
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

Interpretive

Reference Range:	Negative
Critical Values:	N/A
Limitations:	N/A
Methodology:	Capillary sequencing
References:	GeneDx March 2018 (301) 519-2100 • FAX (301) 519-2892