1. GROWTH

ANTHROPOMETRIC MEASUREMENTS

00:01:30

Weight

- Average birth weight of an Indian baby: 2.9 kg
- Birth weight:
  - Doubles at: 5 months
  - Triples at: 1 year
  - Quadruples at: 2 years

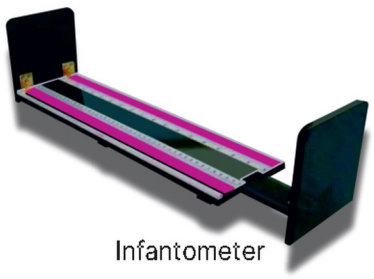
Rate Of Weight Gain

Age Group	Weight Gain per Day	Weight Gain per Month
1st 3 months	30 g/day	~ 900 g/month
3-6 months	20 g/day	~ 600 g/month
6-9 months	15 g/day	~ 450 g/month
9-12 months	12 g/day	–
1-3 years	8 g/day	–

Length / Height

00:03:42

- Infantometer: Used for children < 2 years old to measure supine or recumbent length.
- Stadiometer: Used for measuring standing height.



Infantometer

Stadiometer Requirements

- No footwear.
- No headgear.
- Occiput, shoulder, buttocks, and heel must touch the rod behind.
- Looking at the Frankfurt plane.

Important Information

- Standing Height is 0.7 cm less than supine or recumbent length due to gravity.

Usual Length Or Height

NEET PG 2021, Neet PG 2023

- At birth: 50 cm
- At 1 yr: 75 cm (An increase of 25 cm from birth)
- At 2 yrs: 90 cm
- At 4-4.5 yrs: 100 cm

Percentage Increase

- Length increases by 50% in the 1st year of life.
- Length increases by 100% in 4-4.5 yrs.
- Maximum growth takes place in the 1st year of life, followed by puberty.

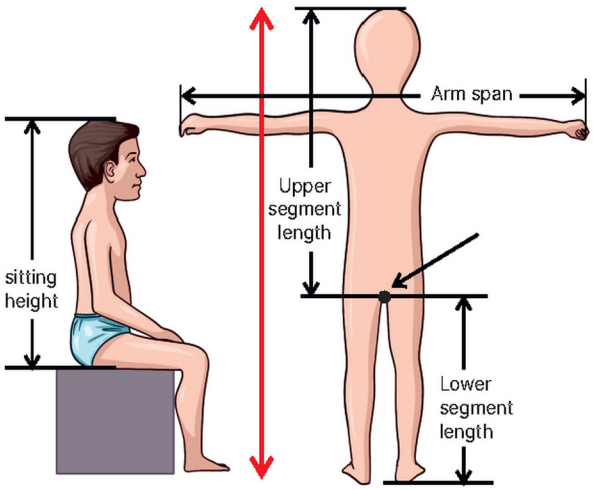
Increase In Length With Age

- 1st 3 months: 3.5 cm/month
- 3-6 months: 2 cm/month
- 6-9 months: 1.5 cm/month
- 9-12 months: 1.2 cm/month
- 1-3 years: 0.8 - 1 cm/month

Body Segments

00:10:20

- Sitting height = Upper segment length
- Height Prediction Formula:
  - The expected height of a child is calculated using
    → 6x + 77cm
  - x is the age in years
- Alternative Parameter: Arm span
  - Also used as a clinical indicator to estimate a child's height.
- A child typically reaches half of adult height at 2 years of age.



MCQ

Q. Best sign of adequate infant growth in an infant with 2.8 kg birthweight?
A. Weight gain of 300 gm per month till 1 year
B. Increase in length of 25cm in the first year
C. Anterior fontanelle closure by 6 months
D. Weight under 75th percentile and length under 25th percentile

Answer: B. Increase in length of 25cm in the first year

SHORT STATURE

00:13:30

- Definition:
  - Height of a child < 3rd percentile or < -2 z-score, according to the age & sex of the child.
  - Growth Chart Standards:
    → < 5yr → WHO Growth charts
    → > 5yr → IAP Growth charts
- Classification:
  - Proportionate: Upper Segment: Lower Segment ratio → Normal.
  - Disproportionate

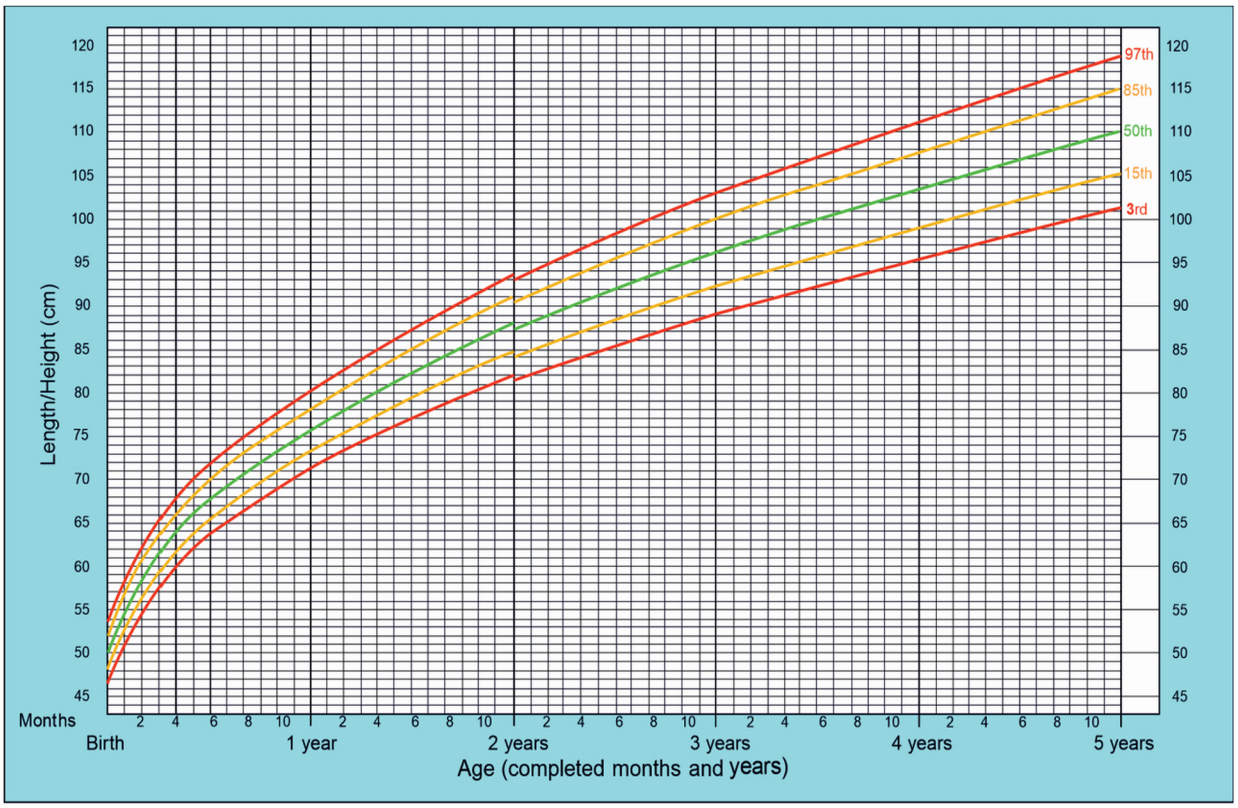
Etiology Of Proportionate Short Stature

Normal Variants	Intrauterine	Post-natal / Acquired

• Familial Short Stature
• CDGP (Constitutional Delay in Growth & Puberty)
	• IUGR / SGA
	• Intrauterine infections
	• Genetic Syndromes:
	  ○ Turner Syndrome
	  ○ Down Syndrome
	  ○ Seckel (Bird-headed dwarfism)
		• Chronic systemic illness (Any)
		• Long-standing/chronic malnutrition
		• Celiac disease
		• Endocrine:
		  ○ Type 1 Diabetes
		  ○ GH deficiency
		  ○ Cushing syndrome
		• Psychosocial: Maternal deprivation

Interpretation Of The Height Of A Child

FMGE 2022, FMGE 2023



Who Growth Charts

INICET 2021
00:21:35

Basis	MGRS (Multicentre Growth Reference Study)
Year of Implementation	1st came into use in 2006
Included Countries	Brazil, Oman, Norway, Ghana, the US, and India (6 countries total)
Feeding Standard	Exclusively breastfed
Exclusion Criteria	Maternal smoking or Alcohol

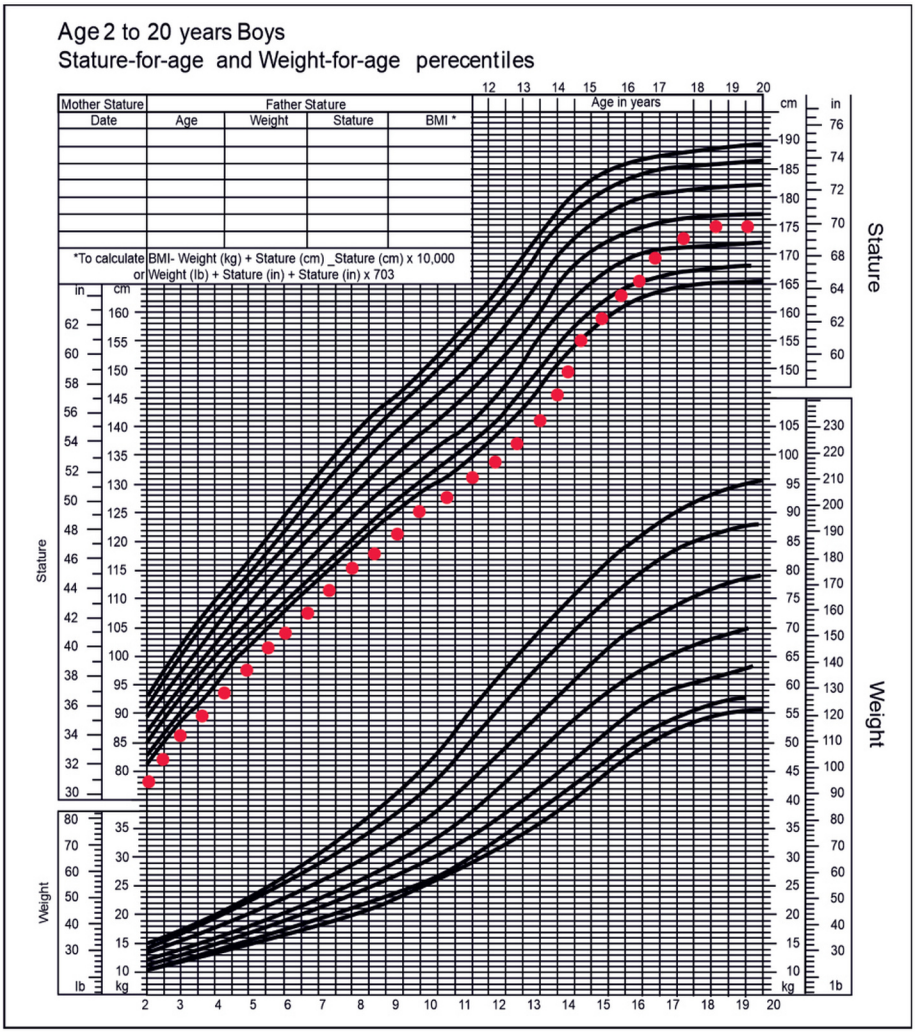
Yourwish

Case Scenario

FMGE 2023, FMGE 2025

- A 7-year-old otherwise well child presents with short stature. His weight is appropriate as per his age. His bone age is less than his chronological age. The height of his parents is normal. What is the diagnosis?
  - Impression: CDGP

Feature	CDGP (Constitutional Delay in Growth & Puberty)	Familial Short Stature
What is it?	During childhood, there is short stature, but the final adult height attained is normal.	The child has short stature, but his height is normal as per the height of his parents.
Puberty	H/o Delayed puberty	Normal
Family history	No H/o Short stature	H/o Short stature
Bone age (BA)	BA < CA (Chronological Age)	BA = CA



Constitutional Delay in Growth & Puberty

Mid Parental Height (mph) Formulas

00:28:28

- MPH is used to calculate the target height of a child based on parental measurements
  - FH= Father's Height
  - MH= Mother's Height

Boys	Girls
$\frac{FH+MH+13}{2}$ cm	$\frac{FH+MH-13}{2}$ cm

Growth Hormone (gh) Deficiency

00:30:20

- At Birth: Normal weight and length at birth
- Type of Short Stature: Proportionate
- US: LS Ratio: Normal
- Bone Age: BA < CA
- Investigations: GH stimulation test (using Insulin, Clonidine)
- Management: Recombinant GH Injection (s/c)
  - Adverse Effects: Pseudotumor cerebri

Q. Which factor does not cause fetal growth in the uterus?

A. Insulin

B. GH

C. IGF-1

D. Thyroxine

Answer: B. GH

Upper Segment: Lower Segment (us: Ls) Ratio

Age	Normal Ratio
Birth	1.7 - 1.9: 1
3 years	1.3: 1
7-10 years	1: 1

Disproportionate Short Stature

00:33:40

Short Trunk (US: LS ratio Decreases)	**Short Limbs** (US: LS ratio Increases)
• Mucopolysaccharidosis • Spondyloepiphyseal dysplasia • Congenital vertebral defects (e.g., Hemivertebra, Butterfly vertebra) ○ Butterfly vertebra associated with Alagille syndrome. • Caries Spine / Pott's disease	• Achondroplasia • Osteogenesis imperfecta • Rickets • Congenital Hypothyroidism

Achondroplasia

00:37:00

Q. A 10-year-old child presents with short stature. The upper segment to lower segment ratio is 1.4:1. He has adequate dietary intake. What is the most likely diagnosis?

A. Achondroplasia

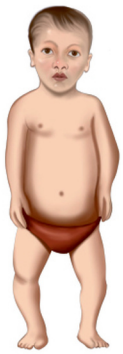
B. Spondyloepiphyseal dysplasia

C. Severe PEM

D. Growth hormone deficiency

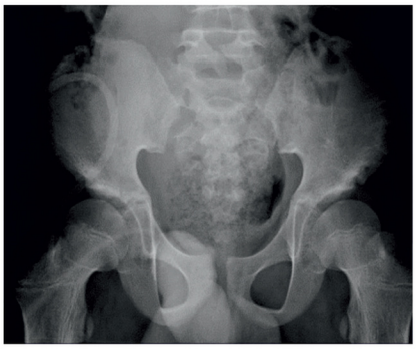
Answer: A. Achondroplasia

- Genetics: Autosomal Dominant
  - FGFR3 gene.
- Clinical Features:
  - Large head.
  - Short limbs
    → US: LS ratio ↑
  - Trident hand.
  - Delayed motor milestones.
- Radiology: "Champagne glass" pelvis



A

B



C

HEAD CIRCUMFERENCE (HC)

00:39:22

INICET 2024

- Occipito-Frontal Circumference
- At Birth: 33 - 35 cm
- Normal Rate of Increase:
  - 1st 3 months: 2 cm/month
  - Next 3 months: 1 cm/month
  - Next 6 months: 0.5 cm/month
  - Next 2 years: 0.2 cm/month
- If HC increases by >2 cm in any month, suspect Hydrocephalus or Intracranial Tumor.

Microcephaly

- Definition: HC of a child <-3 z-score as per the age & sex of the child

Etiology Of Primary Microcephaly

- Edwards Syndrome: Trisomy 18
- Patau Syndrome: Trisomy 13
- Cri-du-chat Syndrome: 5p⁻ (deletion in short arm of chromosome 5)
- Smith-Lemli-Opitz Syndrome: Deficiency of the enzyme 7-dehydrocholesterol
- Familial

Etiology Of Secondary Microcephaly

Maternal Factors	• Maternal Infections: TORCH ○ Toxoplasmosis can also cause macrocephaly due to hydrocephalus • Maternal Drugs: e.g., Phenytoin • Maternal metabolic disorders: e.g.,Phenylketonuria • Maternal Radiation • Maternal Alcohol: Fetal Alcohol Syndrome → ASD • Maternal Smoking
Baby / Infant Factors	• Perinatal Asphyxia / HIE → Cerebral Palsy • Severe Malnutrition • Meningoencephalitis during infancy • Acquired Causes (normal at birth) ○ e.g., Rett Syndrome, Angelman Syndrome

Rett Syndrome INICET 2023

- Inheritance: X-linked Dominant
- M/c seen in Girls
- M/c affected gene is the MECP2 gene
- Head Circumference at birth: Normal
- Clinical Features:
  - Developmental Delay
  - Intellectual Disability
  - Acquired microcephaly
  - Stereotypic hand wringing movements
  - May have Seizures or breathing irregularities
- Treatment:
  - FDA Approved Drug: Trofinetide

Macrocephaly (>+2sd) 00:48:07

- Definition: HC > 2 standard deviations above the mean.

Etiology Of Macrocephaly

- Increased thickness of cranial bones:
  - Rickets
  - Chronic hemolytic anemia (e.g., Thalassemia)
  - Osteogenesis Imperfecta
- Subdural Effusion / Empyema
- Megalencephaly
  - Increased size of the brain parenchyma
- Hydranencephaly
  - Cerebral hemispheres are absent and replaced by fluid-filled sacs
  - Tranillumination +
- Hydrocephalus
  - Increased size of the ventricles inside the brain

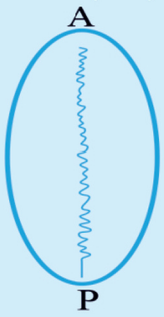
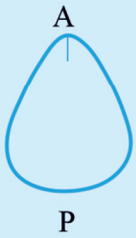
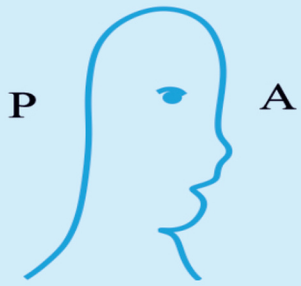
Etiology Of Megalencephaly

- Benign Familial Megalencephaly.
- Amino acid disorders: Glutaric aciduria Type I.
- Lysosomal Storage Disorders: Mucopolysaccharidosis.
- Neurodegenerative diseases → Regression of previously attained milestones
  - Alexander disease
    → GFAP gene mutation
    → Accumulation of Rosenthal fibers
  - Canavan disease
    → Aspartoacylase deficiency → Accumulation of N-acetylaspartic acid (NAA) in the brain.
    → Magnetic Resonance Spectroscopy (MRS) → NAA peak
- Genetic Syndromes: e.g., Sotos syndrome (Cerebral Gigantism)

Craniosynostosis

00:53:00

- Abnormal head shape due to the premature fusion of cranial sutures.

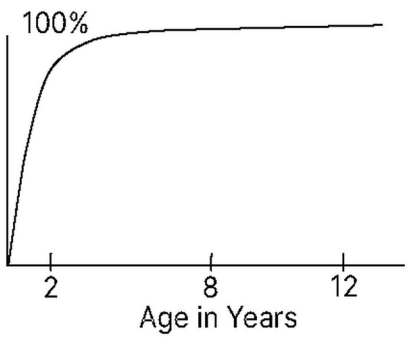
Dolichocephaly 	Premature fusion of the Sagittal suture Results in a long, narrow head.
Trigonocephaly 	Premature fusion of the Metopic suture Results in a triangular forehead.
Turricephaly / Oxycephaly 	Premature fusion of multiple sutures at the base of the skull Results in a tall, tower-like head.

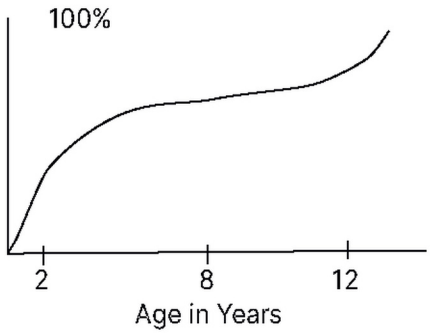
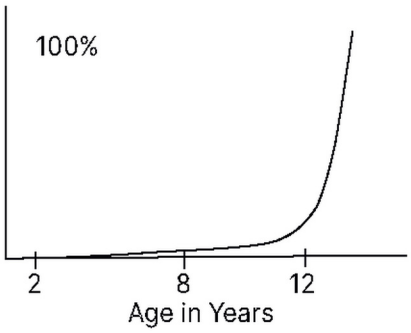
Syndromes Associated With Craniosynostosis

- Apert Syndrome: Characterized by "mitten" hands and feet (syndactyly)
- Crouzon Syndrome
- Carpenter Syndrome
- Pfeiffer Syndrome: Often presents with a "cloverleaf skull."

PATTERNS OF GROWTH

00:55:25

CNS Growth	Rapid early growth 

Somatic Growth (Increase in stature)	Rapid in infancy, slows in childhood, and surges again at puberty 
Lymphoid Growth	Rapid growth at 4-8 years, then undergoes involution 
Gonadal Growth	Latent during childhood with a very sharp increase at puberty 

DENTITION

00:56:30
FMGE 2021

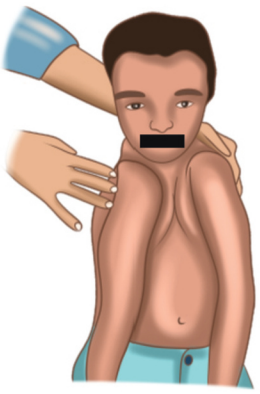
Feature	Primary (Milk / Temporary)	Secondary (Permanent)
Begins at	6-7 months	6 years
1st Tooth	Lower central incisor	1st molar
Last Tooth	2nd molar	3rd molar or wisdom tooth
Completes by	2.5-3 years	12 years (except 3rd molar: 18-25 years)
Dental Formula (Incisors, Canines, Premolars, Molars)	2 1 0 2	2 1 2 3
Total Count	20	28-32

- Mixed Dentition Period: 6-12 years.
- Permanent Teeth Calculation: (Age - 5) x 4
  - 6 years: (6-5) x 4 = 4 teeth
  - 10 years: (10-5) x 4 = 20 permanent teeth (plus 4 temporary).

Delayed Dentition

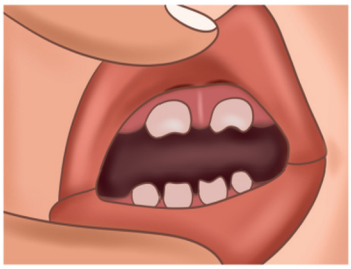
- Definition: When no tooth erupts by 13 months of age.
- Etiology:
  - Familial, Idiopathic, Rickets, Down syndrome, Cleidocranial dysostosis.
  - Endocrine Causes:
    → Hypopituitarism
    → Hypothyroidism
    → Hypoparathyroidism

Cleidocranial Dysostosis



- Autosomal dominant
- Large anterior fontanelle with delayed closure
- Absent clavicles
- Supernumerary teeth.

Hutchinson's Triad



- Late manifestation of Congenital Syphilis
  - Hutchinson's teeth (notched incisors)
  - Interstitial keratitis
  - Sensorineural hearing loss (SNHL)

Natal Teeth

- Seen in:
  - Pierre Robin Sequence
  - Ellis-Van Creveld syndrome
  - Sotos syndrome.

2. DEVELOPMENT AND PUBERTY

DEVELOPMENT

00:00:07

- Cephalocaudal direction
- Certain milestones appear only after specific primitive reflexes are lost
  - Palmar grasp lost → Voluntary grasp
  - Asymmetric tonic neck reflex (ATNR) lost → Rolling over

Major Gross Motor Milestones

- On Ventral Suspension:
  - 1 month → Head is below the plane of the rest of the body
  - 2 months → Head is in the plane of the rest of the body
  - 3 months → Head is above the plane of the rest of the body

Neck holding:	
• Partial	3m
• Complete	5m
Rolls over:	5m (ATNR lost)
Sitting:	
• With support	6m (Tripod position)
• Without support	8m
Pivoting, Cruising:	10-11m
Standing:	
• With support	9m
• Without support	12m
Walking:	
Walk with support	12m
Walking without support	13m
Creeps upstairs:	15m
Rides Tricycle: 	3yr

Yourwish

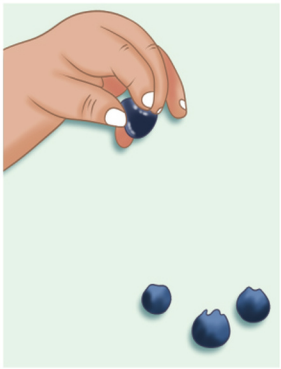
Upstairs & downstairs:	
• 2 feet/step • Runs and Kicks a ball	2yr
• Up with alternate feet & downstairs 2 feet/step	3yr
• Alternate feet upstairs & downstairs	4yr
Hops:	4yr

INICET 2021, INICET 2024, FMGE 2024, FMGE 2025

Major Fine Motor Milestones

INICET 2020, AIIMS 2020, INICET 2023, FMGE 2021, FMGE 2023, INICET 2024, FMGE 2024, INICET 2025, FMGE 2025

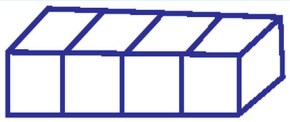
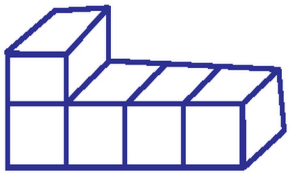
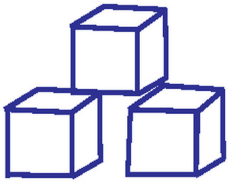
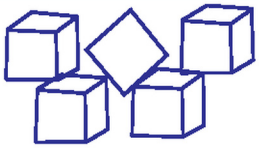
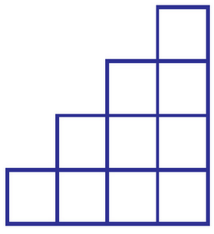
00:04:44

Hand regard 	3m
Reaches out for objects	4m
Bidextrous grasp	5m
Unidextrous / palmar grasp (Appears after the palmar grasp reflex disappears)	6m
Transfers objects (Appears after the palmar grasp reflex disappears)	7m
Pincer grasp (Immature) 	9m
Pincer grasp (Mature)	12m
Casting	12m
Feeding self	18m
Handedness (Appears)	2yr
Handedness (Established)	3yr
Dresses and undresses self	3yr

Ties shoelace 	5yr

Tower Of Cubes

- Formula: Tower of cubes = Age x 3
  - 2years → tower of 6-7 cubes
  - 3years → tower of 9-10 cubes

2 yr	Train without chimney	
2½ yr	Train with chimney	
3 yr	Bridge	
4 yr	Gate	
5-6 yr	Steps	

Copying

2 yr	Horizontal & Vertical lines	–
3 yr	Circle	○
4 yr	Rectangle & Plus sign	□ and +
5 yr	Triangle & Cross	Δ and ×
6 yr	Diamond	◊

Social Milestones

00:10:10

INICET 2021, INICET 2024, FMGE 2024, FMGE 2025

2 m	Social smile
6 m	Mirror play 
7 m	Stranger anxiety
9 m	Bye-bye
10 m	Peek-a-boo 
12 m	Kisses / Plays a simple ball game 
15 m	Points to objects
18 m	Domestic mimicry Dry during the daytime
2 yr	Parallel play
3 yr	Joins in play & Night-time continence
3 yr	Knows name, age, and gender
4 yr	Goes to the washroom alone
5 yr	Helps with household tasks

Nocturnal Enuresis (ne)

00:11:53

- Involuntary urination at night beyond 5 years of age, irrespective of being a boy or a girl.
- Gender Prevalence: Males > Females.
- Positive family history in >50% of cases.

Treatment

- 1st line: Behavioral therapy
  - Lifestyle changes
  - Motivational therapy (e.g., Star Chart)
- 2nd line: Bed and alarm technique
- 3rd line: Drugs
  - Imipramine
  - Desmopressin

FMGE 2023, INICET 2024, FMGE 2025

Language Milestones

00:14:40

2 m	Vocalises
3 m	Cooing
6 m	Monosyllables
9 m	Bisyllables / Bisyllabic babbling
1 yr	2 to 3 words with meaning
15 m	Jargon speech
18 m	Vocabulary of 10 words
2 yr	Vocabulary of 50-100 words Forms 2-word sentences & uses pronouns
3 yr	3-word sentences Can repeat 3 digits
4 yr	Tells a poem/rhyme/story or sings a song

Developmental Quotient (dq)

00:16:10

- DQ = (Developmental Age/Chronological Age) x 100
- Example: If a 6-year-old child can only do the milestones of a 3-year-old
  - DQ= (3/6) x 100 = 50%
- DQ<70% → Developmental delay
  - In 2 or more milestones → Global developmental delay

Q. A 9-month-old infant is brought to the pediatrician due to concerns that the child cannot sit without support. The child is otherwise able to roll over, says "mama" and "dada", and demonstrates stranger anxiety. Which of the following is the most appropriate next step in management?

a. Immediate referral for developmental assessment
b. Order an immediate MRI
c. Reassure the parents, as this is normal for the age
d. Initiate physiotherapy

Answer: c. Reassure the parents, as this is normal for the age

Red Flag Signs Of Developmental Delay

00:19:00
INICET 2025

Milestone	Normal	Red flag
Sitting with support	6 m	9 m
Walking with support	12 m	15 m
Social smile	2 m	3 m
Waves bye-bye	9 m	12 m
Pincer grasp	9 m	12 m
Meaningful words	1 yr	15 - 16 m

Developmental Implications

- Hand regard → Self-discovery of hands.
- Reaches for objects → Visuomotor co-ordination.
- Transfers objects → Comparison of objects.
- Points to objects →Interactive communication.
- Uncovers hidden toy → Object permanence.
- Pretends to drink from a cup → Symbolic thought

PUBERTY

00:21:54

- Adolescent age group: 10 - 19 years of age
- Tanner's staging or Sexual Maturity Rating (SMR):
  - Stage 1 to 5
    → Stage 1: Prepubertal
    → Stage 5: Mature adult
  - Boys → Development of Testes, Penis, Pubic hairs
  - Girls → Breast, pubic hair

Changes In Puberty In Girls

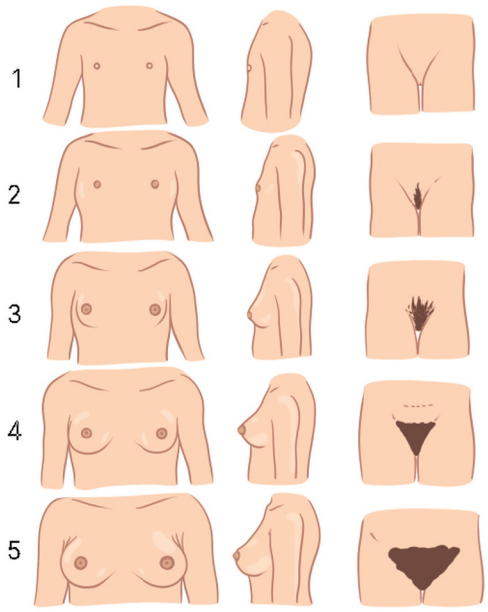
00:23:47

Thelarche (Breast development): 1st sign of puberty in girls
↓
Pubarche (Pubic hairs)
↓
Growth spurt (Peak ↑ in Growth velocity)
↓
Menarche

Tanner Staging

Stage	Breast Development	Pubic Hair Growth
1	Prepubertal	No pubic hair
2	Elevation of the breast bud	Sparse thin growth along the medial border of the labia.

3	Further enlargement of the breast and areola	Considerably coarser and curlier hair spreads sparsely over the mons pubis
4	Projection of the areola and nipple to form a secondary mound above the level of the breast.	Adult-type hair, but the area covered is considerably smaller than in most adults.
5	Areola flattens and merges with the rest of the breast and nipple projects	Adult in quantity and type with spread to the medial surface of the thighs.



Changes In Puberty In Boys

00:27:40

Testicular Enlargement (1st sign of puberty in boys)
↓
Penile enlargement
↓
Pubic hairs
↓
Peak increase in Growth velocity (Growth spurt)
↓
Axillary hairs
↓
Facial hairs

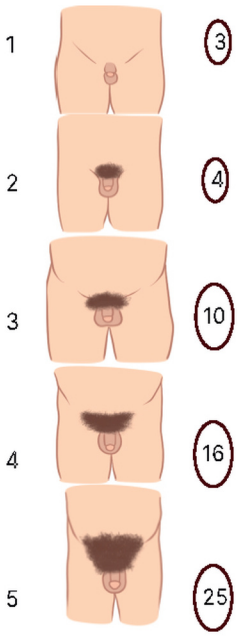
- Deepening or ripening of the voice occurs in the later part of puberty

Orchidometer

- To assess testicular size
- Gently pull the scrotal skin taut to isolate the testis, then hold the orchidometer beads side-by-side to the testicle to find the volume that most closely matches its size.

Tanner Staging

Stage	Physical Development	Testicular size
1	Prepubertal Testicular Volume: < 4 cc	3cc
2	Sparse hair at the base of the penis. Testicular size ≥ 4 cc (1st sign of puberty)	4cc
3	Testicular enlargement, stretching of scrotal skin, and increased penile length.	10 cc
4	Coarser, curlier, abundant pubic hair; penis increases in length and girth.	16 cc
5	Mature adult development.	25 cc



Growth Spurt

00:33:01

- The growth spurt occurs later and lasts longer in boys than in girls
- Females: Occurs during SMR Stage 3
  - Growth rate: 8-9 cm/yr
  - Maximum growth occurs just 6 months before menarche
- Males: Occurs during SMR Stage 4
  - Growth rate: 9-10 cm/yr
- The earliest stage where sperm may be seen in a boy's urine is SMR Stage 3
- Pubertal Gynecomastia:
  - Symmetrical bilateral breast tissue growth is seen in 40-65% of males during SMR Stage 3 and 4.
  - This is due to an excess of estrogenic stimulation.
  - Requires reassurance only in a normal child

Head-ss Assessment

00:36:05

- This psychosocial screening tool for adolescents covers the following areas:
  - Home
  - Education
  - Activities / Alcohol
  - Diet / Drugs
  - Self-esteem / Sexuality
  - Safety / Suicidal tendency/ Sleep

3. NUTRITION,MALNUTRITION,FLUIDS AND ELECTROLYTES

BREAST MILK AND BREASTFEEDING

00:00:20

- Breastfeeding should be initiated within 1 hour of childbirth irrespective of mode of delivery FMGE 2020
- Exclusive breastfeeding
  - Baby should receive only breast milk (not even water, except if medically indicated)
  - Recommended for the first 6 months of life
- Pre-lacteal Feeds
  - Must be strictly avoided
  - No honey, water, or other feeds

Human Colostrum

00:01:35

- Breast milk produced in 1st 72 hours after birth
- Thick and yellowish in color
- Richer in IgA ,proteins,macrophages
- Poor in lactose
- Must be fed to all babies
- Also called 1st immunisation of the baby

Important Information

- Exclusive breastfeeding is recommended for the first 6 months of life
- It can be continued up to 2 years of age or beyond
- Complementary feeding should be introduced after 6 months while continuing breastfeeding

Q. Complementary feeding is initiated for a 7-month-old infant using the "katori method". What is the recommended amount and frequency of food to be given in the first 24 hours?

