

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).

11/3/2021

SECURE LOGIN SYSTEM

GTR Submission

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

OMB NO: 0925-0651
EXPIRATION DATE: 11/30/2021
[Burden statement](#)

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 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 if you have questions or if you need any help.

Thank you for participating in GTR!

FOLLOW NCBI



Follow NLM



National Library of
Medicine
8600 Rockville Pike
Bethesda, MD 20894

Copyright
FOIA
Privacy

Help
Accessibility
Careers

BURDEN STATEMENT

GTR: GENETIC TESTING REGISTRY

[Advanced search for tests](#)

- Overview
- Lab Submission
- Human Test Submission
- Microbe Test Submission
- Search GTR

[Print this document](#)

OMB NO: 0925-0651

EXPIRATION DATE: 11/30/2021

Burden Statement:

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

[Support Center](#)

GETTING STARTED

- [NCBI Education](#)
- [NCBI Help Manual](#)
- [NCBI Handbook](#)
- [Training & Tutorials](#)
- [Submit Data](#)

RESOURCES

- [Chemicals & Bioassays](#)
- [Data & Software](#)
- [DNA & RNA](#)
- [Domains & Structures](#)
- [Genes & Expression](#)
- [Genetics & Medicine](#)
- [Genomes & Maps](#)
- [Homology](#)
- [Literature](#)
- [Proteins](#)
- [Sequence Analysis](#)
- [Taxonomy](#)
- [Variation](#)

POPULAR

- [PubMed](#)
- [Bookshelf](#)
- [PubMed Central](#)
- [BLAST](#)
- [Nucleotide](#)
- [Genome](#)
- [SNP](#)
- [Gene](#)
- [Protein](#)
- [PubChem](#)

FEATURED

- [Genetic Testing Registry](#)
- [GenBank](#)
- [Reference Sequences](#)
- [Gene Expression Omnibus](#)
- [Genome Data Viewer](#)
- [Human Genome](#)
- [Mouse Genome](#)
- [Influenza Virus](#)
- [Primer-BLAST](#)
- [Sequence Read Archive](#)

NCBI INFORMATION

- [About NCBI](#)
- [Research at NCBI](#)
- [NCBI News & Blog](#)
- [NCBI FTP Site](#)
- [NCBI on Facebook](#)
- [NCBI on Twitter](#)
- [NCBI on YouTube](#)
- [Privacy Policy](#)

National Center for Biotechnology Information, U.S. National Library of Medicine
8600 Rockville Pike, Bethesda MD, 20894 USA
[Policies and Guidelines](#) | [Contact](#)



Last updated: 2021-11-10T17:08:11Z

GTR CODE OF CONDUCT



National Library of Medicine
National Center for Biotechnology Information

 genereviews.ncbi@gmail.com

GTR Submission

[Home](#)

[Contact GTR staff](#)

[Help documents](#)

[GTR Homepage](#)

[My profile](#)

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.

For microbe tests, submitters agree that they are submitting molecular or serologic microbe tests for microorganisms involved in human health and disease.

AMA CPT CODE LICENSE AGREEMENT



genereviews.ncbi@gmail.com

GTR Submission

[Home](#) [Contact GTR staff](#) [Help documents](#) [GTR Homepage](#) [My profile](#)

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.

Any use not authorized herein is prohibited, including by way of illustration and not by way of limitation, making copies of CPT for resale 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/cpt.

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 disclose these technical data and/or computer data bases and/or computer software and/or computer software documentation are subject to the limited rights restrictions of DFARS 252.227-7015(b)(2) (November 1995) and/or subject to the restrictions of DFARS 227.7202-1(a) (June 1995) and DFARS 227.7202-3(a) (June 1995), as applicable for U.S. Department of Defense procurements and the limited rights restrictions of FAR 52.227-14 (December 2007) and/or subject to the restricted rights provisions of FAR 52.227-14 (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. CPT 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 CPT, 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 is with 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 clicking below on the button labeled “accept”.

