution name** 

Name of department 

Laboratory Address


* **Country or region**
 United States 


Street & No 


Additional address line 


* **City** 


State or province 
 Alabama 

* **Postal code** 

* Yes No **Make this address public?** 

Phone number: XXX-XXX-XXXX (U.S.A), +(country code)-AreaCode-XXXXXX ext XXXX (International) 

Fax number: XXX-XXX-XXXX (U.S.A), +(country code)-AreaCode-XXXXXX ext XXXX (International) 

*** Email (ex. lab@lab.com) and/or** **URL for lab contact form** 

Lab website URL 

Laboratory Types of Service

Service  **Order code**  **Comment**  


 [Add another service](#)

Laboratory Affiliation(s)

Name of affiliate (example: clinic, research center)  **Website**  

 [Add another affiliation](#)

Laboratory Participation in External Programs

Participation in standardization programs (select all that apply) 

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

Participation in data exchange programs (select all that apply) 

- CETT Program (Collaboration Education and Test Translation)
- ClinVar
- ICCG (International Collaboration for Clinical Genetics) - Previously ISCA
- Locus-specific Databases
- Mutation-specific Databases
- Other

to delete

Submission ID: SUB180239

[Lab information](#)
[Personnel](#)
[Licensure and accreditations](#)
[Default parameters](#)
[Overview](#)

This lab has the following staff members and research personnel:

Category	Name	Title	Action
Lab staff	Brandi Kattman	Genetic Counselor	Delete Edit

- Please add all staff members relevant to your lab registration. Personnel entered here will be available for selection when submitting clinical and research tests. You may specify whether they will display on the public page of the lab.
- Click to add the current submitter as a staff member of the lab.
- Research personnel entered here will be available for selection when submitting research tests. These personnel will display on research tests but will not be displayed on the public page of the lab or on clinical tests. If person is entered as staff member, do not resubmit as research personnel.
-

to delete

Submission ID: SUB180239

[Lab information](#)
[Personnel](#)
[Licensure and accreditations](#)
[Default parameters](#)
[Overview](#)

* Required field. * Completed field. Hover over to display help information.

Basic Information

* First name

Middle initial

* Last name

* Should this person display on the GTR public site?

Yes No

* Is this person the primary lab contact?

Yes No

* Is this person a lab director?

Yes No

Job title

Administrator

Academic degree

- MD
- PHD
- MS
- RN
- BS
- AA
- BA
- BASc
- BAdm
- BEng/BE
- BMedSc/BMedSci
- BPharm
- BS
- BSc
- BTech
- CNA
- CRNA
- CPNP

Professional Certifications

Please select a board first, then select a specialty and subspecialty.

Board Specialty Subspecialty [remove](#)

[Add another professional certification](#)

Professional credentials

- American Society for Clinical Pathology, DLM
- American Academy of Cosmetic Surgery, FACRM
- American Academy of Dermatology, FAAD
- American Academy of Family Physicians, FAAFP
- American Academy of Neurology, FAAN
- American Academy of Ophthalmology, FAAO
- American Academy of Orthopaedic Surgeons, FAAOS
- American Academy of Otolaryngology-Head and Neck Surgery, FAAOS
- American Academy of Pediatrics, FAAP
- American Board of Genetic Counseling, CGC
- American College of Asthma, Allergy & Immunology, FACA
- American College of Emergency Physicians, FACEP
- American College of Medical Genetics, FACMG
- American College of Nuclear Medicine, FACNM
- American College of Physicians, FACP
- American College of Rheumatology, FACRM

Contact information to be displayed on GTR public site

Phone number: XXX-XXX-XXXX (U.S.A.), +(country code)-AreaCode-XXXXXX ext XXXX (International)

Email (ex. person@lab.com)

Fax number: XXX-XXX-XXXX (U.S.A.), +(country code)-AreaCode-XXXXXX ext XXXX (International)

Supplementary public contact information

Contact information for GTR staff to contact you about your submission

[copy contact information from above](#)

Phone number: XXX-XXX-XXXX (U.S.A.), +(country code)-AreaCode-XXXXXX ext XXXX (International)

Email (ex. person@lab.com)

Fax number: XXX-XXX-XXXX (U.S.A.), +(country code)-AreaCode-XXXXXX ext XXXX (International)


to delete

Submission ID: SUB180239

[Lab information](#) [Personnel](#) [Licensure and accreditations](#) [Default parameters](#) [Overview](#)

Hover over  to display help information.