A. ½-1 katori, 4 times a day

B. ½-1 katori, 5 times a day

C. ½-1 katori, 6 times a day

D. ½-1 katori, 8 times a day

Who And Unicef Recommendation

- Early initiation of breastfeeding within 1st hour
- Exclusive breastfeeding for the first 6 months
- Introduction of complimentary feeding at 6 completed months
- 2-3 meals per day for infants 6-8 months of age
- 3-4 meals per day for infants 9-23months of age, with 1-2 additional snacks are required

Important Information

- Breastmilk can provide >50% of a child energy needs between 6-12months and one third of energy needs between 12 and 24 months

Storage of Expressed Breast Milk

FMGE 2023

- At room temp-8hours
- In refrigerator-24hours
- In deepfreezer-3 months
- MAX breast milk output in mother is at around 5 to 6 months age

Factors Affecting Breastmilk Output

INCREASING OUTPUT	DECREASING OUTPUT
• Sight of baby • Touch of baby • Thought of baby • Night time feeds	• Stressed out mother • Baby on top feeds • Pacifiers/feeding bottles(nipple confusion) • No night time feeds

Signs of Good Positioning While Breastfeeding

INICET 2025

- Entire body of baby should be well supported
- Baby should be turned towards the mother
- Occiput shoulder and buttocks of the baby in a straight line
- Abdomen of baby should touch abdomen of mother

Signs of Good Attachment While Breast Feeding

- Mouth of baby should be wide open
- Entire areola should be in baby mouth except small upper part which maybe visible
- Lower lip of baby should be everted
- Chin of baby should touch mothers breast
- Cheeks should appear full

Q. Which of the following statements regarding breastfeeding technique is/are incorrect?

A. The infant's lower lip should be inverted during latching

B. The infant's cheeks should appear full during effective suckling

C. A greater portion of areola should be covered above the breast than below

D. The infant's chin should make contact with the breast during feeding

a. A and C only

b. B and D only

c. B, C and D only

d. All of the above

Contrainidications To Breastfeeding

00:12:28

CONDITIONS IN BABY	CONDITIONS IN MOTHER
• Confirmed Galactosemia • Confirmed primary lactose intolerance	ABSOLUTE CONTRAINDICATIONS • Mother on chemotherapy/radiotherapy

	RELATIVE CONTRAINDICATIONS
	• Maternal HIV
	• Maternal varicella
	• Maternal herpes lesion on breast
	• Maternal active TB
	  ○ Treatment not started
	  ○ Treatment given < 3 weeks
	• Breast abscess

Important Information

Maternal HIV

- Breast feeding Recommendation:
  - In developed countries / where replacement feeding is AFASS (Acceptable, Feasible, Affordable, Sustainable, Safe) → Avoid breastfeeding and give replacement feeds
  - If AFASS criteria are not met → Continue exclusive breastfeeding (with appropriate maternal ART)

Breastmilk V/s Cows Milk

CONTENT	BREAST MILK	COW MILK
CARBOHYDRATE	• 7gm/dl	• 4.5gm/dl
PROTEINS	• 1 g/dL • Whey-rich (lactalbumin) • Adequate AA: cysteine, taurine, methionine → CNS development	• 3-3.5 g/dL • Casein-rich → difficult to digest
LIPIDS	• Rich in PUFA • DHA rich	
MINERALS	• Ca:P ratio favors Ca absorption • Iron highly bioavailable	• Rich in phosphates • ↑ Risk of hypocalcemia

VITAMINS	• Adequate except Vit D, K, B12
	  ○ All babies: 400 IU Vit D/day (1st year)
	  ○ Single 4 mg Vit K IM at birth (prevent HDN)
		• Poor in Vit C → ↑risk of scurvy

Antiinfective Substances In Breast Milk

- TGF beta
- Phagocytic macrophages
- PABA
- Lactoferrin
- Lysosyme
- Antibodies especially IgA
- Antistaphylococcal factor

- Bifidus factor
- Bile stimulated ligase

Breast Milk Protects Against

NEONATES	OLDER CHILDREN
• Neonatal sepsis • NEC (necrotizing enterocolitis)	• Obesity • Metabolic syndrome • Asthma • Allergies

Preterm Breastmilk

- Richer in-
  - Sodium
  - Immunoglobulins
  - Proteins
  - Calories
  - Iron

Foremilk V/S Hindmilk

- Complete emptying of each breast is important
- Baby gets benefit of both formilk and hindmilk

FOREMILK	HINDMILK
• Thin and watery • Satisfies mainly thirst of baby	• End of feeding session • Thicker • Richer in fat • Calorie dense • Satisfies hunger

Checking Adequacy of Breastfeeding In Baby

Case scenario:

A primigravida mother feels that her milk output is inadequate for her baby and the baby is remaining hungry. So, she is thinking of starting top feed for her 2-month-old baby. How will you know whether breastfeeds are adequate for a baby?

Explanation

- Baby sleeps → 1-2 hours after feeding
- Passes urine → 6-8 times per day
- Baby gaining weight
- Reassure the mother if above present

MICRONUTRIENTS

00:25:22

FMGE 2024, FMGE 2025

Vitamin A Deficiency

- Earliest symptom → night blindness
- Earliest sign → conjunctival xerosis
- Other signs
  - Corneal xerosis
  - Bitot's spots
    → Foamy pearly white appearance on both sides of the cornea
  - Corneal scarring
  - Corneal ulcerations

Therapeutic Dose Of Vitamin A

- <6months→ 50000IU/dose
- 6M-1yr/<8kg → 1Lac IU
- >1 year / > 8 kg → 2 lakh IU
- Dosing schedule (Total 3 doses):
  - 1st dose → At diagnosis (0 hour)
  - 2nd dose → After 24 hours
  - 3rd dose → After 2 weeks

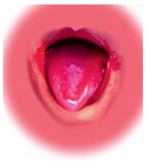
Vitamin A Prophylaxis

- Total: 9 mega doses
  - 1st dose: Given with MR vaccine (at 9 months)

Other Micronutrient Deficiencies

00:28:28

THIAMINE (B1)	• Risk factor: History of polished rice intake
	• Deficiency disease: Beri-beri
	• Types
	  ○ Wet Beri Beri
	  ○ Dry Beri Beri
	• Wet Beri Beri
	  ○ M/C children
	  ○ Features of CHF (Edema, Tachycardia, Tachypnoea, Poor feeding) can be present
	• Dry beri beri
	  ○ Mainly neurological feature (Irritability, Neurodefects, Peripheral nerve involvement)

RIBOFLAVIN(B2)	• Deficiency leads to
	  ○ Oral ulcers (Aphthous stomatitis)
	  ○ Angular chelitis
	  ○ Glossitis
	  ○ Seborrheic dermatitis
	  ○ Conjunctivitis
	 
	 

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NIACIN	• Deficiency leads to Pellagra
	• Features
	  ○ Diarrhoea
	  ○ Dermatitis (casal's necklace)
	  ○ Dementia
	  ○ Death
	 

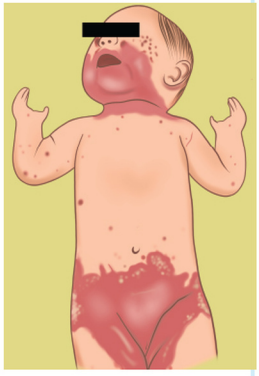
PYRIDOXINE (B6)	• Deficiency cause Neurological features
	  ○ Peripheral neuropathy
	  ○ Very Important cause of refractory seizures in neonate and infant
	• Drugs like INH may predispose to pyridoxine deficiency

BIOTIN	• Deficiency leads to
	  ○ Skin rash
	  ○ Alopecia
	  ○ Neurological manifestations (Developmental delay, Seizures sometimes)
	• H/O raw egg consumption
	  ○ Raw egg contains Avidin, binds with biotin and decrease bioavailability

VITAMIN B 12	• Deficiency leads to
	  ○ Subacute combined degeneration of spinal cord
	  ○ Megaloblastic anemia

VITAMIN K	• Deficiency leads to
	  ○ Hemorrhagic disease of newborn
	  ○ Vit K is required for clotting factors 2,7,9,10

VITAMIN C	• Deficiency leads to ○ Scurvy (Collagen synthesis defect)
VITAMIN D	• Deficiency leads to rickets

ZINC	• Deficiency leads to
	  ○ Diarrhea
	  ○ Dermatitis
	  ○ Hypogonadism
	  ○ Poor wound healing
	  ○ Recurrent infections
	FMGE 2025
	
	ACRODERMATITIS ENTEROPATHICA
	• Pathophysiology
	Mutation in gene SLC39A4
	↓
	Defective intestinal zinc absorption
	↓
	Zinc deficiency
	↓
	Dermatitis (hands, feet, periorificial), Recurrent diarrhea
	• Treatment: High Dose zinc
	 

MALNUTRITION

00:37:00

NEET PG 2021, FMGE 2025

- Best indicator for
  - Acute malnutrition →decrease in weight for height (wasting)
  - Chronic malnutrition →decrease in height for age (stunting)

Who Classification Of Malnutrition

WEIGHT FOR HEIGHT	HEIGHT FOR AGE
Between -2 to -3 z score Or Between 70%-79% of expected	Between -2 to -3 z score Or Between 85%-89% of expected
SEVERE WASTING <-3z score Or <70% of expected	SEVERE STUNTING <-3 Z score Or <85% of expected

Q. A boy whose anthropometry found weight for age −1.8 SD and height for age −2.5 SD and weight for height is of −1.7 SD. What kind of malnutrition is this?

A. Severe acute malnutrition

B. Chronic malnutrition

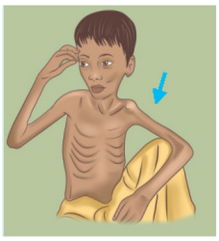
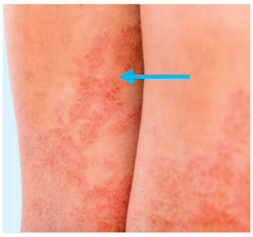
C. Acute on chronic malnutrition

D. Moderate acute malnutrition

FMGE 2020, Neet PG 2020, FMGE 2021, FMGE 2022, FMGE 2023

Kwashiorkar V/S Marasmus

	KWASHIORKAR	MARASMUS
EDEMA	Present	Absent
DEFECIENCY	Protein Deficiency	Calorie Deficiency
APETITE	• Poor appetite • Decompensated state • Poor prognosis	• Voracious / good appetite • Good prognosis
CNS INTERVAL	Present (Apathy)	Active and alert No CNS involved
FATTY HEPATOMEGALY	Present	Absent
SKIN AND HAIR CHANGES	Present	Absent

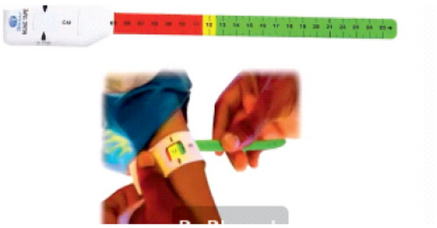
Marasmus	Flaky Paint Dermatosis	Flag Sign
		

Severe Acute Malnutrition (SAM)

INICET 2020, FMGE 2023, INICET 2024, FMGE

- **Definition**: in a child 6M-5yr of age presence of 1 or more of
  - Weight for height <-3 Z score /< 70% of expectant (severe wasting)
    Or
  - Mid upper arm circumference <11.5cm/<115mm
    Or
  - Symmetric bipedal edema of nutritional origin

Shakirs Tape



- Used to measure MUAC by field workers
  - Red Zone → MUAC <11.5CM(severe malnutrition)
  - Yellow Zone →11.5cm to 12.5cm (borderline nutritional status)
  - Green Zone → 12.5cm(normal nutritional status)

Complication Of Sam

- Mnemonic: SHIELDED
  - S - Sugar deficiency (Hypoglycemia)
    → Blood glucose < 54 mg/dL
  - H - Hypothermia
    → Rectal temperature < 35.5°C
  - I - Infections
  - E, L - Electrolyte imbalance
  - D, E - Dehydration
  - D - Deficiency of micronutrients

Management of Sam

- Initial hospitalization of all patients
- Especially those with Poor appetite or Medical complications

HYPOGYLCEMIA	• 10% dextrose orally/NG tube
HYPOTHERMIA	• Remove wet clothing • Warm up the child
INFECTIONS	• Oral antibiotics /IV antibiotics
ELECTROLYTE IMBALANCE	• Supplement potassium

DEHYDRATION	• ReSoMal is used (Rehydration Solution for Malnourished children)
	• Different from ORS
	  ○ ↓ Sodium
	  ○ ↑ Potassium
	• Clinical features:
	  ○ Increased thirst
	  ○ Decreased urine output

MICRONUTRIENTS DEFECIENCY	• Supplement minerals and vitamins in adequate doses • Except iron → interferes with healing ○ Started later in rehabilitation phase

Nutritional Rehabilitation

- Start with low calories and proteins and build up gradually over 1-2 weeks to prevent nutritional recovery syndrome /refeeding syndrome
- Diets: F-75 (Initial diet) → F-100 → RUTF
  - **F-75 DIET**
    → Initial diet
    → 100ml of F-75 contains: 75 kcal, 0.9g protein
  - **F-100 DIET**
    → 100ml contains 100kcal, 3g proteins
  - **RUTF**
    → 100g of RUTF (Ready to use therapeutic food) contain 543kcal and 15g protein

- Refeeding Syndrome
  - Sudden ↑ blood glucose → Sudden ↑ insulin release
    → ↑ Regeneration of tissues (PO_4 requirement) → Abrupt decrease in serum phosphate → **Hypophosphatemia**
    → ↑ Thiamine use → features of beriberi
    → ↑ Uptake of Potassium and Magnesium by cells → Hypokalemia, Hypomagnesemia
  - All these will lead to features of HF
    → Worsening edema
    → Tachypnea
    → Tachycardia
    → Poor feeding
  - Thiamine supplementation is given (prevents complications during refeeding)

Discharging Criteria For Sam

- Child should be accepting well orally
- Should have lost edema and started gaining weight
- All infections and micronutrient deficiencies should have been treated
- Mother should be confident of taking care of child at home

Treatment Failure In Sam

PRIMARY FAILURE TO RESPOND IS INDICATED BY:

- Failure to regain appetite by day 4

- Failure to start losing edema by day 4
- Presence of edema on day 10
- Failure to gain at least 5 g/kg/day by day 10

Secondary Failure To Respond is Indicated By:

- Failure to gain at least 5 g/kg/day for 3 consecutive days during the rehabilitation phase

FLUIDS AND ELECTROLYTES

00:57:27

FMGE 2020, INICEt 2024, FMGE 2024

- Calculation of 24 hour maintenance fluid required in children

'1st 10 kg	100ml/kg
Next 10 kg	50ml/kg
Beyond 20 kg	20ml/kg

- Fluid of Choice: N/2 in 5% dextrose or DNS with added potassium

Intravenous Fluids in Neonates

- Daily requirements during 1st week of life

BIRTH WT	DAY 1	DAY 2	DAY 3	DAY 4	DAY 5	DAY 6	DAY 7
<1500g	80ml/kg/day	95	110	120	130	140	150ml/kg/dl
>1500g	60ml/kg/day	75	90	105	120	135	150ml/kg/dl

- Initial fluids:10% dextrose
- Na and K should be added to IV fluid after 48 hours

Management of Septic Shock In Children

01:02:07

- **0 min**
  - Start high flow oxygen
  - Secure wide bore IV access
  - If no IV access with in 1 min → interosseous access
- **5min**
  - Start IV fluid boluses
    → Crystalloids /isotonic fluids
    → NS/RL
    → 20ml/kg up to 3 times
- Correct hypoglycemia/hypocalcemia
- Start broad spectrum antibiotics
- I/C/O child not improves → Fluid Refractory Shock → Start inotropes
- Common isotonic crystalloids
  - NS
    → Sodium 154meq/L
    → Chloride 154meq/L

→ No potassium
→ Osmolarity → 308

- **RL**
  → Sodium of 131meq
  → Potassium 5meq/L
  → Osmolarity 280mmosm/L

Treatment of Fluid Refractory Shock

- Begin inotropes (dopamine)
- Insert central lines and secure airway
- If not improving consider
  - Cold shock+normal BP →epinephrine
  - Cold shock with low BP →epinephrine +dobutamine
  - Warm shock→ norepinephrine (wide pulse pressure)
- Not improved → Catecholamine Refractory Shock
  - Start steroids (IV hydrocortisone)
  - Monitor CVP
  - Maintain Hb >10g/dl
- Shock still not reversed → ECMO(Extra corporeal membrane oxygenation)

Q. Fluid of choice for shock in a child with severe acute malnutrition
A. Ringer lactate
B. 10% Dextrose
C. Normal Saline
D. Ringer lactate + 5% dextrose

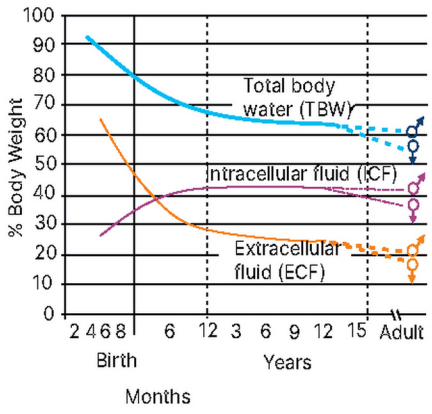
Q. Children easily get dehydrated. What is the difference in ECF and ICF ratio when compared to adults?
A. ICF is higher than ECF
B. Total body water is high
C. ECF is higher than ICF
D. Total body water is low

TOTAL BODY WATER

01:06:54

- TBW= ECF+ICF
- ECF Constitutes
  - Plasma
  - Interstitial fluid
- **TBW Constitutes**
  - 90% body weight in fetal life
  - 75% of weight at birth
  - 60% by the end of 1st year, remains same till puberty
- In fetus and newborn: ECF>ICF
- Children and adults: ICF>ECF

ECF > ICF



ICF >ECF

FLUID RESUSCITATION IN CHILD WITH BURNS

01:09:20

- Identify approximate surface area of the burns
- Preferred fluid: Ringer lactate in 5% dextrose
- Parkland formula:
  - 4 mL/kg/% BSA involved in first 24 hours
    → ½ in first 8 hours
    → ½ in next 16 hours
  - Next 24 hours:
    → Reabsorption of edema fluid & diuresis occurs
    → Give ½ of fluid infused on 1st day

MANAGEMENT OF FOREIGN BODY ASPIRATION IN AN INFANT

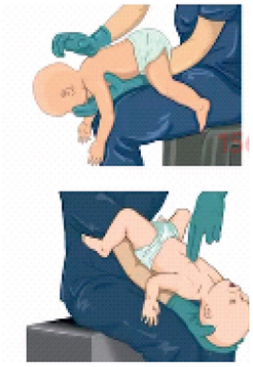
01:10:50

Q. A 3-year child accidentally aspirates a peanut and suddenly starts coughing. He has been brought to the Pediatric Emergency in an unconscious state. What is the appropriate immediate management?

A. Abdominal thrusts (Heimlich maneuver)
B. Back slaps
C. Chest compressions
D. Endotracheal intubation

Responsive Infant

- Use 5 back slaps followed by 5 chest thrusts till the foreign body is expelled or the infant becomes unresponsive
- Do not use abdominal thrusts (Heimlich maneuver) in infants



If The Infant Becomes Unresponsive

- Stop back slaps immediately
- Start CPR
- Begin CPR with chest compressions

Choking Relief Maneuvers For Children (≥ 1year Age)

Responsive Child

- Abdominal thrusts / heilminch maneuvre

Unresponsive Child

- Start CPR
- As per PALS guidelines
  - Airway
  - Breathing
  - Circulation
  - Disability
  - Exposure

Q. A 3-year-old child is on oxygen by nasal cannula at 3 L/min. What will be the FiO_2 delivered?

A. 30%

B. 35%

C. 40%

D. 50%

Explanation:

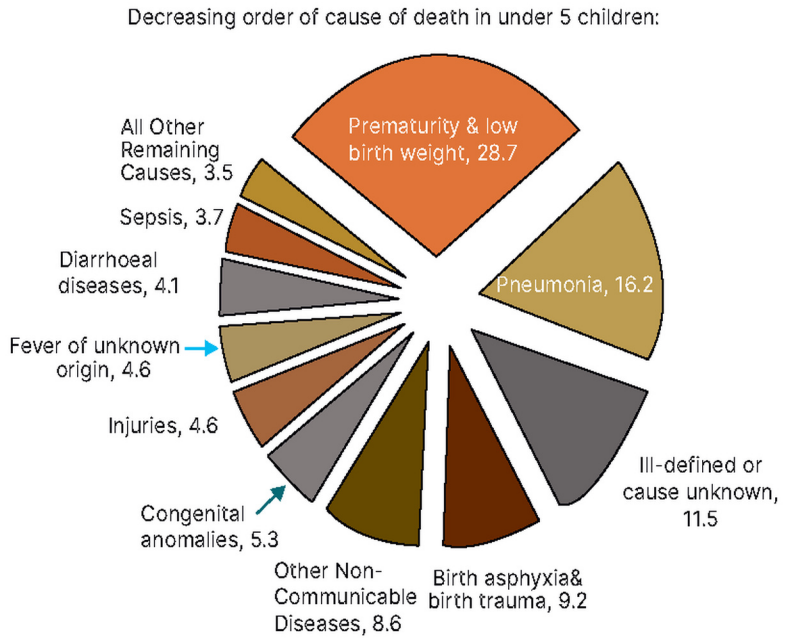
- FiO2= 21 + 3 x flow rate
- 21 + 3 x 3 = 30%

Important Information

- Ratio of Chest compressions : Positive pressure Ventilation in a child
  - Resuscitation of a child with 1 rescuer→ 30:2
  - Resuscitation of a child with 2 rescuers → 15:2

Decreasing Order Of Cause Of Death In Under 5 Children

- M/c cause: prematurity & LBW > Pneumonia > Birth Asphyxia and Birth trauma



Yourwish

4. INFECTIOUS DISEASES IN CHILDREN & IMMUNIZATION

INFECTIOUS DISEASES IN CHILDREN

00:00:08

Measeles

NEET PG 2021, Neet PG 2023, FMGE 2023

Case Scenario

Q. An 18-month-old unimmunized girl had fever with rash (as shown in the given picture), cough, and coryza. There is a history of similar complaints in 2 other children in the neighborhood. The doctor also noticed a few red spots with white central parts in the buccal cavity of this child. Which vitamin has a role in the management of this child?

- Clinical Presentation:
  - 18-month-old and unimmunized (high risk).
  - Fever, cough, coryza, and conjunctivitis
  - Erythematous maculopapular rash
  - "Red spots with white central parts in the buccal cavity" → Koplik spots
- Diagnosis: Measles.

Answer: Vitamin A is crucial in the management of Measles

COMPLICATIONS

- M/c complication: Otitis media
- M/c cause of death: Pneumonia
- Long-standing complication: SSPE (Subacute Sclerosing Panencephalitis)
  - Occurring 7-10 years after primary Measles infection
  - Associated Symptoms of SSPE:
    → Poor school performance
    → Myoclonic jerks
    → Regression of milestones
  - Diagnosis:
    → CSF and Anti-measles antibody
    → Warthin-Finkeldey giant cells on skin biopsy

CASE SCENARIO

Q. A 9-year-old child presents with myoclonic jerks and decreased performance in school. There is a history of fever at the age of 1 year with rash. EEG shows burst suppression. Which investigation should be done?

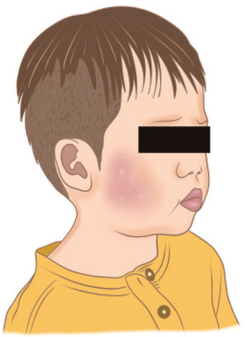
A. Measles IgG
B. MRI Brain for Temporal Sclerosis
C. Electromyography
D. Anti-GQ1b Ab in CSF

- Diagnosis: SSPE

Answer: A. Measles IgG in blood & CSF

Erythema Infectiosum

00:03:00



- Fever with 'slapped cheek' rash
- Caused by Parvovirus B19

Diseases Caused By Parvovirus B19

- Transient Aplastic Crises
- Papular Purpuric Glove & Socks Syndrome
- Non-immune Hydrops Fetalis

Hand Foot Mouth Disease

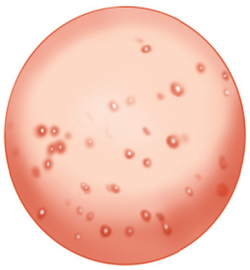
00:03:44

- Causative Agents: Coxsackie A16 or Enterovirus 71
- Clinical Course:
  - Mild and self-limiting
  - Infectious
- Symptoms:
  - Low-grade fever
  - Oral ulcers
  - Vesicles & blisters on palms and soles
- Treatment: Supportive care and isolation

Varicella

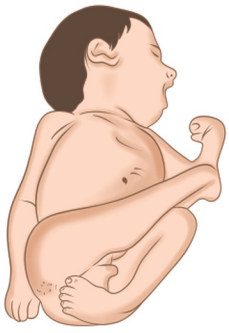
00:04:50

- AKA: Chickenpox
- Incubation Period: 10-21 days
- Characteristic Features:
  - Fever + Pleomorphic rash (macule, papule, vesicle, pustule, and crusted lesions appearing simultaneously)
  - Rash appears in crops
- Infectivity: Patient remains infective until all lesions have crusted
- Complication: Ataxia



Congenital Varicella Syndrome

- History: Fever with rash during pregnancy
- Clinical findings in the baby:
  - Microcephaly
  - Cicatricial Scars
  - Limb defects



Rubella

00:06:54

NEET PG 2021, Neet PG 2023, INICET 2023, FMGE 2025

- AKA: German measles
- Characteristics: Like Measles but milder
- Associated with posterior auricular lymphadenopathy

Congenital Rubella Syndrome

- Risk: Maximum if acquired during the 1st trimester
  - Should be vaccinated before pregnancy

- Classic Triad:
  - Cataract.
  - Deafness.
  - Congenital Heart Diseases
    → M/c: Patent Ductus Arteriosus
    → Least common: Atrial septal defect
- Other Features:
  - Dermal erythropoiesis → Blueberry Muffin Lesions
  - Hepatosplenomegaly
  - Jaundice
  - Microcephaly
  - Chorioretinitis

Congenital Cmv Infection

00:09:10

- M/c cause of non-syndromic SNHL in children
- Asymptomatic cases: >80%.

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- Symptomatic Findings:
  - Microcephaly
  - Periventricular Calcifications
  - SNHL
  - CMV retinitis
  - CMV colitis
  - Hepatosplenomegaly
  - Thrombocytopenia
  - Jaundice
- Risk of transmission during pregnancy:
  - 1° CMV infection → 30%
  - 2° CMV infection →1% (M/c in India)
- Best Sample for CMV PCR of the baby: Urine CMV PCR
- DOC: Ganciclovir

MCQ

Q. Which of the following is least likely to be transmitted perinatally?

A. Hep A

B. HSV

C. Rubella

D. CMV

Answer: A. Hep A

- Hepatitis A is primarily transmitted via the fecal-oral route
  - Presents as Acute Viral Hepatitis with highly elevated liver enzymes
    → SGOT/SGPT in the thousands
  - Diagnosed by positive Anti-HAV IgM
- HSV → Vesicular lesions on the genitalia of mother
  - Baby presents with encephalitis and seizures

Congenital Toxoplasmosis

00:13:44

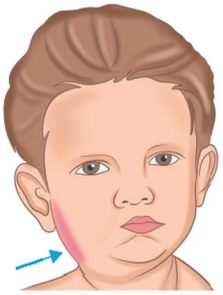
NEET PG 2024

- Findings:
  - Chorioretinitis
  - Hydrocephalus (leading to macrocephaly in baby)
  - Cerebral Parenchymal Calcification
- Treatment:
  - For Exposure / Suspected Infection (Prophylaxis): Spiramycin
  - For Confirmed Fetal Infection:
    → Sulphadiazine and Pyrimethamine with Leucovorin

Mumps

00:15:00

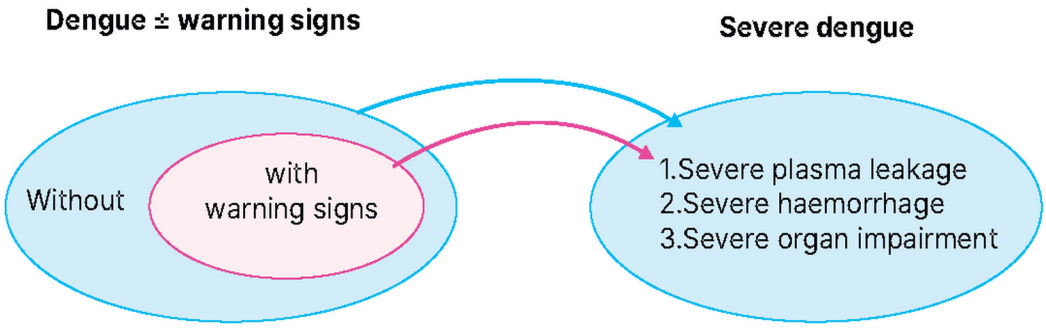
Q. A male child presents to you with swelling below the ear, but no systemic manifestations. There have been two other similar cases in the friends of this child in the same village. What will you do?



- Diagnosis: Mumps
- Management:
  - The condition is self-limiting.
  - Advise Isolation.
  - Provide symptomatic relief with PCM (Paracetamol) and plenty of fluids
- Potential Complication: Aseptic meningitis

Dengue

00:16:00



Criteria for dengue ± warning signs

Probable dengue
Live in/travel to dengue endemic area. Fever and 2 of the following criteria:
- Nausea, vomiting
- Rash
- Aches and pains
- Tourniquet test positive
- Leucopenia
- Any warning sign

Laboratory confirmed dengue

Warning signs
- Abdominal pain or tenderness
- Persistent vomiting
- Clinical fluid accumulation
- Mucosal bleed
- Lethargy: restlessness
- Liver enlargement >2cm
- Laboratory: Increase in HCT concurrent with rapid decrease in platelet count

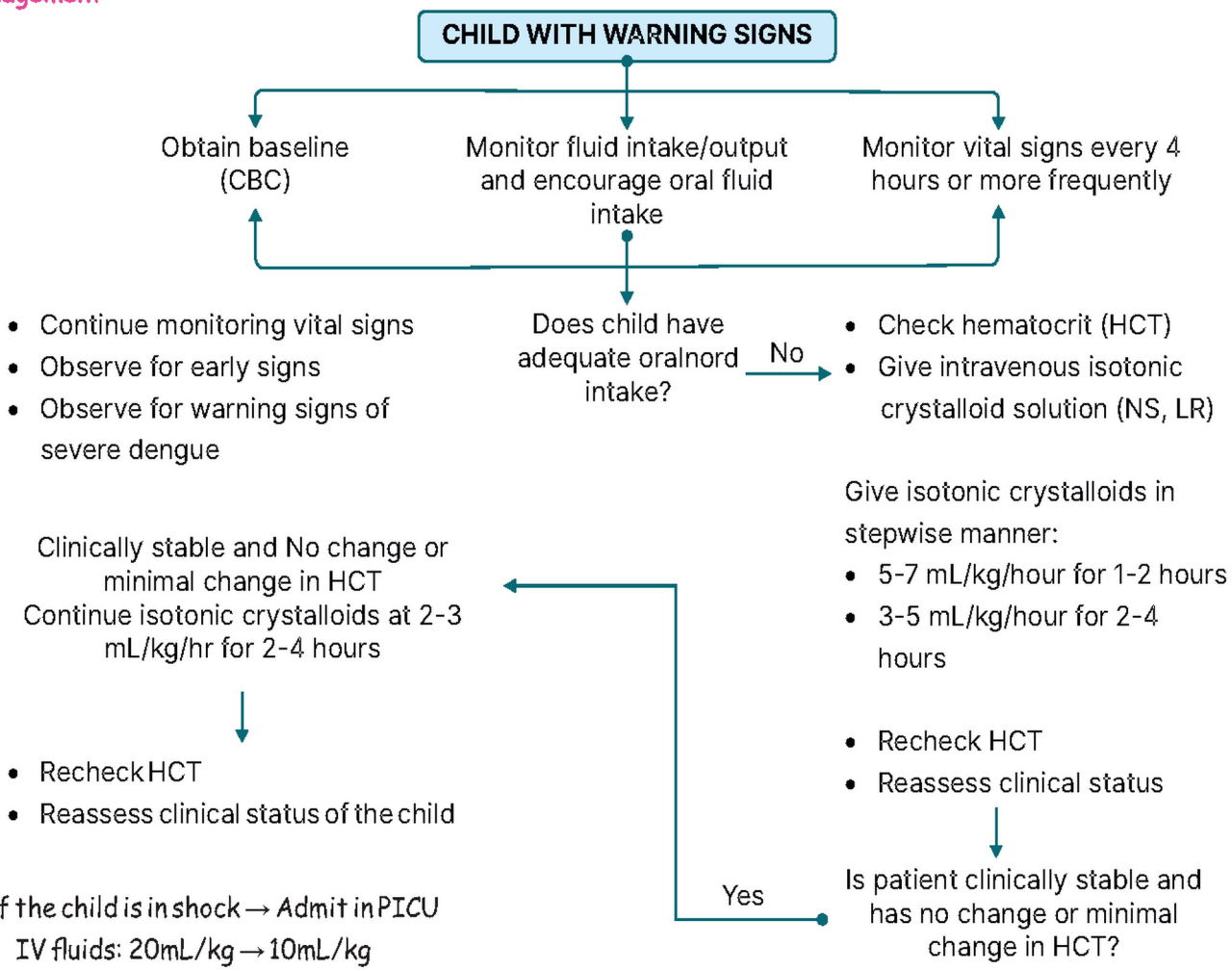
Criteria for severe dengue

1. Severe plasma leakage leading to:
- Shock (DSS)
- Fluid accumulation with respiratory distress

2. Severe bleeding as evaluated by clinician

3. Severe organ involvement
- Liver: AST or ALT>=1000
- CNS: Impaired consciousness
- Heart and other organs

Management



- If the child is in shock → Admit in PICU
  - IV fluids: 20mL/kg → 10mL/kg

Case Scenario

Q: A 10-year-old child, who was previously Anemic, presented with fever and abdominal pain and tested positive for Dengue on day 5 of fever. His BP was 108/70 mm Hg, Hb is 13 g/dl, hematocrit is 40, and platelet counts were 30,000/mm ^{3}, but there were no bleeding manifestations. He has poor oral acceptance. What is the next step?

