
DESCRIPTION

1 ACHONDROPLASIA

Synonyms: Chondrodystrophy, Fetal rickets

Involves the following Biologic System(s):

Genetic/Chromosomal/Syndrome/Metabolic Disorders,
Orthopedic and Muscle Disorders

Achondroplasia is a disorder of the skeletal system that belongs to a group of diseases known as chondrodystrophies. These disorders involve a disturbance in the development of cartilage of the long bones (e.g., arms and legs). They frequently cause a disproportionately short stature (dwarfism). Achondroplasia is a result of a spontaneous change in one gene (mutation) and is transmitted as an autosomal dominant trait. It occurs in approximately 1 out of every 40,000 births. In achondroplasia, there is an abnormality in the conversion of cartilage into bone at the growing ends (epiphyses) of the long bones. This process prevents the bones from further growth which ceases at a relatively early age. Symptoms and characteristic findings include a disproportionately large head with protruding or prominent forehead (frontal bossing); a flattened nasal bridge; an underdeveloped upper jaw and prominent lower jaw (prognathism); a well-developed, shortened trunk; and short, bowed arms and legs. The upper portion of arms and legs are proportionately shorter than other parts of the limbs; the elbows may have a limited range of motion. In addition, the fingers and toes are usually short with a v-shaped space or gap between the third and fourth fingers; however, the hands are relatively wide. As children with achondroplasia grow, the pelvis tilts forward, resulting in a pronounced spinal curvature (lumbar lordosis) characterized by a prominent abdomen and buttocks. Diagnosis is made based on physical examination and skeletal radiograph findings.

Complications associated with achondroplasia may include dental problems such as malocclusion as well as chronic and severe middle ear infections (otitis media) that may lead to conductive hearing loss. Potentially life-threatening complications include the temporary cessation of breathing during sleep (sleep apnea) due to obstruction of the airway as a result of certain craniofacial abnormalities or compression of the spinal cord at its entrance to the vertebral column (foramen magnum). In addition, hydrocephalus, which is a condition characterized by an abnormal accumulation of cerebrospinal fluid around the brain, may also have life-threatening implications. Hydrocephalus may also result from obstruction of the foramen magnum.

The availability of somatropin (recombinant human growth hormone) has revolutionized the treatment of short stature. Growth hormone is currently being used to augment the height of patients with achondroplasia. Other treatment for achondroplasia is directed toward prevention or correction of complications. Monitoring head growth during infancy to ensure that it is within established guidelines is effective to determine the presence of hydrocephalus and the need for surgical intervention or other measures.

To avoid further complications, dental irregularities, ear infections, and some skeletal irregularities may respond to physiotherapy as well as the use of orthopedic appliances such as braces. In addition, emotional and psychologic support may be provided through appropriate counseling.

See also **General Resources** on page 917

Government Agencies

2 NIH/National Institute of Arthritis and Musculoskeletal and Skin Diseases

1AMS Circle
Bethesda, MD 20892

301-402-4484
Fax: 301-718-6366
e-mail: ord@od.nih.gov
rarediseases.info.nih.gov

The mission of the National Institute of Arthritis and Musculoskeletal and Skin Diseases is to support research into the causes, treatment, and prevention of arthritis and musculoskeletal and skin diseases, the training of basic and clinical scientists to carry out this research, and the dissemination of information on research progress in these diseases.

Stephen I Katz MD PhD, Director

3 NIH/National Institute of Environmental Health Sciences (NIEHS)

111 T.W. Alexander Drive
Research Triangle Park, NC 27709

301-496-3511
www.niehs.nih.gov

NIEHS reduces the burden of human illness and dysfunction from environmental causes by defining how environmental exposures, genetics and age interact to affect an individual's health.

4 NIH/National Institute on Drug Abuse (NIDA)

6001 Executive Boulevard/Room 5213
Bethesda, MD 20892

301-443-1124
e-mail: information@nida.nih.gov
www.nida.nih.gov

NIDA leads the nation in bringing the power of science to bear on drug abuse and addiction through support and conduct of research across all disciplines and rapid and effective dissemination of results of that research to improve drug abuse and addiction prevention and treatment.

