

# Achondroplasia

[View the current Achondroplasia InDepth page on BioPortfolio.com](#) (PDF)

(<http://www.bioportfolio.com/indepth/Achondroplasia.html>) - Regularly Updated.

Achondroplasia is a type of autosomal dominant genetic disorder that is a common cause of dwarfism. Achondroplastic dwarfs have short stature, with an average adult height of 131 cm for males and 123 cm for females. The prevalence is approximately 1 in 25,000. ([From the Wikipedia article Achondroplasia](#).)

## Recent Publications on Achondroplasia:



- [New insight on FGFR3-related chondrodysplasias molecular physiopathology revealed by human chondrocyte gene expression profiling.](#)  
*Endochondral ossification is the process by which the appendicular...* 10th November, 2009  
Unite U781, Institut National de la Sante et de la Recherche Medicale,- PLoS One. 2009 Oct 29;4(10):e7633. ([DOI Direct Link](#))
- [Sleep-related respiratory abnormalities and arousal pattern in achondroplasia during early infancy.](#)  
*OBJECTIVE: To assess sleep-disordered breathing (SDB), sleep architecture,...* 23rd October, 2009  
Department of Pulmonary Medicine, Cincinnati Children's Hospital Medical- J Pediatr. 2009 Oct;155(4):510-5. Epub 2009 Jul 16. ([DOI Direct Link](#))
- [Neurosurgical implications of achondroplasia.](#)  
*OBJECT: Achondroplasia is the most common form of human short-limbed...* 21st October, 2009  
Division of Neurosurgery, The Hospital for Sick Children, University of- J Neurosurg Pediatr. 2009 Oct;4(4):297-306. ([DOI Direct Link](#))
- [Editorial. Achondroplasia.](#)  
*OBJECT: Achondroplasia is the most common form of human short-limbed...* 21st October, 2009  
- J Neurosurg Pediatr. 2009 Oct;4(4):295-6; discussion 296. ([DOI Direct Link](#))
- [Acanthosis nigricans and insulin sensitivity in patients with achondroplasia and hypochondroplasia due to FGFR3 mutations.](#)  
*BACKGROUND AND AIMS: Acanthosis nigricans (AN) has been reported in...* 21st October, 2009  
Developmental Endocrinology Research Group, University College London,- J Clin Endocrinol Metab. 2009 Oct;94(10):3959-63. Epub 2009 Jul 21. ([DOI Direct Link](#))
- [Molecular studies of achondroplasia.](#)  
*BACKGROUND: Achondroplasia (ACH) is the most frequent form of short-limbed...* 20th October, 2009  
Department of Genetic Medicine, Sir Ganga Ram Hospital, New Delhi - 110- Indian J Orthop. 2009 Apr;43(2):194-6. ([DOI Direct Link](#))
- [Limb lengthening in short-stature patients using monolateral and circular external fixators.](#)  
*OBJECTIVES: To review the results of distraction osteogenesis in...* 9th October, 2009  
Department of Orthopaedics and Traumatology, Duchess of Kent Children's- Hong Kong Med J. 2009 Aug;15(4):280-4.
- [Chronically elevated plasma C-type natriuretic peptide \(CNP\) level stimulates skeletal growth in](#)

[transgenic mice.](#)

*C-type natriuretic peptide (CNP) plays a critical role in endochondral...* 8th October, 2009

1Chugai Pharmaceutical Co., Ltd.- Am J Physiol Endocrinol Metab. 2009 Oct 6. ([DOI Direct Link](#))

