

# Revised IC Screenshots

---

## Proposed online submission form for the Genetic Testing Registry

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

**2/28/2012**





## SECURE LOGIN SYSTEM

NCBI

**GTR Submission** [Log in](#)

Please [login](#) to create a new submission or to see your existing submissions.

[Copyright](#) | [Disclaimer](#) | [Privacy](#) | [Accessibility](#) | [Contact](#)  
National Center for Biotechnology Information | U.S. National Library of Medicine

  Last Revision: 354643 2012-02-27 18:39:36

## GTR CODE OF CONDUCT

NCBI

**GTR Submission** [Brandi Kattman](#) [Log out](#)

### Code of Conduct

Test submitters providing test information to the Genetic Testing Registry (GTR) agree to abide by a code of conduct. Failure to honor this code of conduct may result in the removal of the submitter's test information from the GTR. Submitters agree to the following terms in the code of conduct:

- To uphold the integrity of the GTR through the submission of information that is accurate and not misleading.
- To assure the accuracy of the data at the time of submission and to review and, if necessary, update the submitted information at least once a year.
- To make no explicit or implicit 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.

[Copyright](#) | [Disclaimer](#) | [Privacy](#) | [Accessibility](#) | [Contact](#)  
National Center for Biotechnology Information | U.S. National Library of Medicine

  Last Revision: 354653 2012-02-28 15:24:28

# YOUR LABS IN GTR

Add a new lab

[Migrate data from GeneTests](#)

## GeneDx

### Lab Submission Status

Submitted, last modified: 4 hours ago. [Review lab submission](#) [Delete this lab](#)

### GeneDx, GeneDx

- o Gaithersburg 118 20877
- o 840
- o Phone: 301-519-2100
- o Fax: 301-519-2892
- o Email: [genedx@genedx.com](mailto:genedx@genedx.com)
- o Website: <http://www.genedx.com/>

### Lab Director(s)

- o Sherri Bale, PhD, Lab Director
- o Anne Maddalena, PhD, Lab Director
- o John Compton, PhD, Lab Director
- o Gabriele Richard, MD, Lab Director
- o Sharon Suchy, PhD, Lab Director
- o Nizar Smaoui, MD, Lab Director
- o Ellen Pfendner, PhD, Lab Director
- o Kathleen Hruska, PhD, Lab Director
- o Swaroop Aradhya, PhD, Lab Director
- o Renkui Bai, MD, Lab Director
- o Daniela Macaya, Lab Director
- o Eden Haverfield, PhD, Lab Director
- o Jeanne Meck, MS, PhD, Lab Director

### Laboratory Affiliations

### Laboratory Credentials

- o CLIA: 21D0969951 exp: 2013-08-28

### Tests

Add a new test

BestTest1	Unfinished	Edit	Delete
None	Unfinished	Edit	Delete
None	Unfinished	Edit	Delete

# ADD A NEW LAB

NCBI

## GTR Submission

Brand Kattman (curator) [Log out](#)

### New

Submission ID: SUB003872

**Lab information** | [Personnel](#) | [Licensure and accreditations](#) | [Default parameters](#) | [Overview](#)

ⓘ \* required field. ⌘ completed field.

#### Laboratory & Institution Name

* Name of laboratory ⓘ	Acronym of lab name ⓘ
<input type="text"/>	<input type="text"/>
GeneTests lab ID, if known ⓘ	
<input type="text"/>	
Name of institution ⓘ	Acronym of institution name
<input type="text"/>	<input type="text"/>
Name of department	
<input type="text"/>	

#### Laboratory Address

* Country or region
<input type="text" value="United States"/>
Street & No ⓘ
<input type="text"/>
<input type="text"/>
Additional address line ⓘ
<input type="text"/>
* City ⓘ
<input type="text"/>
State or province ⓘ
<input type="text" value="Alabama"/>
* Postal code ⓘ
<input type="text"/>
* Make this address public? ⓘ
<input type="radio"/> Yes <input type="radio"/> No
* 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"/>
* Email (ex. lab@lab.com)
<input type="text"/>
Lab website URL ⓘ
<input type="text"/>

