X-Linked Adrenoleukodystrophy: ABCD1 Gene Deletion/Duplication

**Test Code:** DABCD  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1

### Condition Description

X-linked adrenoleukodystrophy (X-ALD) is a disorder of fatty acid oxidation cause by mutations in the ABCD1 gene (Xq28). X-ALD can present at a variety of ages and with different manifestations depending on the presence and type of neurologic findings. The condition has three main phenotypes seen in males. In the childhood cerebral form, symptoms appear between ages four and eight years and include inattention, hyperactivity, and emotional lability. Progressive impairment of cognition, behavior, vision, hearing, and motor function follow the initial symptoms and often lead to total disability within two years. The second phenotype, adrenomyeloneuropathy (AMN), manifests most commonly in the late twenties as progressive paraparesis, sphincter disturbances, sexual dysfunction, and often, impaired adrenocortical function; all symptoms are progressive over decades. The third phenotype, “Addison disease only,” presents with primary adrenocortical insufficiency between age two years and adulthood (most commonly by age 7.5 years), without evidence of neurologic abnormality. Some degree of neurologic disability (most commonly AMN), however, usually develops later. Varying phenotypes often coexist in the same kindred or sibship. Approximately 20% of carrier females develop neurologic manifestations that resemble adrenomyeloneuropathy, but have later onset (age 35 years or later) and milder disease than affected males.

The ABCD1 gene encodes the ATP-binding cassette sub-family D member 1 protein, which is located in the peroxisomal membrane. Gene product is for the GeneTests summary on this condition.

DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes which cover the entire genomic region.

### Methodology

DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes which cover the entire genomic region.

### Detection

Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

### Specimen Requirements

Submit only 1 of the following specimen types

* Preferred specimen type: Whole Blood

**Type:** Whole Blood
Specimen Requirements:

In EDTA (purple top) or ACD (yellow top) tube:
Infants (2 years): 3-5 ml
Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.

Type: Saliva

Specimen Requirements:

Oragene™ Saliva Collection kit (available through EGL) used according to manufacturer instructions.

Specimen Collection and Shipping: Store sample at room temperature. Ship sample within 5 days of collection at room temperature with overnight delivery.

Special Instructions

Submit copies of diagnostic biochemical test results with the sample, if appropriate. Contact the laboratory if further information is needed.

Sequence analysis is required before deletion/duplication analysis by targeted CGH array. If sequencing is performed outside of Emory Genetics Laboratory, please submit a copy of the sequencing report with the test requisition.

Related Tests

- Sequence analysis of the ABCD1 gene is available and is required before deletion/duplication analysis.
- A CGH array-based test for deletion/duplication analysis of 64 different X-linked intellectual disability genes is available.
- Prenatal testing is available to couples who are confirmed carriers of mutations. Please contact the laboratory genetic counselor to discuss appropriate testing prior to collecting a prenatal specimen.