# 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

National Library of Medicine
National Center for Biotechnology Information

Log in

# GTR Submission

Home | Contact GTR staff | Help documents | GTR Homepage

## 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!

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Bethesda, MD 20894

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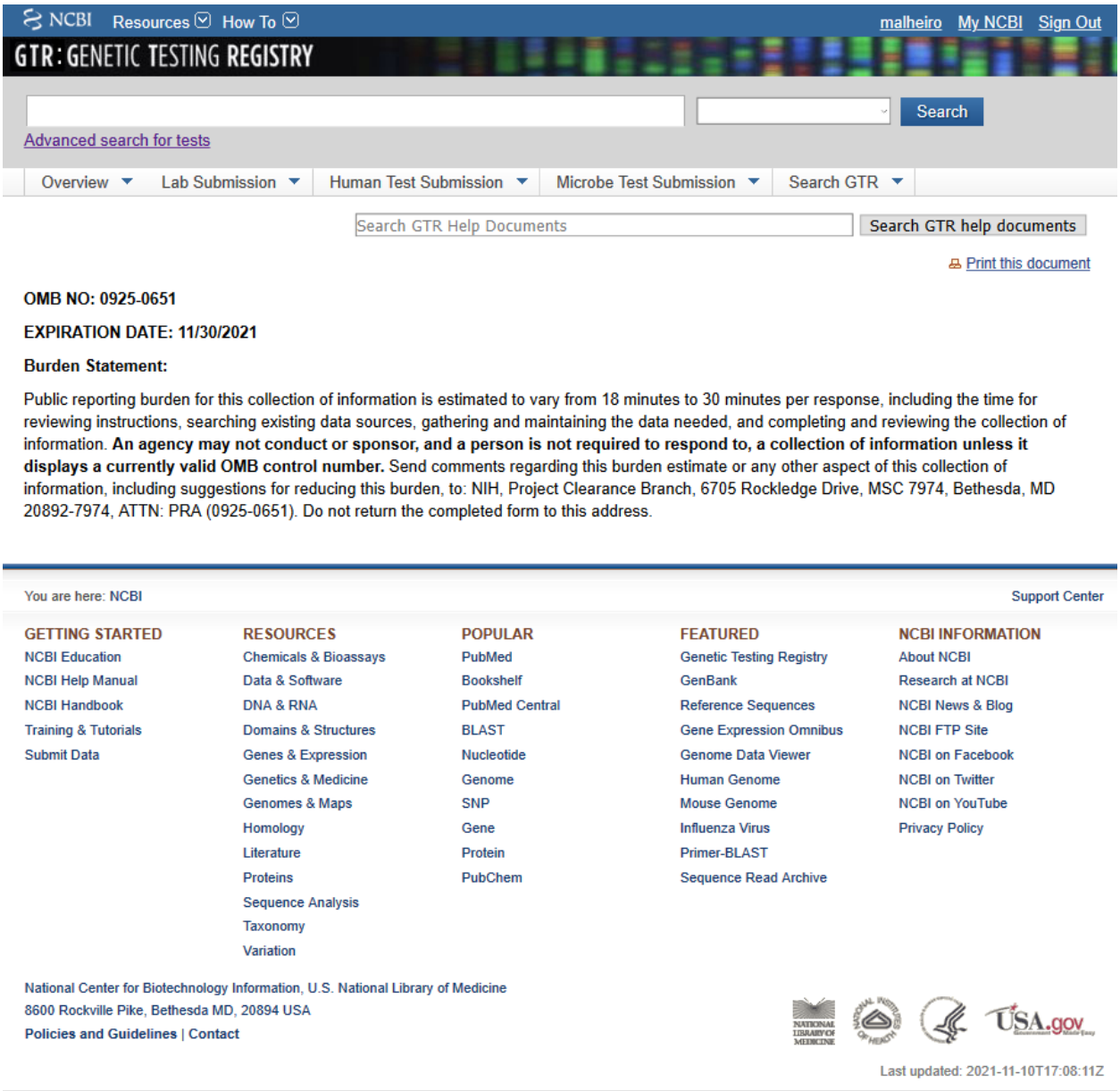
NLM NIH | HHS USA.gov

