

CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY (CACT) REFERENCES
(*SOLUTE CARRIER FAMILY 25 (CARNITINE/ACYLCARNITINE TRANSLOCASE), MEMBER 20;*
CARNITINE-ACYLCARNITINE CARRIER; CACT DEFICIENCY)

1. Al Aqeel AI, Rashid MS, Ruiter JP, Ijlst L, Wanders RJ. "A novel molecular defect of the carnitine acylcarnitine translocase gene in a Saudi patient," *Clin Genet*. 2003 Aug; 64(2): 163-5.
2. al Aqeel AI, Rashed MS, Wanders RJ. "Carnitine-acylcarnitine translocase deficiency is a treatable disease," *J Inherit Metab Dis* 1999 May; 22(3): 271-5.
3. Angelini C, Trevisan C, Isaya G, Pegolo G, Vergani L. "Clinical varieties of carnitine and carnitine palmitoyl transferase deficiency," *Clin Biochem*. 1987 Feb; 20(1): 1-7.
4. Bartlett K, Eaton S. "Mitochondrial beta-oxidation," *Eur J Biochem*. 2004 Feb; 271(3): 462-9.
5. Boles RG, Buck EA, Blitzer MG, Platt MS, Cowan TM, Martin SK, Yoon H, Madsen JA, Reyes-Mugica M, Rinaldo P. "Retrospective biochemical screening of fatty acid oxidation disorders in postmortem livers of 418 cases of sudden death in the first year of life," *J Pediatr* 1998; 132(6): 924-33.
6. Bonnet D, de Lonlay P, Gautier I, Rustin P, Rotig A, Kachaner J, Acar P, LeBidois J, Munnich A, Sidi D. "Efficiency of metabolic screening in childhood cardiomyopathies," *Eur Heart J* 1998; 19(5): 790-793.
7. 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.
8. Bougneres PF, Saudubray JM, Marsac C, Bernard O, Odievre M, Girard J. "Fasting hypoglycemia resulting from hepatic carnitine palmitoyl transferase deficiency," *J Pediatr*. 1981 May; 98(5): 742-6.
9. Brivet M, Boutron A, Slama A, Costa C, Thuillier L, Demaugre F, Rabier D, Saudubray JM, Bonnefont JP. "Defects in activation and transport of fatty acids," *J Inherit Metab Dis* 1999 Jun; 22(4): 428-41.
10. Brivet M, Slama A, Millington DS, Roe CR, Demaugre F, Legrand A, Boutron A, Poggi F, Saudubray JM. "Retrospective diagnosis of carnitine-acylcarnitine translocase deficiency by acylcarnitine analysis in the proband Guthrie card and enzymatic studies in the parents," *J Inherit Metab Dis*. 1996; 19(2): 181-4.
11. Brivet M, Slama A, Ogier H, Boutron A, Demaugre F, Saudubray JM, Lemonnier A. "Diagnosis of carnitine acylcarnitine translocase deficiency by complementation analysis," *J Inherit Metab Dis*. 1994; 17(3): 271-4.
12. Chalmers RA, Stanley CA, English N, Wigglesworth JS. "Mitochondrial carnitine-acylcarnitine translocase deficiency presenting as sudden neonatal death," *J Pediatr* 1997 Aug; 131(2): 220-5.
13. Costa C, Costa JM, Slama A, Boutron A, Vequaud C, Legrand A, Brivet M. "Mutational spectrum and DNA-based prenatal diagnosis in carnitine-acylcarnitine translocase deficiency," *Mol Genet Metab* 2003 Jan; 78(1): 68-73.
14. DiDonato S, Rimoldi M, Moise A, Bertagnoglio B, Uziel G. "Fatal ataxic encephalopathy and carnitine acetyltransferase deficiency: a functional defect of pyruvate oxidation?" *Neurology*. 1979 Dec; 29(12): 1578-83.
15. Galron D, Birk OS, Kazanovitz A, Moses SW, Hershkovitz E. "Carnitine-cylcarnitine translocase deficiency: Identification of a novel molecular defect in a Bedouin patient," *J Inher Metab Dis* 2004; 27(2): 267-273.
16. Guertl B, Noehammer C, Hoefler G. "Metabolic cardiomyopathies," *Int J Exp Pathol* 2000; 81(6): 349-372.
17. Hsu BY, Iacobazzi V, Wang Z, Harvie H, Chalmers RA, Saudubray JM, Palmieri F, Ganguly A, Stanley CA. "Aberrant mRNA splicing associated with coding region mutations in children with carnitine acylcarnitine translocase deficiency," *Mol Genet Metab* 2001 Sep-Oct; 74(1-2): 248-55.
18. Iacobazzi V, Naglieri MA, Stanley CA, Wanders RJ, Palmieri F. "The structure and organization of the human carnitine/acylcarnitine translocase (CACT1) gene2," *Biochem Biophys Res Commun* 1998 Nov 27; 252(3): 770-4.
19. Iacobazzi V, Pasquali M, Singh R, Matern D, Rinaldo P, di San Filippo CA, Palmieri F, Longo N. "Response to Therapy in Carnitine/Acylcarnitine Translocase (CACT) Deficiency Due to a Novel Missense Mutatio," *Am J Med Genet* 2004 126A(2): 150-155.

