Lab Dept: Anatomic Pathology

Test Name: RETT SYNDROME DELETION ANALYSIS

General Information

Lab Order Codes:	RSDA
Synonyms:	MECP2 Deletion Analysis
CPT Codes:	81304 – MECP2 gene analysis, deletion/duplication variants
Test Includes:	N/A

Logistics

Test Indications:	Rett syndrome is an X-linked neurodevelopmental disorder caused by mutations in the MECP2 gene which encodes the Methyl CpG Binding Protein 2 transcriptional repressor. Rett syndrome affects ~1 in 10,000 females with symptoms including loss of speech and purposeful hand use, microcephaly, seizures, ataxia, and stereotypic hand movements. MECP2 mutations manifest a broader spectrum of clinical phenotypes in female and rare male patients, with features overlapping with other mental retardation disorders. Mutations in the MECP2 coding region can be detected by sequence analysis in up to ~85% of Rett cases (see MECP2 Sequencing Analysis). In addition, large MECP2 gene deletions have been identified in approximately 10% of Rett patients. Our laboratory offers Southern analysis to detect gene rearrangements involving MECP2 (exons 1 through 4) for patients with a documented negative MECP2 sequencing study.
Lab Testing Sections:	Anatomic Pathology - Sendouts
Referred to:	Baylor College of Medicine-Medical Genetics Laboratories (Baylor Test: 6069)
Phone Numbers:	MIN Lab: 612-813-6280 STP Lab: 651-220-6550
Test Availability:	Daily, 24 hours
Turnaround Time:	3 weeks
Special Instructions:	This test is usually ordered when the MECP2 sequencing analysis test is negative. Please include completed Baylor Request Form and Consent Form.

Specimen

Specimen Type:	Whole blood
Container:	Lavender top (EDTA) tube
Draw Volume:	3 - 5 mL children/adults 3 mL infants
Processed Volume:	Same as Draw Volume
Collection:	Routine venipuncture
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

Interpretive

Reference Range:	No mutations detected
Critical Values:	N/A
Limitations:	N/A
Methodology:	Southern Blot
References:	Baylor College of Medicine – Medical Genetics October 2013 (800) 411-4363 Fax (713) 798-2787
Updates:	2/11/2013: CPT update