



## Genetic Fact Sheets for Professionals

# 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](http://www.newbornscreening.info)

---

<b>Disease name</b>	<b>Isovaleric acidemia</b>
<b>Alternate name(s)</b>	Isovaleric acid CoA dehydrogenase deficiency
<b>Acronym</b>	IVA
<b>Disease classification</b>	Organic Acid Disorder
<b>Variants</b>	Yes
<b>Variant name</b>	Chronic intermittent form
<b>Symptom onset</b>	Infancy (in the acute neonatal form). The chronic intermittent form presents later in infancy or in childhood.
<b>Symptoms</b>	Episodic overwhelming illness with vomiting, ketosis, acidosis and coma. Hematological abnormalities include leucopenia, thrombocytopenia and possible anemia.
<b>Natural history without treatment</b>	About 50% of patients with the acute neonatal form will die during their first episode. Survivors may have neurological damage though several have made complete recoveries. Patients with the chronic form may have neurologic damage, but the majority of patients are developmentally normal.
<b>Natural history with treatment</b>	Intellectual prognosis depends on early diagnosis and treatment and subsequently on long-term compliance. If treated appropriately, most will have normal development.
<b>Treatment</b>	Low protein diet with restricted leucine intake, glycine supplementation and possible carnitine supplementation.
<b>Other</b>	Sometimes a "sweaty feet" odor is reported during an acute crisis.
<b>Physical phenotype</b>	No obvious dysmorphic features .
<b>Inheritance</b>	Autosomal recessive
<b>General population incidence</b>	1: 230,000

<b>Ethnic differences</b>	None known
<b>Population</b>	N/A
<b>Ethnic incidence</b>	N/A
<b>Enzyme location</b>	N/A
<b>Enzyme function</b>	Isovaleryl-CoA dehydrogenase is the first step in the branched chain organic acid metabolism of leucine.
<b>Missing enzyme</b>	Isovaleryl-CoA dehydrogenase
<b>Metabolite changes</b>	Urinary isovaleryl glycine, 3-hydroxysoroline acid, increased isovaleric acid in blood. During acute attacks, 4-hydroxyisovaleric acid, mesaconic acid, and methylsuccinic acid, isovalerylglycine and 3-hydroxyisovaleric acid are present.
<b>Gene</b>	IVD
<b>Gene location</b>	15q14-15
<b>DNA testing available</b>	No
<b>DNA testing detail</b>	N/A
<b>Prenatal testing</b>	Enzyme analysis by GCMS in amniotic fluid or CVS tissue.
<b>MS/MS profile</b>	Elevated C5 isovaleryl carnitine
<b>OMIM link</b>	<a href="http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=243500">www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=243500</a>
<b>Genetests link</b>	<a href="http://www.genetests.org/servlet/access?prg=j&amp;db=genestar&amp;site=&amp;fcn=d&amp;id=12600&amp;qry=2251&amp;res=nous&amp;res=nointl&amp;key=Issq6RQIfI8i5&amp;show_flag=c">www.genetests.org/servlet/access?prg=j&amp;db=genestar&amp;site=&amp;fcn=d&amp;id=12600&amp;qry=2251&amp;res=nous&amp;res=nointl&amp;key=Issq6RQIfI8i5&amp;show_flag=c</a>
<b>Support group</b>	Organic Acidemia Association <a href="http://www.oaanews.org">www.oaanews.org</a>  Save Babies through Screening Foundation <a href="http://www.savebabies.org">www.savebabies.org</a>  Genetic Alliance <a href="http://www.geneticalliance.org">www.geneticalliance.org</a>

## Document Info

<b>Created by</b>	www.newbornscreening.info
<b>Reviewed by</b>	HI, CA, OR and WA metabolic specialists
<b>Review date</b>	May 2, 2005
<b>Update on</b>	N/A

### **DISCLAIMER:**

THIS INFORMATION DOES NOT PROVIDE MEDICAL ADVICE. All content ("Content"), including text, graphics, images and information are for general informational purposes only. You are encouraged to confer with your doctor or other health care professional with regard to information contained on this information sheet. After reading this information sheet, you are encouraged to review the information carefully with your doctor or other healthcare provider. The Content is not intended to be a substitute for professional medical advice, diagnosis or treatment. NEVER DISREGARD PROFESSIONAL MEDICAL ADVICE, OR DELAY IN SEEKING IT, BECAUSE OF SOMETHING YOU HAVE READ ON THIS INFORMATION SHEET.



This project is supported by a grant from the Maternal and Child Health Bureau,  
Health Resources and Service Administration, Genetic Services Branch,  
MCH Project #:1H46 MC 00189-03 <http://mchb.hrsa.gov>