

## CITRULLINEMIA, TYPE II (ASS TYPE II) REFERENCES

(CITRULLINEMIA, TYPE II, ADULT-ONSET; CTLN2; CITRULLINEMIA, TYPE II, NEONATAL-ONSET; CHOLESTASIS, NEONATAL INTRAHEPATIC, CAUSED BY CITRIN DEFICIENCY)

1. Albayram S, Murphy J, Gailloud P, Moghekar A, Brunberg A. CT findings in the infantile form of citrullinemia. *Am J Neuroradiol* 2002 23:334-336.
2. Au WL, Lim TCC, Seow CC, Koh PL, Loh NK, Lim MSF, Tan IK, Yee WC. Serial diffusion-weighted magnetic resonance imaging in adult-onset citrullinaemia. *J Neurological Sciences* 2003 209:101-104.
3. Bachmann C. Outcome and survival of 88 patients with urea cycle disorders: a retrospective evaluation. *Eur J Pediatr*. 2003 Jun; 162(6): 410-416.
4. Bachmann, C. "Outcome and survival of 88 patients with urea cycle disorders: a retrospective evaluation," *Eur J Pediatr* 2003 162(6): 410-16.
5. Ban K, Sugiyama N, Sugiyama K, Wada Y, Suzuki T, Hashimoto T, Kobayashi K. A pediatric patient with classical citrullinemia who underwent living-related partial liver transplantation. *Transplantation* 2001 71:10 1495-1497.
6. Bennett MJ, Dear PRF, McGinlay JM, Gray RGF. Acute neonatal citrullinaemia. *J Inher Metab Dis* 1984 7:85.
7. Ben-Shalom E, Kobayashi K, Shaag A Yasuda T, Gao HZ, Saheki T, Bachmann C, Elpeleg O. Infantile citrullinemia caused by citrin deficiency with increased dibasic amino acids. *Molecular Genetics and Metabolism* 2002 77:202-208.
8. Burns SP, Woolf DA, Leonard JV, Iles RA. "Investigation of urea cycle enzyme disorders by 1H-NMR spectroscopy," *Clin Chim Acta*. 1992 Jul 31; 209(1-2): 47-60
9. Brusilow SW, Horwich AL. Urea Cycle Enzymes 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), MMBID Online (genetics.accessmedicine.com)
10. Brusilow SW, Maestri NE. "Urea cycle disorders: diagnosis, pathophysiology, and therapy," *Adv Pediatr*. 1996; 43:127-70.
11. Chadifaux-Vekemans B, Rabier D, Chabli A, Blanc A, Aupetit J, Bardet J, Kamoun P. Improving the prenatal diagnosis of citrullinemia using citrulline/ornithine + arginine in amniotic fluid. *Prenat Diagn* 2002; 22: 456-458.
12. Chen YF, Huang YC, Liu HM, Hwu WL. MRI in a case of adult-onset citrullinemia. *Neuroradiology* 2001 43: 845-847.
13. Choi CG, Yoo HW. Localized proton MR spectroscopy in infants with urea cycle defect. *Am J Neuroradiol* 2001 22:834-837.
14. Fletcher JM, Couper R, Moore D, Coxon R, Dorney S. Liver transplantation for citrullinaemia improves intellectual function. *J Inher Metab Dis* 1999 22: 581-586.
15. Fletcher JM, Couper R, Moore D, Coxon R, Dorney S. Liver transplantation for citrullinaemia improves intellectual function. *J Inher Metab Dis* 1999 22: 581-586.
16. Haberle J, Pauli S Linnebank M, Kleijer WJ, Bakker HD, Wanders RJA, Harms E, Koch HG. Structure of the human argininosuccinate synthetase gene and an improved system for molecular diagnostics in patients with classical and mild citrullinemia. *Hum Genet* 2002 110:327-333.
17. Hwu WL, Kobayashi K, Hu YH, Yamaguchi N, Saheki T, Chow SP, Wang JH. A Chinese adult onset type II citrullinaemia patient with 851del4/1638ins23 mutations in the SLC25A13 gene. *J Med Genet* 2001 38e23.
18. Ikeda S, Yazaki M, Takei Y, Ikegami T, Hashikura Y, Kawasaki S, Iwai M, Kobayashi K Saheki T. Type II (adult onset) citrullinaemia: clinical pictures and the therapeutic effect of liver transplantation. *J Neurol Neurosurg Psychiatry* 2001 71:663-670.
