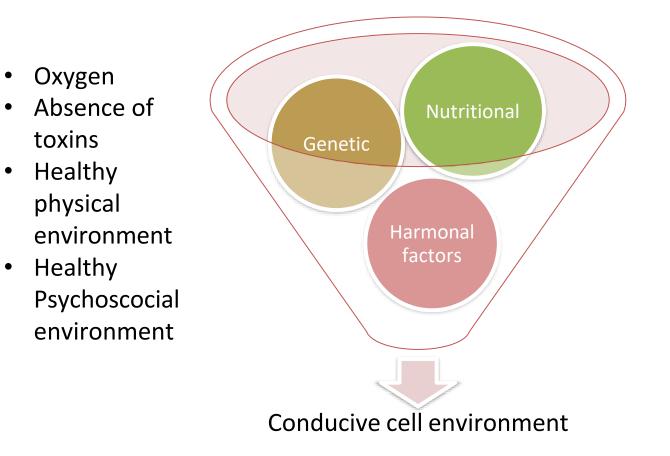
## Short stature

- Normal growth physiology
- Growth monitoring and growth charts
- When to evaluate/refer
- Short stature vs growth failure
- Major phenotypes
- How to evaluate
- Management
- Points to ponder

## Normal growth



•

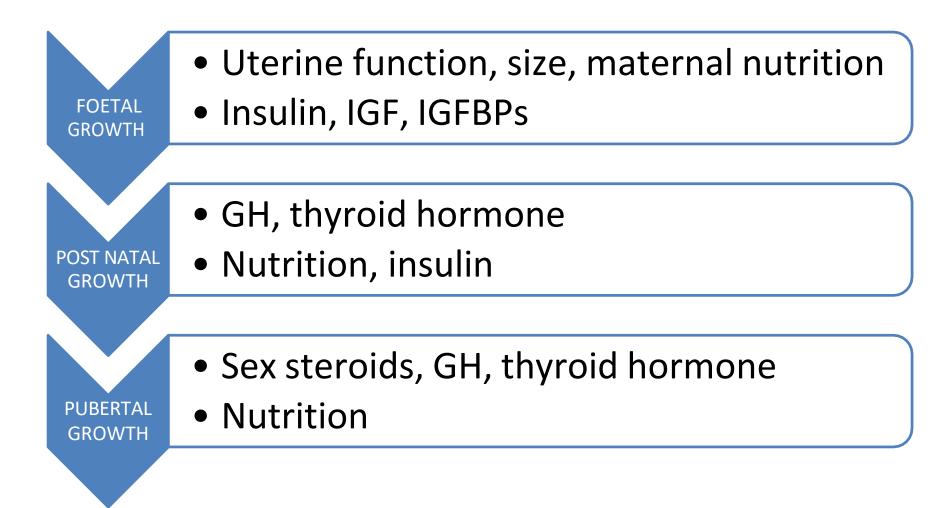
### Age

### Growth velocity per year

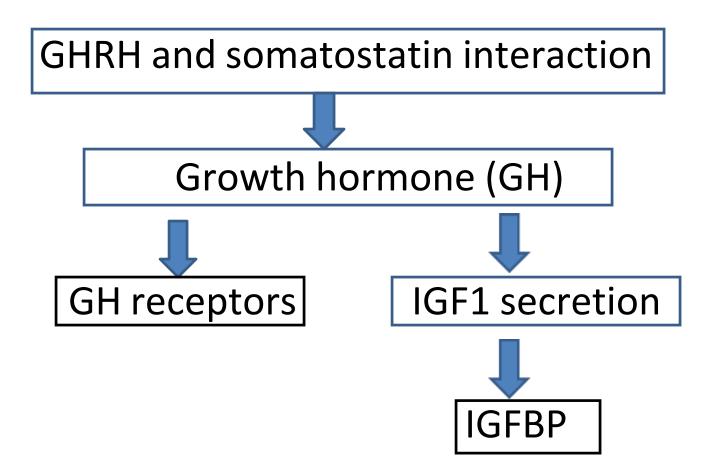
Birth to 12 months 12 months to 1 year 2 to 3 years 3 to 5 years 5 years to puberty Puberty

