
Growth disorders
Growth disorders are problems that prevent children from developing normal height, weight, sexual maturity or other features.

Short stature

- Height less than 3rd percentile of the normal for age, or more than 2 SDs below the mean height for that age, sex and reference population.
- Height velocity less than 25th percentile for age,sex and reference population.

CAUSES OF SHORT STATURE *"IS NICE"*

- **I-** Idiopathic (Most common, constitutional delay, familial short stature)
  - Intrauterine (IUGR, TORCH, Fetal alcohol)
- **S-** Skeletal causes (dysplasia, osteogenesis imperfecta)
  - Spinal defects (scoliosis, kyphosis)
- **N-** Nutritional (under nutrition)
- - Nurturing (deprivation)
- **I-** Iatrogenic (steroids, radiation)
- **C-** Chronic disease
  - Chromosomal (Turner, Down's)
- **E-** Endocrine (GH deficiency, hypothyroidism)

Comparison

Feature	Familial Short Stature	Constitutional Short Stature
1) Sex	Both equally affected	More common in boys
2) Length at Birth	Normal	Normal (starts falling <5th centile in 1st 3yrs of life)
3) Family History	Of short stature	Of delayed puberty
4) Parents Stature	Short (one or both)	Average
5) Height Velocity	Normal	Normal
6) Puberty	Normal	Delayed
7) Bone Age & Chronological Age	BA = CA > Height Age	CA > BA = Height Age
8) Final Height	Short, but normal for target height	Normal

Under nutrition

One of the commonest cause of short stature in India

Protein Energy Malnutrition, anemia and trace element deficiency such as Zinc def are common causes

Weight gain is slow and muscles are wasted. Long standing malnutrition leads to Stunting

BA < CA

Diagnosis: good dietary history, anthropometric measurements

Psychosocial short stature:

- Aka: **emotional deprivation dwarfism, maternal deprivation dwarfism, hyperphagic short stature**
- Functional hypopituitarism- low IGF-1 levels & inadequate response to GH stimulation
- Type1- below 2 yrs, failure to thrive, no Gh deficiency
- Type2- in > 3 yrs
- Other behavioural disorders: enuresis, encorpresis, sleep & appetite disturbances, crying spasms, tantrums
- Dental eruptions & sexual development delayed

Intra-uterine Growth Restriction

- Arrest of fetal growth in early embryonic life causes reduction in total number of cells, leading to diminished growth potential in postnatal life
- BW <10th centile for gestational age
- Most of these babies show catch-up growth by 2yrs of age, but 20-30% may remain short
- Subtle defects in the GH-IGF axis are considered to be responsible
- Growth Velocity is normal
- BA = CA
- Learning disabilities could be present

Steps of assessment

1) ***Accurate height measurement***

- Plotted on an appropriate growth chart.
- Length is measured lying down and should be used for infants and children up to 24 months of age .
- Height is measured standing and should be used for children 2 years and above.

- Below 2 yrs- supine length with

  infantometer

- For older children- **Stadiometer**

3) ***Assessment of height velocity***

Rate of increase in height over a period of time, expressed as cm/year

If low – pathological cause of short stature

4) ***Comparison with population norms***

Height plotted on appropriate growth charts & expressed as centile or SD score

5) ***Comparison with child's own genetic potential***

Mid parental height for boys

= mother's height + father's height /2 + 6.5cm

Mid parental height for girls

= mother's height + father's height /2 – 6.5cm

6) ***Sexual maturity rating (SMR):***

- Also known as *Tanners stages*
- Used in older children
- Total 5 stages included in each gender

DIAGNOSIS

DETAILED HISTORY

CAREFUL EXAMINATION

LABORATORY EVALUATION

Clues to etiology from history

History	Etiology
History of delay of puberty in parents	Constitutional delay of growth
Low Birth Weight	SGA
Neonatal hypoglycemia, jaundice, micropenis	GH deficiency
Dietary intake	Under nutrition
Headache, vomiting, visual problem	Pituitary/ hypothalamic SOL
Lethargy, constipation, weight gain	Hypothyroidism
Polyuria	CRF, RTA
Social history	Psychosocial dwarfism

