Clinical Dilemmas in Evaluating the Short Child

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Abstract

Concerns about a child’s growth are one of the most common topics parents voice during general pediatric office visits and are a leading cause for referral to a pediatric endocrinologist. There are a variety of conditions that lead to short stature in children; however, in the absence of true pathology, idiopathic short stature and constitutional delay are the most frequent causes. This article reviews the general approach to evaluating the short child and clinical signs that should prompt further evaluation and referral. We also address the unique psychological issues that these children face and approaches to counseling families with a child with idiopathic short stature. [Pediatr Ann. 2014;43(8):321-327.]

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Children’s growth patterns are one of the most important aspects of the “well child visit” with general pediatricians, and concerns about growth problems are one of the most common reasons for referrals to pediatric endocrinologists. There is a broad differential of conditions associated with short stature; however, most causes can be excluded with a thorough history, physical examination, and basic screening tests. Overall, most instances of short stature in children are due to constitutional delay of growth, with idiopathic short stature a diagnosis of exclusion. Pediatricians face unique challenges in counseling families of children with short stature, particularly those who experience psychological difficulties and bullying. In this article, we review the general approach to evaluating a short child and discuss the psychological implications of short stature, including methods to counsel families on the natural course of the diagnosis. Our objectives are to (1) discuss the most important components of the history and physical examination when evaluating the short child; (2) review the most valuable laboratory and radiological studies used in determining the cause of short stature; (3) discuss signs and symptoms that indicate pathology and the need for specialty referral in evaluating the short child; (4) describe the unique psychological issues that children with short stature face; and (5) provide resources and approaches to counseling the child and parent on idiopathic short stature.

INITIAL EVALUATION
Medical History

The evaluation of a child with short stature begins with a thorough medical history, including a comprehensive family history and an inquiry into social aspects that can be potential causes of short stature (Table 1). Obtaining a detailed birth history that includes birth weight and length, any perinatal complications, and any maternal health problems during the pregnancy can provide important clues to the etiology of short stature. Children born premature often have intrauterine growth issues and nutritional challenges after birth that may affect future growth. Children born below the 10th percentile for age are small for gestational age (SGA) and can have poor linear growth early on; fortunately, approximately 90% achieve catch-up growth by age 2 years. In addition to illneses, the family history should include both the mother’s and father’s height and timing of puberty. Children of parents who had late onset of puberty are likely to have similar constitutional delays in growth and development. Calculating the target height (Figure 1) enables the physician to determine if the patient is achieving his or her genetic potential for growth. Furthermore, height and growth problems in siblings can provide clues to constitutional delay in families or other genetic syndromes.

Table 1

<table>
<thead>
<tr>
<th>Area Assessed</th>
<th>Important Factors</th>
<th>Indicators of Possible Pathology</th>
</tr>
</thead>
<tbody>
<tr>
<td>Birth history</td>
<td>Weight, length, complications, maternal health problems</td>
<td>SGA, IUGR, low birth weight, maternal illness</td>
</tr>
<tr>
<td>Medical problems</td>
<td>Infections, procedures, diagnoses</td>
<td>Frequent respiratory or gastrointestinal infections, chronic diarrhea or abdominal pain, history of significant trauma</td>
</tr>
<tr>
<td>Family history</td>
<td>Maternal and paternal height, timing of puberty, major illnesses</td>
<td>Consanguinity, familial congenital anomalies</td>
</tr>
<tr>
<td>Developmental history</td>
<td>Major milestones, learning difficulties, age of first tooth eruption</td>
<td>Developmental delays, learning disabilities, delayed tooth eruption</td>
</tr>
<tr>
<td>Medications</td>
<td>Stimulants, steroids, psychiatric medications</td>
<td>Frequent glucocorticoid use, prolonged stimulant use</td>
</tr>
<tr>
<td>Puberty</td>
<td>Age at onset of breast or genital development, presence of pubic and axillary hair, age at menarche</td>
<td>Early menarche, delayed breast development in females or delayed testicular enlargement in males</td>
</tr>
<tr>
<td>Dietary history</td>
<td>Appetite, portions, food allergies</td>
<td>Poor appetite, food intolerance, poor nutritional intake</td>
</tr>
<tr>
<td>Social history</td>
<td>Living environment, access to nutrition and health care</td>
<td>History of neglect or abuse, significant family stressors</td>
</tr>
<tr>
<td>Review of systems</td>
<td>Comprehensive with particular attention to gastrointestinal and neurological symptoms</td>
<td>Diarrhea, constipation, recurrent abdominal pain, headaches, vision changes, polyuria, polydipsia, weight loss, skin or hair changes, temperature intolerance</td>
</tr>
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IUGR = intrauterine growth restriction; SGA = small for gestational age.

Figure 1. Method for calculating the parental target height.

Girl

\[
\text{Target height (cm)} = \text{Mother's height (cm)} + \text{Father's height (cm)} - 13 \text{ cm}
\]

Boy

\[
\text{Target height (cm)} = \text{Mother's height (cm)} + \text{Father's height (cm)} + 13 \text{ cm}
\]
that are associated with short stature.

