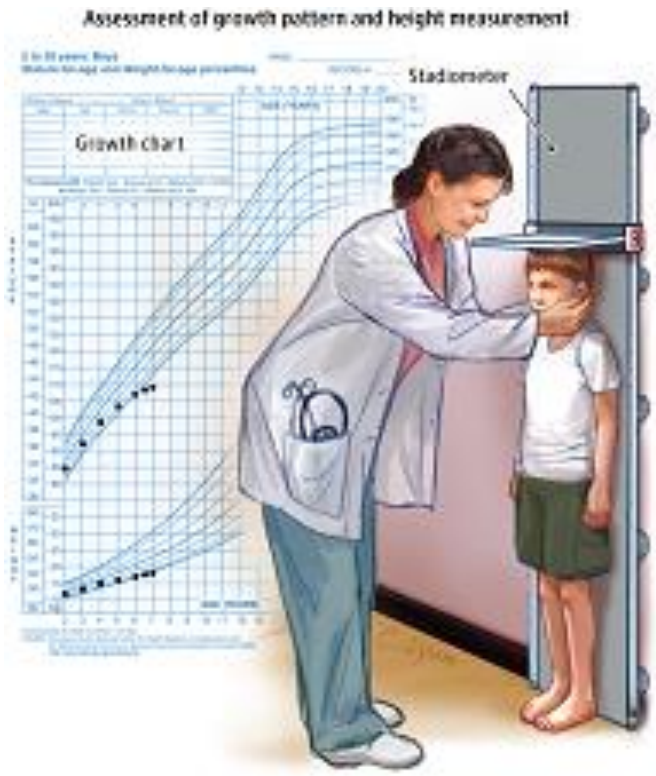
Short stature



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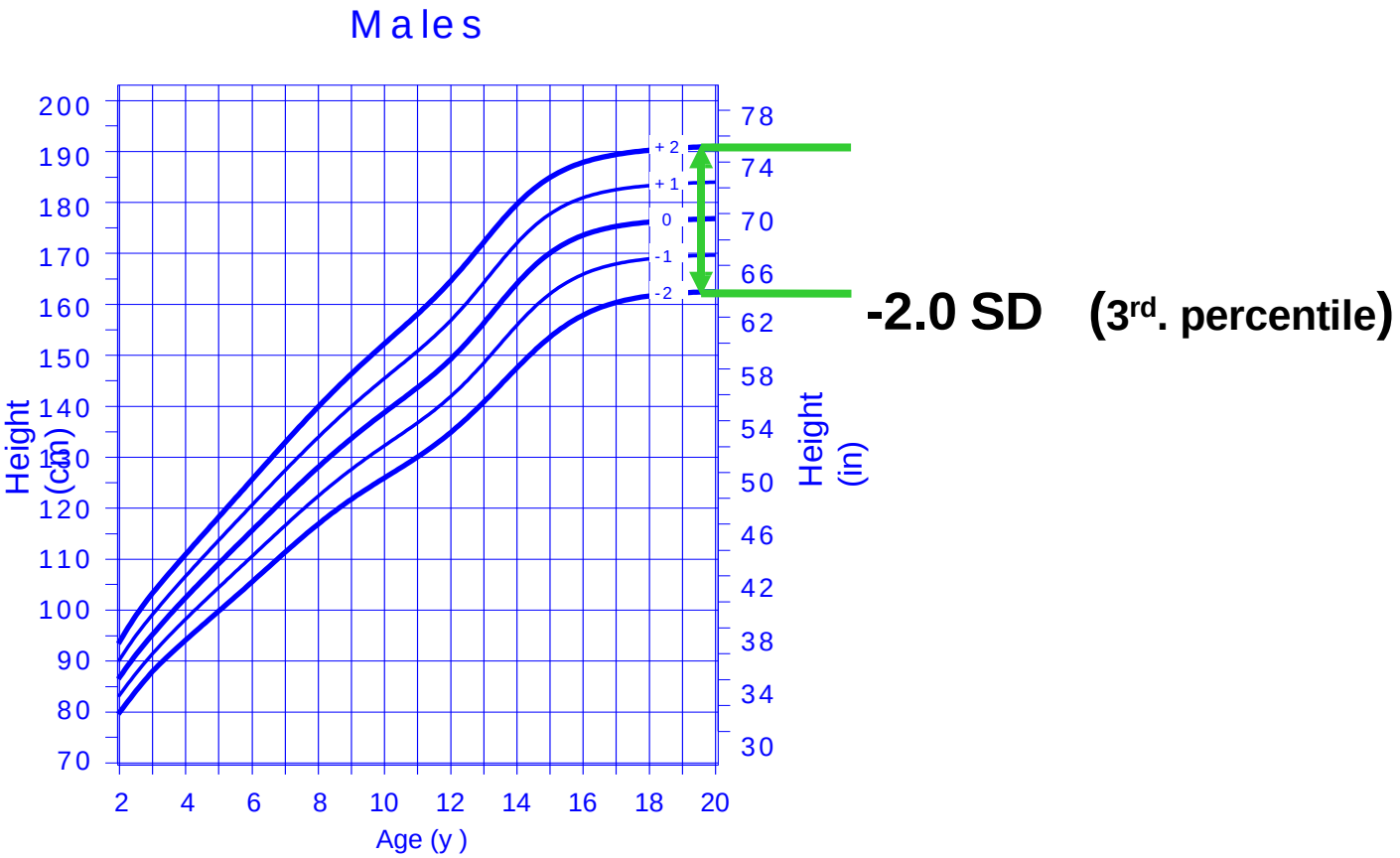
Definition

