



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)

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| Disease name | Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency |
| Alternate name(s) | N/A |
| Acronym | LCHADD/TFP |
| Disease classification | Fatty Acid Oxidation Disorder |
| Variants | Yes |
| Variant name | Mitochondrial trifunctional protein deficiency |
| Symptom onset | Neonatal, infancy |
| Symptoms | Hypoketotic hypoglycemia, hypotonia, cardiomyopathy, hepatic disease, peripheral neuropathy and pigmentary retinopathy, rhabdomyolysis, sudden death |
| Natural history without treatment | Possible developmental delay due to damage from hypoglycemic episodes, possible death due to cardiomyopathy or hepatic failure. |
| Natural history with treatment | Intelligence is usually normal if there is no damage due to hypoglycemic crisis. Peripheral neuropathy, if present, may not improve with treatment. |
| Treatment | Avoidance of fasting, use of uncooked starch, MCT treatments, carnitine supplementation, DHA supplementation (may prevent retinopathy, but this has not been proven) |
| Other | Maternal complications in pregnancy include acute fatty liver of pregnancy, HELLP syndrome, and pre-eclampsia |

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| Physical phenotype | Hypotonia, cardiomyopathy and possible retinal changes |
| Inheritance | Autosomal recessive |
| General population incidence | Rare |
| Ethnic differences | Yes |
| Population | Finnish |
| Ethnic incidence | 1:240 carrier rate for common mutation G1528C in Finland |
| Enzyme location | Inner mitochondrial membrane, liver, heart, fibroblasts |
| Enzyme function | Metabolizes long chain fatty acids (C-12 to C-16 in length) |
| Missing enzyme | Long-chain 3-hydroxyacyl-CoA dehydrogenase or mitochondrial trifunctional protein |
| Metabolite changes | Increased 3-hydroxydicarboxylic acids in urine, increased saturated and unsaturated 3-hydroxy organic acids, possible elevated CPK during acute illness. |
| Gene | HADHA and HADHB (alpha and beta subunits) |
| Gene location | 2p23 |
| DNA testing available | Yes – mutation analysis |
| DNA testing detail | Common mutation, G1528C, accounts for 87% of all mutant alleles in LCHAD deficiency; 70% of affected individuals will be homozygous for this mutation. There is no common mutation in trifunctional protein deficiency. |
| Prenatal testing | Enzyme analysis, protein analysis and direct DNA (when applicable). |
| MS/MS profile | C18:OH, C16:1OH, C16OH |
| OMIM link | www.ncbi.nlm.nih.gov/htbin-post/Omim/dispnim?600890 |
| Genetests link | www.genetests.org/servlet/access?prg=j&db=genetests&site=&id=12600&fcn=c&qry=2927&res=nous&res=nointl&key=z31IF9RXwW9Oo&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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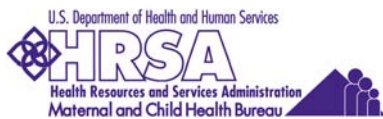
Reviewed by HI, CA, OR, and WA metabolic specialists

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Updated on N/A

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