

## Tay-Sachs Disease

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### Recent Publications on Tay-Sachs Disease:

- [ACOG Committee Opinion No. 442: Preconception and prenatal carrier screening for genetic diseases in individuals of Eastern European Jewish descent.](#)  
*Certain autosomal recessive disease conditions are more prevalent in...*6th November, 2009  
- Obstet Gynecol. 2009 Oct;114(4):950-3. ([DOI Direct Link](#))
- [Population-based Tay-Sachs screening among Ashkenazi Jewish young adults in the 21st century: Hexosaminidase A enzyme assay is essential for accurate testing.](#)  
*Tay-Sachs disease (TSD) carrier screening, initiated in the 1970s, has...*31st October, 2009  
Genetics Division, Albert Einstein Medical Center, Philadelphia,- Am J Med Genet A. 2009 Nov;149A(11):2444-7. ([DOI Direct Link](#))
- [Assessing the Potential Success of Cystic Fibrosis Carrier Screening: Lessons Learned from Tay-Sachs Disease and beta-Thalassemia.](#)  
*Objective: The objective of this study was to identify factors involved in...*30th October, 2009  
Institute for Public Health Genetics, University of Washington, Seattle,- Public Health Genomics. 2009 Oct 26. ([DOI Direct Link](#))
- ['Cherry red spot' in a patient with Tay-Sachs disease: case report.](#)  
*Tay-Sachs disease is an autosomal recessive disorder of sphingolipid...*13th October, 2009  
Universidade de Regensburg, Alemanha.- Arq Bras Oftalmol. 2009 Jul-Aug;72(4):537-9.
- [Development of genomic DNA reference materials for genetic testing of disorders common in people of ashkenazi jewish descent.](#)  
*Many recessive genetic disorders are found at a higher incidence in people...*10th October, 2009  
Laboratory Practice Evaluation and Genomics Branch, National Center for- J Mol Diagn. 2009 Nov;11(6):530-6. Epub 2009 Oct 8. ([DOI Direct Link](#))
- [Miglustat in late-onset Tay-Sachs disease: a 12-month, randomized, controlled clinical study with 24 months of extended treatment.](#)  
*PURPOSE: To evaluate the safety and efficacy of miglustat in patients with...*26th September, 2009  
Department of Neurology, University Hospitals Case Medical Center,- Genet Med. 2009 Jun;11(6):425-33. ([DOI Direct Link](#))
- [Novel human pathological mutations. Gene symbol: HEXA. Disease: Tay-Sachs disease.](#)  
*Heredoataxias are a group of genetic disorders with a cerebellar syndrome...*9th September, 2009  
Human Genetics,- Hum Genet. 2009 Aug;126(2):329. ([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](#))

- [Ataxias with autosomal, X-chromosomal or maternal inheritance.](#)  
*Heredoataxias are a group of genetic disorders with a cerebellar syndrome...*2nd September, 2009  
Krankenanstalt Rudolfstiftung, Vienna, Austria, Europe.- Can J Neurol Sci. 2009 Jul;36(4):409-28.
- [Re: Neurocognitive testing in late-onset Tay-Sachs disease: a pilot study.](#)  
*We report the case of a girl with Tay-Sachs disease who had convulsions...*17th July, 2009  
- J Inherit Metab Dis. 2009 Apr;32(2):310-1. ([DOI Direct Link](#))
- [Design, synthesis, and biological evaluation of enantiomeric beta-N-acetylhexosaminidase inhibitors LABNAc and DABNAc as potential agents against Tay-Sachs and Sandhoff disease.](#)  
*N-Acetylhexosaminidases are of considerable importance in mammals and are...*20th June, 2009  
Chemistry Research Laboratory, Department of Chemistry, University of- ChemMedChem. 2009 Mar;4(3):378-92. ([DOI Direct Link](#))
- [Diltiazem, a L-type Ca\(2+\) channel blocker, also acts as a pharmacological chaperone in Gaucher patient cells.](#)  
*Recently, inhibition of L-type Ca(2+) channels, using either Diltiazem or...*16th May, 2009  
Genetics & Genome Biology Program, Research Institute, The Hospital for- Mol Genet Metab. 2009 Apr;96(4):225-32. Epub 2009 Jan 22. ([DOI Direct Link](#))
- [Serial MR imaging and 1H-MR spectroscopy in monozygotic twins with Tay-Sachs disease.](#)  
*Four-year-old monozygotic female twins with early onset Tay-Sachs disease...*12th May, 2009  
Department of Pediatrics, Gifu Prefectural General Medical Center, Gifu,- Neuropediatrics. 2008 Oct;39(5):259-63. Epub 2009 Mar 17. ([DOI Direct Link](#))
- [Genetic testing of sperm donors: survey of current practices.](#)  
*OBJECTIVE: To determine which genetic tests are being performed on sperm...*4th April, 2009  
California Cryobank, Inc., Los Angeles, California.- Fertil Steril. 2009 Mar 31. ([DOI Direct Link](#))
- [Unilaterally and rapidly progressing white matter lesion and elevated cytokines in a patient with Tay-Sachs disease.](#)  
*We report the case of a girl with Tay-Sachs disease who had convulsions...*13th March, 2009  
Department of Pediatrics, Jichi Medical University, 3311-1 Yakushiji,- Brain Dev. 2009 Mar 9. ([DOI Direct Link](#))