- [Increased p21 expression in chondrocytes of achondroplastic children independently from the presence of the G380R FGFR3 mutation.](#)  
*BACKGROUND: Achondroplasia (ACH) represents the major cause of dwarfism...* 6th October, 2009  
Pathology Department, Orthopaedic Institute Gaetano Pini, Milan, Italy.- J Orthop Sci. 2009 Sep;14(5):623-30. Epub 2009 Oct 3. ([DOI Direct Link](#))
- [Prenatal diagnosis of skeletal dysplasia due to FGFR3 gene mutations: a 9-year experience : Prenatal diagnosis in FGFR3 gene.](#)  
*PURPOSE: Prenatal diagnosis with ultrasound findings compatible with...* 1st October, 2009  
Department of Genetics, Fundacion Jimenez Diaz (CIBERER), Madrid, Spain.- J Assist Reprod Genet. 2009 Sep 30. ([DOI Direct Link](#))
- [Lumbar fusion in achondroplasia: does fusion to the sacrum affect function?](#)  
*BACKGROUND: Stenosis and kyphosis are common in achondroplasia, often...* 24th September, 2009  
Division of Pediatric Orthopaedic Surgery, The Johns Hopkins University,- J Pediatr Orthop. 2009 Jul-Aug;29(5):476-80. ([DOI Direct Link](#))
- [Factors related to progression of thoracolumbar kyphosis in children with achondroplasia: a retrospective cohort study of forty-eight children treated in a comprehensive orthopaedic center.](#)  
*STUDY DESIGN: Retrospective cohort. OBJECTIVE: To assess the period...* 23rd September, 2009  
Department of Orthopaedics, Alfred I duPont Hospital for Children,- Spine (Phila Pa 1976). 2009 Jul 15;34(16):1699-705. ([DOI Direct Link](#))
- [Genetic mouse models for the functional analysis of the perifibrillar components collagen IX, COMP and matrilin-3: Implications for growth cartilage differentiation and endochondral ossification.](#)  
*The mutual interaction of the two supramolecular compartments, the...* 26th August, 2009  
Centre for Biochemistry, Medical Faculty, University of Cologne, Germany.- Histol Histopathol. 2009 Aug;24(8):1067-79.
- [Limb lengthening and deformity correction in children using hexapodal external fixation: Preliminary results for 36 cases.](#)  
*Limb deformities in children can be corrected using different techniques,...* 12th August, 2009  
Service de chirurgie infantile et orthopedique, hopital Timone-Enfants,- Orthop Traumatol Surg Res. 2009 Oct;95(6):425-30. Epub 2009 Aug 8. ([DOI Direct Link](#))
- [Callus features of regenerate fracture cases in femoral lengthening in achondroplasia.](#)  
*OBJECTIVE: We studied the callus features seen in cases of regenerate...* 3rd July, 2009  
Institute for Rare Diseases and Department of Orthopedic Surgery, Korea- Skeletal Radiol. 2009 Jul 2. ([DOI Direct Link](#))

## BioNews Results for Achondroplasia

- [Why older fathers are more likely to have children with genetic disorders](#)  
*Mail Online UK: Oct 26 2009 12:14PM Matching: achondroplasia*
- [Link found between old dads and genetic diseases](#)  
*Times Online: Oct 25 2009 11:45PM Matching: achondroplasia*
- [Scientists discover link between older dads and genetic diseases](#)  
*Times Online: Oct 25 2009 11:33PM Matching: achondroplasia*
- [Testicular tumours linked to offsprings' disease](#)  
*New Scientist: Oct 25 2009 6:07PM Matching: achondroplasia*

## Achondroplasia Clinical Trials:



- [Issues Surrounding Prenatal Genetic Testing for Achondroplasia](#)

