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.

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.

### Related Tests

- Sequence analysis of the **MBD5** 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.