Accept

Skip

YOUR LABS IN GTR

GTR Submission

[Home](#) [Contact GTR staff](#) [Help documents](#) [GTR Homepage](#) [My profile](#)

OMB NO: 0925-0651
EXPIRATION DATE: 11/30/2021
Burden statement

GoPath Laboratories, LLC

[Review lab submission](#)

Lab Submission Status

Lab ID: 507620
Status: **Processed** Public display of this lab
Last modified: 2021-11-08

ⓘ 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: **GoPath Laboratories, LLC**
Address: 1351 Barclay Blvd
Buffalo Grove Illinois 60089
Email: vengelmenn@gopathlabs.com
Website: <http://www.gopathlabs.com/>
Phone: 855-467-2849
Fax: 224-588-9941

Annual Review

Last review performed: 2021-09-22
Next review due: 2022-09-22

[Perform annual review](#) ⓘ

Learn more about annual review
Completion resets the next due date one year forward.

Lab Director(s)

Jim Lu, MD

Lab Credentials

CLIA: 14D2037930 exp: 2023-10-22
IL - Illinois Department of Public Health IDPH: 036.119395 exp: 2023-07-31
College of American Pathologists, CAP: 8662247 exp: 2023-10-22

Usage Reports

07/01/2021 to 09/30/2021 ⓘ These reports show you the usage of lab and test records in the GTR public site (how many times each record was seen).

[Archived reports](#)

Submission of Tests

Human Genetic Tests (molecular, biochemical, cytogenetic) ⌵

Microbe Tests (microbiology, molecular, serologic) ⌵

Tests in this lab 13

Test name	Test type	Submission status	Action	Use to create a new test	Link to public site
BRAF Individual Marker - Ovarian Cancer	Clinical test	Processed	Update test	Copy	ID: GTR000591182
Brain Cancer Profile	Clinical test	Processed	Update test	Copy	ID: GTR000591183
Colon Cancer Profile	Clinical test	Processed	Update test	Copy	ID: GTR000591184
DiabetesNow Comprehensive Genetic Panel	Clinical test	Processed	Update test	Copy	ID: GTR000596384
GIST Profile	Clinical test	Processed	Update test	Copy	ID: GTR000591198
IDH1 IDH2 Individual Marker, Brain Cancer	Clinical test	Processed	Update test	Copy	ID: GTR000591174
KRAS - Individual Marker for Ovarian Cancer	Clinical test	Processed	Update test	Copy	ID: GTR000591201
Lung Cancer Driver Profile	Clinical test	Processed	Update test	Copy	ID: GTR000591172
Lung Cancer Expanded Profile	Clinical test	Processed	Update test	Copy	ID: GTR000591175
LYNCHnow Extended	Clinical test	Processed	Update test	Copy	ID: GTR000591263
Melanoma Profile	Clinical test	Processed	Update test	Copy	ID: GTR000591211
Mismatch Repair Proteins by Immunohistochemistry	Clinical test	Processed	Update test	Copy	ID: GTR000591173
OncoDefender CRC	Clinical test	Processed	Update test	Copy	ID: GTR000591213

ⓘ 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](#)

New submission: SUB10192224

- 1 LAB INFORMATION
- 2 PERSONNEL
- 3 LICENSURE AND ACCREDITATIONS
- 4 DEFAULT PARAMETERS
- 5 REVIEW & SUBMIT

Lab Information

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

Laboratory & Institution Name

* Name of laboratory ⓘ Acronym of lab name ⓘ

Name of institution ⓘ Acronym of institution name ⓘ

Name of department ⓘ

Laboratory Address

* Country or region

Street & No ⓘ

Additional address line ⓘ

* City ⓘ

State or province ⓘ

* 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 ⓘ ** At least one is required

Lab website URL ⓘ

Laboratory Types of Service

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

[Add another service](#)

Laboratory Affiliation(s)

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

[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)
- ICCG (International Collaboration for Clinical Genetics) - Previously ISCA
- Locus-specific Databases
- Mutation-specific Databases
- Other

Save & continue


Example submission: SUB10192224


- 1 LAB INFORMATION
- 2 PERSONNEL
- 3 LICENSURE AND ACCREDITATIONS
- 4 DEFAULT PARAMETERS
- 5 REVIEW & SUBMIT


Personnel

 This lab has no personnel information yet.