A. Platelet transfusion
B. 10 ml/kg/hr RL
C. 7 ml/kg/hr RL
D. 20 ml/kg/hr RL
E. Paracetamol

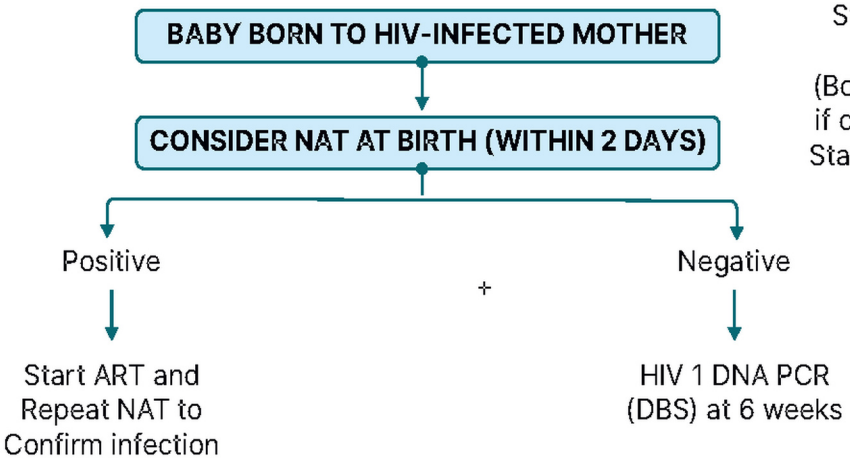
Answer: C and E

- Diagnosis:
  - 1st 3 days → NS1Ag +
  - ≥ 5 days → Dengue IgM +
- SBP Formula = > 70 +2x (x → age)
  - SBP should be > 90 in this case
- Platelet Transfusion:
  - Not indicated here because the count is >10,000/mm ^{3} and there is no mucosal bleeding
- Because the child has "poor oral acceptance" and warning signs (abdominal pain), IV fluids are needed.
  - The rate of 7 ml/kg/hr is appropriate for initial stabilization in non-shock dengue with warning signs

HIV

00:21:21

Management



Start nevirapine 2mg/kg/dose OD at birth
(Both Nevirapine and Zidovudine if classified as a high-risk infant)
Start cotrimoxazole at 4-6 weeks of age

- If negative → Continue prophylaxis with Nevirapine
  - Until 3 months after breastfeeding has completely stopped

High-risk Infant

- Mother not on ART or received <4 weeks ART at delivery
- Mother has unknown or high viral load (RNA > 1000 copies/ml) from 32 weeks of pregnancy to delivery
- Mother newly identified during current pregnancy or within 6 weeks of delivery

MANAGEMENT

- Zidovudine + Nevirapine for the first 12 weeks if exclusively breastfeeding
- Zidovudine + Nevirapine for the first 6 weeks of life if exclusive replacement feeding
- Mothers should start or continue receiving ART

CASE SCENARIO

Q. An HIV-positive mother with a viral load of 1200 copies/mL delivers a baby. What is the most appropriate antiretroviral prophylaxis for the newborn?

A. Nevirapine for 6 weeks

B. Nevirapine + Zidovudine for 12 weeks

C. Nevirapine for 12 weeks

D. Zidovudine for 4 weeks

Answer: **B.** Nevirapine + Zidovudine for 12 weeks (High-risk)

Treatment of HIV in Children

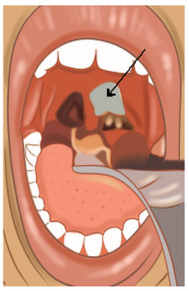
- ART should be given to all children with HIV, irrespective of CD4 count or WHO stage
- High Priority Groups:
  - Children <2 years age
  - WHO stage 3 or 4
  - Children <5 years with CD4 count < 25% or < 750mm^3
- Cotrimoxazole prophylaxis is indicated against Pneumocystis jiroveci for all children

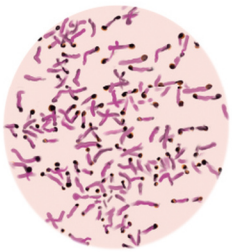
Age	Preferred 1st line treatment
Infant < 2 weeks	Zidovudine + Lamivudine + Raltegravir
Beyond 2 weeks	Abacavir (or Zidovudine) + Lamivudine + Lopinavir / Ritonavir

Diphtheria

00:25:55

Case Scenario





Q. An unimmunized 8-year-old girl presented with fever, sore throat, and swelling of the neck. The examination of the throat revealed a lesion (shown in image 1). The picture seen on Albert stain of the swab taken from the lesion is shown in image 2. What is the probable diagnosis?

- Diagnosis: Diphtheria.
  - Grayish white adherent pseudomembrane.
    → Bleed on removal
  - Albert stain → Characteristic metachromatic granules of Corynebacterium diphtheriae

Management

- Isolation
- Anti-Diphtheritic Serum (ADS)
- Antibiotics and supportive care
- May require intubation and mechanical ventilation (MV)

Complications

- Early:
  - Myocarditis → often presents with disproportionate tachycardia
  - Multiple cranial nerve palsies
    → Weak gag
    → Extraocular muscle weakness
    → Palatal palsy
- Long Term: Post-Diphtheritic Quadriparesis

Care of Close Contacts of Diphtheria Case

- Isolation & Monitoring:
  - Monitor close contacts for a 7-day incubation period
- Chemoprophylaxis:
  - Administer antimicrobial prophylaxis irrespective of immunization status
  - Using a single dose of Inj. Benzathine Penicillin, Erythromycin, Azithromycin
- Vaccination:
  - Adults: If not received diphtheria-containing vaccine in the last 5 years → Tdap or Td
  - Children: If ≤ 3 doses of diphtheria-containing vaccine received → Complete immunization against Diphtheria

Pediatric Tuberculosis

00:32:00

- Ghon Focus:
  - Primary TB: Lungs
  - Congenital TB: Liver
- Treatment: 2HRZE + 4HRE
- Isoniazid Prophylaxis:
  - For all children < 5 years old exposed to an infectious case of TB
  - Dosage: 10 mg/kg/day for 6 months (+ Pyridoxine)
  - After ruling out active TB

Tetanus

00:33:38

Case Scenario

Q. A 4-year-old child presents with difficulty in opening the mouth following a recent ear infection. The child also had an episode of painful tonic spasm of limbs. What is the most likely diagnosis?

A. Meningitis

B. Sigmoid sinus thrombosis

C. Tetanus

D Bezold abscess

Answer: C. Tetanus

Presentation And Treatment

- In unimmunised older Children
  - Secondary to a dirty penetrating injury or an ear infection
- Clinical Features:
  - Difficulty in opening the mouth: Lockjaw or Trismus.
  - Risus Sardonicus
  - Painful spasm of limbs
- Management: Tetanus Ig + Antibiotics + Diazepam

Neonatal Tetanus

- Occurs in babies born to unimmunised mothers
- 1st 2-3 days: Normal
- Day 3 - Day 10:
  - Poor feeding
  - Lethargy
  - Irritability
  - Opisthotonic posturing
- Could be fatal

IMMUNIZATION

00:37:00

National Immunization Schedule of India

Age	Vaccines Given	Route / Notes
Birth	**BCG, OPV-0, Hepatitis B** birth dose	• BCG: Intradermal (Left upper arm)

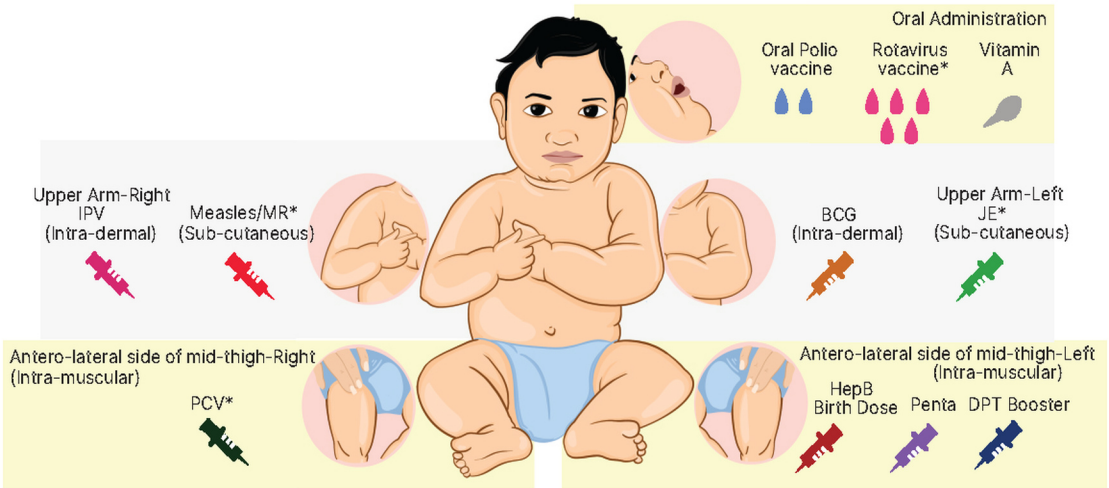
		• If the mother is a known case of Hep B → Hepatitis B immunoglobulin is also given
6 Weeks	**OPV-1, RVV-1, Pentavalent-1, fIPV-1, PCV-1**	• OPV-1, RVV-1: Oral • Pentavalent & PCV: Intramuscular; • fIPV: Intradermal
10 Weeks	**OPV-2, RVV-2, Pentavalent-2**	
14 Weeks	**OPV-3, RVV-3, Pentavalent-3, fIPV-2, PCV-2**	

9-12 Months	**MR-1 , PCV-Booster, fIPV-3, JE-1** (in endemic districts only), **Vitamin A** (1 Lakh IU)	• MR-1: Subcutaneous
		• JE:
		  ○ Live: SC
		  ○ Killed: IM

16-24 Months	**MR-2, DPT-Booster-1, OPV-Booster, JE-2**	
5-6 Years	**DPT-Booster-2**	
10 & 16 Years	**Td**	

- Pentavalent vaccine (Intramuscular) provides protection against five specific diseases:
  - D: Diphtheria
  - P: Pertussis
  - T: Tetanus
  - Hepatitis B
  - Haemophilus influenzae

Site and Route of Vaccines



Catch-up Vaccination in An Unvaccinated Child At 1st Visit

- BCG: For children < 1 year old
- OPV: For children < 5 years old

- DPT: For children < 7 years old
- T dap for the 1st dose in children > 7 years old
- Td for the 2nd and 3rd doses in children > 7 years old
- MMR: 2-3 doses at 6-monthly intervals (any age)
- Hib:
  - 6-12 months: 2 doses + 1 Booster
  - 12-15 months: 1 dose + 1 Booster
  - > 15 months: 1 dose

Adverse effects

- Anaphylaxis can occur with any vaccine

Neet PG 2024

Vaccine	Important Adverse Effects
BCG	BCG lymphadenitis, Osteitis, Disseminated BCG infection
DPT (Pertussis)	Persistent inconsolable cry, Hyperpyrexia, Hypotonic Hyporesponsive Episodes, Seizures, Encephalopathy • Avoid DPT vaccine in progressive encephalopathy
MMR	Thrombocytopenia, Measles-like rash
OPV	VAPP (Vaccine-Associated Paralytic Poliomyelitis)
Rotavirus	Intussusception • Not given beyond 8m →IAP • Can be given up to 12m →NIS

Case Scenario

00:47:50

Q. A child with steroid-dependent nephrotic syndrome has been on daily oral steroids for 3 weeks. All of the following are true about the vaccination of this child EXCEPT?

A. Give all immunization according to schedule

B. Sibling should not be given with OPV

C. Give killed vaccines

D. Pneumococcal vaccine should ideally be given before commencing the treatment

Answer: **A.** Give all immunization according to schedule

- Live vaccines should be avoided (immunocompromised)
- The sibling of such a child should not be given OPV
  - OPV is a live attenuated vaccine
  - OPV virus is excreted in stool
  - Risk of vaccine-derived infection to immunocompromised contacts
- Give Killed vaccine:
  - Inactivated (killed) vaccines are safe
  - They do not contain live organisms
  - Can be given even during immunosuppression
    → Response may be reduced, but vaccine is safe
- Pneumococcal vaccine should be administered before starting steroid treatment.

  - Streptococcus pneumoniae is a leading cause of Spontaneous Bacterial Peritonitis (SBP) in children with Nephrotic Syndrome
- Inhaled or topical steroids are not a contraindication to vaccination.

Vaccine Guidelines

00:49:47

- Vaccines that can only be given beyond 2 years of age: Any Polysaccharide vaccine
- Vaccine that should be given beyond 9 years of age: HPV vaccine
  - 9-14 years: 2 doses
  - >14 years: 3 doses
- Vaccines that can be given to adolescents:
  - Td, Tdap
  - MMR
  - HPV
  - Rabies Vaccine
  - Influenza Vaccine
- Vaccines contraindicated in egg allergy: Influenza vaccine, Yellow fever vaccine
- Vaccines causing Thrombocytopenia: Measles vaccine
- Vaccines that act faster than the incubation period of the disease they prevent:
  - Varicella vaccine
  - Measles vaccine
  - Rabies vaccine

MCQs

00:52:08

Q. Hepatitis B subunit vaccine is derived from which antigen?
A. HBeAg
B. HBcAg
C. HBsAg
D. HBxAg

Answer: **C.** HBsAg
- Anti HBs Ab > 10 mIU/ml → Protective against Hep B

Q. Which is not true regarding the Rota virus vaccine?
A. It is given orally
B. It is not given after 6 months of age
C. Live vaccine
D. Both monovalent and pentavalent variants are available for use

Answer: **B.** It is not given after 6 months of age
- Can be given till 8 months of age
- Currently used in NIS:
  - Monovalent (Rotavac → 116E)
  - Pentavalent (Rotateq)

Q. Protection given by the Yellow fever vaccine is valid for?
A. Lifelong

B. 10 years
C. 5 years
D. 20 years

Answer: **A.** Lifelong

- Yellow Fever (YF) Vaccine:
  - It is given to all unvaccinated travelers aged > 9 months traveling to at-risk or endemic areas.
  - In May 2014, the World Health Assembly adopted an amendment stipulating that the period of protection and validity of the certificate changed from 10 years to the duration of life of the person vaccinated.
  - Since 11 July 2016, the certificate of vaccination against YF is valid for the life of the person vaccinated, beginning 10 days after the date of vaccination.

Q. Shanchol vaccine is:
A. Monovalent 2 doses
B. Monovalent 1 dose
C. Bivalent 2 doses
D. Bivalent 1 dose

Answer: **A.** Monovalent 2 doses

- Oral Cholera Vaccines (OCV):
  - Currently, there are three WHO pre-qualified oral cholera vaccines: Dukoral, Shanchol, and Euvichol.
  - All three vaccines require two doses for full protection.

Q. Which of the following is the recommended dose for HPV vaccination in girls aged 9-14 years according to the WHO-SAGE Protocol?
A. 1 dose
B. 2 doses
C. 3 doses
D. 1 or 2 doses

Answer: **D.** 1 or 2 doses

Q. All of the following vaccines are recommended to be given before splenectomy except?
A. Typhoid vaccine
B. H. influenzae vaccine
C. Meningococcal vaccine
D. Pneumococcal vaccine

Answer: **A.** Typhoid vaccine (not mandatory)

- Vaccines against encapsulated organisms should be given at least 2 weeks before the procedure.

Open Vial Policy

00:54:58

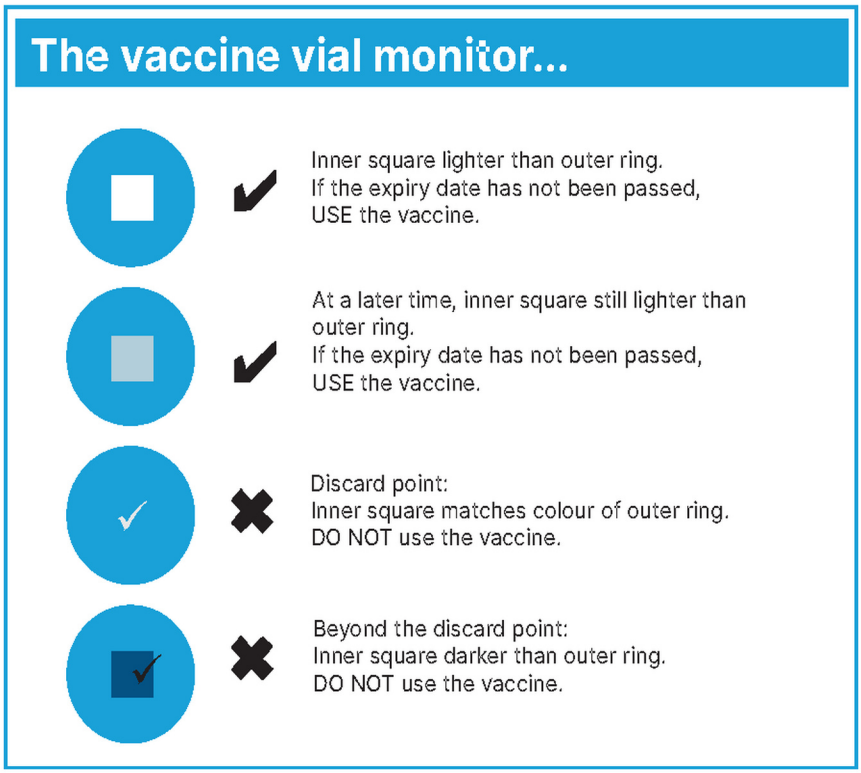
- Allows reuse of partially used multidose vials of certain vaccines, provided:
  - Expiry date has not passed
  - VVM (Vaccine Vial Monitor) → No damage

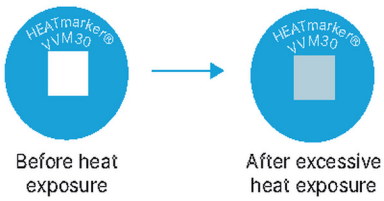
- Asepsis maintained
- Date of opening of the vial mentioned

- The vaccine that follow open vial policy can be reused for up to 4 weeks
  - Prevent vaccine wastage
- Vaccines that do not follow open vial policy: (use within 4 hours)
  - BCG
  - Measles-containing vaccines

Vaccine Vial Monitor (VVM)

- To ensure that no thermal damage has occurred to a vaccine
- Shake Test → To identify if certain vaccines have been damaged by freezing.
  - If sediments develop → Do not use
- VVM30 means:
  - Can remain stable in room temperature (37 °C) up to 30 days





5 GENETICS

TYPES OF GENETIC DISORDERS

00:00:28

INICET 2023

- Chromosomal Disorders: abnormality in
  - Number of chromosomes
  - Structure of chromosomes
- Single gene disorders / Mendelian
  - Autosomal Dominant
  - Autosomal Recessive
  - X-Linked
- Non-Mendelian disorders:
  - For example:- Mitochondrial, Genetic Imprinting, or Trinucleotide Repeat
- Multifactorial:
  - For example:- Neural Tube Defects, Hypertension, and Diabetes

Aneuploidy

- Definition: Chromosome number is not an exact multiple of the haploid number
- Examples:
  - Down Syndrome: 47 chromosomes
  - Turner Syndrome: 45 chromosomes

Q. Which of the following genetic conditions is not classified as an aneuploidy?

A. Trisomy 21

B. Trisomy 13

C. Bloom syndrome

D. Klinefelter's syndrome

Answer: C. Bloom syndrome

Down Syndrome

00:04:05

FMGE 2020, 2021, 2023, 2025

INICET 2023

- Basic Defect: 3 copies of Chromosome 21
- Underlying Mechanisms:
  - Trisomy 21: 95% of cases
  - Translocation of chromosome 21 with other chromosomes: 3% of cases
  - Mosaicism: 1-2% of cases
    - → A condition where an individual has more than one cell line
    - → Usually, less severe clinical features
- Risk: Increases with increased maternal age
- M/c cause: Maternal meiotic non-disjunction

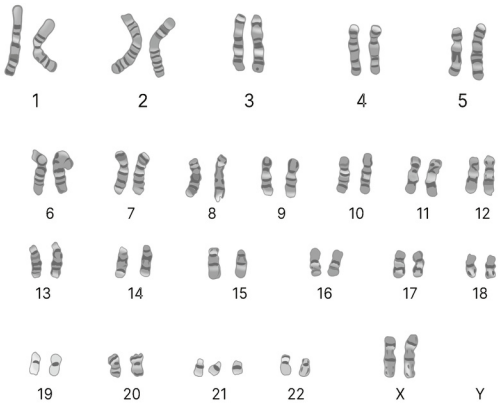
Clinical Features

- Flat occiput
- Protruded tongue
- Epicanthic folds
- Simian crease
  - Single transverse palmar crease
- Sandal gap
- Clinodactyly
- Congenital hypothyroidism

Commonly Associated Problems

- Stature: Short stature
- CNS: Intellectual Disability, Global Developmental Delay
- Eye: Brushfield spots on the iris, refractive errors
- Ear: Deafness
- Neck: Short neck, Hypothyroidism, atlanto-axial instability
- CVS: Increased risk of Congenital Heart Disease (CHD)
  - Most commonly: Endocardial Cushion Defect or AVSD
  - VSD, PDA, ASD, and TOF can also be present
- Hematology:
  - Transient Myeloproliferative Disorder
  - Thrombocytopenia
  - Increased risk of Acute Leukemia
    → < 3 years → AML
    → > 3 years → ALL

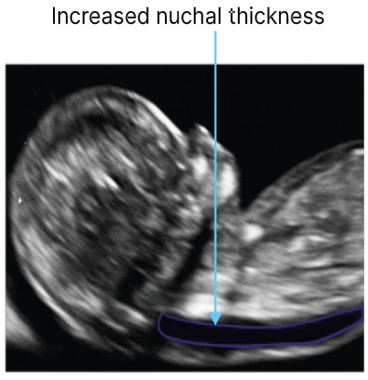
Karyotype



- 47XX (+ 21) → Girl

Antenatal Screening

Radiological Markers



Down syndrome

- Increased nuchal thickness (>3 mm)
- Timing: Typically assessed at 11–13 weeks of gestation

Biochemical Markers

- T1: Dual Markers
  - Beta HCG + PAPP-A (Pregnancy-Associated Plasma Protein A)
- T2:
  - Triple test: Beta HCG, Unconjugated Estriol, AFP
  - Quadruple test: Triple test + Inhibin
- In Down syndrome, Beta HCG and Inhibin are elevated

Integrated Test

- Combines:
  - Maternal age
  - T1 markers (NT + PAPP-A)
  - T2 Quadruple test
- To determine composite risk

Nips

- Non-Invasive Prenatal Screening
- Detects cell-free fetal DNA in maternal blood after 10 weeks of gestation

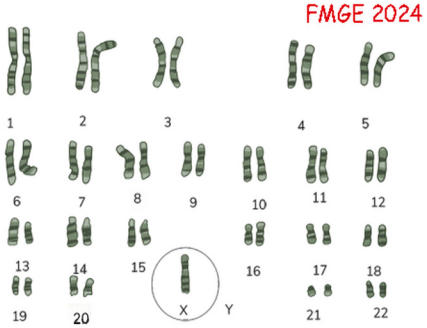
Confirmatory Tests

- Fetal Karyotype:
  - Chorionic Villus Sampling (11-13 weeks)
  - Amniocentesis (14-16 weeks)
  - Cordocentesis (17-20 weeks)
- If any of the parent is a carrier of a t(21;21) translocation, there is a 100% risk of recurrence

Turner Syndrome

00:14:10

FMGE 2024



- Karyotype: 45, XO
- Always seen in females
- Absent Barr body
  - The number of Barr bodies is calculated as X - 1
  - A normal female (XX) has 1
  - Turner (XO) has 0

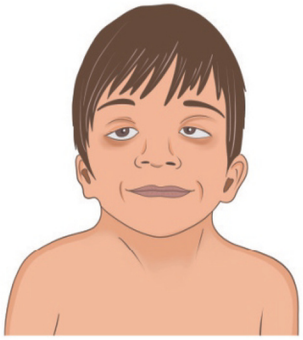
Clinical Features

- Short stature and webbed neck
- Shield-shaped chest with widely spaced nipples
- Cubitus valgus
- Streak ovaries and rudimentary uterus, leading to primary amenorrhea and infertility
- Intelligence: Usually normal

Noonan Syndrome

00:17:30

- Noonan syndrome shares several similarities with Turner syndrome:
  - Short stature
  - Cubitus valgus
  - Webbed neck



Yourwish

Feature	Turner Syndrome	Noonan Syndrome
Gender	Always seen in females	Males or females
Genetics	45, XO; Absent Barr body (X-1=0)	PTPN11 mutation (Autosomal Dominant inheritance); Normal karyotype
Intelligence	Usually normal	Often associated with Intellectual Disability
Dysmorphism	No facial dysmorphism	Antimongoloid slant of eyes; depressed nasal bridge
Fertility	Infertility due to streak ovaries and rudimentary uterus	Normal fertility, though puberty may be delayed
M/c CHD	Bicuspid Aortic Valve > Coarctation of the Aorta	Pulmonary Stenosis

Single Gene Disorders

00:21:24

INICET 2023
NEET PG 2023

Autosomal Dominant Inheritance

- **D**: Ehlers-Danlos syndrome (Collagen defect → hyperextensibility)
- **O**: Osteogenesis imperfecta.
- **M**: Marfan syndrome.
- **I**: Intermittent Porphyria.
- **N**: Noonan syndrome.
- **A**: Achondroplasia.
- **N**: Neurofibromatosis.
- **T**: Tuberous sclerosis

Marfan Syndrome

- Physical Signs: Tall stature, Arachnodactyly (long fingers), Pectus excavatum, and a positive thumb sign
- Eye Findings: Lens dislocation
  - Superolateral direction
- Fibrillin-1 gene (FBN1)

Q. An abnormally tall boy presents with subluxation of the lens, long limbs, and arachnodactyly. Mutation in which of the following genes is most commonly associated with this condition?

A. Fibrillin-1 gene (FBN1)
B. Collagen Type I gene
C. Elastin gene
D. Dystrophin gene

Ans. **A.** Fibrillin-1 gene (FBN1)

Autosomal Recessive Inheritance

- A: Alkaptonuria
- B: Beta Thalassemia and Sickle Cell Disease
- C: Cystic fibrosis and Congenital Adrenal Hyperplasia

- G: Gaucher disease
- H: Homocystinuria
- I: Inborn errors of metabolism
  - Except Hunter syndrome and Fabry's disease

Q. A child with coarse facial features, hypercalcemia, and cardiac findings/murmur suggestive of supravalvular Aortic stenosis, is most likely suffering from?

A. William syndrome

B. Prader-Willi syndrome

C. Angelman syndrome

D. Down syndrome

Ans. **A.** William syndrome

X-linked Dominant

FMGE 2020
NEET PG 2021

- Rett Syndrome
  - Acquired microcephaly
- X-linked Hypophosphatemic Rickets
  - Associated with the PHEX gene

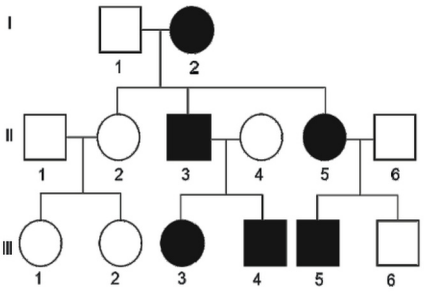
PEDIGREE ANALYSIS

00:26:14

- If all children of an affected female have the disease → Mitochondrial inheritance
- If at least one parent of an affected child has the disease → Dominant.
- Father-to-son transmission:
  - Rules out X-linked inheritance
  - If father-to-son transmission is present → Autosomal

Autosomal Dominant

- Not all children of the affected mother have the disease
  - Rules out Mitochondrial
- One parent of an affected child has the disease → Dominant.
- Father-to-son transmission present → Autosomal



Mitochondrial Inheritance

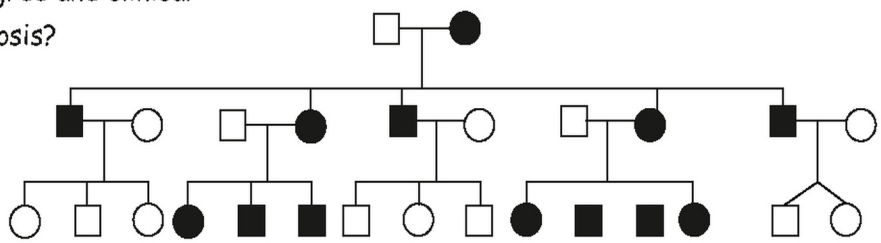
Q. The pedigree diagram of a family is shown below. Affected individuals present with progressive external ophthalmoplegia, pigmentary retinopathy, and cardiac conduction defects. Based on the pedigree and clinical features, what is the most likely diagnosis?

A. Duchenne Muscular Dystrophy

B. Kearns-Sayre Syndrome

C. Prader-Willi syndrome

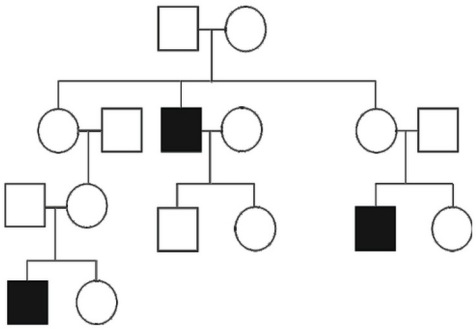
D. William syndrome



Ans. **B.** Kearns-Sayre Syndrome

- Kearns-Sayre Syndrome:
  - A mitochondrial disorder presenting with progressive external ophthalmoplegia, pigmentary retinopathy, and cardiac conduction defects.
- Other Mitochondrial Disorders:
  - Leber's Optic Neuropathy
  - Leigh disease
  - MERRF
  - MELAS (characterized by recurrent strokes)

X-linked Recessive Inheritance



- Most common in males
- Mothers are carriers
- Family history of maternal uncles affected
- None of the parents is affected → Recessive
- No father-to-son transmission
- Examples:
  - DMD (Duchenne Muscular Dystrophy)
  - BMD (Becker Muscular Dystrophy).
  - Chronic Granulomatous Disease
  - Bruton's X-linked Agammaglobulinemia
  - Hunter syndrome
  - Fabry disease
  - Hemophilia A & B
  - G6PD deficiency
  - Wiskott-Aldrich syndrome

Q. A normal couple has a son with Hemophilia, but their daughter is normal. The father's family has no history of Hemophilia, while the mother's family has a positive history of Hemophilia. Which of the following is true?