#### Laboratory Types of Service

Service ⓘ	Order code ⓘ	<a href="#">remove</a>
<input type="text"/>	<input type="text"/>	
<a href="#">Add another service</a>		

#### Laboratory Affiliation(s)

Name of affiliate (example: clinic, research center)	Website ⓘ	<a href="#">remove</a>
<input type="text"/>	<input type="text"/>	
<a href="#">Add another affiliation</a>		

#### Laboratory Participation in External Programs

Participation in standardization programs (select all that apply) ⓘ
<input type="checkbox"/> CETT Program (Collaboration Education and Test Translation)
<input type="checkbox"/> ISCA Consortium (International Standards for Cytogenomic Arrays)
<input type="checkbox"/> Locus-specific Databases
<input type="checkbox"/> Mutation-specific Databases
<input type="checkbox"/> Other
Participation in data exchange programs (select all that apply) ⓘ
<input type="checkbox"/> CETT Program (Collaboration Education and Test Translation)
<input type="checkbox"/> ISCA Consortium (International Standards for Cytogenomic Arrays)
<input type="checkbox"/> Locus-specific Databases
<input type="checkbox"/> Mutation-specific Databases
<input type="checkbox"/> Other

[Save & Continue](#)

### Generic Genetic Laboratory

Submission ID: SUB003872

[Lab information](#) [Personnel](#) [Licensure and accreditations](#) [Default parameters](#) [Overview](#)

This lab has the following personnel information.

Name	Title	Action
Brandi Kattman	Genetic Counselor	<a href="#">Delete</a> <a href="#">Edit</a>

[Add a person](#) [Continue](#)

Generic Genetic Laboratory

Submission ID: SUB003872

[Lab information](#) **Personnel** [Licensure and accreditations](#) [Default parameters](#) [Overview](#)

\* required field. \* completed field.

Basic Information

\* First name

Middle initial

\* Last name

\* Should this person's information be displayed 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

Academic degree

Professional Certifications

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

Board  Specialty  Subspecialty  [remove](#)

[Add another professional certification](#)

Professional credentials

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. lab@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. lab@lab.com)

Fax number: XXX-XXX-XXXX (U.S.A), +(country code)-AreaCode-XXXXXX ext XXXX (International)

[Cancel](#)

Generic Genetic Laboratory

Submission ID: SUB003872

[Lab information](#) [Personnel](#) [Licensure and accreditations](#) [Default parameters](#) [Overview](#)

Laboratory CLIA Certification

Certification # (e.g. 12D1234567)	Exp. Date (YYYY-MM-DD)	
<input type="text" value="12D1234567"/>	<input type="text" value="2013-03-01"/>	<a href="#">remove</a>

[Add another CLIA certification](#)

Laboratory State License(s)

Qual id	License #	Exp. Date (YYYY-MM-DD)	
<input type="text" value="MD - Maryland Department of Health and Mental Hygiene DHMH"/>	<input type="text" value="123"/>	<input type="text" value="2013-03-01"/>	<a href="#">remove</a>

[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"/>	<a href="#">remove</a>

[Add another certification/license](#)

[Save & Continue](#)



## Generic Genetic Laboratory

Submission ID: SUB003872

[Lab information](#)
[Personnel](#)
[Licensure and accreditations](#)
[Default parameters](#)
[Overview](#)

- In this page you can enter information that is common to many of the tests you will submit. This information will pre-populate the corresponding fields on each test so you do not need to enter the same information multiple times. When you see this information on the test submission page, you can edit it as necessary.

