Mental Retardation, Stereotypic Movements, Epilepsy, and/or Cerebral Malformations: MEF2C Gene Deletion/Duplication

**Test Code:** DMEF2

**Turnaround time:** 2 weeks

**CPT Codes:** 81228 x1

**Condition Description**

Le Meur et al. (2010) describe six unrelated children with a deletion or mutation of the MEF2C gene (5q14). All six patients had a similar phenotype including severe intellectual disability, developmental delay, hypotonia, absent speech, and the inability to walk unaided. Variable features include stereotypic movements, epilepsy and/or cerebral malformations, and dysmorphic features. Mutation of the MEF2C gene causes autosomal dominant mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations.

For patients with suspected mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, sequence analysis is recommended as the first step in mutation identification. For patients in whom mutations are not identified by full gene sequencing, deletion/duplication analysis is appropriate.

**References:**

- OMIM #600662: MEF2C gene
- OMIM #613443: Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations

**Genes**

MEF2C

**Indications**

This test is indicated for:

- Confirmation of a clinical diagnosis of mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations in an individual in whom sequence analysis was negative.
- Carrier testing in adults with a family history of mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations in whom sequence analysis was negative.

**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: DNA, Isolated**

**Specimen Requirements:**

- Microtainer
- 3 µg

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.

**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 uncotted cord or cardiac blood

**Specimen Collection and Shipping:**
Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not freeze.

**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.

**Related Tests**

- Sequence analysis of the *MEF2C* gene is available and is required before deletion/duplication analysis.
- Custom diagnostic mutation analysis (KM) is available to family members if mutations are identified by targeted mutation testing or sequencing analysis.
- Prenatal testing is available only for known familial mutations to individuals who are confirmed carriers of mutations. Please contact the laboratory genetic counselor to discuss appropriate testing prior to collecting a prenatal specimen.