A. All males affected

B. All females affected

C. Half of males are affected, and 50% females are carriers

D. All of males are affected, and half of the females are carriers

	XY	X^cX
	X^c	X
X	X X^c	X X
Y	X^c Y	X Y

Ans. C. Half of the males are affected, and 50% females are carriers

TRINUCLEOTIDE REPEAT DISORDER

FMGE 2020

00:35:28

Fragile X Syndrome

- Genetics: Mutation in the FMR1 gene on the X chromosome
- Mechanism: Increase in CGG repeats to > 200
  - Normal: < 40 repeats
  - Premutation: 40-200 repeats
- Anticipation: Worsening of clinical features with each successive generation
- Clinical Features:
  - Large head

- Elongated face
- Large jaw
- Macro-orchidism (large testes) in post-pubertal males
- Low IQ

- Carrier Females may have low IQ and premature ovarian failure

GENOMIC IMPRINTING DISORDERS

00:37:40

- Defect in chromosome 15q (long arm)

Prader-Willi Syndrome	Angelman Syndrome
Mechanisms: • Defective paternal imprinting • Paternal gene deletion • Maternal disomy	Mechanisms: • Defective maternal imprinting • Maternal gene deletion • Paternal disomy
Clinical Features: • Infantile feeding problems • Obesity • Hypotonia • Hypogonadism • Dysmorphism: Almond eyes & tapering fingers • Recurrent pneumonias	Clinical Features: • "Happy Puppet Syndrome" • Intellectual Disability • Microcephaly • Episodes of inappropriate laughter • Seizures • Dysmorphism

Q. Which genetic syndrome is caused by a deletion of the short arm of chromosome 5 (5p deletion)?
A. Edward syndrome
B. Patau syndrome
C. Cri-du-chat syndrome
D. Turner syndrome

Answer: C. Cri-du-chat syndrome

- **Edward syndrome** - Trisomy 18
  - Microcephaly, Rocker bottom foot, Congenital Heart Disease
- **Patau syndrome**- Trisomy 13
  - Scalp defects, Holoprosencephaly, CHD, Renal malformations, polydactyly
  - Incompatible with life
- **Cri-du-chat syndrome**
  - Cat-like cry
  - Microcephaly
  - IQ
  - Seizures

6 INBORN ERRORS OF METABOLISM

GLYCOGEN STORAGE DISORDER

00:00:19

Liver Glycogenoses

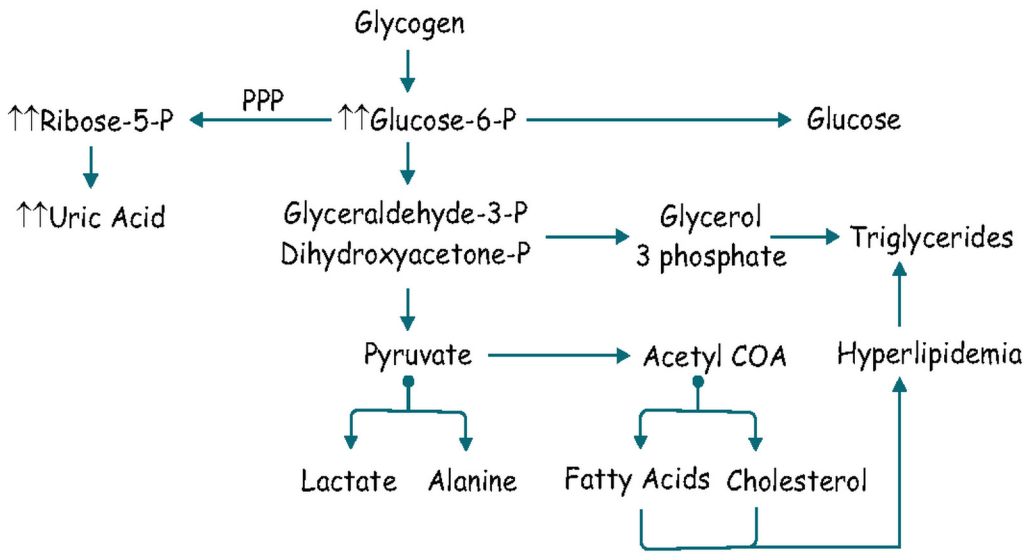
Type	Disease	Enzyme Deficient	Clinical features
I	Von Gierke disease	Glucose 6 phosphatase	• Hypoglycemia • Hyperuricemia • Hypertriglyceridemia ○ Doll-like facies • Lactic acidosis • Hepatomegaly

III	Cori disease	Debranching enzyme	• Along with the liver, the muscle is also involved
			• CPK enzymes elevated
			• When glucagon given
			  ○ Fed state - Blood glucose rises
			  ○ Fasting state - Blood glucose does not rise

IV	Anderson disease	Branching enzyme	
VI	Her's disease	Hepatic phosphorylase	

Von Gierke Disease

00:03:20



- Deficiency of glucose-6-phosphatase
- Glucose 6-phosphate is not converted to glucose
- Hypoglycemia produced, leads to early morning seizure and lethargy
- Accumulation of glycogen in the liver causes hepatomegaly

- Increased Glucose-6-phosphate is directed to other pathways
  - Pentose phosphate pathways
    → Increased uric acid produced leads to hyperuricemia, which manifests as Gout and renal stones
    → Treatment - Allopurinol
  - Glycolytic pathway
    → Increased glyceraldehyde 3-phosphate and Dihydroxy acetone production
      – Glycerol 3 phosphate is formed, producing hypertriglycerides and hyperlipidemia, characteristics of Doll-like facies
    → Increased pyruvate production
      – Leads to increased lactate causes Lactic acidosis
      – Leads to increased acetyl-CoA and fatty acids production

Muscle Glycogenoses

00:06:30

Type	Disease	Enzyme Deficient	Clinical features
II	Pompe disease	Alpha 1,4 glucosidase / Acid maltase	• Affects both skeletal and cardiac muscle • Enzyme replacement therapy is available
V	McArdle disease	Muscle phosphorylase	• Common in adolescents • Easy fatiguability • Calf pain • Muscle breakdown causes myoglobinuria
VII	Tarui disease	Phosphofructokinase	

Q.Match the following

Column A	Column B
A. Cori disease	1. Branched structure in liver biopsy
B. McArdle disease	2. Accumulation of glycogen in lysosomes
C. Pompe disease	3. Exercise intolerance
D. Von Gierke disease	4. Hypoglycemia with lactic acidosis

A. A-1, B-2, C-3, D-4
B.A-1, B-3, C-2, D-4
C. A-3, B-2, C-4, D-1
D. A-4, B-3, C-2, D-1

Ans: B

GALACTOSEMIA

00:09:25

NEET PG 2024
FMGE 2024

Enzyme deficient	• GALT (Galactose 1-phosphate uridyl transferase) • Galactokinase • Epimerase

Clinical features	• Milk and milk products contain lactose, which breaks down into glucose and galactose
	• Asymptomatic at birth
	• Develops clinical features within days to weeks of starting breastfeeding or any milk
	• Galactose metabolism does not occur
	• Galactose converts to
	  ○ Galacitol → accumulates cause
	  → B/L oil drop cataract
	  ○ Galactose 1-phosphate → accumulates causes
	  → Hepatomegaly
	  → Jaundice
	  → Vomiting
	  → Seizures
	  → Developmental delay

Investigations	• Benedict's Test
	  ○ Galactose is a reducing substance that is present in urine
	  ○ On the addition of Benedict's reagent
	  ○ The color of the urine changes to green, orange, and red
	• GALT assay
	  ○ Demonstrate the enzyme deficiency
	  ○ Sample collected from a green vial that contains HEPARIN
	• Genetic diagnosis
	  ○ Gene mutation demonstrated
	  ○ Autosomal recessive inheritance

Management	• Life-long lactose-free diet • Lactose-free soy-based formulae • Absolute C/I to breastfeeding

HEREDITARY FRUCTOSE INTOLERANCE

00:14:16

Enzyme deficient	• Aldolase B / Fructose 1,6 bisphosphate aldolase
Clinical features	• Sweet food contains sucrose, which is converted to glucose and fructose • Inability to metabolize fructose, so fructose and fructose 1 phosphate accumulates • Clinical features develop beyond 6 months of age when started on complementary feeding • The child develops an aversion to sweets

Investigations	• Benedict's Test
	  ○ Positive for reducing substance
	• GALT assay
	  ○ Demonstrate the enzyme deficient
	• Genetic diagnosis

Management	• Life long fructose free diet

PHENYLKETONURIA

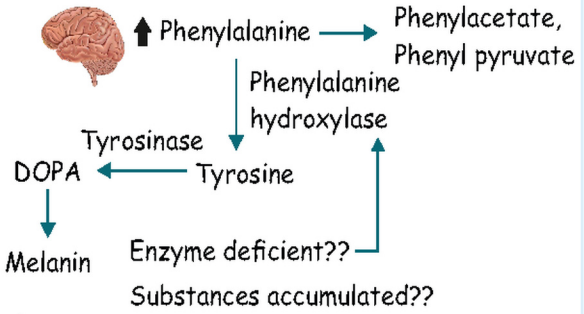
00:16:17

NEET PG 2021

Enzyme deficient	• Phenylalanine hydroxylase

Clinical features	• Phenylalanine accumulates
	  ○ Cause brain damage leads to
	  → Intellectual disability
	  → Seizures
	  → Hypertonia
	• Phenylalanine is converted to phenylacetate and phenyl pyruvate
	  ○ Causes a mousy or musty odor in the urine
	• Tyrosine is not synthesized
	  ○ As a result Melanin is reduced
	  → Hypopigmentation, fair skin, blue iris
	  ○ So tyrosine becomes an essential amino acid in the diet

Management	• Low phenylalanine diet • Supplement tyrosine



ALKAPTONURIA

00:18:51

INICET 2023

Enzyme deficient	• Homogentisic acid oxidase

Clinical features	• Polymers of homogentisic acid accumulate
	  ○ Urine turns black on standing
	  ○ Oochronosis- Black spots on the sclera and eye cartilage
	• Arthritis
	• Cardiac vasculitis

Management	• Low phenylalanine diet • A diet rich in Vitamin C • Nitisinone which blocks excess of homogentisic acid

HOMOCYSTINURIA

00:20:50

Enzyme deficient	• Classical
	  ○ Cystathionine β synthase (pyridoxine is a cofactor)
	• Others
	  ○ Defect in methylcobalamin formation
	  ○ Deficiency of MTHFR (Methylene tetrahydrofolate reductase)

Clinical features	**Similar to Marfan syndrome**
	• Tall stature
	• Skeletal abnormalities
	  ○ Pectus carinatum
	  ○ Pectus excavatum
	  ○ Arachinodactyly
	• Subluxation of the eye lens- Inferomedial
	• Hypercoagulable state- Produce recurrent stroke

Management	• Pyridoxine (Vitamin B6)

Important Information

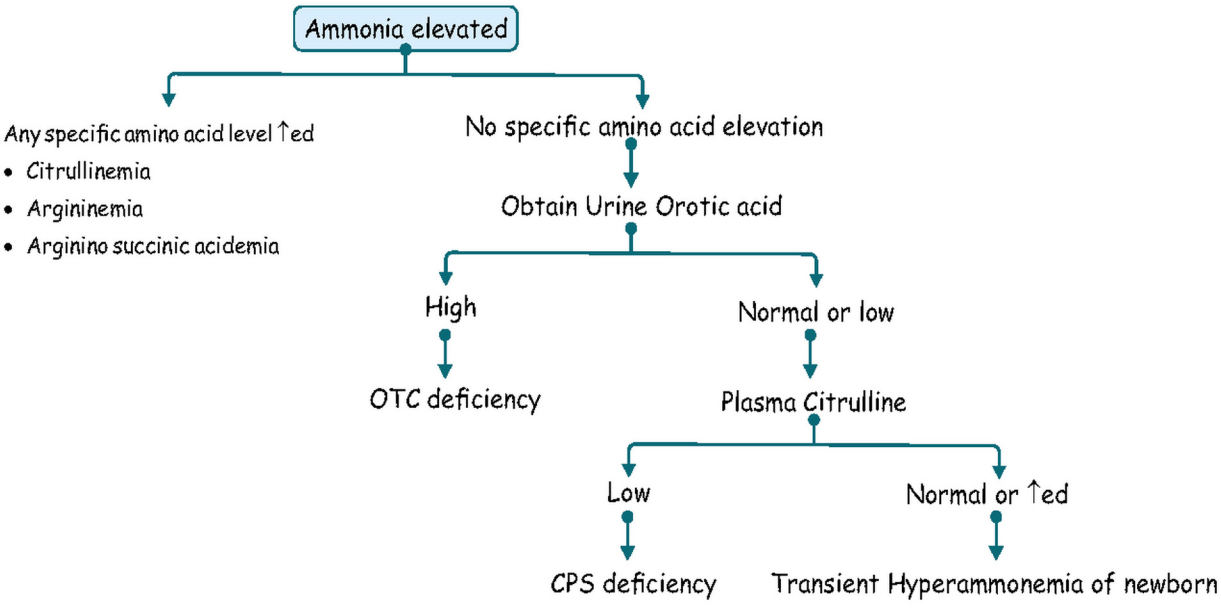
- Marfan syndrome - Supero-temporal dislocation of the lens

Inborn errors of metabolism with a peculiar odour

Inborn error of metabolism	Urine odour
Multiple carboxylase deficiency (Biotin is used as a treatment)	Tomcat
Glutaric aciduria	Sweaty feet
Tyrosinemia	Boiled cabbage
Phenylketonuria	Mousy/musty
MSUD	Burnt sugar or Maple syrup

HYPERAMMONEMIA

00:24:15



- Management of hyperammonemia
  - To reduce the level of ammonia and to prevent encephalopathy
    → Phenylacetate
    → Arginine
    → Benzoate
  - In refractory cases
    → Peritoneal or hemodialysis is used

HARTNUP DISEASE

00:25:48

- Mutation in SLC6A19 gene mutation
- Defect in the transport of neutral amino acids
- Clinical features
  - In most patient it is asymptomatic
  - In a symptomatic individual
    → Photosensitivity
    → Pellagra-like rash - Casal's necklace
- Autosomal recessive
- Investigation
  - Elevation of amino acids like tryptophan, valine, and tyrosine in the urine
- Treatment
  - Supplement with Niacin or nicotinamide
  - High protein diet

MAPLE SYRUP URINE DISEASE

00:27:11

- Deficiency of Branched Chain α ketoacid dehydrogenase (BCKD)
- Accumulation of amino acids like leucine, isoleucine, and valine causes intellectual disability
- Management
  - Dietary restriction of leucine, isoleucine, and valine

SCREENING TEST FOR INBORN ERROR OF METABOLISM

00:27:55

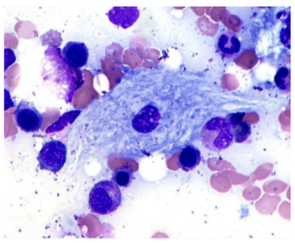
- Tandem mass spectroscopy
  - Dried blood spot collected on filter paper after the heel prick
  - Detect nearly 50 metabolic disorders
- Gas chromatography mass spectroscopy
  - Performed on the urine sample
- High-performance liquid chromatography/electrophoresis
- Definitive diagnosis is genetic

GAUCHER DISEASE

00:28:51

NEET PG 2024

Deficiency of glucocerebrosidase
↓
Accumulation of glucocerebroside
↓
In the cytoplasm of a macrophage
↓



GAUCHER CELL
(Crumpled tissue paper appearance)
↓
Gaucher cell infiltrates in various organs

Bone Marrow
- Anemia
  - Easy fatigability
- Leucopenia
  - Recurrent infection
- Thrombocytopenia
  - Bleeding manifestations
- On BM aspiration
  - Gaucher cell is seen

Spleen
- Massive splenomegaly

Bone
- Erlenmeyer flask deformity
  - Lytic lesions of ends of long bone
- Pathological fractures

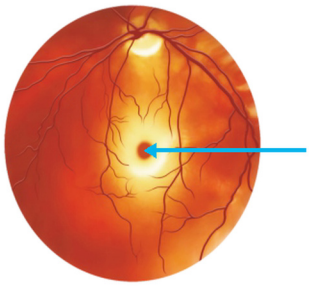
CNS
- Seizures
- Intellectual disability

- Treatment for Gaucher disease
  - Enzyme replacement therapy

Important Information
- Other diseases where enzyme replacement therapy is used
  - Pompe disease
  - Mucopolysaccharidosis
  - Type I - Hurler disease
  - Type II - Hunter disease
  - Type VI - Marateaux lamy syndrome
  - Fabry disease- α-galactosidase deficiency, presents with angiokeratomas

LYSOSOMAL STORAGE DISORDER

00:31:40



Disease	Cherry red spot	Visceromegaly	Skeletal involvement
Gaucher disease	-	Hepatosplenomegaly	Present
Niemann pick disease	+	Splenomegaly	May or may not be present
GM1 Gangliosidosis (beta - galactosidase -1)	+	Splenomegaly	Present
Tay-Sachs disease/ GM2 Gangliosidosis (Hexosaminidase A)	+	-	May or may not be present

Q.A child presents with neurodegeneration and a cherry red spot on fundus examination. Enzyme assays reveal Hexosaminidase A deficiency. What is the most likely diagnosis?

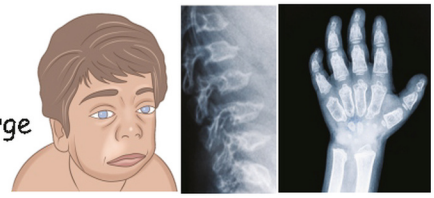
a. GM1 Gangliosidosis
b. GM2 Gangliosidosis
c. Niemann Pick disease
d. Gaucher disease

Ans: b

MUCOPOLYSACCHARIDOSIS

00:33:41

NEET PG 2023, 2024, 2025

Type	Name	Enzyme deficiency	Clinical feature
I	Hurler disease - severe Scheie disease - mild	α-L-Iduronidase	• Accumulation of substances like heparan and dermatan sulfate • Clinical features ○ Coarse facies ○ Global developmental delay ○ Intellectual disability ○ Hepatosplenomegaly ○ Protruded tongue ○ OSA ○ Cardiac involvement ○ Corneal opacity ○ Excessive nasal discharge ○ Dysostosis multiplex 
II	Hunter disease	Iduronate 2-sulfatase	• Features similar to Hurler disease, but no corneal opacity • X-linked recessive inheritance

Important Information

- All Mucopolysaccharidosis have AR inheritance except Hunter disease, which has X-linked recessive inheritance

Q.A child presents with developmental delay and coarse facial features. Enzyme assay reveals a deficiency of α-L-Iduronidase. Which of the following substances is accumulated?

a. Dermatan sulfate + chondroitin sulfate
b. Only Dermatan sulfate
c. Heparan sulfate + Dermatan sulfate
d. Heparan sulfate + chondroitin sulfate

Ans: c

Yourwish

WILSON DISEASE

00:37:54

- Pathogenesis

Defect in ATP7B gene, Chr 13q
↓
Defective incorporation of copper in apo-ceruloplasmin
↓
Copper is not excreted
↓
Excess copper is deposited in the liver, eyes, CNS, and other organs

- Clinical features
  - Liver
    → Acute/chronic hepatitis, hepatomegaly, jaundice, liver failure, portal hypertension
  - Eye
    → Kayser Fleischer ring
      – Copper deposited in descemet membrane of the cornea
      – Correlate with neurological feature
    → Sunflower cataract
  - Neurology
    → Copper is deposited in the basal ganglia
    → Chorea, tremors, dystonia, dysarthria
  - Gonads
    → Delayed puberty, hypogonadism, hypothyroidism, hypoparathyroidism, growth failure, and short stature
  - Hemolytic anemia
- Investigations
  - Screening test
    → Urinary 24-hour copper (Best) → elevated
    → D-penicillamine challenge test
    → Serum Ceruloplasmin → low
  - Confirmatory test
    → Liver biopsy- >250 µg/g of dry weight of liver
    → Genetic analysis
- Management
  - Zinc → Competes with the absorption of copper
  - D-pencillamine
  - Trientine

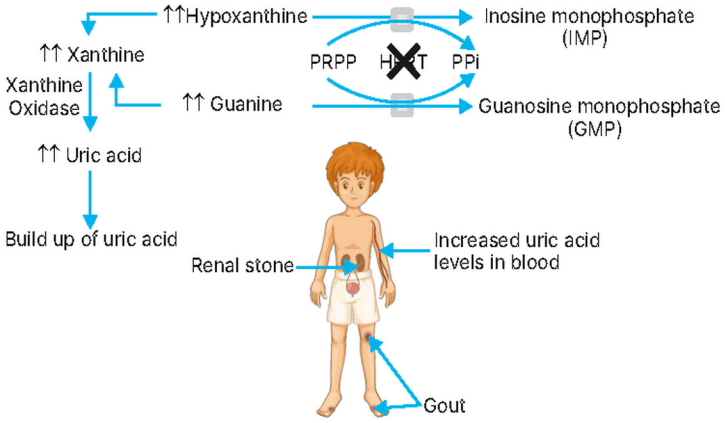
LESCH NYHAN SYNDROME

00:42:58

- Deficiency of Hypoxanthine Guanine Phosphoribosyl Transferase (HGPRTase)
- X-linked recessive
- More common in males
- Increased metabolism of xanthine and increased levels of uric acid in the blood
- Clinical features
  - Asymptomatic at birth
  - Developmental delay, intellectual disability
  - Neurological features

→ Dystonia
→ Dysarthria
→ Spasticity
- Self-injury/self-mutilation

- Diagnosis
  - Increased levels of uric acid
  - Deficient HGPRT enzyme
- Treatment
  - Allopurinol
  - Alkalinization
  - High fluid intake



MENKE DISEASE

00:44:25

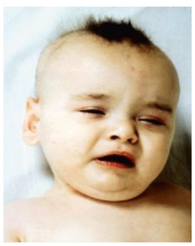
- Pathogenesis

Mutation in ATP7A gene
↓
Gene encoding copper transporting ATPase
↙ ↘
Copper levels decrease in liver & brain
Copper levels increase in fibroblasts & enterocytes

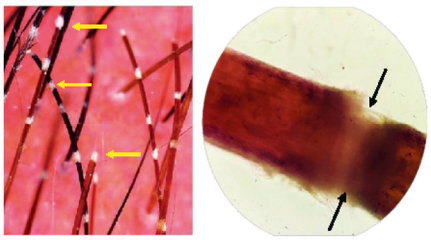
- Clinical features
  - Hair abnormalities
  - Seizures
  - Feeding difficulties
  - Failure to thrive
  - Hypothermia
  - Apneas
  - Poor prognosis - Death usually by 3 years of age

Hair Abnormalities

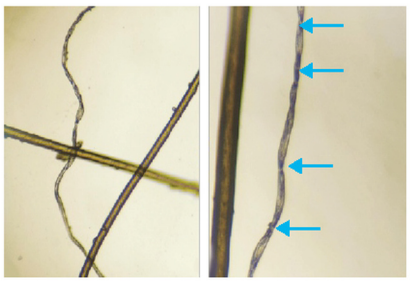
Fuzzy hair



Trichorrhexis nodosa: Splitting of the hair shaft



Pili torti: flattened and twisted hair



WOLMAN DISEASE

00:45:32

- Pathogenesis

Mutation in the LIPA gene

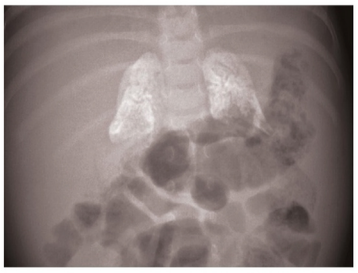
↓

Deficiency of lysosomal acid lipase

↓

Accumulation of TG & cholesterol esters in cells

- Clinical features
  - Hepatosplenomegaly
  - Jaundice
  - Vomiting
  - Diarrhea
  - Bilateral adrenal gland calcification



B/L adrenal gland calcification

7 IMMUNODEFICIENCY AND VASCULITIC DISORDERS IN CHILDREN

IMMUNODEFICIENCY DISORDERS

00:00:13

Severe combined immunodeficiency (SCID)

Q. A 6-month-old infant presents with fever, recurrent infections, and failure to thrive. Laboratory investigations reveals a deficiency of both T cells, B cells, and adenosine deaminase (ADA). Which of the following immunodeficiency disorders is most likely associated?

A. Severe Combined Immunodeficiency (SCID)
B. B cell defect
C. DiGeorge Syndrome
D. Wiskott-Aldrich Syndrome

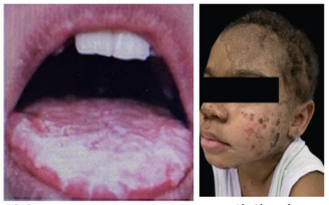
Ans. A

Basic defect

X-linked	Autosomal recessive
• IL7 defect - T cell affected • IL15 defect - NK cell affected	• Adenosine deaminase deficiency - ○ Loss of common precursors of B & T cells due to accumulation of deoxyadenosine in immature lymphocytes • JAK - 3 defect • IL-7 receptor defect

Clinical features

- Present in the 1st few months of life.
- Recurrent diarrhea, pneumonia, sepsis, and otitis media
- Persistent mucocutaneous candidiasis
- BCG vaccine can lead to disseminated BCG infection
  - Avoid all live vaccines



Muco-cutaneous candidiasis

Diagnosis

- Absolute Lymphocyte count <2500/mm^3
- Antibody levels are low
- Lymph node Biopsy - depleted T & B cell zones
- Thymus underdeveloped - devoid of Lymphoid cells
- Confirmation - Demonstration of the underlying genetic defect

Management

- HSCT (Hematopoietic stem cell transplant)
- Gene therapy
- PEG-ADA (specifically for the ADA-deficient type)
- Treatment of infections and avoidance of live vaccines (BCG, OPV, Rota Vaccine, MR)

Q. Which of the following vaccines is contraindicated in a patient diagnosed with Severe Combined Immunodeficiency (SCID)?

A. Hepatitis B

B. Tetanus, Diphtheria, and Pertussis (Tdap)

C. Inactivated Poliovirus (IPV)

D. Measles, Mumps, and Rubella (MMR)

Ans. D

Wiskott aldrich syndrome

00:04:10

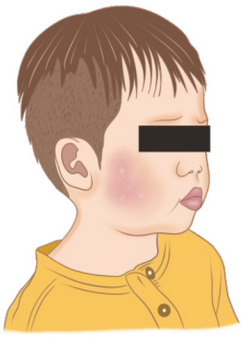
Q. A 5-year-old male child presented with H/O recurrent infection. O/E the child has rashes as shown below. Routine blood investigation reveals the patient has low platelets. Which of the following diagnosis is possible?

A. Job syndrome

B. Wiskott Aldrich syndrome

C. Measles

D. Henoch Schonlein purpura



Ans. B

Genetics

- Inheritance: X-linked recessive (XLR) — most common in males
- Genetic Defect: Mutation in the WASP gene (located on Chromosome Xp11)

Clinical features

- Recurrent infections
- Eczema-like rash
- Thrombocytopenia and small-sized platelets

Laboratory and diagnostic findings

- Microthrombocytopenia
- Immunoglobulins:
  - IgM - Low
  - IgA & IgE - Elevated
- Definitive Diagnosis: Genetic study to confirm WASP gene expression

Management

- IVIg
- Treatment of infection
- Symptomatic management

Bruton's x-linked agammaglobulinemia

00:07:18

Q. A 9-month-old child was admitted in ICU with a history of recurrent sinusitis and otitis media caused by Staphylococcus aureus. Blood test shows decreased serum IgA, IgG, IgM, IgE, and plasma B cells. What is the diagnosis?

A. Ataxia telangiectasia

B. Chronic granulomatous disease

C. Bruton's X-linked Agammaglobulinemia

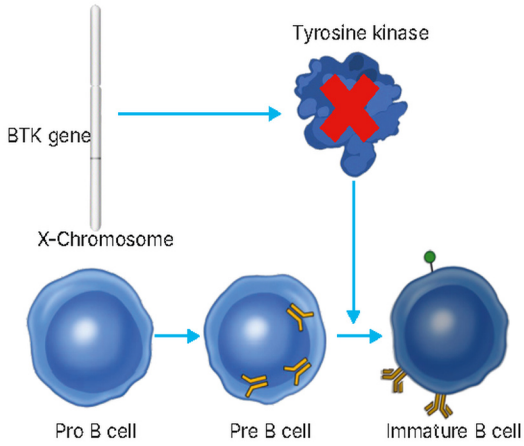
D. DiGeorge syndrome

Ans. C

- Ataxia Telangiectasia:
  - Autosomal recessive
  - Presents with cerebellar ataxia and telangiectasia in addition to combined immune defects
- Chronic Granulomatous Disease (CGD):
  - X-linked recessive
  - It is diagnosed by the NBT or Dihydrorhodamine assay

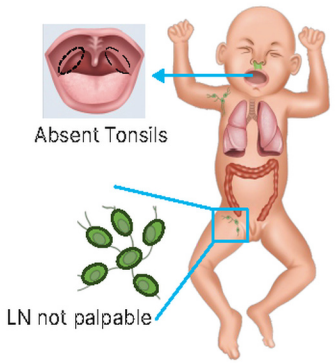
Genetics and pathophysiology

- Genetic Defect: BTK gene mutation on Chromosome Xq21
- Mechanism: Defect in Tyrosine kinase prevents the maturation of Pre-B cells into Immature B-cells



Clinical features

- Usually presents at 6-18 months of age
  - After maternal antibodies wane
- More common in males
- Tonsils and adenoids are absent
- Lymph nodes are not palpable (hypoplastic)



Diagnosis and management

- B-cells: Decreased or absent mature B cells (CD19, CD20)
- T-cells: Normally present
- Plasma cells: Absent
- Antibodies: Low Immunoglobulin (Ig) levels across all classes

- Treatment:
  - IVIg infusion
  - Treatment of infections

Common variable immunodeficiency (CVID) 00:11:00

- Autosomal recessive
- Both males and females are affected

Pathophysiology

- Inability of B cells to differentiate into plasma cells → CD19, CD20+
- Due to defects in BAFF (B cell activation factor for TNF-R) or ICOS (Inducible Co-Stimulator)
  - Immunoglobulins are not produced

Diagnosis and complications

- Hypogammaglobulinemia (Low Ig levels)
- Normal B cells (CD20 positive)
- Lymph nodes may be palpable (normal or enlarged)
- Tonsils and adenoids are present
- Complications: Increased risk of B-cell lymphomas and autoimmune diseases

Q. All of the following are contraindications for vaccinations except:

A. Digeorge syndrome

B. Wiskott Aldrich syndrome

C. Ataxia telangiectasia

D. Complement deficiency

Answer: D

Digeorge syndrome 00:13:11

Q. Case scenario with Facial abnormality, thymic hypoplasia, and Tetralogy of Fallot. Identify the syndrome:

A. Digeorge syndrome

B. Down syndrome

C. Turner syndrome

D. Klinefelter syndrome

Answer: A

Basic defect

- Also known as: Velocardiofacial syndrome
- 22q11 deletion → Failure of development of 3rd and 4th pharyngeal pouches
  - Thymic hypoplasia → Decreased T-cells
  - Parathyroid hypoplasia → Hypocalcaemia
  - Ultimobranchial body not formed
  - Defect in the heart & blood vessels

Clinical features

- **Mnemonic: CATCH-22**

C	Conotruncal/cardiac abnormalities	Tetralogy of Fallot, Truncus arteriosus
A	Abnormal facies	Hypertelorism, antimongoloid short abnormal nose, short philtrum, mandibular hypoplasia
T	Thymic hypoplasia	Absent thymic shadow on chest X-ray
C	Cleft palate	
H	Hypocalcemia	

Diagnostics

- FISH (Fluorescence In Situ Hybridization)
- MLPA (Multiplex Ligation-dependent Probe Amplification)

VASCULITIC DISORDERS

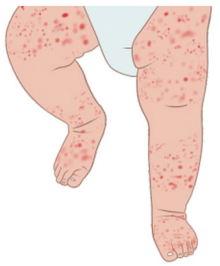
00:15:20

NEET PG 2021, FMGE 2021

Henoch-schönlein Purpura

Case scenario

Q. A child presents with abdominal pain, hematuria, and palpable purpura over the leg as shown in the image. Skin lesions show IgA deposition on biopsy. What is the diagnosis?



- Diagnosis: HSP

Diagnostic criteria

- Palpable Purpura with at least 1 out of 4 of the following criteria:
  - Abdominal pain
  - Joint involvement: Arthritis/arthralgia
  - Renal involvement: Hematuria, Proteinuria
  - Any biopsy showing IgA: Skin / Kidney
- Palpable Purpura is due to the dilatation of blood vessels
- Glomerulonephritis: Occurs in 1/3 of cases
- Thrombocytopenia: Absent

Management

- Self-limited
- Symptomatic management
- Corticosteroids: Used in severe cases, such as:
  - Severe abdominal pain
  - GI bleeding

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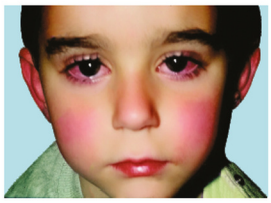
00:17:49

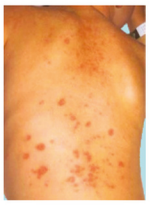
Kawasaki disease

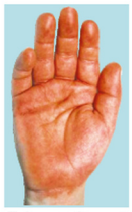
Clinical presentation

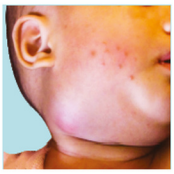
- A 4-year-old child presenting with fever and rash for 6 days, with any 4 of these 5:
  - Eyes: Bilateral non-purulent conjunctivitis
  - Mouth: Mucosal involvement → Strawberry tongue

- Extremities: Edema and erythema of extremities, followed by peeling of fingers
- Skin: Polymorphous rash
- Lymph Nodes: Adenopathy (often cervical)


Conjunctivitis


Rash


Edema & erythema of extremitres


Adenopathy


Mucosal involvement

Treatment

- Drug of Choice: IVIg (Intravenous Immunoglobulin) + Aspirin (High dose followed by low dose)

Complications

- Coronary Artery Aneurysm

Management of coronary artery aneurysm

- Continue Long-term prophylaxis

Severity	Treatment Regimen
Mild	Aspirin
Moderate	Aspirin ± Clopidogrel
Large	Aspirin + Warfarin / LMWH

- Thrombolytic therapy if required

Juvenile dermatomyositis

00:21:33

Q. Identify the given condition seen in a child with these rashes on the hand and bilateral proximal muscle weakness:

A. Scabies

B. Juvenile dermatomyositis

C. Osteoarthritis

D. RF

Answer: B

Diagnostic criteria for jdms

Classic Rash	Heliotrope rash of the eyelids Gottron papules

Plus any 3 of the following:	
Weakness	Symmetric Proximal
Muscle Enzyme Elevation (≥1)	Creatine kinase (CK) Aspartate transaminase (AST) Lactate dehydrogenase (LDH) Aldolase
Electromyographic (EMG) Changes	Short, small polyphasic motor unit potentials Fibrillations Positive sharp waves Insertional irritability Bizarre, high-frequency repetitive discharges
Muscle Biopsy (MRI of muscles is currently done)	Necrosis Inflammation

Systemic lupus erythematosus

00:23:13

Q. A 12-year-old child is diagnosed with systemic lupus erythematosus (SLE) and presents with nephrotic-range proteinuria. Renal biopsy reveals "wire loop lesions". Which of the following is the drug of choice in this case?

