

Fucosidosis

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Fucosidosis is an autosomal recessive metabolic disorder in which fucosidase is not properly used in the cells to break down fucose. There are at least two types of fucosidosis. In type 1, patients have no vascular lesions, but have rapid psychomotor regression, severe and rapidly progressing neurologic signs, elevated sodium and chloride excretion in the sweat, and fatal outcome before the sixth year. In type 2, patients have angiokeratoma, milder psychomotor retardation and neurologic signs, longer survival, and normal salinity in the sweat. ([From the Wikipedia article Fucosidosis](#).)

Recent Publications on Fucosidosis:



- [Severe hypomyelination as the leading neuroradiological sign in a patient with fucosidosis.](#)
Fucosidosis is a rare autosomal recessive lysosomal storage disease... 1st August, 2008
Department of Pediatrics, Städtisches Klinikum Karlsruhe, Karlsruhe,- Neuropediatrics. 2008 Feb;39(1):51-4. ([DOI Direct Link](#))
- [Phenotypic spectrum of fucosidosis in Tunisia.](#)
Fucosidosis (OMIM 230000) is a rare autosomal recessive lysosomal disorder... 25th July, 2008
Paediatric Department, La Rabta Hospital, Jbal Lakhdar, Jebbari, Tunis,- J Inherit Metab Dis. 2008 Jul 27. ([DOI Direct Link](#))
- [Fucosidosis: MRI and MRS findings.](#)
Fucosidosis is a rare, autosomal recessive lysosomal storage disease in... 20th February, 2008
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- [Fucosidosis and anesthesia.](#)
Fucosidosis is a rare, autosomal recessive lysosomal storage disorder... 6th February, 2008
Department of Anesthesia, Children Medical Center, University of Tehran,- *Saudi Med J*. 2007 Sep;28(9):1446-8.
- [\[Neurological presentations of lysosomal diseases in adult patients\]](#)
Lysosomal diseases represent a large group of genetic storage disorders... 17th January, 2008
Federation des maladies du systeme nerveux et centre de reference national- *Rev Neurol (Paris)*. 2007 Oct;163(10):919-29. ([DOI Direct Link](#))
- [Decreased T2 signal in the thalami may be a sign of lysosomal storage disease.](#)
INTRODUCTION: Lysosomal disorders are rare and are caused by genetically... 19th December, 2007
Helsinki Medical Imaging Center, Helsinki University Central Hospital,- *Neuroradiology*. 2007 Jul;49(7):571-8. Epub 2007 Mar 3. ([DOI Direct Link](#))
- [Anesthesia for fucosidosis.](#)
Fucosidosis is an extremely rare, autosomal recessive lysosomal storage... 6th December, 2007
Division of Anesthesia and Pain Medicine, Children's National Medical- *Paediatr Anaesth*. 2007

- Oct;17(10):994-7. ([DOI Direct Link](#))
- [Fucosidosis: a therapeutic challenge.](#)
*Lysosomal diseases represent a large group of genetic storage disorders...*9th November, 2007
- *Pediatr Dermatol.* 2007 Jul-Aug;24(4):442-3. ([DOI Direct Link](#))
 - [Mutation identification and characterization of a Taiwanese patient with fucosidosis.](#)
*Fucosidosis is a rare lysosomal storage disease caused by a defect of the...*21st July, 2007
Division of Genetics and Metabolism, Department of Pediatrics, Mackay- *J Hum Genet.* 2007;52(6):553-6. Epub 2007 Apr 11. ([DOI Direct Link](#))
 - [Profiling oligosaccharidurias by electrospray tandem mass spectrometry: quantifying reducing oligosaccharides.](#)
*A method to semiquantify urinary oligosaccharides from patients suffering...*31st December, 2005
Lysosomal Diseases Research Unit, Department of Genetic Medicine, Women's- *Anal Biochem.* 2005 Oct 1;345(1):30-46. ([DOI Direct Link](#))
 - [Diagnosis of fucosidosis through a skin rash.](#)
*INTRODUCTION: Lysosomal disorders are rare and are caused by genetically...*24th December, 2005
Department of Internal Medicine, Ina Central Hospital, Japan.- *Intern Med.* 2005 Aug;44(8):907-8.
 - [Fucosidosis with angiokeratoma. Immunohistochemical & electronmicroscopic study of a new case and literature review.](#)
*Fucosidosis is a rare lysosomal storage disease due to alpha-L-fucosidase...*28th September, 2005
Department of Dermatology (Pav. R), Ed. Herriot Hospital, Lyon, France.- *J Cutan Pathol.* 2005 Aug;32(7):506-11. ([DOI Direct Link](#))
 - [Allogeneic stem cell transplantation for the treatment of lysosomal and peroxisomal metabolic diseases.](#)
*This is a review of the clinical responses and prospectus of new therapies...*14th June, 2005
University of Minnesota Hospitals and Clinics, 420 Church Street, Box 477,- *Springer Semin Immunopathol.* 2004 Nov;26(1-2):119-32. Epub 2004 Sep 25. ([DOI Direct Link](#))
 - [Parent-child interactions among children with juvenile fibromyalgia, arthritis, and healthy controls.](#)
*Parent-child interactions during pain-inducing exercise tasks among...*19th April, 2005
Psychology, IWK Health Centre and Dalhousie University, Halifax, NS,- *Pain.* 2005 Jan;113(1-2):201-10. ([DOI Direct Link](#))
 - [The expanding spectrum of disorders with elevated plasma chitotriosidase activity: an update.](#)
*A striking elevation of plasma chitotriosidase activity, greater than 150...*25th March, 2005
Department of Enzymology and Cellular Function, Institute of Child Health,- *J Inherit Metab Dis.* 2004;27(5):705-6.