23 to 27 cm (9.06 to 10.63 in) 10 to 14 cm (3.94 to 5.51 in) 8 cm (3.15 in) 7 cm (2.76 in) 5 to 6 cm (1.97 to 2.36 in) Girls: 8 to 12 cm (3.15 to 4.72 in) Boys: 10 to 14 cm (3.94 to 5.51 in)

# Changes with development



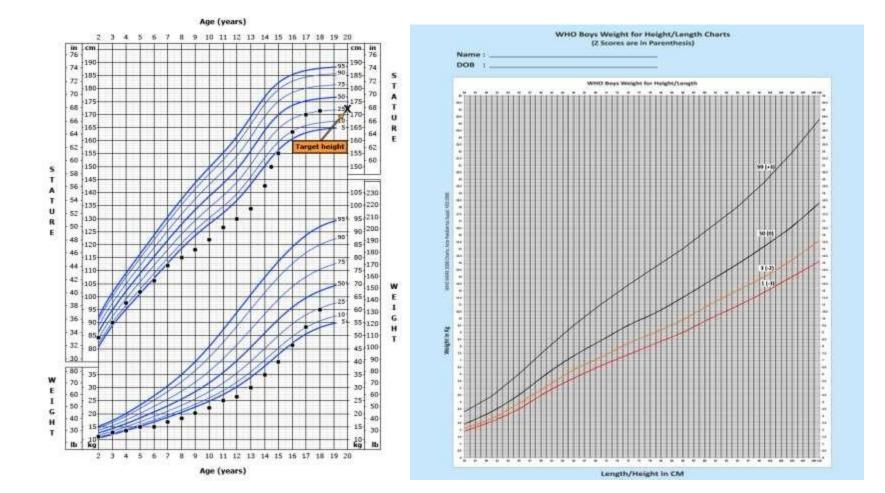
# Physiology of growth hormone



# Growth monitoring

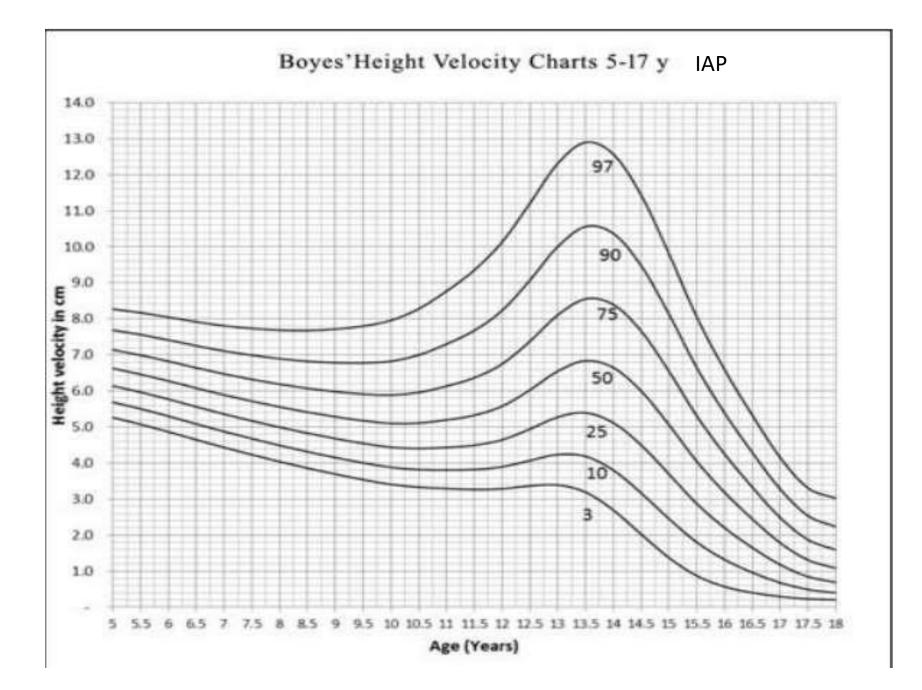
- Absolute height
- Growth velocity
- Weight for height ratio
- Target height
- Body proportion (US:LS ratio)
- Pubertal assessment

