

# Information Collection Screenshots

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
## 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).

01/31/2018



# SECURE LOGIN SYSTEM

 U.S. National Library of Medicine  National Center for Biotechnology Information Sign in to NCBI

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## Submission Portal

### Welcome to the submission site for the NIH Genetic Testing Registry (GTR)!

To log in (or create a new account), please click the link "Sign in to NCBI" on the top right corner of this page. Make sure to always use the same log in account.

Here you will be able to register your laboratory and your clinical and research genetic tests. You can update your information at any time. The information entered here displays publicly at <https://www.ncbi.nlm.nih.gov/gtr/>.

The first time you log in you will see the GTR code of conduct and the AMA CPT code agreement before you reach your submission homepage.

To register your laboratory, click the button "Add a new lab". Once you submit your lab information, GTR staff will review it and contact you for more information.

When approved you will be able to register your clinical and research genetic tests manually by clicking the "Add a new clinical test" or "Add a new research test" or by using one of the two excel files available to register clinical tests in bulk. Please register your tests as represented in your lab's catalog. The more information you provide the more discoverable your test will be by GTR users.

Regardless of how many times you update your data, please submit your annual review once a year as this is a separate action. To submit your annual review, click the "Perform annual review" button to start it and the "Submit" button to finish and submit it.

There is a groups feature where multiple lab staff can work on the same lab and test records, please contact us at [gtr@ncbi.nlm.nih.gov](mailto:gtr@ncbi.nlm.nih.gov) if you would like others in your lab to work on your GTR records.










For more information on how to submit to GTR: <https://www.ncbi.nlm.nih.gov/gtr/docs/submit/>

Please contact us at [gtr@ncbi.nlm.nih.gov](mailto:gtr@ncbi.nlm.nih.gov) if you have questions or if you need any help.

Thank you for participating in GTR!

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	dbGap				

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[Write to the Help Desk](#)

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**NCBI**  
National Center for Biotechnology Information, U.S. National Library of Medicine  
8600 Rockville Pike, Bethesda, MD 20894, USA

Polices and Guidelines | Contact Last revision: 2.6.1 post30HF5d050

# BURDEN STATEMENT

## GTR: GENETIC TESTING REGISTRY

   [Advanced search for tests](#)

- Overview
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- Public Search Site
- Submission
- Lab Submission
- Clinical Test Submission
- Research Test Submission

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OMB NO: 0925-0651

EXPIRATION DATE: 07/31/2018

### Burden Statement:

Public reporting burden for this collection of information is estimated to vary from 18 minutes to 54 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

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- [PubMed Health](#)
- [GenBank](#)
- [Reference Sequences](#)
- [Gene Expression Omnibus](#)
- [Map Viewer](#)
- [Human Genome](#)
- [Mouse Genome](#)
- [Influenza Virus](#)
- [Primer-BLAST](#)
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Last updated: 2017-11-14T17:07:05Z

# GTR CODE OF CONDUCT

## GTR Submission

[Contact GTR Staff](#) [Help Documents](#) [GTR Homepage](#) [Adriana Malheiro](#) [Log out](#)

### Code of Conduct

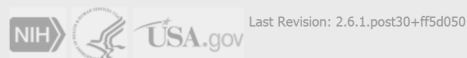
Test submitters providing test information to the Genetic Testing Registry (GTR) agree to abide by a code of conduct. Failure to honor this code of conduct may result in the removal of the submitter's test information from the GTR. Submitters agree to the following terms in the code of conduct:

- 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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# AMA CPT CODE LICENSE AGREEMENT

## GTR Submission

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### AMA CPT Code License Agreement

#### LICENSE FOR USE OF CURRENT PROCEDURAL TERMINOLOGY, FOURTH EDITION ("CPT®")

CPT only copyright 2012 American Medical Association. All rights reserved. CPT is a registered trademark of the American Medical Association.

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.

Registrant, Registrant's employees and agents are authorized to use CPT codes and descriptors only as contained in the Genetic Testing Registry solely for Registrant's own use for the sole purpose of identifying and adding the appropriate CPT code(s) to their registered tests. Registrant acknowledges that the American Medical Association (AMA) holds all copyright, trademark and other rights in CPT.

