Approach to a child with short stature
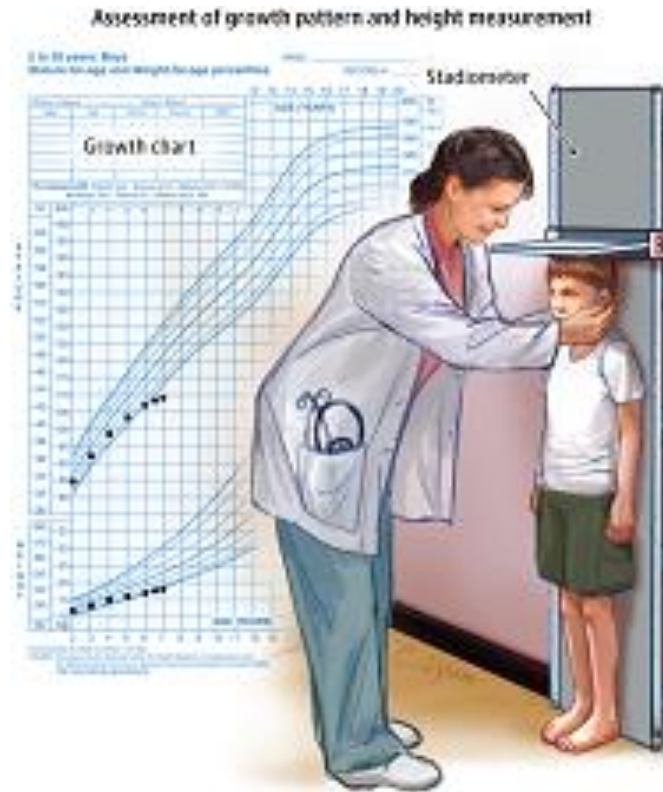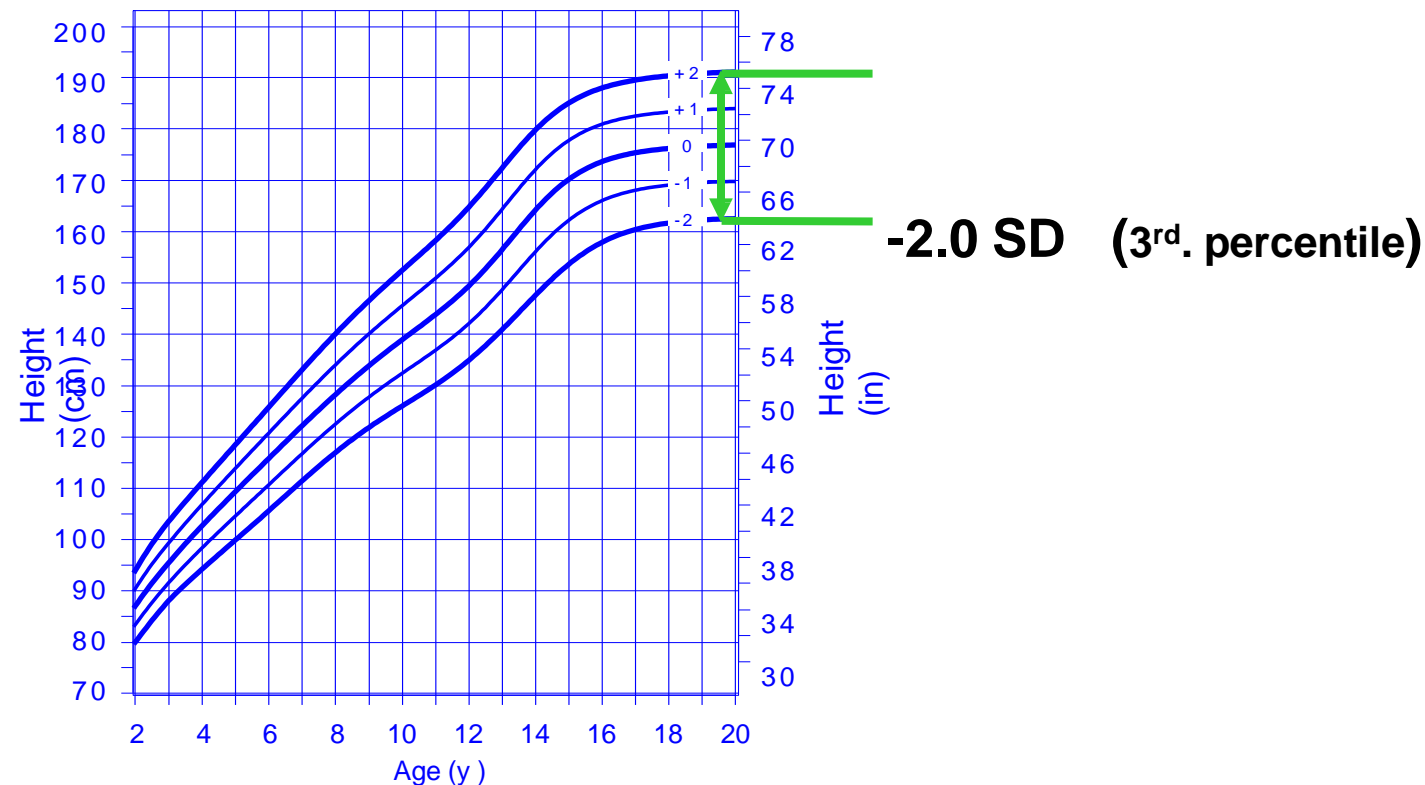

