

# Cystinosis

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Cystinosis is an autosomal recessive genetic disorder of the renal tubules, characterized by the presence of carbohydrates and amino acids in the urine, excessive urination, and low blood levels of potassium and phosphates. ([From the Wikipedia article Cystinosis](#).)



## Recent Publications on Cystinosis:

- [Gastric Acid hypersecretory States: recent insights and advances.](#)  
*Gastric acid hypersecretory states are characterized by basal...*12th November, 2009  
Digestive Diseases Branch, National Institutes of Diabetes and Digestive- Curr Gastroenterol Rep. 2009 Dec;11(6):433-41.
- [Analysis of the CTNS gene in 32 cystinosis patients from Spain.](#)  
*Gastric acid hypersecretory states are characterized by basal...*30th October, 2009  
- Clin Genet. 2009 Nov;76(5):486-9. ([DOI Direct Link](#))
- [Evaluation of treatment with cysteamine eyedrops for cystinosis with confocal microscopy.](#)  
*PURPOSE: The purpose of this study was to evaluate the efficacy of...*30th October, 2009  
Ophthalmology Department, Coimbra University Hospital, Coimbra, Portugal.- Cornea. 2009 Sep;28(8):938-40. ([DOI Direct Link](#))
- [Characterization of CTNS mutations in Arab patients with cystinosis.](#)  
*BACKGROUND: Cystinosis is an autosomal recessive disease characterized by...*27th October, 2009  
Department of Genetics, King Faisal Specialist Hospital and Research- Ophthalmic Genet. 2009 Dec;30(4):185-9.
- [An Indian boy with nephropathic cystinosis: a case report and molecular analysis of CTNS mutation.](#)  
*Cystinosis is a rare autosomal recessive lysosomal storage disorder...*27th October, 2009  
- Genet Test Mol Biomarkers. 2009 Aug;13(4):435-8. ([DOI Direct Link](#))
- [Renal phenotype of the cystinosis mouse model is dependent upon genetic background.](#)  
*BACKGROUND: Cystinosis is caused by mutations in CTNS that encodes...*23rd October, 2009  
1Inserm, U574, Hopital Necker-Enfants Malades, Paris, France.- Nephrol Dial Transplant. 2009 Oct 21. ([DOI Direct Link](#))
- [Successful treatment of the murine model of cystinosis using bone marrow cell transplantation.](#)  
*Cystinosis is an autosomal recessive metabolic disease that belongs to the...*23rd October, 2009  
Department of Molecular and Experimental Medicine, Scripps Research- Blood. 2009 Sep 17;114(12):2542-52. Epub 2009 Jun 8. ([DOI Direct Link](#))
- [Gastrointestinal manifestations of nephropathic cystinosis in children.](#)  
*INTRODUCTION. Cystinosis is an autosomal recessive disorder which is...*21st October, 2009  
Department of Pediatric Gastroenterology, Ali-Asghar Children Hospital,- Iran J Kidney Dis. 2009 Oct;3(4):218-21.

- [Report of a Brazilian Multicenter Study on Nephropathic Cystinosis.](#)  
*Introduction: The Brazilian Multicenter Nephropathic Study Group, founded...*10th October, 2009  
Brazilian Multicenter Cystinosis Study Group, Pediatric Nephrology Unit,- Nephron Clin Pract. 2009 Oct 9;114(1):c12-c18. ([DOI Direct Link](#))
- [Twice-Daily Cysteamine Bitartrate Therapy for Children with Cystinosis.](#)  
*OBJECTIVE: Cystinosis causes renal and other organ failure. Regular...*25th September, 2009  
Department of Pediatrics, University of California San Diego, La Jolla, CA- J Pediatr. 2009 Sep 21. ([DOI Direct Link](#))
- [Current medical aspects of pantethine.](#)  
*Pantethine, the stable disulfide form of pantetheine, is the major...*19th September, 2009  
Department of Neurology, Albert Szent-Gyorgyi Clinical Centre, University- Ideggyogy Sz. 2009 Jul 30;62(7-8):220-9.
- [UK Renal Registry 11th Annual Report \(December 2008\): Chapter 13 Demography of the UK paediatric renal replacement therapy population.](#)  
*AIMS: To describe the demographics of the paediatric RRT population in the...*11th September, 2009  
Manchester Childrens- Nephron Clin Pract. 2009;111 Suppl 1:c257-67. Epub 2009 Mar 26. ([DOI Direct Link](#))
- [Caspase-4 may play a role in loss of proximal tubules and renal injury in nephropathic cystinosis.](#)  
*Nephropathic cystinosis is characterized clinically by generalized...*26th August, 2009  
Department of Pediatrics, Stanford University, G306, 300 Pasteur Drive,- Pediatr Nephrol. 2009 Aug 25. ([DOI Direct Link](#))
- [Cysteamine prevents inhibition of adenylate kinase caused by cystine in rat brain cortex.](#)  
*Cystinosis is a systemic genetic disease caused by a lysosomal transport...*19th August, 2009  
Departamento de Bioquimica, Instituto de Ciencias Basicas da Saude,- Metab Brain Dis. 2009 Sep;24(3):373-81. Epub 2009 Aug 18. ([DOI Direct Link](#))
- [Chitotriosidase plasma activity in nephropathic cystinosis.](#)  
*Chitotriosidase is a fully active chitinase produced and secreted by...*21st May, 2009  
First Department Pediatrics, Athens University, Athens, Greece.- J Inherit Metab Dis. 2009 May 20. ([DOI Direct Link](#))

