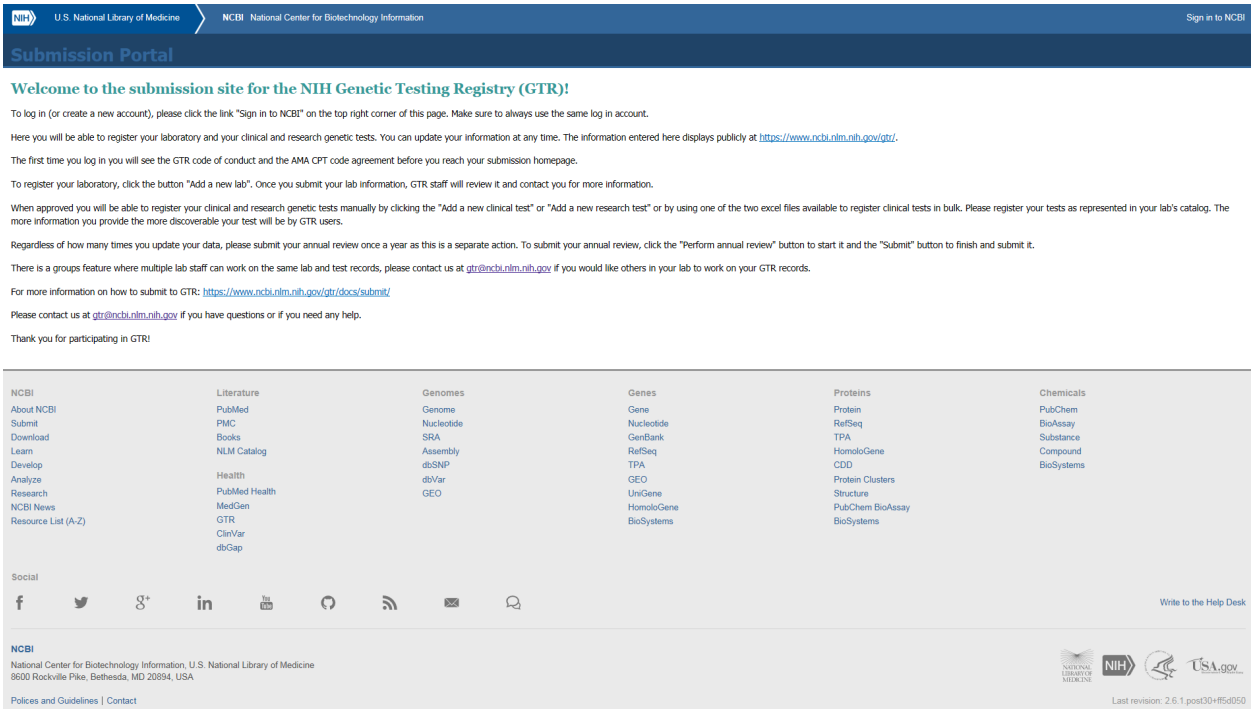
Information Collection Screenshots

Online submission form for the Genetic Testing Registry (GTR)

