Autosomal Dominant Mental Retardation 1: **MBD5** Gene Deletion/Duplication

**Test Code:** DMBD5  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1

### Condition Description

Talkowski et al. (2011) mapped the *MBD5* gene (2q23.1) to the critical region of the 2q23.1 deletion syndrome. Haploinsufficiency of the *MBD5* gene causes Autosomal Dominant Mental Retardation syndrome type 1. Overall, of the features evaluated in individuals with 2q23.1 deletion syndrome and *MBD5*-specific deletions, approximately 84% were observed in both groups. Features associated with the haploinsufficiency of the *MBD5* gene include intellectual disability, developmental delay, motor delay, significant speech impairment, craniofacial manifestations, seizures, constipation, and behavioral problems.

For patients with suspected Autosomal Dominant Mental Retardation syndrome type 1, deletion/duplication analysis is recommended as the first step in mutation identification. For patients in whom mutations are not identified by deletion/duplication analysis, full gene sequencing is appropriate.

### References:

- OMIM #611472: *MBD5* gene  
- OMIM #156200: Autosomal Dominant Mental Retardation Syndrome Type 1  

### Genes

**MBD5**

### Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of Autosomal Dominant Mental Retardation syndrome type 1.
- Carrier testing in adults with a family history of Autosomal Dominant Mental Retardation syndrome type 1.

### 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 72 hours of collection. Do not 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.

### Related Tests

Disclaimer: This information is confidential and subject to change without notice. It may not be reproduced in whole or part unless authorized in writing by an authorized EGL representative.
- Sequence analysis of the *MBDS5* gene is available in those individuals in whom deletion/duplication analysis is negative.
- 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.