

PedsCases Podcast Scripts

This is a text version of a podcast from PedsCases.com on "Short Stature." These podcasts are designed to give medical students an overview of key topics in pediatrics. The audio versions are accessible on iTunes or at www.pedcases.com/podcasts.

Short Stature

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Learning Objectives

After listening to this podcast, the learner will be able to:

1. Define short stature.
2. Define a normal childhood growth pattern.
3. Develop an approach to the history and physical examination of a child with short stature.
4. List common investigations, with a basic understanding of their usefulness.
5. Briefly describe some specific causes of short stature including constitutional delay of growth and puberty, familial short stature, endocrine diseases, chromosomal abnormalities or syndromes, skeletal dysplasias and chronic diseases or malnutrition.

Clinical Scenario

A 12 year-old girl presents to the Pediatric Clinic concerned about her short height. She has always been short but lately she has noticed more of a difference between herself and her peers. Her height was at the 50th percentile at birth, the 10th percentile between 2 to 5 years of age, and then dropped below the 3rd percentile after 5 years of age. Her weight has always plotted on the 25th percentile. She has a history of frequent urinary tract infection and otitis media. School is okay but she doesn't like math. She is an otherwise well, active, and bright young girl.

Her mother is 5 feet and 6 inches and her father is 5 feet and 9 inches.

The patient's weight is 36 kg and height is 129.1 cm.

What is the approach to assessing this girl's short stature?

Definition of Short Stature

Short stature is defined differently in different sources. However, generally it is defined as a height that is two standard deviations below the mean, or less than the 5th percentile. A decreased growth velocity, at less than the 5th percentile on the growth velocity chart, is more concerning than an absolute height measurement. Growth velocity can be determined by plotting repeated heights on a growth chart and noting crossing of percentiles or by plotting the velocity on a growth velocity growth chart. Proper measuring and plotting is essential. Inaccurate measuring and plotting is a frequent reason for referral for short stature. It should also be remembered that by definition, 5 percent of normal children will plot below the 5th percentile.

Inadequate weight gain is termed failure to thrive and is covered in another podcast.

A normal growth pattern entails rapid growth between birth and 2 years of age, followed by slower childhood or pre-pubertal growth of about 5cm per year, and then rapid growth again in puberty. It is normal for children to cross height percentiles in the first two years of life as birth size is dependent on maternal and placental factors more than a child's actual height potential.

Approach to a Child with Short Stature

Differential Diagnosis

The differential diagnosis for a child with short stature can be broken down as follows.

Normal Variants which include

- i. Constitutional Delay of Growth and Puberty and
- ii. Familial Short Stature

Endocrine Diseases which include

- i. GH Deficiency
- ii. Hypothyroidism and
- iii. Cushing's Syndrome

Chromosomal Abnormalities or Syndromes which include, for example

- i. Turner Syndrome
- ii. Russell Silver Syndrome and
- iii. Noonan Syndrome

Skeletal dysplasias of the long bones or spine.

Chronic disease or malnutrition which can include

- i. Gastrointestinal disease such as celiac disease, or inflammatory bowel disease
- ii. Renal disease such as renal tubular acidosis, or renal failure and
- iii. Congenital heart disease

Key Points on History

Growth velocity is the most important piece of information. Therefore, the growth rate and the onset of the short stature should be determined on history. For example, one

may ask if the patient has always been short or if there has been a rapid decrease in growth recently. This can be asked in various ways including asking about the need to change shoe or clothing sizes, or the relative height differences noted between peers or siblings.

The birth history should include birth weight and length and gestation. This will elicit whether the patient was intrauterine growth restricted, small for gestational age, or premature all of which may lead to short stature. One should also ask about the presence of neonatal hypoglycaemia and prolonged jaundice both of which would make one more suspicious of congenital growth hormone deficiency. Pregnancy issues including maternal illness, drugs, smoking, and nutrition should also be determined.

Developmental history should include queries about developmental milestones, learning difficulties, and tooth eruption and loss.

A dietary history is also helpful.

Special attention should be made to the social history including indications of deprivation, and teasing at school.

Important aspects of the past medical history include the presence of chronic disease, radiation, head trauma, or major illnesses. A pubertal onset history is vital. The presence of frequent otitis media or urinary tract infections may make one suspicious of Turner Syndrome in a short female. A medication history focusing on medications known to affect growth such as corticosteroids or stimulants is important.

The parents', siblings', other family members' heights and histories of puberty onset are important aspects of the family history. The target height or mid parental height should be calculated by the formula of fathers height in centimeters plus mother's height in centimeters plus 13 all divided by 2 for boys, or fathers height in centimeters plus mother's height in centimeters minus 13 all divided by 2 for girls. Normal children, with for example familial short stature, should obtain a final height within 10 centimeters of this target height.

Finally a review of symptoms should include an assessment for systemic diseases such as gastrointestinal, cardiac, or renal diseases that can result in short stature; hypothyroid symptoms such as cold intolerance, constipation, and fatigue; and central neurological symptoms such as headaches and vision changes that would be suspicious of a tumor.