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Definition

A child whose height is below (3rd. Percentile) or 2 standard deviations for age , gender &race

Males

-2.0 SD   (3rd. percentile)

- Sometimes, height is within the normal percentiles, but growth velocity slow over 6-12 months of observation (pathological causes should be excluded).
- Short stature should not be confused with failure to gain weight.
- Target height should be calculated & plotted on growth charts.
- Height should be plotted on appropriate growth charts.
- If syndromes, height should be plotted on specific syndrome charts.
  - Down, Turner, Noonan, Russell-silver …..)

Standing Vs supine height measurements

Supine = length

Standing = Height
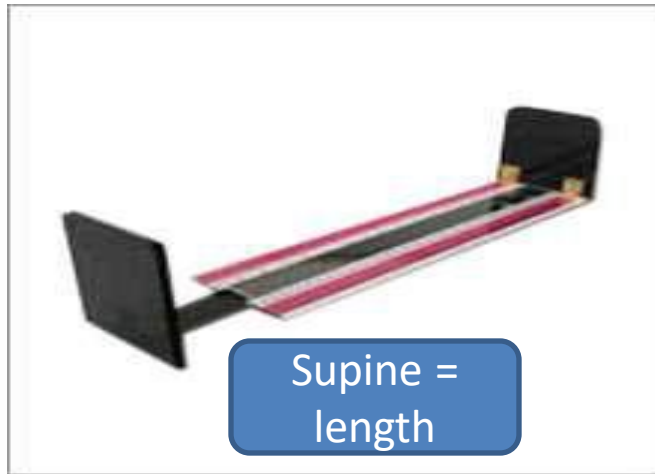
How to measure the height?
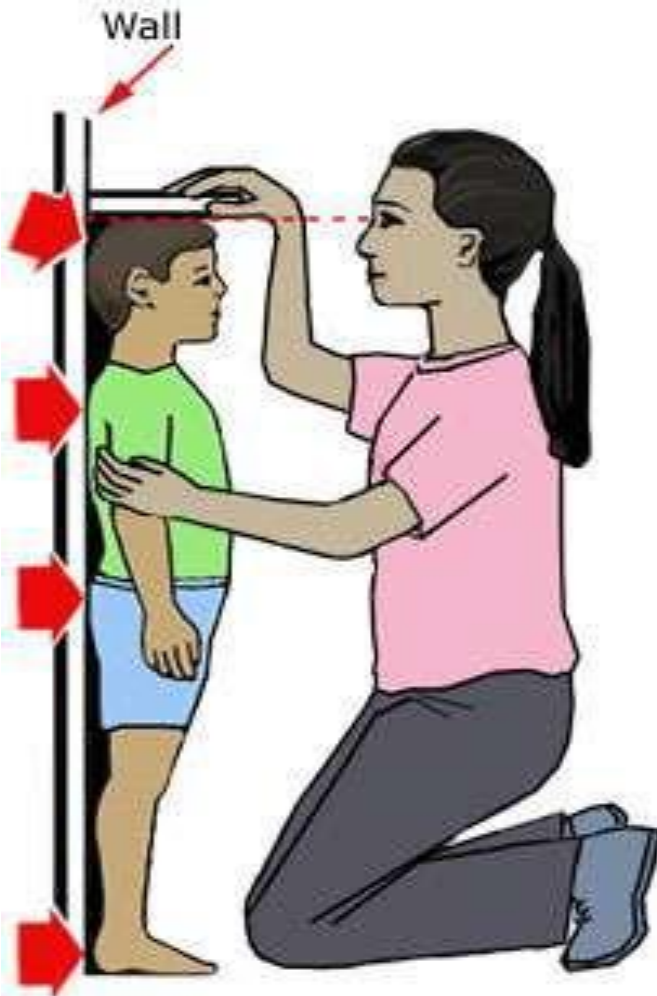
Wall

- Without footwear.
- Heels & back touching the wall.
- Looking straight ahead.
- Gentle but firm pressure upwards applied to the mastoids from underneath.

Body Proportion

- Lower segment (LS): Measure from the symphysis pubis to the floor.

- Upper segment (US): Subtract the LS from the height.

  - US/LS at birth = 1.7.

  - US/LS at 3years = 1.3.

