

Brain Disorders Inborn Genetic

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*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 3. ([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.](#)
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Great Ormond Street Hospital-University College London, London, United- N Engl J Med. 2009 May 7;360(19):1960-70. ([DOI Direct Link](#))

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