

**VERY LONG-CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY (VLCADD)  
REFERENCES**

(VLCAD DEFICIENCY)

1. Andresen SB, Olpin S, Kvittingen EA, Augoustides-Savvopoulou P, Lindhout D, Halley DJJ, Vianey-Saban C, Wanders RJA, IJlst L, Schroeder LD, Bolund L, Gregersen N. "DNA-based prenatal diagnosis for very-long-chain acyl-CoA dehydrogenase deficiency", *J Inher Metab Dis* 1999; 22: 281-285.
2. Andresen BS, Olpin S, Poorthuis BJ, Scholte HR, Vianey-Saban C, Wanders R, IJlst L, Morris A, Pourfarzam M, Bartlett K Baumgartner ER, deKlerk JB, Schroeder LD, Corydon TJ, Lund H, Winter V, Bross P, Bolund L, Gregersen N. "Clear Correlation of Genotype with Disease Phenotype in Very-Long-Chain Acyl-CoA Dehydrogenase Deficiency," *Am J Hum Genet* 1999; 64(2): 479-494.
3. Bartlett K, Eaton S. "Mitochondrial beta-oxidation," *Eur J Biochem.* 2004 Feb;271(3):462-9.
4. Bonnet D, Martin D, deLonlay P, Villain E, Jouvet P, Rabier D, Brivet M, Saudubray JM. "Arrhythmias and Conduction Defects as Presenting Symptoms of Fatty Acid Oxidation Disorders in Children," *Circulation* 1999; 100(22): 2248-2253.
5. Brown-Harrison MC, Nada MA, Sprecher H, Vianey-Saban C, Farquhar J, Gilladoga AC, Roe CR. "Very Long Chain Acyl-CoA Dehydrogenase Deficiency: Successful Treatment of Acute Cardiomyopathy," *Biochem Molec Med* 1996; 58: 59-65.
6. Cairns AP, O'Donoghue PM, Patterson VH, Brown JH. "Very-long-chain acyl-coenzyme A dehydrogenase deficiency- a new cause of myoglobinuric acute renal failure," *Nephrol Dial Transplant* 2000; 15(8): 1232-4.
7. Costa CG, Dorland L, Tavares de Almeida I, Jakobs C, Duran m, Poll-The BT. "The effect of fasting, long-chain triglyceride load and carnitine loan on plasma long-chain acylcarnitine levels in mitochondrial very long-chain acyl-CoA dehydrogenase deficiency," *J Inherit Metab Dis* 1998; 21(4): 391-399.
8. Cox KB, Hamm DA, Millington DS, Matern D, Vockley J, Rinaldo P, Pinkert CA, Rhead WJ, Lindsey JR, Wood PA. "Gestational, pathologic and biochemical differences between very long-chain acyl-CoA dehydrogenase deficiency and long-chain acyl-CoA dehydrogenase deficiency in the mouse," *Hum Mol Genet* 2001; 10(19): 2069-2077.
9. Cox GF, Souri M, Aoyama T, Rockenmacher S, Varvogli L, Rohr F, Hashimoto T, Korson MS. "Reversal of severe hypertrophic cardiomyopathy and excellent neuropsychologic outcome in verylong- chain acyl-coenzyme A dehydrogenase deficiency," *J Pediatr* 1998; 133(2): 247-253.
10. Doi T, Abo W, Tateno m, Hayashi K, Hori T, Nakada T, Fukao T, Takahashi Y, Terada N. "Milder childhood form of very long-chain acyl-CoA dehydrogenase deficiency in a 6-year-old Japanese boy," *Eur J Pediatr* 2000; 159(12): 908-911.
11. Fukao, T, Watanabe H, Orii KE, Takahashi Y, Hirano A, Kondo T, Yamaguchi S, Aoyama T, Kondo N. "Myopathic Form of Very-Long Chain Acyl-CoA Dehydrogenase Deficiency: Evidence for Temperature-Sensitive Mild Mutations in Both Mutant Alleles in a Japanese Girl," *Ped Res* 2001; 49(2): 227-231.
12. Giak SK, Carpenter K, Hammond J, Christodoulou J, Wilcken B. "Quantitative fibroblast acylcarnitine profiles in mitochondrial fatty acid beta-oxidation defects: phenotype/metabolite correlations," *Mol Genet Metab* 2002; 76 (4): 327
13. Gregersen N, Andresen BS, Corydon MJ, Corydon TJ, Olsen RK, Bolund L, Bross P. "Mutation analysis in mitochondrial fatty acid oxidation defects: Exemplified by acyl-CoA dehydrogenase deficiencies, with special focus on genotype-phenotype relationship," *Hum Mutat* 2001; 18 (3): 169-189.
