General Paediatrics

Marrow ss
Paediatrics 2024

General Paediatrics Index

NORMAL GROWTH

Growth phases

00:00:16

Periods of growth :

1. Prenatal period : Growth before birth/intrauterine period.
   - Fertilized ovum/zygote : First 2 weeks of gestation.
   - Embryo : 2 to 8 weeks of gestation.
   - Fetus : 9 weeks of gestation.
2. Postnatal period :
   - Newborn : First 4 weeks.
     Early newborn : First week.
     Late newborn : Next 3 weeks.
   - Infancy : First year.
   - Toddler : 1 to 3 years.
   - Preschool : 3 to 6 years.
   - School age : 6 to 12 years.
   - Adolescent : 10 to 19 years.

Adolescent age group :

Early : 10 to 13 years.
Mid : 14 to 16 years.
Late : 17 to 19 years.

Pubertal changes :

Puberty occurs earlier in females (8 to 13 years) than males (9 to 14 years).

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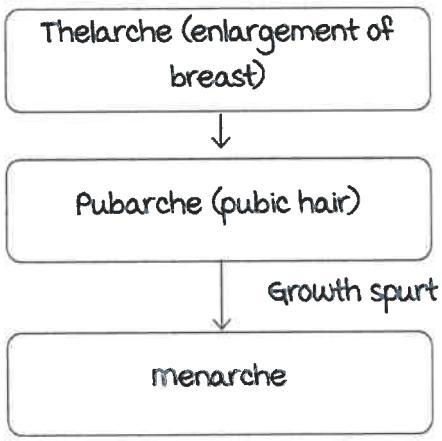
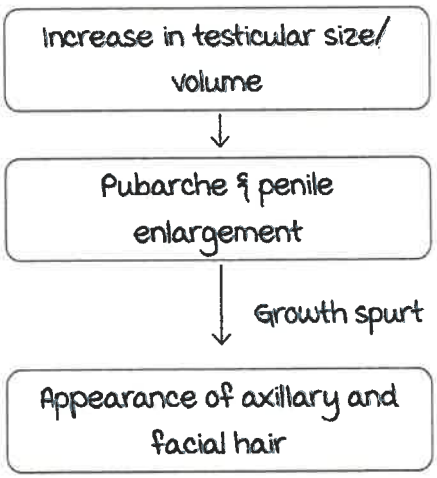
Sequence in females :



Sequence in males :

Assessment of testicular size is by Praders orchidometer.



Changes during the middle part of puberty in males :

- Spermarche (production of sperms).
- Voice cracking/deepening seen.

Assessment of puberty :

Tanner's SMR (Sexual maturity rating).

Done separately in males and females.

In males : Testis, penis, pubic hair, and scrotum.

In females : Breast and pubic hair appearance.

From stages I (prepubertal) to stages V (adult like appearance).

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Tanner's staging in females : Breast appearance.

Stage I : No changes in the breast.

Stage 2 : Appearance/prominence of breast buds.

Stage 3 : Generalized swelling of the breasts (beyond nipple and areola).

Stage 4 : Nipple + areola forming a second mount over the breast.

Stage 5 : Mature/adult type appearance, where the nipple protrudes forward, and areola retracts.

Pubic hair appearance :

Stage I : No pubic hairs.

Stage II : Straight, sparse hairs.

Stage III : Increase in number and curling of hairs.

Stage IV : Dense pubic hair (curly).

Pigmentation starts increasing from stage II to IV.

Stage V : Pubic hairs extends into medial part of thigh region.

Tanner's staging in males :

Pubic hair changes are similar to females.

Testicular volume is represented as number.

Size of the testis is mentioned.

Stage I : Testis size < 2.5 cm and volume < 4 ml.

Stage II : Testis size ≥ 2.5 cm and volume ≥ 4 ml.
Scrotum size increases, skin over scrotum is red in color.

Stage III : Further increase in volume of testis, penis lengthens, and scrotal skin is dark.

Stage IV : Increase in length and breadth of the penis.

Stage V : Volume of testis > 20 ml (adult like appearance).

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Growth patterns

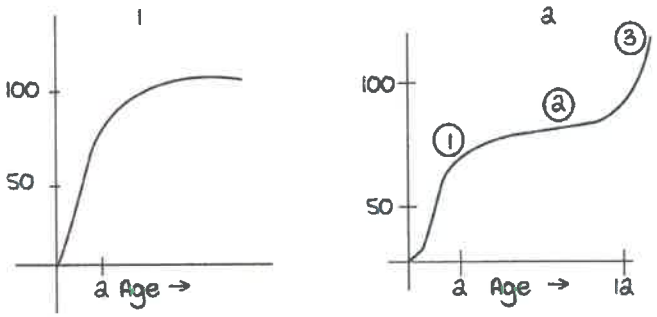
00:15:10

Rules :

Growth is in Cephalocaudal progression.