A child whose height is below 2 standard deviations for age, gender &race



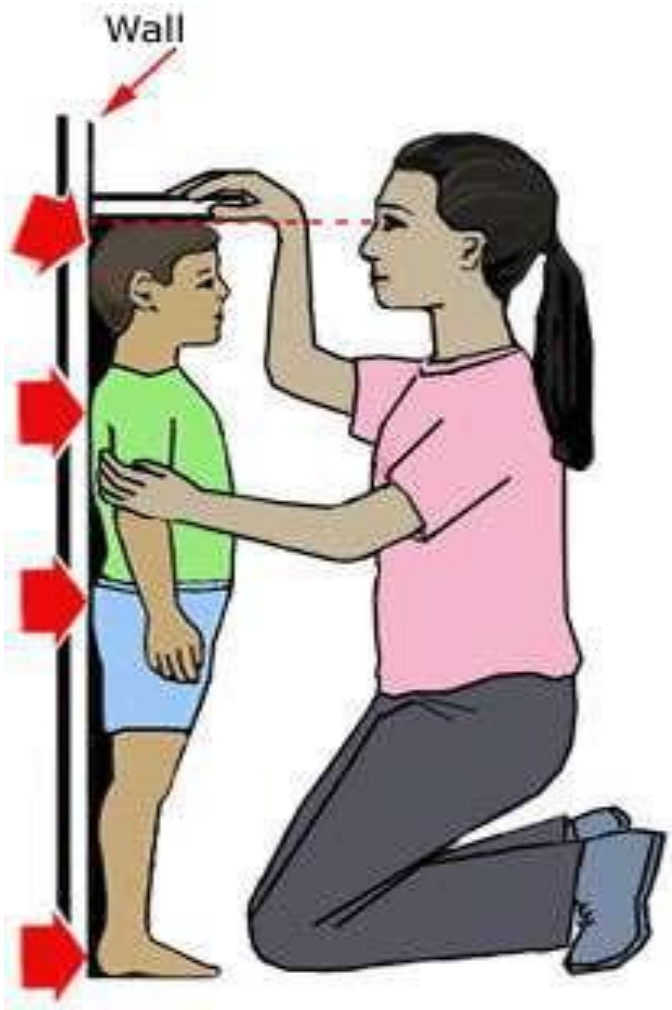
-2.0 SD (3rd. percentile)

The term ***'Dwarfism'*** is no longer used for short Stature.

- Sometimes, height is within the normal percentiles, but growth velocity is consistently below the 25th percentile over 6-12 months of observation.
- It should not be confused with failure to gain weight.
- Target height should be calculated and plotted on growth charts.
- It should be plotted on appropriate growth charts (national chats).
- If syndromes, should be plotted on specific syndrome charts.
  - Turner, Noonan, Russell-silver ……).

Height Measurement

Height Measurement



- Without footwear
- Heels & back touching the wall
- Looking straight ahead in frankfurt plane.
- Gentle but firm pressure upwards applied to the mastoids from underneath
- Record to last 0.1cm

Body Proportion

- Lower segment (LS): Measure from the symphysis pubis to the floor.
- Upper segment (US): Subtract the LS from the height.
  - U/L birth = 1.7
  - U/L 3years = 1.3
  - U/L > 7 years = 1
- Proportionate (involves both the trunk and the lower extremities)
- Disproportionate (involves one more than the other).

Factors affecting Growth

- Genetic factors (important to measure MPH).
- Environmental factors:
  - Intrauterine factors (maternal nutrition, smoking, infections, teratogens, alcohol, HTN, DM,…. Etc.)
  - Extra uterine factors (nutrition, psychological & social, infections, medications …etc.)
- Chronic diseases.
- Endocrine factors (Growth hormone, Thyroid hormone, Gonadotrophins).
- Syndromes / chromosomal anomalies.

Factors affecting height

Intra uterine Growth factors IGF2 Insulin	Nutrition &Thyroid hormone		Nutrition ,Thyroid & Growth Hormone		Nutrition ,Thyroid Growth & Sex Hormones
	Birth	1-2 years	Childhood		Puberty

Genetic Factors

Mid parental height = Target height

Boy (Fa + MO + 13) ÷ 2 = ± 8.5

Girl (MO+ Fa-13) ÷ 2 = ± 8.5

Growth velocity (cm / year)

Age	Normal GV (cm)	Abnormal GV (cm)
1st Year	25	<16
2nd Year	12.5	<10
2 – 5 years	6.5	< 6
5 y – adolescence	5 - 6	<5
Adolescence	10 -12	--

Growth Velocity

- Girls have their peak growth velocity (9 cm / y) during early puberty & before menarche (Tanner II – III)
- Boys have their peak growth velocity (10.3 cm/year) mid puberty (Tanner III-IV)
- Boys' pubertal growth spurt period is longer than girls.

