Autosomal Dominant Optic Atrophy (Kjer Type): OPA1 Gene Deletion/Duplication

| Test Code: KO | Turnaround time: 2 weeks | CPT Codes: 81406 x1 |

**Condition Description**

Autosomal dominant optic atrophy, Kjer type, is the most common form of hereditary optic neuropathy. The incidence of ADOA is approximately 1 in 50,000 to 1 in 10,000 live births. It is a childhood onset disorder typically characterized by a progressive loss in central vision, color vision deficits (dyschromatopsia), decreased visual acuity, decreased sensitivity of the central retinal field (paracentral scotomas), and asymmetric degeneration of the retinal ganglion cells visible as pallor of the optic disk. The underlying defect is retinal ganglion cell degeneration. The disease phenotype displays both inter- and intra-familial variability with incomplete penetrance.

Mutations in the OPA1 gene, located on chromosome 3q28-q29, cause Kjer type autosomal dominant optic atrophy. OPA1 consists of 31 exons and encodes a mitochondrial dynamin-related GTPase, a protein thought to be involved in maintaining the structure and function of mitochondria. Approximately 90% of autosomal dominant optic atrophy patients carry a mutation in OPA1.

**Genes**

OPA1

**Indications**

This test is indicated for patients with a diagnosis of optic atrophy. Sequencing is not appropriate for prenatal samples in which familial mutations have not been identified.

**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. Array CGH will not detect point mutations or intronic mutations. Results of molecular analysis must 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.

**Special Instructions**

Submit copies of diagnostic biochemical test results with the sample. Sequence analysis is required before deletion/duplication analysis by targeted CGH array. If sequencing is performed outside EGL Genetics, please submit a copy of the sequencing report with the test requisition. Contact the laboratory if further information is needed.

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Related Tests

- Leber Hereditary Optic Neuropathy (QC) is a mitochondrial disorder characterized by atrophy of the optic nerve.
- OPA3 gene sequencing.
- Prenatal testing may be available to family members if OPA1 mutations have been identified. Please contact the laboratory genetic counselor to arrange prior to collecting a prenatal specimen.