

LONG-CHAIN HYDROXYACYL-CoA DEHYDROGENASE (LCHAD) REFERENCES
(LCHAD; MITOCHONDRIAL TRIFUNCTIONAL PROTEIN DEFECT; LONG-CHAIN HYDROXYACYL-CoA DEHYDROGENASE; LONG-CHAIN 3-HYDROXYACYL-CoA DEHYDROGENASE DEFICIENCY)

1. Amirkhan RH, Timmons CF, Brown KO, Weinberger MJ, Bennett MJ. "Clinical, biochemical, and morphologic investigations of a case of long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency," *Arch Pathol Lab Med.* 1997 Jul; 121(7): 730-4.
2. Bartlett K, Eaton S. "Mitochondrial beta-oxidation", *Eur J Biochem.* 2004 Feb; 271(3): 462-9.
3. Batey RG. "Acute fatty liver of pregnancy: is it genetically predetermined?" *Am J Gastroenterol.* 1996 Nov; 91(11): 2262-4.
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. Carpenter KH, Wilcken B. "Neonatal diagnosis of long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency and implications for newborn screening by tandem mass spectrometry," *J Inherit Metab Dis.* 1999 Oct; 22(7): 840-1.
6. Chakrapani A, Olpin S, Cleary M, Walter JH, Wraith JE, Besley GT. "Trifunctional protein deficiency: three families with significant maternal hepatic dysfunction in pregnancy not associated with E474Q mutation," *J Inherit Metab Dis.* 2000 Dec; 23(8): 826-34.
7. Costa CG, Dorland L, Holwerda U, de Almeida IT, Poll-The BT, Jakobs C, Duran M. "Simultaneous analysis of plasma free fatty acids and their 3-hydroxy analogs in fatty acid beta-oxidation disorders," *Clin Chem.* 1998 Mar; 44(3): 463-71.
8. Das AM, Fingerhut R, Wanders RJ, Ullrich K. "Secondary respiratory chain defect in a boy with long chain 3-hydroxyacyl-CoA dehydrogenase deficiency: possible diagnostic pitfalls," *Eur J Pediatr.* 2000 Apr; 159(4): 243-6.
9. den Boer ME, Wanders RJ, Morris AA, IJlst L, Heymans HS, Wijburg FA. "Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency: clinical presentation and follow-up of 50 patients," *Pediatrics.* 2002 Jan; 109(1): 99-104.
10. den Boer ME, IJlst L, Wijburg FA, Oostheim W, van Werkhoven MA, van Pampus MG, Heymans HS, Wanders RJ. "Heterozygosity for the common LCHAD mutation (1528g>C) is not a major cause of HELLP syndrome and the prevalence of the mutation in the Dutch population is low," *Pediatr Res.* 2000 Aug; 48(2): 151-4.
11. Dionisi-Vici C, Garavaglia B, Burlina AB, Bertini E, Saponara I, Sabetta G, Taroni F. "Hypoparathyroidism in mitochondrial trifunctional protein deficiency," *J Pediatr.* 1996 Jul; 129(1): 159-62.
12. Divry P, Vianey-Saban C, Mathieu M. "Determination of total fatty acids in plasma: cis-5-tetradecenoic acid (C14: 1 omega-9) in the diagnosis of long-chain fatty acid oxidation defects," *J Inherit Metab Dis.* 1999 May; 22(3): 286-8.
13. 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.
14. Gillingham MB, Connor WE, Matern D, Rinaldo P, Burlingame T, Meeuws K, Harding CO. "Optimal dietary therapy of long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency," *Mol Genet Metab.* 2003 Jun; 79(2): 114-23.
15. Gillingham M, Van Calcar S, Ney D, Wolff J, Harding C. "Dietary management of long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHADD). A case report and survey," *J Inherit Metab Dis.* 1999 Apr; 22(2): 123-31.
16. 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.
17. Grunewald S, Bakkeren J, Wanders RA, Wendel U. "Neonatal lethal mitochondrial trifunctional protein deficiency mimicking a respiratory chain defect," *J Inherit Metab Dis.* 1997 Nov; 20(6): 835-6.
18. Guertl B, Noehammer C, Hoefler G. "Metabolic cardiomyopathies," *Int J Exp Pathol* 2000; 81(6): 349-372.

19. Harding CO, Gillingham MB, van Calcar SC, Wolff JA, Verhoeve JN, Mills MD. "Docosahexaenoic acid and retinal function in children with long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency," *J Inherit Metab Dis*. 1999 May; 22(3): 276-80.