Laboratory CLIA Certification

Certification # (e.g. 12D1234567) 	Exp. Date (YYYY-MM-DD) 	 remove
<input type="text"/>	<input type="text"/>	

 Add another CLIA certification

Laboratory State License(s)

License name 	License # 	Exp. Date (YYYY-MM-DD) 	 remove
<input type="text"/>	<input type="text"/>	<input type="text"/>	

 Add another state license

Other Certification(s)/License(s) that the Lab Holds

Name of certification/licensing body	License #	Exp. Date (YYYY-MM-DD)	 remove
<input type="text"/>	<input type="text"/>	<input type="text"/>	

 Add another certification/license

Save & Continue

to delete

Submission ID: SUB180239

- [Lab information](#)
[Personnel](#)
[Licensure and accreditations](#)
[Default parameters](#)
[Overview](#)

Hover over to display help information.

In this page you can enter information that is common to many of the tests you will submit. This information will pre-populate the corresponding fields on each test so you do not need to enter the same information multiple times. When you see this information on the test submission page, you can edit it as necessary.

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

Test contact policy

- Laboratory can only accept contact from health care providers. Patients/families are encouraged to discuss genetic testing options with their health care provider.
- Post-test email/phone consultation regarding genetic test results and interpretation is provided to patients/families.
- Pre-test email/phone consultation regarding genetic test results and interpretation is provided to patients/families.

Who can order this test?

- Genetic Counselor
- Health Care Provider
- In-State Patients
- Licensed Dentist

How to order (provide a brief explanation about ordering requirements)

URL to lab website with information about how to order this test

Test-specific laboratory services

<input type="text"/>	Order code	Comment	
----------------------	------------	---------	--

Add another test-specific laboratory service

Test-specific laboratory additional services

<input type="text"/>	Order code	
----------------------	------------	--

Add another test-specific laboratory additional service

Specimen source(s) (select all that apply)

- Amniocytes
- Amniotic fluid
- Bone marrow
- Buccal swab
- Cell culture
- Cell-free DNA
- Cerebrospinal fluid
- Chorionic villi
- Cord blood
- Cystic hygroma fluid
- Dried blood spot (DBS) card
- Fetal blood
- Fibroblasts
- Fresh tissue
- Frozen tissue
- Isolated DNA
- Paraffin block
- Peripheral (whole) blood
- Plasma
- Product of conception (POC)
- Saliva
- Serum
- Skin
- Sputum
- Urine
- White blood cell prep
- Other

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?

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

- Yes
 No
 Decline to answer
 Not provided

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

- Yes
 No
 Decline to answer
 Not provided

Comments about the laboratory procedure to re-contact the ordering physician

Upload Sample Reports

Sample negative report

No file selected.

Sample positive report

No file selected.

Sample VUS report

No file selected.

Generic Genetic Laboratory

Submission ID: SUB180239

[Lab information](#)
[Personnel](#)
[Licensure and accreditations](#)
[Default parameters](#)
[Overview](#)

This lab is ready for submission. [Preview how your lab will display](#) [Submit it](#) [Return to homepage](#)

Status: Not submitted, **Last modified:** 16:42.

Lab information

Name	Generic Genetic Laboratory Acronym: GGL
Institution	
Address	1000 Smith Ave Bethesda Maryland 20894
Phone	800-800-8000
Email	info@lab.com
Website	http://www.genericgenetics.com/
Types of service	Custom Sequence Analysis order code: 1234
Affiliations	

Personnel

Brandi Kattman	<p>Lab staff Display this person's information on the GTR public site: yes Primary lab contact Lab director Professional certifications: Contact information to be displayed on GTR public site: Phone number: 800-800-8888 Email: brandi@lab.com Contact information for GTR staff to contact you about the submission: Phone number: 800-800-8888 Email: brandi@lab.com</p>
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Licensure and accreditations

CLIA certification	CLIA: 23D0123456 exp: 2014-10-31
State license(s)	MD - Maryland Department of Health and Mental Hygiene DHMH: 1234 exp: 2014-10-31
Other certification(s)	

Default parameters

Test contact policy	Laboratory can only accept contact from health care providers. Patients/families are encouraged to discuss genetic testing options with their health care provider.
Who can order test	Health Care Provider
Test-specific services	
Specimen source(s)	Buccal swab Peripheral (whole) blood

Tests

This lab is ready for submission. [Preview how your lab will display](#) [Submit it](#) [Return to homepage](#)

Status: Not submitted, **Last modified:** 16:42.