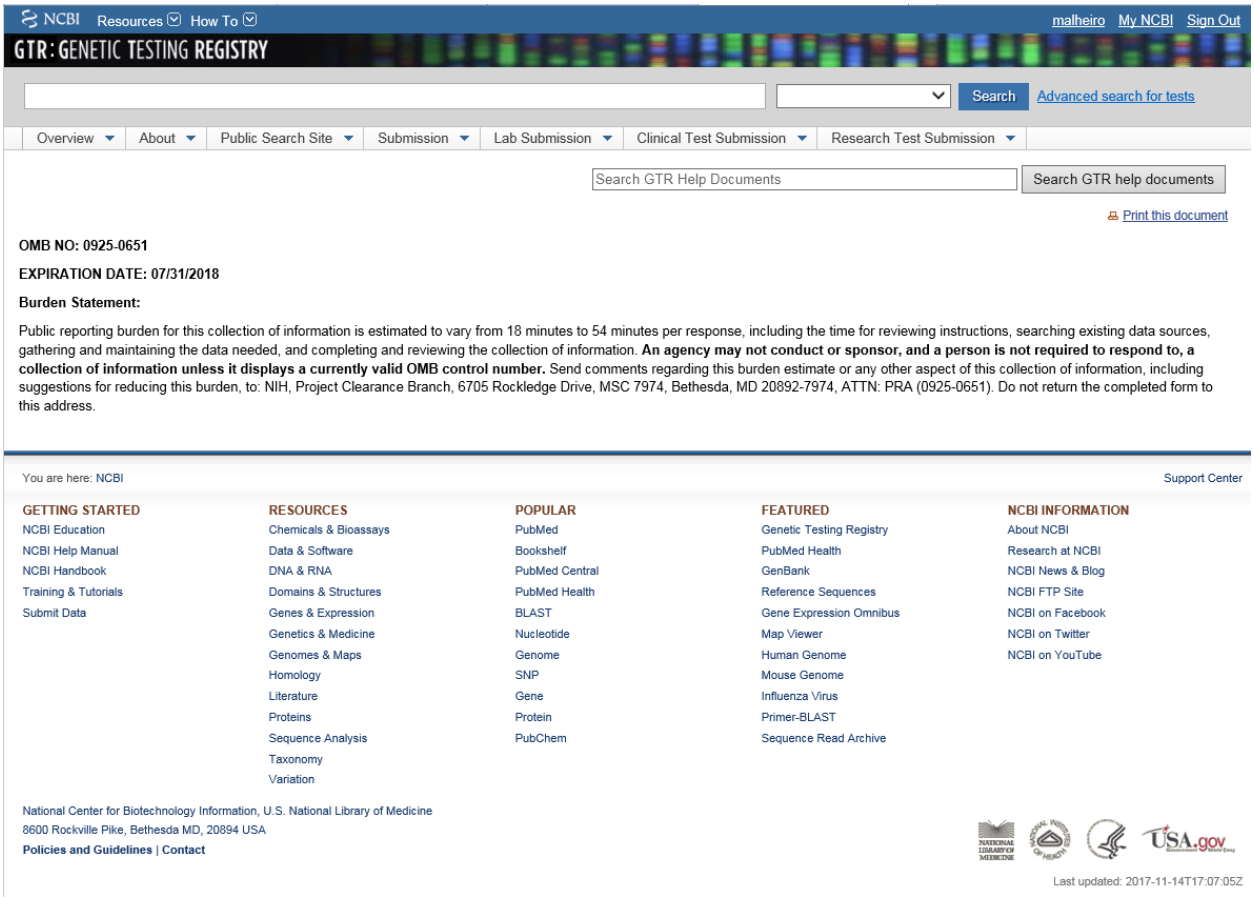
This document provides updated screenshots of the online form for the submission of genetic test information to the Genetic Testing Registry (GTR).

01/31/2018

SECURE LOGIN SYSTEM



BURDEN STATEMENT



GTR CODE OF CONDUCT

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.

I agree | Disagree

AMA CPT CODE LICENSE AGREEMENT

AMA CPT Code License Agreement

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

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 Associations (AMA) holds all copyright, trademark and other rights in CPT.

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This Agreement will terminate upon notice if Registrant violates its terms. The AMA is a third party beneficiary to this Agreement.

Should the foregoing terms and conditions be acceptable to Registrant, please indicate your agreement and acceptance by clicking below on the button labeled "accept".

Accept | Skip

YOUR LABS IN GTR

GTR Submission Contact GTR Staff | Help Documents | GTR Homepage Acting as Lindsey Mighion | Switch back to pda|malheiro | Log out

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.