## Optional: Default Parameters (May be overwritten for specific tests)

## Test contact policy

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

## Who can order this test?

Genetic Counselor  
 Health Care Provider  
 In-State Patients  
 Licensed Dentist

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

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

## Test-specific laboratory services

Confirmation of research findings

## Order code

1234

remove

[Add another test-specific laboratory service](#)

## Test-specific laboratory additional services

Custom mutation-specific/Carrier testing

## Order code

5678

remove

[Add another test-specific laboratory additional service](#)

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

- Amniocytes  
 Amniotic fluid  
 Bone marrow  
 Buccal swab  
 Cell culture  
 Chorionic villi  
 Cord blood  
 Cystic hygroma fluid  
 Dried blood spot (DBS) card  
 Fetal blood  
 Fibroblasts  
 Fresh tissue  
 Frozen tissue  
 Paraffin block  
 Peripheral (whole) blood  
 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

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

 Yes  No  Decline to answer

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

## Upload Sample Reports

## Sample negative report

## Sample positive report

## Sample VUS report

## Generic Genetic Laboratory

Submission ID: SUB003872

[Lab information](#) [Personnel](#) [Licensure and accreditations](#) [Default parameters](#) [Overview](#)This lab is ready for submission. [Submit it](#) [Return to homepage](#)**Lab information** ([edit](#))

<b>Name</b>	Generic Genetic Laboratory Acronym: GGL
<b>Institution</b>	
<b>Address</b>	1000 Smith Ave Bethesda Maryland 20824
<b>Phone</b>	800-111-1000
<b>Fax</b>	301-111-1000
<b>Email</b>	genetic@genericgenetics.com
<b>Website</b>	<a href="http://www.genericgenetics.com/">http://www.genericgenetics.com/</a>
<b>Types of service</b>	Confirmation of research findings order code: 1234 DNA Banking order code: 5678
<b>Affiliations</b>	

**Personnel** ([edit](#))

<b>Brandi Kattman</b>	Display this person's information on the GTR public site: yes Primary lab contact Lab director Job title: Genetic Counselor Academic degree: MS Professional certifications: American Board of Genetic Counseling: Genetic counselor Contact information to be displayed on GTR public site: Phone number: 301-318-1671 Email: brandi.kattman@nih.gov Contact information for GTR staff to contact you about the submission: Phone number: 301-318-1671 Email: brandi.kattman@nih.gov
-----------------------	---

**Licensure and accreditations** ([edit](#))

<b>CLIA certification</b>	CLIA: 12D1234567 exp: 2013-03-01
<b>State license(s)</b>	MD - Maryland Department of Health and Mental Hygiene DHMH: 123 exp: 2013-03-01
<b>Other certification(s)</b>	

**Default parameters** ([edit](#))

<b>Test contact policy</b>	Laboratory can only accept contact from health care providers. Patients/families are encouraged to discuss genetic testing options with their health care provider.
<b>Who can order test</b>	Health Care Provider
<b>Test-specific services</b>	Confirmation of research findings order code: 1234
<b>Specimen source(s)</b>	Buccal swab Peripheral (whole) blood

**Tests**This lab is ready for submission. [Submit it](#)

# LIST OF TESTS

Add a new lab

[Migrate data from GeneTests](#)

## GeneDx

### Lab Submission Status

Submitted, last modified: 4 hours ago. [Review lab submission](#) [Delete this lab](#)

### GeneDx, GeneDx

- o Gaithersburg 118 20877
- o 840
- o Phone: 301-519-2100
- o Fax: 301-519-2892
- o Email: [genedx@genedx.com](mailto:genedx@genedx.com)
- o Website: <http://www.genedx.com/>

### Lab Director(s)

- o Sherri Bale, PhD, Lab Director
- o Anne Maddalena, PhD, Lab Director
- o John Compton, PhD, Lab Director
- o Gabriele Richard, MD, Lab Director
- o Sharon Suchy, PhD, Lab Director
- o Nizar Smaoui, MD, Lab Director
- o Ellen Pfendner, PhD, Lab Director
- o Kathleen Hruska, PhD, Lab Director
- o Swaroop Aradhya, PhD, Lab Director
- o Renkui Bai, MD, Lab Director
- o Daniela Macaya, Lab Director
- o Eden Haverfield, PhD, Lab Director
- o Jeanne Meck, MS, PhD, Lab Director

### Laboratory Affiliations

### Laboratory Credentials

- o CLIA: 21D0969951 exp: 2013-08-28

### Tests

Add a new test

BestTest1	Unfinished	<a href="#">Edit</a>	<a href="#">Delete</a>
None	Unfinished	<a href="#">Edit</a>	<a href="#">Delete</a>
None	Unfinished	<a href="#">Edit</a>	<a href="#">Delete</a>

# ADDING A TEST

## New

Submission ID: SUB003874

[Basics](#) [Ordering](#) [Indication](#) [Methodology](#) [Interpretation](#) [Performance characteristics](#) [Overview](#)

\* required field. \* completed field.