Growth occurs from the distal to proximal direction.

Not all tissues in the body grow at the same rate.



Graph 1 : Till 2 years there is steep increase followed by minimal growth (parabola). Depicts growth of brain.

Graph 2 : Depicts somatic growth.

Sigmoid growth.

Phases : Rapid growth (1 & 3) : Growth spurts.

Phase 3 : Pubertal growth spurt.

Pubertal growth spurts occur early in females.

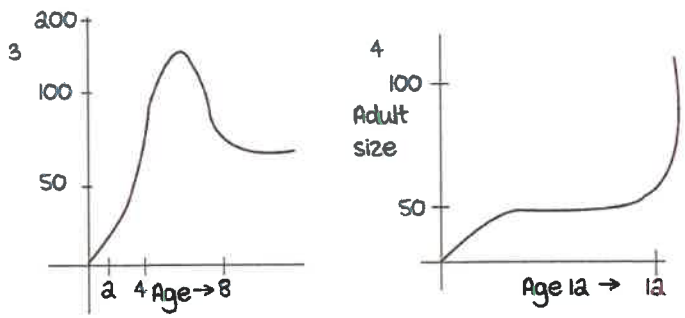
Increase in height is around 16 to 28 cm in females.

Increase in height is 20 to 30 cm in males.

Growth spurts :

Female : Tanner's stage 3.

Males : Tanner's stage 4.



Graph 3 : Peak (4–8 years) is 2 times of adult size. Come back to normal size after 8 years.

Eg : Lymphoid tissues like LN, tonsils, and adenoids.

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Between 4 to 8 years : Physiological lymphoid hyperplasia.

Graph 4 : Increase in size after 10 to 12 years.
Stage of gonadal growth.

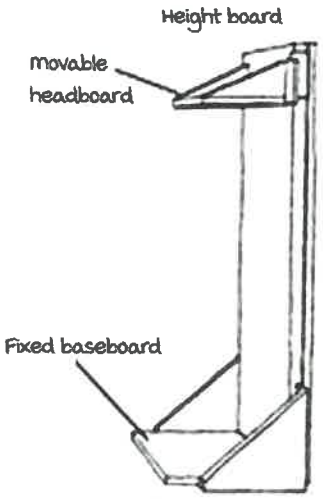
Assessment of growth 00:23:34

Anthropometry : Measurement of growth parameters.
Average birth weight in India : 2.9 Kg.
LBW is defined as weight < 2.5 Kg.
During first week, there is 10% of weight loss in term babies.
For preterm babies : Up to 15% of weight loss.
This is called as physiological weight loss.

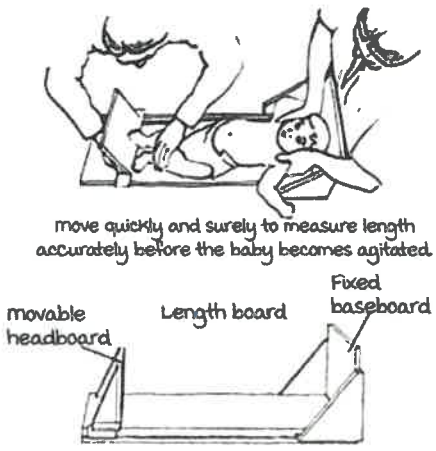
Regaining of birth weight : 10 (term) to 14 days (preterm).
Best way of assessment : Growth charts.

Birth weight doubles by 5 to 6 months.
Birth weight triples by 1 year.
Birth weight quadruples by 2 years.
Height/length assessment :
Length : Measured in supine posture (up to 2 years).
Measured by stadiometer (height) or infantometer (length).

