

Citrullinemia

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Citrullinemia, also called citrullinuria, is an autosomal recessive urea cycle disorder that causes ammonia and other toxic substances to accumulate in the blood. Two forms of citrullinemia have been described, both having different signs and symptoms, and are caused by mutations in different genes. Citrullinemia belongs to a class of genetic diseases called urea cycle disorders. The urea cycle is a sequence of chemical reactions that takes place in the liver. These reactions process excess nitrogen, generated when protein is used by the body, to make a compound called urea that is excreted by the kidneys. ([From the Wikipedia article Citrullinemia](#).)



Recent Publications on Citrullinemia:

- [Use of peritoneal dialysis and mesothelium in non primary renal conditions.](#)
*The management of a handful of non renal clinical conditions includes...*6th November, 2009
Laboratory of Experimental Nephrology, Ha'Emek Medical Center, Afula,- Adv Perit Dial. 2009;25:2-5.
- [Cranial MRI in acute hyperammonemic encephalopathy.](#)
*Cranial magnetic resonance imaging was performed in three cases of acute...*22nd September, 2009
Department of Neurology, National Institute of Mental Health and- Pediatr Neurol. 2009
Aug;41(2):139-42. ([DOI Direct Link](#))
- [The human and mouse SLC25A29 mitochondrial transporters rescue the deficient ornithine metabolism in fibroblasts of patients with the hyperornithinemia-hyperammonemia-homocitrullinuria \(HHH\) syndrome.](#)
*The hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome is a...*10th September, 2009
Department of Pediatrics, University of California-Irvine, Irvine, CA- Pediatr Res. 2009 Jul;66(1):35-41.
([DOI Direct Link](#))
- [Novel human pathological mutations. Gene symbol: ASS1. Disease: Citrullinaemia.](#)
*The management of a handful of non renal clinical conditions includes...*9th September, 2009
Pediatrics, Medical College of Wisconsin, Genetics Lab: HRC PD169,- Hum Genet. 2009
Aug;126(2):342.
- [Novel human pathological mutations. Gene symbol: ASS1. Disease: Citrullinaemia.](#)
*The management of a handful of non renal clinical conditions includes...*9th September, 2009
Pediatrics, Medical College of Wisconsin, Genetics Lab: HRC PD169,- Hum Genet. 2009
Aug;126(2):341.
- [\[A 69-year-old man developing consciousness disorder\]](#)
*The management of a handful of non renal clinical conditions includes...*4th September, 2009
- Nippon Shokakibyo Gakkai Zasshi. 2009 Jun;106(6):840, 843.
- [Citrullinemia type I, classical variant. Identification of ASS-p~G390R \(c.1168G>A\) mutation in families of](#)

[a limited geographic area of Argentina: a possible population cluster.](#)

OBJECTIVE: Citrullinemia type I (CTLN1) is an urea cycle defect caused by... 29th August, 2009
Centro de Estudio de las Metabopatias Congenitas, Clinica Pediatrica,- Clin Biochem. 2009
Jul;42(10-11):1166-8. Epub 2009 Apr 7. ([DOI Direct Link](#))

- [Hypergalactosemia in early infancy: diagnostic strategy with an emphasis on imaging.](#)
BACKGROUND: Portosystemic shunt is one of the main causes of persistent... 26th August, 2009
Department of Radiology, Dokkyo Medical University, Shimotsuga, Tochigi,- *Pediatr Int.* 2009
Apr;51(2):276-82. ([DOI Direct Link](#))
- [A case of adult onset type II citrullinemia with portal-systemic shunt.](#)
A 48-year-old woman who had conscious disturbance and abnormal behaviors... 13th August, 2009
Department of Neurology and Neurobiology of Aging, Kanazawa University- *J Neurol Sci.* 2009 Jun
15;281(1-2):127-9. Epub 2009 Mar 13. ([DOI Direct Link](#))
- [\[Failure to thrive and dyslipidemia caused by citrin deficiency: a novel clinical phenotype\]](#)
Two clinical phenotypes for citrin deficiency (CD) have been reported. One... 12th August, 2009
Department of Pediatrics, First Affiliated Hospital, Jinan University,- *Zhongguo Dang Dai Er Ke Za Zhi.*
2009 May;11(5):328-32.
- [Inherited diseases of Australian Holstein-Friesian cattle.](#)
Inherited disorders are of major importance in Holstein-Friesian cattle, a... 17th July, 2009
Faculty of Veterinary Science, University of Sydney, PMB 3, Camden, New- *Aust Vet J.* 2009
May;87(5):193-9. ([DOI Direct Link](#))
- [Possible mechanism for zonisamide-induced hyperammonemia in a child with citrullinemia type 1.](#)
Cranial magnetic resonance imaging was performed in three cases of acute... 9th July, 2009
- *Indian J Med Sci.* 2009 May;63(5):203-6. ([DOI Direct Link](#))
- [Sustaining hypercitrullinemia, hypercholesterolemia and augmented oxidative stress in Japanese children with aspartate/glutamate carrier isoform 2-citrin-deficiency even during the silent period.](#)
Neonatal intrahepatic cholestasis caused by citrin deficiency (NICCD)... 28th May, 2009
Division of Metabolism, Chiba Children's Hospital, 579-1 Heta Cho,- *Mol Genet Metab.* 2009
May;97(1):21-6. Epub 2009 Jan 25. ([DOI Direct Link](#))
- [Liver transplantation for an infant with neonatal intrahepatic cholestasis caused by citrin deficiency using heterozygote living donor.](#)
NICCD is an autosomal recessive genetic disorder, characterized by... 6th May, 2009
Department of Transplant Surgery, National Center for Child Health and- *Pediatr Transplant.* 2009 Apr
3. ([DOI Direct Link](#))
- [Living donor liver transplantation for type II citrullinemia from a heterozygous donor.](#)
Adult-onset type II citrullinemia (CTLN2) is a rare disorder of the urea... 25th April, 2009
Department of Gastroenterological and General Surgery, Yamagata University-
Hepatogastroenterology. 2008 Nov-Dec;55(88):2211-6.