### Test Information

\* This test is for

Clinical  Research

\* Laboratory test name

Short test name

Manufacturer's test name, if any

Other test name, if any

Name type

remove

Add another test name

\* Purpose of the test

- Diagnosis  
 Drug Response  
 Mutation Confirmation (family-specific or research results, etc)  
 Pre-implantation genetic diagnosis  
 Pre-symptomatic  
 Risk Assessment  
 Screening

Test development

Platforms

Has there been FDA review of the test?

Yes  No

FDA category designation

### Test-Specific License(s)

Licensed by

License #

Exp. Date (YYYY-MM-DD)

remove

Add another test-specific license

L1CAM Gene Testing for X-linked Hydrocephalus

Submission ID: SUB003874

[Basics](#) [Ordering](#) [Indication](#) [Methodology](#) [Interpretation](#) [Performance characteristics](#) [Overview](#)

\* required field. \* completed field.

Ordering Information

Test order code (lab code to order this test, ex. for requisition form) [?](#)

URL of the lab website with information about this test [?](#)

URL of the lab website with information about how to order this test

How to order (provide a brief explanation about ordering requirements) [?](#)

URL to lab website with information about codes related to this test (ex. CPT, ICD9, ICD10) [?](#)

LOINC code(s) [?](#)  
 [search](#)

Who can order this test? [?](#)

- Genetic Counselor
- Health Care Provider
- In-State Patients
- Licensed Dentist
- Licensed Physician
- Nurse Practitioner
- Out-of-State Patients
- Physician Assistant
- Public Health Mandate
- Registered Nurse

Communication

	Required	Not required	As required by law
Informed consent	<input type="checkbox"/>	<input type="checkbox"/>	<input checked="" type="checkbox"/>
Pre-test genetic counseling	<input type="checkbox"/>	<input checked="" type="checkbox"/>	
Post-test genetic counseling	<input type="checkbox"/>	<input checked="" type="checkbox"/>	

Test-specific laboratory services [?](#)  
 Confirmation of research findings  [remove](#)  
[Add another test-specific laboratory service](#)

Test-specific laboratory additional services [?](#)  
 [remove](#)  
[Add another test-specific laboratory additional service](#)

Specimen source(s) (select all that apply) [?](#)

- Amniocytes
- Amniotic fluid
- Bone marrow
- Buccal swab
- Cell culture
- Chorionic villi
- Cord blood
- Cystic hygroma fluid
- Dried blood spot (DBS) card
- Fetal blood
- Fibroblasts
- Fresh tissue
- Frozen tissue
- Paraffin block
- Peripheral (whole) blood
- Product of conception (POC)
- Saliva
- Serum
- Skin
- Sputum
- Urine
- White blood cell prep
- Other

Specimen requirement URL (ex. collection, handling, transportation)

Testing strategy (ex. reflex testing) [?](#)

Citations for testing strategy [?](#)  
 [remove](#)  
[Add another citation \(search PubMed\)](#)

Test-specific contact information

Contact person [?](#)

- 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.

[Save & Continue](#)

127.0.0.3:3390 - Remote Desktop Connection

Firefox

GTR Submission - GeneDx

https://dssubmit.ncbi.nlm.nih.gov/subs/SUB003874/indication/0

NCBI

GTR Submission — GeneDx Brandi Kattman (curator) Log out

### L1CAM Gene Testing for X-linked Hydrocephalus Submission ID: SUB003874

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

🔴 \* required field. 🟢 \* completed field.

