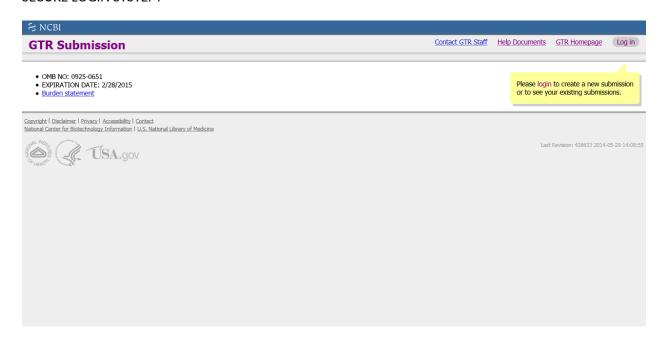
## **Information Collection Screenshots**

# 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** 

Contact GTR Staff Help Documents GTR Homepage Brandi Kattman 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 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

Copyright | Disclaimer Privacy | Accessibility | Contact National Center for Biotechnology Information | U.S. National Library of Medicine

USA.gov

#### AMA CPT CODE LICENSE AGREEMENT

S NCBI

**GTR Submission** 

Contact GTR Staff Help Documents GTR Homepage Brandi Blaisdell Log out

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

Any use not authorized herein is prohibited, including by way of illustration and not by way of limitation, making copies of CPT for reside and/or license, transferring copies of CPT to any party not bound by this Agreement, creating any modified or derivative work of CPT, or making any commercial use of CPT. License to use CPT for any use not authorized herein must be obtained through the American Medical Association, Intellectual Property Services, 515 N. State Street, Chicago, Illinois 60654. Applications are available at the American Medical Association Web site, www.ama-assn.org/go/pt

U.S. Government Rights
This product includes CPT which is commercial technical data and/or computer data bases and/or commercial computer software and/or commercial computer software documentation, as applicable which were developed exclusively at private expense by the American Medical Association, 515 North State Street, Chicago, Illinois, 60654. U.S. government rights to use, modify, reproduce, release, perform, display, or display, or display, or display, or display and data and/or computer data bases and/or computer software and/or computer software documentation are subject to the limited rights restrictions of DEARS 252.227-105(b)(2) (November 1995) and/or subject to the restrictions of PARS 227.702-6, applicable for U.S. Department of Defense procurements and the limited rights restrictions of FAR 5227.14 (December 2007) and FAR 52.227-19 (December 2007), as applicable, and any applicable agency FAR Supplements, for non-Department of Defense Federal procurements.

#### Disclaimer of Warranties and Liabilities.

OTT is provided "as is" without warranty of any kind, either expressed or implied, including but not limited to the implied warranties of merchantability and fitness for a particular purpose. Fee schedules, relative value units, conversion factors and/or related components are not assigned by the AMA, are not part of OTT, and the (AMA)is not recommending their use. The AMA does not directly or indirectly practice medicine or dispense medical services. The responsibility for the content of this product if this company, and no endorsement by the AMA is intended or implied. The AMA disclaims responsibility for any consequences or liability attributable to or related to any use, non-use, or interpretation of information contained or not contained in this product.

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 dicking below on the button labeled "accept".

Accept Skip

Copyright | Disclaimer Privacy | Access bilty | Contact National Center for Biotechnology Information | U.S. National Library of Medicine

#### YOUR LABS IN GTR

#### S NCBI

#### **GTR Submission**

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

Migrate data from GeneTests at NCBI

OMB NO: 0925-0651 EXPIRATION DATE: 2/28/2015 Burden statement

#### **Emory Genetics Laboratory**

#### Review lab submission

#### Submission Status

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

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

If you want to delete this laboratory from the GTR, please contact GTR staff.