Last revision: 3.3.1.post45+48bcc36

# BURDEN STATEMENT



NCBI Resources How To malheiro My NCBI Sign Out

GTR: GENETIC TESTING REGISTRY

Search

Advanced search for tests

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

Search GTR Help Documents | Search GTR help documents

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

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NCBI Handbook
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Domains & Structures
Genes & Expression
Genetics & Medicine
Genomes & Maps
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Literature
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Taxonomy
Variation

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Genome
SNP
Gene
Protein
PubChem

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GenBank
Reference Sequences
Gene Expression Omnibus
Genome Data Viewer
Human Genome
Mouse Genome
Influenza Virus
Primer-BLAST
Sequence Read Archive

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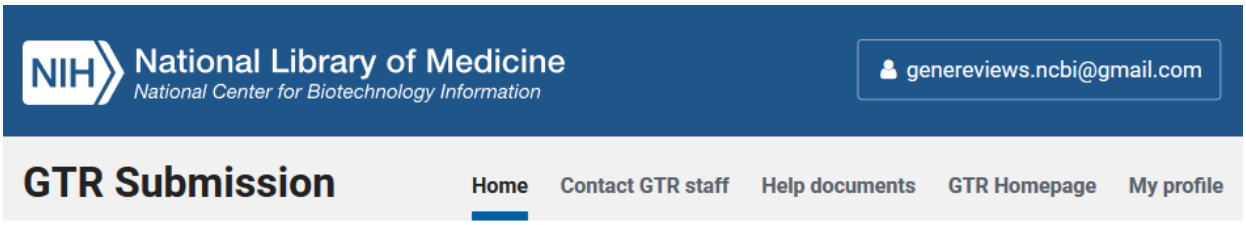
National Center for Biotechnology Information, U.S. National Library of Medicine
8600 Rockville Pike, Bethesda MD, 20894 USA
Policies and Guidelines | Contact

