Genetic Fact Sheets for Professionals

Fatty Acid Oxidation 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

Disease name	**Carnitine transporter deficiency**
Alternate name(s)	Systemic carnitine deficiency, Carnitine deficiency, Carnitine uptake deficiency
Acronym	SCD, CUD
Disease classification	Fatty Acid Oxidation Disorder
Variants	N/A
Variant name	N/A
Symptom onset	Infancy or childhood with fasting hypoglycemia, weakness and/or cardiomyopathy.
Symptoms	Hypoketotic hypoglycemia, seizures, vomiting, lethargy progressing to coma. Chronic muscle weakness, cardiomyopathy, hepatomegaly.
Natural history without treatment	Non-progressive developmental delay due to hypoglycemia, cardiomyopathy and muscle weakness.
Natural history with treatment	Developmental delay, if present, is not reversed by treatment. Cardiomyopathy and muscle weakness can be reversed by treatment.
Treatment	Carnitine supplementation, no fasting.
Other	N/A
Physical phenotype	Cardiomyopathy, muscle weakness.
Inheritance	Autosomal recessive

General population incidence	1/40,000
Ethnic differences	No
Population	N/A
Ethnic incidence	N/A
Enzyme location	Muscle, heart, kidney, leukocytes and fibroblasts
Enzyme function	Transports carnitine into cells
Missing enzyme	Carnitine transporter
Metabolite changes	Decreased free carnitine in plasma, increased carnitine in urine, decreased carnitine in muscle.
Gene	SLC22A5
Gene location	5q33.1
DNA testing available	May be available on a research basis.
DNA testing detail	If a mutation in a proband is detected, DNA carrier screening is possible.
Prenatal testing	Protein analysis in cultured amniocytes, biochemical analyte testing. If a mutation in a proband is detected, DNA prenatal diagnosis via CVS or amniocytes is possible.
MS/MS profile	Reduced concentrations of free carnitine and various acylcarnitine species.
OMIM link	www.ncbi.nlm.nih.gov/htbin-post/Omim/dispmim?212140
Genetests link	www.genetests.org/servlet/access?prg=j&db=genestar&site=&fcn=d&id=12600&qry=3164&res=nous&res=nointl&key=PEbBO3wAQlhnM&show_flag=c
Support group	FOD Family Support Group www.fodsupport.org Save Babies through Screening Foundation www.savebabies.org

Genetic Alliance
www.geneticalliance.org

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