Mental Retardation with Language Impairment and Autistic Features: **FOXP1 Gene Deletion/Duplication**

**Test Code:** DFOX1  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1

### Condition Description

Hamdan et al. (2010) identified two patients with de novo mutations in the **FOXP1** gene (3p14.1). Mutation of the **FOXP1** gene causes autosomal dominant mental retardation with language impairment and autistic features. Common features seen in these patients include global developmental delay with severe language impairment, mild to moderate intellectual disability, autism or autistic features, and internalizing and externalizing behavior problems. The **FOXP1** gene and its closest homolog, the **FOXP2** gene, may regulate common processes. They are both expressed in overlapping regions of the brain including areas associated with the production and processing of vocalization and language.

For patients with suspected mental retardation with language impairment and autistic features, 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 #605515: **FOXP1** gene
- OMIM #613670: Mental Retardation with Language Impairment and Autistic Features

### Genes

**FOXP1**

### Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of mental retardation with language impairment and autistic features in an individual in whom sequence analysis was negative.
- Carrier testing in adults with a family history of mental retardation with language impairment and autistic features 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: 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.

**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 *FOXP1* gene is available and is required before deletion/duplication analysis.
- Sequencing and deletion/duplication analysis of the *FOXP2* gene are available.
- 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.