Dwarfism

Dwarfism is the medical terminology for short-stature. It is defined as height-vertex below two standard deviations (-2SD) or in the third percentile for agiven age and sex. The standing height is measured from ground to the vertex when the head is in the Frankfurt-horizontal plane.

Dwarfism is broadly categorized into two types based on the patient's physical appearance, which are: proportionate short stature (PSS) anddisproportionate short stature (DSS). Proportionate short stature (PSS), as the name suggests, means that the limbs and the trunk are proportionatelysmall. Whereas, disproportionate short stature (DSS) implies that the individual has a significant difference in their sitting and standing height, andeither their trunk or extremities are small. The assessment and evaluation aim to identify the pathological causes of short stature and intervene accordingly.

Disproportionate dwarfism is marked by an average-size torso and shorter arms and legs or a shortened trunk with longer limbs.

In proportionate dwarfism, the body parts are in proportion but shortened.

Along with short stature, dwarfism has many other symptoms that can vary depending the type it is.

Disproportionate dwarfism symptoms

Disproportionate dwarfism usually doesn’t affect intellectual development unless a child has other rare conditions, including hydrocephalus, or excess

fluid around the brain.

Symptoms of disproportionate dwarfism may include:

Adults typically about 4 feet tall

Average-size torso and very short limbs, especially in the upper halves of arms and legs

Short fingers

Wide spaces between the middle and ring fingers

Limited elbow mobility

Disproportionately large head

Prominent forehead

Flattened bridge of the nose

Bowing of legs that worsens over time

Swaying of the back that worsens over time

Some people with disproportionate dwarfism have a rare disorder called spondyloepiphyseal dysplasia congenita (SEDC). Adults with SEDC are around 3 to 4 feet tall and may have these signs too:

Very short trunk

Short neck, arms, and legs, but average-size hands and feet

Broad chest

Flattened cheekbones

Cleft palate, or an opening in the roof of the mouth

Unstable neck bones

Deformed hips where the thigh bones turn inward

Twisted feet

Hunched upper back that worsens

Swayed lower back that worsens

Arthritis

Joint mobility problems

Impaired vision or hearing

Proportionate dwarfism symptoms

Proportionate dwarfism is caused by a medical condition you have at birth or that develops in childhood that hinders growth and development. One common cause is too-low amounts of growth hormone produced by your pituitary gland.

Symptoms of proportionate dwarfism include a smaller head, arms, and legs. But all are in proportion with each other. Organ systems may be smaller too.

Other signs of proportionate dwarfism in children are:

Slower growth rate than expected for their age

Height lower than the third percentile on standard charts for age

Delayed or no sexual development during the teen years.

CAUSES

Causes of proportionate dwarfism include metabolic and hormonal disorders such as growth hormone deficiency. The most common types of

dwarfism, known as skeletal dysplasias, are genetic. Skeletal dysplasias are conditions of abnormal bone growth that cause disproportionate dwarfism.

The short stature can either be due to an underlying medical condition or a standard variant of growth. FSS, CDGP, and ISS described below can be

considered as normal variants of growth, while the rest are pathological causes-

1. Familial Short Stature (FSS): The child's genetic height potential can be measured by measuring the mid-parental height, which is a child's projectedadult height based on parental height measurements. A positive family history, and the absence of underlying pathological etiologies of dwarfism, can

be diagnosed as a case of familial short stature. This child has an expected growth velocity, and his bone age is consistent with the chronological age.

X-Ray of the hand and wrist can establish the bone age and is a frequently used modality.

2. Constitutional delay of growth and puberty (CDGP): The child presents with short height in childhood, but attain their target height until adulthood,

also known as a late bloomer. They even enter puberty at later ages. Unlike in familial cases, these children have bone age lagging behind the

chronological age. Malnutrition in gestational age or childhood, even genetics, could be the plausible cause for this short stature pattern.

3. Idiopathic Short Stature (ISS): Short stature is said to be idiopathic when no other

etiology like endocrine/metabolic can be determined. With the advancements in genomic studies, it is found that many cases previously established as idiopathic can be explained by hundreds of genetic mutations with small or large effects.

4. Endocrine disorders: Growth hormone deficiency is a frequent cause of dwarfism. The hypothalamic-pituitary axis maintains the levels of growth hormones in the body, which then directly or indirectly, through Insulin-like growth factor-1 (IGF-1), stimulates bone elongation, and growth of soft tissue and cartilage. Lower levels of IGF-1 are linked with short stature. Precocious puberty is defined as the attainment of puberty before the age of 8 years in girls and nine years in boys due to the premature release of gonadotropins centrally or peripherally. These gonadotropins lead to an initial growth spurt but also lead to the early maturation of the child's skeletal system resulting in short adult height.

