Achondroplasia

By the Human Growth Foundation

Acknowledgements

The Human Growth Foundation is a nonprofit volunteer organization. Its mission is to help individuals with growth-related disorders, their families, and health-care professionals through education, research, and advocacy. It is composed of concerned parents and friends of children with growth problems and interested health professionals. Its objectives include:

* Support of research
* Family education, service, and support
* Public education
* Education of health-care professionals
* Advocacy on behalf of individuals with growth problems and their families

This booklet was written by Charles I. Scott Jr., MD.

This edition was revised and edited by Clair A. Francomano, MD; William Horton, MD; Patricia Rieser, FNP-C; Ruth E. Ricker; and the Education Committee of the Human Growth Foundation.

Publication sponsored by Human Growth Foundation and the National Center for Human Genome Research. Revised August 1996.

In memory of Eric Vonderlieth

Introduction

There are many conditions and diseases that can cause short stature. Some of these conditions involve a primary bone disorder -- the bones do not grow and develop normally. These conditions are called *skeletal dysplasias* or *chondrodystrophies*. People with disproportionate short stature often refer to themselves as dwarfs, little people, or short-statured persons. More than 100 specific skeletal dysplasias have been identified. Of these, achondroplasia is the most common. It occurs in all races and with equal frequency in males and females, and affects about one in every 25,000 children. It is estimated that there are about 10,000 individuals with achondroplasia in the United States.

Signs of achondroplasia

An individual with achondroplasia has disproportionate short stature: the head is large and the arms and legs are short when compared to the trunk length. This shortness is particularly noticeable in the upper arms and thighs. Other signs are a prominent forehead, a flat or even depressed area at the base of the nose (between the eyes), a protruding jaw, and sometimes poor dental structure -- the teeth are crowded and the upper and lower teeth may be poorly aligned. An adult with achondroplasia usually has an exaggerated forward curve to the lower spine, which presents a swaybacked appearance. The legs of a person with achondroplasia almost always become bowed and the elbows often cannot be straightened completely. Sometimes the person has limited twisting ability at the elbows. The hands are short, and the feet are short, broad, and flat. Another sign is "double" jointedness, caused by lax (loose) ligaments in some joints. Many achondroplastic children can flex their finger, wrist, hip, and knee joints to an extreme degree because of ligamentous laxity. These signs are usually apparent at birth, and achondroplasia can be diagnosed at that time in most cases. Intelligence is generally normal.

Affected men average 51.8 inches in height, while women average 48.6 inches. There seems to be little or no relationship between the height of the parents and the adult height of their children with achondroplasia.

Related physical conditions

Children with achondroplasia may reach motor milestones of development slowly. For instance, good head control may not occur until the infant is three or four months old, because it take longer to develop the muscular strength necessary to control the large head. Though there are exceptions, many of these children do not walk until relatively late, often between 24 and 36 months. Ultimately, overall development is usually normal.

Weight control is a frequent and lifelong problem for many people with this disorder. Both children and adults must be careful of their nutrition because they are prone to add excess weight.

Children with achondroplasia have a tendency toward middle-ear infections in the first five or six years of life, probably due to abnormal drainage of the tube from the middle ear to the throat. The basic cause is faulty development of the bone structure. If these infections are not recognized and treated, or are resistant to treatment, the child may sustain significant hearing loss. Each infection should be treated promptly and the child's hearing tested regularly. Many children with achondroplasia require ear "tubes" for treatment of recurrent ear infections.

Dental problems caused by overcrowding of teeth (especially those of the upper jaw) may occur. Malocclusion (poor bite) often results and makes good oral hygiene difficult. In addition to ordinary dental care, orthodontic treatment may be necessary.

The large head seen in achondroplasia is often confused with hydrocephalus ("water on the brain"). Hydrocephalus may develop in achondroplasia, but it rarely requires surgical treatment. It is important to measure the young child's head circumference regularly to distinguish hydrocephalus from normal head growth.

Young children with achondroplasia are at increased risk for specific neurologic and respiratory problems in some cases. These result from smaller openings in the skull, particularly at the foramen magnum (or "large window") at the base of the skull. In some cases, surgery is required to enlarge this opening and relieve pressure on the base of the brain and spinal cord.

