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

GTR Submission

Log in

Please login to create a new submission or to see your existing submissions.

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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.

I agree   Disagree

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

YOUR LABS IN GTR

GTR Submission

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

- Gaithersburg 118 20877
- 840

- Phone: 301-519-2100
- Fax: 301-519-2892
- Email: genedx@genedx.com
- Website: http://www.genedx.com/

Lab Director(s)

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

Laboratory Affiliations

Laboratory Credentials

- 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

GTR Submission

New

Submission ID:SUB003872

| Lab information | Personnel | Licensure and accreditations | Default parameters | Overview |

❷ ✱ required field. ✱ completed field.

Laboratory & Institution Name

✱ Name of laboratory ❷

Acronym of lab name ❷

GeneTests lab ID, if known ❷

Name of institution ❷

Acronym of institution name

Name of department

Laboratory Address

✱ Country or region
United States

Street & No ❷

Additional address line ❷

✱ City ❷

State or province ❷
Alabama

✱ Postal code ❷

✱ Make this address public? ❷
○ 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)

Lab website URL ❷

Laboratory Types of Service

Service ❷

Order code ❷   ⊖ remove

⊕ Add another service

Laboratory Affiliation(s)

Name of affiliate (example: clinic, research center)

Website ❷   ⊖ remove

⊕ Add another affiliation

Laboratory Participation in External Programs

Participation in standardization programs (select all that apply) ❷
☐ 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) ❷
☐ CETT Program (Collaboration Education and Test Translation)
☐ ISCA Consortium (International Standards for Cytogenomic Arrays)
☐ Locus-specific Databases
☐ Mutation-specific Databases
☐ Other

Save & Continue

USA.gov

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GTR Submission

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	Delete Edit

[Add a person] [Continue]

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GTR Submission

Generic Genetic Laboratory

Submission ID:SUB003872

Lab information | Personnel | Licensure and accreditations | Default parameters | Overview

❷ ✱ required field. ✱ completed field.

Basic Information

✱ First name

Middle initial

✱ Last name

✱ Should this person's information be displayed on the GTR public site? ❷
⦿ Yes   ○ No

✱ Is this person the primary lab contact? ❷
○ Yes   ⦿ No

✱ Is this person a lab director? ❷
○ Yes   ⦿ No

Job title ❷

```
Administrator
Genetic Counselor
Lab Associate Director
Lab Director
```

Academic degree ❷

```
MD
PhD
MS
RN
```

Professional Certifications

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

Board	Specialty	Subspecialty
▾	▾	▾ ⊖ remove

⊕ Add another professional certification

Professional credentials
```
American Academy of Cosmetic Surgery, FACRM
American Academy of Dermatology, FAAD
American Academy of Family Physicians, FAAFP
American Academy of Neurology, FAAN
```

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)

Save   Cancel

USA.gov

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)
`12D1234567`   `2013-03-01`   ⊖ remove

⊕ Add another CLIA certification

Laboratory State License(s)

Qual id

`MD - Maryland Department of Health and Mental Hygiene DHMH ▾`   License # `123`   Exp. Date (YYYY-MM-DD) ❓ `2013-03-01`   ⊖ remove

⊕ Add another state license

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

Name of certification/licensing body

`▾`   License #   Exp. Date (YYYY-MM-DD)   ⊖ remove

⊕ Add another certification/license

[Save & Continue]

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GTR Submission

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 ❓   Order code ❓
Confirmation of research findings   1234   ⊖ remove
⊕ Add another test-specific laboratory service

Test-specific laboratory additional services ❓   Order code ❓
Custom mutation-specific/Carrier testing   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 ❓
Browse..
Sample positive report ❓
Browse..
Sample VUS report ❓
Browse..