LIST OF TESTS



GTR Submission

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[Migrate data from GeneTests at NCBI](#)

OMB NO: 0925-0651
EXPIRATION DATE:
[Burden statement](#)

[Claritas Genomics](#)

[Review lab submission](#)

Submission Status

Lab ID: 1179
Status: processed-ok
Last modified: 2014-10-27

Click here for the public display of this lab. Please note: It takes 24-48 hours to process a submission. If the lab record was submitted or edited in the last 48 hours, the public site may not display your currently submitted data.

If you want to delete this laboratory from the GTR, please [contact GTR staff](#).

Lab General Information

Lab name: **Claritas Genomics**
Address: 99 Erie St
Cambridge Massachusetts 02139
Email: clientservices@claritasgenomics.com
Website: <http://www.claritasgenomics.com/>
Phone: 617-553-5880
Fax: 617-553-5842

Annual Review

Last review performed: 2013-10-31
Next review due: 2014-10-31

[Perform Annual Review](#) [read more](#)

Completion resets the next due date one year forward.

Lab Director(s)

Mark Kellogg, PhD

Lab Credentials

CLIA: 22D0950490 exp: 2015-05-23

Submission of Tests

Clinical test: [Add a new clinical test](#)
[Add tests by spreadsheet](#)
for complex clinical tests or many tests
Research test: [Add a new research test](#)

Tests in this lab(6)

Test name	Test type	Submission status	Action	Use to create a new test	Link to public site
CHD7 Sequencing	Clinical test	Unfinished	Continue editing		ID: 334060
New test	Clinical test	Unfinished	Continue editing		ID: 330317
New test	Clinical test	Unfinished	Continue editing		ID: 334061
New test	Clinical test	Unfinished	Continue editing		ID: 5326
New test	Clinical test	Unfinished	Continue editing		ID: 270348
New test	Clinical test	Unfinished	Continue editing		ID: 304229

Want to submit another lab to GTR? Please check first if the lab can be [migrated from GeneTests](#). Migrating it will avoid duplicated display of your lab in GTR. If the lab is NOT in GeneTests, [click here to submit it to GTR](#).

ADDING A CLINICAL TEST

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GTR Submission — Generic Genetics

[Contact GTR Staff](#) [Help Documents](#) [GTR Homepage](#) [Brandi Kattman](#) [Log out](#)

New

Submission ID: SUB180908

[Basics](#) [Ordering](#) [Indication](#) [Methodology](#) [Interpretation](#) [Performance characteristics](#) [Overview](#)

* Required field. * Completed field. Hover over ? to display help information.

Test Information

* This test is for ?

Clinical

* Laboratory test name ?

Short test name ?

Manufacturer's test name, if any ?

Search terms, if any ?

For definitions of the terms in the list below, please go to [Help Documents](#)

* Purpose of the test ?

- Diagnosis
- Drug Response
- Monitoring
- Mutation Confirmation
- Pre-implantation genetic diagnosis
- Pre-symptomatic
- Risk Assessment
- Screening
- Predictive
- Prognostic
- Recurrence
- Therapeutic management

Target population for this test ?

Quotations for target population ?

Test development ?

Has there been FDA review of the test? ?

Yes No

FDA category designation ?

New York State CLEP (NYS CLEP) certification

Status ?

License # ?

Exp. Date (YYYY-MM-DD) ?

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Last Revision: 8bf4bde926c828d760b01f2265ef1dbb7d50baa Thu Oct 23 11:50:49 2014 -0400

New Clinical Test

Submission ID: SUB180908

[Basics](#) [Ordering](#) [Indication](#) [Methodology](#) [Interpretation](#) [Performance characteristics](#) [Overview](#)

Hover over  to display help information.

Ordering Information

Test order code (lab code to order this test, ex. for requisition form) 

URL of the lab website with information about this test 

URL of the lab website with information about how to order this test 

How to order (provide a brief explanation about ordering requirements) 

URL to lab website with information about codes related to this test (ex. CPT, ICD9, ICD10) 

LOINC code(s) 

 [search](#)

Who can order this test? 

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

Ordering requirements
Indicate whether the laboratory requires an informed consent form to be signed and/or proof of pre-test genetic counseling before performing this test. Indicate whether the laboratory requires proof of post-test genetic counseling for release of test results.