Staiometer



Infantometer



Normal length at birth : Approximately 50 cm.
In the first year : Increase of 25 cm. So, at 1 year : 75 cm.
In the second year : Increase of 10 to 15 cm. So, at 2 years :

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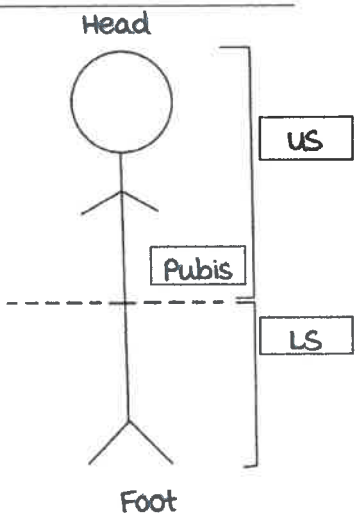
85 to 90 cm.
There is a steady increase of 6 cm/year till 12 years.
After 12 years (pubertal growth spurts) :
Females : 8cm/year.
Males : 10 cm/year.

Height doubles by 4 years.
Height triples by 12 years.

Half of adult's height in a normal child (80-85 cm) : Around 2 years (18 to 24 months).

Body proportions 00:32:28



Upper Segment (US): Lower Segment (LS).
At birth it is 1.7 : 1
At 3 years it is 1.3 : 1
At 7 to 10 years it is 1 : 1.
After >10 years it is 0.9 : 1.

They are useful in the evaluation of short stature.
In tall children : Arm span.

At birth : Height > Arm span (difference is 2.5 cm).
At 11 years : Height = Arm span.
>11 years : Arm span > Height (difference is 1 to 2 cm).

Circumferences 00:36:50

Head circumference (HC) : Also called as occipitofrontal circumference.
It indicates brain growth.

At birth : Approximately 34 cm (33-35 cm).

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First 3 months after birth : Increase of 2 cm/month.
At 3 months : 40 cm.
Next 3 months : Increase in 1 cm/month. At 6 months : 43 cm.
Next 6 months : Increase in 0.5 cm/month. At 12 months : 46 cm.
Next 1 year : Increase in 2 cm. At 2 years : 48 cm.
At around 12 years : 52 cm.

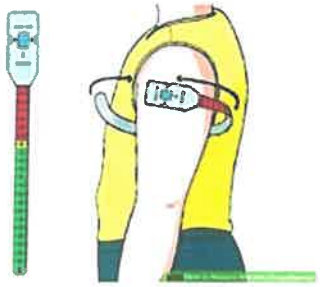
Chest circumference (CC) : measured at the level of nipples midway between inspiration and expiration.

Chest circumference

Birth : HC > CC (3cm).
1 year : HC = CC.
> 1 year : CC>HC.

Mid arm circumference :

Mid arm circumference



measured in children < 5 years.
It is useful for community assessment of growth, because it can be done by health workers.

Shakir's tape :
Midpoint of the arm : Mid point between 2 bony prominences.
Green : Nutrition is normal.
Yellow : Malnutrition.

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Red : Severe malnutrition (≤ 11.5 cm).

Skin fold thickness measurement :
Thickness of subcutaneous fat is measured.
It is an indicator of nutritional status in children.
It is done by Harpenden's caliper.

≥ 10 mm : Normal.
< 6 mm : Severe malnutrition.

Charts for skin fold thickness : Interpreted as percentile value.
< 5th percentile : Low value (malnutrition).

Growth charts

00:46:06

WHO growth is most preferred and used commonly.
In India, between 0 to 5 years : Only WHO charts are used.
5 to 19 years : WHO or IAP charts can be used.

WHO charts can be used as international comparison tools.
Types of WHO charts :

- Weight/age charts.
- Weight/height charts.
- Height/age charts.
- HC for age charts.
- MAC for age charts.
- BMI charts (weight in kg/height in m^2) : Used in > 5 years of age.

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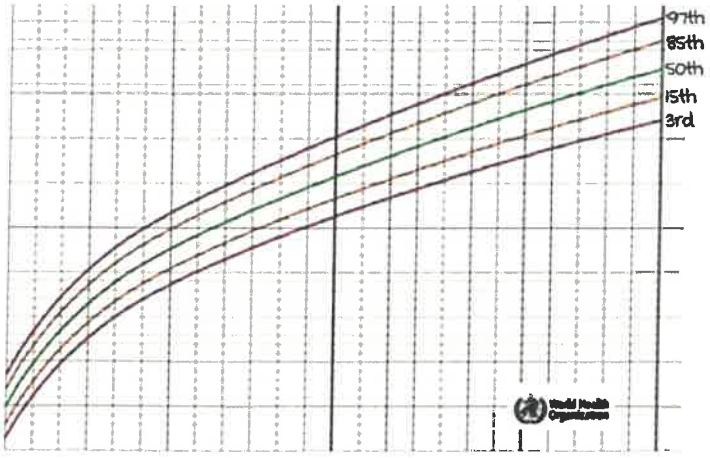
Best tool for assessment in < 5 years : Weight / Height chart.



Growth chart in

Blue colour : Boys.

Pink colour : Girls.

In a height for age chart,

Major percentiles are recorded.

50th percentile : mean value.

High value (tall stature) : Value above 97th percentile.

Low value (short stature) : Below 3rd percentile.

Between 97th percentile and 3rd percentile : Normal values.

Standard deviation / Z scores : Deviation from mean value.

Eg : 97th percentile (+2) SD

85th percentile is (+ 1) SD.

15th percentile is (- 1) SD.

3rd percentile is (- 2) SD.

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ABNORMALITIES IN HEAD SIZE AND HEAD SHAPE

Microcephaly (small head) :
Based on head circumference for age chart, head circumference < -3SD is considered as microcephaly.
This is an exception to general rule : Considering < -2SD in the definition would mean including normal children as well.

Primary/Genetic microcephaly :
Seen due to any insult during brain development.
Refers to anomalies affecting the brain like anencephaly or neuronal migration disorders.
Genetic syndromes like down's Syndrome (trisomy 21).

Familial microcephaly : Small head size runs in the family members. Usually inherited as autosomal recessive.

Secondary microcephaly (normal brain development) :
Acquired microcephaly : Extrinsic factors cause microcephaly.
Extrinsic factors include :

- Maternal factors :
    1. Infections during intra uterine life.
       Example : TORCH infection.
    2. Radiation during Ist trimester (impaired organogensis).
    3. Toxins (drugs/chemicals) taken durig pregnancy.
    4. Teratogens like phenytoin or alcohol.
- Perinatal factor like birth asphyxia.
- Post natal factors :
  Trauma or infection in the child up to the age of 2 years (active brain growth occurs till 2 years of age).
- Metabolic disorders (metabolite accumulation in brain) :
    1. Phenylketonuria.
    2. Lysosomal storage disorders :

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Hepatosplenomegaly and low IQ are seen.

a. Gaucher disease.

b. Niemann pick disease.

Both have autosomal recessive inheritance.

Gaucher's disease	Niemann pick disease
Deficiency in glucocerebrosidase enzyme	Deficiency of sphingomyelinase
Can also affect the bones : Causes expansile lytic lesions also called as Erlenmeyer flask deformity and pancytopenia (defective bone marrow).	Does not involve the bones
No cherry red spots.	Cherry red spots seen in macula of eye.
Accumulation of glucocerebrosides as typical cytoplasmic inclusions called wrinkled/crumpled tissue paper appearance on biopsy.	Accumulation of sphingomyelin gives foamy appearance of cells.

Late onset syndromes associated with microcephaly :

- Rett syndrome (acquired microcephaly).
- Angelman Syndrome.
- Seckel's Syndrome.

Rett syndrome 00:12:56

X-linked dominant disorder.

- Genetic disorder characterized by defect in MECP2 gene.
- From birth till 1 year of age, head circumference is normal. After 1 year, deceleration of brain growth results in microcephaly.
- Developmental regression :
  Up to 1 year of age, the child achieves normal milestones but after 1 year of age, the child loses previously acquired milestones (eg : stand without support in 1 year but not able to, even with support at 1.5 years).

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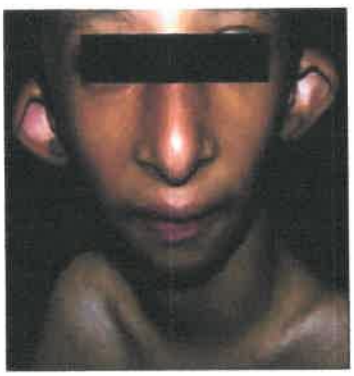
- Stereotypies : Characteristic hand wringing purposeless repetitive movements in the midline.
- Speech defects.
- Ataxia (worsen over time and adolescents become wheelchair-dependent.


Stereotypies

Seckel syndrome :

- Bird headed dwarfism.
- Beak like nose.


Seckel syndrome

Q. A child with microcephaly, low IQ, seizures and hypopigmented hair is being evaluated. The child is born out of consanguinous marriage. Which of the following is the likely cause of microcephaly?

- Familial microcephaly.
- Inborn error of metabolism.
- TORCH infections.
- Neural tube defect.

Diagnosis : Phenyl Ketonuria.

Phenyl Ketonuria :

Most common amino acid metabolic disorder.

Autosomal recessive in inheritance.

Normally, phenylalanine $\xrightarrow[\text{hydroxylase}]{\text{Phenylalanine}}$ Tyrosine $\longrightarrow$ melanin

Phenylketonuria $\longrightarrow$ deficient/absent phenylamine hydroxylase $\longrightarrow$ deficient tyrosine $\longrightarrow$ deficient melanin $\longrightarrow$ hypopigmentation $\longrightarrow$ fair skin, blonde hair, blue eyes.

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Increasing phenylalanine ⟶ converted to metabolites like phenylactate, phenylpyruvate & phenylacetate.

↓

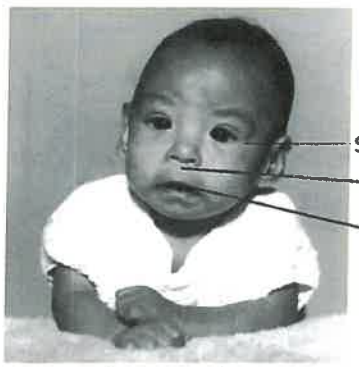
Brain damage and microcephaly in these patients.

musty/mousy odor of the urine ⟶ due to phenylacetate excretion.

management :

- Restrict diet containing phenylalanine.
- Enhance activity of phenylalanine enzyme co-factor, via synthetic BH4 (tetrahydrobiopterin) called as sapropterin.

Q. Phenotypic appearance of two infants with microcephaly is shown below. Identify the possible teratogen incriminated in these scenarios ?



Fetal alcohol syndrome

Philtrum : Area between nose & mouth. Normally, markings/ impressions are seen.
In fetal alcohol syndrome, there is increased risk of heart defects (Ventricular Septal Defect is common).

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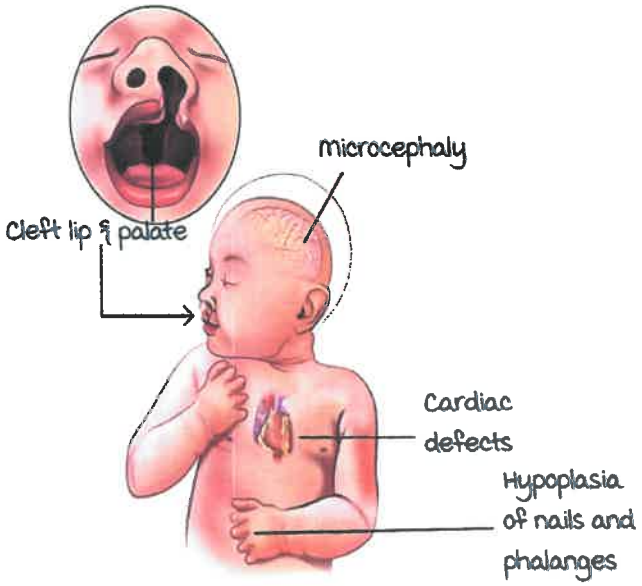

Fetal hydantoin syndrome :

- Phenytoin (diphenylhydantoin) exposure.
- Cleft lip/palate.
- Heart defects (commonly VSD).
- Small hands/feet.
- Mid facial hypoplasia (mid face structures smaller when compared to other structures in face).

Macrocephaly :

Head circumference > +2SD for the age.

Causes of macrocephaly :

- Increased amount of CSF :
    1. Hydrocephalus.
    2. Hydranencephaly : Absent brain tissue is replaced by sacs containing fluid (CSF). Transillumination (illiminate upwards from below the skull) is positive.
- Increased bleeding within skull :
    1. Sub dural hemorrhage.
    2. Extra dural hemorrhage.
    3. Intraventricular hemorrhage.
    4. Sub arachnoid hemorrhage.
- Increase in size of bony compartment :

