

PREP XI RR

STUDY NOTES

PEDIATRICS

14

VIDEOS

152

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PREMIUM STUDY MATERIAL

PREP XI RR

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1. GROWTH/ DEVELOPMENT/PUBERTY

BIG QUESTION 1: What is growth, and how is it measured?

Broad Answer:

- Growth refers to an increase in size or mass of the body or any of its parts.
- It is measured by:
 - o Weight
 - o Length or height
 - o Upper segment & lower segment ratio

DETAILED QUESTIONS

Q1.1: What is the average weight of an Indian baby?

Answer:

- The average birth weight of an Indian baby is 2.9 kg

Q1.2: At what ages does a child's birth weight double, triple, and quadruple?

Answer:

- Birth weight
 - o Doubles at: 5 months
 - o Triples at: 1 year
 - o Quadruples at: 2 years

Q1.3: What is the average rate of weight gain in children during different age periods?

Answer:

Age	Rate of weight gain
1 st 3 months	30 g/day
3-6 months	20 g/day
6-9 months	15 g/day
9-12 months	12 g/day
1-3 years	8 g/day

Q1.4: What are the formulas used to calculate the expected weight of a child at different ages?

Answer:

Age	Formula to calculate the expected weight of a child
< 1 year	$\frac{x+9}{2}$, where x is the age in months
2-6 years	$2x + 8$, where x is the age in years
7-12 years	$\frac{7x-5}{2}$, where x is the age in years

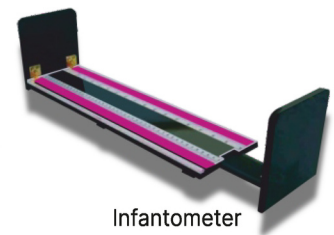
Q1.5: How is length and height measured?**Answer:**

- <2 years: Measured as length using an infantometer
- >2 years: Measured as height using a stadiometer

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Q1.6: What is an infantometer, and how is it used to measure length in children?**Answer:**

- Infantometer is a horizontal board on which the baby is made to lie down, where one person is made to fix the baby on the board with legs extended, and another person can move the plate and read the length

**Q1.7: What precautions should be taken while measuring the height of a child above 2 years?****Answer:**

- Remove footwear
- Remove headgear
- Stand erect so that the eyes are parallel to the Frankfurt plane
- The occiput, back of the shoulder, buttocks, and heel should be touching the vertical wall behind

Q1.8: What is the difference between standing height and recumbent length in children, and why?**Answer:**

- Standing height is approximately 0.7 cm less than recumbent length due to the effect of gravity.

Q1.9: What is the expected length/height of a child at various ages?**Answer:**

Age	Expected length/height of a child
At birth	50 cms
At 1 year	75 cms
At 2 years	90
At 4 - 4.5 years	100 cms

Q1.10: What are the key milestones in length/height increase during childhood?**Answer:**

- Length increases by 50% in the 1st year
- Length doubles (100%) by 4-4.5 years
- Maximum growth occurs in the 1st year of life, followed by puberty

Q1.11: How does a child's length increase during the first 3 years of life?**Answer:**

Age	Increase in length with age
1 st 3 months	3.5 cm/month
3-6 months	2 cm/month
6-9 months	1.5 cm/month

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9-12 months	1.2 cm/month
1-3 years	0.8 - 1 cm/month

Q1.12: What is the formula for calculating the expected height of a child?

Answer:

- Expected height of a child = $6x + 77$, where x is the age in years

Q1.13: What does arm span indicate about a child's height, and how does it vary with age?

Answer:

- Arm span is another parameter that gives an idea about the height of the child
- Arm span is the distance between the middle finger of one arm to another when arms are outstretched (perpendicular to the rest of the body)
 - < 10 years: 1-2 cms less than the height
 - > 10 years: 1-2 cms more than the height
- Normally, the difference between the arm span and the height is 3 cms

Q1.14: At what age is the child's height half of adult height?

Answer:

- Average height of an Indian adult: 160-170 cms
- Age at which the child will be half the adult height (80-85 cms): 20-24 months

Q1.15: What is meant by the Upper Segment and Lower Segment in children?

Answer:

- Upper segment: Part of the body above the symphysis pubis
- Lower segment: Part of the body below the symphysis pubis

Q1.16: What is the normal Upper Segment to Lower Segment (US:LS) ratio at birth, 3 years, and 7-10 years of age?

Answer:

- At birth- 1.7-1.9 : 1
- 3 years- 1.3 : 1
- 7-10 years: 1 : 1

Q1.17: What growth charts are used to interpret the height of a child?

