Lab Dept: Anatomic Pathology

Test Name: X-LINKED HYDROCEPHALUS (L1CAM) KNOWN MUTATION

General Information

Lab Order Codes: XLHK (Blood)

Synonyms: L1CAM Gene

CPT Codes: 81403 – Molecular Pathology Level 4

Test Includes: X-Linked Hydrocephalus (L1CAM Gene) and related syndrome specific mutation testing in situations such as prenatal diagnosis, carrier testing and pre-symptomatic testing on family members at risk for the mutation. There must be a confirmed mutation analysis on an individual to proceed with this testing.

Logistics

Test Indications: Once a mutation is confirmed in an affected individual, prenatal diagnosis, carrier testing, and presymptomatic testing can be performed on family members who are at risk for the mutation. In some cases, particularly those with time constraints such as prenatal diagnosis, mutation confirmation in an affected family member can run concurrently with other samples from the same family.

Reasons for referral:
1. Clinical laboratory confirmation of one or more mutations identified in a research lab.
2. Carrier testing in female relatives
3. Genetic counseling and risk assessment
4. Prenatal diagnosis in at-risk pregnancies

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: GeneDx, Inc.(GDX Test: 9011)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours. Specimens collected Saturday or Sunday will be held for shipment on Monday.

Turnaround Time: 2 - 4 weeks

Special Instructions:	A GeneDx request form must be sent with any patient or specimen to the laboratory.

Specimen

Specimen Type:	Whole blood
Container:	Lavender top (EDTA) tube
Draw Volume:	3 - 5 mL (Minimum: 1 mL) blood
Processed Volume:	Same as Draw Volume
Collection:	Routine venipuncture. Mix specimen thoroughly by gentle inversion.
Special Processing:	Lab Staff: Do Not centrifuge: Ship whole blood in original collection container, with a cool pack in warm temperatures, via overnight or second-day courier so that the sample will arrive at GeneDx Monday through Saturday. Note: Specimens can be held at refrigerated temperature for up to 7 days.
Patient Preparation:	None
Sample Rejection:	Unrefrigerated specimens older than 48 hours; frozen specimens; mislabeled or unlabeled specimens

Interpretive

Reference Range:	No mutations detected
Critical Values:	N/A
Limitations:	In the group of patients with a positive family history and more than one typical associated finding for L1CAM-associated disease, the detection rate of mutations is greater than 90%. Buccal specimens are not accepted for this testing.
Methodology:	Capillary sequencing
References:	GeneDx, Inc.March 2018 (301) 519-2100 Fax: (301) 519-2892
Updates:	2/11/2013: CPT update 3/7/2018: CPT update