#### Condition/Phenotype Information

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

x-linked hy

- HIGM1: X-Linked Hyper IgM Syndrome
- HSAS: X-linked hydrocephalus syndrome
- XLHR: Familial x-linked hypophosphatemic vitamin D refractory rickets
- XLHR: X-Linked Hypophosphatemia

Acronym to be used for display in the GTR test page, if different from above 🔗

Suggest new acronyms 🔗

Start | VAR-MD: A tool to an... | Inbox - Mailbox - Katt... | McKesson and NCBI fi... | McKesson & NCBI fiel... | ssh.ncbi.nlm.nih.gov -... | 127.0.0.3:3390 - Re... | Submission site scree... | 2:21 PM

## L1CAM Gene Testing for X-linked Hydrocephalus

Submission ID: SUB003874

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

🔗 \* required field. \* completed field.

## Condition/Phenotype Information

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

HSAS: X-linked hydrocephalus syndrome

Note: the condition/phenotype is in [one or more hierarchies](#).

## Indication type

disease

Condition/Phenotype name to be used for display in the GTR test page, if different from above 🔗

## Synonyms

- Aqueductal stenosis, X-linked
- Hydrocephalus due to congenital stenosis of aqueduct of sylvius
- Hydrocephalus, X-linked
- X-Linked Hydrocephalus with Stenosis of the Aqueduct of Sylvius
- X-linked hydrocephalus

## Suggest new synonyms

🔗 Add another synonym

## Preferred acronym

- HSAS

Acronym to be used for display in the GTR test page, if different from above 🔗

## Acronyms

- HSAS1
- HYCX
- XLAS

## Suggest new acronyms 🔗

🔗 Add another acronym

## Mode of inheritance 🔗

X-linked inheritance

## Disease mechanism

loss of function

## Prevalence

## URL for prevalence

## Citations for prevalence

🔗 Add another citation for prevalence (search [PubMed](#))

## Target population for this test 🔗

## Citations for target population

🔗 Add another citation for target population (search [PubMed](#))

## Private comment about the condition/phenotype to GTR staff 🔗

Save Cancel

### L1CAM Gene Testing for X-linked Hydrocephalus

Submission ID: SUB003874

[Basics](#) | [Ordering](#) | [Indication](#) | [Methodology](#) | [Interpretation](#) | [Performance characteristics](#) | [Overview](#)

This test has the following indications. The primary indication will be displayed as part of the test record.

Condition/Phenotype	Primary	Actions
HSAS: X-linked hydrocephalus syndrome	<input checked="" type="radio"/>	<a href="#">Delete</a>   <a href="#">Edit</a>

[Add another indication](#) | [Continue](#)



### L1CAM Gene Testing for X-linked Hydrocephalus

Submission ID: SUB003874

[Basics](#) | [Ordering](#) | [Indication](#) | [Methodology](#) | [Interpretation](#) | [Performance characteristics](#) | [Overview](#)

Target is associated with  
 HSAS: X-linked hydrocephalus syndrome

\* Target is identified by [?](#)

Gene: please specify [?](#)

Please enter  
 Associated Reference Sequences and Exons  
 Variants

#### Associated Reference Sequences and Exons

Associated Reference Sequence  Relevant exons for each associated reference sequence  [remove](#)

[Add another associated reference sequence](#)

#### Sequence data

Associated Reference Sequence

OR

Copy & paste sequence

OR

Upload FASTA file  [Browse...](#)

[Save Target](#) | [Cancel](#)





## L1CAM Gene Testing for X-linked Hydrocephalus

Submission ID: SUB003674

[Basics](#) [Ordering](#) [Indication](#) [Methodology](#) [Interpretation](#) [Performance characteristics](#) [Overview](#)

\* required field. \* completed field.