## Tay-Sachs Disease Patents:



- 7456015- [Tetracycline-regulated adeno-associated viral \(AAV\) vectors for gene delivery to the nervous system](#)
- 7507413- [Albumin fusion proteins](#)
- 7507414- [Albumin fusion proteins](#)
- 7511052- [Pyrimidinyl amide compounds which inhibit leukocyte adhesion mediated by VLA-4](#)
- 7514099- [Lipid nanoparticle based compositions and methods for the delivery of biologically active molecules](#)
- 7514453- [Method for enhancing mutant protein activity](#)
- 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](#)

- 7526114- [Analysis, secure access to, and transmission of array images](#)
- 7528153- [Deoxynojirimycin analogues and their uses as glucosylceramidase inhibitors](#)
- 7534606- [Placental stem cell and methods thereof](#)
- 7501123- [Human G-protein chemokine receptor \(CCR5\) HDGNR10](#)
- 7498171- [Modulation of stem and progenitor cell differentiation, assays, and uses thereof](#)
- 7488721- [Real time methylumbelliferone-based assay](#)
- 7456149- [ALK7 and myostatin inhibitors and uses thereof](#)
- 7459152- [Erythropoietin administration to improve graft survival](#)
- 7459531- [50 Human secreted proteins](#)
- 7465548- [Methods of screening for potential therapeutics for diseases associated with G-protein coupled receptor HM74a](#)
- 7465583- [Duplexed parvovirus vectors](#)
- 7468276- [Placental stem cells](#)
- 7482013- [Albumin fusion proteins](#)
- 7485433- [Determination and quantification of red blood cell populations in samples](#)
- 7485435- [Simultaneous detection of metabolic enzyme activity and metabolite levels](#)
- 7485755- [Acid addition salt of carbasugar amine derivative](#)
- 7541466- [Tetrahydroisoquinoline derivatives for treating protein trafficking diseases](#)
- 7544518- [Rapid quantitative analysis of proteins or protein function in complex mixtures](#)
- 7589065- [Catalysis of the cis/trans-isomerisation of secondary amide peptide compounds](#)
- 7592010- [Albumin fusion proteins](#)
- 7592321- [Methods of treating lysosomal storage related diseases by gene therapy](#)
- 7595318- [Polyethylene glycol conjugates of heterocycloalkyl carboxamido propanoic acids](#)
- 7595874- [Method of condensed cell slide preparation and detection of rarely occurring cells on microscope slides](#)
- 7598055- [N-acetylglucosaminyltransferase III expression in lower eukaryotes](#)
- 7608394- [Methods and compositions for phenotype identification based on nucleic acid methylation](#)
- 7608749- [Monitoring APP cleavage in transgenic rodents comprising an APP Swedish mutation](#)
- 7611839- [Methods for diagnosing RCC and other solid tumors](#)
- 7612040- [Lefty polypeptides and derivatives thereof](#)
- 7582292- [Adipose tissue derived stromal cells for the treatment of neurological disorders](#)
- 7579466- [Pyrimidinyl sulfonamide compounds which inhibit leukocyte adhesion mediated by VLA-4](#)
- 7579166- [Glycoprotein and process for producing the same](#)
- 7547545- [Directed in vitro differentiation of marrow stromal cells into neural cell progenitors](#)
- 7556924- [Detection of nucleic acid sequence differences using coupled ligase detection with padlock probes and polymerase chain reactions](#)
- 7557144- [Modulation of protein expression using carbocyclic aryl alkenoic acid derivatives](#)
- 7560424- [Targeted therapeutic proteins](#)
- 7560431- [Methods of increasing delivery of active agents to brain comprising administering receptor associated protein \(RAP\) fragments conjugated to active agents](#)
- 7560449- [Methods and therapeutic combinations for the treatment of demyelination](#)
- 7563591- [Method for assaying the activity of lysosomal enzymes](#)
- 7569384- [Albumin fusion proteins](#)
- 7569544- [Methods of increasing delivery of active agents to brain comprising administering receptor associated protein \(RAP\) fragments conjugated to active agents](#)
- 7576101- [Composition for and treatment of demyelinating diseases and paralysis by administration of remyelinating agents](#)
- 7612107- [Treatment and inhibition of disease conditions using flexible heteroarotinoids](#)

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