

## ARGINASE DEFICIENCY (ARG) REFERENCES

### (ARGINASE DEFICIENCY; HYPERARGININEMIA)

1. Antonozzi I, Leuzzi V. "Hyperargininaemia," *J Inherit Metab Dis* 1987; 10(2): 200.
2. Ash DE, Scolnick LR, Kanyo ZF, Vockley JG, Cederbaum SD, Christianson DW. "Molecular Basis of Hyperargininemia: Structure-Function Consequences of Mutations in Human Liver Arginase," *Molec Genet Metab* 1998; 64: 243-249.
3. Bachmann C, Colombo JP. "Diagnostic value of orotic acid excretion in heritable disorders of the urea cycle and in hyperammonemia due to organic acidurias," *Eur J Pediatr* 1980 Aug; 134(2): 109-13.
4. Bernar J, Hanson RA, Kern R, Phoenix B, Shaw KN, Cederbaum SD. "Arginase deficiency in a 12-year-old boy with mild impairment of intellectual function," *J Pediatr* 1986 Mar; 108(3): 432-5.
5. Braga AC, Vilarinho L, Ferreira E, Rocha H. "Hyperargininemia Presenting as Persistent Neonatal Jaundice and Hepatic Cirrhosis," *J Pediatr Gastroenterol Nutr* 1997; 24(2): 218-221.
6. Brockstedt M, Smit LM, de Grauw AJ, van der Klei-van Moorsel JM, Jakobs C. "A new case of hyperargininaemia: neurological and biochemical findings prior to and during dietary treatment," *Eur J Pediatr*. 1990 Feb; 149(5): 341-3.
7. 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).
8. Brusilow SW, Maestri NE. "Urea cycle disorders: diagnosis, pathophysiology, and therapy," *Adv Pediatr*. 1996; 43:127-70.
9. Burton BK. "Urea Cycle Disorders," *Clin Liver Dis* 2000; 4(4): 815-830.
10. Candito M, Bebin B, Vianey-Saban C, Rabier D, Bekri S, Sebag F, Chambon P, Kamoun P. "Arginase deficiency in two brothers," *J Inherit Metab Dis*. 1993; 16(6): 1054-6.
11. Cardoso ML, Martins E, Vasconcelos R, Vilarinho L, Rocha J. "Identification of a Novel R21X Mutation in the Liver-Type Arginase Gene (ARG1) in Four Portuguese Patients with Argininemia," *Hum Mutat* 1999; 14(4): 355-356.
12. Cederbaum SD, Moedjono SJ, Shaw KN, Carter M, Naylor E, Walzer M. "Treatment of hyperargininaemia due to arginase deficiency with a chemically defined diet," *J Inherit Metab Dis* 1982; 5(2): 95-9.
13. Cederbaum SD, Shaw KN, Spector EB, Verity MA, Snodgrass PJ, Sugarman GI. "Hyperargininemia with arginase deficiency," *Pediatr Res* 1979 Jul; 13(7): 827-33.
14. Cederbaum SD, Shaw KN, Valente M. "Hyperargininemia," *J Pediatr* 1977 Apr; 90(4): 569-73.
15. Cederbaum SD, Yu H, Grody WW, Kern RM, Yoo P, Iyer RK. "Arginases I and II: do their functions overlap?" *Mol Genet Metab*. 2004 Apr; 81 Suppl: 38-44.
16. Cowley DM, Bowling FG, McGill JJ, van Dongen J, Morris D. "Adult-onset Arginase deficiency," *J Inher Metab Dis* 1998; 21: 677-678.
17. Cutler P. "Arginase deficiency and phenylketonuria," *J Neurol Neurosurg Psychiatry* 1986 Sep; 49(9): 1090.
18. Endres W, Schaller R, Shin YS. "Diagnosis and treatment of argininaemia. Characteristics of arginase in human erythrocytes and tissues," *J Inherit Metab Dis* 1984; 7(1): 8.
19. Fuchshuber A, Marescau LB, Roth B, De Deyn PP, Sprenger HJ, Michalk DV. "Haemodialysis and continuous veno-venous haemofiltration in a patient with hyperargininaemia and acute renal failure," *J Inherit Metab Dis*. 1993; 16(5): 909-10.
20. Gatti R, Cerone R, Caruso U, Schiaffino MC, Ciccone O. "Biochemical diagnosis and follow-up in a new Italian patient with hyperargininaemia," *J Inherit Metab Dis*. 1993; 16(6): 1050.
21. Grillo MA, Colombatto S. "Arginine revisited: minireview article," *Amino Acids*. 2004 Jul; 26(4): 345-51.
22. Grody WW, Kern RM, Klein D, Dodson AE, Wissman PB, Barsky SH, Cederbaum SD. "Arginase deficiency manifesting delayed clinical sequelae and induction of a kidney arginase isozyme," *Hum Genet*. 1993 Mar; 91(1): 1-5.
23. Harrington JW, Stiefel M, Gianos E. "Arginase deficiency presenting with cerebral oedema and failure to thrive," *J Inher Metab Dis* 2000; 23: 517-518.

