

| Disease Name | | | |
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| 3-METHYLGLUTACONIC ACIDURIA TYPE III (MGA 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 19q13.2-q13.3 | | |
| 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 nonprogressive 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: | | | |
| Credit: | <i>Prepared by the North West Regional Newborn Screening Program Judith Tuerck, RN, MS, and Lorinda Paradise at Oregon Health Services University in Portland, Oregon and by Sara Copeland MD, Iowa Neonatal Metabolic Screening Program.</i> | | |
| Sites of Reference: | OMIM - Methylmalonic Aciduria www.ncbi.nlm.nih.gov/htbin-post/Omim/dispim?251000 | | |
| Support Groups: | <table border="0" style="width: 100%;"> <tr> <td style="width: 50%; vertical-align: top;"> Organic Acidemia Association www.oaanews.org/ 13210 35th Avenue Plymouth, MN 55441 Contact: Kathy Stagni </td> <td style="width: 50%; vertical-align: top;"> Children Living with Inherited Metabolic Diseases (CLIMB) Climb Building 176 Nantwich Road Crewe, CW2 6BG </td> </tr> </table> | Organic Acidemia Association www.oaanews.org/ 13210 35th Avenue Plymouth, MN 55441 Contact: Kathy Stagni | Children Living with Inherited Metabolic Diseases (CLIMB) Climb Building 176 Nantwich Road Crewe, CW2 6BG |
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