5. Genetic disorders: Many genetic conditions that are associated with short stature are- Down's, Turner's, Noonan's, Prader-Willi, Russell-Silver, and short stature homeobox gene deficiency syndrome. Short stature is one clinical manifestation among several others.

Turner syndrome. This genetic condition only affects females. It's caused by a missing or partial X chromosome. Girls with Turner syndrome only inherit one fully functioning X chromosome from their parents, instead of one from each parent.

6. Bone diseases: The faulty formation of bone can also lead to short stature. The bone disorders linked to dwarfism are-

Achondroplasia: It is an autosomal dominant genetic condition due to the mutation in the Fibroblast growth factor receptor-3 (FGFR-3) gene. This gene typically limits the conversion of cartilage to bone, particularly in the long bones. In achondroplasia, this gene becomes overactive and bone formation is severely impacted in the extremities. Another cause of short-limb dwarfism is diastrophic dysplasia.

Achondroplasia. The most common form of dwarfism – accounting for 70% of cases – achondroplasia occurs in about 1 out of 26,000 to 40,000 babies
and is evident at birth. People with achondroplasia have a relatively long trunk and shortened upper parts of their arms and legs. Other features of
achondroplasia include:

A large head with a prominent forehead

A flattened bridge of the nose

Protruding jaw

Crowded and misaligned teeth

Forward curvature of the lower spine

Bowed legs

Flat, short, broad feet

Double-jointedness

Spondyloepiphyseal dysplasia: It is a cause of short-trunk dwarfism with the vertebraes primary involvement and the proximal epiphyseal centers.

Type-2 collagen matrix is abnormal, but it remains unclear how this abnormality spares the distal epiphysis, given that collagen is ubiquitous.

Spondyloepiphyseal dysplasia (SED). A less common form of dwarfism, SED affects about 1 in 95,000 babies. Spondyloepiphyseal dysplasia refers to
a group of conditions marked by a shortened trunk, which may not become apparent until a child is between ages 5 and 10. Other features can include:

Club feet

Cleft palate

Severe osteoarthritis in the hips

Weak hands and feet

Barrel-chested appearance

Diastrophic dysplasia. A rare form of dwarfism, diastrophic dysplasia occurs in about 1 in 100,000 births. People who have it tend to have shortened
forearms and calves (this is known as mesomelic shortening).

Other signs can include

Deformed hands and feet

Limited range of motion

Cleft palate

Ears with a cauliflower appearance

7. Systemic disorders: Other systemic diseases which have a secondary effect on growth are undernutrition, juvenile idiopathic arthritis, inflammatory bowel disease (IBD), celiac disease, chronic kidney disease (CKD), pulmonary/cardiac/immunologic/metabolic diseases, cancers, and glucocorticoid therapy.Malnutrition can reduce the growth velocity and cause short stature. It can affect the baby in the gestational period, leading to underweight or small for gestational age (SGA) baby, or in childhood leading to stunted growth.

EVALUATION

Dwarfism Diagnosis

Some forms of dwarfism are evident in utero, at birth, or during infancy and can be diagnosed through X-rays and a physical exam. A diagnosis of achondroplasia, diastrophic dysplasia, or spondyloepiphyseal dysplasia can be confirmed through genetic testing. In some cases, prenatal testing is done if there is concern for specific conditions.

Sometimes dwarfism doesn't become evident until later in a child's life, when dwarfism signs lead parents to seek a diagnosis. Here are signs and symptoms to look for in children that indicate a potential for dwarfism:

A larger head

Late development of certain motor skills, such as sitting up or walking

Breathing problems

Curvature of the spine

Bowed legs

Joint stiffness and arthritis

Lower back pain and numbness in the legs

Crowding of teeth

Appearance. Children with possible dwarfism may have changes to their skeleton or facial structures as they develop.

Chart comparisons. At regular check-ups, your child's height, weight, and head circumference will be measured and compared to percentiles for

standard development for their age. If your child shows any signs of abnormal growth, they may need more frequent measurements.

Imaging. Doctors may spot signs of achondroplasia, such as shorter limbs, or other causes of dwarfism on ultrasounds of a fetus during pregnancy. X-

rays of babies or children may show that their arms or legs are not growing at a normal rate, or that their skeleton shows signs of dysplasia. MRI scans

can show any problems with the pituitary gland or hypothalamus, which affect hormone production.

Genetic testing. DNA tests may be done before or after birth to look for genetic mutations linked to dwarfism. Girls with suspected Turner syndrome

may need DNA tests to check their X chromosomes. DNA testing may help parents with family planning if they wish to have more children.

