

Achondroplasia

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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.



Recent Publications on Achondroplasia:

- [Skeletal dysplasia in ancient Egypt.](#)
The ancient Egyptian civilization lasted for over 3000 years and ended in... 14th November, 2008
Department of Pediatrics, Georgetown University Hospital, Washington,- *Am J Med Genet A.* 2008 Nov 12. ([DOI Direct Link](#))
- [The time of onset of abnormal calcification in spondylometaphyseal dysplasia, short limb-abnormal calcification type.](#)
A 1-month-old boy with shortness of extremities on prenatal US was... 13th November, 2008
Department of Pediatric Genetics, Cerrahpasa Medical School, Istanbul- *Pediatr Radiol.* 2008 Nov 11. ([DOI Direct Link](#))
- [Skeletal dysplasias: 38 prenatal cases.](#)
OBJECTIVE: To assess the prenatal diagnosis of skeletal dysplasias in a... 11th November, 2008
Center for Human Genetics, University of Leuven, Belgium.- *Genet Couns.* 2008;19(3):267-75.
- [Hypothesis for lateral ventricular dilatation in communicating hydrocephalus: New understanding of the Monro-Kellie hypothesis in the aspect of cardiac energy transfer through arterial blood flow.](#)
Many theories have been postulated to date regarding mechanisms involved... 4th November, 2008
Department of Neurosurgery, Ajou University School of Medicine, San 5,- *Med Hypotheses.* 2008 Oct 29. ([DOI Direct Link](#))
- [\[Causes and clinical implications of sperm DNA damages.\]](#)
Numerous recent studies involve DNA damages associated with poor... 31st October, 2008
Service de gynecologie-obstetrique, maternite Aline-de-Crepy, hopital- *Gynecol Obstet Fertil.* 2008 Oct 27. ([DOI Direct Link](#))
- [FGFR3 promotes synchondrosis closure and fusion of ossification centers through the MAPK pathway.](#)
Activating mutations in FGFR3 cause achondroplasia and thanatophoric... 17th October, 2008
Department of Orthopaedics, Case Western Reserve University, 10900 Euclid- *Hum Mol Genet.* 2008 Oct 15. ([DOI Direct Link](#))
- [Nordihydroguaiaretic acid inhibits an activated fibroblast growth factor receptor 3 mutant and blocks downstream signaling in multiple myeloma cells.](#)
Activating mutations within fibroblast growth factor receptor 3 (FGFR3), a... 15th October, 2008
Department of Chemistry and Biochemistry, Moores UCSD Cancer Center,- *Cancer Res.* 2008 Sep 15;68(18):7362-70. ([DOI Direct Link](#))

- [Foramen magnum stenosis from overgrowth of the opisthion in a child with achondroplasia.](#)
Achondroplasia has a known association with foramen magnum stenosis that... 10th October, 2008
Harvard Medical School, Boston, Massachusetts, USA.- J Neurosurg Pediatrics. 2008 Aug;2(2):136-8.
([DOI Direct Link](#))
- [Tibial Lengthening: Extraarticular Calcaneotibial Screw to Prevent Ankle Equinus.](#)
Between 2003 and 2006, we used an extraarticular, cannulated, fully... 19th September, 2008
SPARSH Hospital, Bangalore, India.- Clin Orthop Relat Res. 2008 Sep 18. ([DOI Direct Link](#))
- [Age-appropriate body mass index in children with achondroplasia: interpretation in relation to indexes of height.](#)
BACKGROUND: Achondroplasia is the most common short stature skeletal... 16th September, 2008
Johns Hopkins- Am J Clin Nutr. 2008 Aug;88(2):364-71.
- [Scott sign: a clinical measure of ligamentous laxity in achondroplastic infants.](#)
Achondroplasia has a known association with foramen magnum stenosis that... 16th September, 2008
Division of Genetics, Department of Pediatrics, A.I. DuPont Hospital for- Am J Med Genet A. 2008 Sep 1;146A(17):2291-2. ([DOI Direct Link](#))
- [The population-based prevalence of achondroplasia and thanatophoric dysplasia in selected regions of the US.](#)
There have been no large population-based studies of the prevalence of... 13th September, 2008
Houston Health Science Center, The University of Texas, Houston, Texas- Am J Med Genet A. 2008 Sep 15;146A(18):2385-9. ([DOI Direct Link](#))
- [Progressive joint limitations as the first alarming signs in a boy with short - limbed dwarfism: A case report.](#)
ABSTRACT: INTRODUCTION: Contracture is a condition of abnormal shortening... 21st August, 2008
Ludwig-Boltzmann Institute of Osteology at the Hanusch Hospital of WGKK- Cases J. 2008 Aug 19;1(1):112. ([DOI Direct Link](#))
- [Achondroplasia manifesting as enchondromatosis and ossification of the spinal ligaments: a case report.](#)
ABSTRACT: INTRODUCTION: A girl presented with achondroplasia manifested as... 13th August, 2008
Ludwig Boltzmann Institute of Osteology, Hanusch Hospital of WGKK and AUVA- J Med Case Reports. 2008 Aug 11;2:263. ([DOI Direct Link](#))
- [\[Prenatal diagnosis of achondroplasia\]](#)
OBJECTIVE: To diagnose achondroplasia prenatally by FGFR3 gene detection.... 7th August, 2008
Department of Obstetrics and Gynecology, the First Affiliated Hospital,- Zhonghua Yi Xue Yi Chuan Xue Za Zhi. 2008 Aug;25(4):427-9.

