Rett Syndrome: MECP2 Gene Deletion/Duplication

**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.

Please note that a "backbone" of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

**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

* Preferred specimen type: Whole Blood

**Type: Whole Blood**

Specimen Requirements:

In EDTA (purple top) tube:
- Infants (2 years): 3-5 ml
- Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.

**Type: Saliva**

Specimen Requirements:

Oragene™ Saliva Collection kit (available through EGL) used according to manufacturer instructions.

Specimen Collection and Shipping: Store sample at room temperature. Ship sample within 5 days of collection 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.