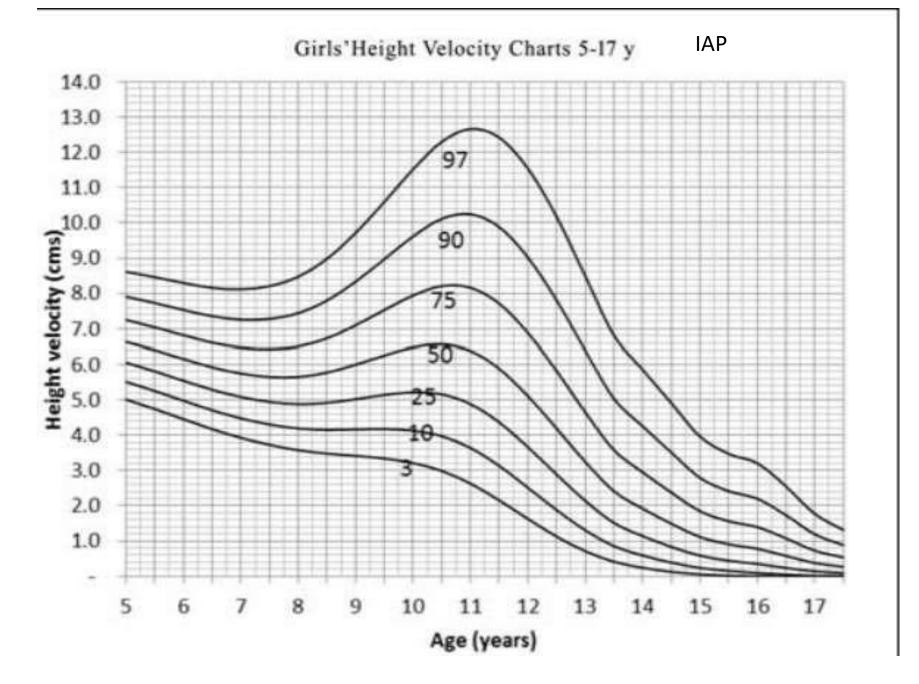
## Growth charts



#### Weight for age and height for age

#### Weight for height/length





## When to evaluate

- Severe short stature (height SDS <-3 SD)
- Severe growth deceleration (height velocity SDS <-2 SD over 12 months).</li>
- Height <-2 SD and height velocity <-1.0 SD over 12 m
- Height <-1.5 SD and height velocity <-1.5 SD over 2 years.
- Risk factors for growth hormone defciency

# Short stature vs growth failure

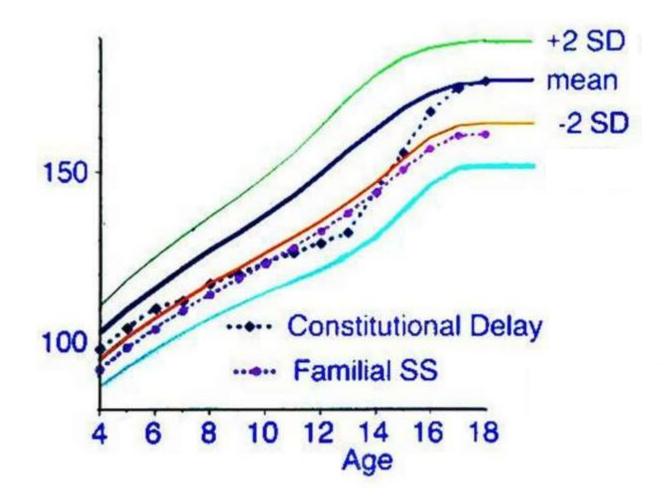
- Short stature -height that is two standard deviations below the mean height for age and sex (less than the 3rd percentile)
- Growth failure -pathologic state of abnormally low growth rate over time
- -may manifest as height deceleration across two major percentile lines on the growth chart.
- Short stature may be normal, but growth failure is always abnormal

## **MAJOR CLASSES OR PHENOTYPES**

## 1. Normal growth variants

Age	FSS	CGD
Sex		Commoner in boys
Birth Height	Less	Normal
Family History	History of short statur -positive	e Delayed puberty especially in father
Growth Velocity	Growth Velocity normal	Growth Velocity normal for bone age.
Bone Age	Normal	Slight delay/BA = HA
Final adult height	short	Normal

