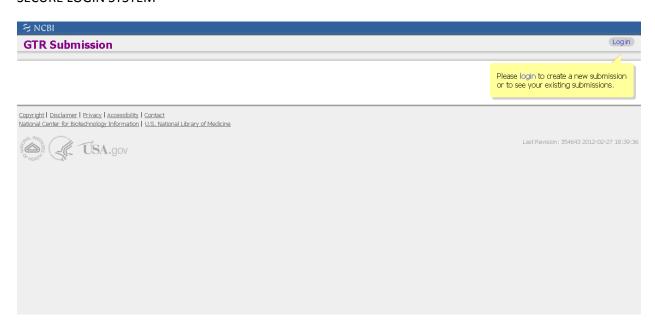
Revised IC Screenshots

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 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.

I agree Disagree



YOUR LABS IN GTR

SNCBI

GTR Submission

Brandi Kattman (Log out)

Add a new lab

Migrate data from GeneTests

<u>GeneDx</u>

Lab Submission Status

Submitted, last modified: 4 hours ago. Review lab submission Delete this lab

- o Gaithersburg 118 20877
- o 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 Sudvy, PhD, Lab Director
 Nizar Smaoui, MD, Lab Director
 Rizar Smaoui, MD, Lab Director
 Ellen Pfendher, 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