20. Hintz SR, Matern D, Strauss A, Bennett MJ, Hoyme HE, Schelley S, Kobori J, Colby C, Lehman NL, Enns GM. "Early neonatal diagnosis of long-chain 3-hydroxyacyl coenzyme a dehydrogenase and mitochondrial trifunctional protein deficiencies," *Mol Genet Metab*. 2002 Feb; 75(2): 120-7.
21. Ibdah JA, Bennett MJ, Rinaldo P, Zhao Y, Gibson B, Sims HF, Strauss AW. "A fetal fatty-acid oxidation disorder as a cause of liver disease in pregnant women," *N Engl J Med*. 1999 Jun 3; 340(22): 1723-31.
22. Ibdah JA, Dasouki MJ, Strauss AW. "Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency: variable expressivity of maternal illness during pregnancy and unusual presentation with infantile cholestasis and hypocalcaemia," *J Inherit Metab Dis*. 1999 Oct; 22(7): 811-4.
23. Ibdah JA, Tein I, Dionisi-Vici C, Bennett MJ, IJlst L, Gibson B, Wanders RJ, Strauss AW. "Mild trifunctional protein deficiency is associated with progressive neuropathy and myopathy and suggests a novel genotype-phenotype correlation," *J Clin Invest*. 1998 Sep 15; 102(6): 1193-9.
24. Ibdah JA, Yang Z, Bennett MJ. "Liver disease in pregnancy and fetal fatty acid oxidation defects," *Mol Genet Metab*. 2000 Sep-Oct; 71(1-2): 182-9.
25. Ibdah JA, Zhao Y, Viola J, Gibson B, Bennett MJ, Strauss AW. "Molecular prenatal diagnosis in families with fetal mitochondrial trifunctional protein mutations," *J Pediatr*. 2001 Mar; 138(3): 396-9.
26. IJlst L, Oostheim W, Ruiten JP, Wanders RJ. "Molecular basis of long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency: identification of two new mutations," *J Inherit Metab Dis*. 1997 Jul; 20(3): 420-2.
27. IJlst L, Ruiten JP, Hoovers JM, Jakobs ME, Wanders RJ. "Common missense mutation G1528C in long chain 3-hydroxyacyl-CoA dehydrogenase deficiency. Characterization and expression of the mutant protein, mutation analysis on genomic DNA and chromosomal localization of the mitochondrial trifunctional protein alpha subunit gene," *J Clin Invest*. 1996 Aug 15; 98(4): 1028-33.
28. Jones PM, Burlina AB, Bennett MJ. "Quantitative measurement of total and free 3-hydroxy fatty acids in serum or plasma samples: short-chain 3-hydroxy fatty acids are not esterified," *J Inherit Metab Dis*. 2000 Nov; 23(7): 745-50.
29. Jones PM, Butt Y, Bennett MJ. "Accumulation of 3-hydroxy-fatty acids in the culture medium of long chain L-3-hydroxyacyl CoA dehydrogenase (LCHAD) and mitochondrial trifunctional protein-deficient skin fibroblasts: implications for medium chain triglyceride dietary treatment of LCHAD deficiency," *Pediatr Res*. 2003 May; 53(5): 783-7. Epub 2003 Mar 05.
30. Jones PM, Butt YM, Bennett MJ. "Effects of odd-numbered medium-chain fatty acids on the accumulation of long-chain 3-hydroxy-fatty acids in long-chain L-3-hydroxyacyl CoA dehydrogenase and mitochondrial trifunctional protein deficient skin fibroblasts," *Mol Genet Metab*. 2004 Feb; 81(2): 96-9.
31. Jones PM, Moffitt M, Joseph D, Harthcock PA, Boriack RL, Ibdah JA, Strauss AW, Bennett MJ. "Accumulation of free 3-hydroxy fatty acids in the culture media of fibroblasts from patients deficient in long-chain l-3-hydroxyacyl-CoA dehydrogenase: a useful diagnostic aid," *Clin Chem*. 2001; 47(7): 1190-4.
32. Johnson DW, Trinh MU. "Analysis of isomeric long-chain hydroxy fatty acids by tandem mass spectrometry: application to the diagnosis of long-chain 3-hydroxyacyl CoA dehydrogenase deficiency," *Rapid Commun Mass Spectrom*. 2003; 17(2): 171-5.
33. Koneke GC, Wanders RJ, Handfed F. "Striking improvement of muscle strength under creative therapy in a patient with long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency," *J Inherit Metab Dis*. 2003; 26(1): 67-8.
