



State of CT
Genetics Newborn Screening Program
Health Care Provider Fact Sheet

Long chain acyl-CoA dehydrogenase (LCAD) deficiency

Until the discovery of Very Long Chain Acyl-CoA Dehydrogenase Deficiency in the early 1990s, patients diagnosed with deficiencies in VLCAD were considered to have a defect in long-chain acyl-coenzyme A dehydrogenase (LCAD). Thus far, no specific mutations have been identified in the gene encoding human LCAD (*ACADL*), and no definitively diagnosed cases of human LCAD deficiency are known. The failure to identify patients with LCAD deficiency is surprising given the recognition of disease caused by deficiencies in all other members of this gene family. This has put into question both the role of LCAD in the metabolism of long-chain fatty acids and the potential of LCAD deficiency to produce disease. Human LCAD deficiency may not cause clinical disease or it could result in gestational lethality, either of which could account for the lack of identified human cases.

There is limited information available on this disease. The following are related articles and links:

Targeted disruption of mouse long-chain acyl-CoA dehydrogenase gene reveals crucial roles for fatty acid oxidation

David M. Kurtz*, Piero Rinaldo[†], William J. Rhead[‡], Liqun Tian*, David S. Millington[§], Jerry Vockley[¶], Doug A. Hamm*, Amy E. Brix*, J. Russell Lindsey*, Carl A. Pinkert*, William E. O'Brien^{||}, and Philip A. Wood
<http://www.pnas.org/cgi/content/full/95/26/15592?ikey=95ed755ad78c0437174e868e8ea63b2b56641bf4>

Regulation of Fatty Acid Transport Protein and Mitochondrial and Peroxisomal β -Oxidation Gene Expression by Fatty Acids in Developing Rats

FETTA OUALI, FATIMA DJOUADI, CLAUDIE MERLET-BÉNICHOU, BÉATRICE RIVEAU and JEAN BASTIN
<http://www.pedresearch.org/cgi/content/full/48/5/691>

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201460 ACYL-CoA DEHYDROGENASE, LONG-CHAIN, DEFICIENCY OF

Alternative titles; symbols

ACADL DEFICIENCY

LCAD DEFICIENCY

HYPOGLYCEMIA, NONKETOTIC, DUE TO DEFICIENCY OF ACYL-CoA DEHYDROGENASE

ACYL-CoA DEHYDROGENASE, LONG-CHAIN, INCLUDED; ACADL, INCLUDED

<http://www.ncbi.nlm.nih.gov/entrez/dispmim.cgi?id=201460>

Human Molecular Genetics, 2001, Vol. 10, No. 19 2069-2077 © 2001 Oxford University Press

Gestational, pathologic and biochemical differences between very long-chain acyl-CoA dehydrogenase deficiency and long-chain acyl-CoA dehydrogenase deficiency in the mouse

Keith B. Cox, Doug A. Hamm, David S. Millington¹, Dietrich Matern², Jerry Vockley³, Piero Rinaldo², Carl A. Pinkert, William J. Rhead⁴, J. Russell Lindsey and Philip A. Wood[‡]

<http://hmg.oupjournals.org/cgi/content/full/10/19/2069>



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