Lab Dept: Anatomic Pathology

Test Name: RETT SYNDROME (MECP2) SEQUENCING

General Information

Lab Order Codes: RSSA

Synonyms: MECP2 Sequencing Analysis

CPT Codes: 81302 – MECP2 gene analysis; full sequence analysis

Test Includes: DNA sequencing of MECP2 exons 1-4 in both directions using 96-capillary sequencer

Logistics

Test Indications: Confirmation of clinical diagnosis or carrier testing. 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. Medical Genetics offers DNA sequencing analysis of MECP2 exons 1 through 4.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: Baylor College of Medicine-Medical Genetics Laboratories (Baylor Test: 6068)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: 3 weeks

Special Instructions: 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, ship at room temperatures. **Note:** For specimens collected Saturday – Sunday (or on a holiday), they should 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:	Previously, testing laboratories had performed sequence analysis of the MECP2 coding region which was thought to consist of exons 2 through 4. Recently, rare mutations in MECP2 exon 1 have been reported in a few Rett patients. For patients who had previously tested negative by sequence analysis of exons 2 through 4, targeted sequencing analysis of MECP2 exon 1 can be requested through our laboratory. More significantly, testing may be considered for MECP2 gene deletions which have been identified in approximately 10% of Rett patients. (See MECP2 Deletion Analysis which is offered as a separate test).
Methodology:	Capillary sequence analysis is performed in both forward and reverse directions.
References:	Baylor College of Medicine – Medical Genetics October 2013 (800) 411-4363 Fax (713) 798-2787
Updates:	2/11/2013: CPT update