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 General Information

Lab name: **EGL Genetic Diagnostics, Eurofins Clinical Diagnostics**
Address: 2460 Mountain Industrial Boulevard
Tucker Georgia 30084
Email: eglcs@egl-eurofins.com
Website: http://www.egl-eurofins.com/
Phone: 470-378-2200
Fax: 470-378-2250

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: 800026872 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

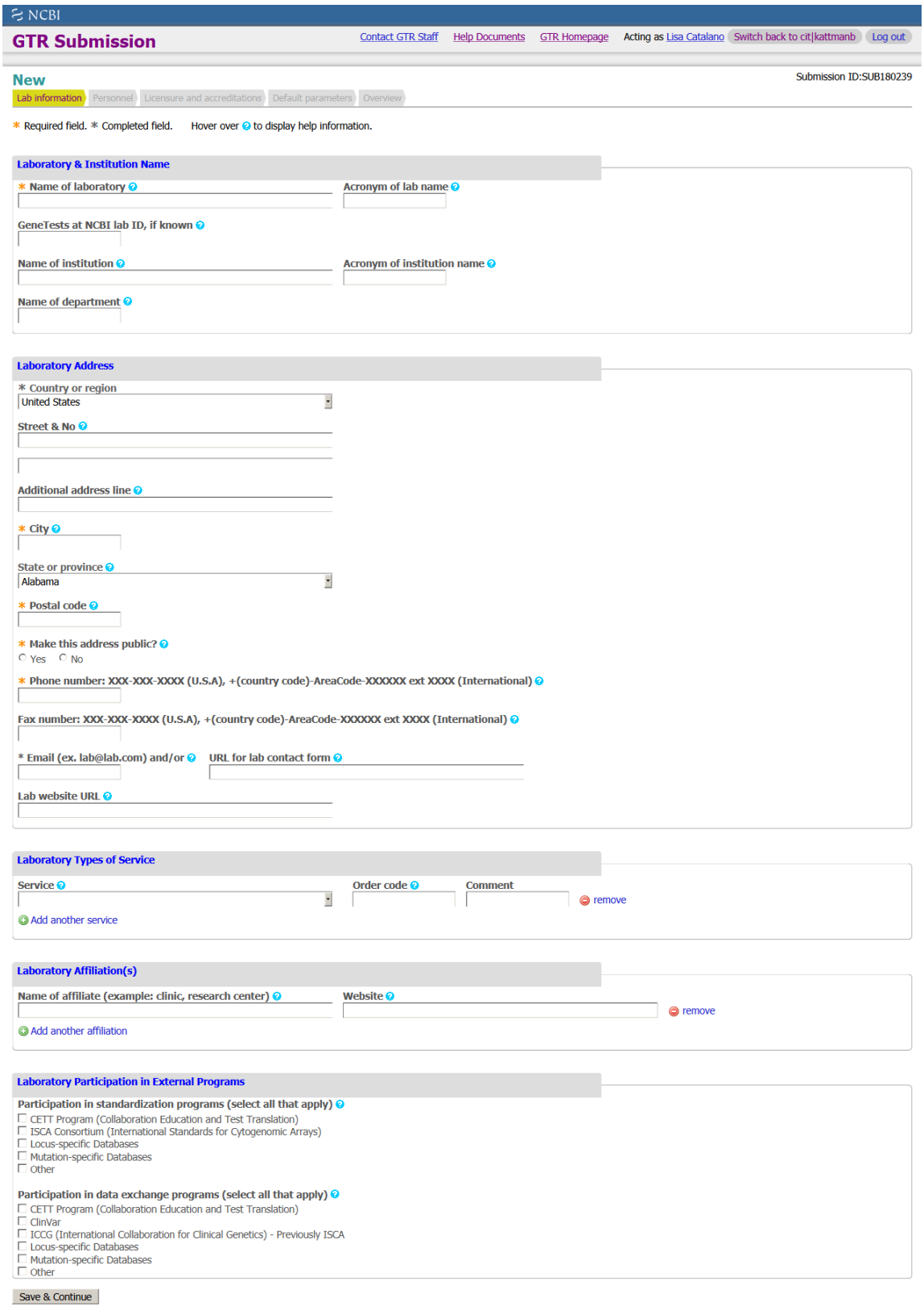
Tests in this lab(937)