Category	Name	Title	Actions
----------	------	-------	---------

Add a person		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.	
------------------------------	---	--	--

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](#)

Example submission: SUB10192224

- 1 LAB INFORMATION
- 2 PERSONNEL
- 3 LICENSURE AND ACCREDITATIONS
- 4 DEFAULT PARAMETERS
- 5 REVIEW & SUBMIT

Personnel Details

* 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 ⓘ Please specify
 Administrator

Academic degree ⓘ Please specify

 MD
 PhD
 MS
 RN
 BS
 AA
 BA
 BAsC
 BAdm
 BEng/BE
 BMedSc/BMedSci
 BPharm
 BS

Professional Certifications

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

Board Specialty Subspecialty

[Add another professional certification](#)

Professional credentials ⓘ Please specify

 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

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

Example submission: SUB10192224**1** LAB INFORMATION **2** PERSONNEL **3** LICENSURE AND ACCREDITATIONS **4** DEFAULT PARAMETERS **5** REVIEW & SUBMIT**Add Research Personnel**

★ Required field. ★ Completed field. Hover over ⓘ to display help information.

Enter a Person responsible for the study (Minimal), a Study contact (Minimal) and Co-investigator(s) (Optional). Research personnel entered here will be available for selection when submitting research tests.

★ First name	★ Last name
<input type="text"/>	<input type="text"/>
Academic degree ⓘ	Please specify
<input type="text" value="MD"/> PhD MS RN BS AA BA BASC BAdm BEng/BE BMedSc/BMedSci BPharm BS	<input type="text"/>
Institution, City, State/Province, Country ⓘ	
<input type="text"/>	

Contact information

ⓘ Email is REQUIRED for Person responsible for the study. NO contact information for this person will display publicly.

Either Email or Phone number are REQUIRED for Study contact(s). All contact information for Study contact will display publicly.

Contact information is optional for Co-investigator(s) and will NOT display publicly.

Email (ex. person@lab.com) ⓘ
<input type="text"/>
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"/>

Save

Cancel


Example submission: SUB10192224



- 1 LAB INFORMATION
- 2 PERSONNEL
- 3 LICENSURE AND ACCREDITATIONS
- 4 DEFAULT PARAMETERS
- 5 REVIEW & SUBMIT


Licensure And Accreditations

Hover over  to display help information.

Laboratory CLIA (or CLIP) Certification




 Submission of clinical tests by US labs requires provision of CLIA (or CLIP) number

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

 Add another CLIA certification

 Add CLIP certification (applicable to DoD labs only)


Laboratory State License(s)

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

 Add another certification/license

[Save & continue](#)

Example submission: SUB348876

- 1 LAB INFORMATION
- 2 PERSONNEL
- 3 LICENSURE AND ACCREDITATIONS
- 4 DEFAULT PARAMETERS
- 5 REVIEW & SUBMIT

Default Parameters

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) ⓘ

Text input field for ordering requirements.

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

Text input field for lab website URL.

Test-specific laboratory services ⓘ

Dropdown menu for test-specific laboratory services.

Order code ⓘ

Text input field for order code.

Comment ⓘ

Text input field for comment.

Add another test-specific laboratory service

Test-specific laboratory additional services ⓘ

Dropdown menu for test-specific laboratory additional services.

Order code

Text input field for order code.

Comment ⓘ

Text input field for comment.

Add another test-specific laboratory additional service

Specimen source(s) (select all that apply) ⓘ

- Amniocytes
- Amniotic fluid
- Bone marrow
- Buccal swab
- Buffy coat
- 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
- Nasal aspirates
- Nasopharyngeal washes
- Oropharyngeal swab
- 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? ⓘ

Text input field for VUS protocol.

What software is used to interpret novel variations? ⓘ

Text input field for VUS software.

What is the laboratory's policy on reporting novel variations? ⓘ

Text input field for VUS reporting policy.

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 ⓘ

Text input field for VUS re-contact comments.

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

Example submission: SUB348876

1 LAB INFORMATION 2 PERSONNEL 3 LICENSURE AND ACCREDITATIONS 4 DEFAULT PARAMETERS 5 REVIEW & SUBMIT

Summary

Submit

Lab ID: 506435 Status: Not submitted, Last modified: 20:08.

