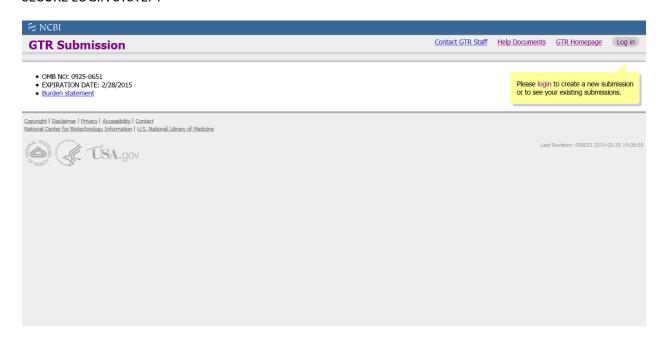
Information Collection Screenshots

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



BURDEN STATEMENT



A 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 GETTING STARTED RESOURCES POPULAR FEATURED NCBI INFORMATION NCBI Education Chemicals & Bioassays PubMed Genetic Testing Registry About NCBI NCBI Help Manual Data & Software Bookshelf PubMed Health Research at NCBI NCBI Handbook DNA 8 RNA PubMed Central GenBank NCBI News

GTR CODE OF CONDUCT

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 dains 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

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 daims or any other breaches of this Code of Conduct on our Contact GTR page

I agree Disagree

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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.

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 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/egl/

855-831-7447 Phone: 404-778-8559 Fax:

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

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

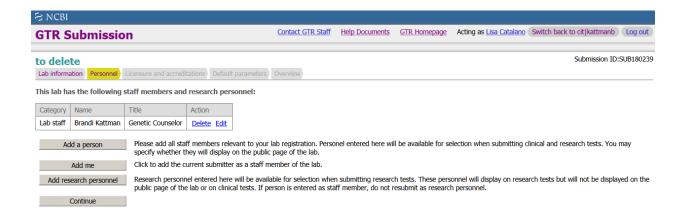
College of American Pathologists, CAP: 7181693 exp: 2014-08-02

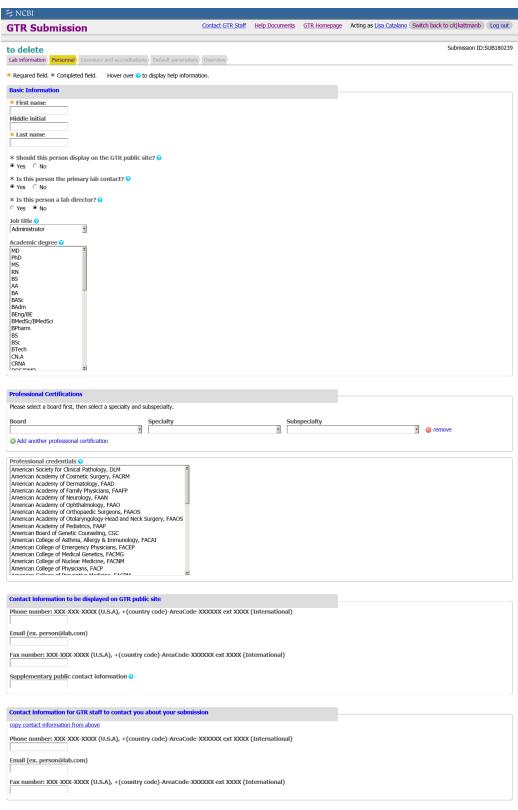
Tests in this lab(875)

