FACT SHEET - ACHONDROPLASIA

Achondroplasia is:

- the most common type of dwarfism
- a genetic condition that results in short stature with disproportionately short limbs

Achondroplasia is not:

- an intellectual disability
- a disease that requires a "cure"
- a reason to assume someone is incapable

Characteristics

Achondroplasia is a disorder of bone development in which some bones in the body grow at a much slower pace than usual, resulting in short stature. It occurs in approximately one of every 20,000 live births. The bones most severely affected are those of the thighs and upper arms, resulting in significant shortening. The lower legs and forearms are affected to a lesser degree, while the hands and feet are also short with a characteristic separation between the third and fourth digits. The head is larger than in average-size people, with a prominent forehead and flat nose. The average height of adult males with achondroplasia is 4 feet, 4 inches. The average height of adult females with achondroplasia is 4 feet, 1 inch.

Causes

Achondroplasia is caused by a mutation (change) in a gene (instruction) that regulates the growth of several bones in the body. The gene responsible for achondroplasia is known as FGFR3; when it is changed it leads to shorter bones. Males and females are equally affected. The majority of achondroplasia cases (80%) are the result of a new mutation that occurs in the sperm or ovum of one of the parents. In this case both parents are of average height and do not have the abnormal gene. The chance of a new mutation rises with the age of the father.

People with achondroplasia have a 50% chance to pass the changed gene to a child. If both parents have achondroplasia, with each pregnancy there is a 50% chance to have a child with achondroplasia, a 25% chance that the child will be of average height and not inherit the changed gene, and a 25% chance that the child will inherit the changed gene from each parent. The latter can lead to severe skeletal problems and may result in early death.

Diagnosis

Achondroplasia can be diagnosed before birth by fetal ultrasound during the third trimester, or after birth by body measurements and by studying x-ray images; this usually happens in the context of a clinical genetics evaluation. It is important to get a diagnosis early, to understand which skeletal dysplasia the child has. While most children are just fine in the very early stages, being aware of what potentially might happen can prevent serious problems down the road. What doctors are looking for early on are problems with compression of the upper cervical cord at the level of the base of the skull. These could lead to breathing problems and/or accumulation of fluid in the head, a problem known as hydrocephalus. There are genetic tests available that can confirm the mutation (change) in the gene in an affected person; they can be used for early prenatal testing if desired.

Treatment

Children and adults with achondroplasia can lead normal lives provided they receive attentive, informed care by their physicians and parents. Monitoring should include careful measurements of growth (length/height and weight) and head circumference using curves specially standardized for achondroplasia [for specialized growth charts check out the LPO website]. Young children are usually bow legged, but bowing does not usually result in any serious long term problems. Treatment with growth hormone does not substantially affect the height of a person with achondroplasia. Leg-lengthening surgeries may be considered in some very specialized cases.

Middle ear infections are frequent and can lead to mild to moderate hearing loss. Therefore, ear infections should be readily suspected and promptly treated. Monitoring and controlling weight is also important because excess weight aggravates back and joint problems.

Assessment by a geneticist to confirm a diagnosis, and genetic counselling for discussion of the natural history of the disease, the risk of recurrence in future pregnancies, and available options for management is highly recommended.

Misconceptions

People of average height often have misperceptions about people with dwarfism. Many wrongly believe that people of short stature have limited intellectual abilities or personality disorders. In fact, the vast majority of short statured individuals achieve a full and rich life, and are able to be very productive members of society.

Help and Advice

As a non-profit organization, Little People of Ontario (LPO) provides people of short stature and their families with a life-long, supportive community. From the first moments when a child is diagnosed with dwarfism, to the first day of school, through the teen years, young adulthood, career, marriage, parenting and aging, the organization provides mentorship and valuable information. Check out our website at www.lpo.on.ca or contact us at:

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References:

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Generously funded by
The Ontario Trillium Foundation / La Fondation Trillium de l'Ontario