[Perform annual review](#)

Lab information

Name Example
 Institution
 Address Bethesda Maryland 21842
 Phone 301-555-5555
 Email GTRlab@lab.com
 Types of service
 Affiliations

Personnel

Gregor Mendel 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@lab.com
 Contact information for GTR staff to contact you about the submission:
 Email: person@lab.com

Licensure and accreditations

CLIA or CLIP certification CLIA: 12D1234567 Exp: 2023-12-01
 State license(s) MD - Maryland Department of Health and Mental Hygiene DHMH: 1234567 Non-expiring
 Other certification(s) College of American Pathologists, CAP: 112233 Exp: 2023-11-15

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 Genetic Counselor
 Test-specific services Clinical Testing/Confirmation of Mutations Identified Previously
 Order code: 234
 Specimen source(s) Peripheral (whole) blood

Submit

Lab ID: 506435 Status: Not submitted, Last modified: 20:08.

LIST OF TESTS

GTR Submission

OMB NO: 0925-0651
 EXPIRATION DATE: 11/30/2021
[Burden statement](#)

University of Minnesota Physicians Outreach Laboratory

[Edit this lab](#)

Lab Submission Status

Lab ID: 1130
 Status: ⓘ Unfinished at the Overview step
 Last modified: 2021-10-15
ⓘ If you want to delete this laboratory from the GTR, please contact GTR staff.

Lab General Information

Lab name: University of Minnesota Physicians Outreach Laboratory, University of Minnesota
 Address: Mayo Building, Room D210
 420 Delaware St SE
 Minneapolis Minnesota 55455
 Email: mbower1@fairview.org
 Website: https://mphysicians.org/outreach-laboratories
 Phone: 612-273-8445
 Fax: 612-273-8959

Annual Review

Last review performed: 2021-04-26
 Next review due: 2022-04-26
[Perform annual review](#) ⓘ Learn more about annual review
 Completion resets the next due date one year forward.

Lab Director(s)

Sophia Yohe, MD

Lab Credentials

CLIA: 24D0688128 exp: 2022-10-19
 CLIA: 24D2043116 exp: 2023-04-14
 College of American Pathologists, CAP: 1906001 exp: 2022-09-01

Usage Reports

07/01/2021 to 09/30/2021 ⓘ These reports show you the usage of lab and test records in the GTR public site (how many times each record was seen).
[Archived reports](#)

Submission of Tests

- Human Genetic Tests (molecular; biochemical; cytogenetic) ⌵
- Microbe Tests (microbiology; molecular; serologic) ⌵

Tests in this lab 5

Test name ^	Test type ⚙	Submission status ⚙	Action	Use to create a new test	Link to public site
Breast Actionable panel	Clinical test	✔ Processed	Update test	Copy	ID: GTR000592961
Breast Expanded	Clinical test	✔ Processed	Update test	Copy	ID: GTR000395973
New test	Clinical test	ⓘ Unfinished	Continue editing		
PAH sequencing	Clinical test	✔ Processed	Update test	Copy	ID: GTR000027152
PAX2 sequencing	Clinical test	✔ Processed	Update test	Copy	ID: GTR000282123

ⓘ 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

GTR Submission

[Home](#) [Contact GTR staff](#) [Help documents](#) [GTR Homepage](#) [My profile](#)

Example submission: SUB704030

Clinical test: Genetic test example for PRA

1 **BASICS** 2 ORDERING 3 INDICATION 4 METHODOLOGY 5 INTERPRETATION 6 PERFORMANCE CHARACTERISTICS 7 REVIEW & SUBMIT

Basics

★ Required field. ✖ Completed field. Hover over ⓘ to display help information.

Test Information

★ Laboratory test name ⓘ	Short test name ⓘ
<input type="text" value="Genetic test example for PRA"/>	<input type="text"/>
Manufacturer's test name, if any ⓘ	<input type="text"/>
Search terms, if any ⓘ	<input type="text" value=""/>
Add another search term	
For definitions of the terms in the list below, please go to Help Documents	
★ Purpose of the test ⓘ	
<input checked="" type="checkbox"/> Diagnosis	
<input type="checkbox"/> Drug Response	
<input type="checkbox"/> Monitoring	
<input type="checkbox"/> Mutation Confirmation	
<input type="checkbox"/> Pre-implantation genetic diagnosis	
<input type="checkbox"/> Pre-symptomatic	
<input type="checkbox"/> Predictive	
<input type="checkbox"/> Prognostic	
<input type="checkbox"/> Recurrence	
<input type="checkbox"/> Risk Assessment	
<input type="checkbox"/> Screening	
<input type="checkbox"/> Therapeutic management	
Target population for this test - Recommended ⓘ	
<input type="text" value="Not Provided"/>	
Enter citation(s) for target population (one on each box) - Recommended ⓘ	
<input type="text" value="Not Provided"/>	
Add another citation for target population (search PubMed) Remove	
Test development - Recommended ⓘ	
<input type="text"/>	
Has there been FDA review of the test? ⓘ	
<input type="radio"/> Yes <input checked="" type="radio"/> No	
FDA category designation - Recommended ⓘ	
<input type="text"/>	