### Test Method(s)

Method # 1 [remove](#)

- \* Major method category  
Molecular Genetics
- \* Method category  
Sequence analysis of the entire coding region
- \* Primary Test Methodology  
Bi-directional Sanger Sequence Analysis
- Instruments
  - Agilent SureSelect
  - Applied Biosystems 3730 capillary sequencing instrument
  - Applied Biosystems 7900HT Sequence Detection System
  - Applied Biosystems SOLID v4 System Sequencer

[add another method](#)

Test procedure or protocol

Citations for test procedure or protocol

[Add another citation for test procedure or protocol](#) (search [PubMed](#))

Confirmation of test results (ex. how does the lab confirm positive results: using new sample/different method)

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

[Add another test comment](#)

### \* Test Targets

1 target(s) in this test

gene: L1CAM: L1 cell adhesion molecule | HSAS: X-linked hydrocephalus syndrome [Delete](#) [Edit](#)

[Add Test Target](#) | [Add Test Target\(s\) In Batch](#)

[Save & Continue](#)

## L1CAM Gene Testing for X-linked Hydrocephalus

Submission ID: SUB003674

[Basics](#) [Ordering](#) [Indication](#) [Methodology](#) [Interpretation](#) [Performance characteristics](#) [Overview](#)

🔗 \* required field. \* completed field.

## Upload Sample Reports

Sample negative report

Sample positive report

Sample VUS report 🔗

## 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

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

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

 Yes  No  Decline to answer

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

Research performed after clinical testing is complete 🔗

L1CAM Gene Testing for X-linked Hydrocephalus

Submission ID: SLB003874

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

\* required field. \* completed field.

Availability

\* Test performance location(s)

Test work	In-house	Externally	Both in-house & externally
Entire test	<input checked="" type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Specimen preparation	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Wet lab work	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Interpretation	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Generate report	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

Comment on test performance location(s)

\* Analytical validity

Using the method described, this test is 99% sensitive.

Citations to support analytical validity

Add another citation to support analytical validity (search PubMed)

Assay limitation(s)

Citations to support assay limitation

Add another citation to support assay limitation(s) (search PubMed)

Quality Control

Is proficiency testing performed for this test?

Yes No

Proficiency testing method

Inter-Laboratory

Provider for proficiency testing

Major CAP category

CAP category

CAP test list

add another CAP test

Category for my test is not listed here

Description of proficiency testing method

Citations to support the above statement

Add another citation to support proficiency testing method (search PubMed)

Description of internal test validation method

Citations to support the above statement

Add another citation to support internal test validation method (search PubMed)

Clinical Validity

Statement of clinical validity

Citations to support the above statement

Add another citation to support clinical validity (search PubMed)

Clinical Utility

Clinical utility # 1

remove

\* Category of clinical utility  
Establish or confirm diagnosis

URL to explain the clinical utility

Citations to support the clinical utility

Add another citation to support the clinical utility (search PubMed)

add another clinical utility

Save & Continue

## L1CAM Gene Testing for X-linked Hydrocephalus

Submission ID: SUB003874

[Basics](#) [Ordering](#) [Indication](#) [Methodology](#) [Interpretation](#) [Performance characteristics](#) [Overview](#)[Submit](#) [Delete](#)[Return to homepage](#)**Basics**

This test is for	Clinical
Name	L1CAM Gene Testing for X-linked Hydrocephalus
Short name	L1CAM
Test purpose	Diagnosis
Test development	Test developed by laboratory (no manufacturer test name)
FDA category designation	FDA exercises enforcement discretion

**Ordering**

Test order code	7890
Who can order the test	Health Care Provider
Informed consent required	as required by law
Pre-test generic counseling required	no
Post-test generic counseling required	no
Test-specific services	Confirmation of research findings    order code: 1234
Test-specific additional services	
Specimen source(s)	Buccal swab Peripheral (whole) blood
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: HSAS: X-linked hydrocephalus syndrome**

Mode of inheritance	X-linked inheritance
Disease mechanism	loss of function

**Methodology**

Test method(s)	Molecular Genetics, Sequence analysis of the entire coding region, Bi-directional Sanger Sequence Analysis Instruments: Applied Biosystems 3730 capillary sequencing instrument
----------------	---

**Test target(s)**

Identified by	gene: L1CAM: L1 cell adhesion molecule
Reference Sequence(s)	
Variant(s)	

**Interpretation****Performance characteristics**

Test performance location(s)	Entire test: internal
Analytical validity	Using the method described, this test is 99% sensitive.
Proficiency testing is performed for this test?	yes
Proficiency testing method	Inter-Laboratory

[Submit](#) [Delete](#)[Return to homepage](#)

# MIGRATION FROM GENETESTS

## Migrate Lab Data from GeneTests

Based on your login account (gt\_guest), we have identified your registered labs in [GeneTests](#).