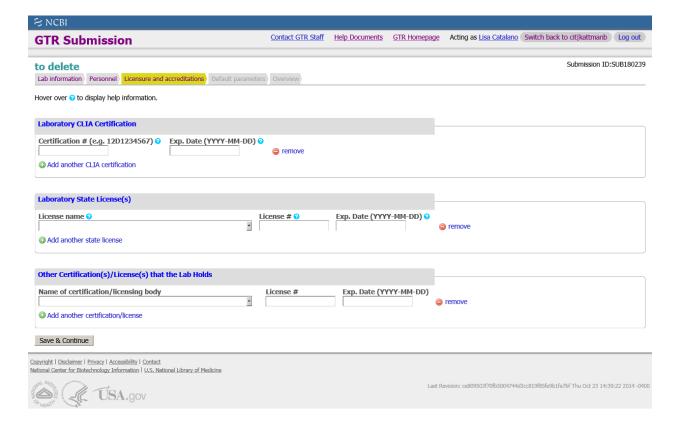
| Test name | ÷ | Test type | Submission status | Action | Use to create a new test | Link to public site |
|--|---|------------------|------------------------|--------------------|--------------------------|---------------------|
| 1q21.1 Deletion/Duplication Analysis | | Clinical test | Processed successfully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

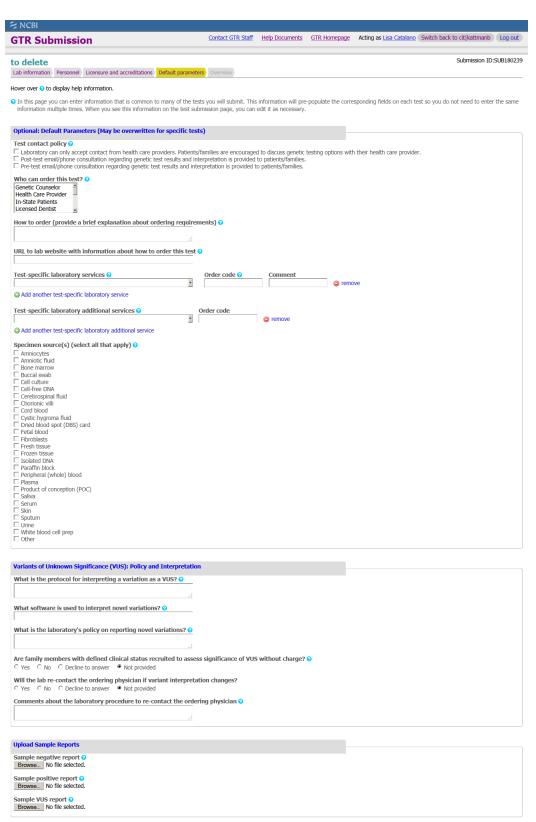
ADD A NEW LAB

| € NCBI | | | | | | |
|--|------------------------|-------------------|--------------|--|--|--|
| GTR Submission | Contact GTR Staff | Help Documents | GTR Homepage | Acting as <u>Lisa Catalano</u> Switch back to cit kattmanb Log out | | |
| | | | | Cohesiana VII. Ci INVONO | | |
| New Lab information Personnel Licensure and accreditations Default parameters. | ers Overview | | | Submission ID:SUB180239 | | |
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| Laboratory & Institution Name | | | | | | |
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| GeneTests at NCBI lab ID, if known | | | | | | |
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| Name of department 2 | | | | | | |
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| Laboratory Address | | | | | | |
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| Additional address line 0 | | | | | | |
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| Alabama | | | | | | |
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| | • | , - | | | | |
| * Email (ex. lab@lab.com) and/or @ URL for lab contact form @ |) | | | | | |
| Lab website URL 2 | | | | | | |
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| Laboratory Types of Service | | | | | | |
| Service ② | Order code 2 | Comment | ⊚ remo | | | |
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| Laboratory Affiliation(s) | | | | | | |
| Name of affiliate (example: clinic, research center) @ | Website 2 | | | | | |
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| Add another affiliation | | | | | | |
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| 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 ISC ☐ Locus-specific Databases | A | | | | | |
| ☐ Mutation-specific Databases ☐ other | | | | | | |
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GTR Submission

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Submission ID:SUB180239

Generic Genetic Laboratory

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 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: brand(@lab.com Contact information for GTR staff to contact you about the submission: Phone number: 800-800-8888