	Decline to Answer	Required	Not required	Based on applicable state law
Informed consent	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Pre-test genetic counseling	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Post-test genetic counseling	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

Test-specific laboratory services 

	Order code 	Comment	
<input type="text"/>	<input type="text"/>	<input type="text"/>	

[Add another test-specific laboratory service](#)

Test-specific laboratory additional services 

	Order code	Comment	
<input type="text"/>	<input type="text"/>	<input type="text"/>	

[Add another test-specific laboratory additional service](#)

Specimen source(s) (select all that apply) 

- Amniocytes
- Amniotic fluid
- Bone marrow
- Buccal swab
- Cell culture
- Cell-free DNA
- Cerebrospinal fluid
- Chorionic villi
- Cord blood
- Cystic hygroma fluid
- Dried blood spot (DBS) card
- Fetal blood
- Fibroblasts
- Fresh tissue
- Frozen tissue
- Isolated DNA
- Paraffin block
- Peripheral (whole) blood
- Plasma
- Product of conception (POC)
- Saliva
- Serum
- Skin
- Sputum
- Urine
- White blood cell prep
- Other

Specimen requirement URL (ex. collection, handling, transportation)


Testing strategy (ex. reflex testing) 

Citations for testing strategy 

[Add another citation \(search PubMed\)](#)

Test-specific contact information

Contact person, please select one or more persons from this list 

Contact policy 

- Laboratory can only accept contact from health care providers. Patients/families are encouraged to discuss genetic testing options with their health care provider.
- Post-test email/phone consultation regarding genetic test results and interpretation is provided to patients/families.
- Pre-test email/phone consultation regarding genetic test results and interpretation is provided to patients/families.

AMA CPT code(s)

[Add AMA CPT code](#)

[Save & Continue](#)

Search AMA CPT code

Search

Show all

Codes found:

Select

Cancel

New Clinical Test

Submission ID: SUB180908

[Basics](#)
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This test has the following conditions/phenotypes

There is no indication information in this test. Please add an indication.

[Condition/Phenotype](#)
[Primary](#)
[Actions](#)

Add other conditions/phenotypes

Limit to: pharma/cogenetic response conditions

HINT: Type the generic drug name, 'response' and/or 'hypersensitivity'

* Type Condition/Phenotype to search

Select conditions/phenotypes included in this test:


- Bronchiectasis
- Cystic fibrosis with helicobacter pylori gastritis, megaloblastic anemia and subnormal mentality
- Ashkenazi Jewish disorders
 - Spongy degeneration of central nervous system
 - Niemann-Pick disease, type A
 - Familial dysautonomia
 - Bloom syndrome
 - Fanconi anemia, complementation group C
 - Ganglioside sialidase deficiency
 - Cystic fibrosis
 - Tay-Sachs disease
 - Hexosaminidase A deficiency, adult type
 - Juvenile (Subacute) Hexosaminidase A Deficiency
 - Torsion dystonia
 - Gaucher's disease, type 1

Do you want to add a novel condition not in GTR?
 to the methodology section

New Clinical Test

Submission ID: SUB180908

[Basics](#) [Ordering](#) [Indication](#) [Methodology](#) [Interpretation](#) [Performance characteristics](#) [Overview](#)

* Required field. * Completed field. Hover over  to display help information.

Condition/Phenotype Information

* Condition/Phenotype name, please select from the autocomplete list 

Indication type

Condition/Phenotype name to be used for display in the GTR test page, if different from above 

Suggest new synonyms

[Add another synonym](#) 


Acronym to be used for display in the GTR test page, if different from above 

Suggest new acronyms 


[Add another acronym](#) 


Mode of inheritance 

Disease mechanism

Prevalence 

URL for prevalence

Citations for prevalence 

[Add another citation for prevalence \(search PubMed\)](#) 

Private comment about the condition/phenotype to GTR staff 


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* Required field. * Completed field. Hover over  to display help information.