Pointers to etiology of short stature

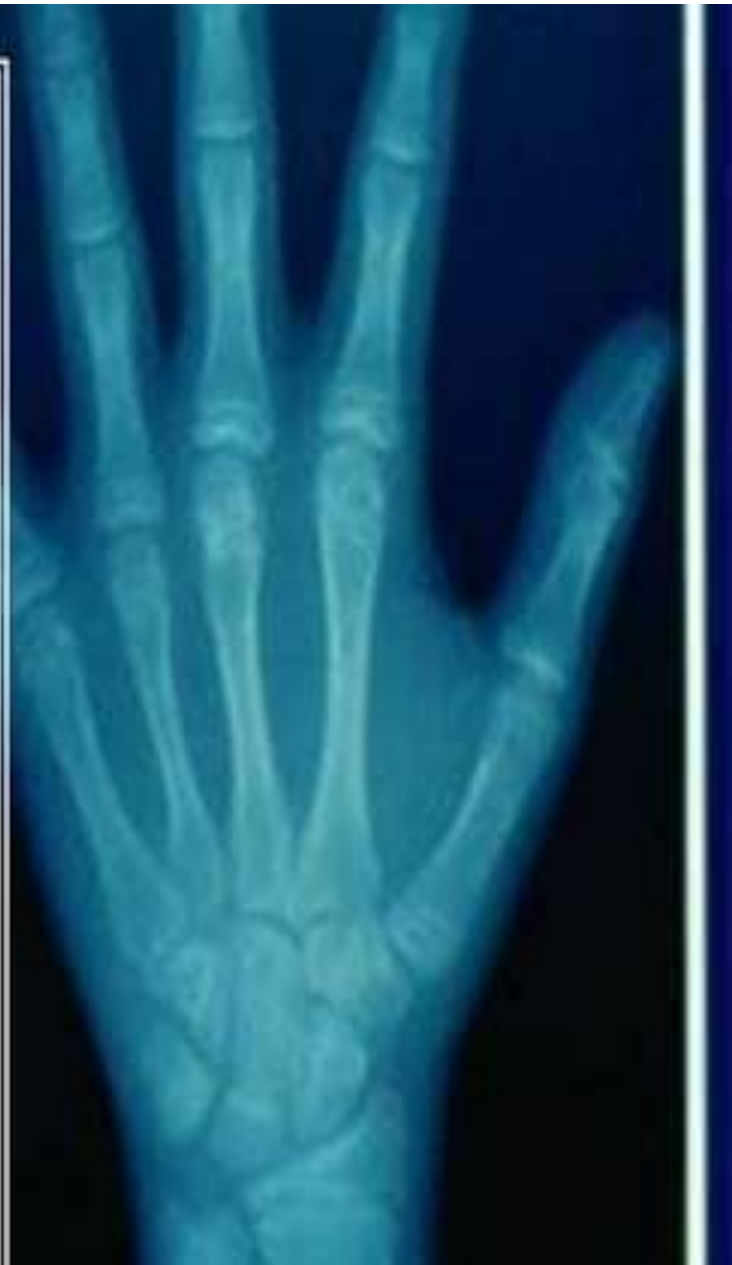
Pointer	Etiology
Midline defects, micropenis, Frontal bossing, depressed nasal bridge, crowded teeth,	GH deficiency
Rickets	Renal failure, RTA, malabsorption
Pallor	Renal failure, malabsorption, nutritional anemia
Malnutrition	PEM, malabsorption, celiac disease, cystic fibrosis
Obesity	Hypothyroidism, Cushing syndrome, Prader Willi syndrome
Metacarpal shortening	Turner syndrome, pseudohypoparathyroidism
Cardiac murmur	Congenital heart disease, Turner syndrome

Clues to etiology from examination

Examination finding	Etiology
Disproportion	Skeletal dysplasia, rickets, hypothyroidism
Dysmorphism	Congenital syndromes
Hypertension	CRF
Goitre, coarse skin	Hypothyroidism
Central obesity, striae	Cushing syndrome

BONE AGE

- Bone age assessment should be done in all children with short stature
- Appearance of various epiphyseal centers & fusion of epiphyses with metaphyses tells about the skeletal maturity of the child
- Conventionally read from Xray of hand & wrist using **Gruelich-Pyle** atlas or **Tanner- Whitehouse** method



