

GROWTH HORMONE DEFICIENCY (DWARFISM)

Common Names for Disorder:

- Little people
- Short stature

Causes/Etiology:

Two disorders, Achondroplasia and growth hormone deficiency (also known as pituitary dwarfism), are responsible for the majority of dwarfism cases.

The most recognizable and most common form of dwarfism is Achondroplasia, which accounts for 70% of dwarfism cases and produces rhizomelic short limbs, increased spinal curvature, and distortion of skull growth. Achondroplasia is an autosomal dominant disorder caused by the presence of a faulty allele in a person's genome. If a pair of achondroplasia alleles are present, the result is fatal. Achondroplasia is a mutation in the gene 3 (*FGFR3*) which is an inhibitor that regulates bone growth. In cases of Achondroplasia, the FGR3 gene is too aggressive, negatively impacting bone growth.

Growth hormone deficiency (GHD) is a medical condition in which the body produces insufficient growth hormone. Growth hormone, also called somatropin, is a polypeptide hormone, which stimulates growth and cell reproduction. If this hormone is lacking, stunted or even halted growth may become apparent. Children with this disorder may grow slowly and puberty may be delayed by several years, or even indefinitely. Growth hormone deficiency has no single definite cause. It can be caused by mutations of specific genes, damage to the pituitary gland, Turner's Syndrome, poor nutrition, or even stress (leading to psychogenic dwarfism)

Other causes of dwarfism include spondyloepiphyseal dysplasia congenita, diastrophic dysplasia, pseudoachondroplasia, hypochondroplasia, primordial dwarfism, Turner syndrome, and osteogenesis imperfecta (OI) and hypothyroidism. Severe shortness with skeletal distortion also occurs in several of the Mucopolysaccharidoses and other storage disorders.

Incidence:

Dwarfism is listed as a "rare disease" by the Office of Rare Diseases (ORD) of the National Institutes of Health (NIH). This means that Dwarfism, or a subtype of Dwarfism, affects less than 200,000 people in the US population.

Characteristics:

- Usually defined as an adult height of less than 4 feet 10 inches (147 cm)
- Children with growth hormone deficiency have a slow rate of growth, usually less than 2 inches per year. The slow growth may not appear until a child is 2 or 3 years old.
- Children with growth hormone deficiency still have normal body proportions, as well as normal intelligence. However, their face often appears younger than children of the same age. They may also have a chubby body build.

- Children with physical defects of the face and skull, such as cleft lip or cleft palate, can also have decreased growth hormone levels.
- Physical maleffects of malformed bones vary according to the specific disease. Many involve joint pain caused by abnormal bone alignment, or from nerve compression. Early degenerative joint disease, exaggerated lordosis or scoliosis can cause pain and disability. Other reductions in size can restrict lung growth and reduce pulmonary function. Some more severe forms of dwarfism are associated with disordered function of other organs, such as the brain or liver.

Primordial Dwarfism

- Primordial Dwarfism is a rare form of dwarfism that results in a smaller body size in all stages of life beginning from before birth. The major difference between Primordial Dwarfism and Dwarfism is that those with Primordial Dwarfism have bones and organs of that are proportionally smaller than in an average person.
- Having Primordial Dwarfism includes specific types of profoundly proportionate dwarfism, in which individuals are extremely small for their age, even as a fetus.
- Most individuals with primordial dwarfism are not diagnosed until they are about 3 years of age.
- Typically, people with primordial dwarfism are born with very low birth weights. After birth, growth continues at a stunted rate, leaving individuals with primordial dwarfism perpetually years behind their peers in stature and in weight.
- There are several subtypes of Primordial Dwarfism: Majewski osteodysplastic primordial dwarfism (MOPD) type I, Majewski osteodysplastic primordial dwarfism (MOPD) type II, Majewski osteodysplastic primordial dwarfism (MOPD) type III, Meyer-Gorlin Syndrome, Seckel Syndrome, and Russell-Silver Syndrome.
- Primordial Dwarfism is the most severe subtype of Dwarfism.
- Those diagnosed with Primordial Dwarfism usually don't live past age 30.

IDEA Category:

- Growth Hormone Deficiency is classified as "Other Health Impaired" (OHI)
- It may also be possible to classify someone as "Developmentally Delayed," assuming they meet the criteria necessary for fulfillment (e.g. low cognitive functioning).

