Lab Dept: Anatomic Pathology

Test Name: X-LINKED HYDROCEPHALUS (L1CAM) SEQUENCING IN MALES

General Information

Lab Order Codes: XLH

Synonyms: L1CAM Gene; MASA Syndrome; CRASH Syndrome; L1 Adhesion Molecule; L1 Syndrome

CPT Codes: 81407 – Molecular Pathology, Level 8

Test Includes: **In males**, analysis is performed by bi-directional sequencing of all 28 coding exons and their exon/intron splice junctions of the L1CAM gene. Large deletions of one or more exons are detectable by sequencing in males; however, partial gene duplications would not be identified by sequencing. Targeted array CGH analysis with exon-level resolution is available to evaluate for partial gene duplication.

Logistics

Test Indications: This group of related neurological syndromes (X-linked Hydrocephalus, MASA syndrome, X-linked spastic paraplegia type 1) are allelic and are due to mutation in the L1CAM gene. Congenital hydrocephalus and resultant macrocephaly due to stenosis of the aqueduct of Sylvius may occur in isolation but is frequently associated with other features including hypoplastic or flexed, adducted thumbs. Patients are mentally retarded and have spastic paraplegia.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: GeneDx, Inc.(GDX Test: 2251)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours (Preferred draws are Sunday – Friday) as specimens can only be received at the reference lab Monday - Saturday. Specimens collected Saturday will be held for shipment on Sunday or Monday.)

Turnaround Time: Approximately 9 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 blood
Processed Volume:	Same as Draw Volume
Collection:	Routine venipuncture for blood specimens
Special Processing:	Lab Staff: Send whole blood in original collection container, including signed consent form and requisition, with a cool pack in warm weather, via overnight or second-day courier so that the sample will arrive at GeneDx, Inc. Monday through Saturday. Samples drawn on Saturday should be held at refrigerated temperatures for shipment on Sunday or Monday. **Do not** freeze. Note: Specimens can be held for 7 days at refrigerated temperatures.
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 acceptable for this testing.
Methodology:	Bi-directional sequence analysis
References:	GeneDx, Inc. October 2012 (301) 519-2100 Fax (301) 519-2892
Updates:	2/11/2013: CPT update 3/26/2014: CPT update