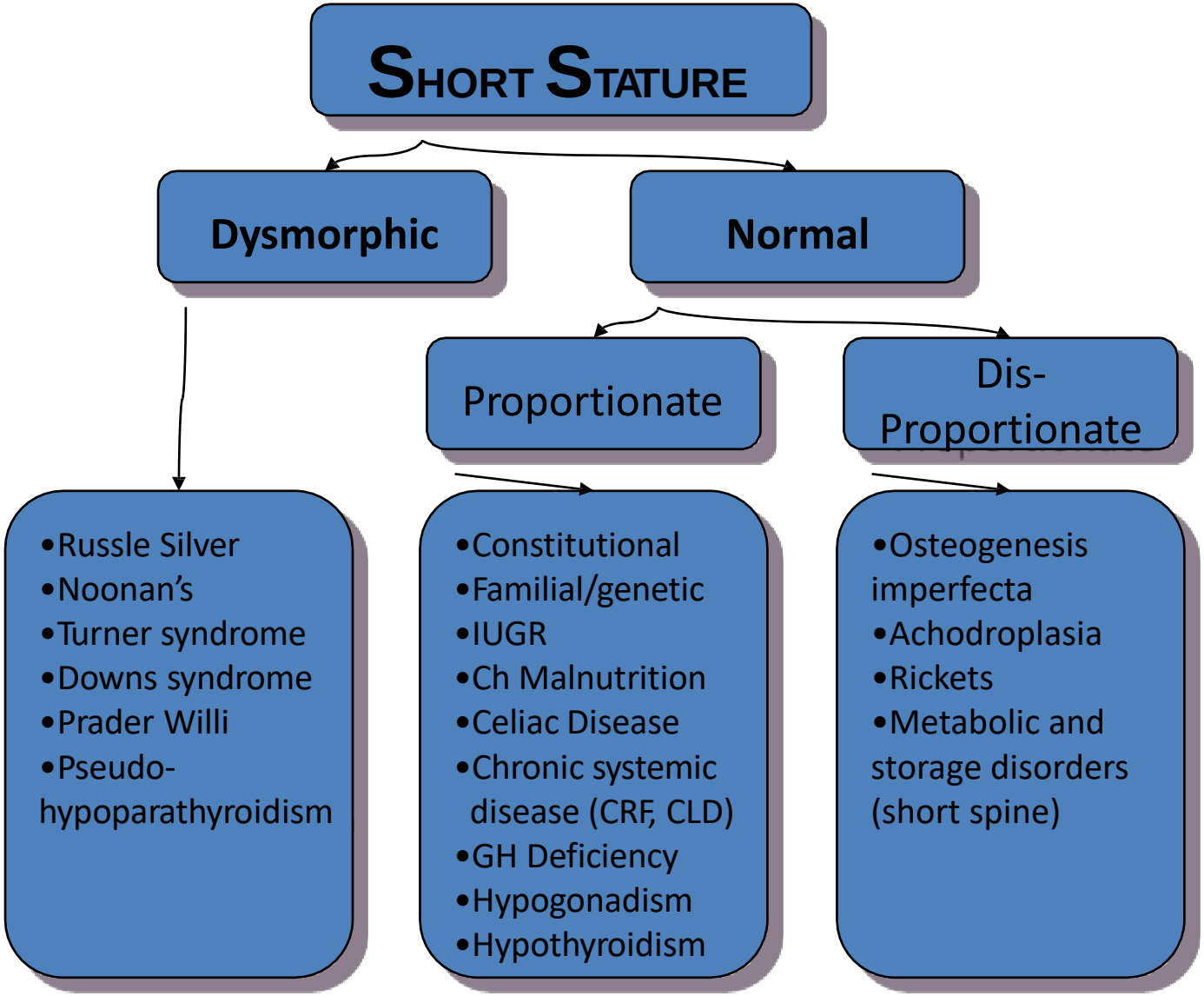
Normal heights in children

- Birth length — 50cm
- One year — 75 cm
- Two yrs — 87.5 cm
- Three yrs — 93.75 cm
- 4 yrs — 100 cm
- 8 yrs — 125 cm
- 12 yrs — 150 cm

growth velocity 6 cm per year

Important notices !!!

- The most critical factor in evaluating the growth is determining “growth velocity”.
- Observation of a child’s height pattern in the form of “crossing down percentile” on a linear growth curve is the simplest method of observing abnormal growth velocity.
- At least 3 measurements with preferably 6 months interval in between is necessary to comment on the growth pattern.
- A short child with delayed bone age is of much more concern.


SHORT STATURE
Dysmorphic
Normal
Proportionate
Dis-Proportionate
•Russle Silver
•Noonan's
•Turner syndrome
•Downs syndrome
•Prader Willi
•Pseudo-hypoparathyroidism
•Constitutional
•Familial/genetic
•IUGR
•Ch Malnutrition
•Celiac Disease
•Chronic systemic disease (CRF, CLD)
•GH Deficiency
•Hypogonadism
•Hypothyroidism
•Osteogenesis imperfecta
•Achodroplasia
•Rickets
•Metabolic and storage disorders (short spine)

Proportionate Short Stature

- Normal Variants (physiological)
  - Familial short stature
  - Constitutional delay of growth & puberty
- Prenatal Causes
  - Intra-uterine growth retardation
    - Placental causes, Infections, teratogens
    - Intra-uterine Infections
  - Genetic Disorders (Chromosomal & Metabolic Disorders)
- Malnutrition.
- Malabsorption e.g., Celiac disease, cystic fibrosis.