  - US/LS at 7 – 14 years = 1.

- Proportionate (involves both the trunk and the lower extremities)

- Disproportionate (involves one more than the other).

Factors affecting Growth

- Genetic factors.

- Environmental factors including:

  - 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).

- Congenital malformations / syndromes.

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: (Father height + Mother height + 13 cm) ÷2
Girl: (Mother height + Father height -13 cm)÷ 2

Results will be plotted on growth charts ± 8.5 cm.

Growth velocity (cm / year)

Age	Normal GV (cm)	Abnormal GVm(cm)
1st Year	25	< 16
2nd Year	12.5	< 10
2 – 5 years	6-7	< 6
5 y – onset of puberty	5 - 6	< 5
Pubertal growth spurt	10 -12	< 8

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 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 -12 months interval is necessary to comment on growth velocity.

- A short child with delayed bone age is of much more concern.

SHORT STATURE

Dysmorphic

- Russle Silver
- Noonan's
- Turner syndrome
- Downs syndrome
- Prader Willi
- Pseudo-hypoparathyroidism

Normal

Proportionate

- Constitutional
- Familial/genetic
- IUGR
- Ch Malnutrition
- Celiac Disease
- Chronic systemic disease (CRF, CLD)
- GH Deficiency
- Hypogonadism
- Hypothyroidism

Dis-Proportionate

- Osteogenesis imperfecta
- Achodroplasia
- Rickets
- Metabolic and storage disorders (short spine)
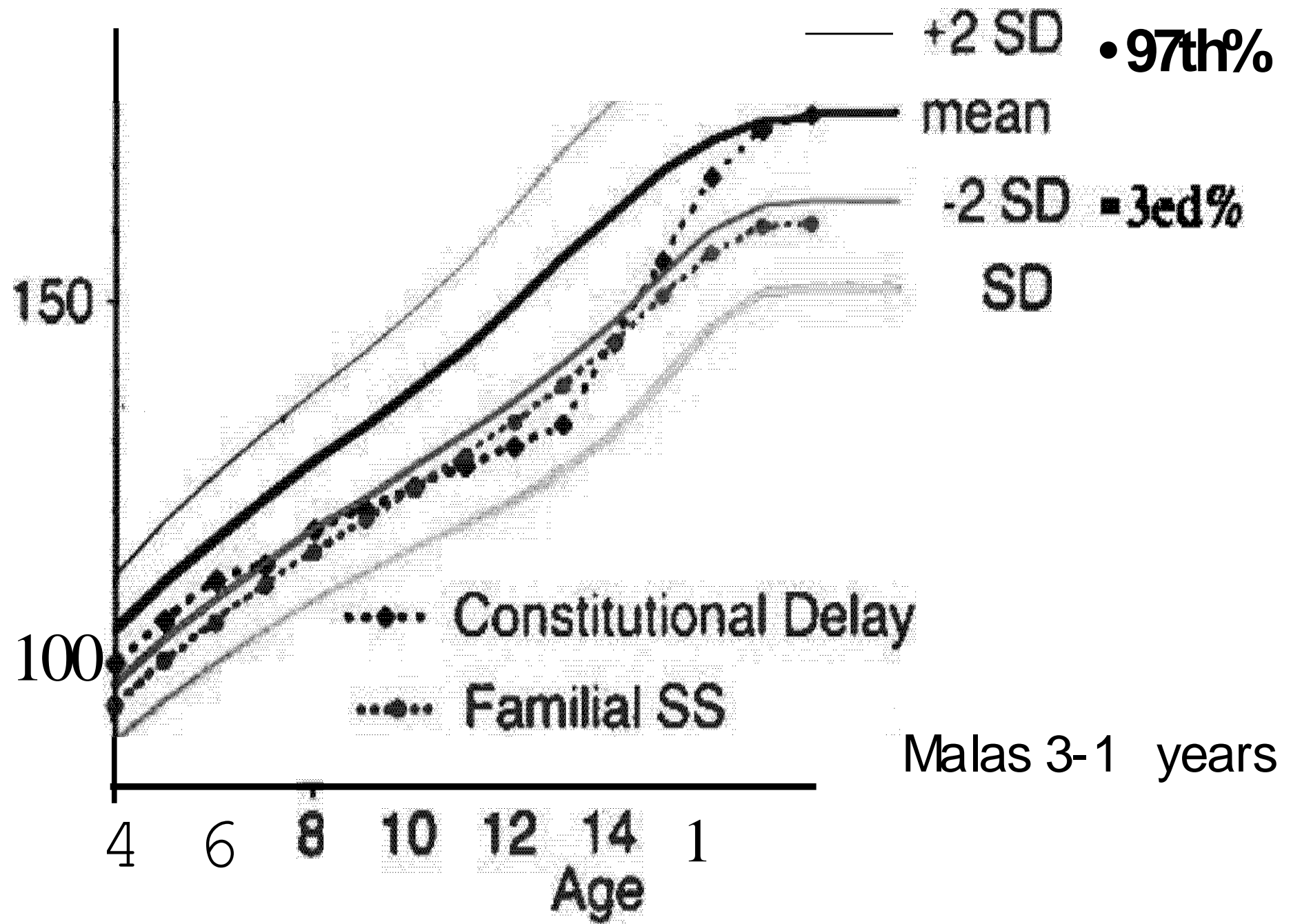
Proportionate Short Stature