14. Gregersen N, Bross P, Andresen BS. "Genetic defects in fatty acid beta-oxidation and acyl-CoA dehydrogenases. Molecular pathogenesis and genotype-phenotype relationships," *Eur J Biochem.* 2004 Feb;271(3):470-82.
15. Guertl B, Noehammer C, Hoefler G. "Metabolic cardiomyopathies," *Int J Exp Pathol* 2000; 81(6): 349-372.
16. Hasegawa T, Hori N, Du W. "A case of impairment of mitochondrial fatty acid beta-oxidation," *Keio J Med* 2002; 51(2): 100-106.

17. He G, Yang BZ, Roe DS, Teramoto R, Aleck K, Grebe TA, Roe CR, Ding JH. "Identification of Two Novel Mutations in the Hypoglycemic Phenotype of Very Long Chain Acyl-CoA Dehydrogenase Deficiency," *Biochem Biophys Res Commun* 1999; 264(2): 483-487.
18. Kluge S, Kuhnelt P, Block A, Merkel M, Gocht A, Lukacs Z, Kohlschutter A, Kreymann G. "A young woman with persistent hypoglycemia, rhabdomyolysis, and coma: Recognizing fatty acid oxidation defects in adults", *Crit Care Med* 2003; 31(4): 1273-1276.
19. Martinez G, Jimenez-Sanchez G, Divry P, Vianey-Saban C, Riudor E, Rodes M, Briones P, Ribes A. "Plasma free fatty acids in mitochondrial fatty acid oxidation defects", *Clinica Chimica Acta* 1997; 67:143-154.
20. Mathur A, Sims HF, Gopalakrishnan D, Gibson B, Rinaldo P, Vockley J, Hug G, Strauss AW. "Molecular Heterogeneity in Very-Long-Chain Acyl-CoA dehydrogenase Deficiency Causing Pediatric Cardiomyopathy and Sudden Death", *Circulation* 1999; 99: 1337-1343.
21. Merinero B, Pascual SIP, Perez-Cerda C, Gangoiti J, Castro M, Garcia MJ, Castroviejo IP, Vianey-Saban C, Andresen B, Gregersen N, Ugarte M. "Adolescent myopathic presentation in two sisters with very long-chain acyl-CoA dehydrogenase deficiency", *J Inherit Metab Dis* 1999; 22(7): 802-810.
22. Millington DS. "Interpretation and follow-up of abnormal newborn screening results from MS/MS", 2004 Newborn Screening & Genetics Testing Symposium, May 3, 2004, Atlanta, GA
23. Morris AA, Leonard JV. "Early recognition of metabolic decompensation", *Arch Dis Child*. 1997 Jun;76(6):555-6.
24. Morris AAM, Lenoard JV. "Improving the Outcome for Fatty Acid Oxidation Disorders", *J Pediatr Gastroenterol Nutr* 2000; 31(4): 367-370.
25. Morris AA, Turnbull DM. "Fatty acid oxidation defects in muscle", *Curr Opin Neurol*. 1998 Oct;11(5):485-90.
26. Nada MA, Vianey-Saban C, Roe CR, Ding J-H, Mathieu M, Wappner RS, Bialer MG, McGlynn JA, Mandon G. "Prenatal Diagnosis of Mitochondrial Fatty Acid oxidation Defects" *Prenatal Diagn* 1996; 16: 117-124.
27. Onkenhout W, Venizelos V, Scholte HR, DeKlerk JBC, Poorthuis BJHM. "Intermediates of unsaturated fatty acid oxidation are incorporated in triglycerides but no in phospholipids in tissues from patients with mitochondrial B-oxidation defects". *J Inherit Metab Dis* 2001; 24: 337-344.
28. OMIM- Online Mendelian Inheritance in Man; ACYL-CoA DEHYDROGENASE, VERY LONGCHAIN, DEFICIENCY OF- \*201475
29. Osorio JH, Lluch M, Ribes A. "Analysis of organic acids after incubation with (16-2H3)palmitic acid in fibroblasts from patients with mitochondrial beta-oxidation defects", *J Inherit Metab Dis*. 2003;26(8):795-803.
30. Parini R, Menni F, Garavaglia B, Fesslova V, Melotti D, Massone ML, Lamantea E, Rimoldi M, Vizziello P, Gatti R. "Acute, severe cardiomyopathy as main symptom of late-onset very long-chain acyl-coenzyme A dehydrogenase deficiency", *Eur J Pediatr* 1998; 157: 992-995.