- Chronic systemic diseases (detailed systemic review).
- Psychosocial / emotional deprivation.
- Idiopathic short stature.
- Endocrine Causes:
  - Growth Hormone Deficiency/ insensitivity
    - Hypothyroidism
    - Poorly controlled type 1 diabetes mellitus
    - Cushing Syndrome
    - Pseudohypoparathyroidism

Disproportionate Short Stature

- Short Limbs:
  - Achondroplasia, Hypochondroplasia, or any other types of skeletal dysplasia.
  - Osteogenesis imperfecta.
  - Hereditary Rickets.

- Short trunk:
  - Spondyloepiphyseal dysplasia, mucolipidosis, mucopolysaccharidosis, Hemi vertebrae

Diagnosis

- Detailed history
- Careful examination
- Laboratory evaluation

History

- Date of onset.
- Birth history.
- Past medical & surgical history.
- Systemic inquiry.
- Developmental history.
- Family and social history.
- Nutritional history.
- Allergies/ medication.

Clues to etiology from history

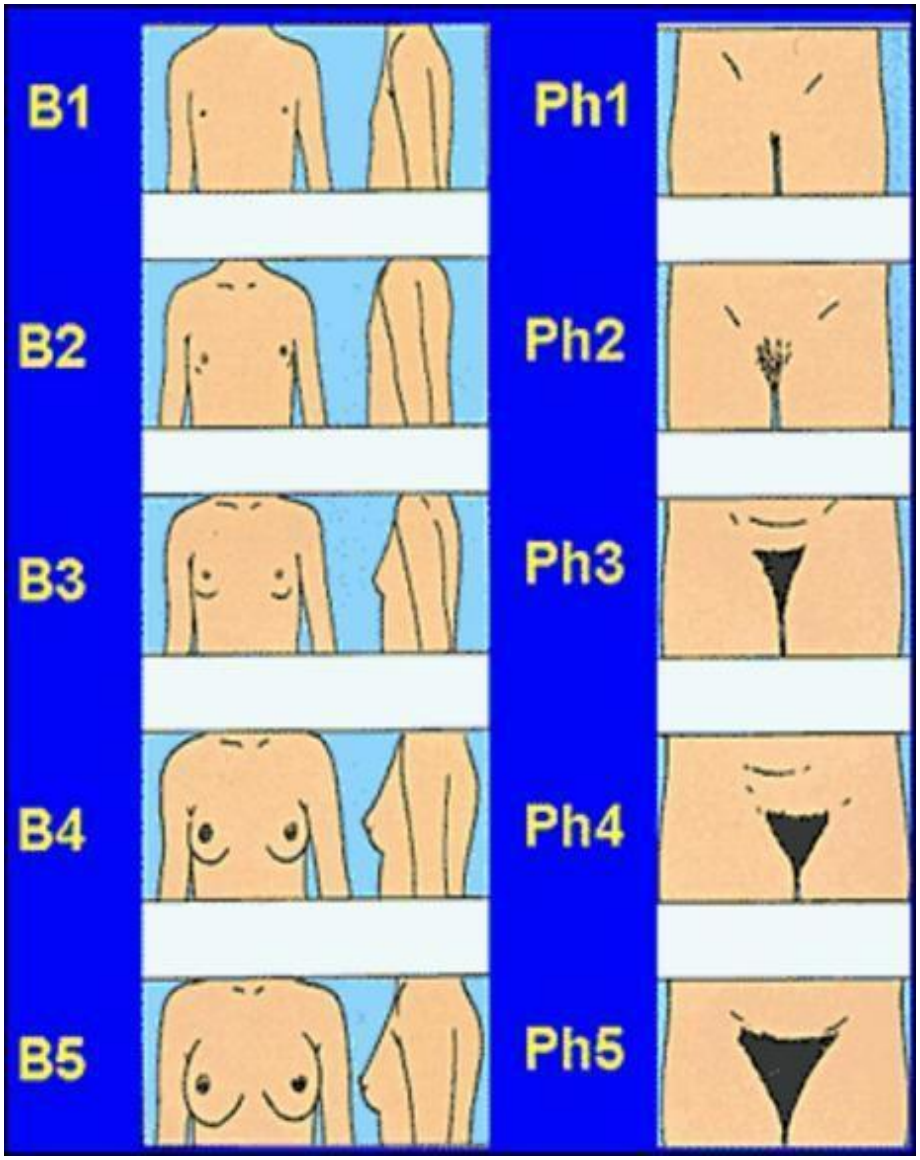
History	Etiology
History of delay of puberty in parents Low	Constitutional delay of growth
Birth Weight	SGA
Neonatal hypoglycemia, jaundice, micropenis	GH deficiency/ Hypopituitarism
Dietary intake	Malnutrition
Headache, vomiting, visual problem	Pituitary/ hypothalamic tumors
Lethargy, constipation, weight gain	Hypothyroidism
Social history	Psychosocial dwarfism
Diarrhea, greasy stools	Malabsorption
Detailed systemic review	Chronic illnesses