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
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
Aarskog-Scott Syndrome: FGD1 Gene Deletion/Duplication	Clinical test	Processed successfully	Update test	Copy	ID: GTR000501833
ACAD9 Deficiency: ACAD9 Full Gene Sequencing	Clinical test	Processed successfully	Update test	Copy	ID: GTR000501494
ACAD9 Deficiency: ACAD9 Gene Deletion/Duplication	Clinical test	Processed successfully	Update test	Copy	ID: GTR000502599

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
* Make this address public?
Yes
No
* 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
remove
Add another service
Laboratory Affiliation(s)
Name of affiliate (example: clinic, research center)
Website
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

NCBI

GTR Submission

Contact GTR Staff | Help Documents | GTR Homepage | Acting as Lisa Catalano | Switch back to cit|kattmanb | Log out

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

Add a person — Please add all staff members relevant to your lab registration. Personel 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.

Add me — Click to add the current submitter as a staff member of the lab.

Add research personnel — 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.

Continue

NCBI

GTR Submission

Contact GTR Staff | Help Documents | GTR Homepage | Acting as Lisa Catalano | Switch back to cit|kattmanb | Log out

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
CN.A
CRNA

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, FACAI
American College of Emergency Physicians, FACEP
American College of Medical Genetics, FACMG
American College of Nuclear Medicine, FACNM
American College of Physicians, FACP

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)

Save Cancel

NCBI

GTR Submission

Contact GTR Staff | Help Documents | GTR Homepage | Acting as Lisa Catalano | Switch back to cit|kattmanb | Log out

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

Add another CLIA certification

Laboratory State License(s)

License name ? | License # ? | Exp. Date (YYYY-MM-DD) ?

remove

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

Add another certification/license

Save & Continue

Last Revision: ce809503f70fb5004744d3cc819f85fe9b1fa7bf Thu Oct 23 14:39:22 2014 -0400

NCBI

GTR Submission

to delete

Submission ID:SUB180239

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 ? | **Order code** ? | **Comment** | remove

Add another test-specific laboratory service

Test-specific laboratory additional services ? | **Order code** | 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

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 ?
Browse... No file selected.

Sample positive report ?
Browse... No file selected.

Sample VUS report ?
Browse... No file selected.

Save & Continue

GTR Submission

GTR Example Laboratory

Submission ID:SUB128509

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 | Submit it | 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: 45W9811 exp: 2019-02-28
Other certification(s)	College of American Pathologists, CAP: 112521 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 | Search 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	Update test	Copy	ID: GTR000523335
GTR Research Test	Research test	Unfinished	Continue editing		ID: GTR000500589
GTR Research Test #2	Research test	Processed successfully	Update test	Copy	ID: GTR000552655
New test	Clinical test	Unfinished	Continue editing		

This lab is ready for submission. Preview how your lab will display | Submit it | Return to homepage

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

LIST OF TESTS

GTR Submission Contact GTR Staff | Help Documents | GTR Homepage Acting as pamela gerrol | Switch back to pda|malheiro | Log out

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: clientservices@claritasgenomics.com
Website: http://www.claritasgenomics.com/
Phone: 617-553-5880
Fax: 617-553-5842

Lab Director(s)

Hayk Hovhannisyan, 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)

