

Short Stature-An Etiological Approach

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Abstract

Short stature is a complex problem in orthopaedics. There are many causes for it. Short stature is divided into 'proportionate' and 'disproportionate' varieties. The causes can vary from familial to racial. There are so many syndromes associated with short stature. The etiology and salient features of some common conditions associated with short stature are described below.

Keywords: Short Stature; Proportionate short stature; Disproportionate short stature

Introduction

The term 'short stature' is used when the height of an individual is below the third percentile of a normal population of their sex and age. Dwarfism is the condition, when the height of the individual is below 2 Standard Deviations (SD) [1-3]. We must avoid using the term dwarfism. The height of 95% of normal children lies between the 3rd and 97th percentile or within 2 standard deviations above or below the mean height for the age [4]. Short stature can be either proportionate as in genetic short stature or constitutional delay in growth or disproportionated as in skeletal dysplasias [5]. Disproportionate short stature can be either short limb or short trunk [6]. Achondroplasia, Hypochondroplasia, Diastrophic dysplasia are examples short limb variety [7]. They are again divided into rhizomelic, mesomelic and acromelic varieties according to the areas of involvement [5]. Mucopolysaccharidosis, Spondyloepiphyseal dysplasia and spinal deformities are included in the short trunk variety [8]. Here we are going to describe the features of some common causes of short stature.

Genetic short stature

Short parental height is the main factor for short stature in most children. We can see this by plotting the child's height on a growth chart and comparing it with the parent's height [9].

Growth delay

This is usually associated with delayed puberty. It can be familial. There is delay in bone age by 3 years or more. Usually normal height and sexual development will be attained at a later date [9].

Chromosomal anomalies

Turner's syndrome is a chromosomal anomaly causing short stature in girls [10]. The karyotype is 45/X0. The average height is 140 cm and usually not above 150 cm. Prevalence is one per 3000 female births. Internal and external genitalia are rudimentary, no breast development and having primary amenorrhoea. Webbing of neck, low hair line, cubitus valgus, short 4th and 5th metacarpals, Madelung deformity, and abnormal finger prints are other features [11]. Another condition resembling Turner's syndrome is Noonan's syndrome which can occur in both boys and girls. Normal karyotype, short stature, webbed neck, pectus carinatum, antimongoloid slant of the eyes, triangular face are the common features. Cubitus valgus, undescended testis, sometimes mental retardation can be seen [12]. Sometimes, delayed puberty is reported, but eventually it becomes normal.

Trisomy 21 or 'Down Syndrome' is a common cause of short stature. Large and protruding tongue, epicanthic folds in the eyes, Simian crease, Wormian bones in the skull are some of the features. Even though rare, Trisomy 18 – Edward's Syndrome and Trisomy 13 - Patau's Syndrome can produce short stature [13].

Chronic debilitating diseases and malnutrition

Childhood occurrence of tuberculosis, bronchiectasis, and cystic fibrosis can lead to short stature [14]. Children with chronic asthma on steroid treatment can have a stunted growth and short stature [15]. Mal-absorption due to coeliac disease, crohn's disease, lactose intolerance can lead to short stature [16,17]. Infection with giardiasis, hook worms can also produce mal-absorption. Chronic renal failure, cyanotic heart disease can cause stunning of growth [18]. Protein energy malnutrition like marasmus and kwashiorkor and rickets can lead to short stature [19]. Chronic infections like tuberculosis, malaria, leishmaniasis, chronic pyelonephritis can cause short stature especially in developing countries. Haematological conditions like thalassemia and sickle cell disease are other causes [20].

Psycho-social short stature

Adverse psychological environment, child abuse and emotional deprivation have shown to produce suppression of pituitary function and temporary retardation of growth. It is reversible, once the child is transferred to a happier and secure environment. In type 1 psychosocial short stature, which is seen below 2 years, behavioural development is retarded, whereas, growth hormone is normal. Type 2 occurs in children older than 3 years [21].

Orthopaedic anomalies

There are many inherited and non-inherited orthopaedic causes for short stature. Achondroplasia is an autosomal dominantly inherited condition. It is due to defect in the endochondral ossification. It is a classical variety of short limb dwarfism. It is characterised by rhizomelic short stature, frontal bossing, depressed nose, trident hand, genu varus, and spinal canal stenosis. Hypochondroplasia is similar to achondroplasia but face is normal. The inheritance is also autosomal dominant [7,22,23]. Multiple epiphyseal dysplasia is an inherited condition both autosomal dominant and recessive modes. They have short stubby digits, enlargement of epiphysis, flaring of the diaphyseal ends deformity of knee, coxa vara and multiple joint osteoarthritis developing at an earlier age. It is due to defective ossification of epiphysis resulting in fragmentation of it [24,25]. Spondyloepiphyseal dysplasia, predominantly a short trunk variety even though the limbs are also affected. There is an autosomal dominant and X- linked recessive pattern of inheritance. It is characterised by short trunk, short and stubby fingers, vertebra plana, narrowing of intervertebral disc space with enlargement of posterior vertebral bodies, coxa valga and dysplasia acetabulum and in most cases odontoid hypoplasia [26-29]. Diaphyseal aclasis is an autosomal dominant condition with multiple Osteochondromas, bony deformities, short stature, trumpeting of the metaphysis. It is due to defective remodelling of bone [30,31]. Chondrodystrophia Calcificans Congenita or Conradi's syndrome is an autosomal recessive condition, where there is early fusion of epiphyseal centres resulting in short stature [32]. Ellis van creval syndrome is characterised by short stature with polydactyly, hypoplasia of hair, teeth and nail, short fingers sometimes associated with cardiac disease [33]. Diastrophic dwarfism is rare autosomal recessive disease. Here in most cases child may not walk. They have generalised ligamentous laxity, talipes equinovarus deformity, severe kyphoscoliosis, dislocation of hip, hitch- hiker thumb and cauliflower ears [34].