19. Ito T, Shiraki K, Sekoguchi K, Yamanaka T, Sugimoto K, Takase K, Tameda Y, Nakano T. Hepatocellular carcinoma associated with adult-type citrullinemia. *Digestive Disease and Sciences* 2000 45:11 2203-2206.
20. Kakinoki H, Kobayashi K, Terazono H, Nagata Y Saheki T. Mutations and DNA diagnoses of classical citrullinemia. *Human Mutation* 1997 9:250-259.
21. Kasahara N, Ohwada S, Takeichi T, Kaneko H, Tomohasa T, Morikawa A, Yonemura K, Asonuma K Tanaka K, Kobayashi K, Saheki T, Takeyoshi I, Morishita Y. *Transplantation* 2001 71:10 1495-7.

22. Kasahara M, Ohwada S, Takeichi T, Kaneko H, Tomomasa T, Morikawa A, Yonemura K, Asonuma K, Tanaka K, Kobayashi K, Saheki T, Takeyoshi I, Morishita Y. Living-related liver transplantation for type II citrullinemia using a graft from heterozygote donor. *Transplantation* 2001 71:1 157-159.
23. Kayler LK, Merion RM, Lee S, Sung RS, Punch JD, Rudich SM, Turcotte JG, Campbell DA Jr, Holmes R, Mcgee JC. Long-term survival after liver transplantation in children with metabolic disorders. *Pediatr Transplantation* 2002 6: 295-300.
24. Kawamoto S, Strong R, Kerlin P, Lynch SV, Steadman C, Kobayashi K, Nakagawa S, Matunami H, Akatsu T, Saheki T. Orthotopic liver transplantation for adult-onset type II citrullinaemia. *Clin Transplant* 1997 11:5 453-458.
25. Kobayashi K, Horiuchi M, Saheki T. Pancreatic secretory trypsin inhibitor as a diagnostic marker for adult-onset type II citrullinemia. *Hepatology* 1997 25:5 1160-1165.
26. Kobayashi K, Nakata M, Terazono H, Shinsato T, Saheki T. Pancreatic secretory trypsin inhibitor gene is highly expressed in the liver of adult-onset type II citrullinemia. *BEBS Letters* 1995 372:69-73.
27. Kobayashi K, Sinasac DS, Iijima M, Boright AP, Begum L, Lee JR, Yasuda T, Ikeda S, Hirano R, Terazono H, Crackower MA, Kondo I, Tsui LC, Scherer SW, Saheki T. The gene mutated in adult-onset type II citrullinaemia encodes a putative mitochondrial carrier protein. *Nature Genetics* 1999 22: 159-163.
28. Lee B, Goss J. Long-term correction of urea cycle disorders. *J Pediatr* 2001 138:1 S62-S71.
29. Li CM, Chao HK, Liu YF, Su TS. A nonsense mutation is responsible for the RNA-negative phenotype in human citrullinaemia. *European J Hum Genetics* 2001 9:685-689.
30. Maruyama H, Ogawa M, Nishio T, Kobayashi K, Saheki T, Sunohara N. Citrullinemia type II in a 64-year-old man with fluctuating serum citrulline levels: mutations in the SLC25A13 gene. *J of Neurological Sciences* 2001 193:63.
31. Morris AA, Leonard JV. "Early recognition of metabolic decompensation," *Arch Dis Child*. 1997 Jun; 76(6): 555-6.
32. Naito E, Ito M, Matsuura S, Yokota I, Saijo T, Ogawa Y, Kitamura S, Kobayashi K, Saheki T, Nishimura Y, Sakura N, Kuroda Y. Type II citrullinaemia (citrin deficiency) in a neonate with hypergalactosaemia detected by mass screening. *J Inherit Metab Dis* 2002 25:71-76.
33. Ohura T, Kobayashi K, Abukawa D, Tazawa Y, Aikawa J, Sakamoto O, Saheki T, Inuma K. A novel inborn error of metabolism detected by elevated methionine and/or Galactose in newborn screening: neonatal intrahepatic cholestasis caused by citrin deficiency. *Eur J Pediatr* 2003 162:317-322.