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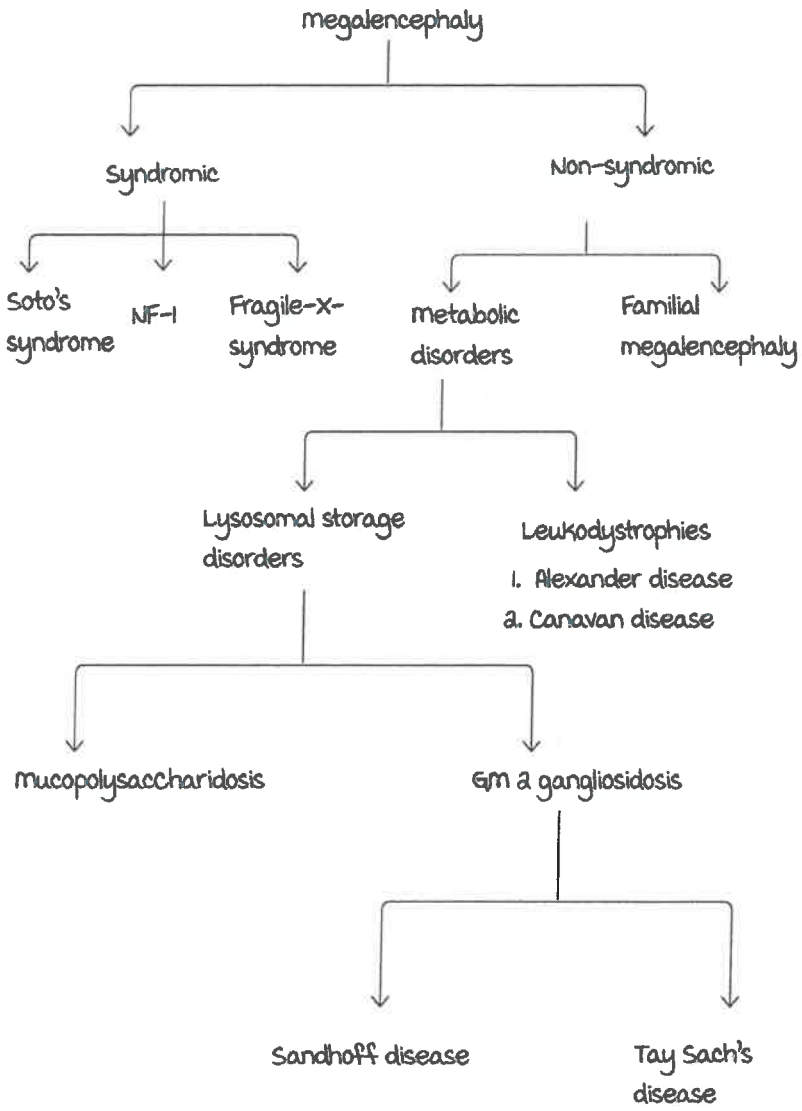
Associated with primary bone disorders like :

1. Achondroplasia.
2. Osteogenesis imperfecta.
3. Abnormal skull expansion : Beta thalassemia major (skull also involved in erythropoiesis).

- Increased brain tissue :
  1. megalencephaly



Soto's syndrome (cerebral gigantism) : Increase in head size and height are noted.

Fragile X Syndrome (CGG trinucleotide repeat) :

- Low IQ.
- Macro-Orchidism.
- Elongated face and large ears.

NF-1 (Neurofibromatosis -1) : Neurocutaneous syndrome.

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Familial megalencephaly :
Normal IQ and normal development seen. All family members have large heads.

Metabolic disorders : Patients have low IQ and delayed development.

↓

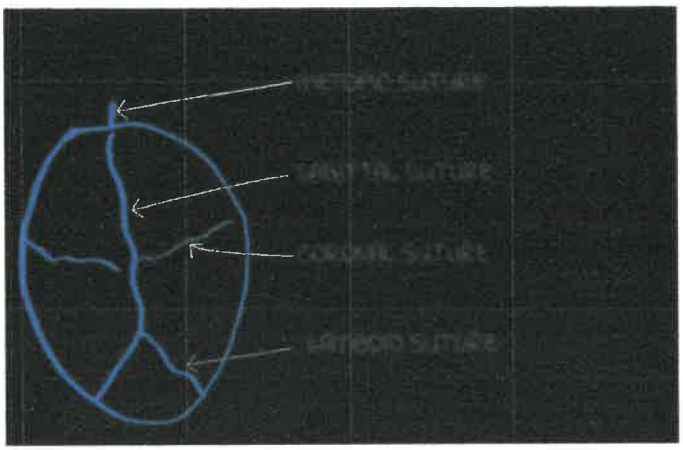
Tay Sach's disease : Hexosaminidase A deficiency.
No hepatosplenomegaly is seen.
Sandhoff disease : Hexosaminidase A and B deficiency.
Hepatosplenomegaly is seen.

Both Tay Sach's and Sandhoff disease have cherry red spots in the macula.

Craniosynostosis 00:37:12

Synostosis : fusion, Cranio : skull.
In this condition, there is premature fusion of sutures.
Growing brain needs space ⟶ skull expands in an irregular manner.

Whenever the suture fuses, the skull elongates along the direction of the fused suture.
most common suture to undergo premature fusion : Sagittal suture.



Active space