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 adrenomyeloneuropthay (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

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