Major illnesses or trauma can also affect a child’s linear growth. Parents should be asked about any frequent infections, hospital admissions, or surgical procedures. These questions can provide important clues about underlying diagnoses or chronic illnesses, such as renal disease, that may affect linear growth. For example, a child with undiagnosed cystic fibrosis may present with poor growth and a history of recurrent respiratory infections. Similarly, a child with celiac disease or inflammatory bowel disease may present with poor linear growth and frequent abdominal complaints. Children who are taking medications for chronic illnesses may also experience growth effects due to the medication. Current or prior medication use should thus always be investigated, particularly for agents that effect weight gain or metabolism. For example, some children with attention-deficit/hyperactivity disorder treated with stimulant medications have been shown to have poor weight gain and suboptimal growth; moreover, slow linear growth may also occur in those with autoimmune diseases or inflammatory conditions (such as asthma) that require frequent treatment with systemic glucocorticoids.

A child’s developmental history, including achievement of major milestones, should also be obtained. Specifically, the age at which a child had their first tooth eruption and loss is valuable information that can help determine elements of constitutional delay. Children with delayed tooth eruption often have skeletal immaturity that is seen in constitutional delay of growth.

Physical Examination

A thorough physical examination specifically looking for features of genetic syndromes that cause short stature should be performed (Table 2). The examiner should pay particular attention to facial appearance, head shape, palate shape, and ear and eye placement. A thorough skin examination should also be performed, as several genetic syndromes associated with short stature can feature distinct birth marks. Chest and genitourinary examinations should be completed to determine pubertal development, especially because delayed pubertal development can be associated with pituitary hormone deficiencies or genetic syndromes. The extremities should be inspected for abnormalities in shape, size, and movement, including the carrying angle of the upper extremities and genu varum or valgus of the lower extremities. A general examination of the heart, lungs, and abdomen should also be completed.

Evaluating the Growth Curve

Changes in growth velocity can be seen when reviewing a child’s growth chart. Accurate height measurements should be plotted at all visits to observe and calculate a height velocity. A child’s height should be measured using a stadiometer and proper technique; this includes measuring a recumbent length for children younger than age 2 years using a fixed headboard and movable footboard and standing height for children older than 2 years. Often the patient’s height is measured inaccurately, which can falsely appear as short stature when viewed on the growth chart. In fact, it has been estimated that only 30% of children seen in primary care offices are measured using the proper technique, and that on average, there is a 2.2-cm difference in measured length between proper stadiometer use and simple tape measure use. Short stature due to poor nutrition or gastrointestinal illnesses typically demonstrate a decrease

<table>
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<tr>
<th>TABLE 2. Physical Examination of a Child with Short Stature</th>
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<tr>
<td><strong>Area Assessed</strong></td>
</tr>
<tr>
<td>Head, ears, eyes, nose, and throat</td>
</tr>
<tr>
<td>Neck</td>
</tr>
<tr>
<td>Cardiovascular</td>
</tr>
<tr>
<td>Chest, back, and lungs</td>
</tr>
<tr>
<td>Abdomen</td>
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<tr>
<td>Puberty</td>
</tr>
<tr>
<td>Extremities</td>
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<tr>
<td>Neurologic</td>
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<tr>
<td>Skin</td>
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in weight gain followed by a decrease in height velocity (Figure 2). In contrast, the typical growth pattern of a child with an endocrinopathy causing short stature, such as hypothyroidism or growth hormone deficiency, reveals a decrease in linear growth velocity with normal or increased weight (Figure 3).

Plotting the parental target height on the growth curve can allow the physician to visualize if the child is growing along a curve consistent with their genetic potential, which is seen in familial short stature (Figure 4). Alternatively, children with constitutional delay will have a normal growth velocity, on average about 5 cm per year in a school-aged child, but will have a delayed bone age and be growing along a curve below their parental target height until they achieve their pubertal growth spurt (Figure 5).

**LABORATORY STUDIES AND IMAGING**

Laboratory studies and imaging should be used judiciously, and the work-up should be tailored to the level of clinical suspicion. If the history, growth chart, and physical examination are consistent with constitutional delay, then a simple bone age X-ray may be helpful and no other investigation needed. In this case, the bone age would be expected to be “delayed,” indicating skeletal immaturity. Should the physical examination findings show dysmorphic features or signs of a genetic syndrome, then the appropriate genetic tests should be ordered and referrals made. In the case of a child with failure to thrive or weight loss in addition to short stature, celiac disease and systemic illness such as renal tubular acidosis and anemia should be evaluated by obtaining a complete blood count, comprehensive metabolic profile, and total immunoglobulin A and tissue transglutaminase or endomysial antibody levels. A history consistent with poor weight gain, short stature, and respiratory infections should prompt an evaluation for cystic fibrosis with a sweat chloride test or genetic studies.

If an endocrine disorder is suspected, it is appropriate to evaluate the different hormonal axes that can cause poor growth. This includes evaluating for hypothyroidism and growth hormone deficiency by measuring levels of thyroid-stimulating hormone, free thyroxine (T4), and insulin-like growth factor-1 (IGF-1) and its binding protein, IGFBP-3. Obtaining a serum growth hormone level is not useful due to the varying levels in the body throughout the
day. Moreover, short females should have a karyotype performed to rule out Turner syndrome, as short stature may be the only physical manifestation of this disorder.

**NEED FOR SUBSPECIALTY REFERRAL**

In cases of familial short stature or constitutional delay, further evaluation and referral is generally not indicated. However, should any of the historical or physical examination findings be concerning for pathology, then referral to the appropriate subspecialist is warranted. For example, the child with suspected celiac disease should be referred to a pediatric gastroenterologist for further evaluation. Moreover, a child with dysmorphic features should be evaluated by a geneticist. Several syndromes qualify for growth hormone treatment (Table 3), and patients with these disorders should be evaluated by an endocrinologist. Short children with mid-line defects such as a single central incisor or cleft palate are at risk for growth hormone deficiency and should also be evaluated by an endocrinologist. Moreover, pediatricians and general practitioners should be concerned and consider referral when any child has a decline in growth velocity with no apparent clinical cause.

**PSYCHOLOGICAL ISSUES**

Reports of bullying and poor outcomes in children due to teasing and psychological stress are common in the media and increasing in frequency. Children who are bullied have been shown to have higher rates of depression, anxiety, school absenteeism, and suicidal ideation; yet, only about one-third of victimized children report the bullying to parents or teachers. Fear of these negative psychological effects often prompts parents to discuss growth concerns with the pediatrician. However, the literature regarding the psychosocial functioning of children with short stature provides varied results. Most studies that address the psychological effects of short stature in children review only parental reports of stress and adjustment problems in the child. However, one study examining student responses found that short children are more likely to be bullied and also feel a greater degree of social isolation when compared with their taller peers, and another found that the frequency of bullying was negatively correlated with the height of the child, indicating that the
shorter the child, the more likely they are to be teased. Risk factors associated with being teased for short stature include male gender, low socioeconomic status, low intelligence, and having a younger but taller sibling. It is clear that significant parental and child anxiety is related to the diagnosis of short stature; however, more recent reviews of the literature show that in general, short children are well adjusted and experience no long-term consequences associated with their stature. In the setting of short stature, parents also tend to rate their children’s well-being lower than do the patients themselves. Thus, both the parent and child should be asked about any psychosocial difficulties including bullying, teasing, abuse, anxiety, or reduced self-esteem.

It is important for pediatricians to evaluate the psychological impact of short stature on their patients. We recommend that pediatricians routinely discuss bullying with their patients and parents and appropriately screen for anxiety and depression in these patients. Should parents report significant social problems or depression, then the child should be evaluated for mental health treatment.

COUNSELING PARENTS

Once the medical evaluation is complete and any pathology excluded as the cause of short stature, efforts to counsel the parents become the focus of the visit. Many parents will inquire about the use of medication, namely growth hormone, as a treatment for their child’s idiopathic short stature, particularly if the child is experiencing negative psychosocial effects from low self-esteem or bullying. Often, the expectation is that achieving a taller stature will improve self-esteem, confidence, and even behavioral difficulties; however, this is not always the case. On average, treating children who have idiopathic short stature with growth hormone has not been shown to improve psychosocial functioning.

The psychosocial impact of short stature is a complex interplay of the child’s functioning, temperament, cultural stereotypes, and adaptation to stress. The coping strategies of the child and family affect how the child emotionally deals with any teasing or bullying he or she may experience. Assessment of other stressors on the family and child, such as financial problems, major life events, or learning difficulties can help determine to what extent further emotional stress will impact the child. When counseling parents about their child’s growth, determining if there are other significant social stressors will help the physician decide what additional support, if any, is needed. Involving social workers, school counselors, and mental health professionals may be beneficial if the family is experiencing significant stress that is exacerbated by psychosocial problems the child is facing due to their short stature.

Importantly, a realistic discussion of the parent’s and child’s expectations
regarding their height should be undertaken during the counseling portion of the visit. We see that children often have unrealistic expectations of achieving tall stature and that parents will also overestimate their stature. A discussion of growth patterns and final height can help parents and children develop more realistic expectations about final height. It is important to emphasize that short stature is not an illness or a medical problem itself. Encouraging parents to focus on the child’s attributes and talents, not just their height, will also help children focus on their positive traits rather than their stature.

Table 4 lists resources that are available to parents and patients who are struggling with bullying and for physicians who are counseling these families. It also lists resources for parents regarding growth and idiopathic short stature to assist in their understanding of the diagnosis.

CONCLUSION

After a thorough history, physical examination, and basic laboratory evaluation, most pathologic causes of short stature can be excluded in the general pediatrician’s office. Evaluating the patient’s growth chart and growth velocity is paramount to making the correct diagnosis. Concerning physical examination findings and changes in growth velocity should prompt a referral to an endocrinologist. When pathology is excluded and the healthy child continues to be short for familial expectations, counseling on the course of idiopathic short stature should be performed. Although children with short stature are at greater risk of being bullied and experiencing psychological difficulties, they function well overall when compared with their peers. There are also several resources available to counsel both patients and their parents when psychological stress is a problem.

REFERENCES