Bone age gives an idea as to what proportion of adult height has been achieved by the child & what is remaining potential for height gain

BA is delayed compared to chronological age in almost all causes of short stature

Exceptions: Familial short stature,

Precocious puberty


Short stature*
Detailed medical history and physical examination
Dysmorphism or disproportionate short stature
Proportionate short stature without dysmorphism
Turner syndrome (Girls)
Down' syndrome
Noonan's syndrome
Prader Willi syndrome
Russel Silver syndrome
SHOX deficiency syndromes
Skeletal dysplasias
Mucopolysaccharidoses
Others
Systemic illnesses
Malabsorption syndromes
Chronic infections/ inflammatory conditions
Chronic renal disease
Chronic liver disease
Pseudohypoparathyroidism
Rickets
Step 1 Investigations
CBC, Albumin
ESR, Tuberculin test, Chest X-ray
Serum creatinine
Liver function tests
Serum calcium, PO4, PTH, Radiology
Normal test results
Birth weight and/or length <-2SD
Small for gestational age with failure to catch up
Endocrine disorders
Hypothyroidism
Turner syndrome (Girls)
Distal renal tubular acidosis
Celiac disease
Cushing's syndrome
Step 2 Investigations
Free Thyroxine, TSH
FSH, Karyotype
Blood gas analysis
IgA, IgA tTG
Basal Cortisol, LDDST
Normal test results and BA>CA
Ht SDS within target Ht SDS
Familial short stature
IGF deficiency syndromes
Growth hormone insufficiency/ insensitivity
Step 3 Investigations
IGF1, IGFBP3,
GH stimulation tests
MRI pituitary
Normal test results and BA=HA<CA
Similar pubertal tempo in parents
Constitutional delay in growth and puberty
Normal tests

Management

- Counselling of parents
  (for physiological causes)
- Dietary advice
  (Undernutrition, Celiac disease, RTA)
- Limb lengthening procedures
  (skeletal dysplasias)
- Levothyroxine (In Hypothyroidism)
- GH s/c injections (GH deficiency, Turner syndrome, SGA, CRF prior to transplant)
- Monitoring with regular & accurate recording of height is mandatory for a good outcome in any form of therapy

Failure to thrive

- Failure to thrive (FTT) is a chronic, potentially life threatening disorder of infants and children who fail to gain and may even lose weight. Children are considered as failing to thrive when their rate of growth does not meet the expected growth rate for a child of their age. More specifically, the term characterized those whose weight is below the 3rdor 5th percentile on an appropriate growth chart.

CLASSIFICATION

Traditionally, classified as

1.Organic FTT

- Secondary to underlining medical illnesses
- Account for less than 20% of cases

2.Nonorganic FTT (NOFT)

- Psychosocial FTT
- No known medical condition that causes poor growth
- Inadequate food or undernutrition
- Accounts for over 70% of cases

Table 2.19: Causes of failure to thrive

Organic

Gastrointestinal: Gastroesophageal reflux, malabsorption, inflammatory bowel disease, pyloric stenosis

Neurological: Mental retardation, cerebral palsy

Renal: Renal tubular acidosis, chronic kidney disease

Cardiopulmonary: Congenital heart disease, cystic fibrosis, asthma

Endocrine: Hypothyroidism, diabetes mellitus

Infections: Chronic parasitic infections of gastrointestinal tract, tuberculosis, human immunodeficiency virus

Genetic: Inborn errors of metabolism, chromosomal anomalies

Miscellaneous: Lead poisoning, malignancy

Nonorganic

Poverty

Misperceptions or lack of knowledge about diet and feeding

Lack of breastfeeding, feeding diluted formulae

Dysfunctional parent child relationship

Clinical features

Most common presentation is poor growth

Accompanied by ***physical signs;***

- Alopecia
- Reduced subcutaneous fat or muscles Dermatitis
- Syndromes of marasmus or kwashiorkor

- Neglect of hygiene
- Delays in social and speech development

  Expressionless face and hypotonic
- Recurrent infections
- Depending on the severity infants with FTT may exhibit
  - Thin

    extremities

    Narrow face
  - Prominent ribs and wasted buttocks

LABORATORY EVALUATION

LABORATORY EVALUATION FOR ORGANIC DISEASE SHOULD BE GUIDED BY THE SIGNS AND SYMPTOMS FOUND IN THE INITIAL EVALUATION.