2: Skeletal Genetics Laboratory, Cedars-Sinai Medical Center	<a href="#">Migrate Lab Data</a>
255: Laboratory of Dr. Aida Metzberg, California State University, Northridge	<a href="#">Migrate Lab Data</a>
432: Laboratory for Molecular Medicine, Harvard Medical School and Partners Healthcare	<a href="#">Migrate Lab Data</a>
495: Molecular Genetics Laboratory, ARUP Laboratories	<a href="#">Migrate Lab Data</a>
506: GeneDx, GeneDx	<a href="#">View/Edit Lab Submission</a> <a href="#">Migrate Test Data</a>
577: Ambry Genetics, Ambry Genetics Corp	<a href="#">Migrate Lab Data</a>
749: Signature Genomic Laboratories, Signature Genomic Laboratories (a subsidiary of PerkinElmer)	<a href="#">Migrate Lab Data</a>
892: Aldred Lab, Genomic Medicine Institute - Cleveland Clinic	<a href="#">Migrate Lab Data</a>

## Migrate Test Data from GeneTests

- Migrate your lab first
- Download and edit the supplemental data file
- Upload the supplemental data file and migrate the tests

506: GeneDx, GeneDx ([View/Edit Lab Submission](#))

- 17-alpha-Hydroxylase-Deficient Congenital Adrenal Hyperplasia
  - 17-Beta-Hydroxysteroid Dehydrogenase X Deficiency
  - 22q11.2 Deletion Syndrome
  - 22q11.2 Duplication
  - 3-Hydroxy-3-Methylglutaryl-CoA Synthase 2 Deficiency
  - 3-Hydroxy-3-Methylglutaryl-Coenzyme A Lyase Deficiency
  - 3-Hydroxyacyl-Coenzyme A Dehydrogenase Deficiency
  - 3-Methylglutaconic Aciduria Type 1
  - 3-Methylglutaconic Aciduria Type 2
  - 3-Methylglutaconic Aciduria Type 3
  - 3-Methylglutaconic Aciduria Type 5
  - 46,XX Testicular Disorder of Sex Development
  - 6-Pyruvoyltetrahydropterin Synthase Deficiency
  - ABCA12-Related Autosomal Recessive Congenital Ichthyosis
  - Williams Syndrome
  - Williams-Beuren Region Duplication Syndrome
  - Wolf-Hirschhorn Syndrome
  - X-Linked Agammaglobulinemia
  - X-Linked Hypophosphatemia
  - X-Linked Juvenile Retinoschisis
  - X-Linked Leigh Syndrome
  - X-Linked Severe Combined Immunodeficiency
  - X-Linked Sideroblastic Anemia and Ataxia
  - Zaspopathy
  - ZIC2-Related Holoprosencephaly
- Select All

[Download](#) supplemental data template as a tab-delimited-values file.

Upload supplemental data file (tab-delimited format):

[Browse...](#)

[Migrate Selected Test\(s\) - Interactive Mode](#)

[Validate and Migrate using Supplemental Data File - Batch Mode](#)

[Back](#)

- KRT1-Related Epidermolytic Hyperkeratosis
- KRT6A-Related Pachyonychia Congenita
- KRT6B-Related Pachyonychia Congenita
- L1 Syndrome
- L-2-Hydroxyglutaric Aciduria
- Lactate Dehydrogenase B Deficiency
- LAMA3-Related Junctional Epidermolysis Bullosa

[View/Edit Test Submission](#)