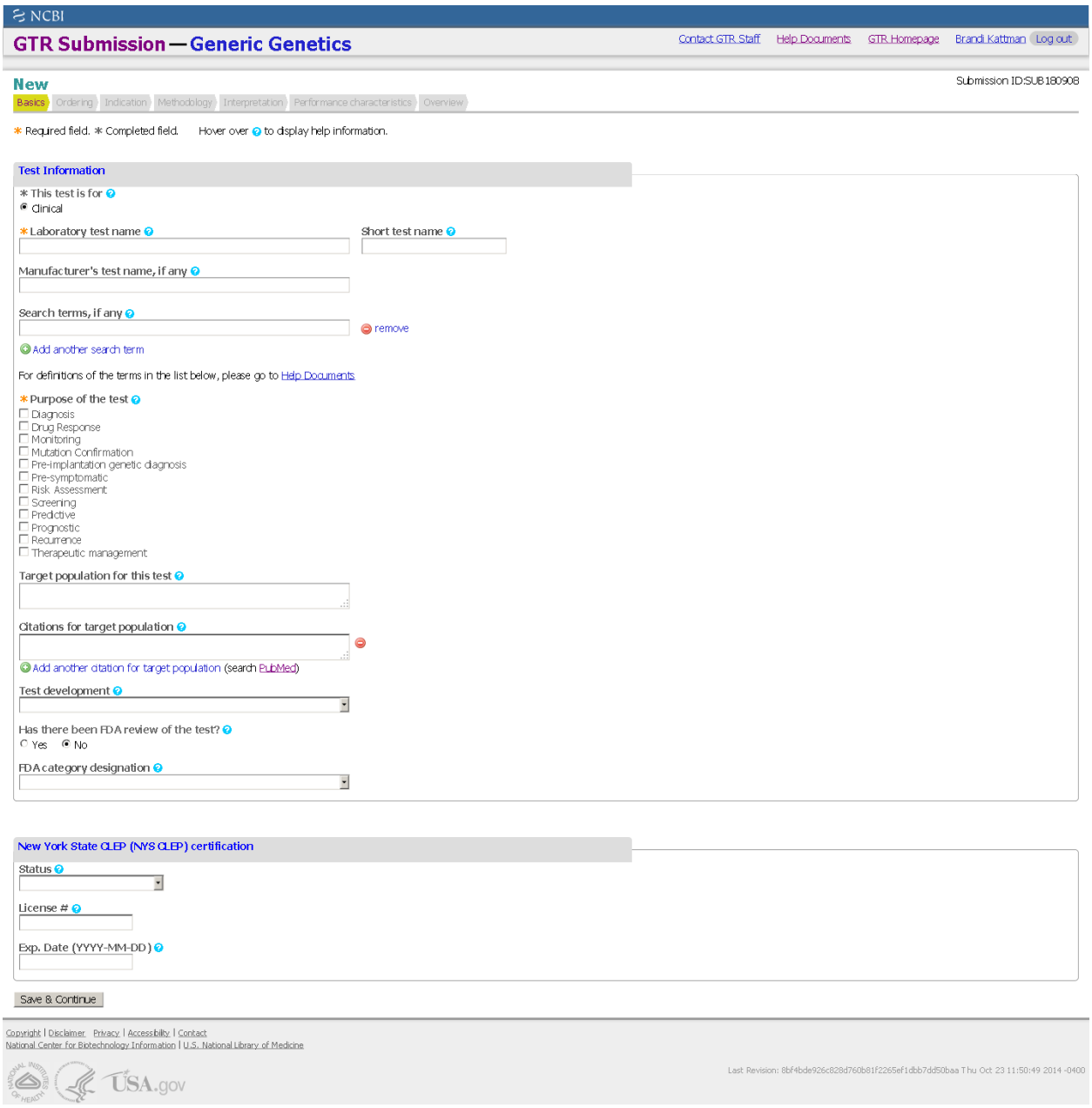
test name | Search tests | Delete tests

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

If you have started submitting a lab in GTR but do not see it in this page, Please log in 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 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	◉	○	○	○
Pre-test genetic counseling	◉	○	○	
Post-test genetic counseling	◉	○	○	

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 ?

