Disease Name	**2-Methylbutyryl-CoA Dehydrogenase Deficiency**
Acronym	2MBDH / SBCADD / 2-MBADD
Disease Classification	Organic Acid Disorder
Variants	Yes
Symptom onset	Few days after birth or later in childhood
Symptoms	Neonatal - Poor feeding, lethargy, vomiting, irritability, hypotonia, hypoglycemia Later in childhood – learning disabilities, motor delay, muscular atrophy, visual deficits**.**
Natural History w/o treatment	Ranges from asymptomatic to acute neonatal decompensation with neurological deficits
Natural History with treatment	With prompt and careful treatment, symptomatic children have a good chance of living healthy lives with typical growth and development.
Treatment	Protein restriction, avoidance of fasting, and carnitine supplement
Emergency Medical Treatment	Management of hypoglycemic episodes; management of metabolic crises May include an aggressive use of glucose and electrolytes.
Inheritance	Autosomal recessive pattern. Effects both boys and girls equally
General population incidence	Rare. Actual incidence unknown.
Ethnic Difference	Increased in Hmong population; 1/500 among people of Hmong ancestry
Enzyme location	2-Methylbutyryl-CoA Dehydrogenase-second step in isoleucine metabolism – enzyme irreversibly dehydrogenates 2-methylbutyryl-CoA to tiglyl Co-A.
Enzyme function	Helps process the amino acid Isoleucine
Metabolite changes	Isoleucine unable to be converted into energy; 2Methylbutyrylglycine builds up to harmful levels
Prenatal testing	Yes. Isoleucine metabolism in cultures fibroblasts
MS/MS Profile	Acylcarnitine profile C5; urine organic acid – marked elevation of isovalerylglycine
OMIM Link	www.omom.org ID=600301
Genetests Link	www.genetest.org
Support Group	**Organic Acidemia Association** www.oaanews.org/ **Save Babies through Screening Foundation** www.savebabies.org **Genetic Alliance** www.geneticalliance.org