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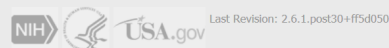
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# YOUR LABS IN GTR

## GTR Submission

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OMB NO: 0925-0651  
 EXPIRATION DATE: 07/31/2018  
[Burden statement](#)

### EGL Genetic Diagnostics

[Review lab submission](#)

#### Submission Status

Lab ID: 500060  
 Status: processed-ok  
 Last modified: 2018-01-17

[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: **EGL Genetic Diagnostics, Eurofins Clinical Diagnostics**  
 Address: 2460 Mountain Industrial Boulevard  
 Tucker Georgia 30084  
 Email: [egcs@eg-eurofins.com](mailto:egcs@eg-eurofins.com)  
 Website: <http://www.eg-eurofins.com/>  
 Phone: 470-378-2200  
 Fax: 470-378-2250

#### Annual Review

Last review performed: 2018-01-30  
 Next review due: 2019-01-30

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

Completion resets the next due date one year forward.

#### Lab Director(s)

Lora Bean, PhD  
 Patricia Hall, PhD  
 John Alexander, PhD  
 Hussain Askree, MD, PhD  
 Arunkanth Ankala, PhD

#### Lab Credentials

CLIA: 11D0683478 exp: 2018-03-15  
 MD - Maryland Department of Health and Mental Hygiene DHMH: 1346 exp: 2018-06-30  
 PA - Pennsylvania Department of Health PADOH: 031676 exp: 2018-08-15  
 FL - Florida Agency for Health Care Administration AHCA: B00026872 exp: 2019-03-30  
 GA - Georgia Department of Community Health DCH: 044-174 exp: 2018-02-28  
 NY - New York State Department of Health NYSDOH: 8951 exp: 2018-06-30  
 College of American Pathologists, CAP: 7181693 exp: 2018-03-31

#### 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(937)

test name	Search tests	Delete tests	Items 1 - 50 of 937	<< First	< Prev	Page 1 of 19	Next >	Last >>
Test name	Test type	Submission status	Action	Use to create a new test	Link to public site			
Iq21.1 Deletion/Duplication Analysis	Clinical test	Processed successfully	<a href="#">Update test</a>	<a href="#">Copy</a>	ID: GTR000508266			
3-Hydroxy-3-Methylglutaryl CoA Lyase Deficiency (HMG): HMGCL Full Gene Sequencing	Clinical test	Processed successfully	<a href="#">Update test</a>	<a href="#">Copy</a>	ID: GTR000502886			
3-Hydroxy-3-Methylglutaryl CoA Lyase Deficiency (HMG): HMGCL Gene Deletion/Duplication	Clinical test	Processed successfully	<a href="#">Update test</a>	<a href="#">Copy</a>	ID: GTR000502887			
3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC): MCCC1/MCCC2 Full Gene Sequencing	Clinical test	Processed successfully	<a href="#">Update test</a>	<a href="#">Copy</a>	ID: GTR000502913			
3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC): MCCC1/MCCC2 Gene Deletion/Duplication	Clinical test	Processed successfully	<a href="#">Update test</a>	<a href="#">Copy</a>	ID: GTR000502918			
Aarskog-Scott Syndrome: FGD1 Full Gene Sequencing	Clinical test	Processed successfully	<a href="#">Update test</a>	<a href="#">Copy</a>	ID: GTR000501493			
Aarskog-Scott Syndrome: FGD1 Gene Deletion/Duplication	Clinical test	Processed successfully	<a href="#">Update test</a>	<a href="#">Copy</a>	ID: GTR000501833			
ACAD9 Deficiency: ACAD9 Full Gene Sequencing	Clinical test	Processed successfully	<a href="#">Update test</a>	<a href="#">Copy</a>	ID: GTR000501494			
ACAD9 Deficiency: ACAD9 Gene Deletion/Duplication	Clinical test	Processed successfully	<a href="#">Update test</a>	<a href="#">Copy</a>	ID: GTR000502599			

# ADD A NEW LAB

NCBI


## GTR Submission

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### 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 
<input type="text"/>	<input type="text"/>
GeneTests at NCBI lab ID, if known 	
<input type="text"/>	
Name of institution 	Acronym of institution name 
<input type="text"/>	<input type="text"/>
Name of department 	
<input type="text"/>	


