Short Stature

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Short stature is one of the common referral to a paediatric endocrine unit. It is important to note that short stature is not a diagnosis by itself but a manifestation of many physiological and pathological causes. The term 'dwarfism' is no longer used for short stature. Apart from being a matter of social concern, it could also be a clinical indicator of an underlying pathology. Hence, it is important to distinguish pathological from the non-pathological forms of short stature.

Definition : Short stature or growth failure is considered when

- Height 2 SD or more below the mean height for that gender and chronological age in a given population, or
- Height more than 1.5 SD below the mid-parental height , or
- Fall in growth velocity below the 25th percentile.

Severe short stature is defined as height 3SD below the mean height for that gender, age and population. Severe short stature is almost always associated with a pathological cause which needs evaluation.

Factors influencing growth in different periods of life (Karlberg model)[1]

1. During Infancy: nutrition
2. During Childhood : growth hormone and thyroid hormones
3. During Puberty : sex hormones

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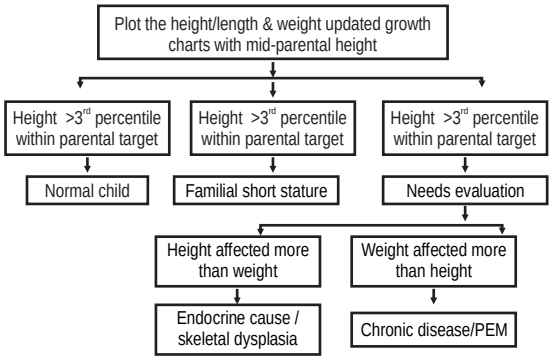
4. Causes of short stature

Dysmorphism	No Dysmorphism	
	Proportionate	Disproportionate
1. Turner Syndrome	1. Constitutional delay	1.Skeletal Dysplasias
2. Prader - Willi Syndrome	2. Familial	2.Metabolic Disorder
3. Noonan Syndrome	3. Chronic Disease/ PEM 3.OI	
4. Down's syndrome	4. SGA	
	5. Hypothyroidism	
	6. GH deficiency	

Initial assessment of a child referred with Short Stature



Pointers in history

1. Low birth weight : small for gestational age
2. Breech delivery: GH deficiency
3. Neonatal jaundice : hypothyroidism (indirect) ; cortisol deficiency(direct)
4. Oedema of hands / feet during neonatal period: Turner syndrome
5. Feeding difficulties, snoring, hyperphagia : Prader-Willi Syndrome
6. Persistent diarrhoea: Coeliac Disease
7. Constipation : Hypothyroidism
8) Polyuria / polydipsia: Diabetes Mellitus , Diabetes Insipidus
9. Headache, vomiting, visual disturbances : intracranial tumours

10. Family History of short stature : FSS, GHD, Skeletal dysplasia
11. Consanguinity : genetic causes
12. Family history of delayed puberty : constitutional delay in growth

Pointers in examination

1. Frontal bossing, micropenis, crowded frontal teeth, midfacial hypoplasia: Growth Hormone Deficiency
2. Pallor : renal failure, undernutrition, Coeliac disease
3. Obesity : Hypothyroidism, Cushing's syndrome, Prader-Willi syndrome
4. Metacarpal shortening : Turner syndrome, SHOX deficiency
5. Mentally subnormal : Down's syndrome, hypothyroidism

Clinical Assessment of child with short stature

1. Accurate weight & height measurement (< 2 yrs- infantometer > 2yrs stadiometer)
2. Upper segment, Lower segment measurement; Arm span
3. Comparison with his / her own genetic potential : Mid-Parental Height[2]
   MPH for boys = (mother's ht + father's ht) / 2 + 6.5
   MPH for girls = (mother's ht + father's ht) / 2 - 6.5
   The target range is 1.5 SD above and below this target.
4. Sexual Maturity Rating : using Tanner staging

Investigations

The investigations required to reach a diagnosis could be exhaustive. Hence, these are considered level-wise as not all are warranted at once.

Level 1

1. CBC, LFT, RFT
2. Stool (parasites, occult blood, steatorrhoea)
3. Calcium profile (Ca, Phosphorus, Alkaline phosphatase, PTH)
4. Thyroid function test
5. Bone age (read using Greulich Pyle atlas / Tanner Whitehouse method)
6. Karyotype in female child to rule out Turner syndrome

All the tests showing normal results, the patient should be followed up after 6 months and a growth chart following the same curve or a static height should be considered for further evaluation.

Level 2 (best left to the specialist as interpretation is difficult)

1. IGFBP-3 (in < 5 yrs) ; IGF-1 (in > 5yrs)
2. Anti-transglutaminase / anti-endomysial(more specific) antibodies[3]
3. MRI Brain (special reference to pituitary)[4,5]

Level 3

1. GH stimulation Test
2. Duodenal Biopsy

Management

Depends upon the cause of short stature: e.g.

1. Counselling of parents (for physiological causes)
2. Dietary advice (Undernutrition, Coeliac disease)
3. Levothyroxine (Hypothyroidism)
4. Limb lengthening procedure (skeletal dysplasia)
5. Growth Hormone

Growth Hormone Therapy : The main goal of initiating growth hormone therapy is to allow the child to achieve his / her maximum genetic potential and hence ensure physical and psychological well being of the child. It needs to be

administered as a subcutaneous injection every night. Apart from the 10 approved indications GH therapy by FDA (GH deficiency, Turner syndrome, Noonan syndrome, short bowel syndrome, acquired immunodeficiency syndrome, SGA, chronic renal failure, Prader-Willi syndrome, SHOX gene defects, idiopathic short stature)[6] there are many evolving indications for GH therapy. Growth hormone therapy is an expensive therapy and hence thorough investigations and family evaluation is necessary before the treatment is initiated. GH is generally well tolerated medication and adverse effects are rare and mild. Serious side effects such as worsening of scoliosis, slipped capital femoral epiphyses and benign intracranial hypertension are rarely seen. Growth hormone does not increase the risk of recurrence of malignancy or a second malignancy and is used in cancer survivors or intracranial tumour survivors with growth failure due to growth hormone deficiency.

Key Points

1. Plotting the child's height and weight along with the mid-parental height on a growth chart should be the first step.[7]
2. Serial readings are more conclusive than a single reading of height.
3. Pubertal status must be considered while evaluating the child.
4. Bone age gives information regarding the potential time left for height gain.[8]
5. Hormonal studies should only be undertaken after ruling out the Level 1 investigations

Abbreviations : SD- standard deviation, GH- growth hormone, SGA- small for gestational age, PEM- protein energy malnutrition, OI- Osteogenesis Imperfecta, CBC - complete blood count, LFT- liver function test, RFT-renal function test, PTH-parathyroid hormone, Ca- calcium, P- phosphorus, ALP- alkaline phosphatase, IGFBP-3 - insulin like growth factor binding protein 3, IGF- insulin like growth factor

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