New York State CLEP (NYS CLEP) test approval

Status ⓘ
<input type="text"/>
Test approval # ⓘ
<input type="text"/>

[Save & continue](#)

Example submission: SUB704030

Clinical test: Genetic test example for PRA

- 1 BASICS
- 2 ORDERING
- 3 INDICATION
- 4 METHODOLOGY
- 5 INTERPRETATION
- 6 PERFORMANCE CHARACTERISTICS
- 7 REVIEW & SUBMIT

Ordering

Hover over ⓘ to display help information.

Ordering Information

Test order code (lab code to order this test, ex. for requisition form) - Recommended ⓘ
 Not Provided

URL of the lab website with information about this test - Recommended ⓘ
 Not Provided

URL of the lab website with information about how to order this test - Recommended ⓘ
 Not Provided

How to order (provide a brief explanation about ordering requirements) - Recommended ⓘ
 Not Provided

URL to lab website with information about codes related to this test (ex. CPT, ICD9, ICD10) - Recommended ⓘ
 Not Provided

LOINC code(s) ⓘ
 Not Provided

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 - Recommended

ⓘ 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
Clinical Testing/Confirmation of Mutations Identified Previous		
Add another test-specific laboratory service		

Test-specific laboratory additional services ⓘ

	Order code	Comment
Custom mutation-specific/Carrier testing		
Add another test-specific laboratory additional service		

Specimen source(s) (select all that apply) - Recommended ⓘ

- Amniocytes
- Amniotic fluid
- Bone marrow
- Buccal swab
- Buffy coat
- 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
- Nasal aspirates
- Nasopharyngeal washes
- Oropharyngeal swab
- 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) - Recommended
 Not Provided

Testing strategy (ex. reflex testing) - Recommended ⓘ
 Not Provided

Enter citation(s) for testing strategy (one on each box) - Recommended ⓘ

Not Provided Remove

Add another citation for testing strategy (search PubMed)

Test-specific contact information

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

A M
George Zhou

Contact policy - Recommended ⓘ

- 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

Select AMA CPT Code

Search AMA CPT code

Search

Show all

Codes found:

Select

Cancel

Example submission: SUB704030

Clinical test: Genetic test example for PRA

1 BASICS 2 ORDERING 3 INDICATION 4 METHODOLOGY 5 INTERPRETATION 6 PERFORMANCE CHARACTERISTICS 7 REVIEW & SUBMIT

Indication

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

This test has the following conditions/phenotypes

- ⓘ The primary condition will be used to name the record and link to resources. If conditions are hierarchical, pick the top level name

Condition/Phenotype	Primary	Actions
Cystic fibrosis	<input checked="" type="radio"/>	Edit Delete

* Add other conditions/phenotypes

Limit to:

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

Type Condition/Phenotype to search ⓘ

[Continue](#) to the methodology section

Example submission: SUB704030

Clinical test: Genetic test example for PRA

- 1 BASICS
- 2 ORDERING
- 3 INDICATION
- 4 METHODOLOGY
- 5 INTERPRETATION
- 6 PERFORMANCE CHARACTERISTICS
- 7 REVIEW & SUBMIT

Condition/Phenotype Information

★ Required field. ★ Completed field. Hover over ⓘ to display help 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 ⓘ

Synonyms

Suggest new synonyms

+ Add another synonym

Acronyms

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

Enter citation(s) for prevalence (one on each box) ⓘ

+ Add another citation for prevalence (search PubMed) Remove

Private comment about the condition/phenotype to GTR staff ⓘ

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

Example submission: SUB704030

Clinical test: Genetic test example for PRA

- 1 BASICS
- 2 ORDERING
- 3 INDICATION
- 4 METHODOLOGY
- 5 INTERPRETATION
- 6 PERFORMANCE CHARACTERISTICS
- 7 REVIEW & SUBMIT

Methodology

★ Required field. ★ Completed field. Hover over ⓘ to display help information.

