Rett Syndrome, MECP2, Comprehensive Analysis

Test Code: 2402

Test Synonyms: MECP2 Duplication Syndrome, MECP2-Related Disorders, MLPA-Rett Testing, Rett Syndrome Congenital Variant

CPT Code(s):
- 83891
- 83892 x 7
- 83898 x 7
- 83900
- 83904
- 83909 x 7-9
- 83914 x 7

Background:
Rett syndrome (RTT) is a severe progressive neurodevelopmental disorder that presents early in childhood and affects primarily females (OMIM #312750). This X-linked dominant disorder affects 1:10,000-1:15,000 girls. In classic RTT, normal growth and development occurs during approximately the first 6-18 months of life. This is followed by an arrest in development and subsequent progressive loss of acquired motor skills and language and the onset of stereotypical hand movements. Other variants of RTT include congenital Rett syndrome, “forme fruste” and preserved speech variant. RTT has generally been considered to be lethal in males, but recent studies have identified males with RTT with varying degrees of severity. Rett syndrome is caused by mutations in the methyl CpG binding protein 2 (MECP2) located at chromosome Xq28. This comprehensive analysis includes Sequencing and Del/ Dup Analysis.

Reasons for Referral:
- Confirmation of clinical diagnosis in patients with classical Rett syndrome.
- Diagnostic testing in patients with variants of Rett syndrome.
- Testing of family members of Rett syndrome patients or those who have non-syndromic mental retardation or autistic spectrum disorder.
- Prenatal diagnosis.

Methodology:
**Sequencing**: Full sequence analysis of the MECP2 gene. 80% detection rate in females with classical Rett syndrome.

**MLPA**: Gross Deletion and duplication analysis is done by MLPA (multiplex ligation-dependent probe amplification). The deletion/duplication analysis is done as a reflex test after full gene sequence analysis of the MECP2 gene.

Test reporting follows the American College of Medical Genetics guidelines.

Specimen Requirements:
- **Blood**: EDTA or ACD (Solution A or B):
  - Adult: 5 mL
  - Child: 5 mL
  - Infant: 2-3 mL
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A REQUISITION FORM MUST ACCOMPANY ALL SAMPLES. Please include detailed clinical information, including ethnicity, clinical history, and family history.

Test Performed (Days):
Weekly

Turn Around Time:
14 – 21 days

Shipment Sensitivity Requirements:
Package and ship specimen to remain cool, but not frozen. Please use the cold pack provided in the kit. Ship via overnight express, using the FedEx priority overnight label provided. Contact Client Services for shipping kits and instructions at (855) 535-1522.

Additional Information:
Prior to any genetic testing we recommend genetic counseling. Please contact Client Services at (855) 535-1522 for forms and information about prenatal diagnostic testing.

References: