



POSNA

The Core Curriculum

Short stature

Objectives

1. Define short stature
2. Discuss the etiology(ies) of short stature, and their relative frequency
3. Discuss an approach to evaluation of the child with short stature
4. Discuss problems associated with short stature

Discussion

It is sometimes difficult to ascertain exactly what is meant by short stature. Normal variant short stature (NVSS) is precisely defined as being characterized by a current height and adult height prediction < 3rd percentile, a birth weight > 2.5 Kg, no organic cause from growth retardation, and average peak serum growth hormone > 12ng/ml. If this definition is expanded to include all types of short stature, by definition 3% of the population is short statured. Absolute short stature has also been defined as being less than 60 inches tall at skeletal maturity for a male, and 58 inches for a female. About 40% of short-statured children in the general population have NVSS.

Short stature is generally subdivided in proportional and disproportional short stature. This subdivision is also somewhat subjective, as the line between normal and disproportional is not always clear. Practically, the subdivision is useful in the differential diagnosis of short stature. Disproportionate short stature is further subdivided into short-limb short stature (includes achondroplasia and other chondrodysplasias) and short-trunk dwarism (includes spondyloepiphyseal dysplasia, Morquio's, and metatropic dwarfism. The normal difference in body proportion with age must be considered. The infant's head is proportionally much larger than the adult's, and the limbs are shorter. At birth the ratio of sitting height to total height is about 70%, decreasing to 57% at age 3, and 52% at skeletal maturity.

Differential diagnosis includes environmental factors (nutritional disorders, irradiation, infection can all affect growth plate function, rickets and scurvy would be included in this category), and genetic disorders which affect protein synthesis. These could result in defective formation and/or function of chondrocytes (spondyloepiphyseal dysplasia, multiple epiphyseal dysplasia, diastrophic dysplasia), osteoblasts (osteogenesis imperfecta); or defects in calcification (hypophosphatasia), cell proliferation (achondroplasia), or bone remodelling (osteopetrosis).

Evaluation of the child with short stature includes documenting the family history, presence or absence of gastrointestinal, renal, or neurologic signs and symptoms. Menarchal status is helpful for the older girl. Documentation of height, weight, body proportion (sitting height, trunk-limb proportion), signs of secondary sexual characteristics (Tanner staging), and an exam of craniofacial

features. Serial documentation of height and weight are helpful, as children who fall proportionally lower in percentile with time are more likely to have a hormonal problem than the child who stays on the same percentile. Routine laboratory values would include CBC, urinalysis, glucose, creatinine, and alkaline phosphatase. Radiographs should include a wrist for bone age in the proportionate group, and a lateral of the spine, neck and skull, and an additional anteroposterior view of the pelvis in the disproportionate group. Consultation with an endocrinologist and/or dysmorphologist is generally a good idea.

More severe types of short stature result in functional problems in everyday life. The child may have difficulty navigating steps in school, desks, or playground equipment. Sinks, light switches, public transportation, and cabinets can all be hindrances for individuals with short stature.

References

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