## Achondroplasia Patents:



- 7288392- [Human protein kinases and uses therefor](#)
- 7385034- [Complementary DNAs encoding proteins with signal peptides](#)
- 7385036- [Human tango 509 polypeptides](#)
- 7402434- [Splice choice antagonists as therapeutic agents](#)
- 7410777- [Non-endogenous, constitutively activated human G protein-coupled receptors](#)
- 7411051- [Antibodies to HDPPA04 polypeptide](#)
- 7414109- [FIAT nucleic acids and proteins and uses thereof](#)
- 7425531- [Bone polypeptide-1](#)
- 7442506- [Methods for detection of genetic disorders](#)
- 7442708- [1,2,3-substituted indolizine derivatives, inhibitors of fgfs, method for making same and pharmaceutical compositions containing same](#)
- 7470665- [Methods for targeting cells that express fibroblast growth receptor-3 or -2](#)
- 7381409- [Antibodies specific for sclerostin and methods of screening and use therefor](#)
- 7368531- [Human secreted proteins](#)
- 7368527- [HADDE71 polypeptides](#)
- 7288406- [Active variants of FGF with improved specificity](#)
- 7291461- [Methods for identifying small molecules that modulate premature translation termination and nonsense mRNA decay](#)
- 7291483- [FGF-CX polynucleotide sequences and methods of producing same](#)
- 7297492- [LMNA gene and its involvement in Hutchinson-Gilford Progeria Syndrome \(HGPS\) and arteriosclerosis](#)
- 7297493- [Fibroblast growth factor receptor activating gene 1 and related compositions and methods](#)
- 7297774- [Use of inhibitors for the treatment of RTK-hyperfunction-induced disorders, particularly cancer](#)
- 7301016- [Human transferase family members and uses thereof](#)
- 7329529- [Ubiquitin proteases](#)
- 7332277- [Methods for detection of genetic disorders](#)
- 7348142- [Cancer diagnostic panel](#)
- 7470668- [Method of use of specific natriuretic peptide receptor c ligands, transgenic non-human mammals expressing specific natriuretic peptide receptor c antagonists and cells thereof](#)
- 7479367- [In vivo assay and molecular markers for testing the phenotypic stability of cell populations and selected cell populations for autologous transplantation](#)
- 7553941- [Long-acting polypeptides and methods of producing same](#)
- 7563769- [FGF variants and methods for use thereof](#)

- 7572899- [Compositions and methods for increasing bone mineralization](#)
- 7578999- [Antibodies specific for sclerostin](#)
- 7592429- [Sclerostin-binding antibody](#)
- 7593820- [Crystal structure of human Janus Kinase 2 \(JAK2\) and uses thereof](#)
- 7598027- [Botulinum toxin screening assays](#)
- 7598060- [Rapid analysis of variations in a genome](#)
- 7598272- [Kinase inhibitors](#)
- 7601501- [Controlling osteogenesis by inhibition of osteogenic suppressors](#)
- 7553940- [Long-acting EPO polypeptides and derivatives thereof and methods thereof](#)
- 7553845- [Substituted indolizine 1,2,3,6,7,8 derivatives, FGFs inhibitors, a method for the preparation thereof and pharmaceutical compositions containing said derivatives](#)
- 7550432- [Recombinant fusion proteins to growth hormone and serum albumin](#)
- 7482114- [In vivo assay and molecular markers for testing the phenotypic stability of cell populations, and selected cell populations for autologous transplantation](#)
- 7482147- [Ubiquitin protease](#)
- 7482367- [Substituted benzimidazoles and methods of their use](#)
- 7488710- [SFRP and peptide motifs that interact with SFRP and methods of their use](#)
- 7498416- [Antibodies that block receptor protein tyrosine kinase activation, methods of screening for and uses thereof](#)
- 7510881- [Method and kit for the quantitative and/or qualitative detection of components in a sample](#)
- 7511047- [Kinase inhibitors](#)
- 7517890- [GTPase inhibitors and methods of use](#)
- 7524813- [Selectively conjugated peptides and methods of making the same](#)
- 7550300- [Prediction of bare metal stent restenosis](#)
- 7615349- [Melanoma gene signature](#)

\*\*\*

Resources from the [NCBI](#) used in this document, [NCBI's standard disclaimer applies](#).

Nothing in this document should be used in place of personal medical advice from your own qualified medical practitioner. See BioPortfolio.com [User Agreement](#)

Send comments and feedback to:

Peter Barfoot Managing Director, BioPortfolio Ltd.

UK Tel: (+44) 1300 321501

USA Voicemail and Fax: (+1) 415 680 2472

[Peter Barfoot peter.barfoot@bioportfolio.com](mailto:Peter.Barfoot@bioportfolio.com)

All rights reserved. All other trademarks recognized.

BioPortfolio Limited is registered in England & Wales at Stafford House, 10 Prince of Wales Road, Dorchester, Dorset, DT1 1PW, UK. No.3312883 VAT No. GB 744 6483 10

Copyright 1997-2009 - BioPortfolio Limited.

