



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

Disease name	Carnitine acylcarnitine translocase deficiency
Alternate name(s)	Carnitine acylcarnitine carrier (CAC), CAT deficiency
Acronym	CAT or CACT
Disease classification	Fatty Acid Oxidation Disorder
Variants	N/A
Variant name	N/A
Symptom onset	Neonatal
Symptoms	Hypoketotic hypoglycemia, hyperammonemia, hepatomegaly, SIDS, muscle weakness, cardiomyopathy with ventricular arrhythmia.
Natural history without treatment	Hypoglycemia crisis leading to coma and death, heart abnormalities can also cause death.
Natural history with treatment	Efficacy of treatment unknown. Disease reported to be commonly fatal. Individuals with residual enzymatic activity may have a better outcome.
Treatment	Carnitine supplementation, restriction of long chain fatty acids. Cornstarch therapy, high carbohydrate/low-fat diet, avoidance of fasting.
Other	Maternal pre-eclampsia during pregnancy has been reported, but may be coincidental.
Physical phenotype	None
Inheritance	Autosomal recessive
General population incidence	Rare
Population	N/A
Ethnic incidence	N/A

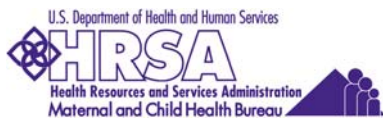
Enzyme location	Heart, liver, skeletal muscles
Missing enzyme	Carnitine acylcarnitine translocase
Metabolite changes	Low plasma free carnitine, elevated esterified carnitine in blood and urine, long-chain acylcarnitine fraction elevated in plasma.
Gene	CAC
Gene location	3p21.31
DNA testing available	Yes
DNA testing detail	DNA testing available in the UK
Prenatal testing	Acylcarnitine analysis on supernatant of cultured amniocytes, protein analysis in CVS and amnio, DNA if mutation is known.
MS/MS profile	C18:1, C18:2, C16, C16DC, C18:2DC, C18:1DC
OMIM link	www.ncbi.nlm.nih.gov/htbin-post/Omim/dispnim?212138
Genetests link	www.genetests.org/servlet/access?prg=j&db=genestar&site=&fcn=d&id=12600&qry=16912&res=nous&res=nointl&key=xQoEYvoBMHPor&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

Document Info

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