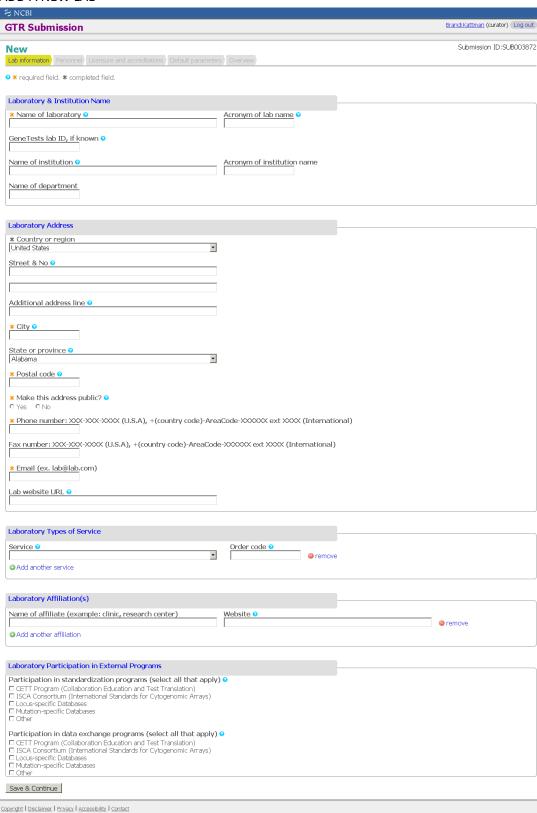
o CLIA: 21D0969951 exp: 2013-08-28

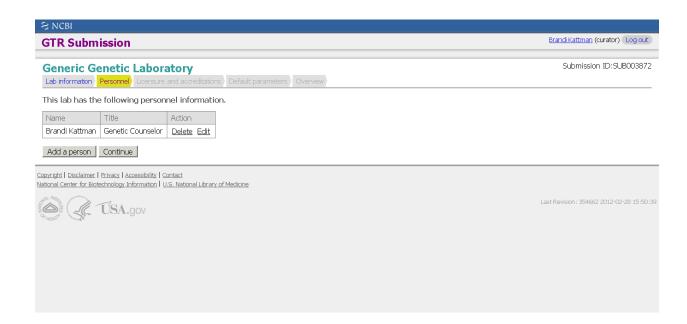
Tests

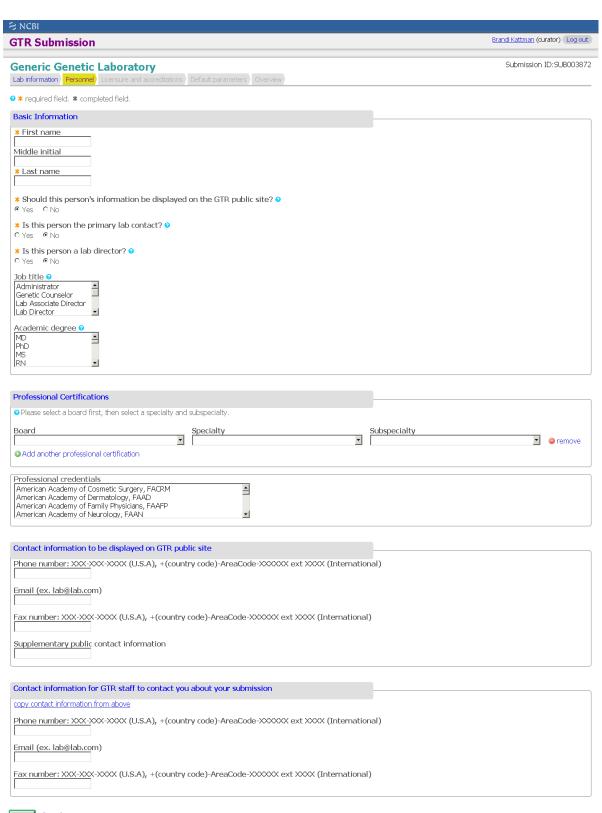
Add a new test

BestTest1	Unfinished	<u>Edit</u>	<u>Delete</u>
None	Unfinished	<u>Edit</u>	<u>Delete</u>
None	Unfinished	Edit	Delete

ADD A NEW LAB

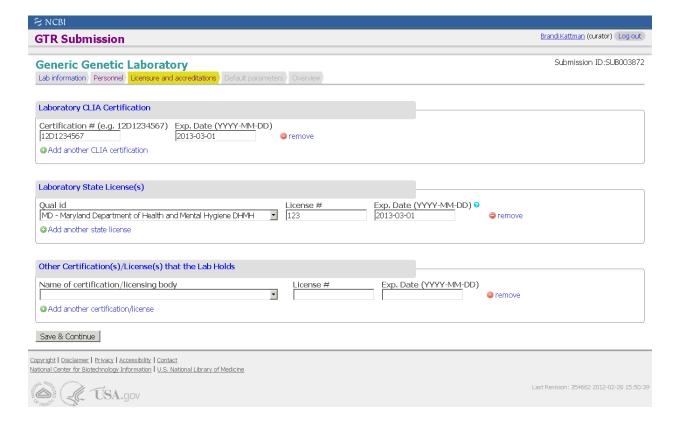






Save Cancel





Upload Sample Reports Sample negative report Browse. Sample positive report Browse. Sample VUS report Browse.

Save & Continue



S NCBI

Brandi Kattman (curator) Log out **GTR Submission**

Generic Genetic Laboratory

Lab information Personnel Licensure and accreditations Default parameters Overvie

Submission ID:SUB003872

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 301-111-1000

Fmail genetic@genericgenetics.com Website http://www.genericgenetics.com/

Confirmation of research findings order code: 1234 DNA Banking order code: 5678 Types of service

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 American both of centuc counseling: centuit 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



Add a new lab Migrate data from GeneTests

<u>GeneDx</u>

Lab Submission Status

Submitted, last modified: 4 hours ago. Review lab submission Delete this lab

- o Gaithersburg 118 20877 o 840

- Phone: 301-519-2100
 Fax: 301-519-2892
 Email: genedx@genedx.com
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Lab Director(s)