Key Points on Physical Exam

A full physical exam is essential but there are some specific key points that should be emphasized. Weight and height measurements should be done on an accurate

scale and stadiometer and then plotted on a growth chart. One should also measure the upper to lower segment ratio and arm span.

Attention should be given to possible dysmorphic features including irregular eye shape, low set ears, high arched palate, wide space nipples, and short 4th or 5th metacarpals.

Assessment of pubertal development with Tanner staging is essential.

Investigations

Investigations should be based on the clinical assessment and differential diagnosis.

Often, no investigations are required if one is for example considering a diagnosis of familial short stature.

A child with the likely diagnosis of constitutional delay of growth and puberty should have a bone age x-ray for further confirmation.

In a short female, a karyotype to rule-out Turner Syndrome should be strongly considered regardless of the presence or absence of other Turner stigmata, especially if she is pre-pubertal.

Children with a presentation suspicious of growth hormone deficiency, or other endocrine disease should have their appropriate hormone levels measured. This may include screening baseline levels with insulin-like-growth-factor-I and BP3 as surrogates of growth hormone, free T4, TSH, cortisol, ACTH, LH, FSH, and testosterone or estradiol. More sensitive and specific stimulation testing under the guidance of an Endocrinologist should be considered if isolated growth hormone deficiency or hypopituitarism is a possibility.

X-rays to rule out skeletal dysplasia, and other blood work such as a celiac screen may be beneficial depending on the clinical suspicion.

Regardless of the investigations, the most important follow up plan should include carefully following the height and growth velocity. This is usually done in 6 month intervals and can be of great assistance in focusing the diagnosis.

Brief Description of Specific Causes of Short Stature

Certain findings on the history, physical exam, or investigations may help in focusing in on the possible diagnosis of a child with short stature.

A child with **Constitutional Delay of Growth and Puberty** should have a history of a normal pre-pubertal growth velocity, late onset of puberty, and delayed bone age.

There is often a similar history in other family members. These children should achieve normal final height without intervention.

A child with **Familial Short Stature** should have a normal pre-pubertal and pubertal growth velocity, short parents, normal timing of pubertal onset, and normal bone age. The final adult height should be within the expected range of the mid-parental or target height without intervention.

A child with short stature secondary to an **Endocrine Disease** tends to be short, with an increased weight for height ratio. That is to say they are short and "chubby". Growth hormone deficiency can be congenital or acquired, and may be associated with other pituitary hormone deficiencies. Congenital growth hormone deficiency is often associated with other midline anomalies such as cleft lip or palate, or micropenis. It is also often associated with hypoglycaemia and prolonged jaundice in the neonatal period. Growth deceleration usually does not occur until after 1 year of age. Acquired growth hormone deficiency may be idiopathic in etiology, but may also be due to a brain tumour, severe head injury, CNS radiation, CNS infection, or CNS surgery. Clinically there is a deceleration of the growth velocity in a child with a young facial appearance. Untreated primary hypothyroidism may lead to short stature but there are usually other thyroid symptoms present also. Glucocorticoid excess or Cushing's syndrome can lead to decreased growth velocity and excess weight gain. In children, the etiology is usually exogenous corticosteroids, such as high dose corticosteroids in an asthmatic, and not endogenous Cushing's disease.

The possibility of a **Chromosomal Abnormality or Syndrome** should also be considered in a child with short stature. The consideration of Turner Syndrome is very important in any short female, especially with pubertal delay, and should not be missed. These girls may have other dysmorphic features of Turner Syndrome but alternatively short stature may be the only feature. Girls with Turner Syndrome have delayed puberty due to primary ovarian failure.

Skeletal dysplasias lead to short stature due to shortening of the long bones or spine and therefore result in disproportionate short stature when measuring the upper and lower segments. These dysplasias can often be suspected following a careful physical exam.

Chronic disease or malnutrition can lead to short stature but usually these children have significant poor weight gain as well. With this clinical scenario one should consider the possibility of conditions such as celiac disease, inflammatory bowel disease, renal disease, and congenital heart disease.

Summary and Take-home Points

In summary, short stature is common in children but true pathological short stature is rare. Familial short stature and constitutional delay of growth and puberty are the

most common etiologies of short stature yet, a high clinical suspicion is required to not miss true pathology, such as growth hormone deficiency. The most important aspect of assessing a child with short stature is a careful evaluation of their growth velocity. A child with true pathology will have a decreased growth velocity evident by crossing of percentiles on a growth curve. A key diagnostic point is not to miss the diagnosis of Turner syndrome in a short female. Finally, slow growing but thin children are more likely to have a chronic disease, such as celiac disease, as an etiology of their short stature rather than an Endocrine etiology such as growth hormone deficiency.

Back to the Case

The girl mentioned in the case at the start of this podcast was found to have significantly elevated levels of LH and FSH, low estradiol and a 45XO karyotype. A diagnosis of Turner Syndrome was made as the cause of her short stature.

References

References available upon request.