A. IV Steroids only

B. IV Steroids + Cyclophosphamide

C. Mycophenolate mofetil

D. Cyclophosphamide only

Answer: B

Diagnosis of sle in children (EULAR 2019)

Clinical Domains	Immunologic Domains
Constitutional Neuropsychiatric (seizures) Musculoskeletal (non-erosive arthritis) Hematologic (pancytopenia) Renal Mucocutaneous (alopecia, oral ulcers, butterfly rash) Serosal (pleuritis, pericarditis)	Antiphospholipid Ab Low Complement factors SLE-specific Ab (e.g., anti-dsDNA or anti-Sm)

- Total Score: ≥ 10 points
  - At least one clinical criterion must be present.
  - ANA ≥ 1:80

Classification and management of sle

Severity	Clinical Presentation	Recommended Treatment
Mild SLE	No renal or other life-threatening organ involvement	Weekly Methotrexate / HCQS
Mod. SLE	Clinically significant but no life-threatening involvement of the kidneys or other organs	Steroids / Immunomodulators
Severe SLE	Major organ-threatening disease	High-dose corticosteroids & immunomodulators

Who classification of lupus nephritis

Class	Type	Histological Findings
Class I	Minimal mesangial lupus nephritis	LM: Normal; EM/IF: Mesangial immune deposits
Class II	Mesangial proliferative nephritis	LM: Mesangial hypercellularity & increased matrix; EM/IF: Mesangial deposits containing immunoglobulin and complement
Class III	Mesangial and endocapillary lesions	EM/IF: Immune deposits in mesangium and subendothelial areas; • Class III affects <50% of glomeruli • Class IV affects ≥50% of glomeruli
Class IV		
Class V	Membranous lupus nephritis	Resembles idiopathic membranous nephropathy with subepithelial immune deposits

Signs of active disease

- Capillary walls that are thickened secondary to subendothelial deposits (wire-loop lesion)
- Necrosis
- Crescent formation

Important Information

- WHO class IV nephritis is associated with poorer outcomes but can be successfully treated with aggressive immunosuppressive therapy

8 PEDIATRIC HEMATO-ONCOLOGY

ANEMIA IN CHILDREN

00:00:07

NEET PG 2024

Who's Definition Of Anemia For Children

AGE	Hb CUT OFF
6 months-5 years	<11 gm/dl
5 years-11 years	<11.5 g/dl
12 years-14 years	<12 g/dl

Clinical Approach To Anemia In Children

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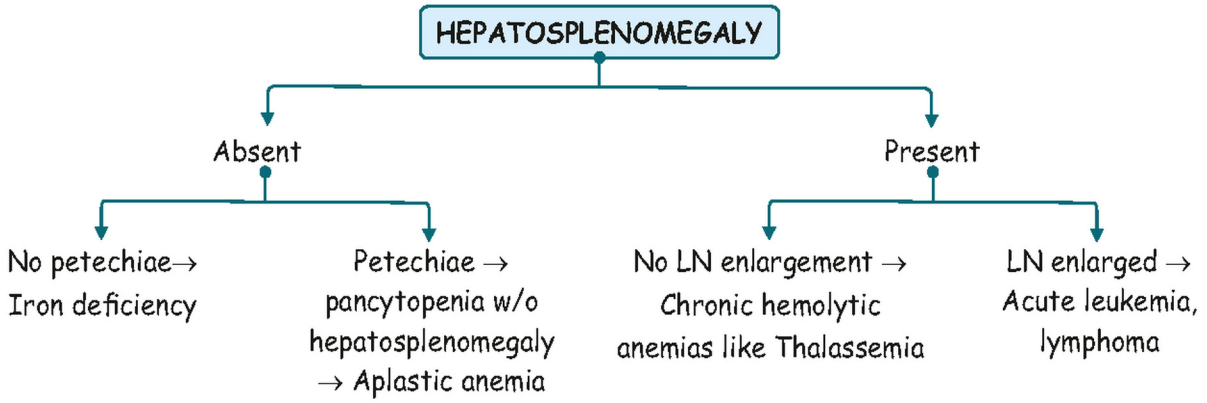
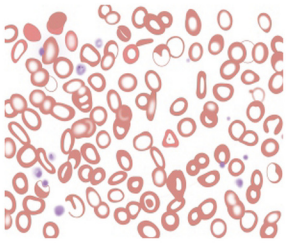


Microcytic Hypochromic Anemia

- Increased central pallor- Pale cells
- Smaller RBC cells



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D/D Of Microcytic Hypochromic Anemia

Parameter	Normal value	Iron deficiency	B-thalassemia trait	Anemia of chronic disease
Rdw	12-15	↑	normal	normal
S.iron	60-170 ug/dL	↓	normal/↑	↓
S.ferritin	15-300 ng/ml	↓	normal/↑	normal/↑
Tibc	250-400 ug/dl	↑	normal	↓

Mentzer Index

- Mentzer index= $\frac{\text{MCV (fl)}}{\text{RBC count (million/uL)}}$

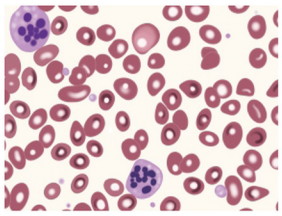
Yourwish

Mentzer index	Diagnosis
>13	Iron deficiency anemia
<13	Beta thalassemia trait

- Iron deficiency anemia, the RBC count is low→ index is high
- RBC count is normal in beta thalassemia trait→ index is less

Megaloblastic Anemia

00:06:40



- Hypersegmented neutrophils
- RBC large/macroovalocytes
- Cause
  - Vitamin B12 deficiency
  - Folic acid deficiency

Beta-Thalassemia

00:07:40

FMGE 2019

- Basic defect: Quantitative decrease in β-globin production
- Autosomal recessive
- Common mutations in India
  - IVS 1-5G→ C
  - IVS1-1G→ T
  - Codon 41/42

Clinical Features

- Chronic hemolytic anemia
- Unconjugated hyperbilirubinemia→ icterus
- Hemolytic facies
  - Frontal bossing
  - Parietal bossing
  - Depressed bridge of the nose
  - Maxillary prominence
  - Teeth prominence
- Hepatosplenomegaly
- Huge/massive splenomegaly

Iron Overload Complications

- Due to repeated transfusions
- Arrhythmias and heart failure
- Stunted growth and delayed puberty
- Hypothyroidism and diabetes
- Hepatitis and fibrosis

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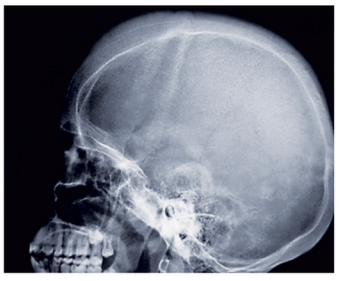

Radiological Features

- Crew cut appearance/Hair on end appearance
- Increased thickness of skull bone
- Can be seen in any chronic hemolytic anemia

Q. A child with Thalassemia on regular blood transfusion goes into an aplastic crisis. The virus responsible attaches to a receptor on erythroblasts. Which is the organism responsible?

a. Parvo B19
b. EBV
c. CMV
d. HIV

Ans: a. Parvo B19

Investigations

- Hb levels are low
- TLC and Platelets are usually normal
- Microcytic hypochromic, Anisopoikilocystosis, target cells on PS
- Unconjugated/indirect bilirubin rises
- Increased LDH
- Coomb's test negative
- Nestroft
  - Screening test previously used for thalassemia
  - Naked eye single tube red cell osmotic fragility test
- HbHPLC
  - HbA ($\alpha_2\beta_2$ decreased)
  - HbA_2 ($\alpha_2\delta_2$ increase)
  - HbF ($\alpha_2\gamma_2$ increased)
  - Beta thal trait $HbA_2 \rightarrow$ 3.5 to 7%
- Globin gene mutation
  - Confirmatory test
  - Prenatal diagnosis

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Management

- Regular PRBC transfusions
  - Pretransfusion Hb maintained between 9.5-10.5 g/dl
- Iron chelation- Started S.ferritin >1000 ng/ml
  - Desferroxamine parenterally
  - Deferiprone- oral drug
  - Deferasirox- oral drug
- Definitive therapy
  - Hematopoietic stem cell transplant

Sickle Cell Disease

00:18:25

- MC structural hemoglobinopathy
- Basic defect: 6^{th} position of β-globin gene glutamate replaced valine$\rightarrow$ Formation of HbS
- Autosomal recessive

Pathophysiology

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HbS formed with different physicochemical properties

↓

On deoxygenation, HbS forms long polymers

↓

Shape of RBC entered

- Microvascular obstruction
- Hemolysis

Clinical Features

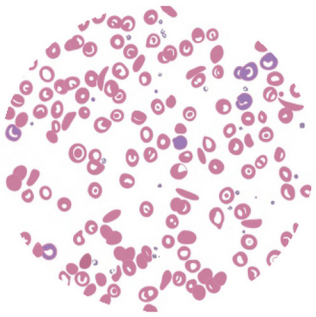
- **Anemia**
  - Jaundice
  - Initially splenomegaly→ splenic infarcts→ autospenectomy
- **CNS**- Stroke
- **Eye**- Retinitis, Vascular occlusion
- **Fingers**- Dactylitis
- **Spleen**- Splenic infarcts, Immunisation against capsulated organisms
- **Kidneys**- Acute papillary necrosis
- **Males**- Priapism

Crisis Situation

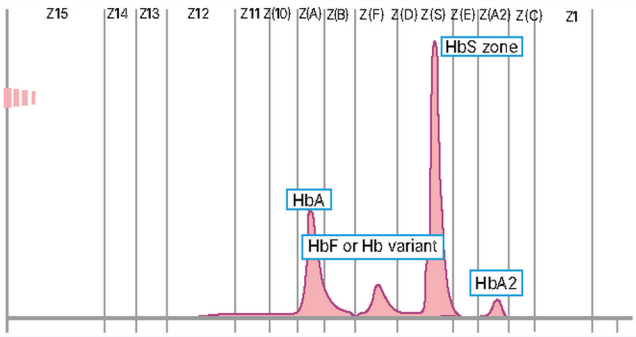
- Acute chest syndrome
  - Sudden chest pain
  - Breathing difficulty
- Vasoocclusive crisis
- Splenic sequestration
  - Pancytopenia
  - Increase in anemia
- Aplastic crisis
  - Due to parvovirus

Diagnosis

- PS shows abnormal, elongated sickle-shaped RBCs
- Sickling test
  - Add sodium metabisulfite
  - Sickling gets precipitated
- Hemoglobin electrophoresis



HB Fractions	%
HbA	29.0
HbF or Hb variant	8.3
HbS zone	59.8
HbA_2	2.9



- Confirmatory diagnosis
  - Genetic analysis
  - β-globin gene analysis

Treatment

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- Hydration
- Avoid/treat infections
- Analgesia
- Hydroxyurea- Increases HbF levels

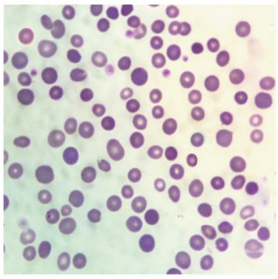
Newer Drugs

- Voxelator
  - Increases oxygen affinity
  - Decreases HbS polymerization
- L-Glutamine
  - Increases the level of antioxidants
  - Decreases oxidative stress in sickle RBC
  - Decreases frequency of painful crisis
- Crizanlizumab
  - Blocks P selectin
  - Reduces vaso-occlusion
- Casgevy
  - CRISPR-Cas9 edited autologous gene therapy
  - Curative treatment

Hereditary Spherocytosis

00:26:14

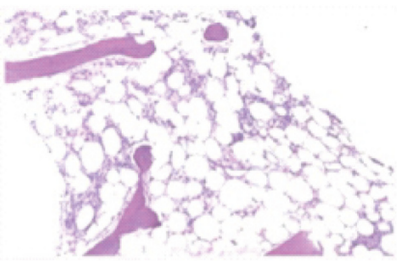
Q. A 2-year-old boy is brought in with episodes of icterus & pallor. On examination, splenomegaly is present, family history of similar illness in father; PS is shown below. What is the diagnosis?



- Autosomal dominant inheritance
- RBCS are spherical shaped
  - Loss of central pallor
  - A mutation in the coding region of proteins for the RBC membrane
- H/O pigment gallstones present

Q. A child with severe pallor presents with bleeding manifestations. On examination, no hepatoplenomegaly. His bone marrow aspiration revealed a dry tap, and the bone marrow biopsy showed that the marrow is full of fat globules. What is the probable diagnosis?

Ans: Aplastic anemia



- Bone marrow shows fat globules

Yourwish

Immunothrombocytopenic Purpura

Q. An otherwise well child presented with generalised petechiae & gum bleeding. There is h/o viral URI 2 weeks back. What is the diagnosis?

Ans: Immunothrombocytopenic purpura

- Low platelet count
- Bone marrow is normal
- Increased megakaryocytes

Treatment Of Acute Thrombocytic Purpura

- Wait and watch in children with no/mild bleeding
- Start treatment for moderate/severe bleeding
  - Oral corticosteroid- Prednisolone 2-4 mg/kg/day for 5-7 days
  - IVIg
- In refractory cases→ Splenectomy
- Platelet transfusion avoided
  - Antibodies present in the body
  - Only in life threatening bleed

Hemophilia A

00:30:37

Q. A mile child presented with recurrent episodes of knee/ankle swelling following trauma. There is a similar history in one of the maternal uncles. What is the diagnosis?

Ans: Hemophilia A

- Deficiency of clotting factor 8
  - Hemophilia B- Deficiency of factor 9
- X-linked recessive inheritance

Clinical Features

- Deep bleeding
  - In joints
  - In abdomen
  - Intracranial
  - Following surgery/trauma
  - In spontaneous and severe disease
- Superficial bleeds are not usually seen
- Muscle hematomas

Investigations

- Elevated apTT
- Normal PT
- Factor 8 assay
- Genetic analysis

Treatment

- Recombinant factor 8 concentrates as replacement therapy
- FFP/cryoprecipitates (cannot use in Hemophilia B d/t lack of factor 9)
- Avoid IM injections
  - Prefer the SC route
  - Schedule IM if necessary after factor replacement therapy

Q. A 2-year-old came with h/o epistaxis, recurrent rectal bleed, and bleeding from umblicus/. Platelet count and PT/APTT are normal, clot urea solubility test is positive. What is the deficient factor?

a. 10
b. 11
c. 12
d. 13

Ans: d. 13

Acute Leukemia

00:34:18

- MC hematological malignancy in children
- MC type pre B cell type
- T cell type is usually associated medicastinal mass

FMGE 2020

Clinical Features

- Fever
- Pancytopenia
  - Pallor
  - Leucopenia→ infections
  - Thrombocytopenia→ bleeding manifestations
- Lymphadenopathy
- Splenomegaly
- Joint pains

ALL	AML
• Lymphadenopathy • Joint involvement • CNS involvement	**M3** • DIC • BLeeding **M4/M5** • Gum hypertrophy • chloroma

Diagnostic Criteria

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- Blasts in PS or BM ≥20%
- ALL→ lymphoblast
- AML→ auer rods (myeloblast)
- Cytochemistry

Stain	All	Aml
Myeloperoxidase (mpo)	Negative	Positive
Sudan black b	Negative	Positive
Periodic acid schiff stain	Block positivity	Diffuse/negative
Tdt (terminal deoxynucleotidyl transferase)	Positive	Negative
Non specific esterase	Negative	Positive in m4/m5

- Flowcytometry
  - Blasts- Cd34
  - B Cells- Cd19
  - T cells- Cd3
  - NK cells- CD16, Cd56
  - MYELOID- MPO

Management Of All

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- Induction
  - Vincristine
  - Prednisolone
  - Anthracyclines
  - L-asparaginase
- CNS prophylaxis
  - Intrathecal methotrexate
  - Intracranial irradiation
- Consolidation
  - High-dose methotrexate
- Mantainence
  - 6-MP
  - Methotrexate

Prognostic Markers In All

Characteristics	Good	Bad
Age	1-10 years	<1yr, >10yr
Gender	Female	Male
Cytogenetics	t(12:21), hyperdiploidy	t(9:22), t(4:11)
Immunophenotype	pre-B cell	T cell ALL
Response to steroids (Day 8)	Present	Absent
Measurable residual disease	<0.01%	>0.01%

Management Of Aml

- **M3 type**
  - ATRA (All trans retinoic acid)
  - Arsenic
- Other types
  - Ara-c
  - Anthracyclines
  - Triple intrathecal therapy

Hodgkin Disease

00:41:30

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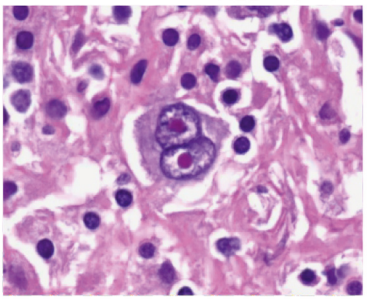
Clinical Features



- Fever
- Generalised lymphadenopathy
- Pancytopenia
- Hepatosplenomegaly
- Reed-Sternberg cells on lymph node biopsy

Treatment

- Adriamycin
- Bleomycin
- Vinblastin
- Dicarbazine

Langerhans Cell Histiocytes

00:42:34

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- Due to the clonal proliferation of Langerhans cells
  - Coffee bean-shaped nuclei

Clinical Features

- Single site- Bone/Skin/Lymph nodes
- Multiple sites
- Risk of organ involvement- Liver/Spleen/Bone marrow

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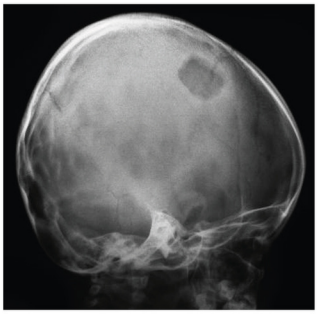
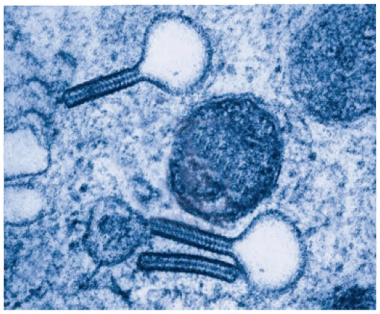
Diagnosis

- Punched out lesion in the skull
- CD1a positive
- Langerin positive Cd207
- Tennis racquet-shaped birkbeck granules





Treatment

- Prednisone
- Vinblastine

PEDIATRIC GASTROENTEROLOGY

Acute Diarrhea In Children

00:44:55

FMGE 2023

- MC Cause is rotavirus
- MC metabolic abnormality- Metabolic acidosis
  - No bicarbonate given
  - Correct dehydration

Classification

Parameter	No dehydration	Some dehydration	Severe dehydration
Sensorium	Active and alert	Irritable	Lethargic
Thirst	Normal	Excessive thirst	Not able to drink and breastfeed
Skin punch	Goes back quickly	Slowly <2 sec	Very slowly >2sec
Mucosa	Moist	Dry	Very dry
Eyes	Normal	Sunken	Very sunken
Tears	Present	Absent	Absent

Q. Best indicator of some dehydration in SAM is?

a. Sunken eyes

b. Thirst

c. Lethargy

d. Skin pinch

Ans: b

How Much Fluid Is To Be Given To A Child With

- **NO DEHYDRATION**
  - WHO ORS
  - 5-10 ml/kg/loose stool
- **SOME DEHYDRATION**
  - WHO ORS
  - 75 ML/g over 4 hrs
- **SEVERE DEHYDRATION**
  - IV RL in 5 % dextrose
  - IV RL alone
  - 100 ml /kg
  - Hospitalise the child

Fluid Rx In Severe Dehydration

- Total- 100ml/kg

AGE	1ST 30 ml/kg	NEXT 70 ml/kg	TOTAL (100 ml/kg)
<1 yr	1 hr	5 hr	6 hr
>1 yr	1.5 hr	2.5 hr	3 hr

- RL in 5 % dextrose-fluid of choice
- Give IV fluids I/C/O shock

Composition Of The Who Ors

NEET PG 2023
FMGE 2023

Composition	Quantities(mmol/l)
Na^+	75
glucose	75
Cl^-	65
K^+	20
Citrate	10
Total	**245**

Management Of Acute Diarrhea In Children

INICET 2022, 2025

- Treatment /prevention of dehydration
- Zinc-oral
  - 2M-6M age
  - >6M 20 mg/day
  - For a total of 14 days
- Continue a normal diet in children

Q. A child with 3 days of watery diarrhea, vomiting, and altered sensorium presented to the pediatric OPD. All of the following can be included in the differential diagnosis, except:

a. Hyponatremia
b. Severe dehydration
c. HUS
d. Cerebral vein occlusion

Ans: c

Persistent Diarrhea

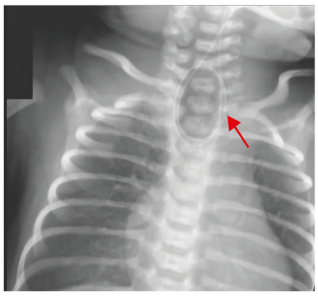
00:52:48

- A diarrhea of acute onset lasting for >14 days

Management

Dietary modifications	Supplementation
• **PLAN A** - Low lactose diet • **PLAN B**- Lactose-free diet • **PLAN C**- Elemental diet	• Zinc • Vitamin A

Q. A neonate presents with excessive frothing from the mouth & difficulty in feeding. There is also a h/o polyhydramnios in the antenatal period. What is the diagnosis?



Ans: Esophageal atresia with tracheo-esophageal fistula

- X-ray shows coiling of the feeding tube in the upper part of the oesophagus
- MC type- Type C
  - Esophageal atresia with distal tracheo-esophageal fistula

Congenital Hypertrophic Pyloric Stenosis

00:55:05

Q. A 5-week-old baby presents with recurrent episodes of non-bilious, projectile vomiting and hard pellet stools. O/E, visible peristalsis is seen in the abdomen. and a small olive-shaped mass is intermittently palpable in the abdomen. What electrolyte abnormality is expected in the child?

Ans: Hypochloremic metabolic acidosis with paradoxical aciduria

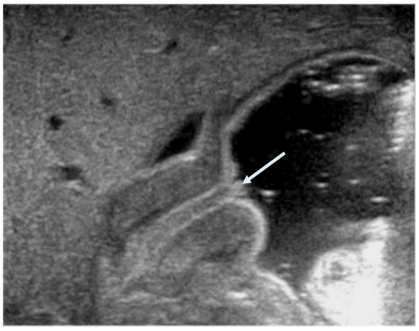
- Presents at 2-8 weeks
- 1st born males are more commonly affected
- Risk factors- Macrolide exposure

Clinical Features

- Non-bilious projectile vomiting
- Thickened pylorus→ olive shaped lump in the abdomen
- Left to right visible peristalsis
- Hypochloremic metabolic acidosis with paradoxical aciduria
  - Treatment fluid of choice: NS

Q. A 6-week-old baby with repeated non-bilious vomiting after feeds. The USG image given shows antral nipple sign or target sign. Diagnosis?

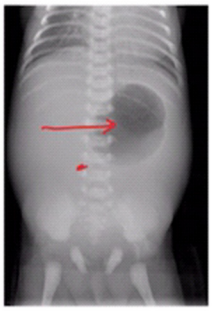
a. Congenital hypertrophic pyloric stenosis
b. Duodenal atresia
c. Annular pancreas
d. Jejunal atresia



Ans: a

Q. A 6-week-old baby is brought by her mother with a complaint of vomiting. The X-ray showed a single bubble appearance. Diagnosis?

a. Duodenal atresia
b. Annular pancreas
c. Congenital hypertrophic pyloric stenosis
d. Jejunal atresia



Ans: c

- X-rays show a single bubble appearance- **Congenital hypertrophic pyloric stenosis**
- Double bubble appearance- **Duodenal atresia**
- Tripple bubble appearance- **Jejunal atresia**

Treatment

- Hydration with NS
- Ramstedt's pylorostomy

Celiac Disease

01:00:40

Q. A 3-year-old girl presents with recurrent episodes of loose stools, failure to thrive, short stature, and anemia. Duodenal biopsy reveals villous atrophy. What is the probable diagnosis?

Ans: **Celiac disease**

- Gluten-containing products, such as wheat and wheat products, are not easily digested.
- Avoid BROW- **B**arley, **R**ye, **O**ats, **W**heat

Clinical Features

- Recurrent episodes of loose stools
- Anemia
- Short stature
- Sensitivity to wheat products
- Failure to thrive
- Duodenal biopsy reveals villous atrophy

Recommended Screening Test For Celiac Disease

Anti ttg iga	Total iga	Inference
Positive (>10 times ULN)	N	Anti-EMA (most specific) No biopsy diagnosis of celiac disease
Positive (<10 times ULN)	N	UGI endoscopy Duodenal biopsy
Negative	low	IgG-based test
Negative	Normal	Follow up with the patient

Yourwish

Duodenal Biopsy Findings

- Villous atrophy
- Crypt hyperplasia
- Increase in intraepithelial lymphocytes

Treatment Of Choice

- Lifelong gluten-free diet

Intususseption

01:04:50

Q. An 11-month-old baby was brought in with a H/o incessant crying and abnormal stool with blood & mucus. A tender mass palpable in the right lumbar region. Diagnosis?

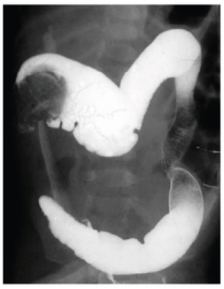
Ans: Intususseption

- Telescoping of bowel into the adjacent segment

Clinical Features

- Incessant crying
- Sausage-shaped abdominal mass
- Red currant jelly stools
- Recurrent severe abdominal pain
- Abnormal stool with blood and mucus
- A tender mass palpable in the right lumbar region

NEET PG 2022



Radiological Signs

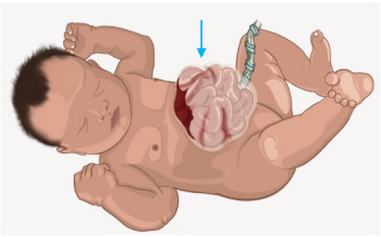
- **Claw Sign**
- Coiled Spring Sign -On Barium Studies

Omphalocele

01:05:40

Q. A neonate is found to have a swelling in the abdomen as shown in the picture. What is the diagnosis?

a. Omphalocele
b. Gastroschisis
c. Ectopia cordis
d. Umbilical hernia



Ans: a

- Gastroschisis- Swelling usually to the right side of the umbilical cord, not covered with cord.
- Ectopia cordis- Heart is exposed outside
- Umbilical hernia- Protrusion of bowels in the umbilical or paraumbilical region

Clinical Features

- Swelling in the abdomen
  - Sac containing bowel loops
  - Present in midline

- May contain part of the liver, bowel loops
- Tip of the umbilical cord is attached

- Commonly associated with congenital abnormalities

Treatment

- Definitive treatment is surgery

Hirschsprung Disease

01:07:13

Q. Which of the following is a cause of Hirschsprung's disease in a patient?

a. Failure of migration of neural crest cells
b. Excessive peristalsis of the affected part of the gut
c. Failure of involution of the vitelline duct
d. Obstruction secondary to an infectious agent

Ans: a

- AKA, Congenital aganglionic megacolon
- MC cause of lower intestinal tract obstruction in neonates and infants
- Defective migration of neural crest cells to the intestine
  - Absence of ganglion cells in the bowel
- The affected part of the bowel is not able to relax
  - The proximal part gets distended
- Limited to the rectosigmoid area in 80% of patients

Pathophysiology

Neonatal period with feed intolerance, bilious emesis, or failure to pass meconium

↓

Dilation of the proximal bowel and abdominal distension

↓

Intraluminal pressure goes on increasing

↓

Decreased blood flow

↓

Deterioration of the mucosal barrier and stasis

↓

Proliferation of bacteria

↓

enterocolitis

Diagnosis

- Rectal suction biopsy is the gold standard
- Acetylcholinesterase nerve bundles to show the hypertrophied nerve bundles
- Anorectal manometry→ internal anal sphincter fails to relax
- Contrast enema
  - Abrupt narrow transition zone between the normal dilated proximal and a smaller-caliber obstructed aganglionic segment

Q. Which of the following is/are seen in Hirschsprung disease?
1. Failure of migration of ganglion cells
2. Child will not pass meconium
3. Proximal segment constricted
4. Non-coordinated peristalsis

a. 1, 2, 3
b. 1, 2, 4
c. 1, 2
d. 3, 4

Ans: b

Poisoning In Children

01:10:12

- MC age group- under 5
- MC route- oral

NEET PG 2024
INICET 2024

Poison	Antidotes
Organophosphates	Atropine
Paracetamol	N-acetylcysteine
BZD	Flumazenil
Opioids	Naloxone
Iron	Desferroxamine
Heparin	Protamine sulphate

RESPIRATORY DISORDERS IN THE PEDIATRIC AGE GROUP

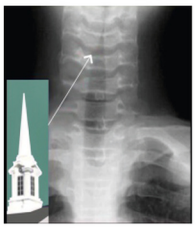
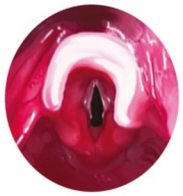
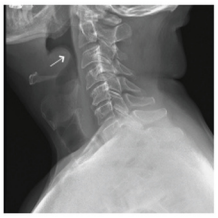
01:11:20

INICET 2020

- MC cause of stridor in infants- Laryngomalacia
- MC cause of common cold- Rhinovirus
- MC cause of bronchiolitis- RSV
- MC cause of bacterial pneumonia- Streptococcus pneumoniae/Pneumococcus
- Pneumonia in HSV positive child-pneumocystis jerovici
- MC cause croup- Parainfluenza virus
- MC cause of Acute epiglottitis-
  - Immunized- Streptococcus
  - Unimmunised- H.influenzae

Croup Vs Acute Epiglotitis

	Croup	Acute epiglotits
Clinical features	• Low-grade fever • Malaise • Corrhyza	• Bacterial infection • Toxic-looking child • Drooling of saliva

	• Stridor • Respiratory distress is mild to moderate • X-ray- Steeple sign  	NEET PG 2023, 2024 FMGE 2025 INICET 2024 • Stridor • Upper airway infection • Thumb sign on X-ray  
Treatment	• Dexamethasone-DOC Single dose • Nebulised epinephrine	• IV third-generation cephalosporins like ceftriaxone

Imnci Guidelines (Integrated Management Of Neonatal & Childhood Illnesses)

01:15:50

- Approach to a child with a cough and/or difficulty in breathing

Fast breathing	General danger signs
<2 months • RR >60 bpm	• Convulsions • Lethargy • Inability to feed • Persistent vomiting
2-12 months • RR >50 bpm	
1-5 yrs • RR >40 bpm	

Classification

- **In children 2 months -5 yrs**

Fast Breathing	Chest Indrawing	General Danger Signs	Category	Rx
±	±	+ or stridor in a calm child	Severe pneumonia/very severe disease	1st dose inj ampicillin and gentamycin
±	±	-	Pneumonia	Oral amoxicillin for 5 days Follow up after 2 days
-	-	-	No pneumonia cough/cold only	Home-based supportive care

Bronchial Asthma In Children

01:20:20
NEET PG 2021

Diagnosis

- H/o cough nocturnal
- H/o wheezing/rhonchi
- H/o Chest tightness
- H/o noisy breathing
- H/o difficulty breathing
- Positive family history

On Examination

- Pulmonary function test
  - PFT: FEV1/FVC- Decreased
  - Increase in FEV1 by 12% after bronchodilator
  - Exercise decreases FEV1
  - Diurnal PEFR variability increases

Latest Change In Gina Guidelines

NEET PG 2019
FMGE 2022

- Never use SABA MDI inhalers alone
  - Always add a steroid
- In children >6 years of age
  - Foracort inhaler (formoterol (LABA) + budesonide)
    → Metered dose inhalers
    → Components are
      – Blue is for SABA
      – Mask
      – Spacer

Management Of Status Asthmaticus

NEET PG 2024
INICET 2022, 2024

- Severe exacerbation of asthma

Hospitilise the patient
↓
Take care of airway/breathing/circulation
↓
Start oxygen humidification if required
↓
Continuous nebulisation with salbutamol + ipratropium bromide
↓
Still severe inj steroids
↓
Inj magnesium sulphate if still severe
↓
Theophylline may be used- narrow therapeutic index
↓
Mechanical ventilation and intubation if no improvement

9. NEONATOLOGY

IMPORTANT TERMINOLOGIES

00:00:50

Baby Born At	• <37 completed weeks of gestation: Preterm Neonate • 37-42 weeks gestation: Term Neonate • ≥42 weeks gestation: Post-term Neonate
Birth Weight	• <2500g: LBW (Low Birth Weight) baby • <1500g: VLBW (Very Low Birth Weight) • <1000g: ELBW (Extremely Low Birth Weight)

Birth weight percentile of gestational age	• <10th percentile of expected- SFD (Small for date) or SGA (Small for Gestational Age)
	  ○ Associated with IUGR
	• >90th percentile of expected - LFD (Large for date) or LGA babies
	  ○ IDM (Infant of a Diabetic Mother)
	  ○ Soto's syndrome
	  ○ Constitutional

NORMAL TERM NEONATE

00:03:55

- HR: 120-140/min
- RR: 40-60/min
- Peripheral/Acrocyanosis: +ve at birth
- Short systolic murmur: +ve
  - In fetal life, pulmonary vascular resistance is very high, but after birth, it goes down, and there is an increase in pulmonary blood flow, giving rise to a short systolic murmur

Assessment of gestational age

- Using the ENBS (Expanded New Ballard Score) for infants between 20 and 44 weeks
- The score consists of two main categories:
  - Physical Maturity
  - Neuromuscular Maturity
    - → Scarf sign
    - → Square window
    - → Popliteal angle
    - → Heel to ear

PRETERM NEONATE

00:07:08

GENERAL	HEAD TO TOE
• Small/emaciated- Hypothermia, Hypoglycemia	• Head appears to be relatively large

- Skin is thin, fragile- Neinatal sepsis
- Generalized Hypotonia- Extended posture
- Abundant Lanugo
- But little vernix caseosa

- Anterior fontanel large & wide open
- Ear cartilage- poorly formed
- Breast buds <5mm
- Soles- Absent deep creases on soles
- Genitalia
  - Males- Undescended testes & poorly formed scrotum
  - Female- Labia majora widely separated, labia minora clearly visible

CONDITIONS IN NEONATE NOT REQUIRING ANY SPECIFIC TREATMENT

00:10:49

SKIN / MUCOSA	OTHER CONDITIONS
• Erythema toxicum- Days 3-5 on trunk • Mongolian spots- bluish-black; lower back, buttocks • Stork bites- Capillary hemangioma • Epstein pearls- pearl-like lesions on the palate; epithelial inclusion cysts	• Mastitis neonatorum- Days 1-3 of life - B/L breast engorgement, in both male/female; ± milky discharge • Vaginal bleeding- Days 5-7, in female neonates due to withdrawal of maternal hormones • Physiological phimosis • Hymenal tags