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

# GTR CODE OF CONDUCT




## 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.

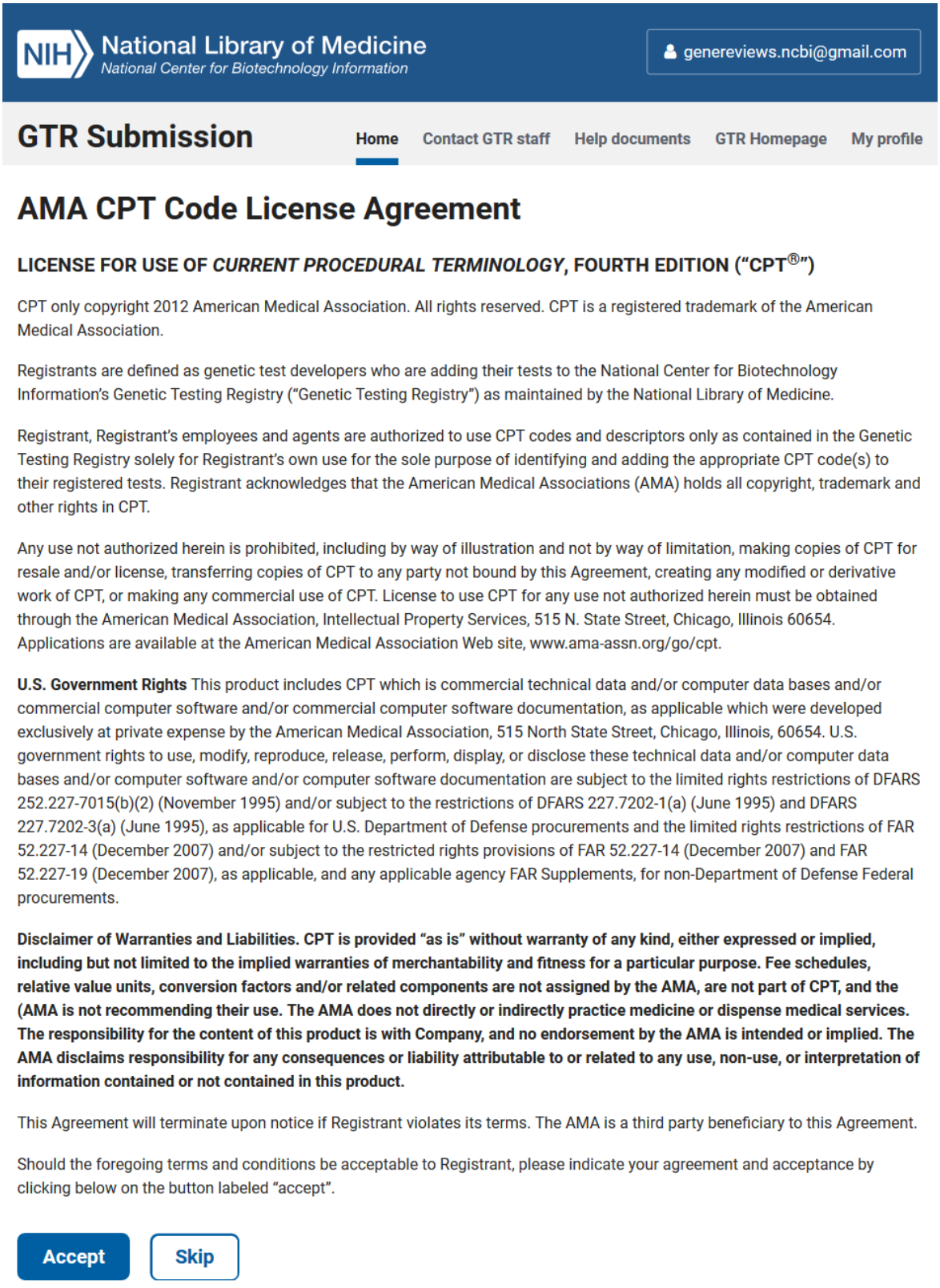
To reference their participation in the GTR, test submitters may refer to the fact that information about their tests is available in the GTR and provide the relevant URL(s) but make no explicit or implicit claims that their tests listed in the GTR, or other information submitted to the GTR, have been 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.

I agree | Disagree

# AMA CPT CODE LICENSE AGREEMENT




## AMA CPT Code License Agreement

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Registrants are defined as genetic test developers who are adding their tests to the National Center for Biotechnology Information's Genetic Testing Registry ("Genetic Testing Registry") as maintained by the National Library of Medicine.