Brandi Kattman

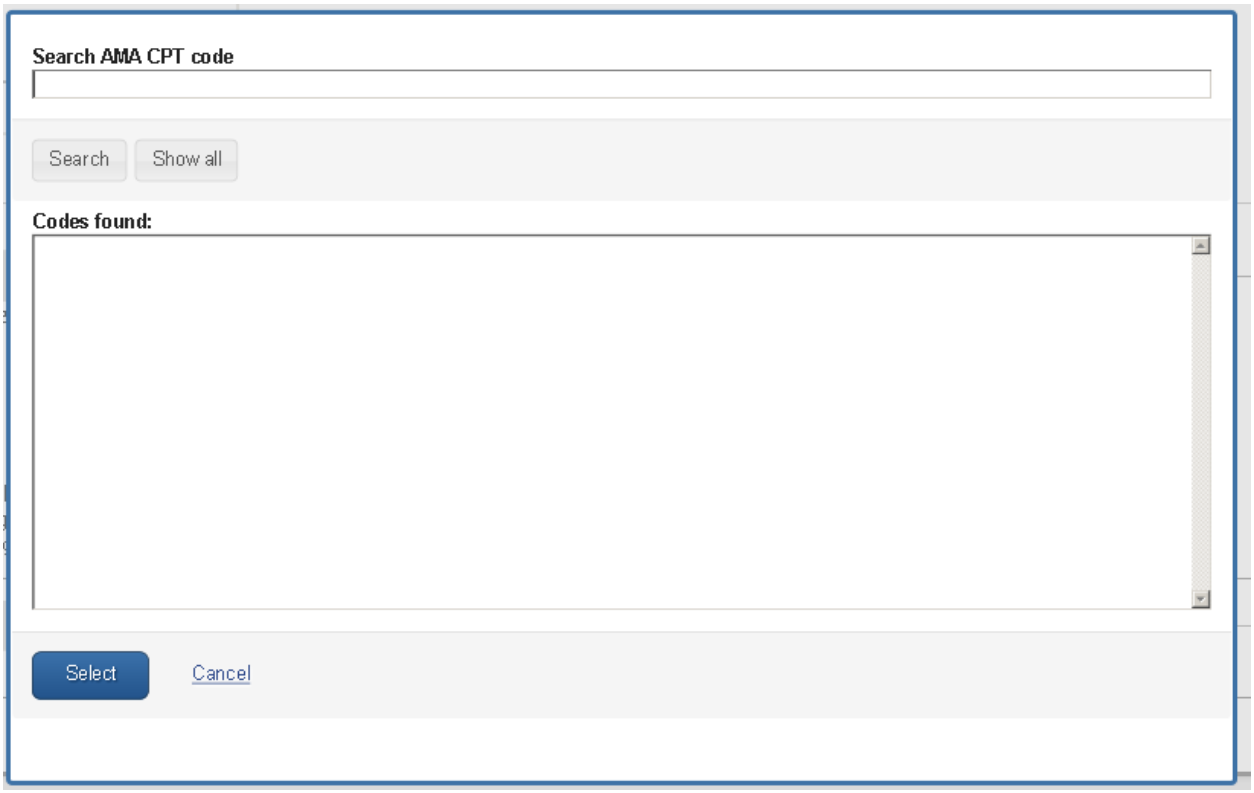
Contact policy ?

- ☐ 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

NCBI

GTR Submission — Generic Genetics

Contact GTR Staff | Help Documents | GTR Homepage | Brandi Kattman | Log out

New Clinical Test

Submission ID:SUB180908

Basics | Ordering | Indication | Methodology | 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: ☐ pharmacogenetic 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

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GTR Submission — Generic Genetics

New Clinical Test

Submission ID:SUB180908

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

Condition/Phenotype Information

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

Indication type
disease

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

Save Cancel

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GTR Submission — Generic Genetics

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 QuantiGene 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)