#### 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

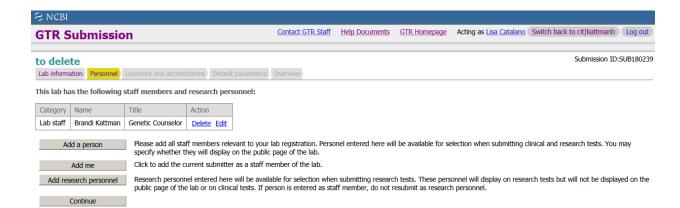
College of American Pathologists, CAP: 7181693 exp: 2014-08-02

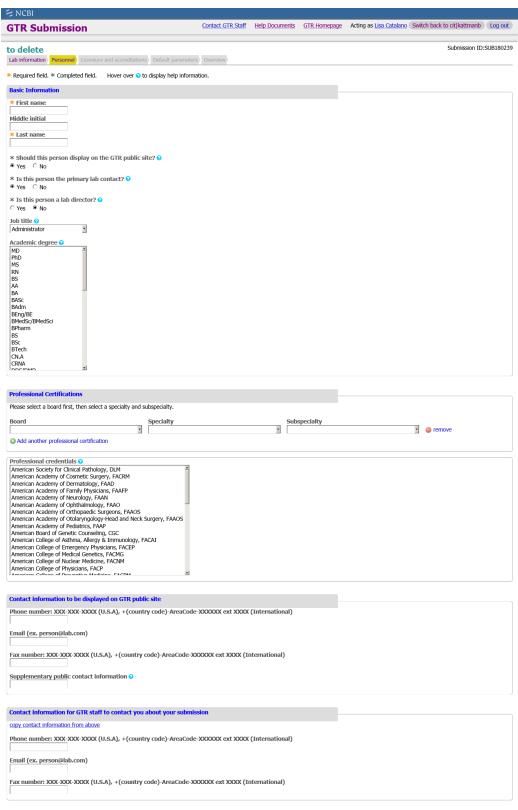
#### Tests in this lab(875)

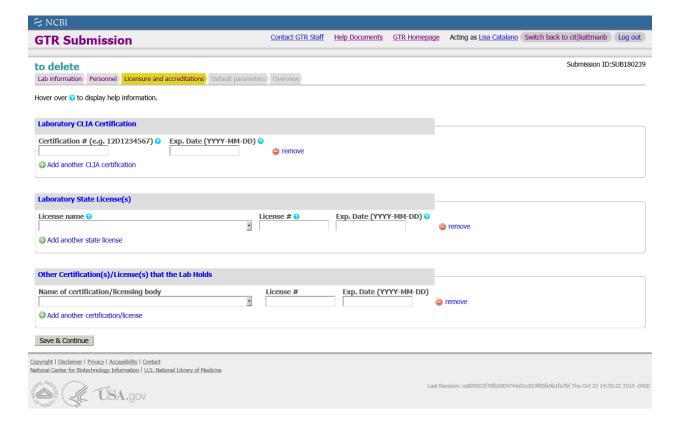
Test name	÷	Test type	Submission status	Action	Use to create a new test	Link to public site
1q21.1 Deletion/Duplication Analysis		Clinical test	Processed successfully	Update test	Сору	ID: GTR000508266
3-Hydroxy-3-Methylglutaryl CoA Lyase Deficiency (HMG): HMGCL Full Gene Sequencing		Clinical test	Processed 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