34. Ohura T, Kobayashi K, Tazawa Y, Nishi I, Abukawa D, Sakamoto O, Inuma K, Saheki T. Neonatal presentation of adult-onset type II citrullinemia. *Hum Genet* 2001 108:87-90.
35. Okeda R, Tanaka M, Kawahara Y, Tokushige J, Imai T, Kameya K. Adult-type citrullinemia. *Acta Neuropathol* 1989 78:96-100.
36. Oshiro S, Kochinda T, Tana T, Yamazato M, Kobayashi K, Komine Y, Muratani H, Saheki T, Iseki K, Takishita, S. A patient with adult-onset type II citrullinemia on long-term hemodialysis: reversal of clinical symptoms and brain MRI findings. *Am J Kidney Dis* 2002 39:189-92.
37. Palmieri L, Pardo B, Lasorsa FM, del Arco A, Kobayashi K, Iijima M, Runswick MJ, Walker JE, Saheki T, Satrustegui J, Palmieri F. Citrin and aralar1 are Ca<sup>2+</sup>-stimulated aspartate/glutamate transporters in mitochondria. *The EMBO Journal* 2001 20:18 5060-5069.
38. Patejunas G, Lee B, Dennis JA, Healy PJ, Reeds PJ, Yu H, Frazer M, Mull B, Warman AW, Beudet AL, O'Brien WE. Evaluation of gene therapy for citrullinaemia using murine and bovine models. *J Inher Metab Dis* 1998 21(Suppl 1) 138-150.
39. Ruitenbeek W, Kobayashi K, Iijima M, Smeitink JAM et al. Moderate citrullinaemia without hyperammonaemia in a child with mutated and deficient argininosuccinate synthetase. *Annals of Clinical Biochemistry* 2003 40:1 102-111.
40. Saheki T, Kobayashi K, Iijima M, Horiuchi M, Begum L, Jalil MA, Li MX, Lu YB, Ushikai M, Tabata A, Moriyama M, Hsiao KJ, Yang Y. "Adult-onset type II citrullinemia and idiopathic neonatal hepatitis caused by citrin deficiency: involvement of the aspartate glutamate carrier for urea synthesis and maintenance of the urea cycle," *Mol Genet Metab*. 2004 Apr; 81 Suppl: 20-6.
41. Saheki T, Kobayashi K, Iijima M, Nishi I, Yasuda T, Yamaguchi N, Gao HZ, Jalil MA, Begum L, Li MX. Pathogenesis and pathophysiology of citrin (a mitochondrial aspartate glutamate carrier) deficiency *Metabolic Brain Disease* 2002 17:4 335-346.

42. Saheki T, Kobayashi K. Mitochondrial aspartate glutamate carrier (citrin) deficiency as the cause of adult-onset type II citrullinemia (CTLN2) and idiopathic neonatal hepatitis (NICCD). *J Hum Genet* 2002 333-341.
43. Sander J, Janzen N, Sander S, Steuerwald U, Das AM, Scholl S, Trefz FK, Koch HG, Häberle J, Korall H, Marquardt I, Korenke C. "Neonatal screening for citrullinaemia," *Eur J Pediatr* 2003 162(6): 417-20.
44. Sander J, Janzen N, Sander S, Steuerwald U, Das AM, Scholl S, Trefz FK, Koch HG, Häberle J, Korall H, Marquardt I, Korenke C. *Eur J Pediatr* 2003 162:6; 417-20.
45. Sass JO, Skladal D. Plasma concentrations and renal clearance of orotic acid in argininosuccinic acid synthetase deficiency. *Pediatr Nephrol* 1999 13:912-916.
46. Sauduray JM, Touati G, Delonlay P, Jouvett P, Narcy C, Laurent J, Rabier D, Kamoun P, Jan D, Revillon Y. Liver transplantation in urea cycle disorders. *Eur J Pediatr* 1999 158 [Suppl 2]: S55-S59.
47. Summar M, Tuchman M. "Urea Cycle Disorders Overview," [www.geneclinics.org](http://www.geneclinics.org)
48. Tamamori A, Fujimoto A, Okano Y, Kobayashi K, Saheki T, Tagami Y, Takei H, Shigematsu Y, Hata I, Ozaki H, Tokuhara D, Nishimura Y, Yorifuji T, Igarashi N, Ohura T, Shimizu T, Inui K, Sakai N, Abukawa D, Miyakawa T, Matsumori M, Ban K, Kaneko H, Yamano T. "Effects of citrin deficiency in the perinatal period: feasibility of newborn mass screening for citrin deficiency," *Pediatr Res*. 2004 Oct; 56(4): 608-14. Epub 2004 Aug 04.