Answer:

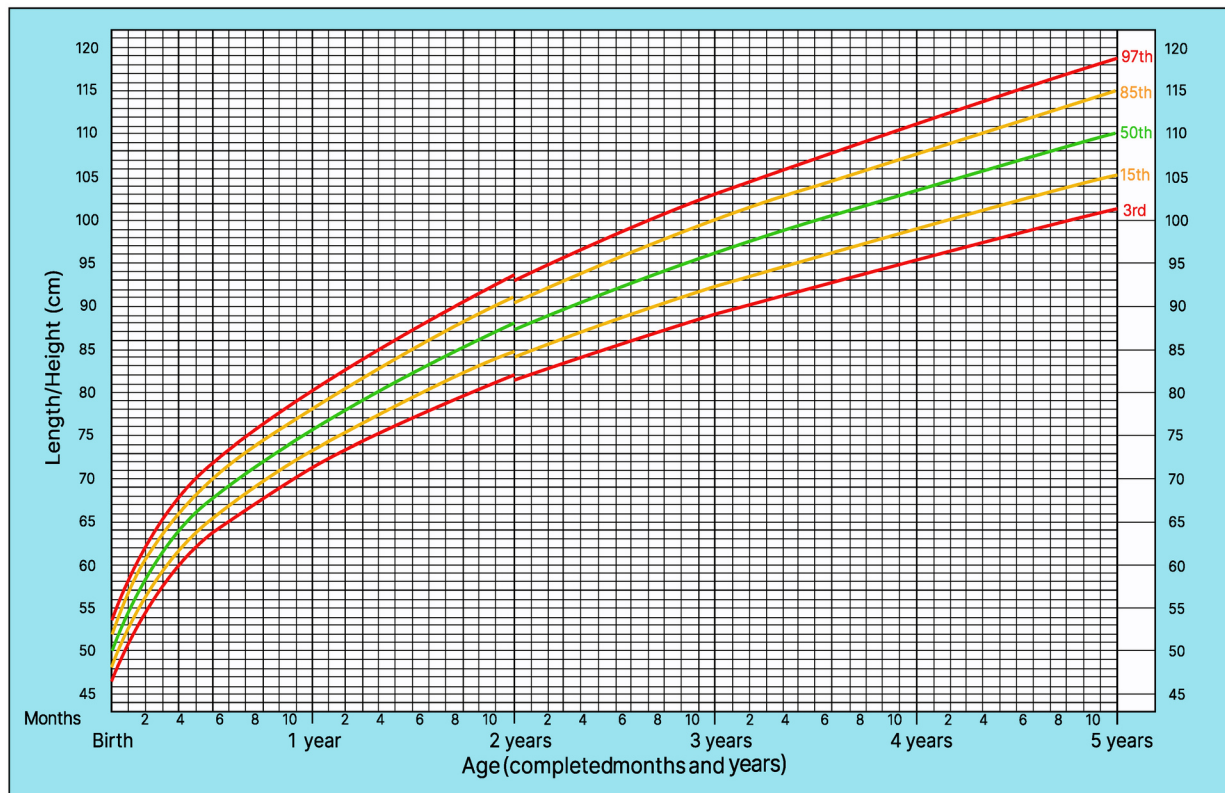
- <5 years: WHO growth charts
- >5 years: IAP growth charts

Q1.18: What does it indicate if a 2-year-old child's height is 70 cm according to the WHO growth chart?

Answer:

- At 2 years of age the height of the child is 70 cms, corresponding co-ordinates are marked in the growth chart, here it falls below the 3rd percentile, which means that the child has short stature

Length/height-for-age BOYS Birth to 5 years (percentiles)



BIG QUESTION 2: What are the WHO Growth charts, and why are they important in pediatrics?

Broad Answer:

- The WHO Growth charts are a crucial set of charts used globally in pediatrics to assess the growth and development of children from birth to 5 years of age. They describe how healthy children should grow under optimal conditions, rather than just how children do grow in a particular population

DETAILED QUESTIONS

Q2.1: What is the basis for the WHO Growth charts?

Answer:

- WHO growth charts are based on the Multicentre Growth Reference Study (MGRS)

Q2.2: Which countries were included in the Multicentre Growth Reference Study (MGRS) used to develop the WHO Growth charts?

Answer:

- Children from 6 countries were studied
 - o Brazil
 - o Oman
 - o Norway
 - o Ghana
 - o USA
 - o India

Q2.3: When were the WHO Growth charts first introduced, and what were the key inclusion and exclusion criteria for the children studied?