- Normal Variants:
    - Familial
    - Constitutional Growth Delay
- Prenatal Causes:
    - Intra-uterine Growth Restriction
        - Placental causes, Infections, teratogens
        - Intra-uterine Infections
    - Genetic Disorders (Chromosomal & Metabolic Disorders)
- Malnutrition.
- Malabsorption: e.g. Celiac disease, cystic fibrosis.

- Chronic systemic diseases.
- Psychosocial short stature.
- Emotional deprivation.
- Endocrine causes including:
  - Growth Hormone Deficiency/ insensitivity.
  - Hypothyroidism.
  - Diabetes Mellitus.
  - Cushing Syndrome.
- Idiopathic short stature.

Familial Vs Constitutional

- Familial (genetic) short stature is characterized by normal bone age; normal growth velocity & child's height lies within mid-parental height range.

- By contrast, constitutional growth delay is characterized by delayed bone age & delayed appearances of pubertal signs with positive family history of delayed puberty.

Malas 3-1 years

Disproportionate Short Stature

- **Short Limbs:**
  - Achondroplasia, Hypochondroplasia, Chondrodysplasia punctata, Chondroectodermal Dysplasia, Diastrophic dysplasia, Metaphyseal Chondrodysplasia
  - Osteogenesis Imperfecta, Hereditary Rickets

- **Short trunk:**
  - Spondyloepiphyseal dysplasia, mucolipidosis, mucopolysaccharidosis, hemi vertebrae

Diagnosis

- Detailed history.
- Careful examination.
- Laboratory evaluation.

History

- Onset of short stature.

- Antenatal, natal & post natal histories including birth measurements).

- Past medical & surgical history.

- Family history of short stature including parents height and timing of their puberty.

- Systemic review.

- Developmental history.

- Family history.

- Nutritional history.

- Medication & Allergic histories.