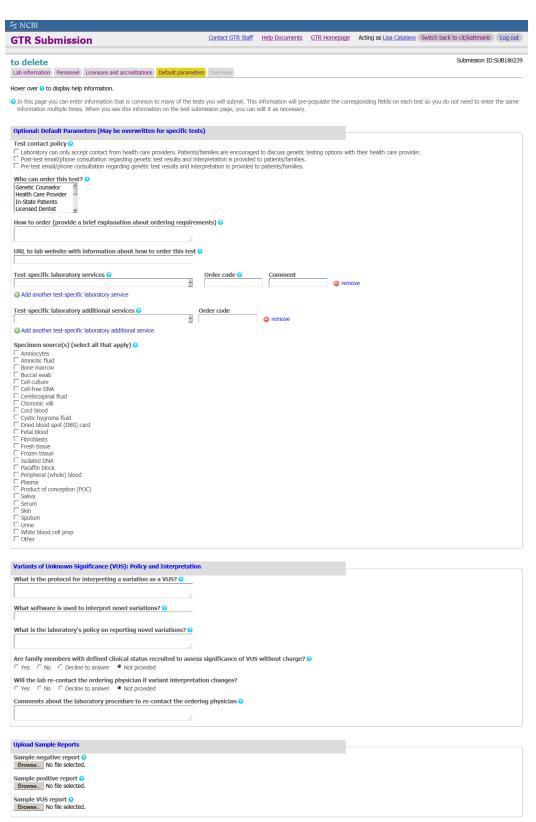
### **ADD A NEW LAB**

€ 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		
* Required field. * Completed field. Hover over © to display help information.						
Required field. To Completed field. Proved over the display fielp fillor	mauon.					
Laboratory & Institution Name						
* Name of laboratory @	Acronym of lab name	:0				
GeneTests at NCBI lab ID, if known						
Generalist at NCDI Iau 1D, II known						
Name of institution @	Acronym of institution	n name 🛭				
None of depote the O						
Name of department 2						
Laboratory Address						
* Country or region						
United States						
Street & No O						
Additional address line 0						
* City ©						
State or province 2						
Alabama						
* Postal code @						
* Make this address public? •						
C Yes C No						
* Phone number: XXX-XXXX-XXXXX (U.S.A), +(country code)-Area	Code-XXXXXXX ext XXX	X (International)	0			
Fax number: XXX-XXX-XXXX (U.S.A), +(country code)-AreaCode-	XXXXXX ext XXXXX (In	ternational) 🔞				
	•	, -				
* Email (ex. lab@lab.com) and/or @ URL for lab contact form @	)					
Lab website URL 2						
Laboratory Types of Service						
Service ②	Order code 2	Comment	⊚ remo			
Add another service	I		i remo	ove		
Laboratory Affiliation(s)						
Name of affiliate (example: clinic, research center) @	Website 2					
				⊜ 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 ISC ☐ Locus-specific Databases	A					
☐ Mutation-specific Databases ☐ other						
- Out-of						









Save & Continue

& NCBI

**GTR Submission** 

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

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

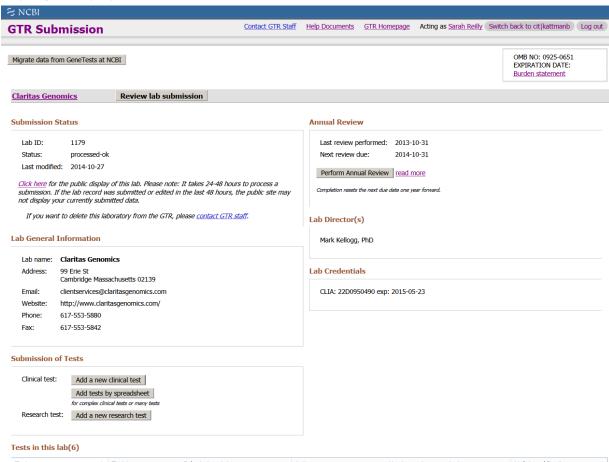
Status: Not submitted, Last modified: 16:42.

Copyright | Disclaimer | Privacy | Accessibility | Contact National Center for Biotechnology Information | U.S. National Library of Medicine



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

#### LIST OF TESTS



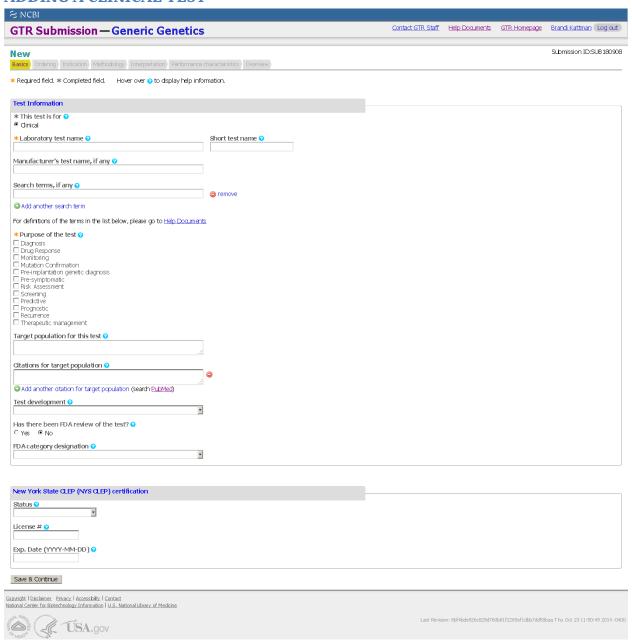
Test name	<b>*</b>	Test type	Submission status	Action	Use to create a new test	Link to public site
CHD7 Sequencing		Clinical test	Unfinished	Continue editing		ID: 334060
New test		Clinical test	Unfinished	Continue editing		ID: 330317
New test		Clinical test	Unfinished	Continue editing		ID: 334061
New 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