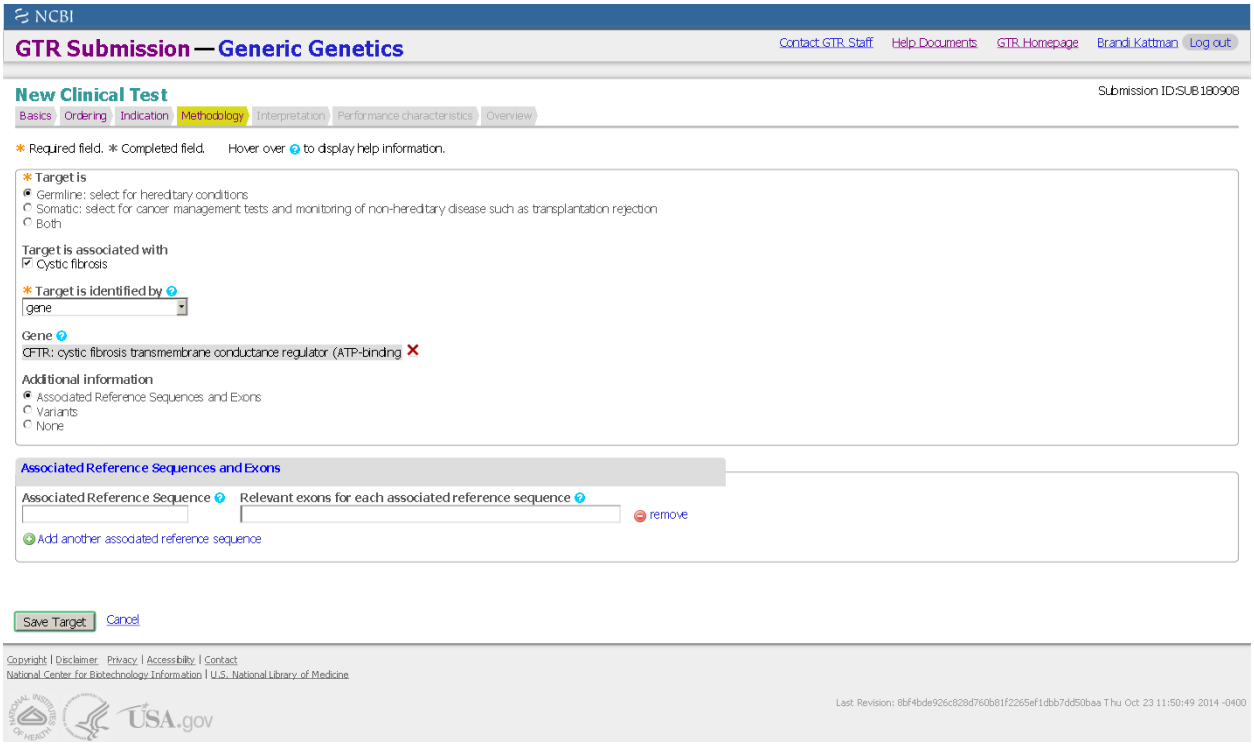
Add another test comment

* 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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GTR Submission — Generic Genetics
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GTR Homepage
Brandi Kattman
Log out
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
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USA.gov
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GTR Submission — Generic Genetics

New Clinical Test

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Hover over ? to display help information.

Upload Sample Reports

Sample negative report ?
Browse... No file selected.

Sample positive report ?
Browse... No file selected.

Sample VUS report ?
Browse... 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

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GTR Submission — Generic Genetics

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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	☑	
Specimen preparation	☐	☐
Wet lab work	☐	☐
Interpretation	☐	☐
Generate report	☐	☐

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 | remove

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

remove

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

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GTR Submission — Generic Genetics

New Clinical Test

Submission ID:SUB180908

Preview how your test will display | Submit

Return to homepage

Basics

This test is for	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	Brandi 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

