

Multiple Carboxylase Deficiency

[View the current Multiple Carboxylase Deficiency InDepth page on BioPortfolio.com \(PDF \)](#)
(http://www.bioportfolio.com/indepth/Multiple_Carboxylase_Deficiency.html) - Regularly Updated.

Recent Publications on Multiple Carboxylase Deficiency:



- [A case of holocarboxylase synthetase deficiency with insufficient response to prenatal biotin therapy.](#)
*Holocarboxylase synthetase (HCS) deficiency is an inborn error of biotin...*18th November, 2009
Department of Pediatrics and Neonatology, Nagoya City University, Graduate- Brain Dev. 2009 Nov;31(10):775-8. Epub 2009 Feb 6. ([DOI Direct Link](#))
- [\[Gene mutation analyses in Chinese children with multiple carboxylase deficiency.\]](#)
*OBJECTIVE: To confirm the diagnosis of multiple carboxylase deficiency...*7th October, 2009
Department of Pediatric Endocrinology and Genetic Metabolism, Xinhua- Zhonghua Yi Xue Yi Chuan Xue Za Zhi. 2009 Oct;26(5):504-10. ([DOI Direct Link](#))
- [Selective screening for organic acidemias by urine organic acid GC-MS analysis in Brazil: fifteen-year experience.](#)
*BACKGROUND: The gas chromatography/mass spectrometry (GC/MS) method for...*14th February, 2009
Servico de Genetica Medica, Hospital de Clinicas, Porto Alegre, RS,- Clin Chim Acta. 2009 Feb;400(1-2):77-81. Epub 2008 Nov 1. ([DOI Direct Link](#))
- [Impaired biotinidase activity disrupts holocarboxylase synthetase expression in late onset multiple carboxylase deficiency.](#)
*Biotinidase catalyzes the hydrolysis of the vitamin biotin from...*5th February, 2009
Departamento de Biologia Molecular y Biotecnologia, Instituto de- J Biol Chem. 2008 Dec 5;283(49):34150-8. Epub 2008 Oct 9. ([DOI Direct Link](#))
- [Management of a patient with holocarboxylase synthetase deficiency.](#)
*We investigated in a patient with holocarboxylase synthetase deficiency,...*24th December, 2008
Department of Pediatrics, University of Colorado at Denver, CO, USA.- Mol Genet Metab. 2008 Dec;95(4):201-5. Epub 2008 Oct 29. ([DOI Direct Link](#))
- [Determination of 3-hydroxyisovalerylcarnitine and other acylcarnitine levels using liquid chromatography-tandem mass spectrometry in serum and urine of a patient with multiple carboxylase deficiency.](#)
*Due to its increased concentration in blood, 3-hydroxyisovalerylcarnitine...*23rd October, 2008
Laboratory of Hospital Pharmacy, Graduate School of Pharmaceutical- J Chromatogr B Analyt Technol Biomed Life Sci. 2008 Jul 15;870(2):154-9. ([DOI Direct Link](#))
- [Selective screening for inborn errors of metabolism and secondary methylmalonic aciduria in pregnancy at high risk district of neural tube defects: a human metabolome study by GC-MS in China.](#)
*,OBJECTIVE: Urease pretreatment-gas chromatography-mass spectrometry...*22nd October, 2008

Department of Pediatrics, First Affiliated Hospital, Jinan University,- Clin Biochem. 2008 May;41(7-8):616-20. Epub 2008 Feb 12. ([DOI Direct Link](#))

- [Microbial biotin protein ligases aid in understanding holocarboxylase synthetase deficiency.](#)
*The attachment of biotin onto the biotin-dependent enzymes is catalysed by...*30th August, 2008
School of Molecular and Biomedical Science, University of Adelaide, North- Biochim Biophys Acta. 2008 Jul-Aug;1784(7-8):973-82. Epub 2008 Apr 9. ([DOI Direct Link](#))
- [Organic acid disorders detected by urine organic acid analysis: twelve cases in Thailand over three-year experience.](#)
*BACKGROUND: Disorders of organic acid (OA) metabolism are generally...*1st August, 2008
Division of Medical Genetics, Department of Pediatrics, Faculty of- Clin Chim Acta. 2008 Jun;392(1-2):63-8. Epub 2008 Feb 23. ([DOI Direct Link](#))
- [\[Biotin deficiency in amino acid formula nutrition for an infant with milk protein allergy\]](#)
*We reported a 4-month-old girl with biotin deficiency caused by amino acid...*31st July, 2008
Department of Pediatrics, Mizonokuchi Hospital, Teikyo University School- Arerugi. 2008 May;57(5):552-7.
- [Reduced half-life of holocarboxylase synthetase from patients with severe multiple carboxylase deficiency.](#)
*Multiple carboxylase deficiency is a clinical condition caused by defects...*17th July, 2008
School of Molecular and Biomedical Science, The University of Adelaide,- Hum Mutat. 2008 Jun;29(6):E47-57. ([DOI Direct Link](#))
- [Selective screening for inborn errors of metabolism on clinical patients using tandem mass spectrometry in China: a four-year report.](#)
*We have initiated clinical selective screening for inborn errors of...*27th September, 2007
Department of Pediatric Endocrinology and Genetic Metabolism, Xinhua- J Inherit Metab Dis. 2007 Aug;30(4):507-14. Epub 2007 Mar 8. ([DOI Direct Link](#))
- [Biotin.](#)
*Holocarboxylase synthetase (HCS) is an enzyme that catalyzes biotin...*20th July, 2007
- Altern Med Rev. 2007 Mar;12(1):73-8.
- [1H NMR spectra of methylcitric acid in urine.](#)
*Methylcitric acid (3-carboxy-3-hydroxy-2-methyl-pentanedioic acid; MCA) is...*27th April, 2007
Faculty of Chemistry, Warsaw University of Technology, Warsaw, Poland.- J Inherit Metab Dis. 2007 Apr;30(2):263. Epub 2007 Feb 14. ([DOI Direct Link](#))
- [Holocarboxylase synthetase deficiency: report of one case.](#)
*Holocarboxylase synthetase (HCS) is an enzyme that catalyzes biotin...*25th April, 2007
Department of Pediatrics, China Medical University Hospital, Taichung.- Acta Paediatr Taiwan. 2006 Nov-Dec;47(6):309-11.

Multiple Carboxylase Deficiency Patents:



- 7613343- [Nosologic system of diagnosis](#)
- 7433520- [Nosologic system of diagnosis](#)
- 7368527- [HADDE71 polypeptides](#)
- 7291461- [Methods for identifying small molecules that modulate premature translation termination and nonsense mRNA decay](#)

- 7232667- [Keratinocyte growth factor-2 polynucleotides](#)
- 6693077- [Keratinocyte growth factor-2](#)
- 6503530- [Method of preventing development of severe metabolic derangement in inborn errors of metabolism](#)
- 5719035- [Assay for enzyme activity from a red blood sample using a direct microfluorometric assay](#)
- 5538857- [Assay for enzyme activity from a red blood sample using a direct microfluorometric assay](#)

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.