Test Method(s)

Method

★ Major method category
Molecular Genetics

★ Method category
Sequence analysis of select exons

★ Primary Test Methodology
Next-Generation (NGS)/Massively parallel sequencing (MPS)

Instruments - Recommended

- Affymetrix GeneChip Scanner 3000 7G Whole-Genome Asso...
- 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 3730XL HT Capillary Sequencing System

⊕ Add another method

Platforms - Recommended ⓘ

- 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 A...
- Affymetrix Genome-Wide Human SNP Array 6.0
- Affymetrix QuantiGene 2.0 Assay
- Affymetrix Quantiplex Assay

Test procedure or protocol ⓘ

Enter citation(s) for test procedure or protocol (one on each box) ⓘ

Remove

⊕ 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) - Recommended ⓘ

Not Provided

ⓘ 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) ⓘ

Remove

⊕ Add another test comment

★ Test Targets

Please add a test target to continue. Each test only needs one condition-test target relationship.

- Test must have at least one explicit condition-target relationship

Add test target

Example submission: SUB704030

Clinical test: Genetic test example for PRA

1 BASICS 2 ORDERING 3 INDICATION 4 METHODOLOGY 5 INTERPRETATION 6 PERFORMANCE CHARACTERISTICS 7 REVIEW & SUBMIT

Target

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

Target is associated with

Cystic fibrosis

Target is (To enable this section, please select a condition from the list above. Data entry will enhance discovery of your test in GTR.)

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

* Target is identified by ?

gene

Gene ?

CFTR: CF transmembrane conductance regulator

Delete

Add another gene

Additional information

- Associated Reference Sequences and Exons
- Variants
- None

Save target

Cancel

Example submission: SUB704030

Clinical test: Genetic test example for PRA

- 1 BASICS
- 2 ORDERING
- 3 INDICATION
- 4 METHODOLOGY
- 5 INTERPRETATION
- 6 PERFORMANCE CHARACTERISTICS
- 7 REVIEW & SUBMIT

Interpretation

Hover over  to display help information.


Upload Sample Reports

Sample negative report - Recommended 

No file selected.


Sample positive report - Recommended 


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? - Recommended 


What software is used to interpret novel variations? 

What is the laboratory's policy on reporting novel variations? - Recommended 


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


Yes No Decline to answer Not provided

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

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

Yes No Decline to answer Not provided

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

Research performed after clinical testing is complete - Recommended 

Example submission: SUB704030

Clinical test: Genetic test example for PRA

- 1 BASICS 2 ORDERING 3 INDICATION 4 METHODOLOGY 5 INTERPRETATION 6 PERFORMANCE CHARACTERISTICS 7 REVIEW & SUBMIT

Performance Characteristics

★ Required field. ★ Completed field. Hover over ⓘ to display help information.

Availability

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 performance location(s)
Test work In-house At an outside lab
Entire test [checked]
Specimen preparation
Wet lab work
Interpretation
Generate report
Comment on test performance location(s)

★ Analytical validity ⓘ
Analytical specificity is 99%, sensitivity is 98%.
Enter citation(s) to support analytical validity (one on each box)
Not Provided
Add another citation to support analytical validity (search PubMed)
Assay limitation(s) - Recommended ⓘ
Not Provided
Enter citation(s) to support assay limitation(s) (one on each box) - Recommended ⓘ
Not Provided
Add another citation to support assay limitation(s) (search PubMed)

Quality Assurance

Is proficiency testing performed for this test? - Recommended ⓘ
Yes No
Proficiency testing method - Recommended ⓘ
Provider for proficiency testing - Recommended ⓘ
Major CAP category CAP category CAP test list
Add another CAP test
Description of proficiency testing method - Recommended ⓘ
Not Provided
Enter citation(s) to support the above statement (one on each box) - Recommended ⓘ
Not Provided
Add another citation to support proficiency testing method (search PubMed)
Description of internal test validation method - Recommended ⓘ
Not Provided
Enter citation(s) to support the above statement (one on each box) - Recommended ⓘ
Not Provided
Add another citation to support internal test validation method (search PubMed)

