Achondroplasia

Achondroplasia is the most common type of short-limbed dwarfism. The condition occurs in 1 in 15,000 to 40,000 newborns.

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Major

Aspects of

Growth

In

Children

The MAGIC Foundation is a national nonprofit organization created to provide support services for the families of children afflicted with a wide variety of chronic and/or critical disorders, syndromes and diseases that affect a child's growth. Some of the diagnoses are quite common while others are very rare.

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Achondroplasia

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Dedicated to the growth and overall development of children

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Achondroplasia

Is a form of short-limbed dwarfism. The word achondroplasia literally means "without cartilage formation." Cartilage is a tough but flexible tissue that makes up much of the skeleton during early development. However, in achondroplasia the problem is not in forming cartilage but in converting it to bone (a process called ossification), particularly in the long bones of the arms and legs. Achondroplasia is similar to another skeletal disorder called hypochondroplasia, but the features of achondroplasia tend to be more severe.

All people with achondroplasia have short stature. The average height of an adult male with achondroplasia is 131 centimeters (4 feet, 4 inches), and the average height for adult females is 124 centimeters (4 feet, 1 inch). Characteristic features of achondroplasia include an average-size trunk, short arms and legs with particularly short upper arms and thighs, limited range of motion at the elbows, and an enlarged head (macrocephaly) with a prominent forehead. Fingers are typically short and the ring finger and middle finger may diverge, giving the hand a three-pronged (trident) appearance. People with achondroplasia are generally of normal intelligence.

Achondroplasia is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. About 80% of people with achondroplasia have average-size parents, these cases result from new mutations in the FGR3 gene. In the remaining cases, people with achondroplasia have inherited an altered FGR3 gene from one or two affected parents. Individuals who inherit two altered copies of this gene typically have a severe form of achondroplasia that causes extreme shortening of the bones and an underdeveloped rib cage.

Health problems commonly associated with achondroplasia include episodes in which breathing slows or stops for short periods (apnea), obesity, and recurrent ear infections. In childhood, individuals with the condition usually developed a pronounced and permanent sway of the lower back (lordosis) and bowed legs. Some affected people also develop abnormal front-to-back curvature of the spine (kyphosis) and back pain. A potentially serious complication of achondroplasia is spinal stenosis, which is a narrowing of the spinal canal that can pinch (compress) the upper part of the spinal cord. Spinal stenosis is associated with pain, tingling, and weakness in the legs that can cause difficulty with walking. Another uncommon but serious complication of achondroplasia is hydrocephalus, which is a building of fluid in the brain in affected children that can lead to increased head size and related brain abnormalities.

Mutations in the *FGFR3* gene cause achondroplasia. The *FGFR3* gene provides instructions for making a protein that is involved in the development and maintenance of bone and brain tissue. Two specific mutations in the *FGFR3* gene are responsible for almost all cases of achondroplasia. Researchers believe that these mutations cause the FGFR3 protein to be overly active, which interferes with skeletal development and leads to the disturbances in bone growth seen with this disorder.

As new drug applications are in the pipeline for achondroplasia, keeping in touch with the MAGIC Foundation will enable you to follow the progress. The goal of new therapy is to lessen the overall complications, not just height. Additionally, through the foundation, groups are available to communicate with other families of children with achondroplasia.