Answer:

- WHO growth charts were first used in 2006
- Used exclusively in breastfed children
- Exclusions:
 - o Maternal alcohol
 - o Smoking

BIG QUESTION 3: Define short stature.

Broad Answer:

- Short stature is defined as “the height of a child is $<3^{\text{rd}}$ percentile or <-2 Z score of expected according to the age and sex of the child”

DETAILED QUESTIONS

Q3.1: What are the etiologies of proportionate short stature?

Answer:

Normal variants	• Familial short stature • Constitutional delay in growth and puberty
Intra-uterine causes	• IUGR • Infection by TORCH • Genetic syndromes: Turner's syndrome, Down's syndrome, Seckel syndrome
Postnatal/ acquired causes	• Chronic malnutrition • Any chronic systemic illness • Celiac disease • Maternal deprivation • Psychosocial • Endocrine: Growth hormone deficiency

Q3.2: What is the difference between acute and chronic malnutrition in terms of their effect on a child's growth?

Answer:

- Acute malnutrition affects the weight of the child, while Chronic malnutrition affects the height of the child

Q3.3: 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?

Answer:

- Height of parents normal- rules out familial short stature
- Bone age $<$ chronological age- delay in bone maturation- points towards constitutional delay in growth and puberty (CDGP)

Q3.4: Which X-ray sites are used to assess bone age in infants, children, and older children?

Answer:

- Children: X-ray left hand and wrist
- Infants: X-ray of knees
- Older children: X-ray of shoulders

Q3.5: What are the differences between Constitutional Delay of Growth and Puberty (CDGP) and Familial Short Stature?

Answer:

🔍 PYQ: FMGE 2020

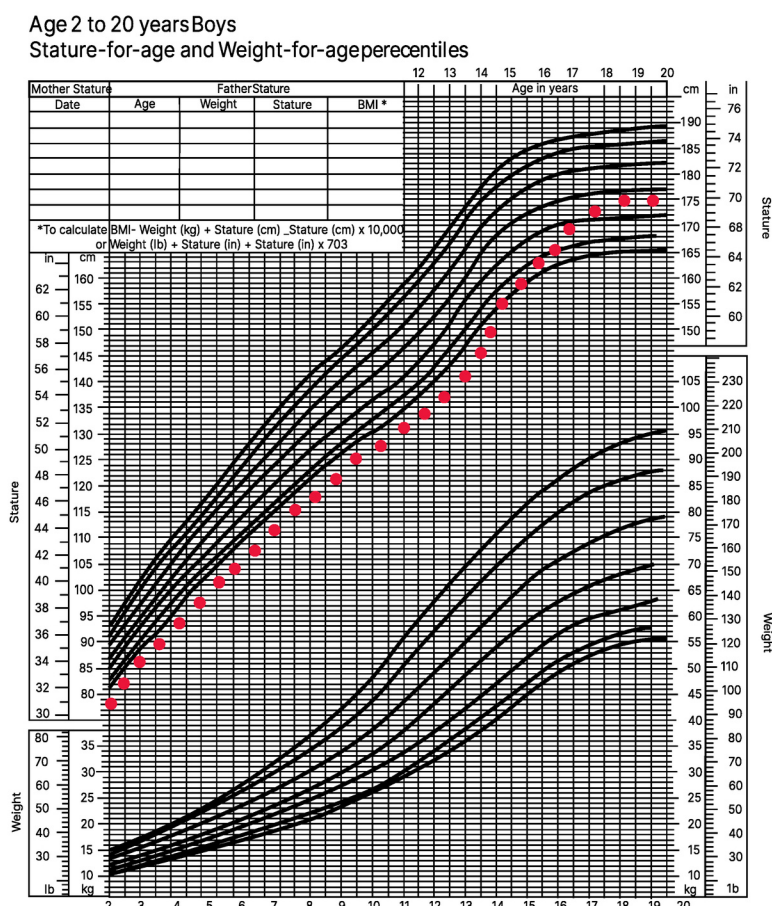
	CDGP	Familial Short Stature
What is it?	Child has short stature during childhood but final height attained is normal	Child has short stature but the height is normal as per his target height (according to the mid parental height)
Puberty	Delayed puberty	Normal
Family history	Family history of delayed puberty	Family history of short stature
Bone age	Bone age < Chronological age	Bone age = Chronological age

Q3.6: How is mid-parental height calculated for boys and girls?

Answer:

- For boys: $\frac{\text{Average height of parents} + 13}{2}$
- For girls: $\frac{\text{Average height of parents} - 13}{2}$

Q3.7: What does the growth pattern shown in the height chart indicate, and in which condition is such a pattern typically seen?