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

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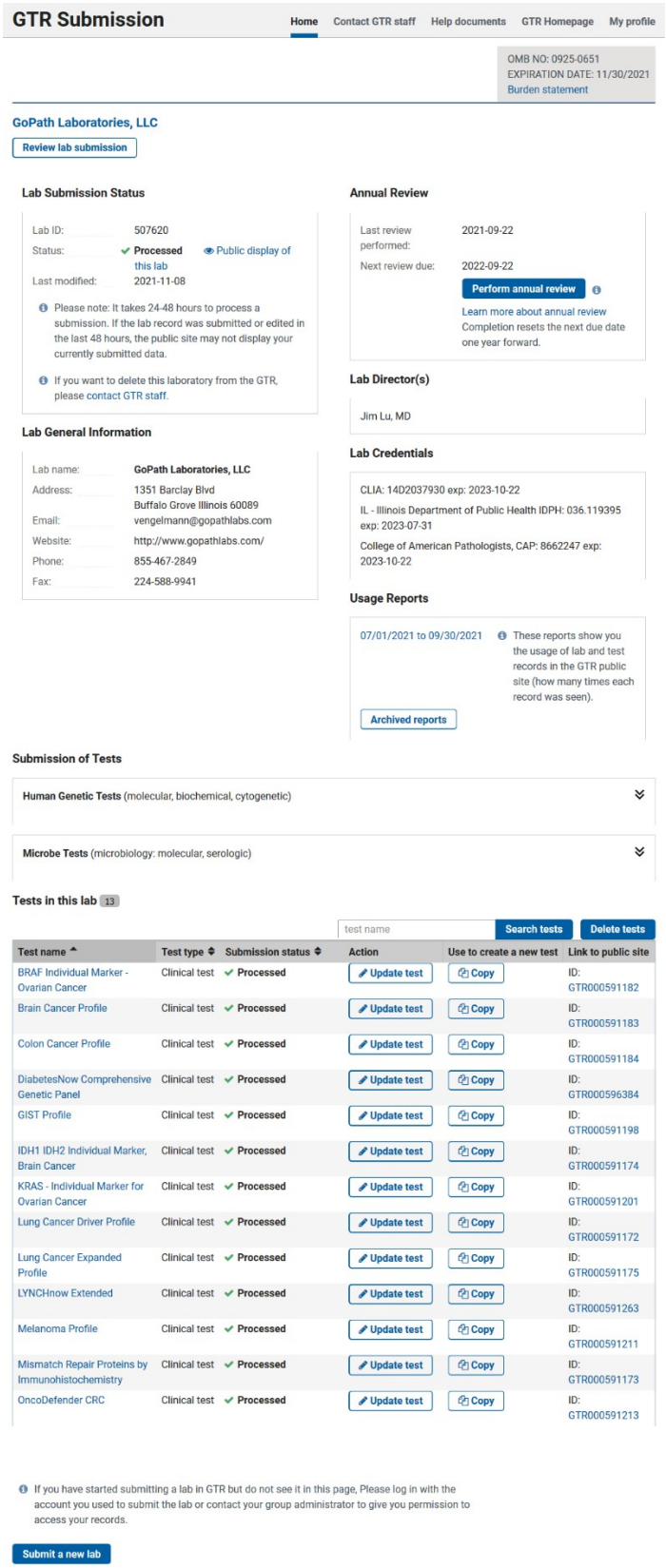
This Agreement will terminate upon notice if Registrant violates its terms. The AMA is a third party beneficiary to this Agreement.

