

Amino Acid Metabolism Inborn Errors

[View the current Amino Acid Metabolism Inborn Errors InDepth page on BioPortfolio.com](http://www.bioportfolio.com/indepth/Amino_Acid_Metabolism_Inborn_Errors.html) (PDF)
(http://www.bioportfolio.com/indepth/Amino_Acid_Metabolism_Inborn_Errors.html) - Regularly Updated.

Recent Publications on Amino Acid Metabolism Inborn Errors:



- [Long-term needs of adult patients with organic acidaemias: outcome and prognostic factors.](#)
*BACKGROUND: With improvements in the treatment of children with organic...*17th November, 2009
Charles Dent Metabolic Unit, National Hospital for Neurology and- J Inherit Metab Dis. 2009
Aug;32(4):523-33. Epub 2009 Jul 23. ([DOI Direct Link](#))
- [Inherited disorders in the conversion of methionine to homocysteine.](#)
*During the last decade much important new information relating to the...*17th November, 2009
Department of Pediatrics, University Hospital Center and School of- J Inherit Metab Dis. 2009
Aug;32(4):459-71. Epub 2009 Jul 7. ([DOI Direct Link](#))
- [Cerebellar hemorrhage in a patient with propionic acidemia.](#)
*Cerebellar hemorrhage (CH) is a well-known complication in newborns. Among...*10th November, 2009
Neurology and Metabolism Department, Hospital Sant Joan de Deu,- Cerebellum. 2009
Sep;8(3):352-4. Epub 2009 May 26. ([DOI Direct Link](#))
- [Metabolic and hemodynamic advantages of an acetate-free citrate dialysate in a uremic case of congenital methylmalonic acidemia.](#)
*During the last decade much important new information relating to the...*3rd October, 2009
Division of Nephrology, Department of Medicine, Jichi Medical University,- Am J Kidney Dis. 2009
Oct;54(4):764-9. Epub 2009 Jul 4. ([DOI Direct Link](#))
- [Fractional anisotropy for assessment of white matter tracts injury in methylmalonic acidemia.](#)
*BACKGROUND: Methylmalonic acidemia (MMA) is a multifactorial autosomal...*3rd October, 2009
Department of Radiology, Xinhua Hospital, Medical School of Shanghai Jiao- Chin Med J (Engl). 2009
Apr 20;122(8):945-9.
- [Enzymatic evaluation of glutaric acidemia type 1 by an in vitro probe assay of acylcarnitine profiling using fibroblasts and electrospray ionization/tandem mass spectrometry \(MS/MS\).](#)
*Glutaric acidemia type 1 (GA1) is usually diagnosed with an accumulation...*30th September, 2009
Department of Pediatrics, Shimane University Faculty of Medicine, Izumo,- J Chromatogr B Analyt
Technol Biomed Life Sci. 2009 Sep 1;877(25):2648-51. ([DOI Direct Link](#))
- [Cystathionine gamma-lyase: Clinical, metabolic, genetic, and structural studies.](#)
*We report studies of six individuals with marked elevations of...*12th September, 2009
Department of Pediatrics, University of Colorado School of Medicine,- Mol Genet Metab. 2009
Aug;97(4):250-9. Epub 2009 Apr 9. ([DOI Direct Link](#))
- [Dynamic changes of striatal and extrastriatal abnormalities in glutaric aciduria type I.](#)
*In glutaric aciduria type I, an autosomal recessive disease of...*10th September, 2009
Department of Neuroradiology, University of Heidelberg, Heidelberg,- Brain. 2009 Jul;132(Pt

- 7):1764-82. Epub 2009 May 11. ([DOI Direct Link](#))
- [Epilepsy spectrum in cerebral creatine transporter deficiency.](#)
*Diaminopimelate (DAP) epimerase is a key enzyme for the biosynthesis of...*2nd September, 2009
- *Epilepsia*. 2009 Sep;50(9):2168-70. ([DOI Direct Link](#))
 - ['Glutaric aciduria type I--an easily diagnosable and treatable metabolic disorder'.](#)
*BACKGROUND: Acrodermatitis acidemica is a recently proposed term for the...*14th August, 2009
- *Indian J Pediatr*. 2009 May;76(5):562-3. ([DOI Direct Link](#))
 - [Mitochondrial trifunctional protein deficiency with recurrent rhabdomyolysis.](#)
*Rhabdomyolysis is an important clinical diagnosis. The differential...*11th August, 2009
Department of Pediatrics B, Schneider Children's Medical Center of Israel,- *Pediatr Neurol*. 2009 Jun;40(6):465-7. ([DOI Direct Link](#))
 - [Seizures versus dystonia in encephalopathic crisis of glutaric aciduria type I.](#)
*In more than two thirds of cases, glutaric aciduria type I begins in the...*11th August, 2009
Neurology Department, Sant Joan de Deu Hospital, Barcelona, Spain.- *Pediatr Neurol*. 2009 Jun;40(6):426-31. ([DOI Direct Link](#))
 - [Acrodermatitis enteropathica-like eruption in metabolic disorders: acrodermatitis dysmetabolica is proposed as a better term.](#)
*BACKGROUND: Acrodermatitis acidemica is a recently proposed term for the...*23rd July, 2009
Department of Dermatology, Faculty of Medicine, Hacettepe University,- *Pediatr Dermatol*. 2009 Mar-Apr;26(2):150-4. Epub 2009 Mar 11. ([DOI Direct Link](#))
 - [L-2-Hydroxyglutaric aciduria: pattern of MR imaging abnormalities in 56 patients.](#)
*PURPOSE: To describe the pattern of magnetic resonance (MR) imaging...*27th June, 2009
Dept of Child Neurology, VU Univ Medical Ctr, De Boelelaan 1117, 1081 HV- *Radiology*. 2009 Jun;251(3):856-65. ([DOI Direct Link](#))
 - [Creatine metabolism in combined methylmalonic aciduria and homocystinuria disease revisited.](#)
*BACKGROUND: Acrodermatitis acidemica is a recently proposed term for the...*20th May, 2009
- *Ann Neurol*. 2009 Apr;65(4):481-2; author reply 482-3. ([DOI Direct Link](#))

Resources from the [NCBI](#) used in this document, [NCBI's standard disclaimer applies](#).

Nothing in this document should be used in place of personal medical advice from your own qualified medical practitioner. See [BioPortfolio.com User Agreement](#)

Send comments and feedback to:

Peter Barfoot Managing Director, BioPortfolio Ltd.

UK Tel: (+44) 1300 321501

USA Voicemail and Fax: (+1) 415 680 2472

[Peter Barfoot peter.barfoot@bioportfolio.com](mailto:peter.barfoot@bioportfolio.com)

All rights reserved. All other trademarks recognized.

BioPortfolio Limited is registered in England & Wales at Stafford House, 10 Prince of Wales Road, Dorchester, Dorset, DT1 1PW, UK. No.3312883 VAT No. GB 744 6483 10

Copyright 1997-2009 - BioPortfolio Limited.

