

Disease Name:

3-METHYLGLUTACONIC ACIDURIA TYPE III
(OPTIC ATROPHY PLUS SYNDROME; IRAQI-JEWISH 'OPTIC ATROPHY PLUS';
MGA, TYPE III; COSTEFF SYNDROME)

Classification:

Organic aciduria

Genetic Information:

Inheritance:

Autosomal recessive

Population Incidence:

Unknown- only been diagnosed among Jewish kindred

Ethnic Incidence:

1:10,000 among Iraqi Jewish kindred

Gene & Location:

OPA3 gene on 19q12.2-13.3

Common Mutation:

A G-to-C founder mutation has been identified

OMIM #

#258501; *606580

Disease Information:

Symptom Onset:

Presents in infants

Symptoms:

The disease presents with infantile bilateral optic atrophy, choreoathetosis, spastic paraparesis, cerebellar ataxia and nystagmus. Some patients have mental retardation. The course of the disease is non-progressive beyond childhood. Most develop spastic paraparesis by the second decade of life. About one-half of patients have non-progressive ataxia. Some patients have been noted to have dysarthria. The life span is normal.

Physical Findings:

No dysmorphisms.

Treatment:

There are no effective treatments. Coenzyme Q10 therapy has been tried without any change in the clinical status.

Natural History without treatment:

Spastic paraparesis with blindness and possible mental retardation. Possibly some ataxia.

Natural History with treatment:

Same as for untreated group

Metabolic Information:

Missing Enzyme & Location:

The basic enzyme defect is unknown

MS/MS profile:

C5-OH (3-hydroxyisovaleryl carnitine) - elevated

Prenatal testing:

Possible mutation analysis for at risk pregnancies.

**Miscellaneous
Information:**

Prepared for the NW Regional Newborn Screening Program by Sara Copeland MD, Judith Tuerck RN MS and Lorinda Paradise at OHSU in Portland, OR.

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