Test Method(s)


Method # 1 


- * Major method category
Molecular Genetics
- * Method category
Sequence analysis of the entire coding region
- * Primary Test Methodology
Bi-directional Sanger Sequence Analysis
- Instruments
Affymetrix GeneChip Scanner 3000 7G Whole-Genome Association Sys=
Affymetrix GeneTitan® MC
Affymetrix HotStart-IT Probe qPCR Master Mix with UDG (2x)
Agilent 2100 Bioanalyzer
Agilent SureSelect
Applied Biosystems 3730 capillary sequencing instrument
Applied Biosystems 7900HT Sequence Detection System
Applied Biosystems SOLID v4 System Sequencer


 add another method

Platforms 

- Affymetrix CytoScan HD Array
- Affymetrix Gene Profiling Array cGMP U133 P2
- Affymetrix GeneChip Human Genome U133 Plus 2.0 Array
- Affymetrix GeneChip Human Mitochondrial Resequencing Array 2.0
- Affymetrix Genome-Wide Human SNP Array 6.0
- Affymetrix QuantGene 2.0 Assay
- Agilent Human CpG Island Microarray Kit, 1x244K
- Agilent Human ENCODE ChIP-on-chip Microarray
- Agilent Human miRNA Microarray Kit Release 16.0, 8x60K


Test procedure or protocol 


Citations for test procedure or protocol 

 Add another citation for test procedure or protocol (search PubMed)

Confirmation of test results (ex. how does the lab confirm positive results; using new sample/different method) 

You may store or edit a comment here to describe the test, but to maximize connectivity with other databases we strongly recommend you provide test target data in the test targets section below. Example: the comment 'Bi-directional sequencing of exons 1-5 with concurrent analysis of NP_000000.0:p.Glu234Gly' can be entered as exons 1-5 as one test target, and NP_000000.0:p.Glu234Gly as another. See the details [\[tag\]](#).

Test comment(s) (ex. is there additional information users should know about this test) 

 Add another test comment.

* Test Targets

Please add a test target to continue.

Some condition(s) are not connected to a test target. Please add a target for each condition.

[Add Test Target](#)

New Clinical Test

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[Interpretation](#)
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[Overview](#)

* Required field. * Completed field. Hover over  to display help information.

* Target is

- Germline: select for hereditary conditions
- Somatic: select for cancer management tests and monitoring of non-hereditary disease such as transplantation rejection
- Both

Target is associated with

- Cystic fibrosis

* Target is identified by 

gene 





Gene 

CFTR: cystic fibrosis transmembrane conductance regulator (ATP-binding) 

Additional information

- Associated Reference Sequences and Exons
- Variants
- None

Associated Reference Sequences and Exons

Associated Reference Sequence 	Relevant exons for each associated reference sequence 	
<input type="text"/>	<input type="text"/>	 remove
 Add another associated reference sequence		


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Hover over  to display help information.

Upload Sample Reports


Sample negative report 
 No file selected.


Sample positive report 
 No file selected.


Sample VUS report 
 No file selected.

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? 

Are family members with defined clinical status recruited to assess significance of VUS without charge? 
 Yes No Decline to answer Not provided

Comments about recruiting family members to assess significance of VUS without charge

Will the lab re-contact the ordering physician if variant interpretation changes? 
 Yes No Decline to answer Not provided

Comments about the laboratory procedure to re-contact the ordering physician 

Research performed after clinical testing is complete 

New Clinical Test

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* Required field. * Completed field. Hover over to display help information.

Availability

* Test performance location(s)

Identify where all components of the test are performed. Tests which are performed entirely out-of-house should not be registered. 'In-house' means within the lab/facility covered by the same CLIA certification number. Use text box to briefly describe components performed at an outside facility e.g., Specimen preparation - DNA isolation done in [location]. For Wet lab work done at an outside facility, briefly describe methodology and location performed.

Test work	In-house	At an outside lab
Entire test	<input checked="" type="checkbox"/>	<input type="checkbox"/>
Specimen preparation	<input type="checkbox"/>	<input type="checkbox"/>
Wet lab work	<input type="checkbox"/>	<input type="checkbox"/>
Interpretation	<input type="checkbox"/>	<input type="checkbox"/>
Generate report	<input type="checkbox"/>	<input type="checkbox"/>

Comment on test performance location(s)

* Analytical validity

99% sensitivity and specificity.

Citations to support analytical validity

Add another citation to support analytical validity (search PubMed)

Assay limitation(s)

Citations to support assay limitation

Add another citation to support assay limitation(s) (search PubMed)

Quality Assurance

Is proficiency testing performed for this test?

Yes No

Proficiency testing method

Provider for proficiency testing

Major CAP category

CAP category

CAP test list

add another CAP test

Description of proficiency testing method

Citations to support the above statement

Add another citation to support proficiency testing method (search PubMed)

Description of internal test validation method

Citations to support the above statement

Add another citation to support internal test validation method (search PubMed)

Clinical Validity

Statement of clinical validity

Citations to support the above statement

Add another citation to support clinical validity (search PubMed)

Clinical Utility

How likely the test is to improve patient outcomes significantly. Provide references to recommendations or practice guidelines that have been issued by authoritative groups. If none available, labs may display a statement explaining that sufficient research has not been conducted to demonstrate the utility of the test.

Clinical utility # 1

Category of clinical utility

URL to explain the clinical utility

Citations to support the clinical utility

Add another citation to support the clinical utility (search PubMed)

add another clinical utility

Save & Continue

New Clinical Test

Submission ID: SUB180908

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Preview how your test will display

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Basics

This test is for
Name Clinical
Name New Clinical Test
Test purpose Diagnosis
Test-specific licenses License#:

Ordering

Informed consent required Decline to answer
Pre-test generic counseling required Decline to answer
Post-test generic counseling required Decline to answer
Test-specific services
Test-specific additional services
Contact person Brandt Kattman

Condition/Phenotype: Cystic fibrosis

Mode of inheritance Autosomal recessive inheritance
Disease mechanism loss of function

Methodology

Test method(s) Molecular Genetics, Sequence analysis of the entire coding region, BI-directional Sanger Sequence Analysis

Test target(s)

Target is germline
Identified by gene: CFTR: cystic fibrosis transmembrane conductance regulator (ATP-binding cassette sub-family C, member 7)
Reference Sequence(s)
Variant(s)

Interpretation

Are family members recruited to assess significance of VUS not provided
Will the lab re-contact the ordering physician if variant interpretation changes not provided

Performance characteristics

Test performance location(s) Entire test: internal
Analytical validity 99% sensitivity and specificity.
Proficiency testing is performed for this test? yes

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ADDING A RESEARCH TEST

NCBI

GTR Submission — Generic Genetics

[Contact GTR Staff](#) [Help Documents](#) [GTR Homepage](#) [Brandi Kattman](#) [Log out](#)

New











Submission ID: SUB180909

[Basics](#) [Participation](#) [Condition](#) [Methodology](#) [Overview](#)





* Required field. * Completed field. Hover over  to display help information.

* Laboratory test name 	Short test name 
<input type="text" value="New Research Test"/>	<input type="text"/>
* What is the purpose of the research test? 	
<input checked="" type="radio"/> Contribute to generalizable knowledge	
<input type="radio"/> For the laboratory to generate data in order to make technical improvements to a test	

Study related to this test

Research study name 	Short study name 
<input type="text"/>	<input type="text"/>
ClinicalTrials.gov identifier 	<input type="text"/>
URL for the study 	<input type="text"/>
If the study is approved by a research ethics committee (e.g., IRB), please provide the protocol number 	
<input type="text"/>	
What is the study type? 	
<input type="radio"/> Interventional study (or Clinical Trial)	
<input type="radio"/> Observational study	
<input type="radio"/> Expanded access	
<input type="radio"/> Not applicable	
* Study description 	
<input type="text"/>	
Citations for study description 	
<input type="text"/>	
Add another citation for study description (search PubMed)	
Study aims and hypotheses 	
<input type="text"/>	
Upload study protocol 	
<input type="button" value="Browse..."/> No file selected.	

Researchers

* Person responsible for the study 
<input type="text"/>
* Study contact 
<input type="text"/>
Co-Investigator 
<input type="text"/>
add another co-Investigator <input type="button" value="remove"/>
Research contact policy 
<input type="text"/>


[Save & Continue](#)

New Research Test


Submission ID: SUB180909

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* Required field. * Completed field. Hover over  to display help information.


Upload participant consent form 

No file selected.

Is the study currently recruiting participants? 

Yes
 No
 Unknown

Eligibility criteria 

Recruitment sites 

New Research Test

Submission ID: SUB180909

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There is no indication information in this test. Please add an indication.