A CAREFUL HISTORY AND PHYSICAL EXAMINATION IN THE CHILD WITH FAILURE TO THRIVE (FTT) MAY SUGGEST CLUES TO AN ***ORGANIC DISEASE***

LABORATORY STUDIES THAT ARE NOT SUGGESTED ON THE BASIS OF THE INITIAL HISTORY AND EXAMINATION ***RARELY*** ARE HELPFUL.

ONE STUDY REVEALED THAT ONLY **1.4 %** OF THE LABORATORY STUDIES PERFORMED IN EVALUATING CHILDREN WITH FTT WERE USEFUL DIAGNOSTICALLY

Simple routine tests

- ✓ Random Blood Sugar(RBS)
- ✓ complete blood count(CBC)
- ✓ Urinalysis(U/A)
- ✓ electrolyte levels
- ✓ stool exam
- ✓ PIHCT
- ✓ TB

Abnormalities of head and shape

Macrocephaly

- Definition: Head circumference (occipito frontal)
- > 2 standard deviation(SD) above the mean for age and sex.

Hydrocephalus

- Pathological increase in ventricular volume due to abnormal CSF accumulation
- Imbalance between CSF production and flow leading to ventricular enlargement

Table 2.21: Causes of macrocephaly

Megalencephaly

Benign familial
Neurocutaneous syndromes: Neurofibromatosis, tuberous sclerosis, Sturge-Weber, Klippel-Trenaunay-Weber, linear sebaceous nevus
Leukodystrophies: Alexander, Canavan diseases; megalencephalic leukoencephalopathy
Lysosomal storage diseases: Tay-Sachs disease, mucopolysaccharidosis, gangliosidosis
Others: Sotos disease, fragile X syndrome

Increased cerebrospinal fluid

Hydrocephalus
Benign enlargement of subarachnoid space
Hydranencephaly, choroid plexus papilloma

Enlarged vascular compartment

Arteriovenous malformation
Subdural, epidural, subarachnoid or intraventricular hemorrhage

Increase in bony compartment

Bone disease: Achondroplasia, osteogenesis imperfecta, osteopetrosis, hyperphosphatasia, cleidocranial dysostosis
Bone marrow expansion: Thalassemia major

Miscellaneous causes

Intracranial mass lesions: Cyst, abscess or tumor

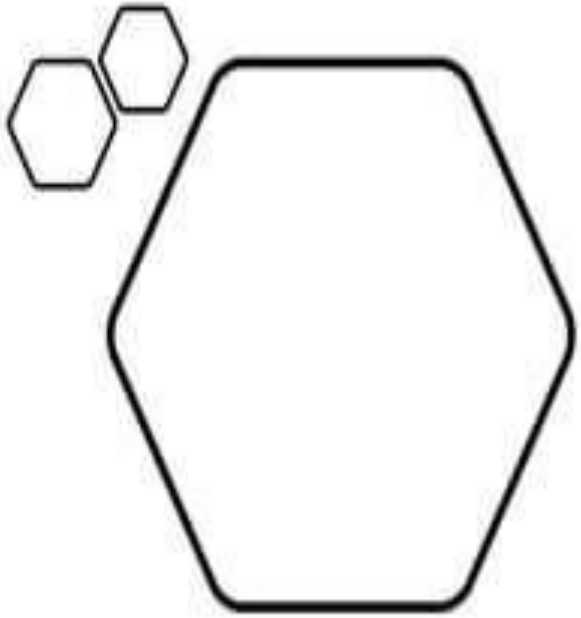
Clinical Features

- 50% may be asymptomatic

Symptoms

- Vomiting, headache
- Drowsiness
- Failure to thrive, poor appetite
- Shrill cry, irritability, lethargy
- Delayed motor milestones – mainly motor
- Progressive enlargement of head
- Abnormal shape of head – inverted triangle
- Slow mental deterioration

Investigations



- USG: when ant fontanelle is open. Assesses ventricular size, detects IVH
- Plain skull films- shows sign of ICP:
  - separation of sutures
  - erosion of the post. Clinoid process
- Increased convolutional markings (beaten silver appearance)
- Flat enlarged sella tursica

Investigation:

- CT:helps to identify the cause
- MRI:better visualization of post fossa pathologies
- Opthalmological evaluation
- Psychomotor assessment:using different dev scales

TREATMENT

- Generally require no tx
- Infants with hydrocephalus may require neurological intervention(e.g. placement of a ventriculo-peritoneal shunt).