34. Lee, JE, Yoon HR, Paik KH, Hwang SJ, Shim JW, Chang YS, Park WS, Strauss AW, Jin DK. "A case of mitochondrial trifunctional protein deficiency diagnosed by acylcarnitine profile and DNA analysis in a dried blood spot of a 4-day-old boy," *J Inherit Metab Dis*. 2003; 26(4): 403-6.
35. Lund AM, Leonard JV. "Feeding difficulties in long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency," *Arch Dis Child*. 2001 Dec; 85(6): 487-8.
36. Lund AM, Lund AM, Dixon MA, Vreken P, Leonard JV, Morris AAM. "What is the role of medium chain triglycerides in the management of long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency?" *J Inherit Metab Dis*. 2003; 26(4): 353-360.

37. Lund AM, Lund AM, Dixon MA, Vreken P, Leonard JV, Morris AAM. "Plasma and erythrocyte fatty acid concentrations in long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency," *J Inherit Metab Dis.* 2003; 26(4): 410-2.
38. Lundy CT, Shield JP, Kvittingen EA, Vinorum OJ, Trimble ER, Morris AA. "Acute respiratory distress syndrome in long-chain 3-hydroxyacyl-CoA dehydrogenase and mitochondrial trifunctional protein deficiencies," *J Inherit Metab Dis.* 2003; 26(6): 537-41.
39. Maitra A, Domiati-Saad R, Yost N, Cunningham G, Rogers BB, Bennett MJ. "Absence of the G1528C (E474Q) mutation in the alpha-subunit of the mitochondrial trifunctional protein in women with acute fatty liver of pregnancy," *Pediatr Res.* 2002 May; 51(5): 658-61.
40. 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; 267:143-154.
41. Matern D, Schehata BM, Shekhawa P, Strauss AW, Bennett MJ, Rinaldo P. "Placental floor infarction complicating the pregnancy of a fetus with long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency," *Mol Genet Metab.* 2001 Mar; 72(3): 265-8.
42. Mauriala T, Herzig KH, Heinonen M, Idziak J, Auriola S. "Determination of long-chain fatty acid acylcoenzyme A compounds using liquid chromatography-electrospray ionization tandem mass spectrometry," *J Chromatogr B Analyt Technol Biomed Life Sci.* 2004 Sep 5; 808(2): 263-8.
43. 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
44. Miyajima H, Kohno S, Tomiyama H, Kaneko E. "Effects of a low-dose L-carnitine supplement on an adult patient with mitochondrial trifunctional protein deficiency," *Muscle Nerve.* 1999 Mar; 22(3): 429-30.
45. Miyajima H, Orii KE, Shindo Y, Hashimoto T, Shinka T, Kuhara T, Matsumoto I, Shimizu H, Kaneko E. "Mitochondrial trifunctional protein deficiency associated with recurrent myoglobinuria in adolescence," *Neurology.* 1997 Sep; 49(3): 833-7.
46. Morris AA, Clayton PT, Surtees RA, Leonard JV. "Clinical outcomes in long-chain 3-hydroxyacylcoenzyme A dehydrogenase deficiency," *J Pediatr.* 1997 Dec; 131(6): 938.
47. Morris AA, Leonard JV. "Early recognition of metabolic decompensation," *Arch Dis Child.* 1997 Jun; 76(6): 555-6.
48. Morris AAM, Lenoard JV. "Improving the Outcome for Fatty Acid Oxidation Disorders," *J Pediatr Gastroenterol Nutr* 2000; 31(4): 367-370.
49. Morris AA, Turnbull DM. "Fatty acid oxidation defects in muscle," *Curr Opin Neurol.* 1998 Oct; 11(5): 485-90.
50. 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.
51. 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.
52. OMIM- Online Mendelian Inheritance in Man; HYDROXYACYL-CoA DEHYDROGENASE/3-KETOACYL-CoA THIOLASE/ENOYL-CoA HYDRATASE, ALPHA SUBUNIT; HADHA- *600890
53. OMIM- Online Mendelian Inheritance in Man; HYDROXYACYL-CoA DEHYDROGENASE/3-KETOACYL-CoA THIOLASE/ENOYL-CoA HYDRATASE, BETA SUBUNIT; HADHB- *143450
54. Orii KE, Aoyama T, Souri M, Jiang LL, Orii KO, Hayashi S, Yamaguchi S, Kondo N, Orii T, Hashimoto T. "Formation of the enzyme complex in mitochondria is required for function of trifunctional beta-oxidation protein," *Biochem Biophys Res Commun.* 1996 Feb 27; 219(3): 773-7.