Family history. Pediatricians may check the height and size of other family members, such as siblings, to compare with the child with suspected

dwarfism.

Hormone tests. Tests of growth hormone levels can confirm if they're low.

child under evaluation undergoes a series of biochemical and radiological tests:

Biochemical Tests

Complete blood count for hematological diseases like anemia.

Sweat chloride test for cystic fibrosis. Patients usually have a history of meconium ileus and pulmonary symptoms.

Serum thyrotropin (TSH) levels and free thyroxine levels (T4) to test for hypothyroidism.

Wintrobe sedimentation rate for inflammatory bowel disease (IBD)

Antibody testing for celiac sprue includes anti-endomysial immunoglobulin A (IgA), immunoglobulin G (IgG), and anti-gliadin IgG titers. Antiendomysial IgA titers are more sensitive, and IgG titers are more specific.

Serum pre-albumin and transferrin- These values are low in undernutrition.

Imaging

Anteroposterior X-ray of the left hand and wrist: It is used to predict the bone age. The radiologist uses the Greulich and Pyle or Tanner-Goldstein-Whitehouse method to calculate the bone age and forecast adult height.

MRI can provide evidence of any intracranial masses (craniopharyngiomas)or developmental anomalies of the pituitary. Before the GH therapy is initiated, an MRI scan should be ordered to rule out any organic lesions.

Karyotyping: It can detect genetic disorders like Down syndrome, Turner syndrome, and others.

Provocative tests: These tests are used to measure the GH reserve, but their utility is limited. They are not reproducible and do not determine the physiological secretory pattern of GH. These include- insulin tolerance test, Levodopa-propanol HCL test, Arginine HCL test, Glucagon test. Other physiological stimuli that can be used are strenuous exercise, deep sleep, and fasting. The levels of growth hormone fluctuate throughout the day, and peaks after meals, after strenuous exercise, and in a deep sleep. This property makes it an unreliable biomarker for detecting growth hormone deficiency (GHD); however, stimulated GH values of more than 10mg/dl can essentially rule out the deficit. Therefore, more predictive markers are serum IGF-1 levels (somatomedin C) and insulin-like growth factor binding protein-3 (IGFBP-3) levels. IGFBP-3 has more specificity than IGF-1. In patients in the pubertal phase, with CNS neoplasms, and poor nutrition, these markers can be abnormally high even in the presence of GHD and are unreliable.

TREATMENT

Early diagnosis and treatment can help prevent or lessen some of the problems that

come with dwarfism. People with dwarfism related to growth hormone deficiency can be treated with growth hormone. For children with achondroplasia who still have the potential for growth, the FDA has approved vosoritide (Voxzogo) to help stimulate bone growth.

In many cases, people with dwarfism have orthopedic or medical complications. Treatment of those can include:

Insertion of a shunt to drain excess fluid and relieve pressure on the brain

A tracheotomy to improve breathing through small airways

Corrective surgeries for deformities such as cleft palate, club foot, or bowed legs

Surgery to remove tonsils or adenoids to improve breathing problems related to large tonsils, small facial structures, and/or a small chest

Surgery to widen the spinal canal (the opening through which the spinal cord passes) to relieve spinal cord compression

Extended limb lengthening, a controversial surgery, due in part to its risks, involves several procedures. It is only done on adults.

Other treatment may include:

Physical therapy to strengthen muscles and increase joint range of motion

Back braces to improve curvature of the spine

Placement of drainage tubes in the middle ear to help prevent hearing loss due to repeated ear infections

Orthodontic treatment to relieve crowding of teeth caused by a small jaw

Nutritional guidance and exercise to help prevent obesity, which can worsen skeletal problems

Dwarfism Complications

Disproportionate dwarfism causes changes to the limbs, back, and head size that could cause complications like these:

Arthritis

Back pain or breathing difficulties due to a hunched or swayed back

Bowed legs

Crowded teeth

Delayed motor skills development

Frequent ear infections and possibly hearing loss

Hydrocephalus (fluid on the brain)

Pressure on the spine at the base of the skull

Sleep apnea

Spinal stenosis, a narrowing of the spine that may cause leg pain or numbness

Weight gain that may cause back problems.

Proportionate dwarfism may cause you to have smaller or less developed organs. Girls with Turner syndrome may have heart problems. Kids with lower growth hormone levels or Turner syndrome may have emotional or social problems due to delays in sexual development.

Women with disproportionate dwarfism may have pregnancy complications, such as respiratory problems. They almost always need to deliver their babies by C-section, as the shape of their pelvis makes vaginal delivery too difficult.

Anyone with dwarfism may encounter people who call them names or don't understand their condition. Kids with dwarfism who have lower self-esteem may need emotional support from their parents to deal with their feelings.

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