24. Hewson S, Clarke JT, Cederbaum S. "Prenatal diagnosis for arginase deficiency: a case study," *J Inherit Metab Dis*. 2003; 26(6): 607-10.
25. Hyland K, Smith I, Clayton PT, Leonard JV. "Impaired neurotransmitter amine metabolism in arginase deficiency," *J Neurol Neurosurg Psychiatry* 1985 Nov; 48(11): 1188-9.
26. Iyer RK, Yoo PK, Kern RM, Rozengurt N, Tsoa R, O'Brien WE, Yu H, Grody WW, Cederbaum SD. "Mouse Model for Human Arginase Deficiency," *Mol Cell Bio* 2002; 22(13): 4491-4498.
27. Jorda A, Portoles M, Rubio V, Capdevila A, Vilas J, Garcia-Pino J. "Liver fibrosis in arginase deficiency," *Arch Pathol Lab Med* 1987 Aug; 111(8): 691-2.
28. Kato T, Sano M, Mizutani N, Hayakawa C. "Homocitrullinuria and homoargininuria in hyperargininaemia," *J Inherit Metab Dis* 1988; 11(3): 261-5.
29. Kang SS, Wong PW, Melyn MA. "Hyperargininemia: effect of ornithine and lysine supplementation," *J Pediatr* 1983 Nov; 103(5): 763-5.
30. Kim PS, Iyer RK, Lu KV, Yu H, Karimi A, Kern RM, Tai DK, Cederbaum SD, Grody WW. "Expression of the liver form of arginase in erythrocytes," *Mol Genet Metab* 2002; 76: 100-110.
31. Lambert MA, Marescau B, Desjardins M, Laberge M, Dhondt JL, Dallaire L, De Deyn PP, Qureshi IA. "Hyperargininemia: intellectual and motor improvement related to changes in biochemical data," *J Pediatr*. 1991 Mar; 118(3): 420-4.
32. Lavulo LT, Emig FA, Ash DE. "Functional Consequences of the G235R Mutation in Liver Arginase Leading to Hyperargininemia," *Arch Biochem Biophys* 2002; 399(1): 49-55.
33. Marescau B, De Deyn PP, Qureshi IA, De Broe ME, Antonozzi I, Cederbaum SD, Cerone R, Chamoles N, Gatti R, Kang SS, et al. "The pathobiochemistry of uremia and hyperargininemia further demonstrates a metabolic relationship between urea and guanidinosuccinic acid," *Metabolism*. 1992 Sep; 41(9): 1021-4.
34. Marescau B, De Deyn PP, Lowenthal A, Qureshi IA, Antonozzi I, Bachmann C, Cederbaum SD, Cerone R, Chamoles N, Colombo JP, et al. "Guanidino compound analysis as a complementary diagnostic parameter for hyperargininemia: follow-up of guanidino compound levels during therapy," *Pediatr Res*. 1990 Mar; 27(3): 297-303.
35. Morris AA, Leonard JV. "Early recognition of metabolic decompensation," *Arch Dis Child*. 1997 Jun; 76(6): 555-6.
36. Naylor EW, Cederbaum SD. "Urinary pyrimidine excretion in arginase deficiency," *J Inherit Metab Dis* 1981; 4(4): 207-10.
37. OMIM- Online Mendelian Inheritance in Man; ARGININEMIA- \*207800.
