Lab Dept: Anatomic Pathology

Test Name: ALEXANDER DISEASE (GFAP) SEQUENCING

General Information

Lab Order Codes: ALEX

Synonyms: GFAP Sequencing

CPT Codes: 81405 – Molecular pathology Level 6

Test Includes: Sequencing of all 9 exons in the GFAP gene.

Logistics

Test Indications: Confirm a clinical diagnosis; differentiate Alexander disease from Canavan disease.

Alexander disease (AD) is a progressive disorder of the white matter of the central nervous system (CNS). The three types of AD are categorized by age of onset: infantile, juvenile, and adult. Affected infants develop a megalencephalic leukodystrophy, seizures, spasticity, ataxia, and psychomotor retardation. Infantile AD frequently leads to death within a decade after the diagnosis. Juvenile and adult forms of AD have a more slowly progressive course and are characterized by ataxia, bulbar signs and spasticity. Rosenthal fibers, observed in the astrocytes of affected individuals upon autopsy, are a hallmark feature of AD. These cytoplasmic inclusions are made up of glial acidic fibrillary protein (GFAP) and small heat-shock protein.

Lab Testing Sections: Anatomic Pathology – Sendouts

Referred to: GeneDx (GDX Test: 218)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: Results are reported in 3 weeks

Special Instructions: A complete GeneDx request form is required. Please send with the patient or sample to the laboratory.

Specimen
**Specimen Type:** Whole blood

**Container:** Lavender top (EDTA) tube

Alternate specimens: Buccal brushes (GeneDx kit is required) for age 6 months and older.

**Draw Volume:** 2 - 5 mL (Minimum: 1 mL) blood

**Processed Volume:** Same as Draw Volume

**Collection:** Routine venipuncture

**Special Processing:** Lab Staff: Do Not Centrifuge. Blood specimen should remain in the original collection container. Store and ship at ambient temperature.

Buccal Brush: Send with GeneDx kit.

**Patient Preparation:** None

**Sample Rejection:** Mislabeled or unlabeled specimens

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**Interpretive**

**Reference Range:** Negative

**Critical Values:** N/A

**Limitations:** N/A

**Methodology:** Capillary sequencing

**References:**

[GeneDx](https://www.genedx.com) March 2018

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