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**Lab Dept:**                   **Anatomic Pathology**

**Test Name:**               **ALEXANDER DISEASE (GFAP) SEQUENCING**

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***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.

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***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 7-8 weeks

**Special Instructions:**    A complete [GeneDx request form](#) is required. Please send with the patient or sample to the laboratory.

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***Specimen***

<b>Specimen Type:</b>	Whole blood
<b>Container:</b>	Lavender top (EDTA) tube  Alternate specimens: Buccal brushes (GeneDx kit is required) for age 6 months and older.
<b>Draw Volume:</b>	2 - 5 mL (Minimum: 1 mL) blood
<b>Processed Volume:</b>	Same as Draw Volume
<b>Collection:</b>	Routine venipuncture
<b>Special Processing:</b>	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.
<b>Patient Preparation:</b>	None
<b>Sample Rejection:</b>	Mislabeled or unlabeled specimens

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***Interpretive***

<b>Reference Range:</b>	Negative
<b>Critical Values:</b>	N/A
<b>Limitations:</b>	N/A
<b>Methodology:</b>	Capillary sequencing
<b>References:</b>	<a href="#">GeneDx</a> October 2013 (301) 519-2100 • FAX (301) 519-2892