**Management: Explain and reassure** 



# 2. Proportionate short stature

### Increased weight for height BA<<CA

- Hypothyroidism: asymptomatic or -dry skin, constipation, goiter, mental retardation, learning deficits
- GH deficiency: doll facies, high pitched voice, micropenis, marbled abdomen fat -congenital/ acquired
- Glucocorticoid excess: central obesity, hypertension
   -endogenous/ iatrogenic

### Decreased weight for height BA<CA

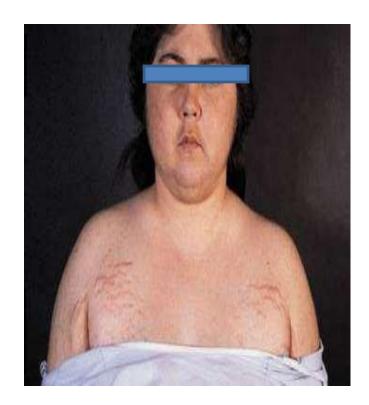
- Malnutritionprotein/calorie/calcium/vitamin D
- Systemic disease
  Can be missed/ not overt in all GI- malabsorption, IBD Renal- RTA, DI Psychosocial
  Usually symptomatic Pulmonary- cystic fibrosis, bronchiectasis
  CVS- Congenital heart disease Others



Congenital hypothyroidism



Growth hormone deficiency



#### Cushing syndrome

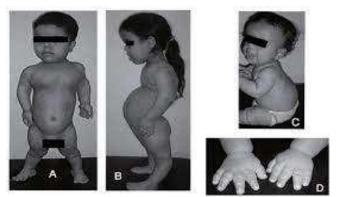
### 3. Disproportionate/ dysmorphic short stature

#### Disproportionate

- BA=CA
- Skeletal dysplasia
   Short limbs: achondroplasia
   Short trunk:spondylodysplasia
- Mucopolysaccharidoses

#### **Clinical syndromes**

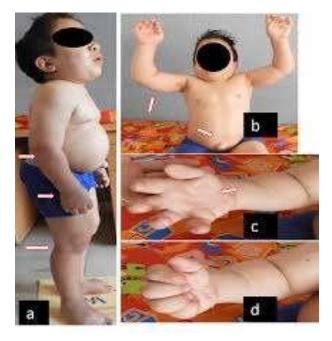
- Bone age minimal delay
- Chromosomal : Turners syndrome, Down, Prader Willi syndrome etc



#### Achondroplasia



Spondylodysplasia



#### Mucopolysachcharidoses



Turner syndrome



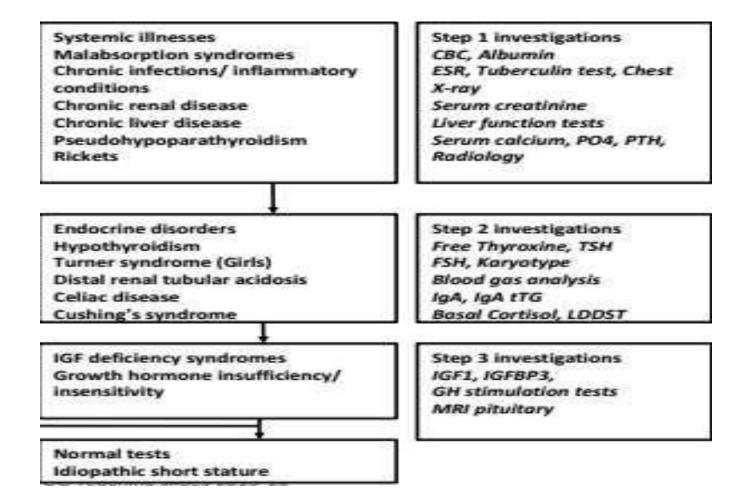
Prader Willi syndrome

## HOW TO EVALUATE

# **Clinical pointers**

- Growth parameters
- Dysmorphic features
- Nutritional status
- Thyroid gland
- Tanner staging for puberty development
- Neurological exam
- visual acuity and visual fields, nystagmus
- signs of hydrocephalus, focal signs
- Signs of any other systemic disease