Email: brandi@lab.com

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

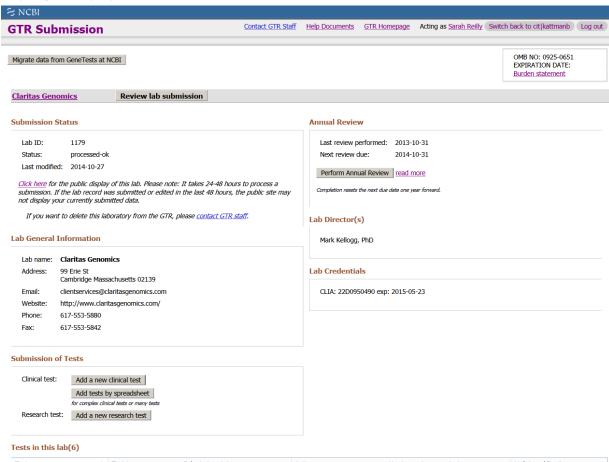
Status: Not submitted, Last modified: 16:42.

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

LIST OF TESTS



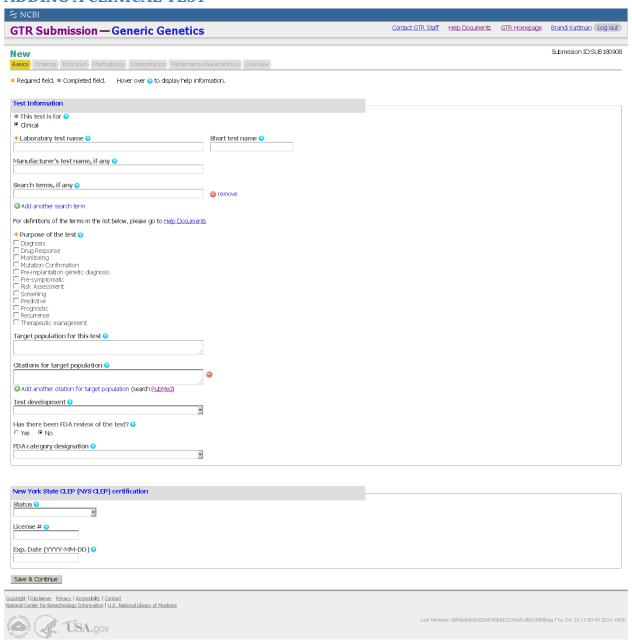
| Test name | * | Test type | Submission status | Action | Use to create a new test | Link to public site |
|-----------------|----------|---------------|-------------------|------------------|--------------------------|---------------------|
| CHD7 Sequencing | | Clinical test | Unfinished | Continue 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 |

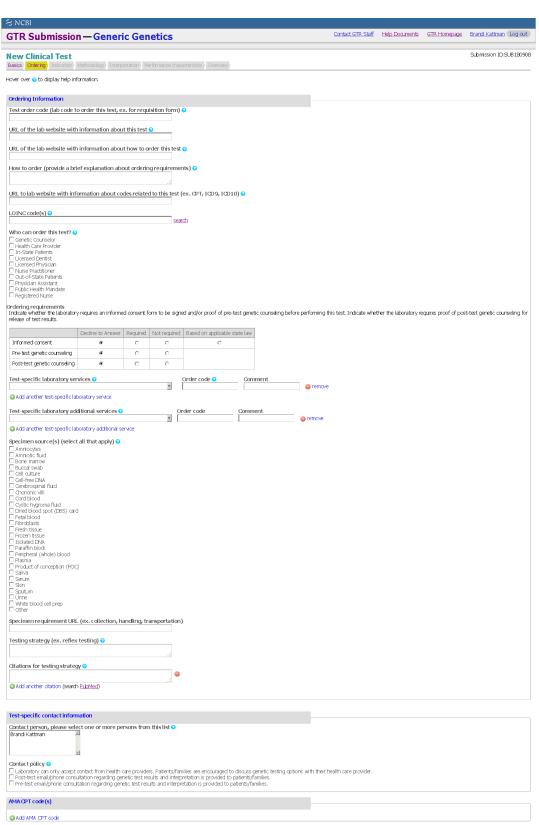
Want to submit another lab to GTR? Please check first if the lab can be <u>migrated from GeneTests</u>. Migrating it will avoid duplicated display of your lab in GTR. If the lab is NOT in GeneTests, <u>click here to submit it to GTR</u>.