31. Pons R, Cavadini P, Baratta S, Invernizzi F, Lamantea E, Garavaglia B, Taroni F. "Clinical and Molecular Heterogeneity in Very-Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency", *Pediatr Neurol* 2000; 22(2): 98-105.
32. Rinaldo P, Matern D. "Disorders of fatty acid transport and mitochondrial oxidation: challenges and dilemmas of metabolic evaluation", *Genet Med* 2000; 2(6): 338-44.
33. Rinaldo P, Matern D, Bennett MJ. "Fatty Acid Oxidation Disorders", *Annu Rev Physiol* 2002; 64: 477-502.
34. Roe CR. "Inherited disorders of mitochondrial fatty acid oxidation: a new responsibility for the neonatologist", *Semin Neonatol* 2002; 7: 37-47.
35. Roe CR, Ding J. Mitochondrial fatty acid oxidation disorders. In: C. Scriver, A.L. Beaudet, W. Sly and D. Valle, Editors, *The Metabolic and Molecular Basis of Inherited Disease* (eighth ed.), McGraw-Hill, New York (2001), pp. 2297-2326.
36. Roe CR, Sweetman L, Roe DS, David F, Brunengraber H. "Treatment of cardiomyopathy and rhabdomyolysis in long-chain fat oxidation disorders using an anaplerotic odd-chain triglyceride", *J Clin Invest* 2002; 110(2): 259-269.
37. Roe CR, Wiltse HE, Sweetman L, Alvarado LL. "Death caused by perioperative fasting and sedation in a child with unrecognized very long chain acyl-coenzyme A dehydrogenase deficiency", *J Pediatr* 2000; 136(3): 397-399.

38. Roe DS, Vianey-Saban C, Sharma S, Zabot MT, Roe CR. "Oxidation of unsaturated fatty acids by human fibroblasts with very-long-chain acyl-CoA dehydrogenase deficiency: aspects of substrate specificity and correlation with clinical phenotype",
39. Ruiz-Sanz JI, Aldamiz-Echevarria L, Arrizabalaga J, Aquino L, Jimeno P, Perez-Nanclares G, Sanjurjo P. "Polyunsaturated fatty acid deficiency during dietary treatment of very long-chain acyl-CoA dehydrogenase deficiency. Rescue with soybean oil", *J Inherit Metab Dis* 2001; 24(4): 493-503.
40. Scholte HR, VanCoster RNA, deJonge PC, Poorthuis BJHM, Jeneson JAL, Andresen BS, Gregersen N, deKlerk JBC, Busch HFM. "Myopathy in very-long-chain acyl-CoA dehydrogenase deficiency: clinical and biochemical differences with the fatal cardiac phenotype", *Neuromuscul Disord* 1999; 9(5): 313-319.
41. Sim KG, Hammond J, Wilcken B. "Strategies for the diagnosis of mitochondrial fatty acid  $\beta$ -oxidation disorders", *Clin Chim Acta* 2002; 323: 37-58.
42. Skladal D, Sass JO, Geiger H, Geiger R, Mann C, Vreken P, Wanders RJA, Trawoger R. "Complications in Early Diagnosis and Treatment of Two Infants With Long-Chain Fatty Acid  $\beta$ -Oxidation Defects", *J Pediatr Gastroenterol Nutr* 2000; 31(4): 448-452.
43. Sluysmans T, Tuerlinckx D, Hubinont C, Verellen-Dumoulin C, Brivet M, Vianey-Saban C. "Very long chain acyl-coenzyme A dehydrogenase deficiency in two sibling: Evolution after prenatal diagnosis and prompt management", *J Pediatr* 1997; 131(3): 444-446.
44. Solis JO, Singh RH. "Management of fatty acid oxidation disorders: A survey of current treatment strategies", *J Am Diet Assoc* 2002; 102(12): 1800-1803.
45. Souri M, Aoyama T, Yamaguchi S, Hashimoto T. "Relationship between structure and substrate-chainlength specificity of mitochondrial very-long-chain acyl-coenzyme A dehydrogenase", *Eur J Biochem* 1998; 257(3): 592-598.
46. Sovik O. "Inborn errors of amino acid and fatty acid metabolism with hypoglycemia as a major clinical manifestation", *Acta Paediatr Scand.* 1989 Mar;78(2):161-70.