20. IJlst L, van Roermund CW, Iacobazzi V, Oostheim W, Ruiten JP, Williams JC, Palmieri F, Wanders RJ. "Functional analysis of mutant human carnitine acylcarnitine translocases in yeast," *Biochem Biophys Res Commun* 2001 Jan 26; 280(3): 700-6.
21. Jakobs BS, Wanders RJ. "Impaired peroxisomal fatty acid oxidation in human skin fibroblasts with a mitochondrial acylcarnitine/carnitine translocase deficiency," *J Inherit Metab Dis*. 1996; 19(2): 185-7.
22. Kerner J, Hoppel C. "Genetic Disorders of Carnitine Metabolism and Their Nutritional Management," *Ann Rev Nutr* 1998; 18: 179-206.
23. Lopriore E, Gemke RJ, Verhoeven NM, Jakobs C, Wanders RJ, Roeleveld-Versteeg AB, Poll-The BT. "Carnitine-acylcarnitine translocase deficiency: phenotype, residual enzyme activity and outcome," *Eur J Pediatr* 2001 Feb; 160(2): 101-4.
24. 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
25. Morris AA, Leonard JV. "Early recognition of metabolic decompensation," *Arch Dis Child*. 1997 Jun; 76(6): 555-6.
26. Morris AA, Olpin SE, Brivet M, Turnbull DM, Jones RA, Leonard JV. "A patient with carnitine acylcarnitine translocase deficiency with a mild phenotype," *J Pediatr* 1998 Mar; 132(3 Pt 1): 514-6.
27. Morris AA, Turnbull DM. "Fatty acid oxidation defects in muscle," *Curr Opin Neurol*. 1998 Oct; 11(5): 485-90.
28. Niezen-Koning KE, van Spronsen FJ, IJlst L, Wanders RJ, Brivet M, Duran M, Reijngoud DJ, Heymans HS, Smit GP. "A patient with lethal cardiomyopathy and a carnitine-acylcarnitine translocase deficiency," *J Inherit Metab Dis*. 1995; 18(2): 230-2.
29. Nuoffer JM, de Lonlay P, Costa C, Roe CR, Chamoles N, Brivet M, Saudubray JM. "Familial neonatal SIDS revealing carnitine-acylcarnitine translocase deficiency," *Eur J Pediatr* 2000 Jan-Feb; 159(1-2): 82-5.
30. Ogawa A, Yamamoto S, Kanazawa M, Takayanagi M, Hasegawa S, Kohno Y. "Identification of two novel mutations of the carnitine/acylcarnitine translocase (CACT) gene in a patient with CACT deficiency," *J Hum Genet* 2000; 45(1): 52-5.
31. Olpin SE, Bonham JR, Downing M, Manning NJ, Pollitt RJ, Sharrard MJ, Tanner MS. "Carnitineacylcarnitine translocase deficiency--a mild phenotype," *J Inherit Metab Dis* 1997 Sep; 20(5): 714-5.
32. OMIM- Online Mendelian Inheritance in Man; CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY, INCLUDED- *212138
33. Pande SV, Brivet M, Slama A, Demaugre F, Aufrant C, Saudubray JM. "Carnitine-acylcarnitine translocase deficiency with severe hypoglycemia and auriculo-ventricular block. Translocase assay in permeabilized fibroblasts," *J Clin Invest*. 1993 Mar; 91(3): 1247-52.
34. Pande SV, Murthy MS. "Carnitine-acylcarnitine translocase deficiency: implications in human pathology," *Biochim Biophys Acta*. 1994 Jul 18; 1226(3): 269-76.
35. Parini R, Invernizzi F, Menni F, Garavaglia B, Melotti D, Rimoldi M, Salera S, Tosetto C, Taroni F. "Medium-chain triglyceride loading test in carnitine-acylcarnitine translocase deficiency: insights on treatment," *J Inherit Metab Dis* 1999 Aug; 22(6): 733-9.
36. Rinaldo P, Matern D, Bennett MJ. "Fatty Acid Oxidation Disorders," *Annu Rev Physiol* 2002; 64: 477-502.
37. 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.
38. Roe DS, Roe CR, Brivet M, Sweetman L. "Evidence for a short-chain carnitine-acylcarnitine translocase in mitochondria specifically related to the metabolism of branched-chain amino acids," *Mol Genet Metab* 2000 Jan; 69(1): 69-75.
39. Roschinger W, Muntau AC, Duran M, Dorland L, IJlst L, Wanders RJ, Roscher AA. "Carnitineacylcarnitine translocase deficiency: metabolic consequences of an impaired mitochondrial carnitine cycle," *Clin Chim Acta* 2000 Aug; 298(1-2): 55-68.
40. Rubio-Gozalbo ME, Vos P, Forget PP, Van Der Meer SB, Wanders RJ, Waterham HR, Bakker JA. "Carnitine-acylcarnitine translocase deficiency: case report and review of the literature," *Acta Paediatr*. 2003 Apr; 92(4): 501-4.
41. 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.

42. Stanley CA. "Dissecting the spectrum of fatty acid oxidation disorders," *J Pediatr* 1998 Mar; 132(3 Pt 1): 384-6.
43. 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.
44. van der Leij FR, Huijkman NC, Boomsma C, Kuipers JR, Bartelds B. "Genomics of the human carnitine acyltransferase genes," *Mol Genet Metab* 2000 Sep-Oct; 71(1-2): 139-53.
45. Yang BZ, Mallory JM, Roe DS, Brivet M, Strobel GD, Jones KM, Ding JH, Roe CR. "Carnitine/acylcarnitine translocase deficiency (neonatal phenotype): successful prenatal and postmortem diagnosis associated with a novel mutation in a single family," *Mol Genet Metab* 2001 May; 73(1): 64-70.