Preview how your test will display

Submit

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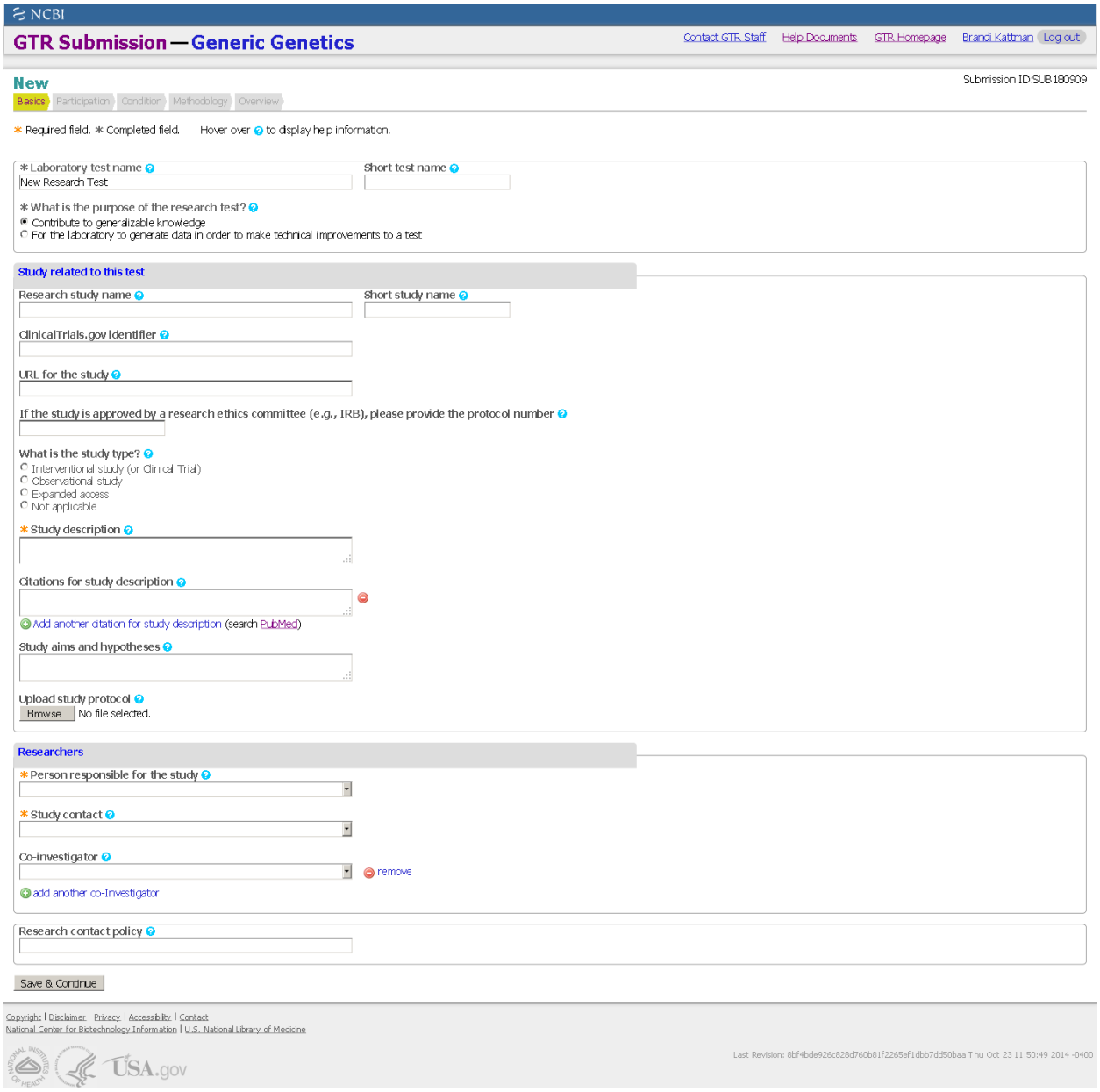
ADDING A RESEARCH TEST



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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 ?

Browse... No file selected.

Is the study currently recruiting participants? ?

- Yes
- No
- Unknown

Eligibility criteria ?

Recruitment sites ?

Save & Continue

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New Research Test

Submission ID:SUB180909

Basics | Participation | Condition | Methodology | Overview

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

Condition/Phenotype | Primary | Actions

Add other conditions/phenotypes

Limit to: pharmacogenetic response conditions

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

* Type Condition/Phenotype to search

CF: Cystic fibrosis Search

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

Add selected conditions/phenotypes

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

Continue to the methodology section

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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
disease

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 ?

Save Cancel

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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 QuantiGene 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

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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
- [x] 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

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GTR Submission — Generic Genetics

New Research Test

Submission ID:SUB180909

Preview how your test will display | Submit

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

Submit

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