

Brain Disorders Inborn Genetic Genetic Brain Disorders

[View the current Brain Disorders Inborn Genetic Genetic Brain Disorders InDepth page on BioPortfolio.com \(PDF\)](http://www.bioportfolio.com/indepth/Brain_Disorders_Inborn_Genetic_Genetic_Brain_Disorders.html) (http://www.bioportfolio.com/indepth/Brain_Disorders_Inborn_Genetic_Genetic_Brain_Disorders.html) - Regularly Updated.

Recent Publications on Brain Disorders Inborn Genetic Genetic Brain Disorders:



- [\[Studies on heredity rule of the first genealogy regarding fatal familial insomnia in Henan province\]](#)
*OBJECTIVE: To investigate the epidemiological, genealogic characteristic,...*11th November, 2009
Henan Provincial Center for Disease Control and Prevention, Zhengzhou- *Zhonghua Liu Xing Bing Xue Za Zhi.* 2009 Jan;30(1):1-5.
- [\[Cerebral creatine deficiency: First Spanish patients harbouring mutations in GAMT gene.\]](#)
*BACKGROUND AND OBJETIVE: Brain creatine (Cr) deficiencies are a group of...*7th November, 2009
Neuropediatria, Hospital Sant Joan de Deu y Centro de Investigacion- *Med Clin (Barc).* 2009 Nov 21;133(19):745-9. Epub 2009 Nov 5. ([DOI Direct Link](#))
- [\[Old and new ideas about genes and behaviour.\]](#)
*Genetic factors have long been recognized as contributors to variantion in...*7th November, 2009
Department of Molecular Biology, Umea University, Umea, Sweden.- *Hereditas.* 2009 Oct;146(5):198-203. ([DOI Direct Link](#))
- [\[Metabolic epilepsies: approaches to a diagnostic challenge.\]](#)
*Although inborn errors of metabolism (IEM) are a relatively rare cause of...*3rd November, 2009
Department of Pediatrics, University of British Columbia, Vancouver,- *Can J Neurol Sci.* 2009 Aug;36 Suppl 2:S67-72.
- [\[Duplication hotspots, rare genomic disorders, and common disease.\]](#)
*The human genome is enriched in interspersed segmental duplications that...*26th September, 2009
Department of Pediatrics, University of Washington, Seattle, WA 98195,- *Curr Opin Genet Dev.* 2009 Jun;19(3):196-204. Epub 2009 May 22. ([DOI Direct Link](#))
- [\[Population programs for the detection of couples at risk for severe monogenic genetic diseases.\]](#)
*Population genetic screening programs for carrier detection of severe...*9th September, 2009
Department of Community Genetics, Public Health Services, Ministry of- *Hum Genet.* 2009 Aug;126(2):247-53. Epub 2009 Apr 24. ([DOI Direct Link](#))
- [\[Primary disorders of metabolism and disturbed fetal brain development.\]](#)
*There exists a link between the in utero metabolic environment and the...*8th September, 2009
Section of Clinical Neurosciences, Department of Pediatrics and Child- *Clin Perinatol.* 2009 Sep;36(3):621-38. ([DOI Direct Link](#))
- [\[Heterogeneity of nervous system mitochondria: location, location, location!\]](#)
*Mitochondrial impairments have been associated with many neurological...*5th September, 2009

- Department of Neuroscience, University of Minnesota, Minneapolis, 55455,- Exp Neurol. 2009 Aug;218(2):293-307. Epub 2009 May 21. ([DOI Direct Link](#))
- [Re: CDKL5 mutations in boys with severe encephalopathy and early-onset intractable epilepsy.](#)
*OBJECTIVE: To investigate the epidemiological, genealogic characteristic,...*18th August, 2009 - Neurology. 2009 Jul 7;73(1):76-7; author reply 77. ([DOI Direct Link](#))
 - [Stroke in children: inherited and acquired factors and age-related variations in the presentation of 48 paediatric patients.](#)
*AIM: Stroke is relatively rare in children and the clinical presentation...*14th August, 2009 Division of Child Neurology, Department of Pediatrics, University La- Acta Paediatr. 2009 Jul;98(7):1130-6. Epub 2009 Apr 30. ([DOI Direct Link](#))
 - [99-mTc-HMPAO single photon emission computed tomography examinations in genetically determined neurometabolic disorders.](#)
*The aim of our study was to determine regional cerebral blood flow (rCBF)...*25th July, 2009 Department of Pediatrics, University of Szeged Albert Szent-Gyorgyi- Ideggyogy Sz. 2009 May 30;62(5-6):168-77.
 - [\[Genome research in pediatric field\]](#)
*Present article concerns about the recent advances of genomic research in...*25th July, 2009 Department of Genetic Disease Research, Tokyo Jikei University School of- Nippon Rinsho. 2009 Jun;67(6):1146-55.
 - [Genetic, molecular and clinical features of familial isolated pituitary adenomas.](#)
*Pituitary adenomas occur in a familial setting in about 5% of all cases,...*25th July, 2009 Department of Endocrinology, Centre Hospitalier Universitaire de Liege,- Horm Res. 2009 Apr;71 Suppl 2:116-22. Epub 2009 Apr 29. ([DOI Direct Link](#))
 - [Newborn screening for genetic disorders.](#)
*Newborn screening has become an integral part of the evaluation of more...*25th June, 2009 Department of Human Genetics, Division of Medical Genetics, Emory- Pediatr Clin North Am. 2009 Jun;56(3):505-13, Table of Contents. ([DOI Direct Link](#))
 - [Epilepsy, ataxia, sensorineural deafness, tubulopathy, and KCNJ10 mutations.](#)
*BACKGROUND: Five children from two consanguineous families presented with...*23rd May, 2009 Great Ormond Street Hospital-University College London, London, United- N Engl J Med. 2009 May 7;360(19):1960-70. ([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.