DSM-IV Category:

- Growth Hormone Deficiency does not have a DSM IV diagnosis.
- It is diagnosed under Axis III with a ICD-9-CM code of 393.3, dwarfism, pituitary

Deficits:

- If left untreated, growth hormone deficiency will lead to short stature and delayed puberty.
- Self-esteem may suffer and family relationships may be affected.
- Extreme shortness (in the low 2–3 foot [60–90 cm] range) can interfere with ordinary activities of daily living, like driving or using countertops built for taller people. Other symptoms of dwarfism such as bowed knees and unusually short fingers can lead to back problems, difficulty in walking, and handling objects.
- Children with dwarfism are particularly vulnerable to teasing and ridicule from classmates. Because dwarfism is relatively uncommon, children may feel isolated from their peers

Growth hormone deficiency may occur with deficiencies of other hormones, including the following:

- Thyrotropins (control production of thyroid hormones)
- Vasopressin (controls water balance in the body)
- Gonadotropins (control production of male and female sex hormones)
- Adrenocorticotrophic hormone or ACTH (controls the adrenal gland and its production of cortisol, DHEA, and other hormones)

These deficiencies might impact the students overall well-being and ability to achieve in school.

Long-term Developmental Outcomes:

The earlier the condition is treated, the better the chance that a child will grow to be a near-normal adult height. Many children gain 4 or more inches over the first year and 3 or more inches during the next 2 years. The rate of growth improvement then slowly decreases. However it is important to note that, growth hormone replacement therapy does not work for all children.

Social prejudice or heightism against extreme shortness may reduce social and marital opportunities. Numerous studies have also demonstrated reduced employment opportunities for individual with growth hormone deficiency. Severe shortness is associated with lower income.

Assessment Approaches:

Approaches in the screening and diagnosis process (conducted by Primary care Physicians/Neurologists):

- A physical examination including weight, height, and body proportions will show signs of slowed growth rate. The child will not follow the normal growth curves.
- Dual energy x-ray absorptiometry (DEXA) scans can also determine bone age.
- Hand x-ray (usually the left hand) can determine bone age. Normally, the size and shape of bones change as a person grows. These changes can be seen on an x-ray and usually follow a pattern as a child grows older.
- Measuring growth hormone and binding protein levels (IGF-I and IGFBP-3) will show whether the growth problem is caused by a problem with the pituitary gland.
- MRI of the head can show the hypothalamus and pituitary glands.
- Tests to measure other hormone levels (lack of growth hormone may not be the only problem) may be done.
- X-ray of the head may show problems with the skull.

Interventions & Treatments:

Treatment involves growth hormone injections given at home. Patients may receive growth hormone several times a week or once a day. Growth hormone is rarely used for shortness caused by bone dysplasias, since the height benefit is typically small.

Serious side effects of growth hormone therapy are rare. The most common side effects are:

- Fluid retention
- Muscle and joint aches

Disability may be alleviated by physical therapy, braces or other orthotic devices, or by surgical procedures (if possible).

The only simple interventions that increase perceived adult height are dress enhancements, such as shoe lifts or hairstyle.

The most effective means of increasing adult height by several inches is distraction osteogenesis, though availability is limited and the cost is high in terms of money, discomfort, and disruption of life. Most people with dwarfism do not choose this option, and it remains controversial.

Contributions of the School Psychologist:

- Growth deficit disorder may interfere with the child's ability to learn. If the student experiences pain or discomfort, he or she may be missing parts of what the teacher is saying. It is important that the teacher, parent, and school nurse be aware of the child's stress.
- Some children may need additional assistance to help them keep up with classmates and function in the classroom setting. Assistance can include adaptations in classroom instruction, and physical environment. All of which should be written in the IEP or 504 Plan.
- It is important that the teachers and school staff are informed about the child's condition, possible teasing and social ostracization, and what to do to counteract these concerns.
- School personnel and the family should work together to monitor the progress the student is making in school as well as any side effects growth hormone treatments might be causing.
- As noted earlier children with dwarfism must also deal with the psychological and social aspects of the condition. These include public misperceptions and inappropriate comments. To help children feel more confident about themselves and accept their growth deficits, the school psychologist can assist by providing dwarfism education programs for staff and students, including information on inclusion of these individuals and their potential. In addition, one on one or group counseling may help the student understand and accept their disorder.

Additional Resources:

<http://www.nlm.nih.gov/medlineplus/dwarfism.html>

<http://www.shortsupport.org/Health/Dwarfism.html>

<http://www.mayoclinic.com/health/dwarfism/DS01012/DSECTION=symptoms>

<http://www.mayoclinic.com/health/dwarfism/DS01012/DSECTION=symptoms>

<http://www.potentialsfoundation.org/>

<http://www.primordialdwarfism.com/>

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