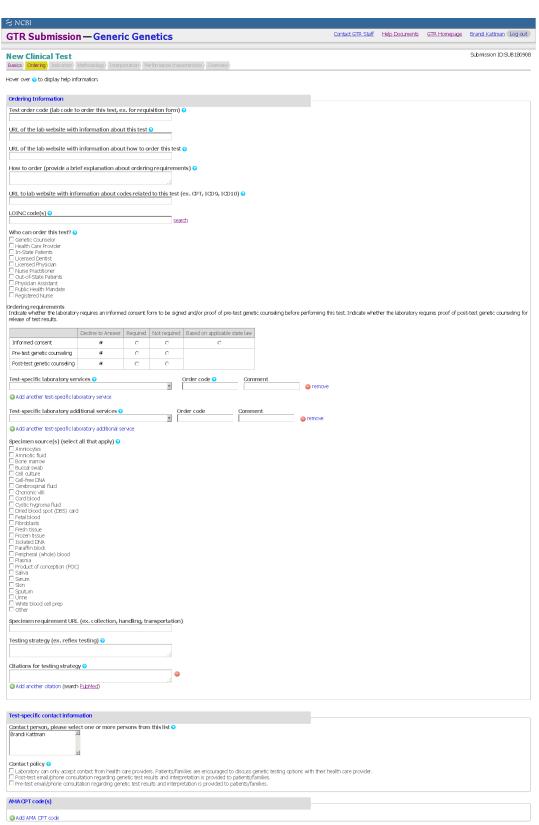
Want to submit another lab to GTR? Please check first if the lab can be <u>migrated from GeneTests</u>. Migrating it will avoid duplicated display of your lab in GTR. If the lab is NOT in GeneTests, <u>click here to submit it to GTR</u>.

Copyright | Disclaimer | Privacy | Accessibility | Contact National Center for Biotechnology Information | U.S. National Library of Medicine