PHYSIOLOGICAL WEIGHT LOSS IN NEONATES

00:15:00

CATEGORY	LOSE UP TO	IN HOW MANY DAYS	REGAINED BY
Term Neonates	10%	3 - 5 days	Day 10
Preterm Neonates	15%	5 - 7 days	Day 15

	CEPHALHEMATOMA	CAPUT SUCCEDANEUM
What is it due to?	Subperiosteal hemorrhage involving cranial bones	Edema in layers of the scalp
Does it cross sutures?	No	Yes
Appears in	24-48 hrs	Immediately at birth
Disappears in	5-7 weeks	48-72 hrs
Predisposes to Jaundice?	Yes	No

Important Information

- Subgaleal hematoma
  - Collection of blood under the muscle aponeurosis
  - The baby appears pale, features suggestive of shock are present, and tender swelling

Yourwish

PRIMITIVE NEONATAL REFLEXES

00:19.38

PRESENT AT BIRTH	APPEARS AT	DISAPPEARS AT
Rooting Reflex	32 wk	Starts disappearing at 1 mth
Moro's Reflex	28 wk - 37 wk	5 - 6 mths
Palmar Grasp Reflex	28 wk	3 mths- Leads to Voluntary grasp
Asymmetric Tonic Neck Reflex (ATNR)	35 wk	5-6 mths- roll over occurs

APPEARING AFTER BIRTH	APPEARS AT	DISAPPEARS AT
STNR (Symmetrical Tonic Neck Reflex)	4 - 6 mths	8 - 12 mths
Parachute	7 - 8 mths	Never disappears

Moros reflex/embrace equivalent

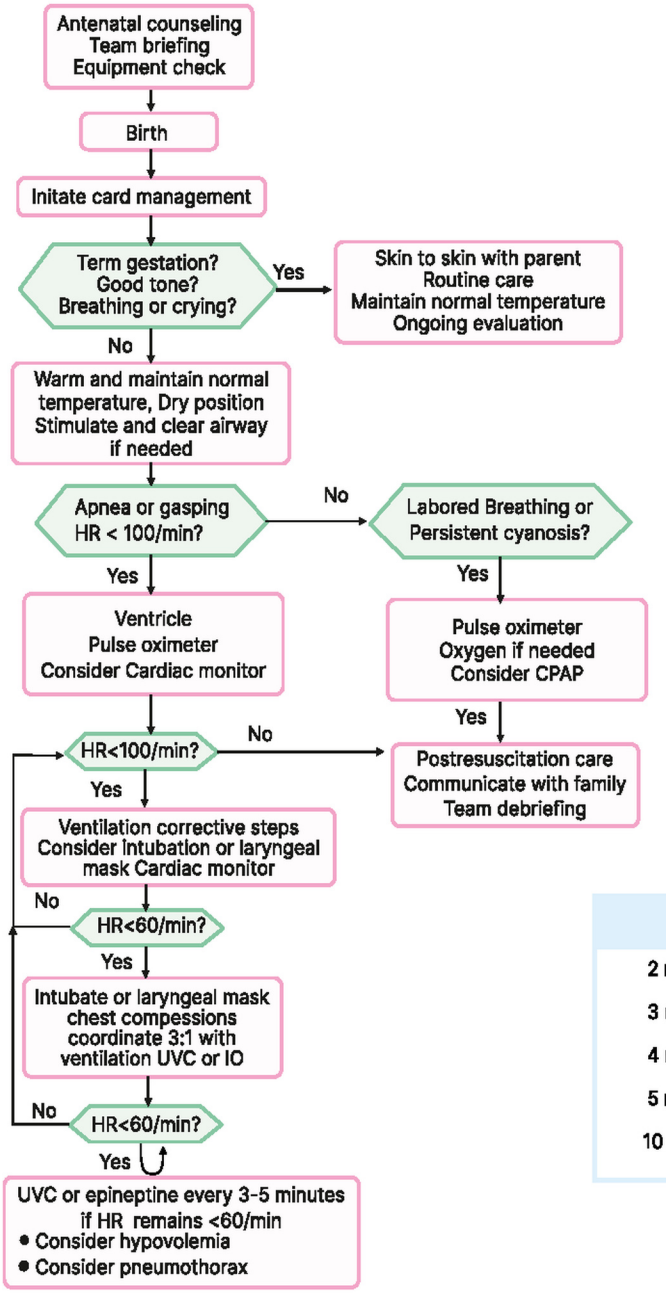
00:22:52

- Symmetrical extension of extension and abduction with opening of hands followed by flexion and adduction, along with extension of head and trunk, movement of lower limbs, and cry
- Begins to appear: 28 wk gestation
- Completely appears: 37 wk
- Disappears: 5-6 mths
- Persistent beyond 6 mths -Cerebral damage
- Absent Moro's Reflex:
  - Generalised Hypotonia- Down syndrome
  - Stage 3 HIE
- Asymmetric Moro's reflex is seen in:
  - Erb's palsy
  - Cong. Hemiplegia
  - Fracture Clavicle (MC bone to fracture in a neonate)
  - U/L Shoulder joint dislocation

NEONATAL RESUSCITATION PROTOCOL

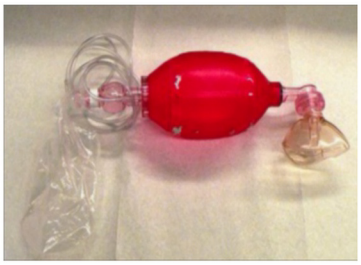
00:25:20

- Ideal temperature of delivery room: 25°C
- Suction during Neonatal resuscitation:
  - Routine suctioning is no longer recommended
  - Mouth followed by Nose
  - Gentle suction- suction pressure 80-100 mmHg
- Indications of PPV during N. resuscitation
  - Apnea
  - Gasping
  - HR <100/min
- PPV done using a self-inflating bag



Target Oxygen Saturation Table	
2 min	60%-65%
3 min	70%-75%
4 min	75%-80%
5 min	80%-85%
10 min	85%-95%

- Initial oxygen is not connected, but if the baby continues to have HR <60/min, oxygen is added
- PPV is done at 40-60 breaths/min
- Pressure for 1st breath- 30-40 cm/H_2O; subsequent breaths- 15-20 cm/H_2O



- Absolute contraindication to Bag & Mask Ventilation: Congenital diaphragmatic hernia

Cdh (congenital diaphragmatic hernia)

- Herniation of bowel loops into the thorax
- X-ray findings
  - Absent diaphragmatic border
  - Gaseous bowel loops in the thorax
  - Mediastinal shift to the right
  - Lungs not visible d/t pulmonary hypoplasia

- Bag and mask ventilation is contraindicated, as the air passing through the oesophagus leads to expansion of the stomach and bowel loops, leaving no space for the lungs to expand
- Posterolateral or Bochdalek type- MC (Most Common)
- MC on the left side
- Insert an NG tube (Nasogastric tube) for PPV
  - Bag & Tube ventilation (Endotracheal intubation is preferred over Bag & Mask)
  - Clinical Presentation at Birth
    → Respiratory distress
    → Scaphoid abdomen
    → Mediastinal shift
  - Complications
    → MC cause of Death: Pulm. Complication -Pulmonary Hypoplasia

Chest compressions in n. Resuscitation

- Indication HR<60/min
- Ratio of CC : PPV = 3 : 1
- Site Midline on the lower 1/3rd of the body of the sternum
- Use of Oxygen 100%

Injection adrenaline

- Indication: Heart Rate (HR)<60/min despite effective ventilation & Chest Compressions (CC) for 60 sec
- The earliest indicator is improvement in HR
- Dose: 0.02 mg/kg/dose or 0.2 ml/kg/dose of 1:10,000 Adrenaline
- Preferred route:
  - UVC / IO (Umbilical Venous Catheter / Intraosseous)
  - Through an endotracheal tube- requires a higher dose

Resuscitation of a baby born through meconium-stained liquor (msl)

- Previously Recommended (No Longer Routine)
  - Intrapartum suction of mouth and nose
  - Routine ET intubation and tracheal suction
- Current Protocol
  - NRP (Neonatal Resuscitation Program): Follow the standard NRP algorithm regardless of meconium presence.
  - At least 1 person trained in ET intubation should be available at the resuscitation

Essential new born care

00:41:25

- Keep baby warm & dry
- Resuscitation, if required
- Care of eyes
- Care of the cord
- Initiation of breastfeeding within 1 hr of delivery
- No care of the ear
- No bathing immediately

Major changes in the neonatal resuscitation 2025 guidelines by aap

- For all term or preterm newborn infants who do not require immediate resuscitation, delayed cord clamping for at least 60 seconds can be beneficial.

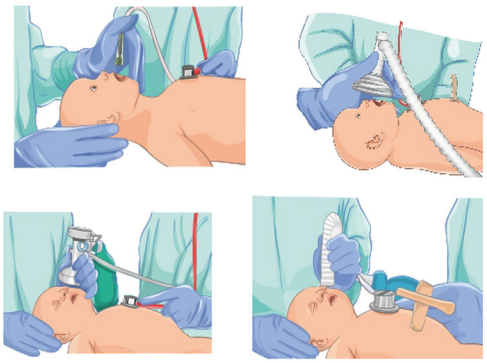
- For non-vigorous term and late preterm infants (35 weeks or more), intact cord milking may be reasonable when compared to immediate cord clamping.
- For newborn infants born at less than 28+0 weeks of gestation, cord milking should not be performed.

Latest changes in the neonatal resuscitation 2025 guidelines by aap for use of oxygen

- Prior neonatal guidelines have used the term positive-pressure ventilation to refer to providing ventilation.
- In this update, we generally use the term ventilation or assisted ventilation in order to simplify terminology.
- Initial Oxygen Concentration for Ventilation
  - Ventilation may be initiated with:
    - → Air (21% oxygen) in term and late preterm newborn infants
    - → 21% to 30% oxygen in preterm babies, 32 to 35 weeks of gestation
    - → 30%-100% in very preterm infants <32 weeks of gestational age
  - Target Oxygen Saturation

Time	Pre-ductal SpO_2 Target
2 min	65%-70%
3 min	70%-75%
4 min	75%-80%
5 min	80%-85%
10 min	85%-95%

Q. Free flow oxygen can be given in a neonate by all EXCEPT?
A. Self-inflating bag and mask
B. Tail/reservoir of self inflating mask
C. Face mask over mouth and nose
D. Tubing over the mouth and nose



FEEDING OF PRETERM NEONATE

00:46:04

GESTATIONAL AGE	RECOMMENDED INITIAL MODE OF FEEDING
<28 weeks	IV fluids only/ TPN MEN- Minimal enteral nutrition can be tried (10-15 ml of expressed breast milk fed
28-31 weeks	OG tube feeding (Gavage feeding)
32-34 weeks	Katori spoon / Paladai feeding
>34 weeks	Direct breastfeeding

Yourwish

NEONATAL SEPSIS

00:48:47

- Most common organism responsible for Neonatal sepsis:
  - In India: Acinetobacter > Klebsiella
  - In hospitals of India: Acinetobacter > Klebsiella
  - In hospitals across the world: E. coli
  - Overall, throughout the world: Group B Streptococcus
  - Early Onset Sepsis: Group B Strep
- Most important method to prevent Neonatal sepsis: Proper handwashing of caregivers
- Sepsis Screen:
  - A sepsis screen is considered positive if two or more of the following parameters are met:
    - → Total Leukocyte count: <5000/mm^3
    - → Absolute Neutrophil count: <1800/mm^3
    - → Immature: Total Neutrophil ratio is >0.2
    - → Micro ESR: >15 mm (in the first hour)
    - → C-Reactive Protein: +ve

- Confirmatory test to diagnose N. Sepsis: Blood culture
- Rx of choice for N. Sepsis:
  - Supportive care
  - IV Ampicillin + Gentamicin
  - IV Cefotaxime (If meningitis is suspected)
- Duration of Antibiotic Therapy

SEPSIS SCREEN	BLOOD C/S	CSF S/O Meningitis	DURATION
-	-	-	3 days
+	-	-	1 week
±	+	-	2 week
±	±	+	3 week

Q. A neonate born at full term, normal vaginal delivery, presents at 14th day of life with B/L conjunctivitis, and her chest x ray show B/L opacities present. What is the probable underlying cause?

A. Streptococcus agalactiae
B. Streptococcus pneumoniae
C. Chlamydia pneumoniae
D. Hemophilus influenzae

Answer: **C**

ETIOLOGY OF NEONATAL MORTALITY IN INDIA

00:52.31

- Prematurity and low birth weight (48%)
- Birth asphyxia and birth trauma (13%)
- Neonatal infections (12%)
- Congenital anomalies (7%)

NEONATAL HYPOTHERMIA

00:53.10

- Definition: Axillary temperature <36.5°C in a neonate
- Classification based on axillary temperature
  - Cold stress: 36 - 36.4°C
    → Clinical definition- Abdomen of the baby is warm, but the soles are cold
  - Moderate Hypothermia: 32 - 35.9°C
  - Severe Hypothermia: <32°C
- Most important mechanism for protection against hypothermia in neonates:
  - Shivering is absent in neonates
  - Non-Shivering Thermogenesis
    → Due to Brown fat - Mitochondria-rich lipid deposits
    → Located in: Axilla, Groin, Nape of neck, Interscapular

Kangaroo mother care (kmc)

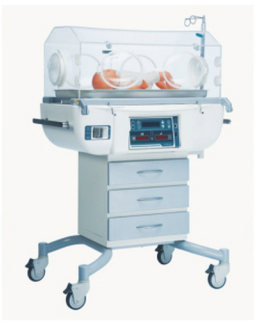
00:56:00

- Indication:
  - All Stable LBW (Low Birth Weight) babies
- Components:
  - K. position (Kangaroo position)
  - K. nutrition (Kangaroo nutrition)
  - Early discharge from the hospital
- Advantages:
  - Higher wt gain/improvement in anthropometry
  - Increase in exclusive breastfeeding rates
  - Lesser N. sepsis- Lesser N. mortality
  - Early discharge from the hospital

Devices used to keep neonates warm:

1. Radiant Warmer

- Mechanism of Heat Transfer: Radiation
- Mechanism of Heat Loss: Convection



2. Incubator

- Mechanism of Heat Transfer: Convection
- Mechanism of Heat Loss: Radiation

Warm chain

- Warm delivery room
- Warm resuscitation
- Immediate drying
- Skin-to-skin contact between the baby and the mother
- Breastfeeding
- Postpone bathing
- Appropriate clothing and bedding
- Keep mother and baby together
- Warm transportation
- Awareness-raising of healthcare provider

NEONATAL HYPOGLYCEMIA

01.00.00

- Definition
  - Blood glucose: <40 mg/dl
  - Plasma glucose: <45 mg/dl
- Clinical Features
  - Most common: Jitteriness/tremors
  - Others: Apnea, seizures
- Treatment
  - Asymptomatic (or BG >20 mg/dl):
    → Offer a feed & recheck blood glucose after 1/2 hr
  - Symptomatic (or BG <20 mg/dl):
    → IV 10% Dextrose bolus, then start continuous IVF (Intravenous Fluids) & BG monitoring

Important causes of persistent hypoglycemia during infancy

Endocrine cause	Metabolic disorders
• Cong. Hypopituitarism • Cong. Adrenal Insufficiency • Cong. Hyperinsulinemia or PHHI (Persistent Hyperinsulinemic Hypoglycemia of Infancy)	• GSD (Glycogen Storage Disease) • HFI (Hereditary Fructose Intolerance) • Galactosemia • FAOD (Fatty Acid Oxidation Disorders) • Mitochondria diseases

PROBLEMS IN INFANT OF DIABETIC MOTHER

01.04.23

- Core Mechanism: Excess insulin is secreted in the fetus/baby
- Large for date/Macrosomia
  - Birth trauma
  - Difficult labour leading to Perinatal asphyxia
- Metabolic
  - Hypoglycemia
  - Hypocalcemia
  - Hypomagnesemia
  - Polycythemia
  - Hyperbilirubinemia
- Respiratory
  - Respiratory Distress Syndrome d/t delayed maturation of surfactant
- Cardiovascular System
  - Most common CHD: VSD
  - Most specific CHD in IDM: TGA
- Neurologic
  - Most Common: Neural Tube Defects
- Most specific congenital abnormality in IDM: Sacral agenesis / Caudal Regression Syndrome

Q. A neonate was born to a diabetic mother. At the time of birth, he was fine; 4 days later, he developed dyspnea with SpO_2 80%. What is the initial management?

A. Give O_2 21% to 30%
B. Give 100% O_2
C. Give 50% O_2
D. Observation

Answer: **B**

- Hyperoxia test- Used to distinguish between hypoxia caused by pulmonary or cardiac causes
  - Give 100% O_2 for 10-15 mins and observe
  - If saturation improves- Pulmonary cause
  - If saturation does not improve- Cardiac cause

PERINATAL ASPHYXIA

01.08.44

- Inability to initiate or sustain breathing
- Parts of the brain affected due to asphyxia:

NEONATAL STATUS	MCINVOLVED AREA	TYPE OF CEREBRAL PALSY
Term neonates	Parasagittal area	Spastic Quadriparesis
Preterm neonates	Periventricular area	Spastic Diplegia

- Diagnosis of severe birth asphyxia:
  - APGAR score: 0-3 for >5 minutes
  - Cord blood pH: <7
  - CNS dysfunction: e.g., seizures, Tone abnormalities
  - Dysfunction of other organs: e.g., AKI, liver injury
- APGAR Score Interpretation
  - >7: Normal
  - 0 - 3: Severely depressed

FEATURE	0	1	2
Appearance	Completely blue or pale	Body pink, extremities blue	Completely pink
Pulse rate	Absent	<100/min	> 100/min
Grimace	No response	Grimaces	Coughs/cries
Activity	Limp and flaccid	Some flexion +	Flexed posture with normal movements
Resp. effort	Absent	Slow & irregular	Good & strong

STAGING OF HIE

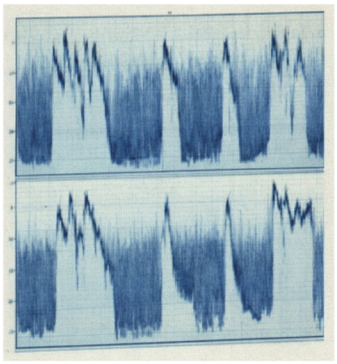
01.12.46

Clinical Feature	Stage 1 (Mild)	Stage 2 (Moderate)	Stage 3 (Severe)
Sensorium	Hyperalert	Lethargic	Comatosed

Yourwish

Tone	Normal	Mild Hypotonia	Severe Hypotonia
Moro's reflex	Exaggerated	Depressed	Absent
Seizures	Absent	Present	Absent
Heart rate & Pupils	Sympathetic overactivity- Tachycardia; mydriasis	Parasympathetic overactivity- Bradycardia; miosis	Both systems depressed- Variable HR & mid-dilated pupils

- Monitoring: an EEG (amplitude integrated Electroencephalogram)
- Management: Supportive
- Newer modality: Therapeutic Hypothermia



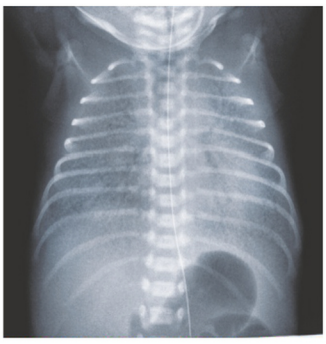
NEONATAL SEIZURES

01.16.39

- MC type: Subtle
- MC cause: Hypoxia
- Type with worst prognosis: Myoclonic
- Best outcome: Focal clonic seizures
- Preferred 1st line neuroimaging in neonates: Transcranial Ultrasound
- Drug of choice:
  - 1st: Phenobarbitone
  - 2nd: Levetiracetam

RESPIRATORY DISTRESS SYNDROME

01.17.30



- MC cause of respiratory distress in a: Preterm Newborn
- Basic defect: Deficiency of mature surfactant
- Surfactant:
  - Produced by: Type 2 Alveolar cells
  - Synthesis begins in fetal lungs at: 20 weeks
  - Begins to appear in Amniotic fluid: 28 weeks
  - Mature & adequate amount: 35 weeks
  - Most important component of surfactant: DPPC or Lecithin
  - Most important surfactant protein: Surf. protein Type B
- Function of surfactant: Reduce surface tension of alveoli & improve lung compliance
- When mature surfactant is deficient:
  - Alveolar collapse during expiration
  - Interstitial edema
  - Diffuse alveolar damage
  - Fibrin deposition
- X-ray findings
  - Ground glass haziness of the lungs

- Air bronchograms
- Lung collapse

Q. A Preterm newborn is found to have respiratory distress at birth. The most probable reason for this is?

A. Increased compliance and surface tension

B. Decreased compliance and surface tension

C. Decreased compliance and increased surface tension

D. Increased compliance & decreased surface tension.

Answer: **C**

Ways to detect the adequacy of mature surfactant in amniotic fluid

- L:S ratio (Lecithin : Sphingomyelin) > 2:1
- Nile blue sulfate test
- Shake test
- Direct estimation of Phosphatidyl glycerol

Silverman-anderson score

- In a preterm neonate with respiratory distress syndrome
  - 0-3: Normal / Mild distress
  - >7: Severe respiratory distress

ASSESSMENT CRITERIA	SCORE 0	SCORE 1	SCORE 2
Upper Chest Retractions	Chest and abdomen move equally with respiration	Chest wall lags behind the abdomen	See-saw / Paradoxical breathing
Lower Chest Retractions	None	Minimal	Marked
Xiphisternal Retraction	None	Minimal	Marked
Nasal Flare	None	Minimal	Marked
Grunt	No grunt	Audible on a stethoscope	Audible without a stethoscope

Downe's score

- In a term neonate for respiratory distress
  - 0-3: Normal
  - >7: Severe Respiratory Distress

COMPONENTS	0	1	2
Cyanosis	No cyanosis	Present on room air	Present at FiO_2 > 40%
Air entry	Normal	Decreased	Barely audible

Respiratory rate	<60/min	60-80/min	>80/min
Grunt	Absent	Loud, Audible with a stethoscope	Audible without a stethoscope
Retraction	None	Mild	Moderate-Severe

Treatment of RDS

- Mild: CPAP (Continuous Positive Airway Pressure)
- Moderate to severe:
  - Intratracheal Surfactant + Resp. support (CPAP / mechanical ventilation)
  - Key Procedures:
    → INSURE: Intubate - Surfactant - Extubate
    → LISA: Lesser Invasive Surfactant Administration

Prevention of RDS

- Antenatal Corticosteroid
  - Indication: Pregnant ladies expected to deliver at 24-34 weeks of gestation
  - Contraindication: Clinical Chorioamnionitis
  - Dosage Regimen

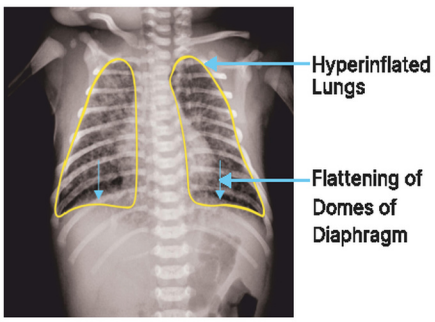
MEDICATION	EACH DOSE	NO OF DOSES	FREQUENCY	Total Dose
Betamethasone lesser side effects	12 mg	2	Every 12 hrs	24 mg
Dexamethasone Cheaper and easily available. Recommended by GOI	6 mg	4	Every 6 hrs	24 mg

- Benefits: Antenatal Corticosteroids decrease the risk of -
  - RDS (Respiratory Distress Syndrome)
  - IVH (Intraventricular Hemorrhage)
  - NEC (Necrotizing Enterocolitis)
  - Neonatal mortality

MECONIUM ASPIRATION SYNDROME (MAS)

01.31.28

- Case Scenario- SGA/IUGR baby born through meconium-stained liquor with respiratory distress appearing soon after birth
- CXR Findings
  - Patchy opacities: Representing areas of atelectasis.
  - Hyperinflation: Seen as flattened diaphragms or increased lung volume.
  - Snowstorm appearance: Coarse, irregular densities scattered throughout the lung fields.



Q. Greenish black stool in a neonate is due to?

A. Amniotic fluid
B. Bile salts
C. Lanugo
D. Bile pigments

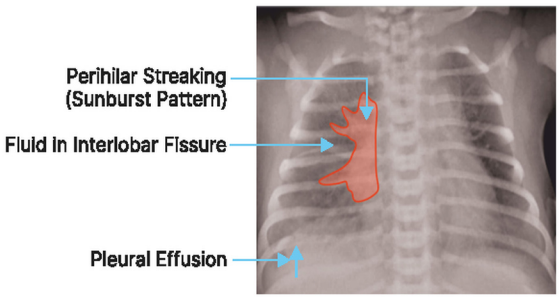
Answer: **D**

- Meconium also contains
  - Mucus
  - Denuded intestinal epithelial cells
  - Water

TRANSIENT TACHYPNEA OF NEWBORN (TTNB)

01.32.45

- Due to: Delayed clearance of lung fluid
- MC in babies born through: Caesarean section
- C/F: Term baby born by LSCS with mild to mod distress soon after birth that improves by 48-72 hrs
- CXR
  - Perihilar streaking
  - Interlobar fissure fluid
- Management: Supportive



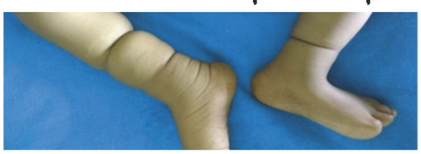
NEONATAL APNEA

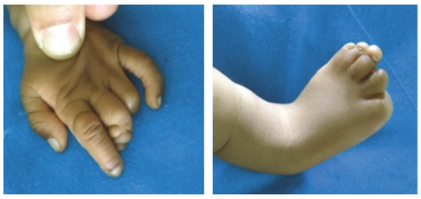
01.34.18

- Definition: Cessation of breathing for at least 20 sec or for any duration in the presence of either Bradycardia or Cyanosis in a neonate
- Etiology:
  - N. Hypothermia
  - N. Sepsis
  - N. Hypoglycemia
  - Polycythemia
  - N. Hypocalcemia
  - N. Hyperbilirubinemia
- Drug of choice for Apnea of Prematurity:
  - Inj. Caffeine citrate

Q. A newborn is noted to have constriction rings around the fingers and indentations over the superior aspect of the knee at birth. What is the most likely diagnosis?

A. Congenital Constriction Band
B. Amniotic Band Syndrome
C. Congenital Talipes Equinovarus
D. Arthrogryposis Multiplex Congenita





Answer: **B**

Yourwish

AMNIOTIC BAND SYNDROME

01.37.38

Fibrous bands from the amniotic sac constrict the body parts of a developing fetus

↓

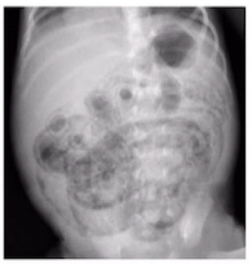
Deformities, including constriction rings around digits or limbs & indentations/amputations

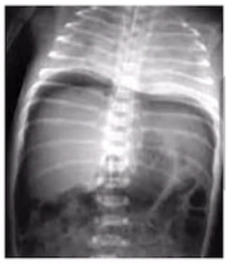
NECROTISING ENTEROCOLITIS (NEC)

01:38:19

- Important risk factors:
  - Prematurity
  - Use of formula feeding / Lack of breastfeeding
- Stages of NEC (Bell's Staging)

Ia	Occult blood +ve in stool
Ib	Fresh blood in stool +ve
IIa	Pneumatosis intestinalis
IIb	Portal vein gas
IIIa	Peritonitis
IIIb	Pneumoperitoneum





- Rx:
  - Supportive + NPO and start IV fluids and IV antibiotics
  - Supportive-O_2/mechanical ventilation, blood products, Ionotropes
  - IIIb - requires surgery

NEONATAL JAUNDICE

01.43.46

- Clinical Jaundice in newborns: Bilirubin level >5mg/dl

PHYSIOLOGICAL JAUNDICE	PATHOLOGICAL JAUNDICE
Icterus never seen in the 1st 24 hrs	May appear on the 1st 24 hrs
Palms & soles are never yellow	Palms & soles may be yellow
Always Unconjugated	May be conjugated or unconjugated
Jaundice does not persist beyond 3 weeks	May last longer

Q.Choose the correct option. Physiological jaundice findings are:-

A. Increased direct bilirubin, increased urobilinogen & increased urine bilirubin

B. Increased direct bilirubin, decreased urobilinogen, decreased urine bilirubin

C. Increased indirect bilirubin, & Increased urine bilirubin & decreased urobilinogen

D. Increased indirect bilirubin, & increased urine urobilinogen & decreased urine bilirubin

Answer : D

- In physiological jaundice (a type of unconjugated hyperbilirubinemia):
  - Indirect (unconjugated) bilirubin is elevated because the liver is not yet fully processing it.
  - Urine bilirubin is absent (decreased/negative)
  - Urine urobilinogen increases because the liver eventually conjugates some bilirubin, which then enters the gut and is recycled.

BREASTFEEDING JAUNDICE	BREASTMILK JAUNDICE
• Due to inadequate breastfeeding	• Due to (d/t) substances in breast milk like pregnanediol & Free Fatty Acids that interfere with the conjugation of bilirubin

- Breastfeeding is continued unless bilirubin levels are >20 mg/dl

Etiology of unconjugated hyperbilirubinemia

01:47:34

Increased production of bilirubin	Decreased Conjugation of Bilirubin	
Hemolytic Causes	**Non-hemolytic causes**	This primarily involves genetic deficiencies of the UDPGT enzyme:
• Rh or ABO incompatibility (Erythroblastosis fetalis)- In severe cases, Hydrops fetalis (Immune Hydrops) • Thalassemia • G6PD deficiency • Hereditary Spherocytosis	• Cephalhematoma • Polycythemia	• Crigler-Najjar Syndrome: Type I: Complete deficiency of the UDPGT enzyme. Type II: Partial deficiency of the UDPGT enzyme. • Gilbert Syndrome

Q. Causes of non-immune hydrops fetalis are all EXCEPT:

A. ABO incompatibility

B. Congestive cardiac failure

C. Parvovirus infection

D. Chromosomal anomalies

Answer: **A**

Etiology of non-immune hydrops fetalis

01:51:05

- Anemia: Alpha-thalassemia
- Bone: OI (Osteogenesis Imperfecta), skeletal dysplasia
- CNS: NTD (Neural Tube Defects)
- Cardiac: HLHS (Hypoplastic Left Heart Syndrome), Congenital heart block, CCF
- Flow-related: Twin-twin transfusion
- Genetic causes: Trisomy 13, 18, 21; Noonan
- Hepatic causes: Congenital hepatic Fibrosis
- Infections: Toxo, Rubella, CMV, Parvovirus
- Inborn Errors of Metabolism: Gaucher, Niemann-Pick Disease, MPS (Mucopolysaccharidosis)
- Kidney: Congenital Nephrotic Syndrome

Conjugated bilirubinemia

01:52:35

- Criteria: Unconjugated Bilirubin >2mg/dl or >20% of total bilirubin
- Non-obstructive Infection
  - Urinary Tract Infection
  - Metabolic-
    - → Alpha 1-AT deficiency
    - → Cystic Fibrosis
    - → Galactosemia
    - → Idiopathic Neonatal Hepatitis
- Obstructive
  - Intrahepatic
    - → Congenital Hepatic Fibrosis
    - → Caroli's disease
    - → Progressive Familial Intrahepatic Cholestasis
    - → Dubin-Johnson syndrome
    - → Rotor syndrome
  - Extrahepatic
    - → Extrahepatic Biliary Atresia

Biliary atresia

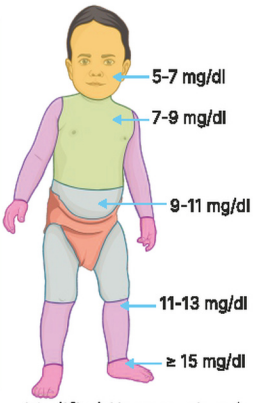
- Screening test for EHBA: HIDA Scan / Hepatic Scintigraphy
- Surgery: Kasai procedure
- Poor prognosis if done >8 wk age
- Most common indication for liver transplantation in children- Biliary atresia

Modified kramer's rule

01:55:23

- Cephalo-caudal progression

BODY REGION	LEMON YELLOW (Mild)-mg/dl	ORANGE YELLOW-mg/dl
Face	5-7	7-9
Chest	7-9	9-11
Lower abdomen & thigh	9-11	13-15
Arms & legs	11-13	15-17
Palms & Soles	13-15	>17



Modified Krammer's rule

Cns manifestations

- Hyperbilirubinemia: Kernicterus
- Part of the brain MC (most commonly) involved: Basal ganglia
- Type of Cerebral palsy seen: Extrapyramidal type
- Features of acute Bilirubin Encephalopathy:
  - Early: Poor feeding, Lethargy, Hypotonia
  - Late: Seizures, Shrill cry, Hypertonia (Opisthotonic posturing)
- Features of chronic Bilirubin Encephalopathy:
  - SNHL (Sensorineural hearing loss)
  - Athetosis
  - Intellectual Disability

- Upward gaze palsy
- Dental enamel changes

Rx of neonatal jaundice

- Phototherapy
- Exchange transfusion
- IVIg

Phototherapy

- Most effective wavelength: 450-460 nm
- Most important mechanism: Structural isomerization (Bilirubin → Lumirubin)
- Ways to increase the effectiveness of Phototherapy:
  - Decrease the distance between the baby & Phototherapy unit
  - More Surface area exposed
  - LED lamps
- When to start phototherapy- Otherwise well-term neonate

AGE	PHOTOTHERAPY CUT OFF (MG/DL)	EXCHANGE TRANSFUSION CUT OFF
24-48 hrs.	≥15 mg/dl	≥20 mg/dl
48-72 hrs	≥18 mg/dl	≥25 mg/dl
>72 hrs.	≥20 mg/dl	
Preterm Neonates	1% of birthweight in grams. Example: For a 1200g baby >12 mg/dl	Phototherapy cut-off + 5. Example: 12 + 5 = 17 mg/dl

Adverse effects of phototherapy

- Retinal toxicity
- Gonadal toxicity/mutation defect
- Dehydration
- Watery diarrhea
- Hypocalcemia
- Impaired maternal-child bonding
- Bronze baby syndrome

Exchange transfusion

- Indication of Exchange Transfusion in a baby with Rh incompatibility:
  - Cord blood bilirubin >5 mg/dl
  - Cord blood Hb <10g/dl
  - HCT <30

Q. A term neonate, with a birth weight of 2700 g, who is otherwise well, and is exclusively breastfed, presents for routine evaluation. His total serum bilirubin is found to be 14mg/dl on day 3. What is the management?