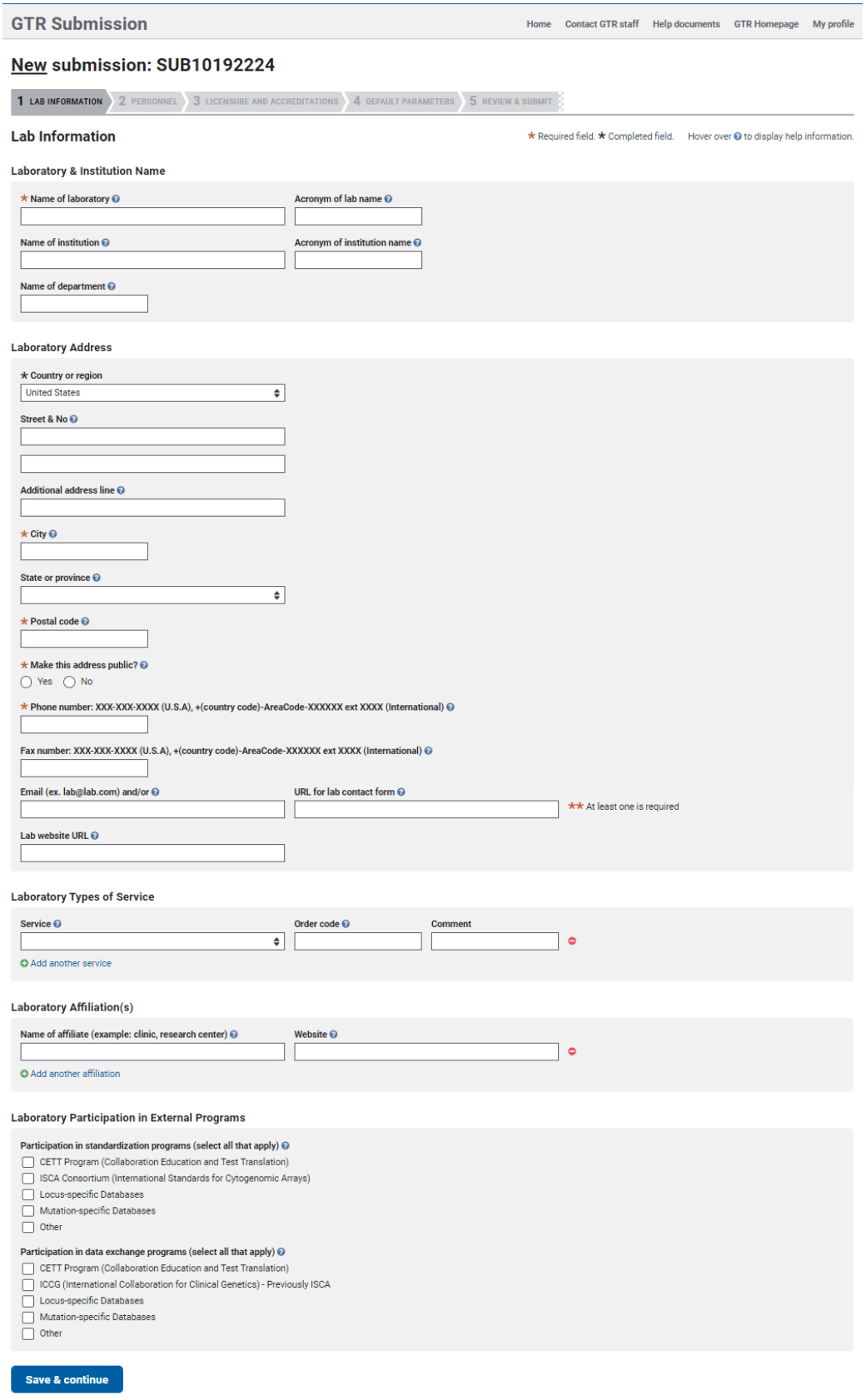
Should the foregoing terms and conditions be acceptable to Registrant, please indicate your agreement and acceptance by clicking below on the button labeled "accept".

Accept Skip

# YOUR LABS IN GTR



# ADD A NEW LAB



GTR Submission

Home | Contact GTR staff | Help documents | GTR Homepage | My profile

**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: United States
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
Add another service

**Laboratory Affiliation(s)**

Name of affiliate (example: clinic, research center) | Website
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. Personel entered here will be available for selection when submitting clinical and research tests. You may specify whether they will display on the public page of the lab.

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

Add research personnel — Research personnel entered here will be available for selection when submitting research tests. These personnel will display on research tests but will not be displayed on the public page of the lab or on clinical tests. If person is entered as staff member, do not resubmit as research personnel.

Continue

GTR Submission

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# 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 ? [Administrator] | Please specify

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

Academic degree

Please specify

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

Institution, City, State/Province, Country

### 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)

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)

Save Cancel

GTR Submission

Home | Contact GTR staff | Help documents | GTR Homepage | My profile

# 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) ?

Add another CLIA certification

Add CLIP certification (applicable to DoD labs only)

### Laboratory State License(s)

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

Add another state license

### Other Certification(s)/License(s) that the Lab Holds

Name of certification/licensing body | License # | Exp. Date (YYYY-MM-DD)

Add another certification/license

Save & continue

# Example submission: SUB348876



## 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**

- [x] Laboratory can only accept contact from health care providers. Patients/families are encouraged to discuss genetic testing options with their health care provider.
- [ ] Post-test email/phone consultation regarding genetic test results and interpretation is provided to patients/families.
- [ ] Pre-test email/phone consultation regarding genetic test results and interpretation is provided to patients/families.

