
Lab Dept: Anatomic Pathology

Test Name: RETT SYNDROME (CDKL5) GENE SEQUENCING

General Information

Lab Order Codes: CDK

Synonyms: CDKL5 Related Atypical Rett Syndrome

CPT Codes: 81406 – CDKL5, full gene analysis (Molecular Pathology procedure, level 7)

Test Includes: Molecular diagnosis of patients with clinical features associated with CDKL5 mutation.

Logistics

Test Indications: Confirmation of clinical diagnosis or carrier testing. Mutation in the CDKL5 gene on Xp22 is associated with mental retardation and severe neurological symptoms. The CDKL5 associated phenotype may be severe, with early-onset encephalopathy, infantile spasms, severe global developmental delay, and profound intellectual impairment seen in female and male patients. At the mild end of the spectrum are patients with mild mental retardation with autistic features. Many of these clinical features meet the criteria for the early-onset variant of Rett syndrome. Rett syndrome is caused by mutations in the MECP2 gene, and clinical symptoms include loss of speech and purposeful hand use, microcephaly, seizures, ataxia, and stereotypic hand movements. Mutation analysis of the CDKL5 gene should be considered in patients with the neurological phenotypes associated with mutations in this gene. In addition, patients who previously tested negative for comprehensive mutation analysis in the MECP2 gene are candidates for CDKL5 sequence analysis.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: Baylor College of Medicine Medical Genetics Laboratory (Baylor test: 6126)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: 4 weeks

Special Instructions: A [requisition form](#) must be sent with any specimen.

Specimen

Specimen Type: Whole blood

Container:	Lavender top (EDTA) tube
Draw Volume:	3 – 5 mL blood (Adults/Children) 3 mL (Infants)
Processed Volume:	Same as Draw Volume
Collection:	Routine venipuncture for blood specimens, invert gently to mix
Special Processing:	Lab Staff: Do Not centrifuge. Specimen should be kept in original collection container. Ship the specimen FedEx Monday through Friday only, ship at room temperatures. Saturday deliveries are accepted at Baylor. Note: For specimens collected Saturday – Sunday (or on a holiday), they will be held in Children’s Laboratories at refrigerated temperatures and shipped at room temperature on Monday (or the next business day Monday – Friday).
Patient Preparation:	None
Sample Rejection:	Mislabeled or unlabeled specimens; frozen or heated specimens

Interpretive

Reference Range:	Interpretive report
Critical Values:	N/A
Limitations:	N/A
Methodology:	Capillary sequence analysis is performed in both forward and reverse directions.
References:	Baylor College of Medicine 2013
Updates:	2/11/2013: CPT updates