Rett Syndrome: MECP2 Gene Deletion/Duplication

Test Code: KT  
Turnaround time: 2 weeks  
CPT Codes: 81304 x1

Condition Description

Rett syndrome is one of the leading causes of mental retardation and developmental regression in girls. Mutations in the MECP2 gene are found in approximately 80% of affected girls. Rett syndrome is inherited as an X-linked dominant trait. Though usually lethal in males, males meeting the clinical criteria for Rett syndrome have been identified. Some of them survive into adulthood with moderate to severe mental retardation, impaired language development, and movement disorders. MECP2 gene mutations may also present as atypical Rett syndrome. Patients previously diagnosed with autism, mild learning disability, clinically suspected but molecularly unconfirmed Angelman syndrome, or mental retardation with spasticity or tremor have been found to carry MECP2 mutations.

The MECP2 gene consists of four exons. Over 200 mutations have been reported in this gene that account for approximately 80% of the causes. About 64% of all MECP2 mutations are caused by C>T transitions at eight CpG dinucleotides. The C-terminal domain is prone to larger multi-nucleotide deletions that account for ~15% of all mutations. Although these deletions tend to affect the same region, completely identical deletions are rare. These may not be detectable in females by sequencing.

Please click here for the GeneClinics summary on this condition.


Genes

MECP2

Indications

Methodology

DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes which cover the entire genomic region.

Detection

Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations.

Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

Specimen Requirements

Submit only 1 of the following specimen types

Type: Whole Blood (EDTA)

Specimen Requirements:
EDTA (Purple Top)
Infants and Young Children (2 years of age to 10 years old): 3-5 ml
Older Children & Adults: 5-10 ml
Autopsy: 2-3 ml unclotted cord or cardiac blood

Specimen Collection and Shipping:
Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.

Type: DNA, Isolated

Specimen Requirements:
Microtainer
3µg
Isolation using the Perkin Elmer™Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping:
Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample at room temperature with overnight delivery.

Special Instructions

Sequence analysis is required before deletion/duplication analysis by targeted CGH array. If sequencing is performed outside of EGL Genetics, please submit a copy of the sequencing report with the test requisition. Contact the laboratory if further information is needed.