#### Laboratory Address

* Country or region 	
<input type="text" value="United States"/>	
Street & No 	
<input type="text"/>	
<input type="text"/>	
Additional address line 	
<input type="text"/>	
* City 	
<input type="text"/>	
State or province 	
<input type="text" value="Alabama"/>	
* Postal code 	
<input type="text"/>	
* Make this address public? 	
<input type="radio"/> Yes <input type="radio"/> No	
* Phone number: XXX-XXX-XXXX (U.S.A), +(country code)-AreaCode-XXXXXX ext XXXX (International) 	
<input type="text"/>	
Fax number: XXX-XXX-XXXX (U.S.A), +(country code)-AreaCode-XXXXXX ext XXXX (International) 	
<input type="text"/>	
* Email (ex. lab@lab.com) and/or  URL for lab contact form 	
<input type="text"/>	<input type="text"/>
Lab website URL 	
<input type="text"/>	


#### Laboratory Types of Service

Service 	Order code 	Comment 	
<input type="text"/>	<input type="text"/>	<input type="text"/>	remove
 Add another service			


#### Laboratory Affiliation(s)

Name of affiliate (example: clinic, research center) 	Website 	
<input type="text"/>	<input type="text"/>	remove
 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

[Save & Continue](#)

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	<a href="#">Delete</a> <a href="#">Edit</a>

- 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.

- 1 LAB INFORMATION
- 2 PERSONNEL
- 3 LICENSURE AND ACCREDITATIONS
- 4 DEFAULT PARAMETERS
- 5 OVERVIEW

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

Lab ID: 505467 **Status:** Not submitted, **Last modified:** 21:46.

**Lab information**

**Name** GTR Example Laboratory  
**Institution** NIH  
**Name of department** NCBI  
**Address** Bethesda Maryland 20892  
**Phone** 555-555-1234  
**Email** GTRlab@lab.com  
**Types of service**  
**Affiliations**

**Personnel**

**Adriana Malheiro** Lab staff  
 Display this person's information on the GTR public site: Yes  
 Is this person the primary lab contact? Yes  
 Is this person a lab director? Yes  
 Job title: Lab Director  
 Professional certifications:  
 Contact information to be displayed on GTR public site:  
 Email: person@GTRlab.com  
 Contact information for GTR staff to contact you about the submission:  
 Email: person@GTRlab.com

**Gregor Mendel** Research personnel  
 Academic degree: MD  
 Institution: University of Vienna, Vienna, Austria  
 Contact information:  
 Email: person@lab.com

**Licensure and accreditations**

**CLIA certification** CLIA : 12D1234567 exp: 2019-06-26  
**State license(s)** MD - Maryland Department of Health and Mental Hygiene DHMH: 45W9B11 exp: 2019-02-28  
**Other certification(s)** College of American Pathologists, CAP: 11Z521 exp: 2020-02-03

**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**

**Tests**

Test name	Test type	Submission status	Action	Use to create a new test	Link to public site
GTR Example Test	Clinical test	Processed successfully	<a href="#">Update test</a>	<a href="#">Copy</a>	ID: GTR000523335
GTR Research Test	Research test	Unfinished	<a href="#">Continue editing</a>		ID: GTR000500589
GTR Research Test #2	Research test	Processed successfully	<a href="#">Update test</a>	<a href="#">Copy</a>	ID: GTR000552655
New test	Clinical test	Unfinished	<a href="#">Continue editing</a>		

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

Lab ID: 505467 **Status:** Not submitted, **Last modified:** 21:46.

# LIST OF TESTS

## GTR Submission

OMB NO: 0925-0651  
 EXPIRATION DATE: 07/31/2018  
[Burden statement](#)

### Claritas Genomics

[Edit this lab](#)

#### Submission Status

Lab ID: 1179  
 Status: **Unfinished: at the Overview step**  
 Last modified: 2017-10-13

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

#### Annual Review

Last review performed: 2017-11-06  
 Next review due: 2018-11-06

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

*Completion resets the next due date one year forward.*

#### Lab General Information

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

#### Lab Director(s)

Hayk Hovhannisyian, PhD  
 Ali Hosseini, PhD, MD

#### Lab Credentials

CLIA: 22D0950490 exp: 2018-02-28  
 RI - State of Rhode Island Department of Health RIDOH: LC000843 exp: 2017-12-30  
 PA - Pennsylvania Department of Health PADOH: 33760 exp: 2018-08-15  
 MA - Executive Office of Health and Human Services EOHHS: 3338 exp: 2018-05-08  
 MD - Maryland Department of Health and Mental Hygiene DHMH: 2147 exp: 2019-06-30  
 FL - Florida Agency for Health Care Administration AHCA: 800027665 exp: 2019-05-12  
 CA - California Department of Public Health CDPH: COS 00800509 exp: 2018-07-16  
 Other: 3712.01 exp: 2017-12-31