Clues to etiology from physical examination

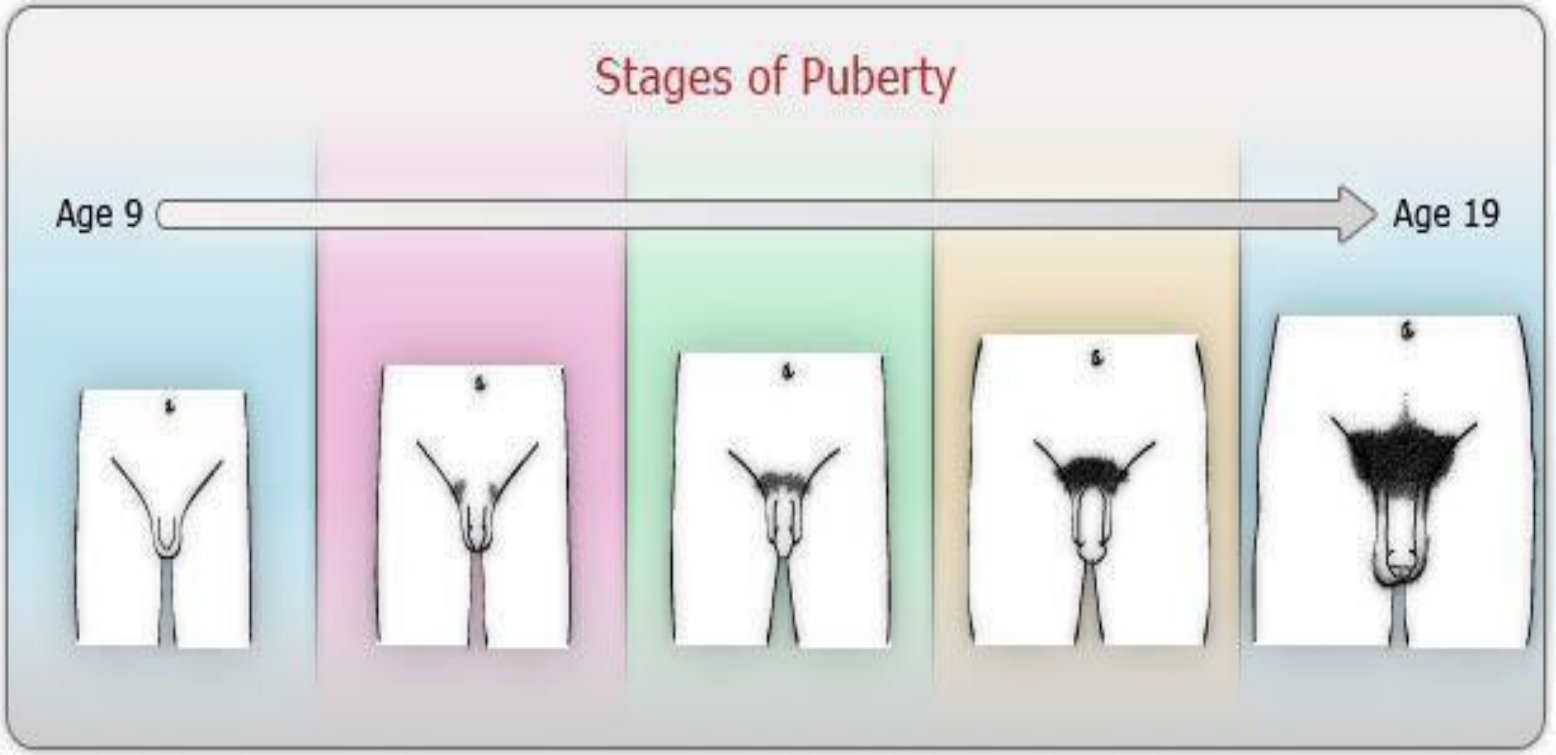
Pointer	Etiology
Midline facial defects, micropenis, frontal bossing, depressed nasal bridge, crowded teeth,	GH deficiency / Panhypopituitarism
Signs of Rickets and / deformities	Renal failure, RTA, malabsorption, Hereditary
Pallor	Renal failure, malabsorption, nutritional anemia
Signs of malnutrition , clubbing, wasting	PEM, malabsorption, celiac disease, cystic fibrosis
Short & obese	Hypothyroidism, Cushing syndrome, Prader Willi syndrome, GH deficiency
Metacarpal shortening	Turner syndrome, Pseudohypoparathyrodism
Cardiac murmur	Congenital heart disease, Turner syndrome
Mental retardation	Hypothyroidism, Down, Pseudohypoparathyrodism