Common orthopedic problems in childhood include a reverse curvature (kyphosis) of the lower spine in infancy, and bowed legs, which may develop any time after children begin to walk. The spinal kyphosis usually resolves without treatment, but may require bracing or surgery if it persists. Bowed legs may be treated by bracing or orthopedic surgery.

Older children and adults frequently experience fatigue, numbness, or pain in the lower back and thighs. Often these complaints are simple muscular problems that do not require special care. If they are persistent or severe, the person should be evaluated by a physician, perhaps a neurologist. Nerve or spinal-cord problems are common because of the narrow spinal canal, particularly in the lower back.

Causes of achondroplasia

Mutations (chemical changes) within a single gene cause achondroplasia. The condition may be passed from one generation to the next or it may result from a new mutation in a gene from average-sized parents. Nine out of ten children born with achondroplasia have average-sized parents, and no other family member is affected. A person with this mutation has achondroplasia; it is not possible to have a little bit of achondroplasia, or "only a touch of it" -- a person either has it or not. In 1994 Dr. John Wasmuth and his colleagues discovered that a mutation of the fibroblast growth factor receptor-3 (FGFR3) gene on human chromosome 4 causes achondroplasia. The exact way in which mutations in FGFR3 cause achondroplasia is not yet known. Much research is being done in this area.

Two average-statured parents have little or no chance of having more than one child with achondroplasia. However, an accurate and very specific diagnosis should be made to establish

that the short-statured child actually has achondroplasia. The tendency in the past was to diagnose everyone of short stature as having achondroplasia when, in fact, they had any of a large number of other conditions causing dwarfism. Before accurate counseling can be provided, the diagnosis must be known with certainty.

At present there is no specific treatment to promote growth in achondroplasia. Growth-hormone treatment seems to increase the rate of growth during the first year of treatment, but may not increase adult height. Surgery to lengthen the legs and arms of people with achondroplasia is being done on an experimental basis in a few centers in the US, but it is not a common practice because complications are frequent and the process is long and arduous.

Current therapy is directed toward preventing or treating complications of achondroplasia. The family physician, pediatrician, or internist, with the help of specialists such as endocrinologists, geneticists, orthopedists, and neurologists, can provide affected individuals with appropriate medical and psychological support. A great deal of research is being done on achondroplasia and other growth problems in medical centers across the country.

It is important for children with achondroplasia to recognize that a wide range of occupational choices exists for them and that their condition need not prevent them from leading a full and satisfying life. Contact with other well-adjusted short people demonstrates these possibilities andencourages the child to "Think Big" (motto of the Little People of America).

Resources

Two organizations concerned with the needs of individuals with growth problems and their families are Human Growth Foundation (HGF) and the Little People of America (LPA). Both groups serve as sources of information and support, and offer opportunities for parents to meet others facing similar challenges. Each organization has close contact with medical professionals to help refer interested individuals to the experts nearest them.

The HGF and LPA chapters exist in many cities and regions throughout the United States; more information about local and national activities can be obtained by contacting the national offices. Many families find membership worthwhile even if no local chapter exists because of newsletters, conferences, booklets, professional directories, pen-pal networks, and other activities that are sponsored nationally.

Human Growth Foundation (HGF) is a nonprofit organization made up of families affected by short stature and other individuals who are interested in growth problems. Its goals include support of research into normal and abnormal growth, family education and support, public education, and education of health professionals. National office: 7777 Leesburg Pike, Suite 202 South, Falls Church, VA 22043, telephone (800) 451-6434. Publications: *Patterns of Growth, Growth Hormone Deficiency, Turner Syndrome, Intrauterine Growth Retardation, Achondroplasia, Short and OK, Precocious Puberty, Growth Hormone Treatment: What To Expect, Growing Children: A Parents' Guide*. Member newsletters: *Outreach for Growth, Fourth Friday*. Website: http://www.hgfound.org. E-mail: hgf1@hgfound.org.

Little People of America (LPA) is a peer support group made up of persons with extreme short stature and their family members, as well as interested friends and professionals. National office: Box 9897, Washington, DC 20016. Publications: *My Child Is a Dwarf, The Idea Machine II, LPA Today* (newsletter). Website: http://www.lpaonline.org.

Short Stature Foundation (SSF) operates a toll-free helpline with information about services available to short people and publishes a catalogue of adaptive devices for people with growth and skeletal disorders. Its goal is to provide services, information, and advocacy to ensure the well-being and independence of short people. Call (800) 24-DWARF.