**Who can order this test?**

Genetic Counselor
Health Care Provider
In-State Patients
Licensed Dentist

**How to order (provide a brief explanation about ordering requirements)**

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

| Test-specific laboratory services | Order code | Comment |
|---|---|---|
| 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)**

- [x] Amniocytes
- [x] Amniotic fluid
- [x] 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?**

**What software is used to interpret novel variations?**

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

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

(•) Yes ( ) No ( ) Decline to answer ( ) Not provided

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

(•) Yes ( ) No ( ) Decline to answer ( ) Not provided

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

### Upload Sample Reports

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

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

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

Save & continue

# Example submission: SUB348876



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

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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: 1806001 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

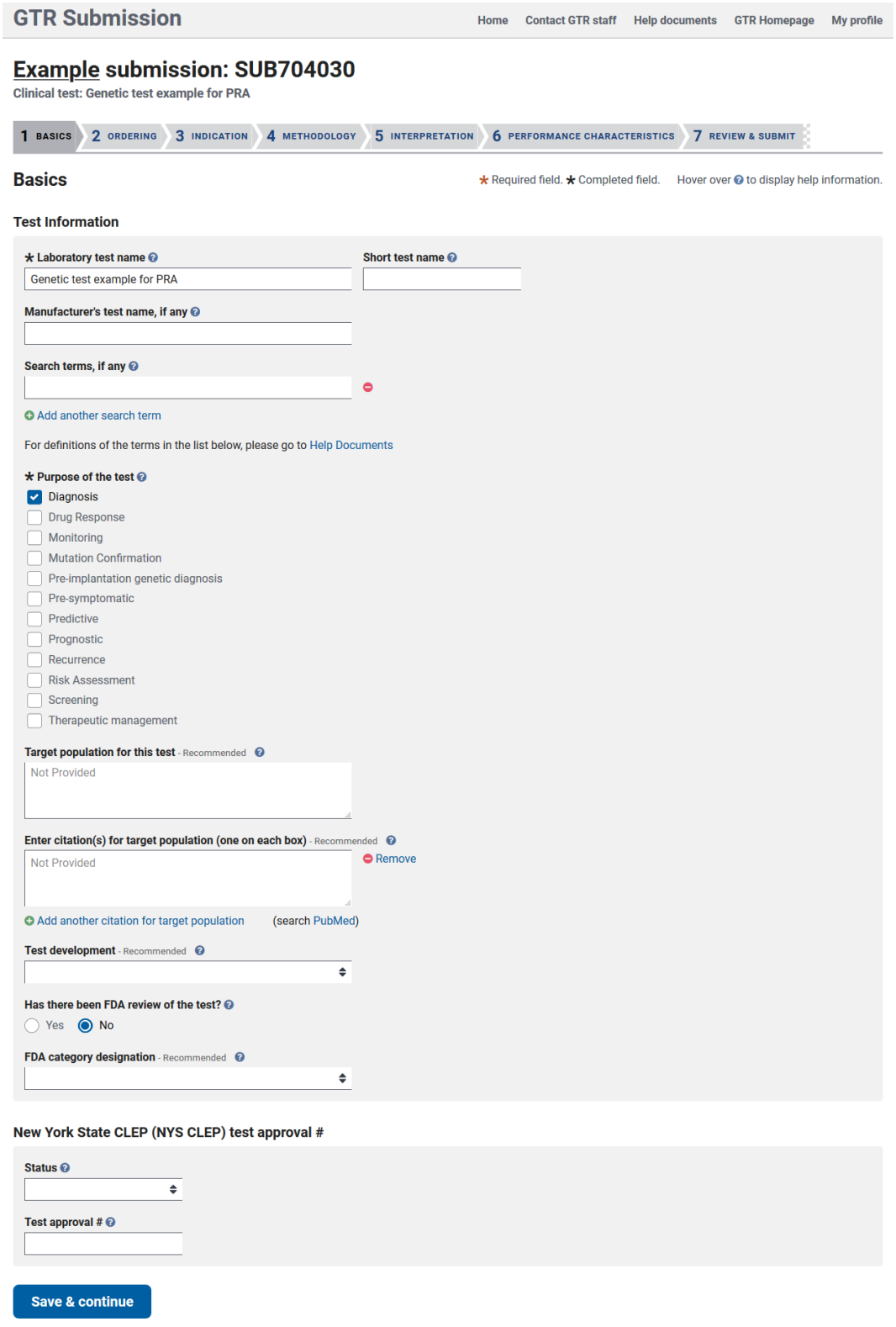
test name | Search tests | Delete tests

| 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: GTR000595973 |
| 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: Genetic test example for PRA

