

# Lesch-Nyhan Syndrome

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## Recent Publications on Lesch-Nyhan Syndrome:



- [Loss of mutant mitochondrial DNA harboring the MELAS A3243G mutation in human cybrid cells after cell-cell fusion with normal tissue-derived fibroblast cells.](#)  
*Mutant mitochondrial (mt) DNA variants are related to human disease and...* 10th March, 2010  
Department of Medical Genome Sciences, Graduate School of Frontier- Int J Mol Med. 2010 Jan;25(1):153-8.
- [Myelination disturbance in a patient with hyperuricemia and hyperserotoninemia combined with 18q deletion syndrome.](#)  
*We previously reported a male patient with an 18q21.3 deletion,...* 17th February, 2010  
Department of Pediatrics and Children's Health Center, Albert- Ideggyogy Sz. 2009 Nov 30;62(11-12):413-7.
- [Abnormal adenosine and dopamine receptor expression in lymphocytes of Lesch-Nyhan patients.](#)  
*Self-injurious behavior is the most outstanding feature of Lesch-Nyhan...* 17th December, 2009  
Divisions of Clinical Biochemistry and Internal Medicine, La Paz- Brain Behav Immun. 2009 Nov;23(8):1125-31. Epub 2009 Jul 25. ([DOI Direct Link](#))
- [Adenosine receptors and neurological disease: neuroprotection and neurodegeneration.](#)  
*Adenosine receptors modulate neuronal and synaptic function in a range of...* 25th September, 2009  
Institute of Biomedical and Life Sciences, University of Glasgow, Glasgow- Handb Exp Pharmacol. 2009;(193):535-87. ([DOI Direct Link](#))
- [Gene expression and mRNA editing of serotonin receptor 2C in brains of HPRT gene knock-out mice, an animal model of Lesch-Nyhan disease.](#)  
*Lesch-Nyhan disease (LND), a genetic disorder associated with motor and...* 24th September, 2009  
Laboratory of Molecular Genetics, International Association of Medical- J Clin Neurosci. 2009 Aug;16(8):1061-3. Epub 2009 May 26. ([DOI Direct Link](#))
- [Reflections on the brainstem dysfunction in neurologically disabled children.](#)  
*This article deals with the neurological basis of brainstem-related...* 23rd September, 2009  
Department of Child Neurology, National Center Hospital of Neurology and- Brain Dev. 2009 Aug;31(7):529-36. Epub 2009 Mar 28. ([DOI Direct Link](#))
- [Severe gouty arthritis and mild neurologic symptoms due to F199C, a newly identified variant of the hypoxanthine guanine phosphoribosyltransferase.](#)  
*A deficiency in hypoxanthine guanine phosphoribosyltransferase (HPRT)...* 15th September, 2009  
Hopital Lariboisiere, INSERM UMR-S 606, Paris, France.- Arthritis Rheum. 2009 Jul;60(7):2201-4. ([DOI Direct Link](#))
- [NAD metabolism in HPRT-deficient mice.](#)

*The activity of hypoxanthine-guanine phosphoribosyltransferase (HPRT) is...*15th September, 2009  
Dipartimento di Biologia Molecolare, Universita di Siena, Siena, Italia.- Metab Brain Dis. 2009  
Jun;24(2):311-9. Epub 2009 Mar 25. ([DOI Direct Link](#))

- [Reflections: neurology and the humanities. You had the choice, Martha.](#)

*The activity of hypoxanthine-guanine phosphoribosyltransferase (HPRT) is...*10th September, 2009  
Southern Connecticut State University, 501 Crescent Street, New Haven, CT- Neurology. 2009 Mar  
24;72(12):1106. ([DOI Direct Link](#))

- [Lesch-Nyhan disease: from mechanism to model and back again.](#)

*Lesch-Nyhan disease (LND) is a rare inherited disorder caused by mutations...*27th August, 2009  
Departments of Neurology and Human Genetics, Emory University School of- Dis Model Mech. 2009  
Mar-Apr;2(3-4):116-21. ([DOI Direct Link](#))

- [Hypoxanthine-guanine phosphoribosyl transferase regulates early developmental programming of dopamine neurons: implications for Lesch-Nyhan disease pathogenesis.](#)

*Hypoxanthine-guanine phosphoribosyltransferase (HPRT) deficiency results...*14th August, 2009  
School of Medicine, Paris Descartes University, 75006 Paris, France.- Hum Mol Genet. 2009 Jul  
1;18(13):2317-27. Epub 2009 Apr 2. ([DOI Direct Link](#))

- [Chronic peritoneal dialysis in a 4-year-old boy with Lesch-Nyhan disease.](#)

*Nephrolithiasis associated with inborn metabolic diseases is a very rare...*17th July, 2009  
- Pediatr Nephrol. 2009 May;24(5):1089-90. Epub 2008 Dec 19. ([DOI Direct Link](#))

- [Efficacy of rasburicase in hyperuricemia secondary to Lesch-Nyhan syndrome.](#)

*We report on a 16-day-old male with metabolic acidosis, hyperuricemia,...*10th April, 2009  
Department of Neurology, Hospital Sant Joan de Deu, Barcelona, Spain.- Am J Kidney Dis. 2009  
Apr;53(4):677-80. Epub 2008 Nov 6. ([DOI Direct Link](#))

- [Uric acid and cardiovascular risk.](#)

*Lesch-Nyhan disease (LND) is a rare inherited disorder caused by mutations...*7th February, 2009  
- N Engl J Med. 2009 Jan 29;360(5):539; author reply 540-1.

