

Maple Syrup Urine Disease

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Recent Publications on Maple Syrup Urine Disease:



- [Use of peritoneal dialysis and mesothelium in non primary renal conditions.](#)
*The management of a handful of non renal clinical conditions includes...*6th November, 2009
Laboratory of Experimental Nephrology, Ha'Emek Medical Center, Afula,- *Adv Perit Dial.* 2009;25:2-5.
- [Utilization of Blood Spot Testing for Metabolic-Genetic Disorders in Honduras: Is it Time for Newborn Screening?](#)
*Honduran infant mortality (20/1000) has fallen below the Latin American...*24th October, 2009
Division of Neonatology, Cincinnati Children's Hospital Medical Center,- *J Child Neurol.* 2009 Oct 21. ([DOI Direct Link](#))
- [Hepatocyte transplantation improves phenotype and extends survival in a murine model of intermediate maple syrup urine disease.](#)
*Maple syrup urine disease (MSUD; OMIM 248600) is an inborn error of...*14th October, 2009
Department of Human Genetics, University of Pittsburgh, Pennsylvania, USA.- *Mol Ther.* 2009 Jul;17(7):1266-73. Epub 2009 May 12. ([DOI Direct Link](#))
- [\[A case report of maple syrup urine disease\]](#)
*Maple syrup urine disease (MSUD) is a defect in the catabolism of the...*7th October, 2009
- *Zhongguo Dang Dai Er Ke Za Zhi.* 2009 Jul;11(7):599-600.
- [Contribution of the diffusion-weighted MRI in the diagnosis and follow-up of encephalopathy caused by maple syrup urine disease in a full-term newborn.](#)
*Honduran infant mortality (20/1000) has fallen below the Latin American...*2nd September, 2009
Department of Radiology, Health Sciences Program FAMERP Medical School,- *Arq Neuropsiquiatr.* 2009 Sep;67(3A):719-23.
- [Maple syrup urine disease in Cypriot families: identification of three novel mutations and biochemical characterization of the p.Thr211Met mutation in the E1alpha subunit.](#)
*We report five mutations, three of them novel, responsible for maple syrup...*1st September, 2009
Department of Biochemical Genetics, The Cyprus Institute of Neurology and- *Genet Test Mol Biomarkers.* 2009 Oct;13(5):657-64. ([DOI Direct Link](#))
- [Hepatocyte transplantation \(HTx\) corrects selected neurometabolic abnormalities in murine intermediate maple syrup urine disease \(iMSUD\).](#)
*Skvorak et al. [1] demonstrated the therapeutic efficacy of HTx in a...*25th August, 2009
Department of Pediatrics, University of Pittsburgh School of Medicine,- *Biochim Biophys Acta.* 2009 Oct;1792(10):1004-10. Epub 2009 Aug 19. ([DOI Direct Link](#))
- [The first use of N-carbamylglutamate in a patient with decompensated maple syrup urine disease.](#)

- Maple syrup urine disease (MSUD) is a defect in the catabolism of the...* 19th August, 2009
Department of Pediatrics, Ege University Medical Faculty, Izmir, Turkey.- *Metab Brain Dis.* 2009 Sep;24(3):409-14. Epub 2009 Aug 18. ([DOI Direct Link](#))
- [Protein phosphatase 2Cm is a critical regulator of branched-chain amino acid catabolism in mice and cultured cells.](#)
The branched-chain amino acids (BCAA) are essential amino acids required... 25th July, 2009
Division of Molecular Medicine, Department of Anesthesiology, David Geffen- *J Clin Invest.* 2009 Jun;119(6):1678-87. ([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](#))
 - [\[HPLC for the diagnosis and monitoring of phenylketonuria and ketoaciduria\]](#)
A simple, rapid, and sensitive HPLC is proposed to test six amino acids in... 18th July, 2009
- *Klin Lab Diagn.* 2009 May;(5):13-5.
 - [Cerebral edema and intracranial hypertension in an adult with maple syrup urine disease.](#)
The branched-chain amino acids (BCAA) are essential amino acids required... 10th July, 2009
- *Eur J Neurol.* 2009 Mar;16(3):e45-6. ([DOI Direct Link](#))
 - [Revisiting MSUD in Portuguese Gypsies: evidence for a founder mutation and for a mutational hotspot within the BCKDHA gene.](#)
Maple syrup urine disease (MSUD) is a rare autosomal recessive disorder of... 9th July, 2009
IPATIMUP- Institute of Pathology and Molecular Immunology of University of- *Ann Hum Genet.* 2009 May;73(Pt 3):298-303. ([DOI Direct Link](#))
 - [Molecular genetics of maple syrup urine disease in the Turkish population.](#)
In maple syrup urine disease (MSUD), disease-causing mutations can affect... 26th June, 2009
Department of General Pediatrics, Heinrich-Heine University, Dusseldorf,- *Turk J Pediatr.* 2009 Mar-Apr;51(2):97-102.
 - [Diagnostic value of proton MR spectroscopy and diffusion-weighted MR imaging in childhood inherited neurometabolic brain diseases and review of the literature.](#)
The purpose of this study is to evaluate parenchymal diffusion properties... 23rd June, 2009
Dokuz Eylul University Faculty of Medicine, Department of Radiology,- *Eur J Radiol.* 2009 Jun 18. ([DOI Direct Link](#))