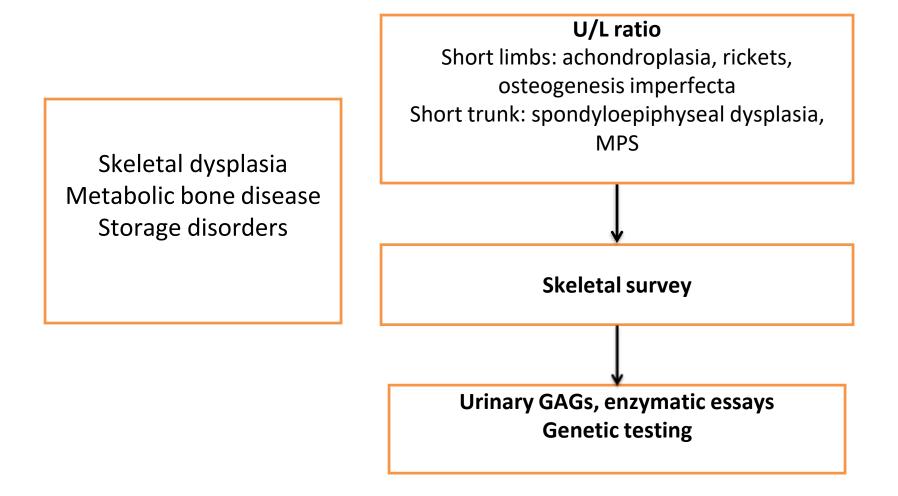
### Proportionate short stature approach



## Evaluation in GH-IGF1 axis

- GH stimulation test
  - -Priming with sex steroids in boys > 11 years and girls > 10 years and not in established puberty
  - -peak concentration< 10 ng/ml in response to 2 stimulation tests(arginine, insulin, clonidine, glucagon)
  - -< 18 ng/ml to combined arginine- GHRH
- IGF-1 levels
- IGFBP-3 levels
- Hypothalamic-pituitary MRI scan in patients with GH deficiency

## Disproportionate short stature



## When to refer to geneticist

- Disproportionate short stature
- Dysmorphism
- Congenital anomalies
- Developmental delay

## Management

- Identifying underlying cause
- Specific disease treatment

Malnutrition: Nutritional rehabilitation

Nutritional rickets: Vitamin D

Distal RTA: Shohl's solution

Celiac disease: Gluten free diet

Psychosocial dwarfism: Good social environment

- Primary hypothyroidism: Thyroxine
- Endogenous Cushing syndrome: Tumorectomy
- Panhypopituitarism: GH, Thyroxine, sex steroids, Glucocorticoid

# GH therapy indication and dosage

Indication	Dose of GHmg/kg/wk	
Growth hormone deficiency	0.18-0.3	
Chronic kidney disease	0.35	
Turner syndrome	0.375	
SGA children with failure to catch up growth	0.47	
Idiopathic short stature	0.48	
Prader Willi syndrome	0.24	
Noonan syndrome	0.46	
SHOX gene haploinsufficiency	0.35	

# Recombinant GH (somatotropin)

- SC route( site rotated daily to avoid lipoatrophy)
- Adverse effects
- -injection site reactions
- -hypersensitivity to the diluent
- -Hypothyroidism
- -Others- edema, arthralgia, myalgia, carpal tunnel syndrome, paraesthesias, and hyperglycemia