47. Spiekertotter U, Schwahn B, Korall H, Trefz FK, Andresen BS, Wendel U. "Very-long-chain acylcoenzyme A dehydrogenase (VLCAD) deficiency: monitoring of treatment by carnitine/acylcarnitine analysis in blood spots", *Acta Paediatr* 2000; 89: 492-495.
48. Spiekertotter U, Sun B, Zytovicz T, Wanders R, Strauss AW, Wendel U. "MS/MS-based newborn and family screening detects asymptomatic patients with very-long-chain acyl-CoA Dehydrogenase deficiency", *J Pediatr.* 2003 Sep;143(3):335-42.
49. Spiekertotter U, Tenenbaum T, Heusch A, Wendel U. "Cardiomyopathy and Pericardial Effusion in Infancy Point to a Fatty Acid  $\beta$ -Oxidation Defect After Exclusion of an Underlying Infection", *Pediatr Cardiol* 2002; 4: epub.
50. Strauss AW, Powell CK, Hale DE, Anderson MM, Ahuja A, Brackett JC, Sims HF. "Molecular basis of human mitochondrial very-long-chain acyl-CoA dehydrogenase deficiency causing cardiomyopathy and sudden death in childhood", *Proc Natl Acad Sci USA* 1995; 92: 10496-10500.
51. Strauss AW, Spiekertotter U, Ding L, Tokunaga C, Zykovitz T, Marsden D, Rinaldo P, Bennett M. "The changing spectrum of fatty acid oxidation disorders post-newborn screening", *Mol Genet and Metab* 2004; 81: 156-7.
52. Straussberg R, Harel L, Varsano I, Elpeleg ON, Shamir R, Amir J. "Recurrent Myoglobinuria as a Presenting Manifestation of Very Long Chain Acyl Coenzyme A Dehydrogenase Deficiency", *Pediatrics* 1997; 99(6): 894-896.
53. Straussberg R, Strauss AW. "A Novel Mutation of Late-Onset Very-Long-Chain Acyl-CoA Dehydrogenase Deficiency", *Pediatr Neurol* 2002; 27(2): 136-137.
54. Takusa Y, Fukao T, Kimura M, Uchiyama A, Abo W, Tsuboi Y, Hirose S, Fujioka H, Kondo, N, Yamaguchi S. "Identification and Characterization of Temperature-Sensitive Mild Mutations in Three Japanese Patients with Nonsevere Forms of Very-Long-Chain Acyl-CoA Dehydrogenase Deficiency", *Mol Genet Metab* 2002; 75(3): 227-234.
55. Tamaoki Y, Kimura M, Hasegawa Y, Iga M, Inoue M, Yamaguchi S. "A survey of Japanese patients with mitochondrial fatty acid  $\beta$ -oxidation and related disorders as detected from 1985 to 2000", *Brain Dev* 2002; 24: 675-680. Touma EH, Rashed MS, Vianey-Saban C, Sakr A, Divry P, Gregersen N, Andresen BS. "A severe genotype with favourable outcome in very long chain acyl-CoA dehydrogenase deficiency", *Arch Dis Child* 2001; 84(1) 58-60.

56. Vianey-Saban C, Divry P, Brivet M, Nada M, Zabot M-T, Mathieu M, Roe C. "Mitochondrial verylong-chain acyl-coenzyme A dehydrogenase deficiency: clinical characteristics and diagnostic considerations in 30 patients", *Clin Chim Acta* 1998; 269: 43-62.
57. Watanabe H, Orii KE, Fukao T, Song X-Q, Aoyama T, IJlst L, Ruiten J, Wanders RJA, Kondo N. "Molecular Basis of Very Long Chain Acyl-CoA Dehydrogenase Deficiency in Three Israeli Patients: Identification of a Complex Mutant Allele With P65L and K247Q Mutations, the Former Being an Exonic Mutation Causing Exon 3 Skipping", *Hum Mutat* 2000; 15(5) 430-438.
58. Wood JC, Magera MJ, Rinaldo P, Seashore MR, Strauss AW, Friedman A. "Diagnosis of Very Long Chain Acyl-Dehydrogenase Deficiency From an Infant's newborn Screening Card", *Pediatrics* 2001; 108(1): e19.
59. Yoon HR, Strauss AW, Yoo HW. "Sudden death in a Korean infant with very long-chain acyl-CoA dehydrogenase deficiency", *J Inherit Metab Dis* 2001; 24: 407-408.
60. Zhang LF, Ding JH, Yang BZ, He GC, Roe C. "Characterization of the bidirectional promoter region between the human genes encoding VLCAD and PSD-95", *Genomics*. 2003 Dec;82(6):660-8.