55. Orii KE, Aoyama T, Wakui K, Fukushima Y, Miyajima H, Yamaguchi S, Orii T, Kondo N, Hashimoto T. "Genomic and mutational analysis of the mitochondrial trifunctional protein beta-subunit (HADHB) gene in patients with trifunctional protein deficiency," *Hum Mol Genet.* 1997 Aug; 6(8): 1215-24.
56. Osorio JH, Lluch M, Ribes A. "Analysis of organic acids after incubation with (16-2H3) politic acid in fibroblasts from patients with mitochondrial beta-oxidation defects," *J Inherit Metab Dis.* 2003; 26(8): 795-803.
57. Otto LR, Boriack RL, Marsh DJ, Kum JB, Eng C, Burlina AB, Bennett MJ. "Long-chain L 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency does not appear to be the primary cause of

- lipid myopathy in patients with Bannayan-Riley-Ruvalcaba syndrome (BRRS),” *Am J Med Genet.* 1999 Mar 5; 83(1): 3-5.
58. 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.
 59. Rinaldo P, Matern D, Bennett MJ. “Fatty Acid Oxidation Disorders,” *Annu Rev Physiol* 2002; 64: 477-502.
 60. Roe CR. “Inherited disorders of mitochondrial fatty acid oxidation: a new responsibility for the neonatologist,” *Semin Neonatol* 2002; 7: 37-47.
 61. 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.
 62. Russell-Eggitt IM, Leonard JV, Lund AM, Manoj B, Thompson DA, Morris AA. “Cataract in long chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHADD),” *Ophthalmic Genet.* 2003 Mar; 24(1): 49-57.
 63. Scaglia F. “Long-Chain Acyl CoA Dehydrogenase Deficiency,” www.emedicine.com 2002.
 64. Schrijver-Wieling I, van Rens GH, Wittebol-Post D, Smeitink JA, de Jager JP, de Klerk HB, van Lith GH. “Retinal dystrophy in long chain 3-hydroxy-acyl-coA dehydrogenase deficiency,” *Br J Ophthalmol.* 1997 Apr; 81(4): 291-4.
 65. Schwab KO, Ensenauer R, Matern D, Uyanik G, Schnieders B, Wanders RA, Lehnert W. “Complete deficiency of mitochondrial trifunctional protein due to a novel mutation within the beta-subunit of the mitochondrial trifunctional protein gene leads to failure of long-chain fatty acid beta-oxidation with fatal outcome,” *Eur J Pediatr.* 2003 Feb; 162(2): 90-5. Epub 2003 Jan 09.
 66. Shen JJ, Matern D, Millington DS, Hillman S, Feezor MD, Bennett MJ, Qumsiyeh M, Kahler SG, Chen YT, Van Hove JL. “Acylcarnitines in fibroblasts of patients with long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency and other fatty acid oxidation disorders,” *J Inherit Metab Dis.* 2000 Feb; 23(1): 27-44.
 67. Sim KG, Hammond J, Wilcken B. “Strategies for the diagnosis of mitochondrial fatty acid oxidation disorders,” *Clin Chim Acta* 2002; 323: 37-58.
 68. Sims HF, Brackett JC, Powell CK, Treem WR, Hale DE, Bennett MJ, Gibson B, Shapiro S, Strauss AW. “The molecular basis of pediatric long chain 3-hydroxyacyl-CoA dehydrogenase deficiency associated with maternal acute fatty liver of pregnancy,” *Proc Natl Acad Sci U S A.* 1995 Jan 31; 92(3): 841-5.
 69. Skladal D, Sass JO, Geiger H, Geiger R, Mann C, Vreken P, Wanders RJ, Trawogger R. “Complications in early diagnosis and treatment of two infants with long-chain fatty acid beta-oxidation defects,” *J Pediatr Gastroenterol Nutr.* 2000 Oct; 31(4): 448-52.
 70. 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.
 71. Spiekerkoetter U, Khuchua Z, Yue Z, Bennett MJ, Strauss AW. “General mitochondrial trifunctional protein (TFP) deficiency as a result of either alpha- or beta-subunit mutations exhibits similar phenotypes because mutations in either subunit alter TFP complex expression and subunit turnover,” *Pediatr Res.* 2004 Feb; 55(2): 190-6. Epub 2003 Nov 19.
 72. Spiekerkoetter U, Khuchua A, Yue Z, Strauss AW. “The early-onset phenotype of mitochondrial trifunctional protein deficiency: A lethal disorder with multiple tissue involvement,” *J Inher Metab Dis* 2004; 27(2): 294-296.