Always Perform Tanner staging

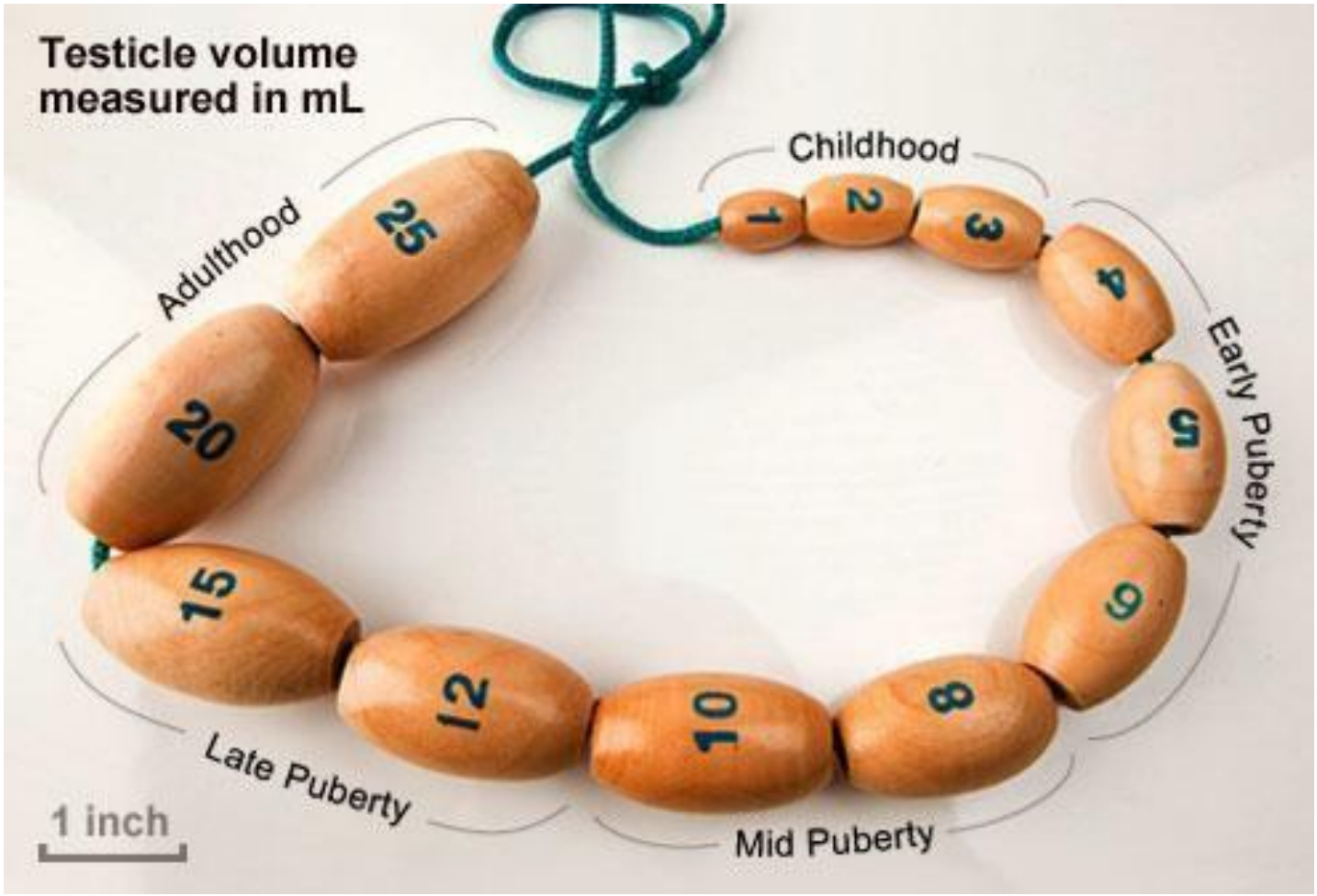
Tanner Stages Females



Tanner Stages Males



Orchidometer



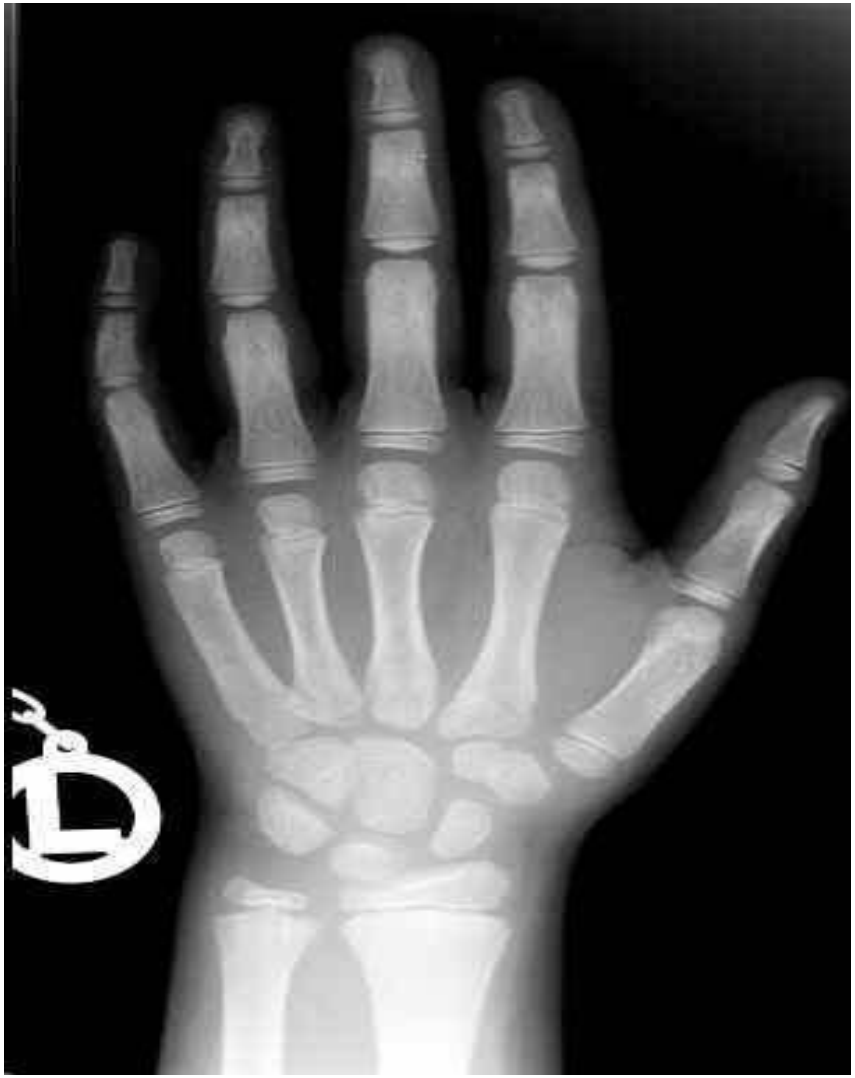
Investigations

Universal for all cases include:

- Bone age (mandatory).
- Thyroid function test (even if no other symptoms).
- Karyotype in girls (even if no dysmorphism).
- CBC, ESR.
- Electrolytes ,Renal &Liver function tests.
- Urinalysis & stool analysis.
- IgA anti-tissue transglutaminase as screening for celiac (even if no other symptoms).

Bone Age

Helpful in differentiating the types of short stature whether delayed or appropriate for chronological age



Investigations

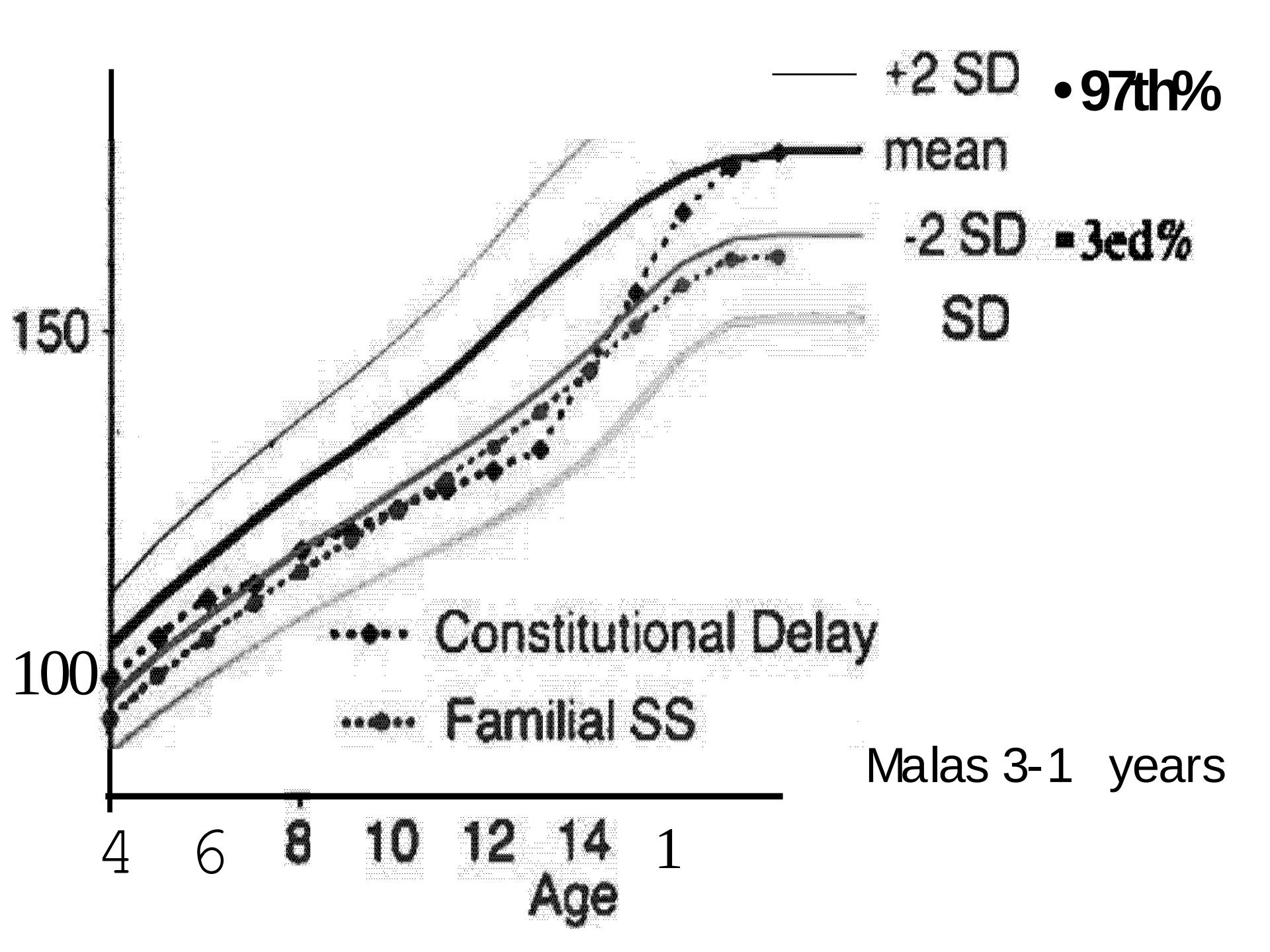
- Apart from universal previous investigations, further ones depend on possible suspicion from history & examinations for example:
  - Skeletal survey: skeletal dysplasia or hereditary rickets
  - Calcium, Phosphate, alkaline phosphatase: Rickets
  - Sweet chloride test: cystic fibrosis.
  - Duodenal biopsy: Celiac disease.
  - Growth factors +/ - GH stimulation test if GH deficiency is suspected.