A. Phototherapy
B. Exchange transfusion
C. Stop breastfeeding for 2 days
D. No active treatment required

Answer: **D**

10. SYSTEMIC PEDIATRICS PART 1

PEDIATRIC CARDIOLOGY

00:00:24

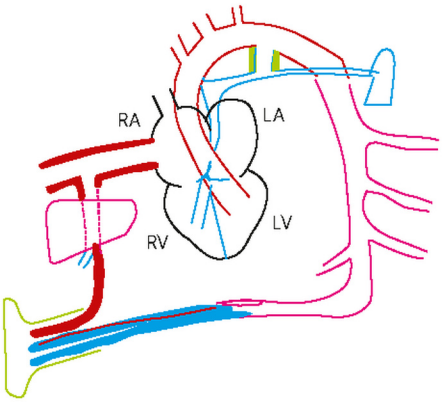
00:00:30

INICET 2020

Fetal Circulation

Placenta
↓
Umbilical cord
(2 Arteries + 1 Vein)
↓
Portal vein
↓
Ductus venosus
↓
IVC
↓
Right atrium → Right ventricle
↓ ↓
Foramen ovale — Pulmonary artery
↓ ↓
Left atrium — Ductus arteriosus
↓ ↓
Left ventricle — Descending aorta
↓ ↓
Aorta — Supplies all other organs
↓ ↓
Branches of arch of aorta — Umbilical artery
↓ ↓
Supplies: Brain, Head, and neck — Placenta

	Percentage of oxygen saturation
Umbilical vein	80%
IVC	70%
Left ventricle	65%
Right ventricle	55-60%



Structure In Fetal Circulation With Decreasing Oxygen Saturation

Umbilical vein
↓
Ductus venosus
↓
IVC
↓
Left ventricle
↓
Ascending aorta
↓
Descending aorta
↓
Umbilical artery
↓
SVC

Changes In Fetal Circulation After Birth

INICET 2020

- Lung become the source of oxygen
- Pulmonary vascular resistance decreases → Pulmonary blood flow increases
- Systemic vascular resistance increases
- The foramen ovale closes
- Ductus venosus closes
- Ductus arteriosus closes
  - Functional closure → Immediately after birth
  - Anatomical closure → Day 10 to Day 21 of life

Pressure In Different Chambers Of The Heart In Adult Circulation

- Established by the age of → 2 to 3 weeks
- In the presence of heart disease (like VSD or PDA) → 6 to 10 weeks

Heart chambers	Pressure (mm Hg)
Right atrium	0-6
Right ventricle • SBP • DBP	25 0-6
Pulmonary artery • SBP • DBP	25 10
Left atrium	6-10

Left ventricle • SBP • DBP	80-120 6-10
Aorta • SBP • DBP	80-120 60-80

Nada's Criteria

INICET 2023

- Used for the assessment of the presence of CHD

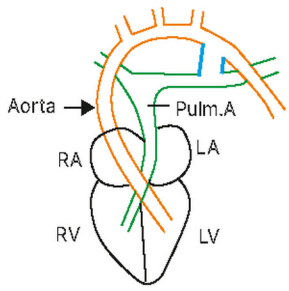
Major criteria	Minor criteria
• Any diastolic murmur • Systolic murmur ≥ grade 3 • Cyanosis • Heart failure	• Systolic murmur < grade 3 • Abnormal BP • Abnormal S2 • Abnormal ECG • Abnormal CXR

CLASSIFICATION OF CHD

00:11:47

INICET 2020

- Left → Right shunt (systemic → pulmonary circulation)
  - Acyanotic heart disease
    - → ASD
    - → VSD
    - → PDA
- Right → Left shunt
  - Eisenmenger syndrome → irreversible changes in pulmonary circulation
  - Tetralogy of Fallot
- Complete mixing of blood of the left and right sides
  - Truncus arteriosus
    - → Pulmonary artery and the aorta arise from a single trunk
  - TAPVC
    - → SVC, IVC, and Pulmonary veins drain into the right atrium
- Parallel circulation
  - TGA
- Ductus dependent CHD
  - Ductus-dependent systemic circulation → Obstruction at the level of systemic circulation
    - → Coarctation of aorta
    - → Interrupted aortic arch
    - → Severe or Critical Aortic Stenosis
  - Ductus-dependent pulmonary circulation → Obstruction at the level of pulmonary circulation
    - → Severe pulmonary stenosis
    - → Pulmonary atresia



→ Severe ToF
→ Ebstein anomaly
→ Tricuspid atresia

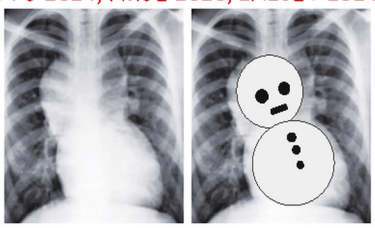
Q. Increased pulmonary blood flow is seen in which of the following congenital heart diseases?
a. Tetralogy of Fallot
b. Ebstein anomaly
c. Transposition of great arteries + Ventricular septal defect
d. Tricuspid atresia
Ans: c

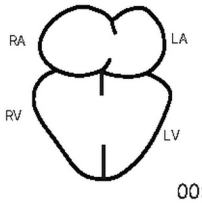
Tapvc

00:18:03

NEET PG 2024, FMGE 2020, INICET 2024

- Supracardiac
  - Complete mixing of the blood of both sides of the heart
  - Alprostadil → PGE1 analogue is not useful
  - On CXR → Snow man or Figure of 8 appearance
- Infracardiac type
  - Obstructive





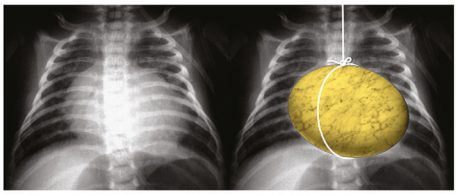
Q. In which heart disease, all 4 chambers have equal Oxygen saturation?
Ans : TAPVC

Transposition Of Great Arteries

00:20:10

FMGE 2025
INICET 2020

- M/c congenital heart disease in infancy
- On CXR → Egg on side appearance
- TGA with intact ventricular septum
  - No communication between the systemic and pulmonary circulation
  - Medical emergency
  - Treatment
    → Balloon atrial septostomy / Rashkind procedure
    → Alprostadil or PGE1 can be used
    → Definitive → Arterial switch operation / Jatene's repair

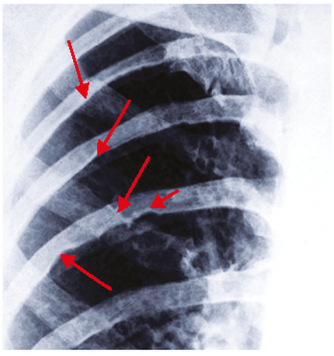


Coarctation Of Aorta

00:21:23

INICET 2022

- Narrowing of aorta
- Juxtaductal
- Decreased blood flow to the lower limb presents as absent or feeble lower limb pulse
- Clinical presentation
  - Severe
    → Present in the neonatal period
    → Heart failure with feeble or absent lower limb pulse
  - Moderate
    → Hypertension present
  - Mild
    → Intermittent claudication of the lower limb
- On CXR

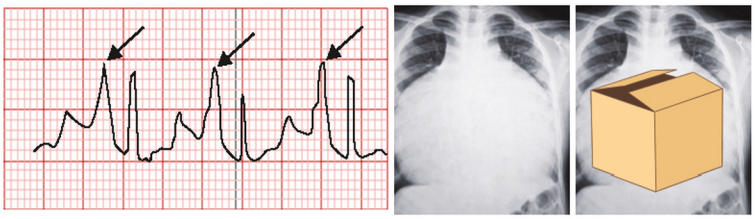


- Notching of the inferior margins of rib → collaterals formed between the anterior and posterior intercostal arteries
- Figure of 3 or E sign → pre- and post-stenotic dilatation

Ebstein Anomaly

NEET PG 2021
FMGE 2021, 2024, 2025
INICET 2024, 2025

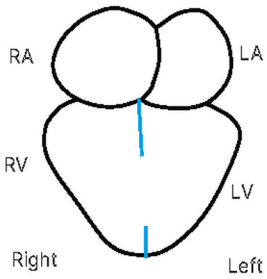
- Tricuspid valve is abnormal
- Downward displacement of the tricuspid valve
- Atrialization of Right ventricle
- Cardiomegaly → mainly the Right atrium
- On CXR → Box-shaped heart
- On ECG → Himalayan P wave



00:23:50
FMGE 2020

Ventricular Septal Defect

- M/c CHD in children
- M/c Congenital acyanotic heart disease in children
- M/c Congenital heart disease affected by Infective Endocarditis
- Part of the ventricular septum most commonly involved → Membranous
- Hemodynamics
  - Abnormal Communication between LV and RV
  - Blood flow from LV → RV
  - More pressure gradient during Systole
- Clinical presentation
  - Presents around the age of 6-10 weeks
  - Pan-systolic murmur
  - Tachypnea
  - Tachycardia
  - Recurrent respiratory tract infection
  - Heart failure → failure to thrive
  - Enlargement of LA and LV
- On CXR
  - Cardiomegaly with LV type Apex
- On ECG
  - Left axis deviation
  - Features of LVH
- Management



NEET PG 2024
INICET 2024

Medical

- Treatment of heart failure
  - Frusemide
  - Enalapril
  - Digoxin
- Treatment of LRTI
- Nutritional rehabilitation

Surgical

- Closure of VSD before puberty
- Indication
  - Refractory heart failure
  - Op : Qs > 2:1

- Complication of VSD

NEET PG 2024
INICET 2024

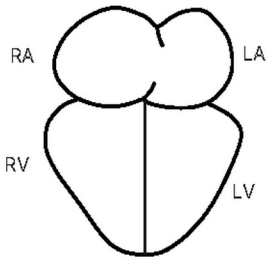
  - Infective endocarditis
    → VSD + Fever
  - Eisenmenger syndrome
    → VSD + Cyanosis + Clubbing

Atrial Septal Defect

00:28:35

NEET PG 2024
FMGE 2020, 2025
INICET 2024

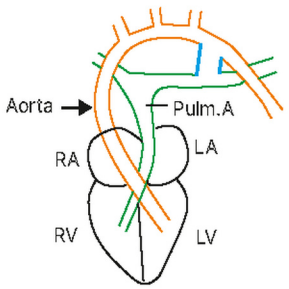
- Hemodynamics
  - Abnormal Communication between LA and RA
  - Blood flow from LA → RA
  - No pressure gradient → Silent Shunt in ASD
  - Wide fixed split of S2
- Types
  - Ostium primum → Lower part of the septum involved
  - Ostium secundum → Upper part of the septum involved
- On ECG
  - Ostium primum → Left axis deviation
  - Ostium secundum → Right axis deviation
- Clinical presentation
  - Asymptomatic throughout life
  - Severe ASD → Similar to VSD

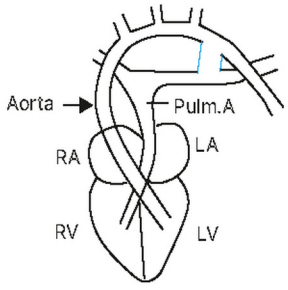


Patent Ductus Arteriosus

00:30:25

- Ductus arteriosus
  - Connects the pulmonary artery to the aorta (distal to the origin of the left subclavian artery)
- Factors predisposing to PDA
  - Hypoxia
  - Prematurity
- Clinical presentation
  - More pressure difference between the aorta and pulmonary artery during both systole and diastole
    → Continuous machinery murmur
  - Recurrent respiratory infection
  - Heart failure → Failure to thrive
  - In Preterm: presents during the 1st week of life with heart failure, bounding pulses
  - In Term: presents during 6-8weeks of life, like VSD
- Management
  - Drug of choice for medical closure of PDA in preterm neonates → PG inhibitors
    → Ibuprofen
    → Indomethacin
    → Paracetamol





Differential Cyanosis

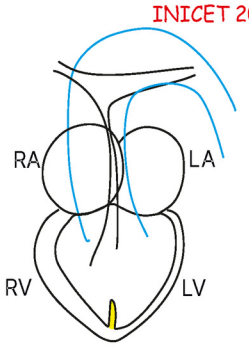
- Seen in PDA with reversal of the shunt
  - No cyanosis in the upper limb

- Cyanosis is present in the lower limb

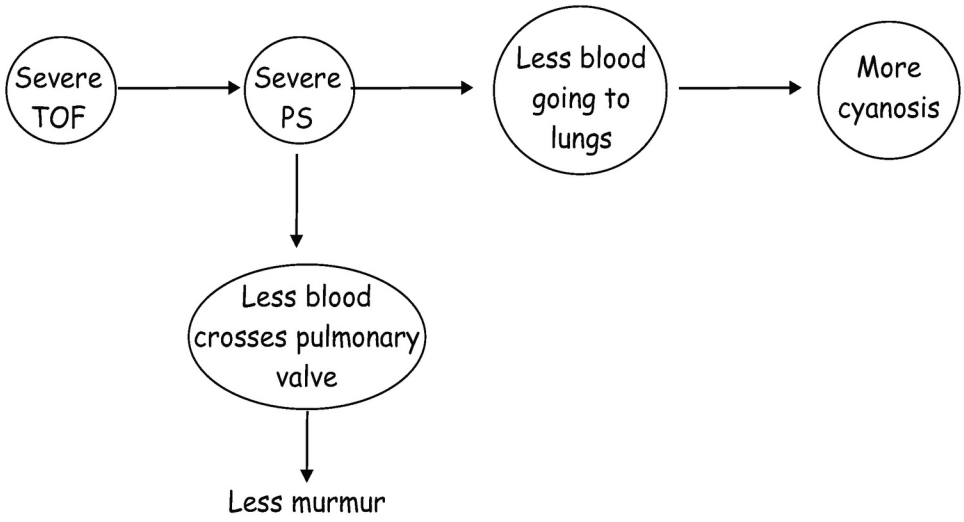
Tetralogy Of Fallot

00:34:48

INICET 2020

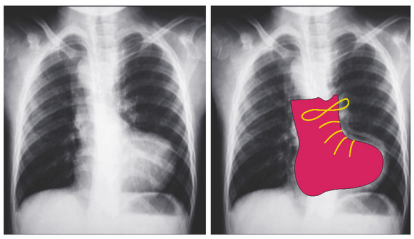


- Components
  - Large VSD
  - Pulmonary stenosis
  - Overriding of aorta
  - RVH
- Clinical presentation
  - Cyanosis
  - Clubbing
  - Polycythemia
  - Ejection systolic murmur → Pulmonary area
  - Single heart sound → because P2 is soft and inaudible



- On CXR

NEETPG 2021



  - Boot-shaped heart
  - Apex upturned and formed by the right ventricle
  - Pulmonary oligemia
- Cyanotic spell
  - Increased R → L shunt due to:
    → Pulmonary infundibular spasm → increased right-sided pressure
    → Systemic vasodilation → decreased left-sided pressure
  - Clinical features
    → Severe hypoxia
    → Hypercarbia
    → Acidosis
    → Murmur decreased or absent
  - Respiratory centre in the brain is stimulated → Hyperpnea → R → L shunt
  - Management
    → Oxygen
    → Inj. Sodium Bicarbonate
    → Inj.Morphine → depress respiratory centre

→ Increasing systemic vascular resistance
- – α agonist → Phenylephrine
- – β blockers → Propanolol
- – Ketamine

→ Squatting position
→ Knee chest position
→ Shunt surgeries

NEET PG 2021
FMGE 2024, 2025
INICET 2020, 2021, 2022, 2024, 2025

Name of the shunt surgery	Pulmonary artery is connected to
Blalock-Taussig surgery	Subclavian artery
Waterson's shunt	Ascending aorta
Pott's shunt	Descending aorta

ACUTE RHEUMATIC FEVER

00:41:13
NEET PG 2021
FMGE 2024

- High-risk countries → India
  - Acute Rheumatic fever Incidence → ≥2/1 lakh school-going children
  - Rheumatic heart disease Prevalence → ≥1 / 1000 population

Major criteria	Minor criteria	Essential criteria

• Clinical/subclinical Carditis
• Polyarthritis / Monoarthritis/ Polyarthalgia
• Chorea
• Erythema marginatum
• Subcutaneous nodule (extensor surfaces)
	• Fever
	• Monoarthalgia
	• Elevated CRP / ESR
	• Prolonged PR interval on ECG
		• Any evidence of streptococcal infection
		  ○ ASO titre
		  ○ Throat swab
		  ○ Rapid antigen detection test or culture

- For initial diagnosis
  - 2 Major or 1 Major + 2Minor + essential criteria
- For the diagnosis of recurrence
  - 2 Major or 1 Major + 2Minor + essential criteria or 3 Minor
- Management
  - Inj.Benzathine Penicillin
  - Aspirin: High dose → Low dose
  - Steroids → Moderate to severe carditis
  - Diuretics, Bed rest → CCF
- Secondary prophylaxis → Inj.Benzathine Penicillin every 3 to 4 weeks IM
  - <27kg → 6lakh IU
  - >27kg → 12lakh IU (OR) 1.2 million IU
  - Allergic to penicillin → Azithromycin or Sulfadiazine
  - Duration → Whichever is later

Yourwish

Clinical presentation	For next ___ yrs	Till the age of (AHA)	Till the age of (WHO)
Without carditis	5	21	18
With carditis but no residual heart disease	10	21	25
With residual heart disease	10	40	40

Important Information

- Tricuspid atresia → Cyanotic Congenital heart disease with left axis deviation

MATERNAL DRUG AND ITS EFFECT ON FETUS

00:48:24

Maternal drug	Defect in child
Lithium	Ebstein anomaly
ACE inhibitors	Renal failure, oligohydramnios, pulmonary hypoplasia
Carbamazepine	Neural tube defect
Carbimazole	Cutis aplasia, Omphalocele, choanal atresia
Warfarin	Nasal hypoplasia, Epiphyseal stripping, limb defects
Phenytoin	Cleft lip/palate, dysmorphism, microcephaly, growth restriction
Thalidomide	Phacomelia

PYQ

00:50:22

Q. What is the earliest time of surgery in cleft lip?

a. 3 months
b. 6 months
c. 1 year
d. 18 months

Ans: a

Time for surgery

- Cleft clip → 3 to 6 months of age
- Cleft palate → 9 to 12 months of age

11. SYSTEMIC PEDIATRICS PART 2

PEDIATRIC NEPHROLOGY

00.00.21

Nephrotic Syndrome

00.00.30

NEET PG 2021

- Massive proteinuria
  - Early morning single spot urine sample
  - Urine protein: Urine creatinine > 2
  - Urine protein ≥3+ → Dipstick method
- Hypoalbuminemia
- Generalized edema
- Hyperlipidemia
- M/c cause of Nephrotic syndrome in children: Minimal change disease
  - Light microscopy → no changes detected
  - Electron microscopy → effacement of the podocytes

Drug of Choice For Nephrotic Syndrome

NEET PG 2025
NICET 2024

- 1st episode → Oral prednisolone

2mg/kg/day for 6weeks (Maximum: 60mg/day)

↓

1.5mg/kg alternate days for 6weeks

  - High protein diet
  - Salt-restricted diet
  - Preventing infections → administering Pneumococcal polysaccharide and Influenza vaccine
- Relapse → Urine protein: Urine creatinine ≥2 for 3 consecutive days in a child who is a K/C/O nephrotic syndrome, who was in remission
  - Oral prednisolone

2mg/kg/day daily till remission

↓

1.5mg/kg alternate days for 4weeks

Terminology	Definition	Management
Steroid-dependent nephrotic syndrome	• Relapse happens on alternate days of steroids or • Within 15days of stopping steroids	• Low-dose alternate-day steroid for 9-18 months ○ S/E of steroid - Prone to infection - Cushing syndrome - Obesity - Cataract - Hyperglycemia - Hypertension - Osteoporosis - Psychotic changes

		• Oral levamisole • Oral cyclophosphamide • Mycophenolate Mofetil
Frequently relapsing nephrotic syndrome	• 2 or more relapses in first 6months or • 4 or more relapses in 1years	

Steroid-resistant nephrotic syndrome	• Absence of remission despite 4weeks of daily steroids (2mg/kg/day)	• Calcineurin inhibitors
		  ○ Cyclosporine
		  ○ Tacrolimus
		• Rituximab (anti-CD20)

Q. A 12-year-old child presents with generalized edema. Urinalysis shows 3+ proteinuria, and 24-hr urine protein is more than 3.5g/day. There is no hypertension or hematuria. Serum albumin is low. Kidney biopsy shows segmental sclerosis and hyalinosis. The child shows no response to steroids. What is the most likely diagnosis?

A. Minimal change disease

B. Focal segmental glomerulosclerosis

C. Membranous glomerulopathy

D. Iga nephropathy

Answer: B

- The child has steroid-resistant nephrotic syndrome

Indications Of A Kidney Biopsy In Nephrotic Syndrome

- Age of onset <1year or >10year
- Hypertension
- Hematuria
- Azotemia → Deranged urea and creatinine levels
- Steroid-resistant nephrotic syndrome
- Before starting calcineurin inhibitor → because calcineurin inhibitors are nephrotoxic
  - The status of kidney should be checked before starting the calcineurin inhibitor and after 2 years

Nephritic Syndrome

00.16.04

- M/c cause in children → Post-streptococcal glomerulonephritis
- Age group → 5-12 years
- Clinical features
  - Oliguria
  - Hypertension
  - Edema (less than nephrotic syndrome)
  - Cola coloured urine
  - H/O preceding streptococcal sore throat or skin infection
- Investigations
  - Nephritic range proteinuria
  - Urine protein: Urine creatinine 0.2 - 2

→ Urine protein 1+ or 2+ → Dipstick method
- ASO titres elevated
- Anti-DNAse B+ in the presence of skin infection
- C3 level low
  → Gets normalized by 6 - 8weeks of onset of infection
  → If not normalized → Kidney biopsy
- Hematuria, RBC casts, Granular casts, Active sediments

Important Information
- Hematuria begins
  - 2-3 weeks after URI → PSGN
  - 2-3 days after URI → IgA nephropathy

- Treatment
  - Supportive treatment
    → Low salt diet
    → Antihypertensive
  - Monitor kidney function

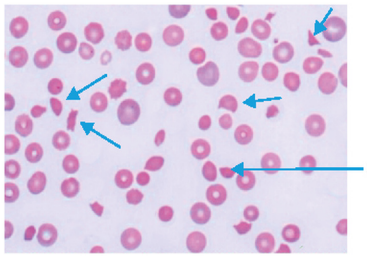
Q.A 3-year-old child with diarrhea presents with a history of epistaxis and not passing urine for the last 12 hours. The child is irritable, and there are petechial spots all over the body. His serum urea is 240mg/dl, and creatinine is 2.8mg/dl. His Hb is 5g/dl, TLC is 10,800cells/mm^3, and platelet count is 36,000cells mm^3. The peripheral smear shows the following picture. What is the probable diagnosis?

Answer: HUS

Hemolytic Uremic Syndrome

00.21.57

- Triad
  - Microangiopathic hemolytic anemia
  - Thrombocytopenia
  - AKI
- Classification
  - Classical HUS: Follows diarrhoea/dysentery → called D+HUS
    → E.coli → produce shiga toxin
    → Shigella
    → Pneumococcal
  - Atypical HUS
    → Defect in alternate complement pathway
- Peripheral blood smear
  - Schistocyte
- Management
  - Supportive treatment
    → Renal replacement therapy → Dialysis (Hemodialysis/Peritoneal dialysis)
  - Atypical HUS
    → Plasma exchangers



→ Immunomodulators → Steroids, azathioprine
→ New drugs α Eculizumab - targets terminal portion of the complement

Q.A 2year old child presented with a history of diarrhea, now comes with anemia, thrombocytopenia, and hematuria. What is the most common organism likely causing this?
A. E.coli O157: H7
B. Salmonella
C. Shigella
D. Yersinia

Answer: A

Kdigo Staging For Acute Kidney Injury (Aki) In Children

FMGE 2020

Stage	Serum Creatinine	Urine Output (Normal: > 1ml/kg/hr)
Stage 1	• Rise in serum creatinine by >50% within 7days • Rise in serum creatine >0.3mg/dL within 48hrs	0.5 - 1 ml/kg/hr
Stage 2	• Rise in serum creatinine by >100%	0.3 - 0.5 ml/kg/hr
Stage 3	• Rise in serum creatinine by >200% • Serum creatinine >4mg/dL	<0.3 ml/kg/hr or Anuria for >12hrs

- Treatment
  - Renal replacement therapy → Dialysis (Hemodialysis / Peritoneal dialysis)
  - Sick child with shock → Continuous renal replacement
- Schwartz formula for calculating estimated Glomerular Filtration Rate
  - Used in AKI and CKD
  - Formula

$$eGFR = \frac{0.413 \times \text{Height (cm)}}{\text{Serum creatinine (mg/dL)}}$$

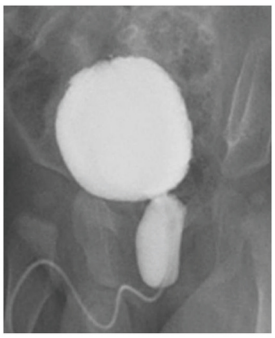
  - Unit → ml/min/1.73 m^2
    → E.g Ht = 100cm , S.creatinine = 1mg/dl

$$eGFR = \frac{0.4 \times 100}{1} = 40 \text{ ml/min/1.73 } m^2$$

  - Normal GFR in children >90

Q.A 5year old child with chronic kidney disease presents with bow legs. Laboratory investigations reveal serum calcium: 8.9mg/dL, serum phosphat: 6.9mg/dL, alkaline phosphatase: elevated, 25(OH) Vitamin D: low. What is the most appropriate next step in management?
A. Calcium supplementation + Phosphate binder
B. Hemodialysis
C. Oral calcium + Vitamin D3
D. Growth hormone therapy
Answer: A

Q.A 6month old male infant is brought with a history of recurrent urinary tract infection and straining while passing urine. MCU image is shown alongside. What is the probable diagnosis?



Posterior urethral valve

- M/c cause of lower urinary tract obstruction in males
- MCU → Distended bladder & dilated posterior urethra
- USG abdomen and pelvis → keyhole sign
- Leads to bilateral hydronephrosis & deranged kidney function
- Management → relieve the obstruction
  - Endoscopic fulguration of the posterior urethral valve
  - Open surgery

Q. Regarding urinary tract infection in children, all of the following are correct, except?

A. Most common cause is Streptococcus pneumoniae

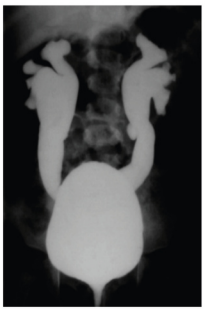
B. Bladder and Bowel dysfunction increases the risk of UTI

C. All children with recurrent UTI should get MCU

D. Ultrasound abdomen must be done in all children with urinary tract infection

Answer: A

Q. A child with a History of recurrent UTI presented to OPD.X ray finding is given below. What is the probable diagnosis?



A. Vesico-ureteric reflux

B. Bladder diverticulum

C. Ectopic kidney

D. Vesico-coli fistula

Answer: A

- Indications for antibiotic prophylaxis in VUR
  - Children < 1 year with febrile UTI
  - Recurrent UTI
  - Associated with bowel and bladder dysfunction
  - Severe VUR → Grade 3, 4 and 5
- Surgery → Ureteric Re-implantation

Potter's Syndrome 00:36:30

- Cause: B/L autosomal recessive PCKD or severe renal congenital malfunction

B/L renal agenesis or dysfunction

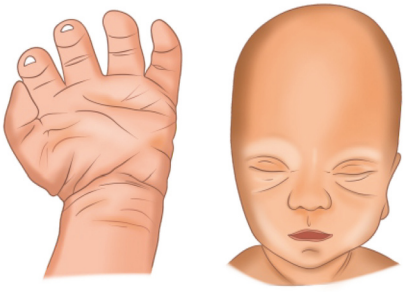
↓

Fetal urine decreased

↓

Oligohydramnios

↓



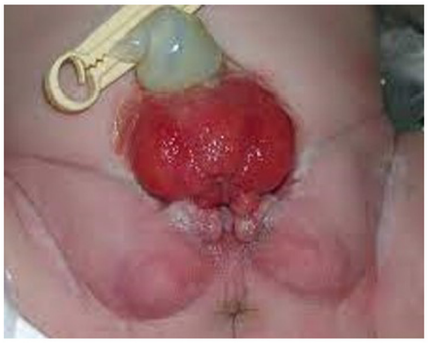
Pulmonary hypoplasia

↓

Death

FACIAL DYSMORPHISM

- Flat facies
- Depressed nasal bridges
- Washerman's hand



Q. Which is the neonatal anomaly shown in the image?

A.Omphalocele

B.Gastroschisis

C.Patent Vitello intestinal duct

D.Exstrophy bladder

Answer: D

- Defect in the anterior abdominal wall
- The anterior wall of the bladder is missing
- The mucosa of the posterior bladder wall is seen

Renal Tubular Acidosis

00.39.22

- Non - anion gap metabolic acidosis
  - Another cause of Non - anion gap metabolic acidosis → Diarrhoea
- Arise from renal tubular impairment
- Normal GFR
  - Serum urea and creatinine are normal
- Types

Features	Type 1 (Distal RTA)	Type 2 (Proximal RTA)	Type 4 (Hyperkalemic)
Basic defect	↓ H^+ secretion in the distal tubule	↓ $HCO3^-$ reabsorption in proximal tubule	↓ Aldosterone effect in cortical collecting duct
Serum K^+	↓	↓	↑

Associations	• SLE
	• Drugs like Amphotericin B
		• Fanconi syndrome
		  ○ Global proximal tubule dysfunction
		  ○ Defect in absorption of albumin, amino acids, phosphate, and bicarbonates
		• Wilson disease
			• Drugs like ACE inhibitors
			• CAH: 21-hydroxylase deficiency

Renal stones (nephrolithiasis, nephrocalcinosis)	Common	Rare	No
Bone disease	Rickets	Rickets (Fanconi)	No

Q. All are true about Renal Tubular Acidosis in children except?
A. Rickets and phosphaturia are seen in Fanconi syndrome
B. Hypokalemia is seen in type 4 RTA
C. RTA -2 has abnormal bicarbonate excretion
D. Type 4 is aldosterone deficiency

Answer: B

PAEDIATRIC NEUROLOGY

00.44.33

Neural Tube Defects

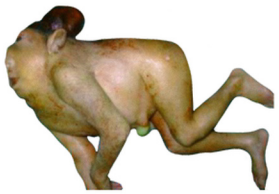
00.44.37

- M/c congenital abnormality of the neurologic system in children
- Multifactorial
- Antenatal markers
  - Maternal serum Alpha-fetoprotein (AFP): Most sensitive test
  - Amniotic fluid acetylcholinesterase level: Most specific confirmatory test
  - Best screening time → 16-18weeks
  - Antenatal USG

Iniencephaly

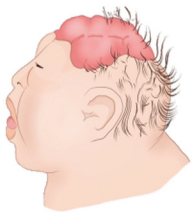
FMGE 2020

- Anencephaly + Defect of the upper part of the cervical spine
- Head is hyperextended
- Absent neck
- Incompatible with life



Anencephaly

- Occur due to defective closure of the anterior neuropore
- Anterior neuropore closes by Day 25 of life
- Scalp and cranial bones are absent
- The brain is exposed to outside

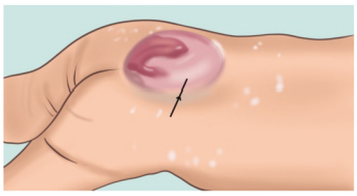


Craniorachischis

- Entire spinal cord is exposed to the outside

Q. Anencephaly occurs due to inability of the neural tube to close in intrauterine life at?
A. 3rd week
B. 4th week
C. 5th week
D. 2nd week
Answer: B

Myelomeningocele



- Swelling in the lower part of the back in a child
- Skin absent
- A sac of meninges with a neural component is present
- M/c in the Lumbosacral region
- Associated with Arnold Chiari malformation type 2
- Treatment → surgery
- In case of ruptured sac → surgery with culture of CSF
- Prophylaxis of NTD NEET PG 2023
  - Dose of folic acid
    → For all women of childbearing age → 400mcg/0.4mg of folic acid per day
    → For high-risk women → H/O previous child being born with NTD, women on antiepileptics → 4000mcg/4mg of folic acid per day
  - To be started at least 1 month before conception

Hydrocephalus

00.50.08

- Enlargement of the ventricles inside the brain
- Due to
  - Increased production
  - Impaired drainage of CSF
- Etiology

Etiology of hydrocephalus

Communicating / Non - obstructive
(All ventricles are uniformly enlarged)

- Choroid plexus papilloma
  - Excess production of CSF
- Meningeal malignancy / metastasis
  - Decreased draining through arachnoid villi
- Post hemorrhagic -Post SAH
- Achondroplasia

Non - Communicating / obstructive
(Some ventricles are enlarged and some are not enlarged)

- ICSOL (Tumor, mass, hematoma, abscess)
- Arnold chiari malformation
  - Herniation of cerebellar tonsil in foramen magnum
- Aqueductal stenosis
  - Lateral and third ventricle Enlarged
  - Fourth ventricle remains normal
- Dandy walker malformation
  - Fourth ventricle dilated
  - Cerebellar hypoplasia
- Vein of galen malformation
- Infection
  - Mumps, TB, Neurocysticercosis

Q. A 7 year old male baby presents with tense bulging fontanelle, bounding pulse, and features S/O heart failure. On auscultation over the anterior fontanelle, a cranial bruit is heard. What is the probable diagnosis?