Short test name:

Manufacturer's test name, if any:

Search terms, if any:

Add another search term

For definitions of the terms in the list below, please go to Help Documents

★ Purpose of the test:

- [x] Diagnosis
- [ ] Drug Response
- [ ] Monitoring
- [ ] Mutation Confirmation
- [ ] Pre-implantation genetic diagnosis
- [ ] Pre-symptomatic
- [ ] Predictive
- [ ] Prognostic
- [ ] Recurrence
- [ ] Risk Assessment
- [ ] Screening
- [ ] Therapeutic management

Target population for this test - Recommended: Not Provided

Enter citation(s) for target population (one on each box) - Recommended: Not Provided — Remove

Add another citation for target population (search PubMed)

Test development - Recommended:

Has there been FDA review of the test?

- ( ) Yes
- (•) No

FDA category designation - Recommended:

### New York State CLEP (NYS CLEP) test approval #

Status:

Test approval #:

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)**

**Who can order this test?**

- [x] 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 | ◉ | ○ | ○ | ○ |
| Pre-test genetic counseling | ◉ | ○ | ○ | |
| Post-test genetic counseling | ◉ | ○ | ○ | |

| 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

- [x] Amniocytes
- [x] Amniotic fluid
- [x] 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

- [x] 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

GTR Submission

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# 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 | ◉ | 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** ?

Search

Continue to the methodology section

GTR Submission

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# 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) ?

Remove

Add another citation for prevalence (search PubMed)

Private comment about the condition/phenotype to GTR staff ?

Save | Cancel

# Example submission: SUB704030

Clinical test: Genetic test example for PRA



## 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

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

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



## Interpretation

Hover over ? to display help information.

### Upload Sample Reports

**Sample negative report** - Recommended

Browse... No file selected.

**Sample positive report** - Recommended

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?** - Recommended

Not Provided

**What software is used to interpret novel variations?**

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

Not Provided

**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

Not Provided

**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

Not Provided

**Research performed after clinical testing is complete** - Recommended

Not Provided

Save & continue

# Example submission: SUB704030

Clinical test: Genetic test example for PRA



## 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 | ☑ | |
| 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)** - Recommended

Not Provided — Remove

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 — Remove

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 — Remove

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 — Remove

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 — Remove

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**



## 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<br>• Amniotic fluid<br>• 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