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Answer:

- In the above image, stature/height is plotted
- During childhood, the child's height runs almost parallel to the 3rd percentile line, but ultimately, a catch-up growth happens, and the final height achieved is normal
- Seen in CDGP

Q3.8: What are the key features of short stature in Growth Hormone Deficiency?**Answer:**

- Type of short stature: Proportionate short stature
- US: LS ratio: Normal
- Bone age: < Chronological age

Q3.9: What are the investigations and management options for Growth Hormone Deficiency, and what is a notable side effect of treatment?**Answer:**

- Investigations:
 - Growth hormone stimulation test
 - Insulin, Clonidine can be used
- Management: Recombinant growth hormone Injection
- Side effect: Pseudotumor cerebri

Q3.10: What are the different types and causes of disproportionate short stature, and how is the upper segment to lower segment (US:LS) ratio affected in each?**Answer:**

Short trunk	Short limbs
• Decreased US:LS ratio • Causes: (Mnemonic- Short Man Climbs High) ◦ Spondyloepiphyseal dysplasia ◦ Mucopolysaccharidosis ◦ Curves spine/Potts' disease ◦ Hemivertebrae/Butterfly vertebra	• Increased US:LS ratio • Causes ◦ Rickets ◦ Osteogenesis imperfecta ◦ Congenital hypothyroidism ◦ Achondroplasia

Q3.11: What are the key clinical features of Alagille syndrome?**Answer:**

- Hemivertebrae/ Butterfly vertebrae
- Conjugated hyperbilirubinemia
- Triangular facies
- Pulmonary stenosis
- Eye abnormalities

BIG QUESTION 4: Why is head circumference an important measurement in pediatric practice?**Broad Answer:**

- Head circumference is a vital anthropometric measurement in pediatrics, particularly during infancy and early childhood. It serves as an important indicator of brain growth and development

DETAILED QUESTIONS

Q4.1: What is the normal head circumference at birth?

Answer:

- Normal head circumference at birth is 33-35 cm

Q4.2: What is the normal rate of increase in head circumference during infancy and early childhood?

Answer:

Age	Normal rate of increase in HC
1 st 3 months	2 cm/month
3 months	1 cm/month
6 months	0.5 cm/month
2 years	0.2 cm/month

Q4.3: When is an increase in head circumference considered abnormal, and what conditions should be suspected?

Answer:

- In any given single month, if the HC increases at a rate >2 cm/month, it is abnormal
- Underlying causes, such as a tumor, hydrocephalus, should be ruled out

Q4.4: How is microcephaly defined?

Answer:

- HC < -3 Z score of expected according to the age and sex is considered Microcephaly

Q4.5: What are the etiologies of Primary microcephaly?

Answer:

- Genetic syndromes (Mnemonic: Cannot See PEFR in a Child)
 - o Cri du chat syndrome(5p-)
 - o Smith-Lemli-Opitz syndrome
 - o Patau syndrome: Trisomy 13
 - o Edward syndrome: Trisomy 18
 - o Familial
 - o Rubinstein-Taybi syndrome-deviated (Z-like)thumb
 - o Cornelia de syndrome- long eyelashes

Q4.6: What are the key features and underlying metabolic defect in Smith-Lemli-Opitz syndrome?

Answer:

- Midline defects: Cleft lip, Cleft palate, holoprosencephaly, gonadal abnormalities
- Metabolic defect: Cholesterol metabolism defects, accumulation of 7-dehydrocholesterol

Q4.7: What are the etiologies of Secondary microcephaly?

Answer:

Maternal	• Alcohol intake • Smoking • Phenytoin • Radiation exposure • Metabolic disorder-Phenylketonuria • TORCH infections
Baby	• Severe malnutrition • Perinatal asphyxia/HIE • Any CNS infection in the first year of life-Meningoencephalitis
Acquired causes	• Rett syndrome • Angelman syndrome • Seckel syndrome

Q4.8: What is the mode of inheritance, the most commonly affected gender, and the gene involved in Rett Syndrome?

Answer:

- Inheritance: X-linked dominant inheritance
- M/c in Females
- M/c gene affected: MECP2 gene

Q4.9: What are the clinical features of Rett syndrome?

Answer:

- HC at birth: Normal
- Gradual developmental delay
- Acquired microcephaly
- Loss of purposeful hand movements
- Stereotypical hand-wringing movements

Q4.10: What is the FDA-approved drug used for treating Rett syndrome?

Answer:

- Trofinetide

Q4.11: How is macrocephaly defined based on head circumference?