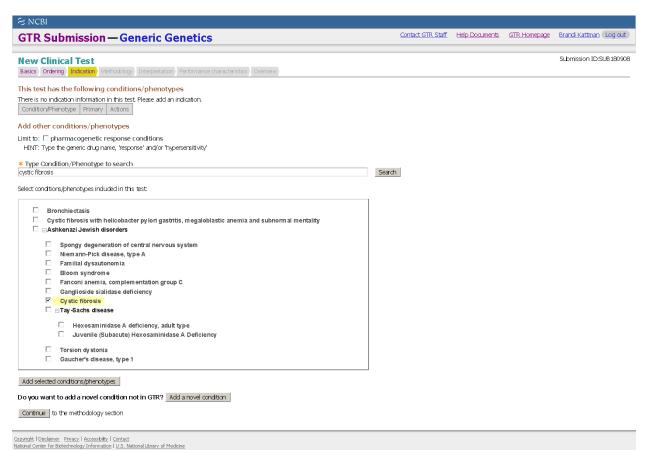
#### ADDING A CLINICAL TEST



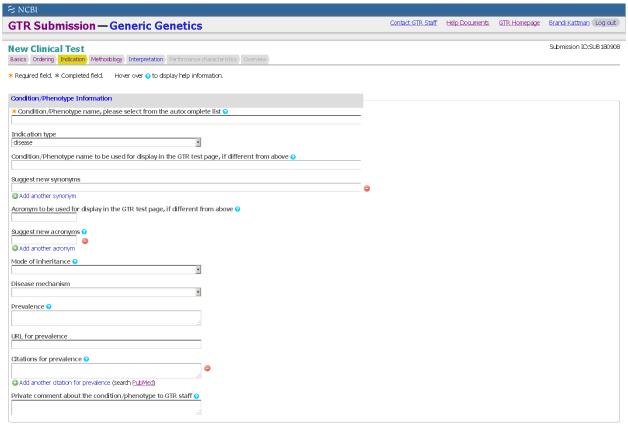


Save & Continue





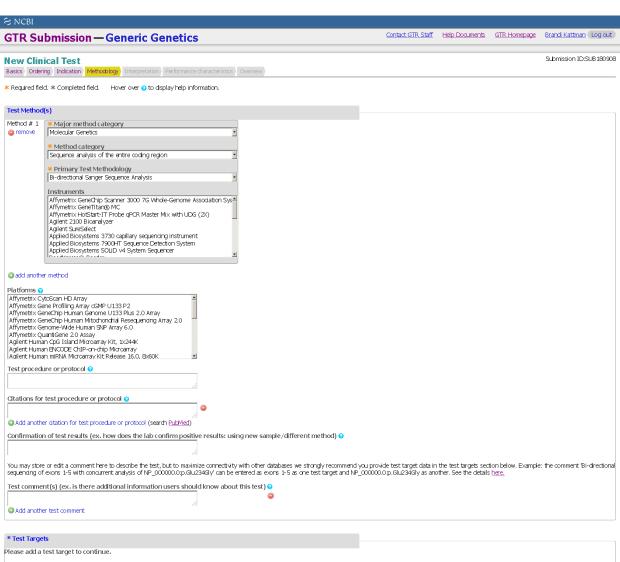
USA.gov



Save Cancel

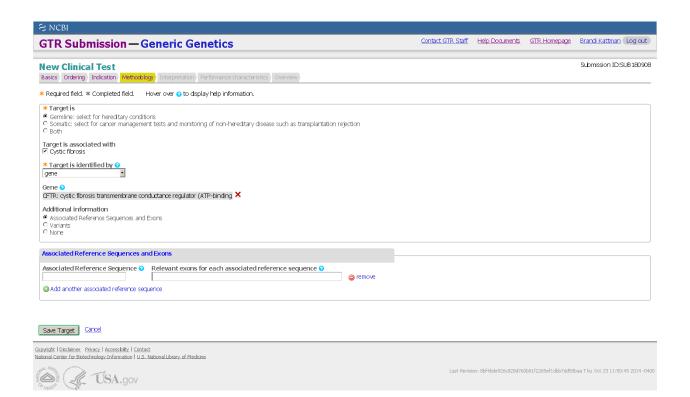
Copyright | Disclaimer Privacy | Accessibility | Contact
National Center for Biotechnology Information | U.S. National Library of Medicine





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





≈ NCBI				
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 🥹				
Save & Continue				

Copyright | Disclaimer Privacy | Access bilty | Contact
National Center for Biotechnology Information | U.S. National Library of Medicine