GH deficiency

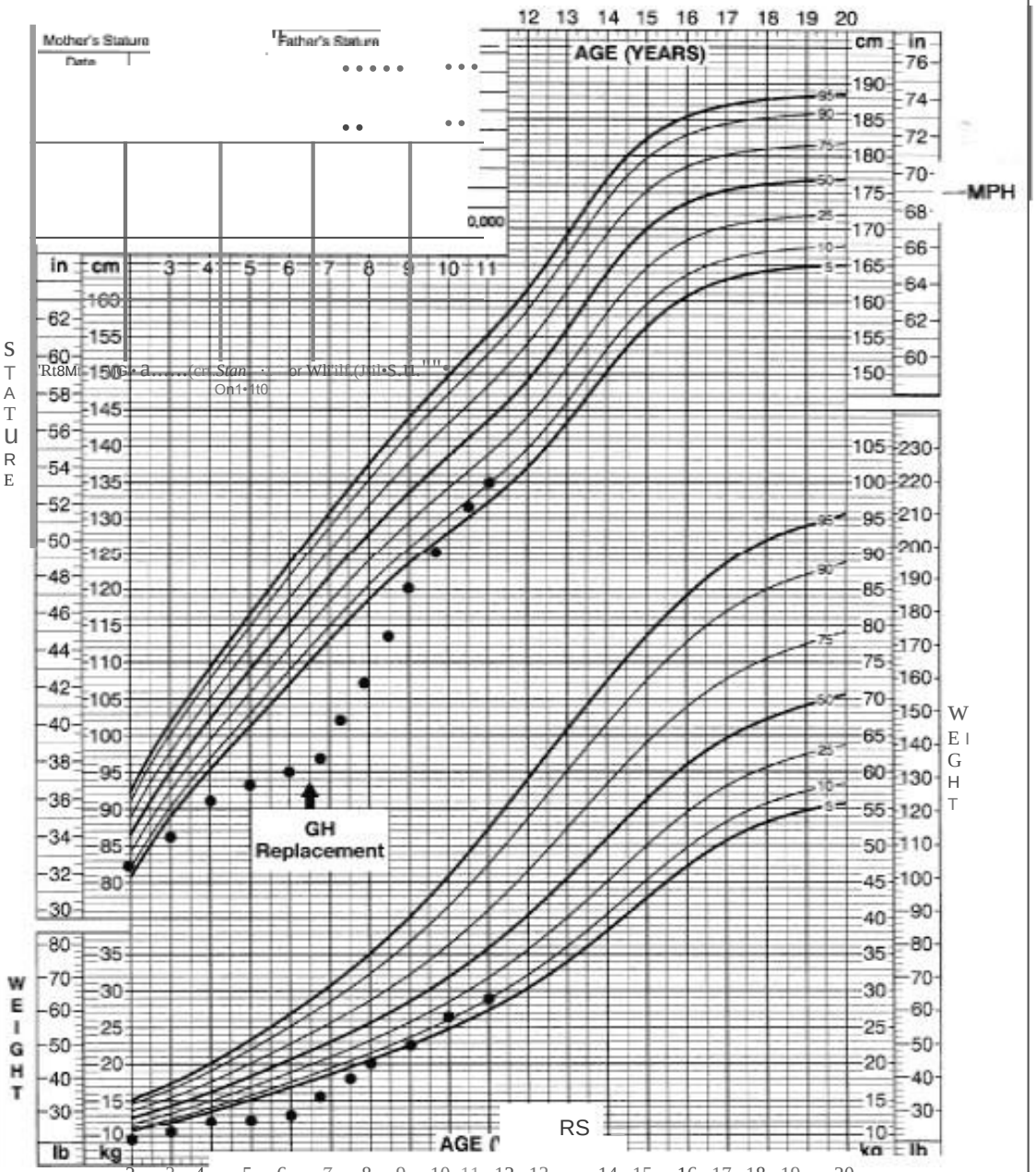
- Screening tests
  - IGF-1, IGFBP3 (Neither are completely sensitive or specific)
  - Physiological GH stimulation
    - exercise / deep sleep (delta wave)
- Pharmacological stimulation tests
  - Preferably two pharmacological tests
  - These tests are best left for the specialized units.
- Neuroimaging
  - only if GH / Pituitary hormone deficiency is confirmed (after not before confirmation).

Familial Vs Constitutional

- The hallmarks of familial (genetic) short stature is normal bone age, normal growth velocity & current height lies within the mid-parental height range.
- By contrast, constitutional growth delay is characterized by delayed bone age & delayed appearances of pubertal signs with positive family history.
- Patients with constitutional growth delay typically have a first or second-degree relative with constitutional growth delay)



Malas 3-1 years


Mother's Stature
Father's Stature
AGE (YEARS)
cm
in
MPH
STATURE
WEIGHT
GH
Replacement
RS
lb
kg

Take Home Messages

In any short child, we have to:

- Height & weight (Accurate & Serial measures) .
- Height velocity.
- Calculate Mid Parental Height (MPH).
- Height age (Height of a person at the 50th percentile for their age).
- Dysmorphic features.
- Systemic examination.
- Pubertal development staging.
- Bone Age.
- appropriates Investigations.


Thank
you