Save & Continue

GTR Submission

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)

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

Personnel (edit)

Brandi Kattman

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)

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

Default parameters (edit)

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

Tests

This lab is ready for submission. Submit it

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LIST OF TESTS

GTR Submission   Brandi Kattman Log out

[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

- Gaithersburg 118 20877
- 840

- Phone: 301-519-2100
- Fax: 301-519-2892
- Email: genedx@genedx.com
- Website: http://www.genedx.com/

Lab Director(s)

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

Laboratory Affiliations

Laboratory Credentials

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

Tests

[Add a new test]

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

ADDING A TEST

GTR Submission — GeneDx

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**
`L1CAM Gene Testing for X-linked Hydrocephalus`

Short test name ❓
`L1CAM`

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 ❓
`Test developed by laboratory (no manufacturer test name)  ▼`

Platforms
```
Affymetrix GeneChip
Affymetrix QuantiGene 2.0 assay
Agilent microarrays
Amersham Codelink UniSet Human I Bioarrays
```

Has there been FDA review of the test? ❓
- ○ Yes   ⦿ No

FDA category designation ❓
`FDA exercises enforcement discretion  ▼`

Test-Specific License(s)

Licensed by
`[          ▼]`

License # ❓
`[            ]`

Exp. Date (YYYY-MM-DD)
`[            ]` ⊖ remove

⊕ Add another test-specific license

[Save & Continue]

GTR Submission — GeneDx

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) ❓
`7890`

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	○	○	◉
Pre-test genetic counseling	○	◉	
Post-test genetic counseling	○	◉	

Test-specific laboratory services ❓
`Confirmation of research findings ▾`

Order code ❓
`1234`   ⊖ remove

⊕ Add another test-specific laboratory service

Test-specific laboratory additional services ❓
`_____ ▾`

Order code ❓
`_____`   ⊖ 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
`_____`   ⊖

⊕ Add another citation (search PubMed)

Test-specific contact information

Contact person ❓
```
Sherri Bale
Anne Maddalena
John Compton
Gabriele Richard
```

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

USA.gov

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Firefox

GTR Submission - GeneDx

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

Google

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 ❓

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

GTR Submission — GeneDx

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	⦿	Delete Edit

Add another indication   Continue

GTR Submission — GeneDx

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 ▼]

Gene: please specify ❓
[L1CAM: L1 cell adhesion molecule]

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

GTR Submission — GeneDx

L1CAM Gene Testing for X-linked Hydrocephalus

Submission ID:SUB003874

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

GTR Submission — GeneDx

L1CAM Gene Testing for X-linked Hydrocephalus

Submission ID:SUB003874

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

❷ ✱ required field. ✱ completed field.

Upload Sample Reports

Sample negative report

[   ] Browse..

Sample positive report

[   ] Browse..

Sample VUS report ❷

[   ] Browse..

Variants of Unknown Significance (VUS): Policy and Interpretation

What is the protocol for interpreting a variation as a VUS? ❷

[   ]

What software is used to interpret novel variations? ❷

[   ]

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

[   ]

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

○ 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 ❷

[   ]

Research performed after clinical testing is complete ❷

[   ]

Save & Continue

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GTR Submission—GeneDx

L1CAM Gene Testing for X-linked Hydrocephalus

Submission ID:SUB003874

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	☑	☐	☐
Specimen preparation	☐	☐	☐
Wet lab work	☐	☐	☐
Interpretation	☐	☐	☐
Generate report	☐	☐	☐

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
[▼]   [▼]   [▼]   ⊖ remove

⊕ 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]

GTR Submission — GeneDx

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

GTR Submission

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

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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]

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- ☐ KRT1-Related Epidermolytic Hyperkeratosis
- ☐ KRT6A-Related Pachyonychia Congenita
- ☐ KRT6B-Related Pachyonychia Congenita
- ☐ L1 Syndrome   View/Edit Test Submission
- ☐ L-2-Hydroxyglutaric Aciduria
- ☐ Lactate Dehydrogenase B Deficiency
- ☐ LAMA3-Related Junctional Epidermolysis Bullosa