38. Patel JS, van't Hoff WV, Leonard JV. "Arginase deficiency presenting with convulsions," *J Inherit Metab Dis*. 1994; 17(2): 254.
39. Picker JD, Puga AC, Levy HL, Marsden D, Shih VE, DeGirolami U, Ligon KL, Cederbaum SD, Kern RM, Cox GF. "Arginase Deficiency with Lethal Neonatal Expression: Evidence for the Glutamine Hypothesis of Cerebral Edema," *J Pediatr* 2003; 142(3): 349-352.
40. Prasad AN, Breen JC, Ampola MG, Rosman NP. "Argininemia: a treatable genetic cause of progressive spastic diplegia simulating cerebral palsy: case reports and literature review," *J Child Neuro* 1997; 12(5): 301-309.
41. Qureshi IA, Letarte J, Ouellet R, Batshaw ML, Brusilow S. "Treatment of hyperargininemia with sodium benzoate and arginine-restricted diet," *J Pediatr* 1984 Mar; 104(3): 473-6.
42. Qureshi IA, Letarte J, Ouellet R, Larochelle J, Lemieux B. "A new French-Canadian family affected by hyperargininaemia," *J Inherit Metab Dis* 1983; 6(4): 179-82.
43. Scarpa P, Faggioli R. "Hyperargininaemia: follow-up of a new case," *J Inherit Metab Dis*. 1995; 18(1): 80-1.
44. Scheuerle AE, McVie R, Beaudet AL, Shapira SK. "Arginase Deficiency Presenting as Cerebral Palsy," *Pediatrics* 1993; 91(5): 995-996.
45. Silva ES, Martins E, Cardoso ML, Barbot C, Vilarinho L, Medina M. "Liver transplantation in a case of argininaemia," *J Inher Metab Dis* 2001; 24: 885-887.
46. Simoni RE, de Oliveira CPH, Braga MJ, De Menezes, CR, Llerena Jr JC, Correia PS, Santa Rosa AA, Horovitz DG, Chaves CRM, de Oliveira MLC. "Hyperargininaemia: A late-diagnosed Brazilian case with increased urinary excretion of homocystine," *J Inher Metab Dis* 1997; 20(5): 715-716.
47. Snyderman SE, Sansaricq C, Norton PM, Goldstein F. "Argininemia treated from birth," *J Pediatr* 1979 Jul; 95(1): 61-3.

48. Summar M, Tuchman M. "Urea Cycle Disorders Overview," [www.geneclinics.org](http://www.geneclinics.org)
49. Terheggen HG, Lowenthal A, Colombo JP. "Clinical and biochemical findings in argininemia," *AdvExp Med Biol* 1982; 153:111-9.
50. Vilarinho L, Senra V, Vilarinho A, Barbosa C, Parvy P, Rabier D, Kamoun P. "A new case of argininaemia without spastic diplegia in a Portuguese male," *J Inherit Metab Dis*. 1990; 13(5): 751-2.
51. Yoshino M, Kubota K, Yoshida I, Murakami T, Yamashita F. "Argininemia: report of a new case and mechanisms of orotic aciduria and hyperammonemia," *Adv Exp Med Biol* 1982; 153:121-5.