Fucosidosis Patents:



- 7196164- [Secreted protein HHTLF25](#)
- 7368527- [HADDE71 polypeptides](#)
- 7368531- [Human secreted proteins](#)
- 7378077- [High dose radionuclide complexes for bone marrow suppression](#)
- 7378231- [Diagnosis of lysosomal storage disorders using saposins and other markers](#)
- 7381800- [Antibodies to HBIMF63 polypeptide](#)
- 7385042- [Therapeutic and diagnostic compounds, compositions, and methods](#)
- 7390637- [Keratinocyte derived interferon](#)

- 7393934- [Human G-protein chemokine receptor \(CCR5\) HDGNR10](#)
- 7396811- [Subcellular targeting of therapeutic proteins](#)
- 7402308- [Method of delivering genes to the central nervous system of a mammal](#)
- 7361481- [Diagnosis of lysosomal storage disorders using saposins and other markers](#)
- 7358086- [Mammalian two-hybrid system for screening for modulators of the accumulation of metabolic products](#)
- 7351410- [Treatment of Pompe's disease](#)
- 7198909- [Myeloid precursor cell useful for gene therapy and for modulation of immune responses](#)
- 7205139- [Tissue plasminogen activator-like protease](#)
- 7232667- [Keratinocyte growth factor-2 polynucleotides](#)
- 7232670- [Targeting proteins to cells expressing mannose receptors via expression in insect cells](#)
- 7235529- [Brain-associated inhibitor of tissue-type plasminogen activator](#)
- 7238660- [Albumin fusion proteins](#)
- 7238667- [Albumin fusion proteins](#)
- 7241442- [Method for producing proteins suitable for treating lysosomal storage disorders](#)
- 7291461- [Methods for identifying small molecules that modulate premature translation termination and nonsense mRNA decay](#)
- 7341720- [Targeting of glycoprotein therapeutics](#)
- 7408046- [Treatment of bone-associated cancers](#)
- 7411051- [Antibodies to HDPPA04 polypeptide](#)
- 7544518- [Rapid quantitative analysis of proteins or protein function in complex mixtures](#)
- 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](#)
- 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](#)
- 7579166- [Glycoprotein and process for producing the same](#)
- 7592010- [Albumin fusion proteins](#)
- 7592321- [Methods of treating lysosomal storage related diseases by gene therapy](#)
- 7605239- [Skeletal-targeted radiation to treat bone-associated pathologies](#)
- 7521424- [Albumin fusion proteins](#)
- 7514453- [Method for enhancing mutant protein activity](#)
- 7507414- [Albumin fusion proteins](#)
- 7442372- [Delivery of therapeutic compounds to the brain and other tissues](#)
- 7446098- [Combination therapy for treating protein deficiencies](#)
- 7446176- [12 human secreted proteins](#)
- 7459531- [50 Human secreted proteins](#)
- 7482013- [Albumin fusion proteins](#)
- 7485435- [Simultaneous detection of metabolic enzyme activity and metabolite levels](#)
- 7485755- [Acid addition salt of carbasugar amine derivative](#)
- 7488721- [Real time methylumbelliferone-based assay](#)
- 7501123- [Human G-protein chemokine receptor \(CCR5\) HDGNR10](#)
- 7507413- [Albumin fusion proteins](#)
- 7615224- [Multiplex-bead complex for determination of lysosomal storage disorders](#)

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