BioNews Results for Achondroplasia

- [Study Finds Clue to Genetic Cause of Fatal Birth Defect](#)
MedIndia: Oct 13 2008 2:52PM Matching: achondroplasia

Achondroplasia Clinical Trials:

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





Achondroplasia Patents:

- 7094565- [21612, novel human dehydrogenases](#)
- 7202025- [Control of gene expression](#)
- 7208274- [Rapid analysis of variations in a genome](#)
- 7217699- [Compositions comprising genome segments and methods of using the same](#)
- 7220406- [Method for promoting bone formation](#)
- 7226902- [Ligands for TGF-beta binding proteins and uses thereof](#)
- 7235241- [Monomer protein with bone morphogenetic activity and medicinal agent containing the same for preventing and treating diseases of cartilage and bone](#)
- 7235634- [2786, a novel human aminopeptidase](#)
- 7250405- [Modified pituitary gland development in offspring from expectant mother animals treated with growth hormone releasing hormone therapy](#)
- 7252974- [Human growth gene and short stature gene region](#)
- 7253266- [Polypeptides of FGF-CX](#)
- 7198790- [Growth differentiation factor-5](#)
- 7192583- [Methods for modulating bone mineralization](#)
- 7189507- [Methods of diagnosis of ovarian cancer, compositions and methods of screening for modulators of ovarian cancer](#)
- 7098015- [27875, 22025, 27420, 17906, 16319, 55092 and 10218 molecules and uses therefor](#)
- 7108984- [Methods of identifying modulators of the FGF receptor](#)
- 7108991- [Human orphan G protein-coupled receptors](#)
- 7119190- [Endogenous and non-endogenous versions of human G protein-coupled receptors](#)
- 7122550- [Protein kinase inhibitors](#)
- 7129072- [Crystal of fibroblast growth factor receptor 1 in complex with fibroblast growth factor](#)
- 7135311- [Means for detecting and treating pathologies linked to FGFR3](#)
- 7135320- [Adaptor protein FRS2 and related products and methods](#)
- 7160916- [Method of increasing bone density or treating osteoporosis](#)
- 7166461- [Polynucleotides encoding a super-active porcine growth hormone releasing hormone analog](#)
- 7259179- [Kinase inhibitors](#)
- 7273724- [Polypeptide-human actin-binding protein 54 and a polynucleotide encoding the same](#)
- 7368531- [Human secreted proteins](#)
- 7381409- [Antibodies specific for sclerostin and methods of screening and use 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](#)
- 7368527- [HADDE71 polypeptides](#)
- 7348142- [Cancer diagnostic panel](#)
- 7332277- [Methods for detection of genetic disorders](#)
- 7276481- [Method and composition for treatment of skeletal dysplasias](#)
- 7288392- [Human protein kinases and uses therefor](#)

- 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](#)
- 7442708- [1,2,3-substituted indolizine derivatives, inhibitors of fgfs, method for making same and pharmaceutical compositions containing same](#)

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