

Brunner Syndrome: MAOA Gene Deletion/Duplication

Test Code: DMAOA

Turnaround time: 2 weeks

CPT Codes: 81228 x1

Condition Description

Mutations in the *MAOA* gene result in the X-linked disorder Brunner syndrome, or monoamine oxidase deficiency, which is characterized in part by severe impulsive behavior and mild nondysmorphic intellectual disability. *MAOA* encodes for monoamine oxidase A, an enzyme that degrades amine neurotransmitters such as dopamine, norepinephrine, and serotonin.

For patients with suspected Brunner syndrome, 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 309850](#): Monoamine Oxidase A; MAOA
- [OMIM 300615](#): Brunner Syndrome
- Gunter et al. Behavioral Genetics in Antisocial Spectrum Disorders and Psychopathy: A Review of the Recent Literature. (2010). Behav. Sci. Law 28: 148-173.

Genes

[MAOA](#)

Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of Brunner syndrome in an individual in whom sequence analysis was negative.
- Carrier testing in adults with a family history of Brunner syndrome 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

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.

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.

Special Instructions

Submit copies of diagnostic biochemical test results with the sample, if appropriate. Contact the laboratory if further information is needed.

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 *MAOA* 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.