## BioNews Results for Cystinosis

- [Third Annual 'Fore a Cure' Golf Tournament Raises \\$165,000 to Fund Cystinosis Research](#)  
*RBC Dain Rauscher: Oct 22 2009 6:46AM Matching: cystinosis*
- [Third Annual 'Fore a Cure' Golf Tournament Raises \\$165,000 to Fund Cystinosis Research](#)  
*Genetic Engineering News: Oct 22 2009 12:18AM Matching: cystinosis*

## Cystinosis Patents:



- 6096536- [In vitro cell culture in media containing beta-alanyl-taurine or carbobenzoxy beta-alanyl-taurine](#)
- 6787321- [Mammalian two-hybrid system for screening for modulators of the accumulation of metabolic](#)

## products

- 6797265- Deleted adenovirus vectors and methods of making and administering the same
- 6946126- Replicating adenovirus vectors
- 6977162- Rapid analysis of variations in a genome
- 6989256- Subfamily of RNA helicases which are modulators of the fidelity of translation termination and uses thereof
- 7029666- Uses for non-autologous mesenchymal stem cells
- 7034010- Use of recombinant gene delivery vectors for treating or preventing lysosomal storage disorders
- 7045303- Screening methods for compounds useful in the treatment of polycystic kidney disease
- 7112668- Polypeptides and nucleic acids encoded thereby
- 7122354- Nucleic acid encoding a chimeric polypeptide
- 6762291- Insect p53 tumor suppressor genes and proteins
- 6759189- Early detection of lysosomal storage disorders
- 6730297- Use of recombinant gene delivery vectors for treating or preventing lysosomal storage disorders
- 6153581- Drug for recovering renal function
- 6323025- In vitro cell culture in media containing beta-alanyl-taurine or carbobenzoxy beta-alanyl-taurine
- 6328958- Deleted adenovirus vectors and methods of making and administering the same
- 6355239- Uses for non-autologous mesenchymal stem cells
- 6395884- Therapy for .alpha.-galactosidase a deficiency
- 6486305- METHOD OF MODULATING THE EFFICIENCY OF TRANSLATION TERMINATION AND DEGRADATION OF ABERRANT MRNA INVOLVING A SURVEILLANCE COMPLEX COMPRISING HUMAN UPF1P, EUCARYOTIC RELEASE FACTOR 1 AND EUCARYOTIC RELEASE FACTOR 3
- 6566099- Nucleic acid encoding a chimeric polypeptide
- 6630294- Subfamily of RNA helicases which are modulators of the fidelity of translation termination and uses thereof
- 6638726- Screening methods for compounds useful in the treatment of polycystic kidney disease
- 6693077- Keratinocyte growth factor-2
- 7208274- Rapid analysis of variations in a genome
- 7226791- Polynucleotides encoding novel guanylate binding proteins (GBP's)
- 7442372- Delivery of therapeutic compounds to the brain and other tissues
- 7442506- Methods for detection of genetic disorders
- 7485435- Simultaneous detection of metabolic enzyme activity and metabolite levels
- 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
- 7569544- Methods of increasing delivery of active agents to brain comprising administering receptor associated protein (RAP) fragments conjugated to active agents
- 7579468- Methods of modulating neurotrophin-mediated activity
- 7592321- Methods of treating lysosomal storage related diseases by gene therapy
- 7598060- Rapid analysis of variations in a genome
- 7411051- Antibodies to HDPPA04 polypeptide
- 7396811- Subcellular targeting of therapeutic proteins
- 7378231- Diagnosis of lysosomal storage disorders using saposins and other markers
- 7232667- Keratinocyte growth factor-2 polynucleotides
- 7232670- Targeting proteins to cells expressing mannose receptors via expression in insect cells
- 7241442- Method for producing proteins suitable for treating lysosomal storage disorders
- 7285631- Human cDNAs and proteins and uses thereof
- 7291461- Methods for identifying small molecules that modulate premature translation termination and nonsense mRNA decay

- 7332277- [Methods for detection of genetic disorders](#)
- 7358086- [Mammalian two-hybrid system for screening for modulators of the accumulation of metabolic products](#)
- 7361481- [Diagnosis of lysosomal storage disorders using saposins and other markers](#)
- 7368527- [HADDE71 polypeptides](#)
- 7368531- [Human secreted proteins](#)
- 7615224- [Multiplex-bead complex for determination of lysosomal storage disorders](#)

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