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 the Emory Genetics Laboratory at (404) 778-8500.

### 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 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) or ACD (yellow 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 Emory Genetics Laboratory, 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.