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Laboratory Affiliations

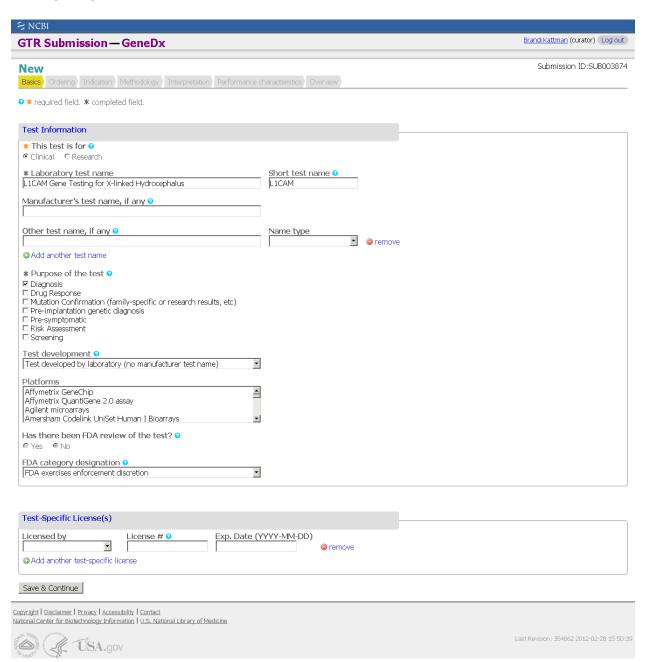
Laboratory Credentials

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

Add a new test

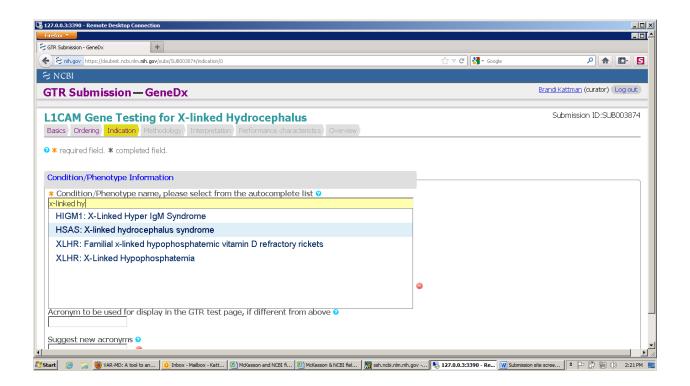
BestTest1	Unfinished	<u>Edit</u>	<u>Delete</u>
None	Unfinished	Edit	<u>Delete</u>
None	Unfinished	Edit	Delete

ADDING A TEST









Save <u>Cancel</u>

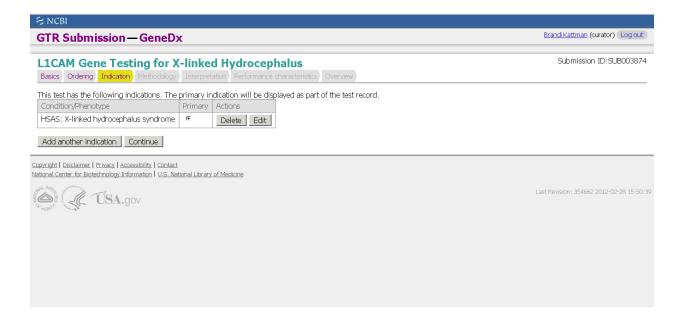
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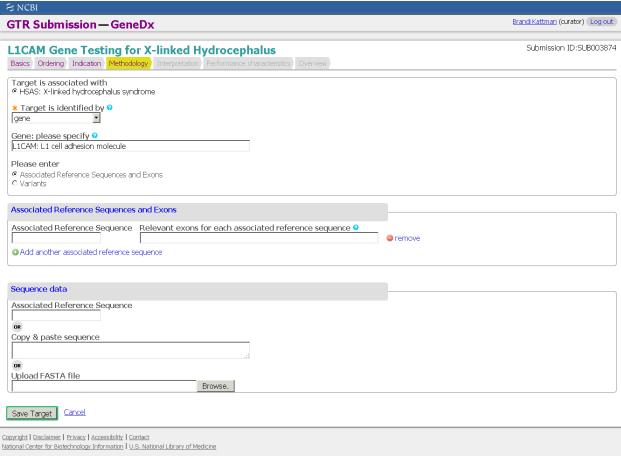
Add another citation for target population (search <u>PubMed</u>)
Private comment about the condition/phenotype to GTR staff @