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

Males Tanner Stages

Stages of Puberty

Age 9 → Age 19
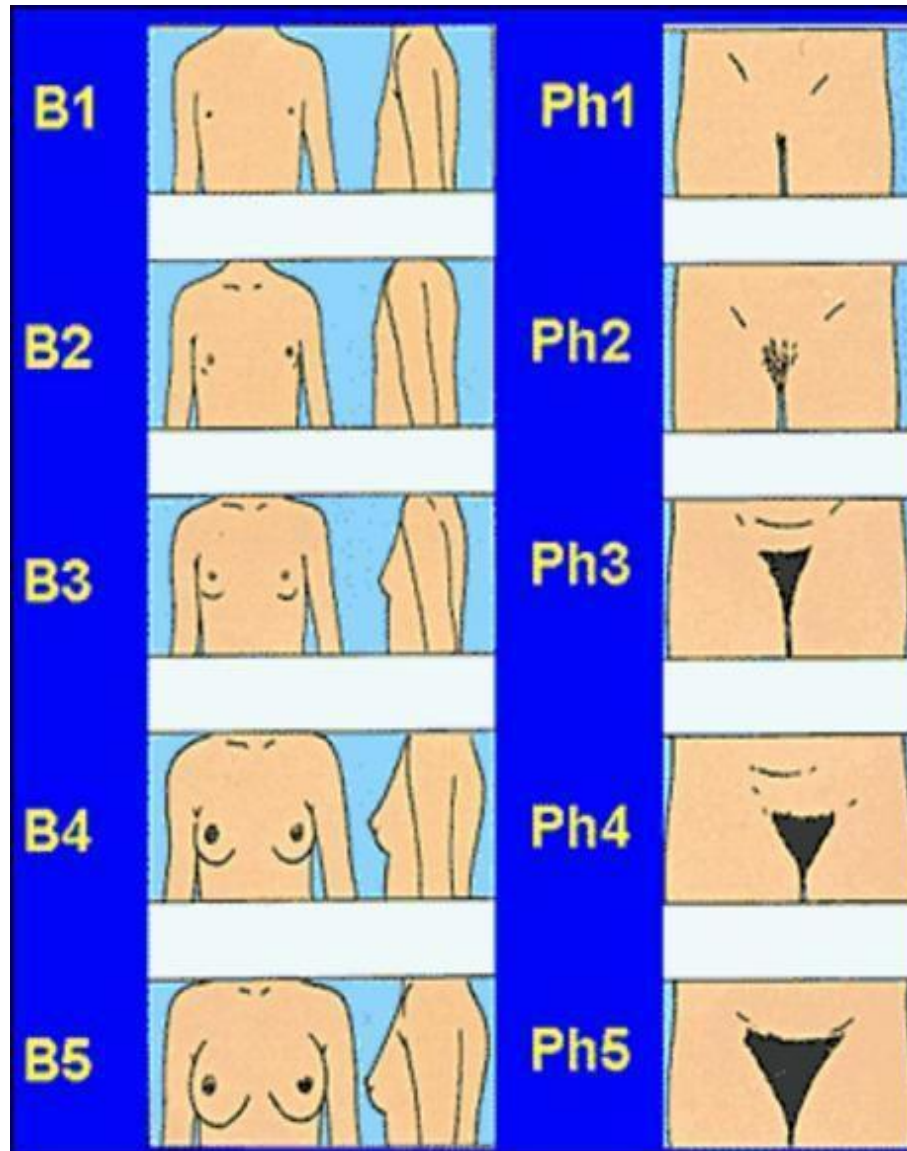