49. Tamamori A, Okano Y, Ozaki H, Fujimoto A, Kajiwarra M, Fukuda K, Kobayashi K, Saheki T, Tagami Y, Yamano T. Neonatal intrahepatic cholestasis caused by citrin deficiency: severe hepatic dysfunction in an infant requiring liver transplantation. *Eur J Pediatr* 2002 161:609-613.
50. Tazawa Y, Kobayashi K, Ohura T, Abukawa D, Nishinomiya F, Hosoda Y, Yamashita M, Nagata I, Kono Y, Yasuda T, Yamaguchi N, Saheki T. Infantile cholestatic jaundice associated with adult-onset type II citrullinemia. *J of Pediatrics* 138:5 735-740.
51. Tazawa Y, Kobayashi K, Ohura T, Abukawa D, Nishinomiya F, Hosoda Y, Yamashita M, Nagata I, Kono Y, Yasuda T, Yamaguchi N, Saheki T. Infantile cholestatic jaundice associated with adult-onset type II citrullinemia. *J of Pediatr* 2001 138:5 735-740.
52. Tomomasa T, Kobayashi K, Kaneko H, Shimura H, Fukusato T, Tabata M, Inoue Y, Ohwada S, Kasahara M, Morishita Y, Kimura M, Saheki T, Morikawa A. Possible clinical and histologic manifestations of adult-onset type II citrullinemia in early infancy. *J of Pediatr* 2001 138:5 741-743.
53. Tsuboi Y, Fujino Y, Kobayashi K, Saheki T, Yamada T. High serum pancreatic secretory trypsin inhibitor before onset of type II citrullinemia. *Neurology* 2001 57:5 933-935.
54. Tuchman M, Yudkoff M. Blood levels of ammonia and nitrogen scavenging amino acids in patients with inherited hyperammonemia. *Molecular Genetics and Metabolism* 1999 66: 10-15.
55. Vilaseca MA, Kobayashi K, Briones P, Lambruschini N, Campistol J, Tabata A, Alomar A, Rodes M, Lluh M, Saheki T. Phenotype and genotype heterogeneity in Mediterranean citrullinemia. *Molecular Genetics and Metabolism* 2001 74: 396-398.
56. Wilson CJ, Lee PJ, Leonard JV. Plasma glutamine and ammonia concentrations in ornithine carbamoyltransferase deficiency and citrullinaemia. *J Inher Metab Dis*. 2001 24:691-695.
57. Yamaguchi N, Kobayashi K, Yasuda T, Nishi I, Iijima M, Nakagawa M, Osame M, Kondo I, Saheki T. Screening of SLC25A13 mutations in early and late onset patients with citrin deficiency in the Japanese population: identification of two novel mutations and establishment of multiple DNA diagnosis methods for nine mutations. *Human Mutation* 2002 19:122-130.
58. Yasuda T, Yamaguchi N, Kobayashi K, Nishi I, Horinouchi H, Jalil A, Li MX, Ushikai M, Iijima M, Kondo I, Saheki T. Identification of two novel mutations in the SLC25A13 gene and detection of seven mutations in 102 patients with adult-onset type II citrullinemia. *Hum Genet* 2000 107:537-545
59. Yasuda T, Yamaguchi N, Kobayashi K, Nishi I, Horinouchi H, Jalil A, Li MX, Ushikai M, Iijima M, Kondo I, Saheki T. Identification of two novel mutations in the SLC25A13 gene and detection of seven mutations in 102 patients with adult-onset type II citrullinemia. *Hum Genet* 2000 107:537-545
60. Ye X, Whiteman B, Jerebtsova M, Batshaw ML. Correction of argininosuccinate synthetase (AS) deficiency in a murine model of citrullinemia with recombinant adenovirus carrying human AS cDNA. *Gene Therapy* 2000 7: 1777-1782.
61. Zamora SA, Pinto A, Scott RB, Parsons HG. Mitochondrial abnormalities of liver in two children with citrullinaemia. *J Inher Metab Dis* 1997 20: 509-516.