Clinical Validity

Statement of clinical validity - Recommended ⓘ
Not Provided
Enter citation(s) to support the above statement (one on each box) - Recommended ⓘ
Not Provided
Add another citation to support clinical validity (search PubMed)

Clinical Utility - Recommended

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
Category of clinical utility
URL to explain the clinical utility
Not Provided
Enter citation(s) to support the clinical utility (one on each box) ⓘ
Not Provided
Remove
Add another citation to support the clinical utility (search PubMed)

Add another clinical utility

Save & continue

Example submission: SUB704030

Clinical test: Genetic test example for PRA

1 BASICS 2 ORDERING 3 INDICATION 4 METHODOLOGY 5 INTERPRETATION 6 PERFORMANCE CHARACTERISTICS 7 REVIEW & SUBMIT

Summary

Submit

Basics

Name Genetic test example for PRA
 Test purpose Diagnosis
 Test-specific licenses License#:

Ordering

Who can order the test Genetic Counselor
 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 Clinical Testing/Confirmation of Mutations Identified Previously
 Test-specific additional services Custom mutation-specific/Carrier testing
 Specimen source(s)
 • Amniocytes
 • Amniotic fluid
 • Bone marrow
 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.

Condition/Phenotype: Cystic fibrosis

Mode of inheritance Autosomal recessive inheritance
 Disease mechanism loss of function

Methodology

Test method(s) Molecular Genetics, Sequence analysis of select exons, Next-Generation (NGS)/Massively parallel sequencing (MPS)

Test target(s)

Target is germline
 Identified by gene: CFTR: CF transmembrane conductance regulator
 Reference Sequence(s)
 Variant(s)

Interpretation

Are family members recruited to assess significance of VUS yes
 Will the lab re-contact the ordering physician if variant interpretation changes yes

Performance characteristics

Test performance location(s) Entire test: internal
 Analytical validity Analytical specificity is 99%, sensitivity is 98%.
 Proficiency testing is performed for this test? no

Submit

ADDING A RESEARCH TEST

GTR Submission

[Home](#) [Contact GTR staff](#) [Help documents](#) [GTR Homepage](#) [My profile](#)

Example submission: SUB704037

Research test: Research test example for PRA

1 **BASICS** 2 PARTICIPATION 3 CONDITION 4 METHODOLOGY 5 REVIEW & SUBMIT

Basics

★ Required field. ★ Completed field. Hover over ⓘ to display help information.

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

Study related to this test

Research study name ⓘ	<input type="text"/>	Short study name ⓘ	<input type="text"/>
ClinicalTrials.gov identifier ⓘ	<input type="text"/>		
URL for the study ⓘ	<input type="text"/>		
If the study is approved by a research ethics committee (e.g., IRB), please provide the protocol number - Recommended ⓘ			
<input type="text" value="Not Provided"/>			
What is the study type? ⓘ			
<input type="radio"/> Interventional study (or Clinical Trial)			
<input type="radio"/> Observational study			
<input type="radio"/> Expanded access			
<input type="radio"/> Not applicable			
★ Study description ⓘ			
<input type="text" value="Example test for screenshots for PRA extension"/>			
Enter citation(s) for study description (one on each box) ⓘ			
<input type="text"/>			
Add another citation for study description (search PubMed) Remove			
Study aims and hypotheses ⓘ			
<input type="text"/>			
Upload study protocol ⓘ			
<input type="button" value="Browse..."/> No file selected.			

Researchers

★ Person responsible for the study ⓘ	<input type="text" value="A M"/>
★ Study contact ⓘ	<input type="text" value="A M"/>
Co-investigator ⓘ	<input type="text"/>
Add another co-investigator	
Research contact policy ⓘ	
<input type="text"/>	

[Save & continue](#)

Example submission: SUB704037

Research test: Research test example for PRA

1 BASICS 2 PARTICIPATION 3 CONDITION 4 METHODOLOGY 5 REVIEW & SUBMIT

Participation

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

Upload participant consent form ?

Browse... No file selected.

Is the study currently recruiting participants? - Recommended ?

- Yes
 No
 Unknown

Eligibility criteria - Recommended ?

Not Provided

Recruitment sites ?