Target population for this test 0

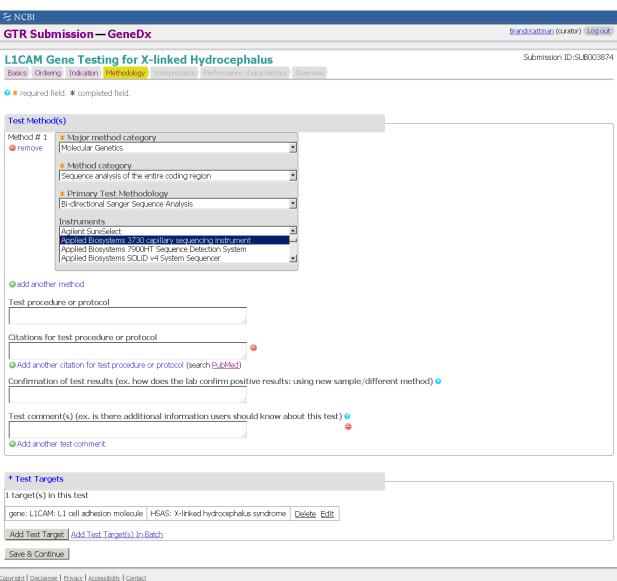
Citations for target population





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F NCBI	
GTR Submission — GeneDx	Brandi Kattman (curator) Log out
L1CAM Gene Testing for X-linked Hydrocephalus Basics Ordering Indication Methodology Interpretation Performance characteristics Overview	Submission ID:SUB003874
∗ required field. * completed field.	
Upload Sample Reports	ļ
Sample negative report Browse	
Sample positive report Browse	
Sample VUS report Browse.	
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 charging of the constant of the control of th	arge? ②
Comments about recruiting family members to assess significance of VUS without charge	
Will the lab re-contact the ordering physician if variant interpretation changes? C Yes C No C Decline to answer	
Comments about the laboratory procedure to re-contact the ordering physician iii	
Research performed after clinical testing is complete	

Save & Continue

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S NCBI

GTR Submission — GeneDx

Brandi Kattman (curator) Log out

L1CAM Gene Testing for X-linked Hydrocephalus

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

Submission ID:SUB003874

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 Health Care Provider test

Informed consent required as required by law no

Pre-test generic counseling required Post-test generic counseling required

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 Reference Sequence(s)

gene: L1CAM: L1 cell adhesion molecule

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? Proficiency testing method

Inter-Laboratory

Submit Delete

Return to homepage

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MIGRATION FROM GENETESTS

SNCBI

GTR Submission

Brandi Kattman (Log out)

Migrate Lab Data from GeneTests

Based on your login account (gt_guest), we have identified your registered labs in <u>GeneTests</u>.

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



Brandi Kattman Log out **GTR Submission**

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
- $\ \square$ 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
- $\hfill\Box$ 3-Methylglutaconic Aciduria Type 5
- ☐ 46,XX Testicular Disorder of Sex Development
- ☐ 6-PyruvoyItetrahydropterin 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...

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- □ KRT1-Related Epidermolytic Hyperkeratosis
- □ KRT6A-Related Pachyonychia Congenita
- $\hfill \square$ KRT6B-Related Pachyonychia Congenita
- □ L1 Syndrome
- □ L-2-Hydroxyglutaric Aciduria
- ☐ Lactate Dehydrogenase B Deficiency
- □ I AMAR-Related 1 inctional Enidermolysis Bullosa

View/Edit Test Submission