Lab Dept: Anatomic Pathology

Test Name: X-LINKED ADRENOLEUKODYSTROPHY (ABCD1) SEQUENCING

**General Information**

Lab Order Codes: ABCD

Synonyms: ABCD1 Sequencing

CPT Codes: 81405 – ABCD1, full gene sequence (Molecular Pathology, Level 6)
81479 – Molecular Pathology Unlisted procedure (MLPA)

Test Includes: ABCD1 gene sequencing

**Logistics**

Test Indications: Confirm diagnosis of X-Linked Adrenoleukodystrophy (X-ALD).

Identification of causative mutations in known or highly suspicious cases of X-ALD, or adrenomyeloneuropathy (AMN) with elevated plasma very long chain fatty acid levels (VLCFA); rule out an ABCD1 defect in the presence of equivocal clinical presentation and/or plasma VLCFA levels; targeted carrier testing of relatives of proband; predictive prenatal testing when familial mutation is known.

Lab Testing Sections: Anatomic Pathology – Sendouts

Referred to: Johns Hopkins DNA Diagnostic Laboratory, Baltimore, MD

Phone Numbers: MIN Lab: 612-813-6280
STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: Results are reported in 4 weeks

Special Instructions: Include a completed Johns Hopkins Form with the specimen or patient to the laboratory. Please include copies of the patient’s biochemical analysis with the sample.

**Specimen**

Specimen Type: Whole blood
**Container:** Lavender top (EDTA) tube

**Draw Volume:** 6 mL (Minimum: 3 mL) blood

**Processed Volume:** Same as Draw Volume

**Collection:** Routine blood collection

**Special Processing:** Lab Staff: Do Not Centrifuge. Blood specimen should remain in the original collection container. Store and ship at refrigerated temperatures with a cool pack by overnight courier. Forward promptly.

Samples can be stored at ambient temperatures if it will arrive less than 36 hours after collection. Samples are stable at 4 degrees Celsius for longer than 1 week.

**Patient Preparation:** None

**Sample Rejection:** Mislabeled or unlabeled specimens

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**Interpretive**

**Reference Range:** Interpretive report

**Critical Values:** N/A

**Limitations:** Sensitivity: Sequencing will identify a mutation in 99% of males (point mutations and indirect evidence of deletions) and 93% of females (point mutations only). Deletions directly identified by MPLA analysis account for 6% of mutations.

**Methodology:** Bidirectional sequencing

**References:** Johns Hopkins Diagnostic Lab March 2018
(410) 955-0483 Fax: (410) 955-0484

**Updates:** 2/11/2013: CPT update