 73. Spiekerkoetter U, Sun B, Khuchua Z, Bennett MJ, Strauss AW. “Molecular and phenotypic heterogeneity in mitochondrial trifunctional protein deficiency due to beta-subunit mutations,” *Hum Mutat.* 2003 Jun; 21(6): 598-607.
 74. Strauss AW, Spiekerkoetter 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.
 75. Tamaoki Y, Kimura M, Hasegawa Y, Iga M, Inoue M, Yamaguchi S. “A survey of Japanese patients with mitochondrial fatty acid oxidation and related disorders as detected from 1985 to 2000,” *Brain Dev* 2002; 24: 675-680.
 76. Thiel C, Baudach S, Schnackenberg U, Vreken P, Wanders RJ. “Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency: neonatal manifestation at the first day of life presenting with tachypnoea,” *J Inherit Metab Dis.* 1999 Oct; 22(7): 839-40.

77. Treem WR, Shoup ME, Hale DE, Bennett MJ, Rinaldo P, Millington DS, Stanley CA, Riely CA, Hyams JS. "Acute fatty liver of pregnancy, hemolysis, elevated liver enzymes, and low platelets syndrome, and long chain 3-hydroxyacyl-coenzyme A dehydrogenase deficiency," *Am J Gastroenterol*. 1996 Nov; 91(11): 2293-300.
78. Tyni T, Johnson M, Eaton S, Pourfarzam M, Andrews R, Turnbull DM. "Mitochondrial fatty acid betaoxidation in the retinal pigment epithelium," *Pediatr Res*. 2002 Oct; 52(4): 595-600.
79. Tyni T, Kivela T, Lappi M, Summanen P, Nikoskelainen E, Pihko H. "Ophthalmologic findings in long chain 3-hydroxyacyl-CoA dehydrogenase deficiency caused by the G1528C mutation: a new type of hereditary metabolic chorioretinopathy," *Ophthalmology*. 1998 May; 105(5): 810-24.
80. Tyni T, Majander A, Kalimo H, Rapola J, Pihko H. "Pathology of skeletal muscle and impaired respiratory chain function in long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency with the G1528C mutation," *Neuromuscul Disord*. 1996 Oct; 6(5): 327-37.
81. Tyni T, Palotie A, Viinikka L, Valanne L, Salo MK, von Döbeln U, Jackson S, Wanders R, Venizelos N, Pihko H. "Long-chain 3-hydroxyacyl-coenzyme A dehydrogenase deficiency with the G1528C mutation: clinical presentation of thirteen patients," *J Pediatr*. 1997 Jan; 130(1): 67-76.
82. Tyni T, Pihko H. "Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency," *Acta Paediatr*. 1999 Mar; 88(3): 237-45.
83. Tyni T, Rapola J, Palotie A, Pihko H. "Hypoparathyroidism in a patient with long-chain 3-hydroxyacylcoenzyme A dehydrogenase deficiency caused by the G1528C mutation," *J Pediatr*. 1997 Nov; 131(5): 766-8.
84. Van Maldergem L, Tuerlinckx D, Wanders RJ, Vianey-Saban C, Van Hoof F, Martin JJ, Fourneau C, Gillerot Y, Bachy A. "Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency and early-onset liver cirrhosis in two siblings," *Eur J Pediatr*. 2000 Jan-Feb; 159(1-2): 108-12.
85. Venizelos N, von Döbeln U, Hagenfeldt L. "Fatty acid oxidation in fibroblasts from patients with defects in beta-oxidation and in the respiratory chain," *J Inherit Metab Dis*. 1998 Jun; 21(4): 409-15.
86. Ventura FV, Ruiten JP, IJlst L, de Almeida IT, Wanders RJ. "Lactic acidosis in long-chain fatty acid beta-oxidation disorders," *J Inherit Metab Dis*. 1998 Aug; 21(6): 645-54.
87. Yamazaki H, Torigoe K, Numata O, Haniu H, Uchiyama A, Ogawa Y, Kaneko U, Imamura M, Hasegawa S. "Mitochondrial trifunctional protein deficiency in a lethal neonate," *Pediatr Int*. 2004 Apr; 46(2): 178-80.
88. Yang Z, Yamada J, Zhao Y, Strauss AW, Ibdah JA. "Prospective screening for pediatric mitochondrial trifunctional protein defects in pregnancies complicated by liver disease," *JAMA*. 2002 Nov 6; 288(17): 2163-6.
89. Yang Z, Zhao Y, Bennett MJ, Strauss AW, Ibdah JA. "Fetal genotypes and pregnancy outcomes in 35 families with mitochondrial trifunctional protein mutations," *Am J Obstet Gynecol*. 2002 Sep; 187(3): 715-20.