

Alkaptonuria

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Alkaptonuria is a rare inherited genetic disorder of tyrosine metabolism. This is an autosomal recessive condition that is due to a defect in the enzyme homogentisate 1,2-dioxygenase, which participates in the degradation of tyrosine. As a result, a toxic tyrosine byproduct called homogentisic acid accumulates in the blood, and is excreted in urine in large amounts. Excessive homogentisic acid causes damage to cartilage and heart valves as well as precipitating as kidney stones. Treatment with nitisinone, which suppresses homogentisic acid production, is being studied. Alkaptonuria is more common in Slovakia and the Dominican Republic than in other countries. ([From the Wikipedia article Alkaptonuria](#).)



Recent Publications on Alkaptonuria:

- [Mutation spectrum of homogentisic acid oxidase \(HGD\) in alkaptonuria.](#)
*Alkaptonuria (AKU) is a rare autosomal recessive metabolic disorder,...*29th October, 2009
Medical Genetics Branch, National Human Genome Research Institute,- Hum Mutat. 2009 Sep 4. ([DOI Direct Link](#))
- [Ochronosis: complicated tear of black meniscus.](#)
*We describe a case of a previously healthy 35-year-old man who presents...*26th September, 2009
Ankara Numune Training and Research Hospital, Talatpasa Blv, Samanpazari,- Knee Surg Sports Traumatol Arthrosc. 2009 Sep 25. ([DOI Direct Link](#))
- [A metabolic cause of spinal deformity.](#)
*A 38-year-old man presented to our clinic with a 6-year history of chronic...*22nd September, 2009
Department of Rheumatology, Basel University, Burgfelderstr. 55, CH 4012- Metabolism. 2009 Sep 16. ([DOI Direct Link](#))
- [\[Intrahepatic gallstones in patient with alkaptonuria\]](#)
*Alkaptonuria is a rare inherited disease with enzyme deficiency in the...*25th August, 2009
Herlev Hospital, Kirurgisk afdeling D.- Ugeskr Laeger. 2009 Jun 22;171(26):2198-9.
- [\[Ochronosis--a rare cause of secondary gonarthrosis\]](#)
*Ochronosis is a manifestation of the rare disease alkaptonuria. The most...*22nd August, 2009
Orthopadische Klinik fur die Universitat Regensburg, Bad Abbach.- Z Orthop Unfall. 2009 May-Jun;147(3):366-8. Epub 2009 Jun 23. ([DOI Direct Link](#))
- [Devastating ochronotic arthropathy with successful bilateral hip and knee arthroplasties.](#)
*Ochronotic arthropathy is a manifestation of long-standing alkaptonuria, a...*8th July, 2009
Department of Orthopaedic Surgery, Mie University Graduate School of- J Clin Rheumatol. 2009 Apr;15(3):138-40. ([DOI Direct Link](#))
- [A novel missense HGD gene mutation, K57N, in a patient with alkaptonuria.](#)
*Alkaptonuria is a rare recessive disorder of phenylalanine/tyrosine...*7th July, 2009
Department of Core Clinical Pathology & Biochemistry, PathWest Laboratory- Clin Chim Acta. 2009

- May;403(1-2):254-6. Epub 2009 Mar 21. ([DOI Direct Link](#))
- [Black bones: a case of incidental discovery of ochronotic arthropathy.](#)
*Alkaptonuria is a rare disease in which the body does not have enough of...*28th May, 2009
Goulburn Valley Health, Shepparton, VIC, Australia.- Med J Aust. 2009 Apr 6;190(7):390.
 - [\[Knee ochronotic arthropathy and arthroscopic findings\]](#)
*Ochronotic arthropathy is a rare condition found in patients with...*27th May, 2009
Haseki Egitim ve Arastirma Hastanesi, Ortopedi ve Travmatoloji Klinigi,- Acta Orthop Traumatol Turc. 2009 Jan-Feb;43(1):67-71. ([DOI Direct Link](#))
 - [Osteoarthritis? Ochronotic arthritis! A case study and review of the literature.](#)
*Alkaptonuria is a rare disease in which the body does not have enough of...*22nd April, 2009
Department of Joint Surgery, Third Hospital of Hebei Medical University,- Knee Surg Sports Traumatol Arthrosc. 2009 Jul;17(7):778-81. Epub 2009 Apr ([DOI Direct Link](#))
 - [Acute anterior uveitis as the initial presentation of alkaptonuria.](#)
*Alkaptonuria is a rare autosomal recessive metabolic disorder that may...*22nd April, 2009
Department of Ophthalmology, Christian Medical College, Vellore, India.- J Postgrad Med. 2009 Jan-Mar;55(1):35-7.
 - [A case of ochronosis with gout and monckeberg arteries.](#)
*Alkaptonuria is an inborn error of amino acid metabolism. A defect in...*28th February, 2009
Rheumatology Department, Isfahan University for Medical Sciences, Alzahra- Rheumatol Int. 2009 Oct;29(12):1507-10. Epub 2009 Feb 27. ([DOI Direct Link](#))
 - [Black aortic valve--ochronosis.](#)
*Alkaptonuria is a rare disorder of metabolism characterized by deficiency...*20th February, 2009
- APMIS. 2008 Nov;116(11):1011-2. ([DOI Direct Link](#))
 - [From darkening urine to early diagnosis of alkaptonuria.](#)
*Alkaptonuria is a rare disorder of metabolism characterized by deficiency...*31st January, 2009
Research Hospital of the Mustafa Kemal University, Hatay, Turkey.- Indian J Dermatol Venereol Leprol. 2008 Nov-Dec;74(6):700.
 - [Three-generational alkaptonuria in a non-consanguineous family.](#)
*OBJECTIVE: Alkaptonuria (AKU) is a rare inborn error of metabolism of...*20th December, 2008
Institute of Human Genetics, Technical University Munich, Trogerstr. 32,- J Inherit Metab Dis. 2008 Dec 22. ([DOI Direct Link](#))