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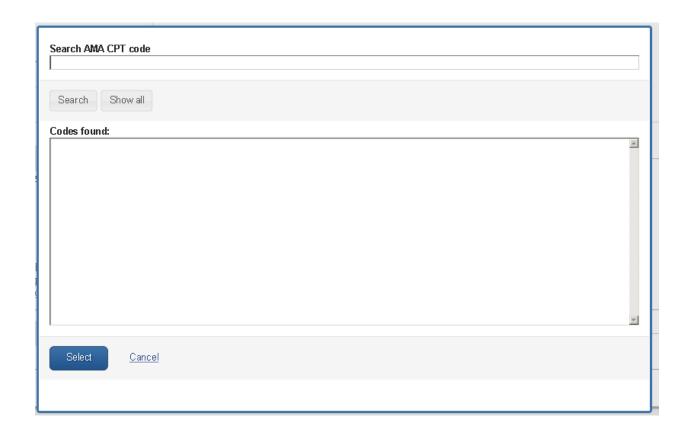


ADDING A CLINICAL TEST



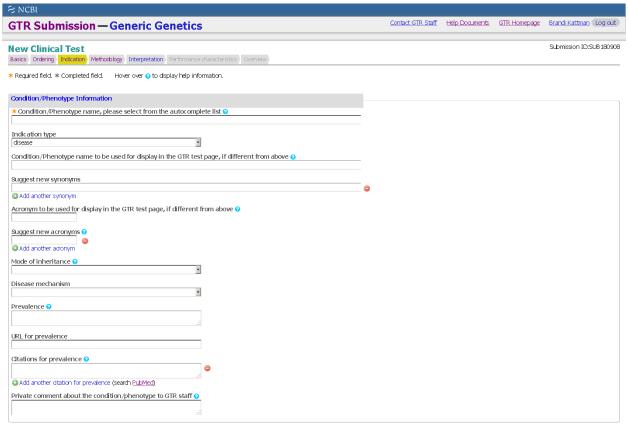


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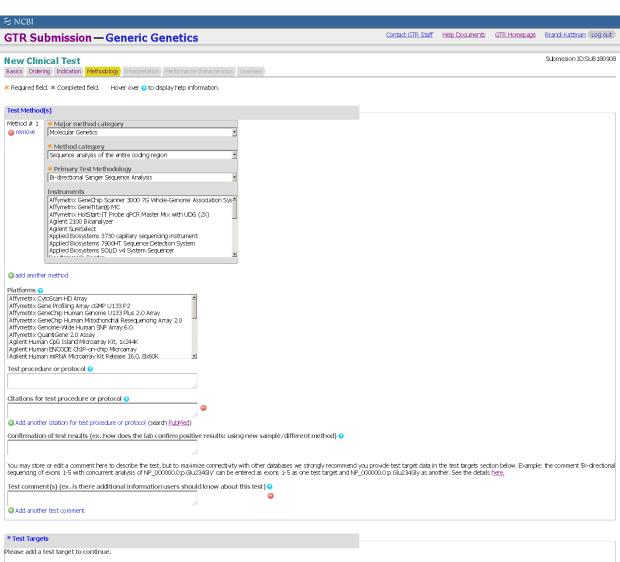
USA.gov



Save Cancel

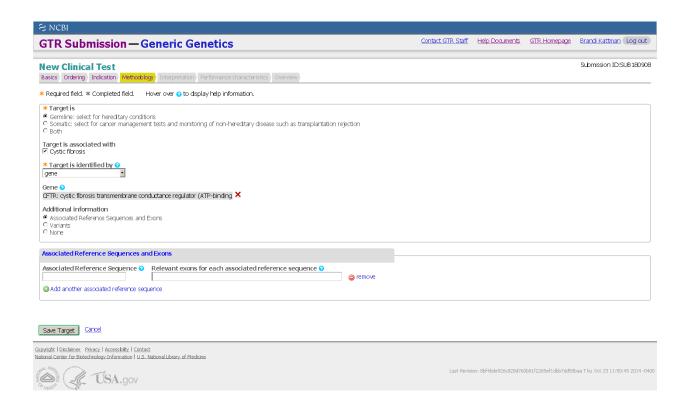
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Some condition(s) are not connected to a test target. Please add a target for each condition. Add Test Target