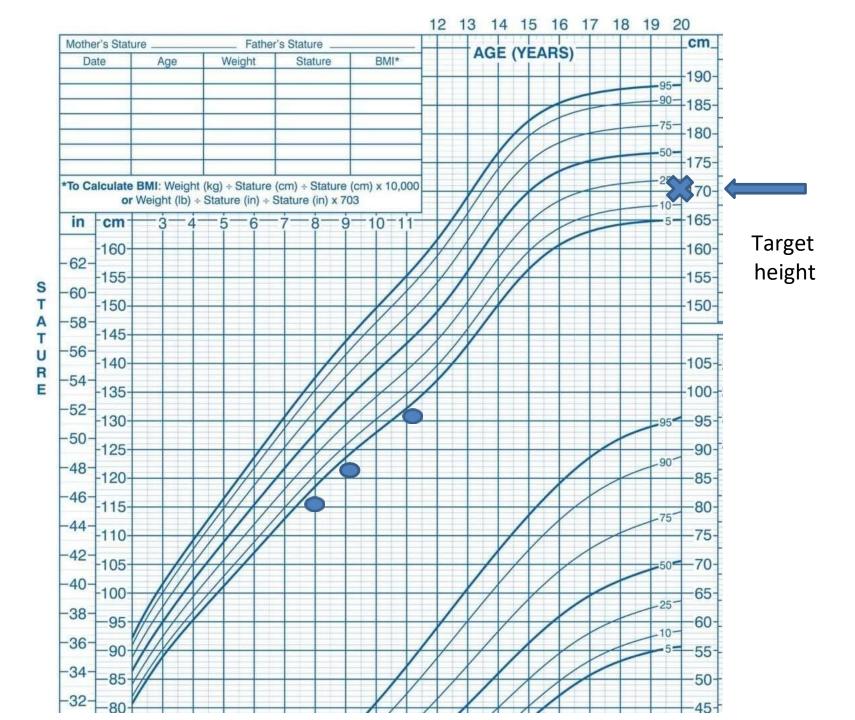
# Points to ponder !

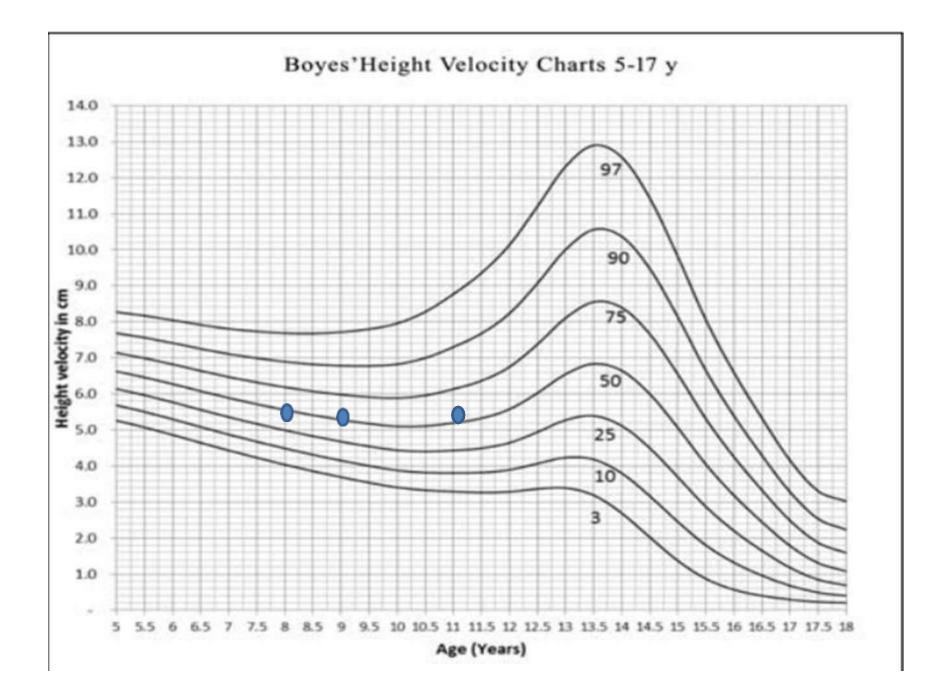
- Deceleration of linear growth in well child/obese childendocrine causes
- Deceleration of linear growth in thin child- systemic causes
- Deceleration of linear growth at adolescencehypogonadism
- Short child with dymorphism/ disproportionate stature- genetic causes
- Short child with normal growth velocity- normal variants

## Case snippet

• A 11 year old boy is brought with concerns of being puny for his age. He is otherwise active, good at school.

Mothers height is 150 cm and fathers is 180 cm





- Weight is on the 10 th centile
- Height is below 3 rd centile
- Height velocity is on 50 th centile
- Systemic examination is normal
- SMR staging is pre pubertal

• He is ABNORMAL

• On re questioning, father admits he too was called short for his age in high school

• Diagnosis : constitutional growth delay