Save & continue

Example submission: SUB704037

Research test: Research test example for PRA

1 BASICS 2 PARTICIPATION 3 **CONDITION** 4 METHODOLOGY 5 REVIEW & SUBMIT

Indication

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

This test has the following conditions/phenotypes

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

* Add 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
 - Mucopolipidosis type IV
 - 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

Example submission: SUB704037

Research test: Research test example for PRA

- 1 BASICS
- 2 PARTICIPATION
- 3 **CONDITION**
- 4 METHODOLOGY
- 5 REVIEW & SUBMIT

Condition/Phenotype Information

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

Synonyms

Suggest new synonyms

+ Add another synonym

Acronyms

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

Enter citation(s) for prevalence (one on each box) ⓘ

 Remove

+ Add another citation for prevalence (search PubMed)

Private comment about the condition/phenotype to GTR staff ⓘ

Example submission: SUB704037

Research test: Research test example for PRA

1 BASICS 2 PARTICIPATION 3 CONDITION 4 METHODOLOGY 5 REVIEW & SUBMIT

Methodology

★ Required field. ★ Completed field. Hover over ⓘ to display help information.

Test Method(s)

Method

★ **Major method category**

Molecular Genetics

★ **Method category**

Deletion/duplication analysis

★ **Primary Test Methodology**

Next-Generation (NGS)/Massively parallel sequencing (MPS)

Instruments - Recommended

Affymetrix GeneChip Scanner 3000 7G Whole-Genome Asso...
Affymetrix GeneTitan® MC
Affymetrix HotStart-HT Probe qPCR Master Mix with UDG (2X)
Agilent 2100 Bioanalyzer
Agilent SureSelect
Applied Biosystems 3730 capillary sequencing instrument
Applied Biosystems 3730XL capillary sequencing instrument

[Add another method](#)

Platforms - Recommended ⓘ

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 A...
Affymetrix Genome-Wide Human SNP Array 6.0
Affymetrix QuantiGene 2.0 Assay

Test procedure or protocol ⓘ

Enter citation(s) for test procedure or protocol (one on each box) ⓘ

Remove

[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) - Recommended ⓘ

Not Provided

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

Remove

[Add another test comment](#)

★ Test Targets

Please add a test target to continue. Each test only needs one condition-test target relationship.

- Test must have at least one explicit condition-target relationship

[Add test target](#)

Example submission: SUB704037

Research test: Research test example for PRA

- 1 BASICS
- 2 PARTICIPATION
- 3 CONDITION
- 4 METHODOLOGY
- 5 REVIEW & SUBMIT

Target

★ Required field. ★ Completed field. Hover over ⓘ to display help information.

Target is associated with

Cystic fibrosis

Target is (To enable this section, please select a condition from the list above. Data entry will enhance discovery of your test in GTR.)

Germline: select for hereditary conditions

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

Both Germline and Somatic

★ **Target is identified by** ⓘ

gene

Gene ⓘ

CFTR: CF transmembrane conductance regulator Delete

Additional information

Associated Reference Sequences and Exons

Variants

None

Variants

Apply to: Cystic fibrosis

Relevant gene variant ⓘ	Clinical significance of variant - Recommended ⓘ	Enter citation(s) to support the clinical significance (one on each box) - Recommended ⓘ
<input type="text"/>	<input type="text"/>	Not Provided Remove
+ Add another variant		+ Add another citation (search PubMed)

Save target Cancel

Example submission: SUB704037

Research test: Research test example for PRA

1 BASICS 2 PARTICIPATION 3 CONDITION 4 METHODOLOGY 5 REVIEW & SUBMIT

Summary

Submit

Basics

Name Research test example for PRA
Test purpose Contribute to generalizable knowledge
Study name
Study Description Example test for screenshots for PRA extension
Person responsible for the study A M
Study contact A M
Contact policy

Participation

Condition/Phenotype: Cystic fibrosis

Mode of inheritance Autosomal recessive inheritance
Disease mechanism loss of function

Methodology

Test method(s) Molecular Genetics, Deletion/duplication analysis, Next-Generation (NGS)/Massively parallel sequencing (MPS)

Test target(s)

Target is germline
Identified by gene: CFTR: CF transmembrane conductance regulator
Reference Sequence(s)
Variant(s) :

Submit