Citrullinemia Clinical Trials:



- [Citrulline Allo. Evaluation of Citrullinemia as a Marker of Bowel Damage After Allogeneic Bone Marrow Transplantation in Children](#)



Citrullinemia Patents:

- 4100160- [Therapeutic compositions comprising alpha-hydroxy analogs of essential amino acids and their administration to humans for promotion of protein synthesis and suppression of urea formation](#)
- 6190887- [Expression of an exogenous gene in a mammalian cell by use of a non-mammalian DNA virus having an altered coat protein](#)
- 6238914- [Use of a non-mammalian DNA virus to express an exogenous gene in a mammalian cell](#)
- 6281009- [Use of a non-mammalian DNA virus to express an exogenous gene in a mammalian cell](#)
- 6338953- [Expression of an exogenous gene in a mammalian cell by use of a non-mammalian DNA virus having an altered coat protein](#)
- 6338962- [Use of a non-mammalian DNA virus to express an exogenous gene in a mammalian cell](#)
- 6455321- [Method for interpreting tandem mass spectrometry data for clinical diagnosis](#)
- 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](#)
- 6498027- [Targeted delivery through a cationic amino acid transporter](#)
- 6503530- [Method of preventing development of severe metabolic derangement in inborn errors of metabolism](#)
- 6586192- [Compositions and methods for use in affecting hematopoietic stem cell populations in mammals](#)
- 6183993- [Complement-resistant non-mammalian DNA viruses and uses thereof](#)
- 6093699- [Method for gene therapy involving suppression of an immune response](#)
- 6093392- [Methods and compositions for use in gene therapy for treatment of hemophilia](#)
- 4100293- [Treatment of hepatic disorders with therapeutic compositions comprising keto analogs of essential amino acids](#)
- 4228099- [Ornithine and arginine salts of branched chain keto acids and uses in treatment of hepatic and renal disorders](#)
- 4320146- [Treatment of hepatic and renal disorders with ornithine and arginine salts of branched chain keto acids](#)
- 5175082- [Method of characterizing genomic DNA](#)
- 5219752- [Process for continuously culturing adherent animal cells](#)
- 5234810- [Diagnostic assays for genetic mutations associated with bovine leukocyte adhesion deficiency](#)
- 5656425- [Screening for mutations by expressing cDNA segments](#)
- 5731182- [Non-mammalian DNA virus to express an exogenous gene in a mammalian cell](#)
- 5766899- [Targeted nucleic acid delivery into liver cells](#)
- 5871986- [Use of a baculovirus to express and exogenous gene in a mammalian cell](#)
- 6610474- [Cells for detection of influenza and parainfluenza viruses](#)
- 6630294- [Subfamily of RNA helicases which are modulators of the fidelity of translation termination and uses thereof](#)
- 7244621- [Method for interpreting tandem mass spectrometry data for clinical diagnosis of genetic disorders such as hypermethionemia](#)
- 7259151- [AAV virions with decreased immunoreactivity and uses therefor](#)
- 7291461- [Methods for identifying small molecules that modulate premature translation termination and nonsense mRNA decay](#)
- 7368527- [HADDE71 polypeptides](#)

- 7368531- [Human secreted proteins](#)
- 7411051- [Antibodies to HDPPA04 polypeptide](#)
- 7427396- [AAV vectors for gene delivery to the lung](#)
- 7433520- [Nosologic system of diagnosis](#)
- 7485435- [Simultaneous detection of metabolic enzyme activity and metabolite levels](#)
- 7566533- [Clinically intelligent diagnostic devices and methods](#)
- 7238531- [Method for interpreting tandem mass spectrometry data for clinical diagnosis](#)
- 7229834- [Method for interpreting tandem mass spectrometry data for clinical diagnosis](#)
- 7223605- [Method for interpreting tandem mass spectrometry data for clinical diagnosis of genetic disorders such as citrullinemia](#)
- 6821741- [Cells for detection of enteroviruses](#)
- 6905816- [Clinically intelligent diagnostic devices and methods](#)
- 6989256- [Subfamily of RNA helicases which are modulators of the fidelity of translation termination and uses thereof](#)
- 6991899- [Cells for detection of influenza and parainfluenza viruses](#)
- 7015037- [Multipotent adult stem cells and methods for isolation](#)
- 7091315- [Protein HDPBQ71](#)
- 7129042- [Compositions and methods for detecting severe acute respiratory syndrome coronavirus](#)
- 7150965- [Cells for detection of influenza and parainfluenza viruses](#)
- 7186504- [Cells for detection of enteroviruses](#)
- 7192933- [Use of a baculovirus to express an exogenous gene in a mammalian cell](#)
- 7613343- [Nosologic system of diagnosis](#)

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