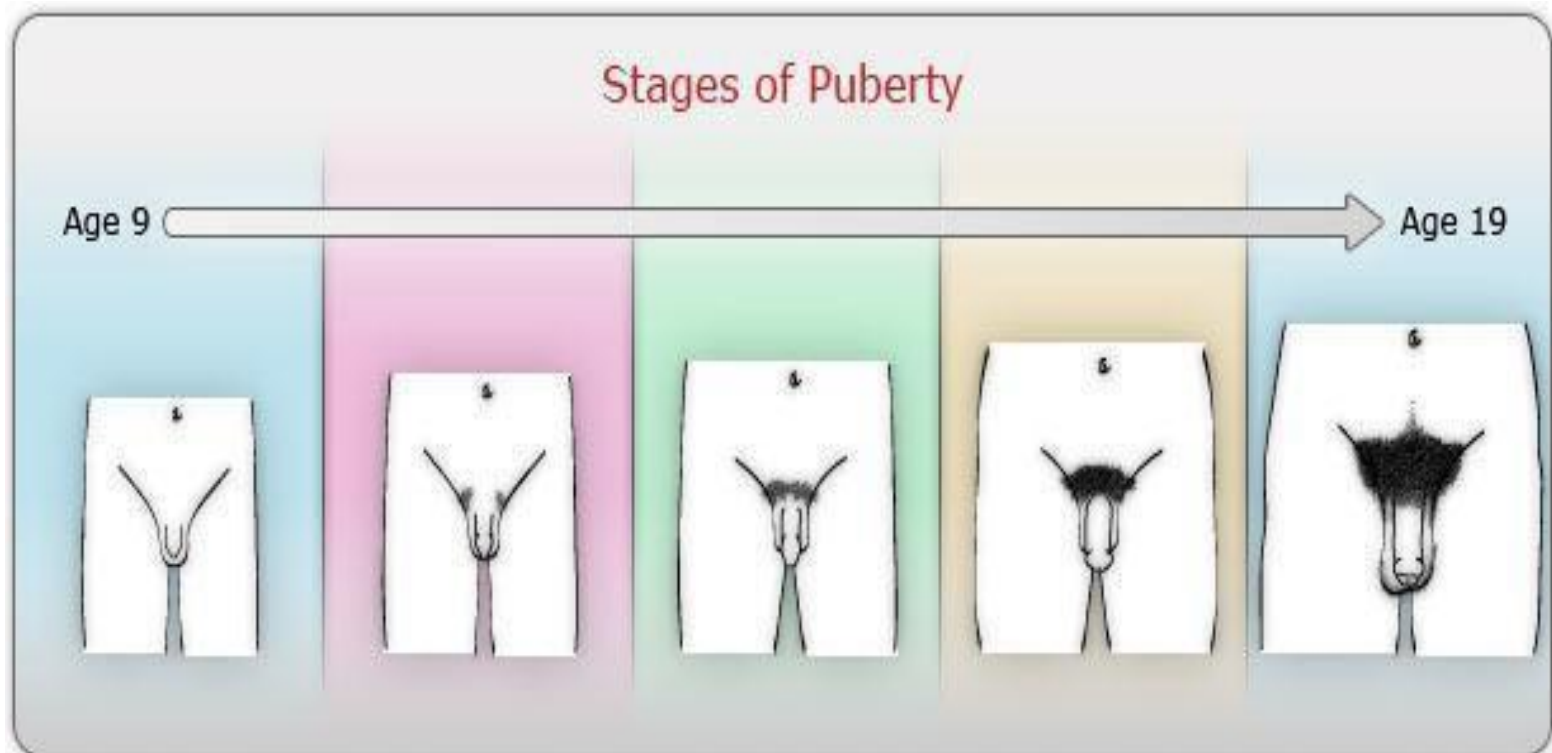
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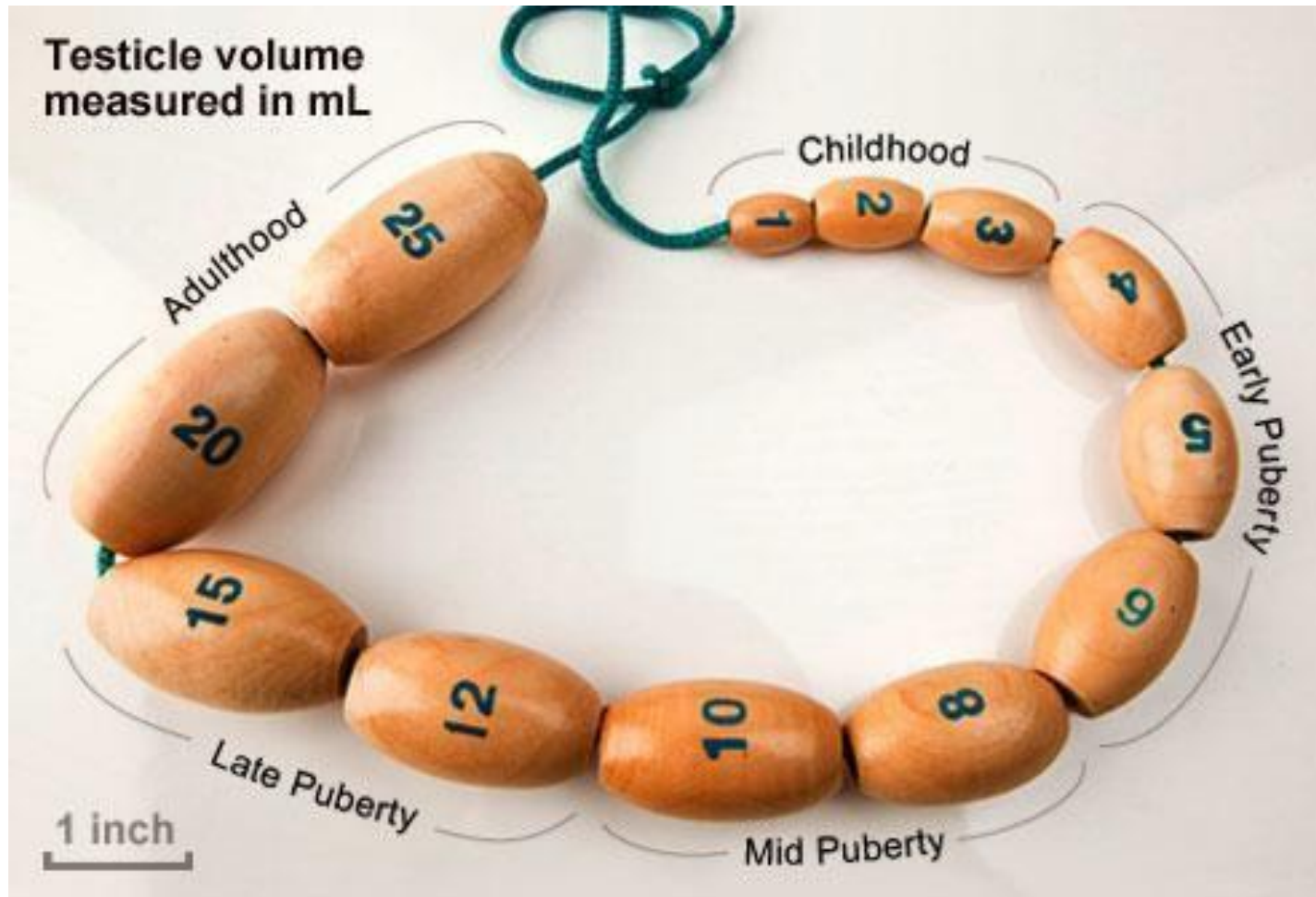
Investigations