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|---|-------------------|----------------|--------------|--------------------------|
| GTR Submission — Generic Genetics | Contact GTR Staff | Help Documents | GTR Homepage | Brandi Kattman (Log out) |
| | | | | |
| New Clinical Test Basks Ordering Indication Methodology Interpretation Performance characteristics Overview | | | | Submission ID:SUB180908 |
| Hover over o 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 ○ Dedine to answer ○ Not provided | | | | |
| Comments about the laboratory procedure to re-contact the ordering physician @ | | | | |
| | | | | |
| Research performed after clinical testing is complete 🥹 | | | | |
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GTR Submission — Generic Genetics

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New Clinical Test

Basics | Ordering | Indication | Methodology | Interpretation | Performance characteristics | Overview

Submission ID:SUB 180908

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 Pre-test generic counseling required Dedine to answer Dedine to answer Post-test generic counseling required Dedine 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 Will the lab re-contact the ordering physician if variant interpretation changes

Performance characteristics

Entire test: internal

Test performance location(s) Analytical validity 99% sensitivity and specificity.

Proficiency testing is performed for this test?

Preview how your test will display

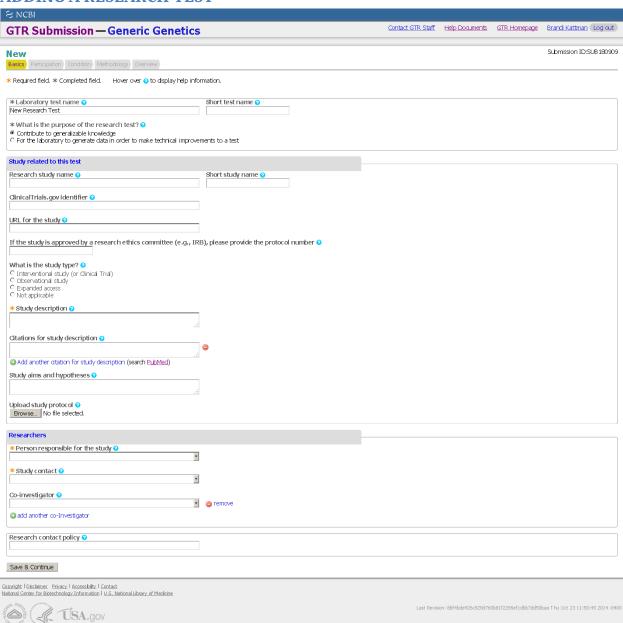
Submit

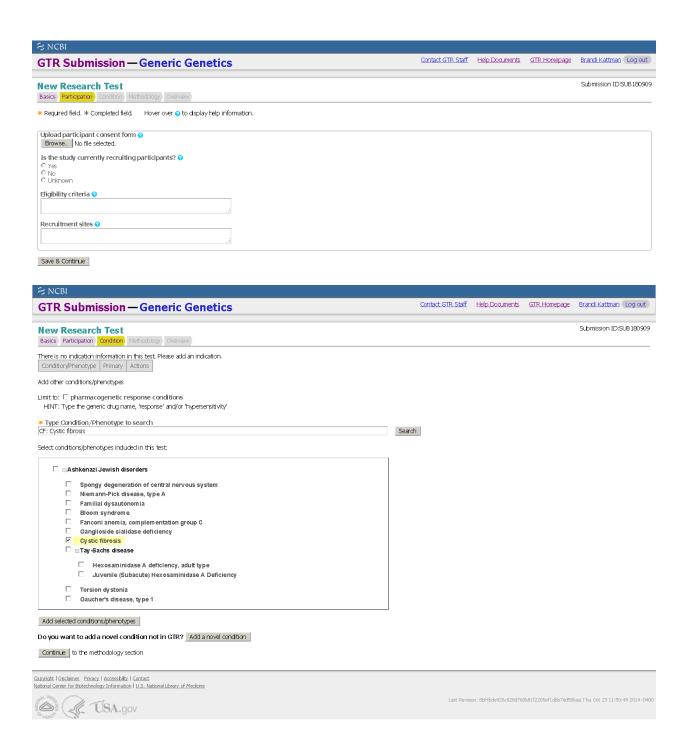
Return to homepage

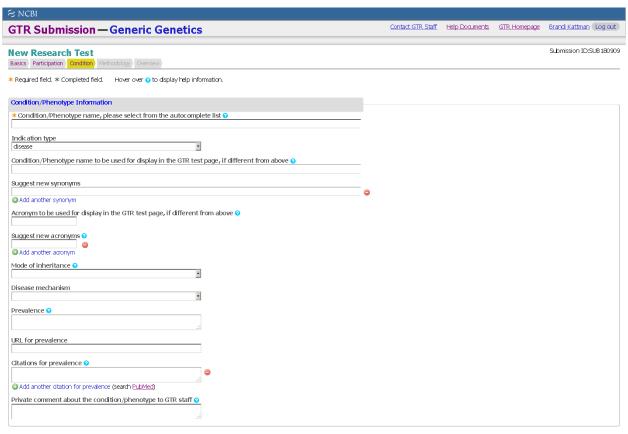
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ADDING A RESEARCH TEST



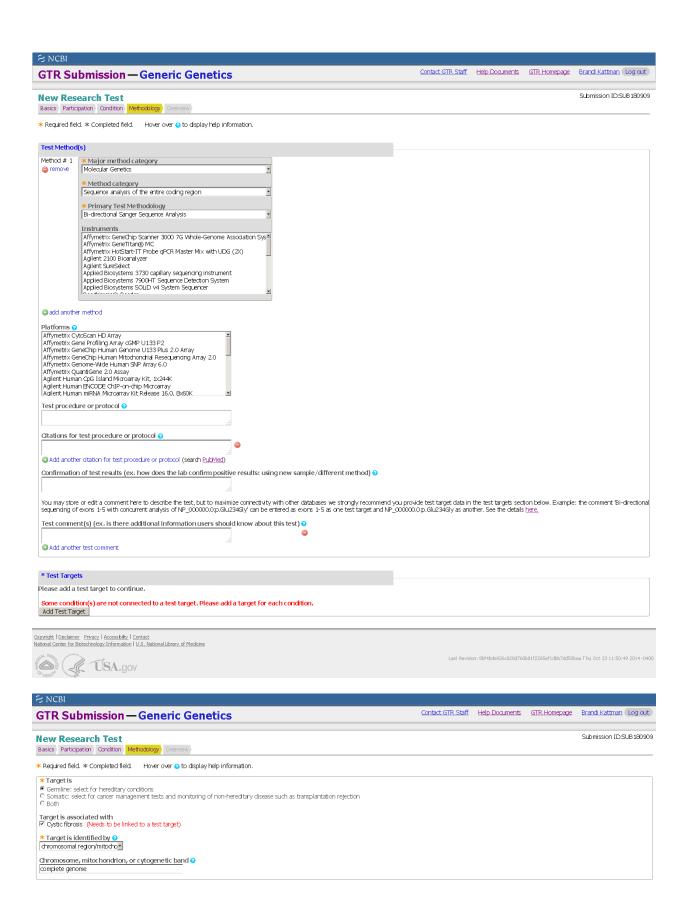




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GTR Submission — Generic Genetics

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Submission ID:SUB 180909

New Research Test

Basics | Participation | Condition | Methodology | Overview

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Basics

This test is for Name

Test purpose Contribute to generalizable knowledge

Study name Study Description

To discover new genes associated with disease.

Person responsible for the Brandi Kattman study 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/mitodhondrion: complete genome

Reference Sequence(s) Variant(s)

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