Disease Name	**Isobutyryl-CoA dehydrogenase deficiency**
Acronym	IBD, IBDH
Disease Classification	Organic Acid Disorder/Fatty Acid Oxidation Defect
Variants	None
Symptom onset	12 months of age
Symptoms	Generally asymptomatic. The initial patient presented with dilated cardiomyopathy, low carnitine and anemia. The child was small for age, normal growth resumed with treatment.
Natural History Without Treatment	Unknown
Natural History With Treatment	Improved symptoms of cardiomyopathy, anemia, Improved growth and normal development
Treatment	Moderate protein restriction. Carnitine therapy
Inheritance	Autosomal recessive
General population incidence	Rare, less than five cases reported
Ethnic Differences	None
Missing Enzyme	Isobutyryl-CoA dehydrogenase
Enzyme location	Mitochondria
Enzyme Function	Metabolism of valine
MS/MS Profile	C4 butyryl carnitine elevation
OMIM Link	www.omom.irg ID# 60473
Genetests Link	None
Support Group	Fatty Acid Oxidation Support Network www.fodsupport.org Save Babies through Screening Foundation www.savebabies.org Genetic Alliance www.genticalliance.org