Universal for all cases include:

- Bone age (mandatory to differentiate between physiological and pathological short stature).

- Thyroid function test (even if no other symptoms).

- Karyotype in girls (even if no dysmorphism).

- CBC.

- 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 bone age or normal.

- Also, good to indicate whether or not epiphysis is still open or closed when assesses pubertal boys or girls.

Investigations

Further investigations depend on suspected possibilities:

- Skeletal survey : Skeletal dysplasia.
- Serum calcium, phosphate, alkaline phosphatase, venous gas, fasting glucose, albumin, transaminases for various types of rickets.
- Sweet chloride test: Cystic fibrosis.
- Jejunal biopsy : Celiac disease (if screening is positive)
- Growth factors: (IGF-1, IGFBP3 (Neither are completely sensitive or specific).
- GH stimulation test if GH deficiency is suspected.
- Pharmacological stimulation tests: Two pharmacological tests
- MRI Brain: if GH hormone deficiency is confirmed.

Take Home Messages

In any short child, we must assess:

- Height & weight (accurate & serial measures).
- Growth velocity.
- Calculate mid parental height (Target Height).
- Dysmorphic features.
- Systemic examination.
- Pubertal Tanner staging.
- Bone Age.
- Appropriates investigations.

Cases

Twelve –year old girl, who has presented with height below 3rd%, with growth velocity of 3 cm/ year. Both parents were of average height (MPH 168 cm between 10th-25th%). On examination, Tanner staging was B3 PH 2. Among which one of the following is most appropriate approach?

a) Observe & follow up in next six months.
b) Re-assure the family, shortly she will catch- up.
c) Do bone age.
d) Do full hormonal assessments workup.

Seven - year old girl, presented to the endocrine clinic because of short stature, which was reported since birth. She continued to be shorter than her schoolmates. On examination (photo). Which one of the following is a diagnostic investigation?

a) Bone age assessment.
b) Skeletal survey.
c) Thyroid function test.
d) Chromosomal analysis.

Achondroplasia

- Occurs due to sporadic mutations in the majority of cases but can be inherited as autosomal dominant condition.
- Achondroplasia is the most common form of disproportionate short stature.
- Generally recognizable intrauterine because of short limbs & macrocephaly.
- Limb shortening is predominantly in the proximal segment (rhizomelic).
- A **trident hand** is a description where the hands are short with stubby fingers, with a separation between the middle and ring fingers.
- Children with achondroplasia have normal intelligence.
- Specialized growth charts have been developed for head circumference, height.
- They have significant midface hypoplasia, which increases the risk of obstructive sleep apnea.
- The narrowing of the foramen magnum can cause brainstem compression with an increased incidence of sudden infant death.

Achondroplasia

- Pelvis is abnormal with small, square iliac wings
- Horizontal acetabular roots and narrowing of the greater sciatic notch
- long bones are short and the metaphyses slope
- because of narrow chest - respiratory problems are frequent
- translucent area at proximal ends of the femora in neonatal period
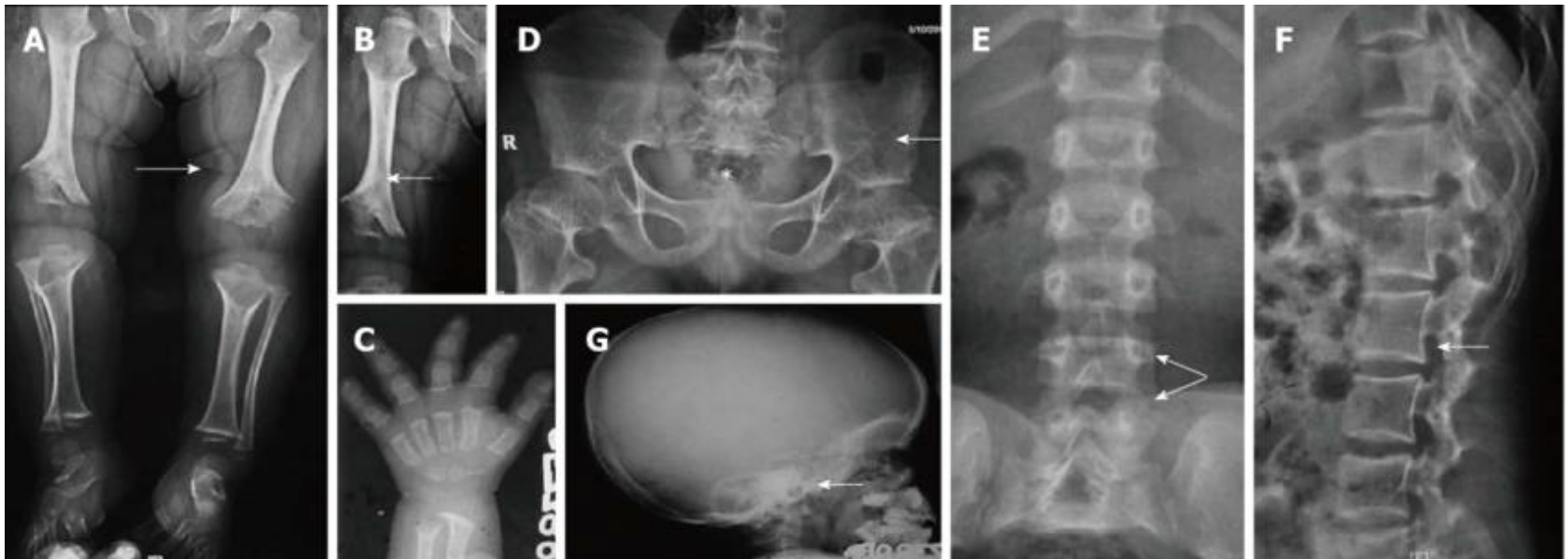
Eight-year-boy recovered from medulloblastoma 4 years ago. Treatment at that time consisted of chemotherapy & craniospinal irradiation. Child' clothes of same size for the last one year. On examination, height was below 3rd%, weight on 25th %. His upper: lower segment was 0.8:1. Which one of the following is the most likely cause of short stature?

a) Acquired growth hormone deficiency.
b) Chemotherapy-induced hypopituitarism.
c) Irradiation-induced spinal epiphyseal fusion.
d) Acquired central hypothyroidism.

