



Organic Acid Disorders

Screening, Technology, and Research in Genetics is a multi-state project to improve information about the financial, ethical, legal, and social issues surrounding expanded newborn screening and genetic testing – <http://www.newbornscreening.info>

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| Disease name | 2-methylbutyryl-CoA dehydrogenase deficiency |
| Alternate name(s) | Short/branched chain acyl-CoA dehydrogenase deficiency, Methylbutyrylglycinuria |
| Acronym | N/A |
| Disease classification | Organic Acid Disorder |
| Variants | None |
| Variant name | N/A |
| Symptom onset | Variable |
| Symptoms | One patient presented with neonatal onset of hypotonia, lethargy, apnea and hypoglycemia. At four years of age, he has developmental delay, choreoathetoid cerebral palsy and visual deficits. Another patient presented in the second year of life with motor delay, muscular atrophy and strabismus. A sibling identified prenatally and 8 Hmong patients identified by newborn screening remain asymptomatic on treatment. At least 4 asymptomatic relatives of these patients have been described with gene mutations and/or elevated excretion of 2-methylbutyrylglycine. |
| Natural history without treatment | Ranges from asymptomatic to acute neonatal decomposition with neurological deficits. The limited number of patients makes it difficult to determine the natural history of the disorder. However, the disorder is not thought to be benign in that asymptomatic individuals may not have been exposed to the environmental stressors (i.e. fasting) that can cause symptoms. |
| Natural history with treatment | Treatment in a symptomatic patient resolved episodic hypoglycemia but the neurologic dysfunction remains. Other patients treated from birth are asymptomatic thus far but the efficacy of the treatment remains to be established. |

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| Treatment | Protein restriction, carnitine supplementation, avoidance of fasting. |
| Other | N/A |
| Physical phenotype | None reported |
| Inheritance | Autosomal recessive |
| General population incidence | Rare; < than 20 patients identified |
| Ethnic differences | Yes |
| Population | Hmong |
| Ethnic incidence | As high as 1/500 |
| Enzyme location | Mitochondria |
| Enzyme function | Metabolism of L-isoleucine |
| Missing enzyme | 2-methylbutyryl-CoA dehydrogenase |
| Metabolite changes | Increased 2-methylbutyryl, increased 2-methylbutyrylcarnitine. |
| Gene | SBCAD |
| Gene location | 10q25-q26 |
| DNA testing available | Research |
| DNA testing detail | A common mutation has been identified in the Hmong population – M356V which causes skipping of exon 10 in the SBCAD. |
| Prenatal testing | Enzyme analysis in amniocytes or CVS |
| MS/MS profile | Elevated C5 isovaleryl-carnitine |
| OMIM link | www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=600301 |
| Genetests link | www.genetests.org/servlet/access?prg=j&db=genetests&site=gt&id=8888891&fcn=c&qry=246672&res=&key=exWOkOLHnxbCE&show_flag=c |

Support group

Organic Acidemia Association
www.oaanews.org

Save Babies through Screening Foundation
www.savebabies.org

Genetic Alliance
www.geneticalliance.org

Document Info

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