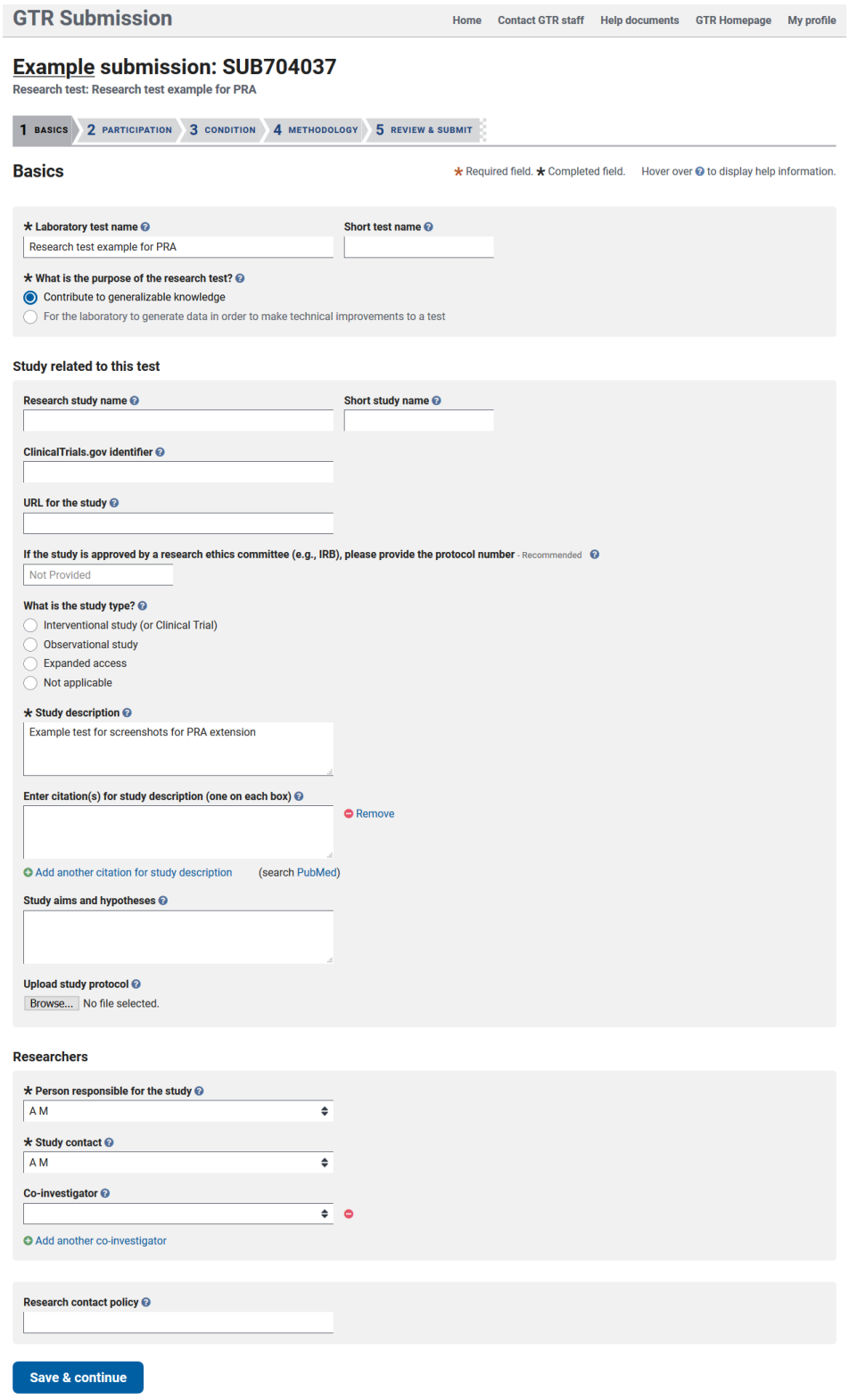
| | |
|---|---|
| Are family members recruited to assess significance of VUS | yes |
| Will the lab re-contact the ordering physician if variant interpretation changes | yes |

### 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



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**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: Research test example for PRA

Short test name:

★ What is the purpose of the research test?

- (•) Contribute to generalizable knowledge
- ( ) For the laboratory to generate data in order to make technical improvements to a test

### Study related to this test

Research study name:

Short study name:

ClinicalTrials.gov identifier:

URL for the study:

If the study is approved by a research ethics committee (e.g., IRB), please provide the protocol number - Recommended

Not Provided

What is the study type?

- ( ) Interventional study (or Clinical Trial)
- ( ) Observational study
- ( ) Expanded access
- ( ) Not applicable

★ Study description: Example test for screenshots for PRA extension

Enter citation(s) for study description (one on each box)

Remove

Add another citation for study description (search PubMed)

Study aims and hypotheses:

Upload study protocol

Browse... No file selected.

### Researchers

★ Person responsible for the study: A M

★ Study contact: A M

Co-investigator:

Add another co-investigator

Research contact policy:

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
  - [ ] Mucolipidosis 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

GTR Submission

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# 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 ?

Save | Cancel

# 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-IT Probe qPCR Master Mix with UDG (2X)
Agilent 2100 Bioanalyzer
Agilent SureSelect
Applied Biosystems 3730 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

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: 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**

- [x] 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 |
|---|---|---|
| | | Not Provided [Remove] |

Add another citation (search PubMed)

Add another variant

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