Autosomal Dominant Optic Atrophy: OPA3 Gene Deletion/Duplication

Test Code: KN  
Turnaround time: 2 weeks  
CPT Codes: 81228 x1

**Condition Description**

Mutations in the OPA3 gene have been associated with different forms of optic atrophy. Autosomal dominant optic atrophy (ADOA) is characterized by progressive visual loss beginning in childhood, loss of visual acuity, and optic nerve pallor. Mutations in the gene OPA3 have been associated with ADOA and cataracts (ADOAC) [1]. Type III 3-methylglutaconic aciduria (MGA; Costeff optic atrophy syndrome) is an autosomal recessive disorder characterized by early onset bilateral optic atrophy and later onset ataxia, spasticity, and cognitive decline. A hallmark of disease is increased urinary excretion of 3-methylglutaconic and 3-methylglutaric acid. A founder OPA3 mutation accounts for the relatively high frequency of Type III MGA in the Iraqi Jewish population [2]. Diagnostic sequencing analysis of the OPA3 gene coding region is available for patients with optic atrophy and their at risk family members. For further information call EGL Genetics at 470-378-2200.

**References:**

**Genes**

OPA3

**Indications**

- Clinical features associated with OPA3 gene mutations
- Prenatal testing after a familial mutation has been identified
- Testing for persons at risk for carrying a familial mutation

**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 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.
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. Contact the laboratory if further information is needed.

Related Tests

- OPA1 gene sequencing
- Custom diagnostic mutation analysis (KM) is available to family members if mutations are identified by sequencing.
- Prenatal testing is available to couples who are confirmed carriers of mutations. Please contact the laboratory genetic counselor to discuss appropriate testing prior to collecting a prenatal specimen.