Evaluation of the Child with Short Stature

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KEYWORDS

• Prenatal evaluation • Children • Short stature • Differential diagnosis • Physical examination

KEY POINTS

- Orthopedic surgeons frequently encounter short statured patients. A systematic approach is needed for proper evaluation of these children.
- The differential diagnosis includes both proportionate and disproportionate short stature types.
- A proper history and physical examination and judicious use of plain film radiography will establish the diagnosis in most cases.
- In addition to the orthopedic surgeon, most of these patients will also be evaluated by other specialists, including endocrinologists and geneticists.

INTRODUCTION

Normal growth and development usually proceed quite smoothly from single-celled zygote all the way to an approximately one hundred trillion–celled adult human being.[1] Three discernable growth spurts occur, with the first being intrauterine (and arguably the most dramatic), the second involving the first 2 years of life (with about a 100% increase in size of the child), and the third is the adolescent (also known as pubescent) growth spurt. The amazing thing may not be that growth aberrations occur, but that they do not occur more frequently. When a child falls 2 standard deviations or more below the average height for age, sex, and ethnic group established norms, they are considered to have short stature.[2]

Short statured humans have been well-recognized by others within society all the way back to antiquity. For instance, the ancient Egyptian sarcophagus carving of Djeho offers a detailed picture of a prominent citizen with achondroplasia[3] (**Fig. 1**). Djeho lived around 360 BC and worked with the chief financial officer of Upper Egypt. There also were powerful short statured Egyptian gods with the god Bes (god of music and warfare) and the god Ptah (a god of creation and master architect of the universe) serving as excellent examples.[4]

In 1951 when Sir Harold Arthur Thomas Fairbank (at 75 years of age) published his classic *Atlas of General Affections of the Skeleton*, he helped bring order to the chaos of this complex mixture of musculoskeletal entities. This article focuses on important orthopedic aspects in the evaluation of short stature in children with a particular focus on skeletal dysplasia. It does not focus on each and every diagnostic entity, but rather the options that must be considered and the process one may undertake to arrive at the most precise diagnosis.

DIFFERENTIAL DIAGNOSIS

The differential diagnosis for short stature is exhaustive and includes what have been referred to as proportionate and disproportionate types.[5–8] Comprehensive evaluation often includes referrals to pediatric subspecialists like

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Fig. 1. Sarcophagus carving of Djeho illustrating typical features of achondroplasia.

endocrinology and genetics.[9] Proportionate short stature may be owing to familial short stature, intrauterine growth retardation (commonly owing to smoking), constitutional delay of growth, occult medical diseases (including endocrinopathy), and idiopathic short stature.[10,11] Multiple evaluations over time may be quite valuable, because skeletal dysplasia may be later confirmed in up to 20% of patients previously labeled as idiopathic short stature or small for gestational age.[12] This paper does not focus on these proportionate short stature types because it is extremely rare for such patients to present undiagnosed to the orthopedic surgeon.

Disproportionate short stature relates to an improper balance between standing height and sitting height. In normal populations, the sitting height/standing height ratio has been shown to be approximately 0.7 at birth and closer to 0.5 at skeletal maturity.[13] Standing height has contributions from both limb length as well as trunk length, whereas sitting height is effectively all about trunk length. One broad (and imperfect) generalization is that disproportionate short stature can be divided into those characterized mainly by shortened limbs and those mainly characterized by a shortened trunk. In the past, these have been referred to as short limb dwarfism and short trunk dwarfism.[14] **Fig. 2** illustrates the striking contrast that can be seen when assessing standing height and sitting height. The sitting height/standing height ratio has been shown to be of significant clinical value.[15,16] In addition to many other more sophisticated methods, the relative femoral and tibial contributions to lower limb length discrepancy can be determined by instantaneous limb length assessment (**Fig. 3**).[17]

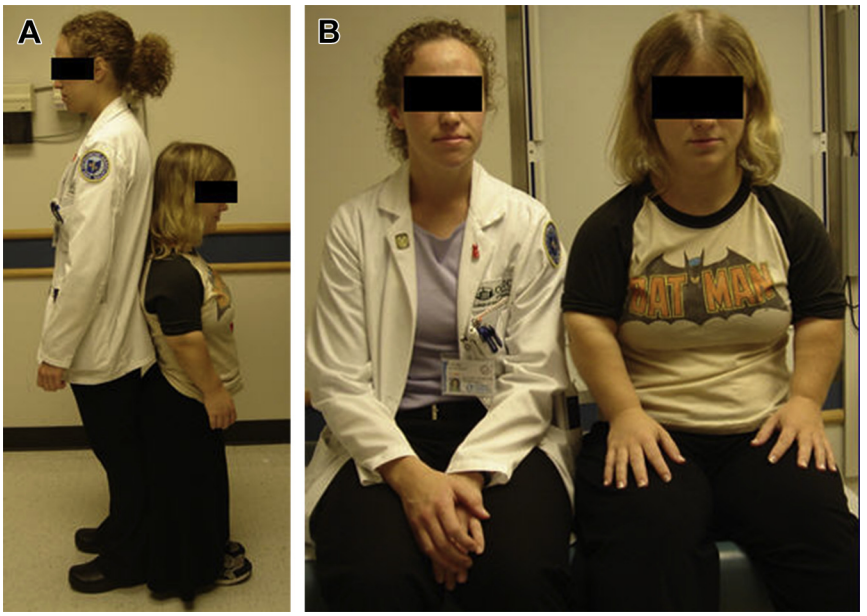

Fig. 2. Standing height versus sitting height. (*A*) A normal statured female standing back to back with a female with skeletal dysplasia. This illustrates a significant difference in standing height. (*B*) The same normal statured female and female with skeletal dysplasia sitting side by side, illustrating nearly identical sitting height.