Craniospinal irradiation

- Is part of treatment of medulloblastoma.
- Has profound effect on spinal growth.
- The younger the child is when given irradiation the greater the subsequent skeletal disproportion.
- Radiation induced growth hormone (GH) deficiency and spinal irradiation are two major adverse factors that contribute to the short adult height achieved by many patients treated for brain tumors in childhood.

Fourteen - year-old boy, who was growing well till 2 years ago, when his family noticed slow growth. He is an otherwise healthy child. On his examination, his height percentile dropped below 3rd. percentile in comparison to 25 %, 2 years ago. His Tanner's stage was prepubertal. Bone age is greater than two standard deviations below the chronologic age. Which one of the following, is the MOST likely cause of short stature?

a) Familial short stature.
b) Growth hormone deficiency.
c) Constitutional delay.
d) Hypogonadism.

Constitutional delay of growth and puberty (CDGP)

- Children with CDGP usually are growing at a low-normal rate (e.g., about 4 to 5 cm/year in preadolescent girls, and 3.5 to 4.5 cm/year in preadolescent boys).

- In addition to a low preadolescent height velocity, they tend to have delayed pubertal development.

- This leads to a marked height discrepancy during the early teenage years compared with their peers, but is followed by catch-up growth when they do enter puberty with normal final adult height.

- In many cases, there is a family history of delayed growth and puberty in one or both parents (sometimes described as being a "late bloomer").

Constitutional delay of growth and puberty
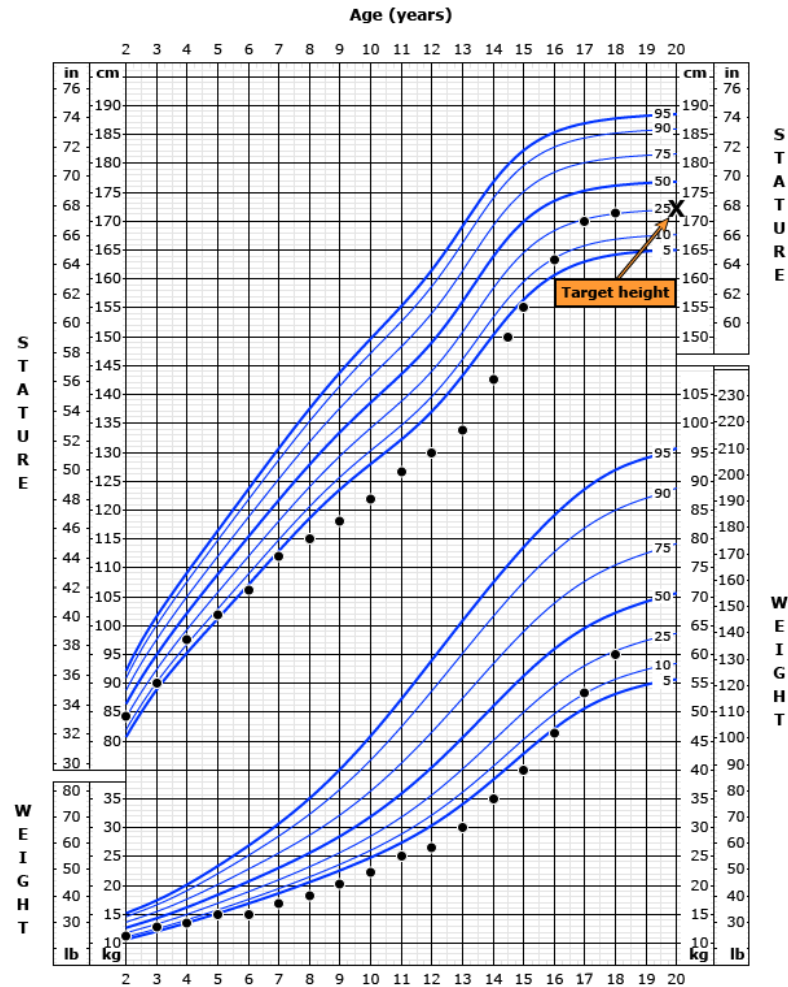
Which one of the following disorders of growth is characterized by normal body proportion ?

a) Achondroplasia.

b) Morquio's syndrome.

c) untreated congenital hypothyroidism.

d) Hypopituitarism.

Thank you