MICROCEPHALY

- **Definition**: Head circumference < 3 SD below the mean for age, sex and gestation.

- Types :
- Primary (Genetic)

- Secondary (non-genetic)

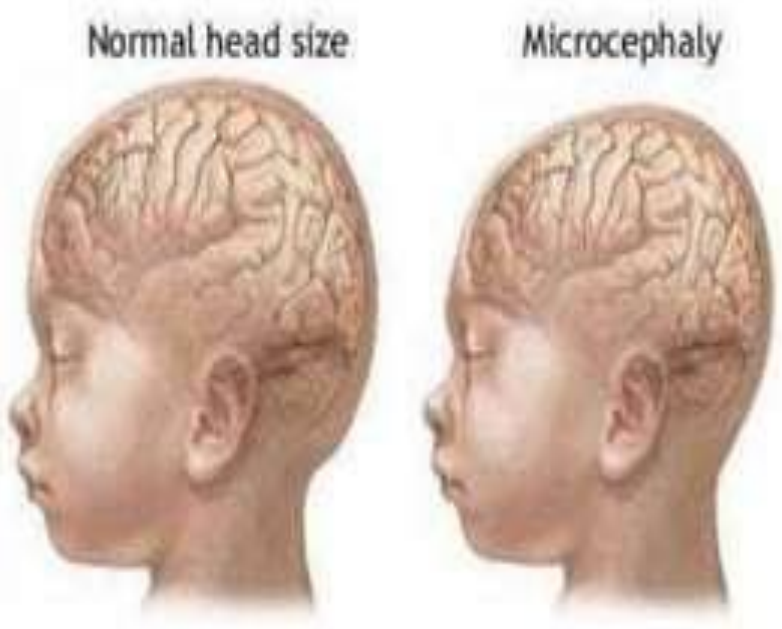


Table 2.22: Causes of microcephaly

Isolated microcephaly

Autosomal recessive, autosomal dominant or X-linked

Syndromic

Trisomies 21, 18, 13
Monosomy 1p36 deletion
Syndromes: William, Cri-du-chat, Seckel, Smith-Lemli-Opitz, Cornelia de Lange, Rubinstein-Taybi, Cockayne, Angelman

Structural diseases

Neural tube defects (anencephaly, hydranencephaly, encephalocele, holoprosencephaly)
Lissencephaly, schizencephaly, polymicrogyria, pachygyria

Metabolic disorders

Phenylketonuria, methylmalonic aciduria, citrullinemia
Neuronal ceroid lipofuscinosis
Maternal: Diabetes mellitus, untreated phenylketonuria

Infections

Congenital: Cytomegalovirus, herpes simplex virus, rubella, varicella, toxoplasmosis, HIV, syphilis, enterovirus
Meningitis

Teratogens

Alcohol, tobacco, marijuana, cocaine, heroin, toluene
Antineoplastic agents, antiepileptic agents
Radiation

Perinatal insult

Hypoxic ischemic encephalopathy, hypoglycemia

Endocrine

Hypothyroidism, hypopituitarism, adrenal insufficiency

APPROACH

- History (perinatal – family history)
- Examination – dysmorphic features – malformations
- Development
- Growth – serial measurements of HC

- **INVESTIGATIONS**
- Baseline biochemistry, metabolic screen
- Genetic testing – karyotype, molecular genetics
- TORCH screen
- Ophthalmology
- MRI brain

Management

- No treatment for microcephaly
- Baby's head cannot be returned to a normal size & shape
- According to the cause
  - Anticonvulsants
  - Physiotherapy
  - Hearing and speech therapy
  - Dietary management for failure to thrive
  - Genetic counseling

CRANIOSYNOSTOSIS

- **Definition**: premature fusion of one or more cranial sutures, either major(e.g metopic, coronal, sagittal, and lambdoid) or minor(frontnasal, temporosquamosal, and frontosphenoidal).