Maple Syrup Urine Disease Patents:



- 7235529- [Brain-associated inhibitor of tissue-type plasminogen activator](#)
- 7368527- [HADDE71 polypeptides](#)
- 7368531- [Human secreted proteins](#)
- 7381800- [Antibodies to HBIMF63 polypeptide](#)
- 7390637- [Keratinocyte derived interferon](#)
- 7393934- [Human G-protein chemokine receptor \(CCR5\) HDGMR10](#)
- 7410799- [In vivo production and delivery of erythropoietin or insulinotropin for gene therapy](#)
- 7411051- [Antibodies to HDPPA04 polypeptide](#)

- 7415358- [Molecular toxicology modeling](#)
- 7426441- [Methods for determining renal toxins](#)
- 7427396- [AAV vectors for gene delivery to the lung](#)
- 7354725- [Kinase peptides and antibodies](#)
- 7329422- [Pharmaceutical compositions](#)
- 7311905- [Embryonic-like stem cells derived from post-partum mammalian placenta, and uses and methods of treatment using said cells](#)
- 7238531- [Method for interpreting tandem mass spectrometry data for clinical diagnosis](#)
- 7238660- [Albumin fusion proteins](#)
- 7238667- [Albumin fusion proteins](#)
- 7244621- [Method for interpreting tandem mass spectrometry data for clinical diagnosis of genetic disorders such as hypermethionemia](#)
- 7250404- [Lipid-mediated polynucleotide administration to deliver a biologically active peptide and to induce a cellular immune response](#)
- 7255874- [Biocompatible polymers and adhesives: compositions, methods of making and uses related thereto](#)
- 7255879- [Post-partum mammalian placenta, its use and placental stem cells therefrom](#)
- 7259151- [AAV virions with decreased immunoreactivity and uses therefor](#)
- 7291461- [Methods for identifying small molecules that modulate premature translation termination and nonsense mRNA decay](#)
- 7297545- [Clinical method for the genetic screening of newborns using tandem mass spectrometry and internal standards therefor](#)
- 7433520- [Nosologic system of diagnosis](#)
- 7446176- [12 human secreted proteins](#)
- 7527966- [Gene regulation in transgenic animals using a transposon-based vector](#)
- 7531300- [Method of diagnosing breast cancer](#)
- 7531364- [Clinical method for the genetic screening of newborns using tandem mass spectrometry](#)
- 7534606- [Placental stem cell and methods thereof](#)
- 7544518- [Rapid quantitative analysis of proteins or protein function in complex mixtures](#)
- 7544669- [Polynucleotide therapy](#)
- 7569384- [Albumin fusion proteins](#)
- 7592010- [Albumin fusion proteins](#)
- 7598031- [Method for the detection of gene transcripts in blood and uses thereof](#)
- 7608451- [Gene regulation in transgenic animals using a transposon-based vector](#)
- 7521424- [Albumin fusion proteins](#)
- 7521234- [Mammalian immortalized liver cell](#)
- 7507414- [Albumin fusion proteins](#)
- 7447594- [Molecular cardiotoxicology modeling](#)
- 7459531- [50 Human secreted proteins](#)
- 7462634- [N-\(pyridin-2-yl\)-sulfonamide derivatives](#)
- 7468276- [Placental stem cells](#)
- 7469185- [Primary rat hepatocyte toxicity modeling](#)
- 7482013- [Albumin fusion proteins](#)
- 7485435- [Simultaneous detection of metabolic enzyme activity and metabolite levels](#)
- 7498171- [Modulation of stem and progenitor cell differentiation, assays, and uses thereof](#)
- 7501123- [Human G-protein chemokine receptor \(CCR5\) HDGMR10](#)
- 7507413- [Albumin fusion proteins](#)
- 7613343- [Nosologic system of diagnosis](#)

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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)

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