Mucopolysaccharidoses is a group of diseases due to lysosomal enzyme deficiency [35]. They are also known as chondro-osteo dystrophy or dysostosis multiplex. There are many variety of MPS depending on the enzyme deficiency. In general, they have microcephaly, thick calvarium, banana shaped ribs, wide clavicle, platyspondyly, dysplastic hip with coxa valga [36]. Generalized osteopenia and clouding of cornea are also seen. Morquio's, which is the commonest one, is characterised by platyspondyly, genu valgum, planovalgus, pectus carinatum, thoracolumbar kyphosis, cloudy cornea, hearing loss, odontoid hypoplasia. Platyspondyly is characterised by anterior breaking, limitation of extension of hip and knee with coxa vara [36,37]. Another MPS Hurler's syndrome is having scaphocephalic head, angular kyphosis, mental retardation and hepato-splenomegaly. Maroteaux-Lamy disease is having severe short stature and grotesque appearance [37].

Certain diseases which produce kyphoscoliosis of spine like poliomyelitis, tuberculosis of spine, congenital scoliosis, Scheuermann's disease, can produce short trunk.

Osteogenesis imperfecta characterised by fragile bones, dentigerous imperfecta and ligamentous laxity can produce short stature. It is associated with blue sclera, deafness in some cases. There can be history of multiple fractures, triangular facies, kyphoscoliosis and Wormian bones [38].

Endocrine diseases

Certain endocrine disorders, like hypopituitarism, hypothyroidism, precocious puberty, Cushing's syndrome, and pseudo hypoparathyroidism can produce short stature [39]. Laurence – Moon-Biedl syndrome with obesity, hypogonadism, polydactyly, mental retardation, retinitis pigmentosa is autosomal recessive condition with short stature [40]. In Laron type of short stature is due to growth hormone receptor defect. Frolich's syndrome or dystrophia adiposogenitalis is characterised by obesity, hypogonadism and normal intelligence is due to a hypothalamic or pituitary dysfunction [41]. Hypothyroidism especially thyroid agenesis causes short stature due to low levels of somatomedin [42]. In precocious puberty increased production of sex hormones leads accelerated growth but leads early fusion of epiphysis leading to short stature. Precocious puberty is more common in girls and majority of the cases are not pathological, whereas, in boys it is less common and more often pathological [43]. Polyostotic fibrous dysplasia with multiple lytic areas in long bones, café au lait spots can produce short stature due to precocious puberty [44]. Pseudopseudohypoparathyroidism is due to a receptor abnormality of PTH in renal tubule. They are usually mentally retarded, and have cataract, ectopic subcutaneous calcification and short fourth and fifth metacarpals [45]. Rickets of any cause can lead to stunted growth and short stature [46]. Insulin has a chondritic effect; hence juvenile diabetes mellitus is associated with growth retardation [47].

Congenital and peri-natal causes

Most preterm babies will be short when compared to normal ones. Those children with intrauterine growth retardation due to maternal or other causes can be of short stature. Russell-Silver syndrome with triangular facies, frontal bossing, micrognathia, low set ears, short 5th finger and loss of subcutaneous fat is associated with short stature [48]. Fetal alcohol syndrome will produce characteristic facies with short stature [49]. Progeria can have mental retardation, beak shaped nose, premature baldness, and loss of subcutaneous fat can lead to early closure of epiphysis between few months to few years leading to short stature [50]. Cornelia de Lange syndrome with hirsutism, bushy eye brows and short nose and mental retardation is also associated with short stature [51]. Prader Willi syndrome is characterised by obesity, hypogonadism, cryptorchidism, hypotonia and mental retardation [52].

There are certain races with short stature. African pygmies, Lappas and Gurkhas are short compared to the normal population.

The approach to a child with short stature is to get a thorough history- including prenatal, natal and postnatal history- Drug intake or substance abuse during pregnancy or intrauterine infections. Developmental history, history of infections or chronic infections, family history of any short stature can be obtained. In a proportionate dwarf, a hormonal work up can be done. Most of the syndromic short stature can be identified from head to foot examination. In disproportionate short stature we have to do a skeletal survey to find out the cause.

Conclusion

Short stature is complex area in the Paediatric Orthopaedics. Simple and thorough clinical examination with very little investigations can make the diagnosis easy.

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