National Associations & Support Groups

5 Human Growth Foundation

997 Glen Cove Avenue, Suite 5
Glen Head, NY 11545

800-451-6434
Fax: 516-671-4055
e-mail: hgfl@hgfound.org
www.hgfound.org

A voluntary, nonprofit organization whose mission is to help children and adults with disorders of growth and growth hormones through research, education, support and advocacy. The foundation is dedicated to helping medical science to better understand the process of growth. It is composed of concerned parents and friends of children and adults with growth problems; and interested health professionals.

Frank Diamond, President
Emily Germain, Vice President

6 Little People of America

5289 NE Elam Young Parkway, Suite F-100
Hillsboro, OR 97124

503-846-1562
888-572-2001
Fax: 503-846-1590
e-mail: info@lpaonline.org
www.lpaonline.org

A nonprofit organization that provides support and information to people of short stature and their families.

Billy Barty, Founder
Lois Gerage-Lamb, President

7 MAGIC Foundation: Major Aspects of Growth in Children

6645 W North Avenue
Oak Park, IL 60302

708-383-0808
800-362-4423
Fax: 708-383-0899
e-mail: mary@magicfoundation.org
www.magicfoundation.org

A national nonprofit organization providing support and education regarding growth disorders in children and related adult disorders. Provides educational information, networking, a national conference, a kids' program and an extensive medical library.

10,000 members

Dianne Tamburrino, Executive Director
Susan Smith, Director Medical Education

State Agencies & Support Groups

Indiana

8 Neurofibromatosis, Inc - Indiana

1173 Hague Court
Franklin, IN 46131

317-736-7577
www.nfinc.org

Dottie Whitehurst, Chapter President

Libraries & Resource Centers

9 NIH/National Library of Medicine (NLM)

8600 Rockville Pike
Bethesda, MD 20894

www.nlm.nih.gov

NLM collects, organizes and makes available biomedical science information to scientists, health professionals and the public. The

library's databases, including PubMed/Medline and MedlinePlus, and used extensively around the world. NLM conducts and supports research in biometric communications; creates information resources for molecular biology, biotechnology, toxicology, and environmental health; and provides grant support for training, medical library resources, and biomedical informatics.

Research Centers

10 Little People's Research Fund

616 Old Edmondson Avenue
Catonsville, MD 21228

410-747-1100
800-232-5773
Fax: 410-747-1374
e-mail: lprf@LPRF.org
www.LPRF.org

Only health organization established to serve people with skeletal dysplasia (dwarfism), providing research, consultation, medical care and family respite.

Web Sites

11 Achondroplasia UK

www.achondroplasia.co.uk

Offers information on health supervision for children of all ages, divided into the following growth stages: newborn; infancy; early childhood; late childhood; and adolescence to early adulthood.

12 Dysmorphic Syndromes

www.hgmp.mrc.ac.uk/dhmd-bin/hum-look-up

13 Human Growth Foundation

www.hgfound.org

Information for parents and friends of children and adults with growth problems, as well as interested health professionals.

14 Little People of America

www.lpaonline.org

Offers resources pertaining to dwarfism and Little People of America, medical data, instructions on how to join an e-mail discussion group, and links to numerous other dwarfism-related sites.

15 MAGIC Foundation: Major Aspects of Growth in Children

www.magicfoundation.org

Provides educational information regarding growth disorders.

16 Medical College of Wisconsin

www.mcw.edu

A private, academic institution dedicated to leadership and excellence in education, research, patient care, and service.

17 Online Mendelian Inheritance in Man

www.ncbi.nlm.nih.gov

This database is a catalog of human genes and genetic disorders.

18 Restricted Growth Association

www.rgaonline.org.uk

Provides medical advice, welfare and counseling services with the support of Regional Coordinators, and offers contact with others and the sharing of helpful information through an information magazine, advisory booklets, meetings, social events and conventions.