Vein of Galen Malformation

- Median prosencephalic vein of Markowsky is affected
  - Precursor of the vein of Galen
  - This malformation occurs around 11 - 13 weeks
- Cerebral angiography → dilated vein seen posteriorly
- Management of hydrocephalus
  - Medical management → decrease the intracranial pressure
    → Acute → IV Mannitol, 3% Hypertonic saline
    → Chronic → Oral glycerol, Oral Acetazolamide
  - Surgical management
    → V-P shunt/Ventriculoperitoneal shunt
    → Treat the underlying cause

Q. A child with hydrocephalus underwent V-P shunt. He developed fever and irritability. A dose of paracetamol was given. What should be the next step?

A. Blood culture with CSF culture via LP

B. Blood culture with CSF culture via shunt

C. Radionuclide scan

D. Imaging

Answer: B

- Shunt infection → M/c cause is Coagulase negative staphylococcus aureus (CONS)

Febrile Seizures

00.57.03

FMGE 2022, INICET 2022

- M/c cause of seizure in under 5 children
- Seizure in a child between 6 months - 5 years of age with significant fever (Temp ≥100.4°F) without any evidence of CNS infection
- Types of febrile seizures

NEET PG 2025

Simple febrile seizure	Complex febrile seizure
• GTCS • <15min • Single episodes during a fever episode • No long-term antiepileptics required	• Focal • ≥15min • Recurrent episode • Start intermittent Clobazam prophylaxis ○ Start Clobazam during first three days of the next fever cycle

- Management of febrile seizure:
  - <5min
    → Left lateral position / Recovery position
    → NPO
  - >5min

→ Home → Rectal Diazepam or Rectal / Nasal Midazolam
→ Hospital → Status epilepticus guidelines

- Factors increasing recurrence risk in a child with febrile seizures INICET 2021
  - Age < 18months
  - Duration of fever <1hour
  - Temperature <39°F
  - Family H/O febrile seizure
  - Complex febrile seizure
  - Low serum Na^+ at presentation
- Risk factor for epilepsy in febrile seizures

Risk factor	Risk of epilepsy
Simple febrile seizure	1%
Complex febrile seizure	6%
Fever for < 1hr before seizure	11%
Family H/O epilepsy	18%
Focal seizures	29%
Preexisting neurodevelopmental abnormalities	33%

Q. Which of the following is not true about febrile seizure?
A. 54% recurrence
B. 6months - 5years
C. Simple febrile seizure last <15minutes & 1episode in 24hour
D. No need for long-term anti-epileptics in simple febrile seizures

Answer: B

Q. A 6year old child is brought to the emergency room with a generalized seizure following a high-grade fever. What is the first-line drug of choice for seizure control in his acute febrile setting?
A. Diazepam
B. Valproate
C. Fosphenytoin
D. Ethosuximide

Answer: A

- Other short-acting drugs
  - Lorazepam
  - Midazolam

Status Epilepticus 01.03.43

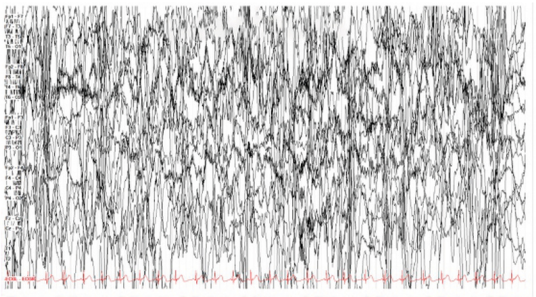
- Seizures lasting for >5min
- Any child presenting with ongoing seizures to a medical facility
- Multiple episodes of seizures without gaining consciousness
- Management:

Airway , Breathing , Circulation
↓
IV / IO access
↓
IV Lorazepam , Midazolam , Diazepam
↓
Rule out Hypoglycemia , Hypocalcemia
↓
IV phenytoin or Fosphenytoin (20 mg/kg)
↓
IV Valproate / Levetiracetam
↓
IV Phenobarbitone

Q.A 9month old infant presents with H/O salaam attacks daily, more frequent during morning hours. On taking a detailed history, the baby is also found to have developmental delay. His EEG picture is provided in the image. What is the probable diagnosis?

West syndrome

- Infantile spasm
- Global developmental delay
- Hypsarrhythmia on EEG
- Treatment
  - Inj.ACTH → DOC
  - Corticosteroids



Q.A premorbidly developmentally normal, unimmunized child presents with gradually deteriorating performance in school and frequent falls. There is a past H/O measles during childhood. O/E the child has myoclonic jerks and EEG shows burst suppression. Diagnosis?

Answer: SSPE - Subacute Sclerosing Panencephalitis

- Develops after 7 to 10years of measles infection

Type of seizure	Drug of choice
Absence seizure • Vacant stare, blank-looking face, daydreaming • EEG → 3Hz spike and wave	• India → Valproate • West → Ethosuximide
Juvenile myoclonic epilepsy	• Valproate
West syndrome	• Inj.ACTH
Infantile Spasm in Tuberous Sclerosis	• Vigabatrin
Status epilepticus	• IV Lorazepam → phenytoin

Q.A 7month old boy presents with H/O fever for 3days along with irritability, poor feeding, and 2 episodes of seizures. On examination, the baby has a tense, bulging fontanelle. What is the probable diagnosis?

Answer: Acute Bacterial Meningitis

Acute Bacterial Meningitis

01.09.15

- Etiology
  - Children
    - → Streptococcus pneumoniae - M/c
    - → Neisseria meningitidis
    - → Haemophilus influenzae
  - Neonates
    - → Group B streptococcus
    - → Acinetobacter
    - → Klebsiella
    - → Listeria
- Clinical features
  - Young children → non-specific
  - Older children
    - → Headache
    - → Irritability
    - → Seizures
    - → Photophobia
- Examination findings
  - Neck rigidity
  - Meningeal sign
    - → Kernig sign → Extension of knee when hip and knee are flexed causes pain
    - → Brudzinsky's sign → Flexion of neck causes involuntary hip flexion
  - Papilledema
- Treatment
  - IV broad-spectrum antibiotics
  - Guided therapy based on CSF study

Guillain-Barre Syndrome

01.11.47

- Prsents with quadriplegia
- Diagnostic criteria in children:

Features required for diagnosis	Features that strongly support diagnosis

• Progressive weakness in > 1 limb (usually starts in the leg)
• Areflexia (or decreased tendon reflex) in weak limbs
	• Progression of symptoms over days to 4 weeks
	• Relative symmetry of symptoms
	• Autonomic dysfunction
	• Cranial nerve involvement
	• Mild sensory symptoms or signs
	• Absence of fever at the onset of neurological symptoms
	• NCV → Typical Electrophysiologic findings
	• CSF Study → Albumino-cytologic dissociation
	  ○ High concentration of protein in CSF
	  ○ Mononuclear cell counts <50/mm^3

Q. Which of the following is not included in the criteria for diagnosing Guillain-Barré Syndrome (GBS) in children?
A. Areflexia
B. Albumino-cytologic dissociation in CSF
C. IVIG or Plasmapheresis in refractory cases
D. Weakness

Answer: C

Q. Tongue fasciculations are a feature of which among the following?
A. Myasthenia gravis
B. Spinal muscular atrophy
C. Guillain-Barré Syndrome
D. Duchenne's muscular dystrophy

Answer: B

Spinal Muscular Atrophy

01.14.28

- Autosomal recessive
- Pathology:

SMN 1 gene on Chr 5q13
↓
Homozygous deletion in survival motor neuron
↓
Loss of motor neurons in the spinal cord

- Clinical presentation
  - Generalized hypotonia → Floppy child (Frog-like posture)
  - Areflexia
  - Delayed motor milestone
  - Recurrent LRTI
- Types
  - Type I - fetal presentation, lethal
  - Type II - Infant presentation, child cannot sit
  - Type III - children presentation, child cannot stand
- Management
  - Nusinersen (Spinraza) → Intrathecal
  - Onasemnogene abeparvovec (Zolgensma) → IV
    → Gene therapy
  - Risdiplam (Evrysdi) → Oral

Brain Death in Children

01.16.43

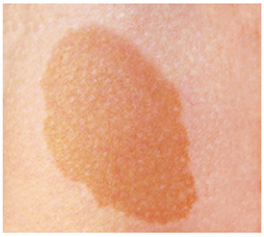
- Irreversible cessation of the function of entire brain, including the brainstem
- Diagnosis
  - Demonstration of irreversible coma
  - Absence of brainstem reflexes

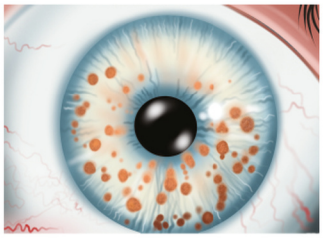
- Apnea
- Finding should remain consistent, at least 2 times, 24 -48 hours apart
- Features incompatible with the diagnosis of brain death
  - Seizures
  - Decerebrate or Decorticate posturing
  - Motor response to painful stimuli

Q.Identify the neurocutaneous syndrome in a family with the father as well as the son having features as shown in the image:

Neurofibromatosis

- Autosomal dominant
- Clinical presentation
  - Café au lait spots
  - Lisch nodules
  - Axillary freckling
  - Neurofibroma
  - Increased risk of tumor



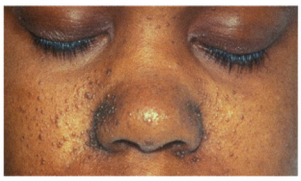


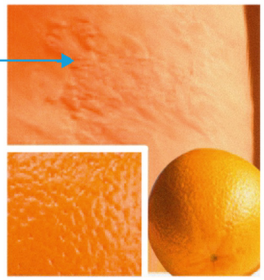
NEET PG 2019

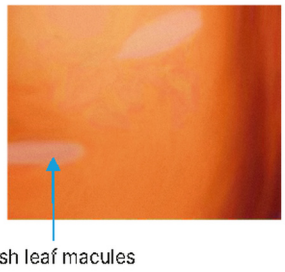
Q.A child with Intellectual disability and seizures has the following features, as shown in the image. What is the diagnosis?

Tuberous sclerosis

- Autosomal dominant
- Clinical presentation
  - Adenoma sebacum
  - Shagreen patch
  - Ash leaf macules







MUSCULOSKELETAL DISORDERS IN CHILDREN

01.19.22

Q. A 4year old boy presents with difficulty in climbing stairs. There is a family history of similar illness in the child's maternal uncle. The child's CPK level is 12,400 U/L. What is the probable diagnosis?

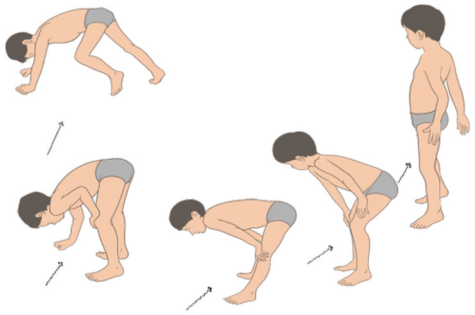
Answer: Duchenne muscular dystrophy

Duchenne Muscular Dystrophy

01.20.14

NEET PG 2020, 2021

- X-linked recessive → Males are commonly affected
- Mutation in the Dystrophin gene
- Clinical presentation
  - Proximal muscle weakness → Difficulty in getting up from the position → Gower sign
  - Exaggerated lumbar lordosis
  - Pseudohypertrophy of calf muscles
  - Valley sign → positive
  - Non ambulatory
  - Recurrent lower respiratory tract infection



- Becker's Muscular Dystrophy
  → A variant of Duchenne muscular dystrophy
  → Present in later age group
- Diagnosis
  - CPK >10,000 U/L
  - Muscle biopsy → Muscle degeneration and fatty deposition
  - Deficient Dystrophin gene staining
  - Genetic studies → MLPA (Multiplex ligation dependent probe amplification)
- Management
  - Supportive → Physiotherapy
  - Alternate days steroids → slow down disease progression
  - Newer drugs
    → Exon skipping antisense oligonucleotide
    - Exondys 51 (Eteplirsen)
    - Amondys 45 (Casimersen)
    - Vyondys 53 (Golodirsen)

    → Gene therapy → Elevidys
    → Novel anti-inflammatory: Vamorolone, Givinostat

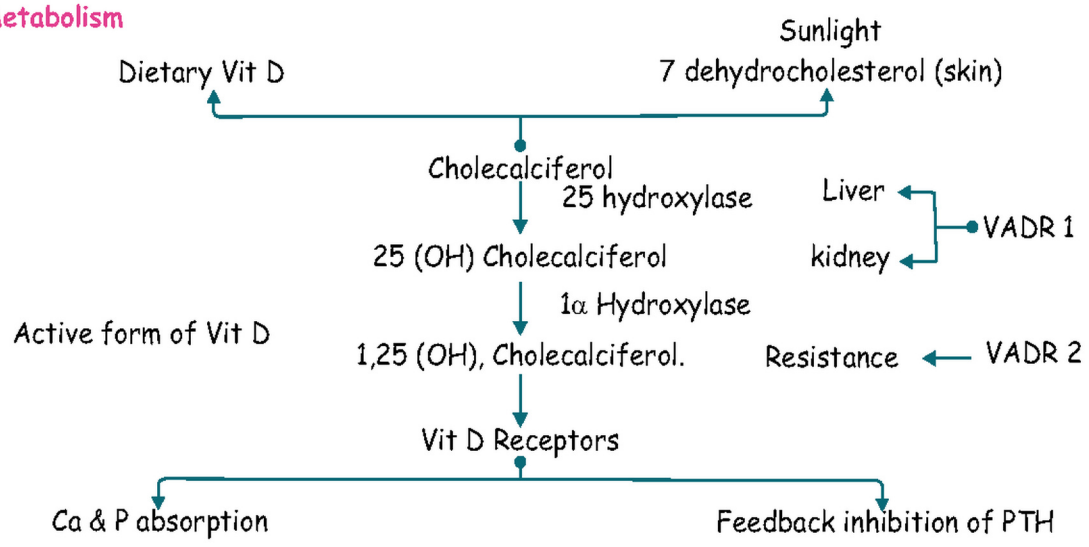
Rickets

01.24.02

FMGE 2020, 2021

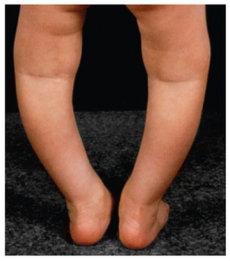
- A disease of growing bone
- Defective mineralization of the bony matrix
- Child with SAM → Changes of rickets not seen
- Etiology
  - Vitamin D deficiency/ Nutritional (M/c cause)
  - Calcium deficiency
  - Phosphate deficiency
  - Renal disease
    → Renal Tubular Acidosis
    → CKD
  - Hypophosphatemic rickets
    → X-linked dominant inheritence
    → PHEX gene is affected

Vitamin D Metabolism



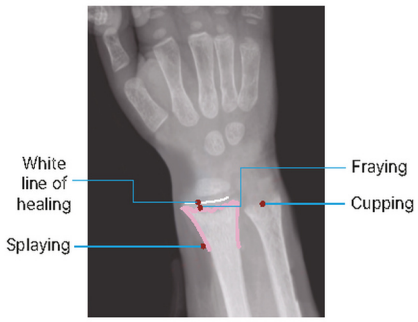
- Vitamin D-Dependent Rickets
  - Type 1
    → Defect in the Liver and Kidney
  - Type 2
    → Resistance at Vitamin D receptors

Clinical features of Rickets	
Chest	• Rachitric rosary • Harrison's sulcus
Limbs	• Genu varum (bow legs) • Genu valgum (knock knees) • Wrist deformity • Wind-swept deformity → Genu varum in one leg and Genu valgum in another leg • Double malleoli
Abdomen	• Pot belly



NEET PG 2023

- Radiological features
  - Density of bone is decreased → Radiolucent
  - Cupping → Concavity
  - Fraying → Irregular border
  - Splaying → widening of the ends of long bone
  - Following treatment with Vitamin D → white line of healing → appears after 6 to 8weeks



S.Ca	S.PO4	ALP	25(OH)Vit.D3	1,25(OH)2 Vit.D3	PTH	Diagnosis
N/↓	N/↓	↑	↓	↓	↑	Nutritional vitamin deficiency
↓	↓	↑	N	↓	↑	VDDR - 1
↓	↓	↑	N	↑↑	↑	VDDR - 2
N	↓	↑	N	N	N/↑	Hypophosphotemic rickets
N/↓	↑	↑	N	N/↓	N/↑	Renal rickets due to CKD

Q. A 6 year old female with joint deformities had received multiple courses of vitamin D with no improvement. Her lab values revealed: Serum calcium -9.5mg/dl, phosphorus -1.6mg/dl, Alkaline phosphatase -814IU/L, with normal serum parathormone and vitamin D3, and normal serum creatinine. What is the probable diagnosis?

A. Vitamin D Dependent Rickets 1
B. Vitamin D Dependent Rickets 2
C. Hypophosphatemic rickets
D. Chronic renal failure

Answer: C

Q. A 1year old exclusively cow milk-fed boy presented with crying with touching of limbs, gum bleeding, and inability to move the right lower limb. Probable diagnosis?

Answer: Scurvy

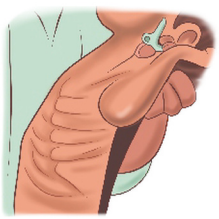
Scurvy

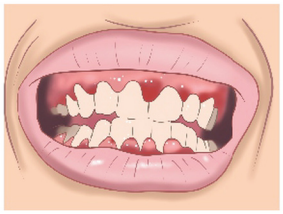
01.33.37

Vitamin C deficiency
↓
Defective collagen synthesis
↓
Blood vessel becomes fragile
↓
Gum bleeding
Subperiosteal haemorrhage in long bone → painful limbs → Pseudoparalysis

NEET PG 2021
INICET 2020

- Clinical features:
  - Scorbutic rosary → enlarged costochondral junction
  - Gum bleeding
  - Painful swelling of the lower limb

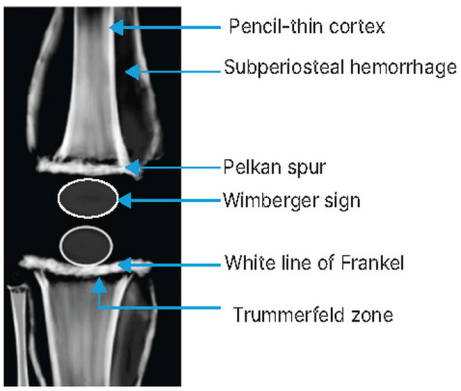




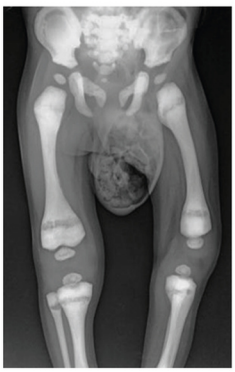
Important Information

- Scorbutic rosary → sharp, angulated, and tender
- Ricketic rosary → Rounded

- Radiological features
  - Subperiosteal hemorrhage
  - Pencil-thin cortex
  - Wimberger sign → ring-shaped epiphysis
  - White line of Frankel
  - Trummerfeld zone
  - Pelkan spur
- Treatment
  - Ascorbic acid therapy



Q. A 11 month old girl presented with pancytopenia and Hepatosplenomegaly. An X-ray of her limb showed the following picture. Diagnosis?



Answer: **Osteopetrosis**

- A/K/A marble bone disease
- Defect in osteoclast → leads to defective resorption of the bone
  - Foramina occluded → neurological features
  - Medullary cavity obscured → pancytopenia
  - Extramedullary hematopoiesis → Hepatosplenomegaly

- Radiological features
  - Increased density of bone
  - Bone within the bone appearance

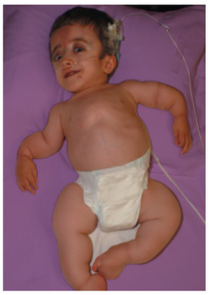
Q.A Baby was born with a limb defect shown in the picture below. There is a H/O Thalidomide ingestion by the mother during her pregnancy. Diagnosis?

Answer: **Phocomelia**

- Proximal limb defect

Q.A 15 month old boy presented with multiple bony deformities and deafness. On enquiring, there was a recurrent H/O limb fracture following trivial trauma in the child. On close examination, the child has blue sclera. What is the probable diagnosis?

Answer: **Osteogenesis Imperfecta**



- A/K/A Brittle bone disease
- Defect of Type I collagen
- Autosomal dominant
- Triad
  - Multiple bony deformities
  - Deafness
  - Blue sclera
- Treatment
  - Bisphosphonates

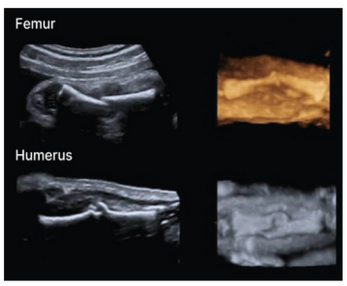
Q.Multiple fracture detected in antenatal ultrasound scan. What could be the diagnosis?

A. Achrondoplasia

B. Osteogenesis imperfecta

C. Marfan syndrome

D. Cretinism



Answer: B

Q.A 5year old girl presented with pain and swelling in the right knee for 3months. There is also a history of pain in the right eye on & off. What is the probable diagnosis?

Answer: JIA (Juvenile Idiopathic Arthritis)

Juvenile Idiopathic Arthritis

01.39.40

- Diagnosis
  - Arthritis of 1 or more joints
  - Lasting for at least 6weeks
  - Child < 16years of age
- Types

- Oligoarticular JIA
  → ≤4 joints are involved
  → Associated with uveitis
  → M/C in girls
- Polyarticular JIA
  → ≥5 joints are involved
- Systemic onset JIA
  → Fever, Hepatosplenomegaly, Rash

- Treatment
  - NSAIDs
  - Weekly Methotrexate and Folic acid therapy
  - Steroids

Q. A 4year girl presented with pain in the left knee and ankle, along with some swelling and restriction of movements. There is a history of fever and blood in stool 4weeks ago. Probable diagnosis?

Answer: Reactive arthritis

Reactive Arthritis

01.41.32

- Joint inflammation due to sterile inflammation reaction following
  - Enteropathic infection
    → Salmonella
    → Shigella
    → Yersinia
    → Campylobacter
  - Urogenital infection
    → Chlamydia
- Clinical presentation
  - Preceding H/O enteric or genitourinary infection 3days to 6weeks before joint symptoms
  - Fever, malaise, fatigue, sterile pyuria, conjunctivitis
  - Asymmetric oligoarthritis
  - Enthesitis
  - HLA B27 positive in most patients

PAEDIATRIC ENDOCRINOLOGY

01.42.31

Q.A 5 year old boy presents with recurrent hypoglycemia, short stature, and micropenis. His bone age is found to be 2.5 years. His serum cortisol was low. What is the probable diagnosis?

- Panhypopituitarism → short child (BA<CA) + hypothyroidism + hypoglycemia + hypogonadism

D/D

- GH deficiency →Short child (BA<CA) + normal genitalia + normal glucose, thyroid, & cortisol
- Craniopharyngioma → child with visual defects + short stature

Yourwish

Isolated GH Deficiency

01.45.09

NEET PG 2021

- Clinical features
  - Short stature with Bone age < chronological age
  - Normal birth weight and length
- Diagnosis → GH stimulation test (Dynamic test) → because GH is pulsatile
  - Done using → Insulin / Clonidine / Arginine
  - Cut off GH level : <10ng/ml → GH deficiency
- Similar clinical features, but level of Growth hormone is high & IGF-1 is low → GH Resistance
- Treatment
  - Recombinant Inj.GH given as Subcutaneous

Q. A 12-year-old child presents with progressive enlargement of hands and feet, coarse facial features, prominent enlarged jaw, and frontal bossing. What is the most definitive test to confirm the diagnosis?

A. Increased IGF-1

B. Increased IGF-1 with oral glucose challenge test

C. Increased GH

D. Increased GH with oral glucose challenge test

Answer: B

Gigantism

01.48.44

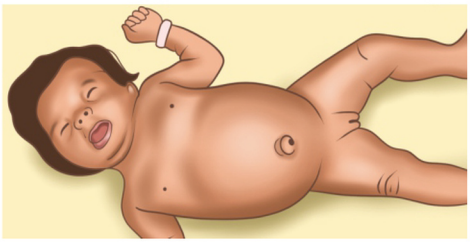
- Excess GH before epiphyseal closure

Excess GH after epiphyseal closure → Acromegaly

- Screening test
  - IGF-1 level → Increased
- Definitive test
  - Oral glucose tolerance test with GH measurement
  - Normally : Oral glucose → suppresses GH to <1ng/ml
  - In Gigantism / Acromegaly → GH fails to suppress

Q.A 4month old male baby presents with puffy looking face, abdominal distention, umbilical hernia, constipation, and prolonged neonatal jaundice. There is also hoarse cry and hypotonia. What is the probable diagnosis?

Answer: Congenital Hypothyroidism



Congenital Hypothyroidism

01.51.11

FMGE 2019

- M/c cause of congenital hypothyroidism → Thyroid Dysgenesis
- M/c cause of congenital hypothyroidism in a child with goitre → Thyroid Dyshormonogenesis
- M/c preventable cause of intellectual disability in children → Hypothyroidism
- Newborn screening for Congenital Hypothyroidism can be done using → TSH
  - TSH >20mIU/L → screening positive

NEET PG 2022, 2025
FMGE 2020, 2024
INICET 2021

→ Cord blood after birth

→ Dried blood spot from heel prick → >48-72hrs after birth → to avoid TSH surge

- The most sensitive is both T4 and TSH

Q. A neonate was found to have TSH >100mIU/L on day 4 of life.USG shows eutopic thyroid gland. What should be the next investigation?

A. Urinary Iodine concentration

B. TSH receptor antibody

C. Thyroid nuclear scan

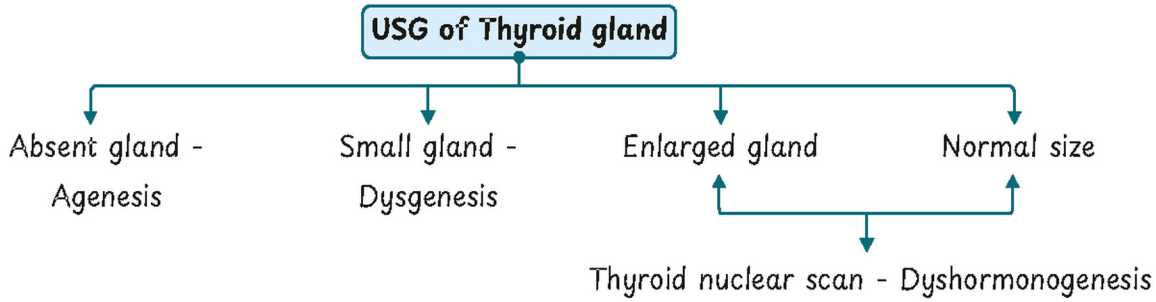
D. Anti-TPO antibody

Answer: C

- Investigations
  - TSH

TSH >20mIU/L
↓
Screening positive
↓
Repeat the test or Repunch
↓
Recall the baby
↓
Repeat on another sample
↓
T3,T4 - low
TSH - High
↓
Congenital hypothyroidism

  - USG -Thyroid
  - I -123 or Tc-99m thyroid scan



T4 - low, TSH - High, thyroid gland is eutopic, and thyroid scan is normal
↓
Possibility of temporary CAH
↓
Reassessment should be done after the age of 3years after stopping treatment for 4weeks and repeating the thyroid function test

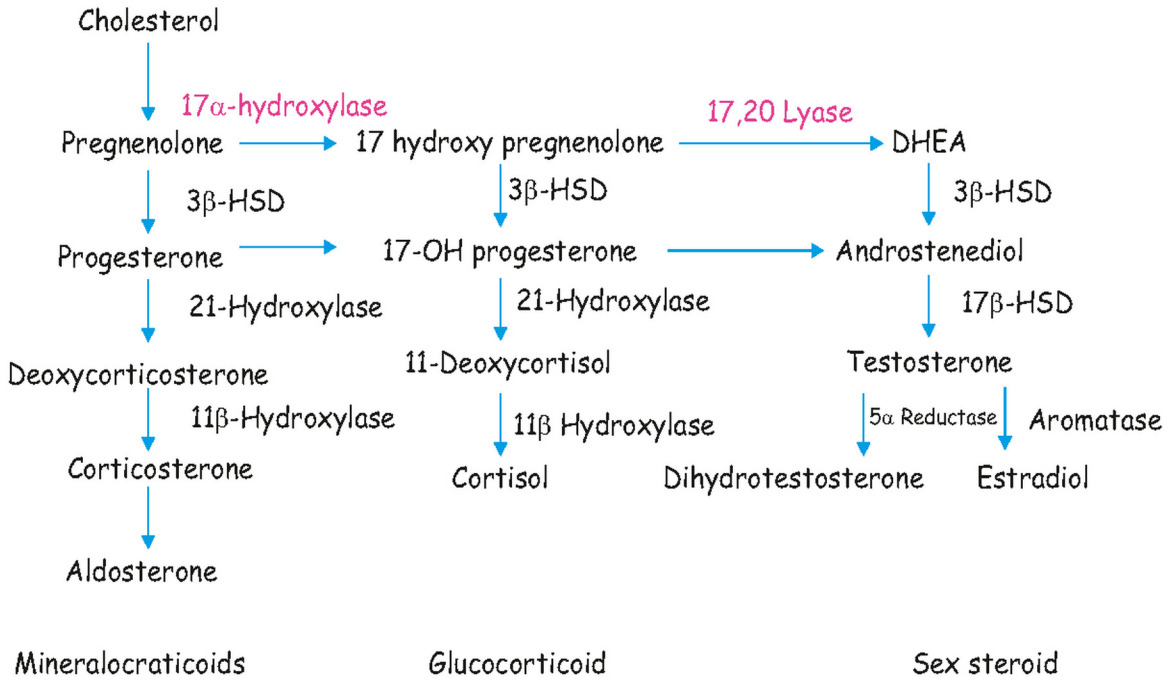
Disorders of Adrenal Gland

01.56.32

- Adrenal cortex of the fetus releases:
  - In early pregnancy → cortisol
  - 2nd trimester → DHEA
- M/c cause of Cushing syndrome in children
  - Iatrogenic / Exogenous steroid
- Clinical features of Cushing syndrome in children:
  - Obesity
  - Buffalo hump
  - Striae
  - Hypertension
  - Hyperglycemia
  - Moon facies
  - Cataract

Q.A 10 day old baby is brought with severe dehydration. O/E the baby has ambiguous genitalia and feeble peripheral pulse. On investigation, there is metabolic acidosis, and serum Na^+ level is 130meq/L and serum K^+level is 7.2 meq/L. Possible diagnosis?

Answer: CAH due to 21 α Hydroxylase deficiency



Congenital Adrenal Hyperplasia

01.58.30

21 α hydroxylase deficiency

- Clinical presentation
  - Low Aldosterone → dehydration and hyperkalemia
  - Low Glucocorticosteroids → shock
  - Increased testosterone
    → Female → virilization → Ambiguous genitalia
    → Normal genitalia in males

- Investigations
  - Hyperkalemia
  - 17 OH progesterone → elevated
  - Genetic analysis to demonstrate gene defect
- Treatment
  - Hydrocortisone
  - Fludrocortisone
  - Pregnant women with H/O previous child with CAH
    → Antenatal Dexamethasone
    → Prenatal diagnosis
  - Extra salt in diet

11 β Hydroxylase Deficiency

- Clinical presentation
  - High deoxycorticosterone → has mineralocorticosteroid action → Hypertension
  - Low Glucocorticosteroids → shock
  - Increased testosterone
    → Female → virilization → Ambiguous genitalia

3 β HSD deficiency

- Clinical presentation
  - Low Aldosterone → dehydration and hyperkalemia
  - Low Glucocorticosteroids → shock
  - Low testosterone
    → Male → under virilized

17 Hydroxylase deficiency

- Clinical presentation
  - High Aldosterone → Hypertension
  - Low Glucocorticosteroids → shock
  - Low testosterone
    → Male → under virilized

	Ambiguous genitalia in female	Under virilization in males
Salt wasting present	21 α Hydroxylase deficiency	3 HSD deficiency
Hypertension present	11 Hydroxylase deficiency	17 Hydroxylase deficiency

Q. A 10 year old boy presents with SMR -5, BP -150/90mmHg with hyper-testosteronism, the enzyme deficient is
A. 11 β Hydroxylase deficiency
B. 21 β Hydroxylase deficiency
C. 3 β Hydroxylase deficiency
D. Cholesterol side chain cleavage enzyme deficiency

Answer: A

Yourwish

Q. A 12 year old child presented with tachypnea and severe dehydration. He had a random blood glucose of 550mg/dl, and urine ketones are 3+. The blood gas showed pH 7.1, Bicarb of 7mmol/L. What is the appropriate management?

A. Stabilize , NS 10ml/kg followed by insulin 0.1U/kg/hr
B. Stabilize , NS 10ml/kg followed by insulin 0.2U/kg/hr
C. Stabilize , NS 20ml/kg followed by insulin 0.1U/kg/hr
D. Stabilize , NS 20ml/kg followed by insulin 0.2U/kg/hr

Answer: C

Diabetic Ketoacidosis

02.05.08
NEET PG 2023

- Hyperglycemia (blood glucose >200mg/dl)
- Venous pH <7.3 or serum bicarbonate <18mmol/L
- Ketonemia (blood β Hydroxybutyrate ≥3mmol/L or moderate or large Ketonuria (≥2+)

Severity of Dka

	pH	Bicarbonate	Dehydration
Severe	≤7.1	≤5mmol/L	Assume 10% dehydration
Moderate	7.1 - 7.2	5 - 10 mmol/L	Assume 7% dehydration
Mild	7.2 - 7.3	10 - 18 mmol/L	Assume 5% dehydration

Treatment of Dka

- IV fluid → NS → in first hour
  - Mild → 10ml/kg
  - Moderate/Severe → 20ml/kg
- Insulin therapy + dehydration correction
  - Start insulin infusion 1 hour after initiation of IV fluid
  - Dose : 0.05 -0.1 U/kg/hr of regular insulin
  - IV insulin bolus should not be used at the start of therapy
  - Infusion tubing should be flushed with the insulin solution before administration