#### 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(8)

test name

[Search tests](#)

[Delete tests](#)

Test name	Test type	Submission status	Action	Use to create a new test	Link to public site
15q13.2-q13.3 Deletory/Duplication	Clinical test	Processed successfully	<a href="#">Update test</a>	<a href="#">Copy</a>	ID: GTR000520904
Claritas Clinical Exome - Proband Only	Clinical test	Processed successfully	<a href="#">Update test</a>	<a href="#">Copy</a>	ID: GTR000531466
Claritas Clinical Exome Trio	Clinical test	Processed successfully	<a href="#">Update test</a>	<a href="#">Copy</a>	ID: GTR000531465
ClariView Array	Clinical test	Processed successfully	<a href="#">Update test</a>	<a href="#">Copy</a>	ID: GTR000531468
Nephrotic Syndrome Region of Interest- Proband Only	Clinical test	Processed successfully	<a href="#">Update test</a>	<a href="#">Copy</a>	ID: GTR000552239
Nephrotic Syndrome Region of Interest-Trio	Clinical test	Processed successfully	<a href="#">Update test</a>	<a href="#">Copy</a>	ID: GTR000531467
New test	Clinical test	Unfinished	<a href="#">Continue editing</a>		
Pediatric Neurology Region of Interest- Trio	Clinical test	Unfinished	<a href="#">Continue editing</a>		

If you have started submitting a lab in GTR but do not see it in this page, Please login with the account you used to submit the lab or contact your group administrator to give you permission to access your records.

[Submit a new lab](#)



# ADDING A CLINICAL TEST

NCBI

GTR Submission — Generic Genetics

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

New

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

Submission ID: SUB180908

\* 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 QLEP (NYS QLEP) certification

Status ?

License # ?

Exp. Date (YYYY-MM-DD) ?

[Copyright](#) | [Disclaimer](#) | [Privacy](#) | [Accessibility](#) | [Contact](#)  
National Center for Biotechnology Information | U.S. National Library of Medicine



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  [remove](#)

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

Test-specific laboratory additional services  Order code  Comment  [remove](#)


[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](#)
[Ordering](#)
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[Interpretation](#)
[Performance characteristics](#)
[Overview](#)

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

cystic fibrosis

[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

[Add selected conditions/phenotypes](#)

Do you want to add a novel condition not in GTR? [Add a novel condition](#)

[Continue](#) 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 

New Clinical Test

Submission ID: SUB180908

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

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

Submission ID: SUB180908

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

 remove

 Add another associated reference sequence

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




New Clinical Test


Submission ID: SUB180908


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

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 

[Save & Continue](#)

New Clinical Test

Submission ID: SUB180908

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

\* 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

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

[Preview how your test will display](#)

[Return to homepage](#)

**Basics**

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

**Ordering**

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

**Condition/Phenotype: Cystic fibrosis**

<b>Mode of inheritance</b>	Autosomal recessive inheritance
<b>Disease mechanism</b>	loss of function

**Methodology**

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

**Test target(s)**

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

**Interpretation**

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

**Performance characteristics**

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

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# ADDING A RESEARCH TEST

NCBI

GTR Submission — Generic Genetics

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New

Submission ID: SUB180909

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

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

* Laboratory test name ⓘ New Research Test	Short test name ⓘ 
* 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 ⓘ 
ClinicalTrials.gov identifier ⓘ 	
URL for the study ⓘ 	
If the study is approved by a research ethics committee (e.g., IRB), please provide the protocol number ⓘ 	
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 ⓘ 	
Citations for study description ⓘ <input type="text"/> ⓘ <a href="#">Add another citation for study description (search PubMed)</a>	
Study aims and hypotheses ⓘ 	
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"/> ⓘ <input type="button" value="remove"/>
<a href="#">add another co-Investigator</a>
Research contact policy ⓘ <input type="text"/>

New Research Test


Submission ID: SUB180909

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


\* 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

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

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

Submission ID: SUB180909

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

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)

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 [\[here\]](#).

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](#)