Add other conditions/phenotypes

Limit to: pharma/genetic response conditions
 HINT: Type the generic drug name, 'response' and/or 'hypersensitivity'

* Type Condition/Phenotype to search

CF: Cystic fibrosis

Select conditions/phenotypes included in this test:

- Ashkenazi Jewish disorders
 - Spongy degeneration of central nervous system
 - Niemann-Pick disease, type A
 - Familial dysautonomia
 - Bloom syndrome
 - Fanconi anemia, complementation group C
 - Ganglioside sialidase deficiency
 - Cystic fibrosis
 - Tay-Sachs disease
 - Hexosaminidase A deficiency, adult type
 - Juvenile (Subacute) Hexosaminidase A Deficiency
 - Torsion dystonia
 - Gaucher's disease, type 1


Do you want to add a novel condition not in GTR?

to the methodology section

New Research Test

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* Condition/Phenotype name, please select from the autocomplete list 

Indication type


Condition/Phenotype name to be used for display in the GTR test page, if different from above 


Suggest new synonyms

 Add another synonym 


Acronym to be used for display in the GTR test page, if different from above 

Suggest new acronyms 

 Add another acronym 

Mode of inheritance 

Disease mechanism

Prevalence 

URL for prevalence

Citations for prevalence 

 Add another citation for prevalence (search PubMed) 

Private comment about the condition/phenotype to GTR staff 

New Research Test

Submission ID: SUB180909

[Basics](#) [Participation](#) [Condition](#) [Methodology](#) [Overview](#)

* Required field. * Completed field. Hover over to display help information.

Test Method(s)

Method # 1 remove

- * Major method category: Molecular Genetics
- * Method category: Sequence analysis of the entire coding region
- * Primary Test Methodology: Bi-directional Sanger Sequence Analysis
- Instruments:
 - Affymetrix GeneChip Scanner 3000 7G Whole-Genome Association Sys=
 - Affymetrix GeneTitan® MC
 - Affymetrix HotStart-IT Probe qPCR Master Mix with UDG (2x)
 - Agilent 2100 Bioanalyzer
 - Agilent SureSelect
 - Applied Biosystems 3730 capillary sequencing instrument
 - Applied Biosystems 7900HT Sequence Detection System
 - Applied Biosystems SOLID v4 System Sequencer

[add another method](#)

Platforms

- Affymetrix CytoScan HD Array
- Affymetrix Gene Profiling Array cGMP U133 P2
- Affymetrix GeneChip Human Genome U133 Plus 2.0 Array
- Affymetrix GeneChip Human Mitochondrial Resequencing Array 2.0
- Affymetrix Genome-Wide Human SNP Array 6.0
- Affymetrix QuantGene 2.0 Assay
- Agilent Human CpG Island Microarray Kit, 1x244K
- Agilent Human ENCODE ChIP-on-chip Microarray
- Agilent Human miRNA Microarray Kit Release 16.0, 8x60K

Test procedure or protocol

Citations for test procedure or protocol

[Add another citation for test procedure or protocol \(search PubMed\)](#)

Confirmation of test results (ex. how does the lab confirm positive results; using new sample/different method)

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Test comment(s) (ex. is there additional information users should know about this test)

[Add another test comment](#)

*** Test Targets**

Please add a test target to continue.

Some condition(s) are not connected to a test target. Please add a target for each condition.

[Add Test Target](#)



New Research Test

Submission ID: SUB180909

[Basics](#) [Participation](#) [Condition](#) [Methodology](#) [Overview](#)

* Required field. * Completed field. Hover over to display help information.

*** Target is**

- Germline: select for hereditary conditions
- Somatic: select for cancer management tests and monitoring of non-hereditary disease such as transplantation rejection
- Both

Target is associated with

- Cystic fibrosis **(Needs to be linked to a test target)**

* Target is identified by

chromosomal region/mitocho

Chromosome, mitochondrion, or cytogenetic band

complete genome

[Save Target](#) [Cancel](#)

New Research Test

Submission ID: SUB180909

[Basics](#) [Participation](#) [Condition](#) [Methodology](#) [Overview](#)[Preview how your test will display](#) [Return to homepage](#)

Basics

This test is for	Research
Name	New Research Test
Test purpose	Contribute to generalizable knowledge
Study name	
Study Description	To discover new genes associated with disease.
Person responsible for the study	Brandi Kattman
Study contact	Brandi Kattman

Contact policy

Participation

Condition/Phenotype: Cystic fibrosis

Mode of inheritance	Autosomal recessive inheritance
Disease mechanism	loss of function

Methodology

Test method(s)	Molecular Genetics, Sequence analysis of the entire coding region, BI-directional Sanger Sequence Analysis
-----------------------	--

Test target(s)

Target is	germline
Identified by	chromosomal region/mitochondrion: complete genome
Reference Sequence(s)	
Variant(s)	

[Preview how your test will display](#)[Return to homepage](#)