



FACT SHEET - DWARFISM

Dwarfism is:

- a genetic condition
- generally defined as an adult height of 4 feet 10 inches or less

Dwarfism is not:

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

Types of Dwarfism

There are more than 200 different types of dwarfism. Most are known as **skeletal dysplasias**, which are conditions of abnormal bone growth. These occur in 1-3 of every 10,000 births. By far, the most common skeletal dysplasia is **achondroplasia**, a short-limb dysplasia that occurs in about 1 of every 20,000 babies of all races and ethnicities. The typical range in adult height among people with dwarfism is 2 feet 8 inches to 4 feet 8 inches. Other relatively common genetic conditions that result in short stature include **spondyloepiphyseal dysplasia congenita (SEDC)**, **diastrophic dysplasia**, **pseudoachondroplasia**, **hypochondroplasia**, and **osteogenesis imperfecta (OI)**.

Causes

Most types of dwarfism are caused by a spontaneous genetic mutation (a new change in a gene) in the egg or sperm cell prior to conception. What prompts the gene to mutate is not yet understood. The change is random and unpreventable, and can occur in any pregnancy. Generally, when average-size parents have a child with short stature due to a spontaneous mutation, it is rare to have a second child of short stature. However, if a parent has some form of dwarfism, depending on the type, there can be a 50% chance of passing the condition on to the children. This is known as dominant inheritance.

Some types of dwarfism are caused by changed genes inherited from both parents (these are called recessive conditions). In these cases, for each pregnancy there is a 25% chance of the condition being present. This means that two average-size parents might have a short statured child, or two short statured parents might have an average-size child. A genetic evaluation may allow a diagnosis to be made and provide opportunity to discuss with the family what the likelihood is for recurrence to occur.

Diagnosis

Some types of dwarfism can be identified through prenatal testing, but most are identified after a child is born. Doctors make a diagnosis based on the child's appearance, growth patterns, and x-rays of the bones. Even then, the diagnosis may be very difficult. Some newborn babies are relatively well proportioned, and it isn't until later on that disproportionate growth patterns become apparent or the characteristic x-ray changes become present. It is important to make a definitive diagnosis of the type of dwarfism in order to make parents aware of any possible health concerns or complications that may occur. An assessment by a geneticist with expertise in skeletal dysplasias may help in making the diagnosis and deciding whether confirmatory genetic testing is available.

Treatment

There is no “cure” for dwarfism. However the vast majority of people who are of short stature go through life without having serious medical problems, and don’t require any aggressive surgical interventions. Each of the conditions that cause dwarfism has its own set of characteristics and possible complications. A small percentage will have difficulties right at birth; therefore, it is important to obtain an early diagnosis of the specific skeletal dysplasia that a child has.

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, most individuals with short stature 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 challenging 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:

- http://medical.lpaonline.org/dwarfism_types/
- <http://kidshealth.org/parent/medical/bones/dwarfism.html>
- <http://www.chkd.org/HealthLibrary/content.aspx?pageid=P01938>

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