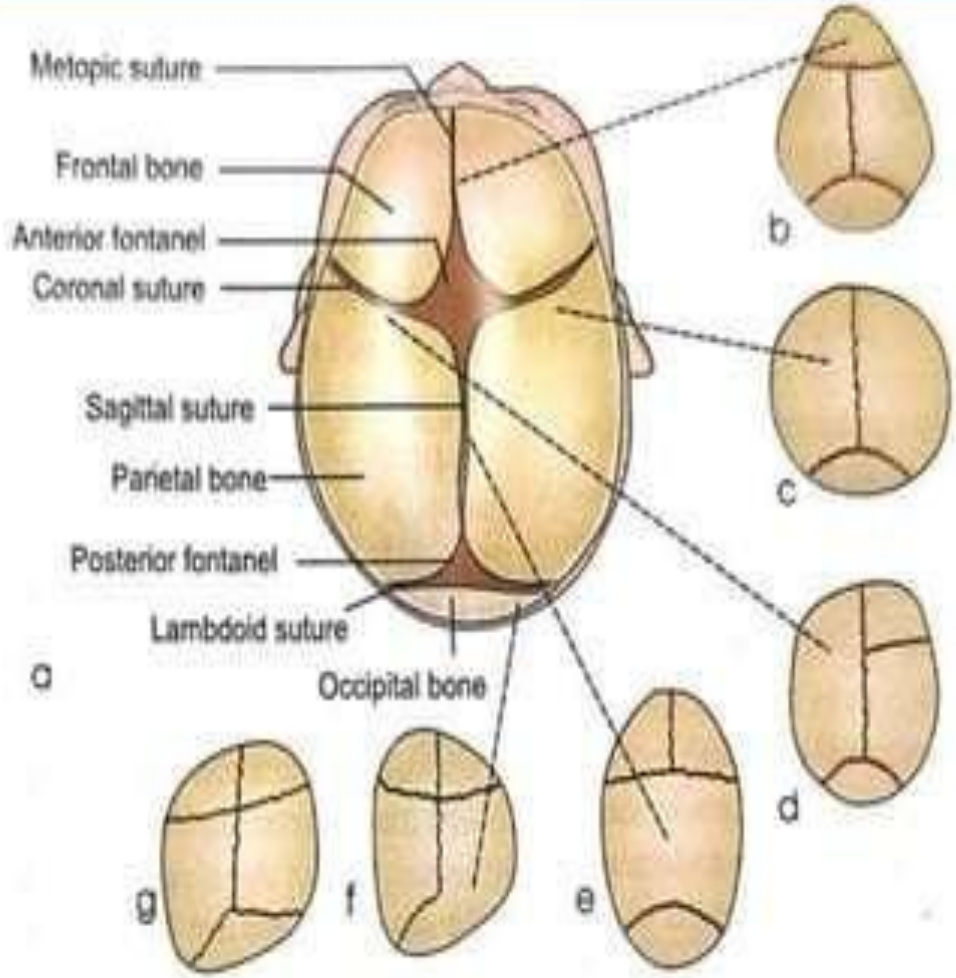


Fig. 2.23: (a) Head of normal neonate showing fontanels and sutures. The common forms of craniosynostosis (secondary to premature fusion of sutures) include: (b) Trigonocephaly (metopic suture); (c) Brachycephaly (bilateral coronal sutures); (d) Left anterior plagiocephaly (left coronal suture); (e) Scaphocephaly (sagittal suture); (f) Right posterior plagiocephaly (right lambdoid

DEFORMITIES OF SKULL

- Plagiocephaly
- Scaphocephaly
- Trigonocephaly
- Turencephaly
- Brachycephaly

Diagnosis

Palpation of suture reveals prominent bony ridge.

Fusion may be confirmed by x-ray skull

Associated syndromes – Crouzon , Alperts, Carpenter

Management

Premature fusion of single suture rarely causes any neurological deficit . Thus, in this situation the only indication is cosmetics.

2 or more suture fusion – more complications eg. ↑ ICT, hydrocephalus, optic atrophy, DNS, choanal atresia --- operative surgery essential – craniectomy with craniofacial correction.

Usually good prognosis with non syndromic infants.....


Thank u