- [Nephrolithiasis related to inborn metabolic diseases.](#)

*Nephrolithiasis associated with inborn metabolic diseases is a very rare...*22nd January, 2009  
Centre de reference des maladies renales rares & Inserm U820, Hospices- Pediatr Nephrol. 2010  
Mar;25(3):415-24. Epub 2009 Jan 21. ([DOI Direct Link](#))

## Lesch-Nyhan Syndrome Clinical Trials:



- [Phase II Pilot Study of Aminoimidazole Carboxamide Riboside \(AICAR\), a Precursor of Purine Synthesis, for Lesch-Nyhan Disease](#)

## Lesch-Nyhan Syndrome Patents:



- 7442506- [Methods for detection of genetic disorders](#)
- 7510881- [Method and kit for the quantitative and/or qualitative detection of components in a sample](#)
- 7514099- [Lipid nanoparticle based compositions and methods for the delivery of biologically active molecules](#)
- 7517864- [RNA interference mediated inhibition of vascular endothelial growth factor and vascular endothelial growth factor receptor gene expression using short interfering nucleic acid \(siNA\)](#)
- 7521424- [Albumin fusion proteins](#)
- 7521446- [Haloaryl substituted aminopurines, compositions thereof, and methods of treatment therewith](#)
- 7541357- [Aryl- and heteroaryl-substituted tetrahydroisoquinolines and use thereof to block reuptake of norepinephrine, dopamine, and serotonin](#)
- 7544518- [Rapid quantitative analysis of proteins or protein function in complex mixtures](#)
- 7550148- [Targeting of Herpes simplex virus to specific receptors](#)
- 7550489- [Substituted pyridyoxo amides](#)
- 7557135- [Inhibitors of cytosolic phospholipase A.sub.2](#)
- 7507414- [Albumin fusion proteins](#)
- 7507413- [Albumin fusion proteins](#)
- 7501123- [Human G-protein chemokine receptor \(CCR5\) HDGMR10](#)
- 7446176- [12 human secreted proteins](#)
- 7446239- [SCA2 knockout animal and methods of use](#)
- 7459531- [50 Human secreted proteins](#)
- 7465580- [Non-cytotoxic oriP replicon](#)
- 7469185- [Primary rat hepatocyte toxicity modeling](#)
- 7473551- [Nano-mechanic microsensors and methods for detecting target analytes](#)
- 7482013- [Albumin fusion proteins](#)
- 7485431- [Identification of a family of secreted proteins in vascular endothelium](#)
- 7485732- [Substituted 3-alkyl and 3-alkenyl azetidine derivatives](#)
- 7488865- [Urocortin-deficient mice and uses thereof](#)
- 7560538- [Porcine isogloboside 3 synthase protein, cDNA, genomic organization, and regulatory region](#)
- 7563617- [Hybrid adenovirus/adeno-associated virus vectors and methods of use thereof](#)
- 7622250- [Clinically intelligent diagnostic devices and methods](#)
- 7635467- [Mammalian multipotent stem cells and compositions, methods of preparation and methods of administration thereof](#)
- 7635565- [Analyzing blood type with identification of patient by genotyping](#)
- 7641915- [Lipid nanoparticle based compositions and methods for the delivery of biologically active molecules](#)
- 7655244- [Targeted delivery of botulinum toxin for the treatment and prevention of trigeminal autonomic cephalgias, migraine and vascular conditions](#)
- 7659113- [Delivery system](#)
- 7667030- [RNA interference mediated inhibition of matrix metalloproteinase 13 \(MMP13\) gene expression using short interfering nucleic acid \(siNA\)](#)
- 7667053- [Bicyclic amides](#)
- 7670783- [Screening for neurotoxic amino acid associated with neurological disorders](#)
- 7683165- [RNA interference mediated inhibition of interleukin and interleukin receptor gene expression using short interfering nucleic acid \(siNA\)](#)
- 7618621- [Mammalian multipotent neural stem cells and compositions, methods of preparation and methods of administration thereof](#)
- 7613343- [Nosologic system of diagnosis](#)
- 7604990- [Methods of regulating differentiation in stem cells](#)
- 7566533- [Clinically intelligent diagnostic devices and methods](#)
- 7569384- [Albumin fusion proteins](#)
- 7569385- [Multipotent amniotic fetal stem cells](#)
- 7572785- [Substituted imidazoles as cannabinoid receptor modulators](#)
- 7576239- [Substituted amides](#)

- 7582292- [Adipose tissue derived stromal cells for the treatment of neurological disorders](#)
- 7592010- [Albumin fusion proteins](#)
- 7595430- [Methods and animal model for analyzing age-related macular degeneration](#)
- 7598060- [Rapid analysis of variations in a genome](#)
- 7599799- [Methods for using co-regulated genesets to enhance detection and classification of gene expression patterns](#)
- 7683166- [RNA interference mediated inhibition of interleukin and interleukin receptor gene expression using short interfering nucleic acid \(siNA\)](#)

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