Speech-Language Disorder 1: **FOXP2 Gene Deletion/Duplication**

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

### Condition Description

Mutations of the FOXP2 gene (7q31) cause a severe form of speech and language disorder. In a large multi-generational family, a heterozygous missense mutation segregates with family members who have verbal dyspraxia or a deficit in sequencing the complex coordinated orofacial movements required for speech. It is also accompanied by a wide range of linguistic and grammatical deficits. Additionally, the FOXP2 gene was found to be disrupted by a de novo balanced translocation in a patient with speech and language disorder.

FOXP2 encodes a transcription factor which likely regulates gene expression in the developing lung, cardiovascular, intestinal, and neural tissue.

**References:**

- OMIM #605317: FOXP2 gene
- OMIM #602081: Speech-Language Disorder 1

### Genes

**FOXP2**

### Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of speech-language disorder 1 in an individual in whom sequence analysis was negative.
- Carrier testing in adults with a family history of speech-language disorder 1 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 FOXP2 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.
- X-Linked Intellectual Disability panels are available for 30, 60, and 90+ genes.