Save & Continue

#### GTR Submission — Generic Genetics

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

**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

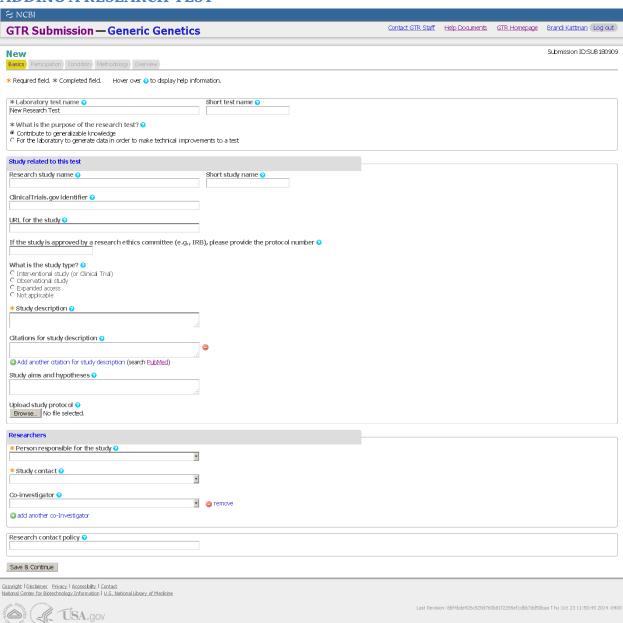
Submit

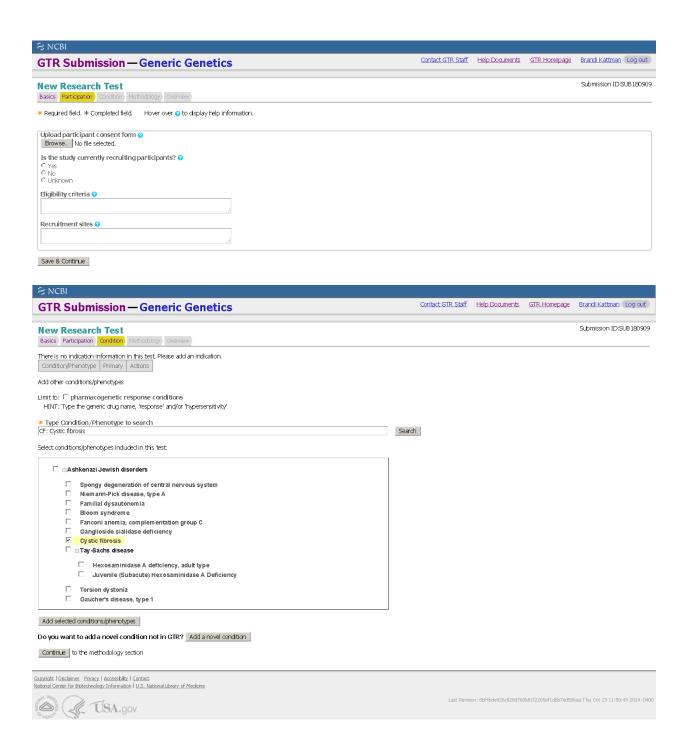
Return to homepage

Copyright | Disclaimer Privacy | Accessibility | Contact National Center for Biotechnology Information | U.S. National Library of Medicine



#### ADDING A RESEARCH TEST



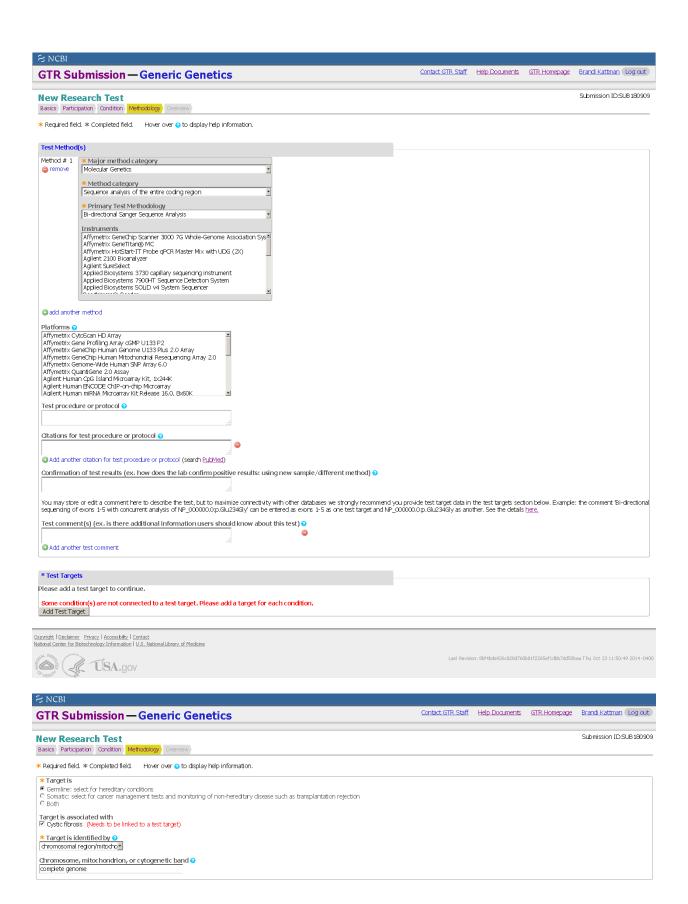




Save Cancel

Copyright | Disclaimer Privacy | Accessibility | Contact
National Center for Biotechnology Information | U.S. National Library of Medicine





#### GTR Submission — Generic Genetics

Contact GTR Staff Help Documents GTR Homepage Brandi Kattman Log out

Submission ID:SUB 180909

**New Research Test** 

Basics | Participation | Condition | Methodology | Overview

Preview how your test will display Submit

Return to homepage

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)

Preview how your test will display

Submit

Return to homepage

Copyright | Disclaimer Privacy | Accessibility | Contact
National Center for Biotechnology Information | U.S. National Library of Medicine