Alkaptonuria Clinical Trials:

- [Study of Alkaptonuria](#)



Alkaptonuria Patents:



- 4889249- [Urine bottle with cap](#)
- 6277974- [Compositions and methods for diagnosing and treating conditions, disorders, or diseases involving cell death](#)
- 6309823- [Arrays of nucleic acid probes for analyzing biotransformation genes and methods of using the same](#)
- 6399383- [Human papilloma virus vectors](#)
- 6468744- [Analysis of genetic polymorphisms and gene copy number](#)
- 6541035- [Nanosheres comprising a biocompatible polysaccharide](#)
- 6562619- [Differentiation of human embryonic germ cells](#)
- 6586410- [Lipid-nucleic acid particles prepared via a hydrophobic lipid-nucleic acid complex intermediate and use for gene transfer](#)
- 6649366- [Methods and compositions related to modulators of annexin and cartilage homeostasis](#)
- 6670194- [Rapid quantitative analysis of proteins or protein function in complex mixtures](#)
- 6689747- [Use of insulin for the treatment of cartilagenous disorders](#)
- 6245566- [Human embryonic germ cell line and methods of use](#)
- 6218169- [Aromatic amino acid catabolism enzymes](#)
- 6214384- [Nanosheres comprising a biocompatible polysaccharide](#)
- 5220302- [NMR clinical chemistry analyzer and method of forming a shield](#)
- 5240415- [Dental bleach system having separately compartmented fumed silica and hydrogen peroxide and method of using](#)
- 5854207- [Compositions and therapeutic methods using morphogenic proteins and stimulatory factors](#)
- 5916870- [Compositions and therapeutic methods using morphogenic proteins and stimulatory factors](#)
- 5948428- [Compositions and therapeutic methods using morphogenic proteins and stimulatory factors](#)
- 5976567- [Lipid-nucleic acid particles prepared via a hydrophobic lipid-nucleic acid complex intermediate and use for gene transfer](#)
- 6027880- [Arrays of nucleic acid probes and methods of using the same for detecting cystic fibrosis](#)
- 6048964- [Compositions and therapeutic methods using morphogenic proteins and stimulatory factors](#)
- 6086913- [Liposomal delivery of AAV vectors](#)
- 6156501- [Arrays of modified nucleic acid probes and methods of use](#)
- 6693077- [Keratinocyte growth factor-2](#)
- 6696410- [Compositions and therapeutic methods using morphogenic proteins, hormones and hormone receptors](#)
- 7291461- [Methods for identifying small molecules that modulate premature translation termination and nonsense mRNA decay](#)
- 7359748- [Apparatus for total immersion photography](#)
- 7368527- [HADDE71 polypeptides](#)
- 7368531- [Human secreted proteins](#)
- 7375198- [Modified nucleic acid probes](#)
- 7399584- [Method of comparing a target nucleic acid and a reference nucleic acid](#)
- 7411051- [Antibodies to HDPPA04 polypeptide](#)
- 7422902- [Lipid-nucleic acid particles prepared via a hydrophobic lipid-nucleic acid complex intermediate and use for gene transfer](#)
- 7423017- [Method for treating cartilage disorders](#)
- 7544518- [Rapid quantitative analysis of proteins or protein function in complex mixtures](#)
- 7268112- [Use of insulin for the treatment of cartilaginous disorders](#)
- 7232667- [Keratinocyte growth factor-2 polynucleotides](#)
- 7189705- [Methods of enhancing SPLP-mediated transfection using endosomal membrane destabilizers](#)
- 6852544- [Rapid quantitative analysis of proteins or protein function in complex mixtures](#)
- 6858224- [Method of preventing aggregation of a lipid:nucleic acid complex](#)
- 6949692- [Method for identifying mutants and molecules](#)
- 7018795- [Hybridization probe and target nucleic acid detecting kit, target nucleic acid detecting apparatus and target nucleic acid detecting method using the same](#)
- 7026292- [Compositions and therapeutic methods using morphogenic proteins and stimulatory factors](#)

- 7033781- [Whole cell engineering by mutagenizing a substantial portion of a starting genome, combining mutations, and optionally repeating](#)
- 7087745- [Three-dimensional structures containing hyaluronic acid derivatives obtained by the supercritical antisolvent technique](#)
- 7101875- [Methods for treating arthritic disorders](#)
- 7115364- [Arrays of nucleic acid probes on biological chips](#)
- 7186698- [Spatial and temporal control of gene expression using a heat shock protein promoter in combination with local heat](#)
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