Answer:

- HC $> +2$ SD of expected according to the age and sex is considered Macrocephaly

Q4.12: What are the causes of macrocephaly?

 PYQ: FMGE 2021

Answer:

- Increased thickness of cranial bones:
 - o Rickets
 - o Osteogenesis imperfecta
 - o Chronic hemolytic anemia- Thalassemia
- Subdural Effusion or empyema (usually a sequel of meningitis)

Q4.13: What is the difference between megalencephaly, hydranencephaly, and hydrocephalus in terms of brain structure and underlying cause?

Answer:

Megalencephaly	• A condition where the size of the brain increases due to the accumulation of storage/abnormal substances
Hydranencephaly	• A condition where the cerebral hemispheres are absent and are replaced by a fluid-filled sac. Transillumination positive. The midbrain and brain stem are intact
Hydrocephalus	• A condition where there is an enlargement of the ventricles, either due to increased production or impaired drainage of CSF

Q4.14: List the etiologies of Megalencephaly.

Answer:

Mnemonic: **BALWANSiNG**

- Benign familial megalencephaly
- Amino acid disorders: Glutaric aciduria
- Lysosomal storage disorders: Mucopolysaccharidosis
- Weavers syndrome
- Achondroplasia
- Neurodegenerative diseases:
 - Alexander disease- deposition of GFAP, Rosenthal fibers
 - Caravan disease- deficiency of aspartoacylase- leads to deposition of N-aspartoacetic acid (NAA) in the brain
- Soto syndrome
- Neurofibromatosis
- Galactosemia

Q4.15: What are the etiologies of communicating Hydrocephalus?

Answer:

Mnemonic: **CAMP**

- Choroid plexus papilloma
- Achondroplasia
- Meningeal malignancy/ metastasis
- Post hemorrhagic

Q4.16: What are the etiologies of non-communicating Hydrocephalus?

Answer:

Mnemonic: **MAAD HIV**

- Mass lesion
- Arnold Chiari syndrome: Herniation of the cerebral tonsils into the foramen magnum
- Aqueductal stenosis
- Dandy-Walker malformation: Dilatation of the 4th ventricle
- Hematomas
- Infections: mumps, neurocysticercosis, TB
- Vein of Galen malformation

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Q4.17: What is Vein of Galen malformation, when does it develop, and what are its key clinical features?

Answer:

- Vein of Galen malformation is an arteriovenous malformation involving the precursor vein of Galen
- Onset- 11-13 weeks of gestation
- Vein involved: Median prosencephalic vein
- On examination: Cranial bruit + on auscultation

Q4.18: How is Hydrocephalus treated?

Answer:

- VP shunt (Ventriculo-Peritoneal shunt)
- Medical management: Mannitol, 3% saline/ hypertonic saline, acetazolamide, glycerol

BIG QUESTION 5: What is Craniosynostosis, and what are the different types of Craniosynostosis?

Broad Answer:

- Craniosynostosis is an abnormal shape of the head due to the premature fusion of one or more cranial sutures
- Types:
 - o Dolichocephaly
 - o Trigonocephaly
 - o Oxycephaly or Turricephaly

DETAILED QUESTIONS

Q5.1: Which cranial sutures are involved in dolichocephaly, trigonocephaly, and oxycephaly, and what head shapes do they produce?

Answer:

- Colichocephaly:
 - o Anteroposteriorly oblong head
 - o Most common
 - o Premature fusion of the sagittal suture
- Trigonocephaly
 - o Premature fusion of the metopic suture
- Oxycephaly or Turricephaly
 - o Tower-like head due to the fusion of multiple sutures in the base of the skull

Q5.2: What are the syndromes associated with Craniosynostosis?

Answer:

- Crouzon syndrome: Prominent eyes, midfacial hypoplasia, bulging eyes, small mandible
- Pfeiffer syndrome: Clover leaf shape of the skull
- Carpenter syndrome
- Apert syndrome: Syndactyly - Mitten hands and toes

BIG QUESTION 6: What are the different patterns of growth?

Broad Answer:

- Different patterns of growth include:

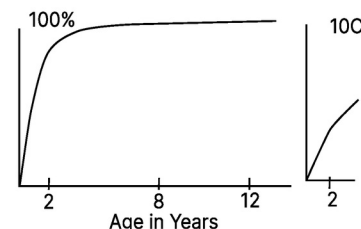
- o Brain growth
- o Lymphoid growth
- o Somatic growth
- o Gonadal growth

DETAILED QUESTIONS

Q6.1: How does brain growth look on a graph?

Answer:

- In the above graph, maximum growth takes place in the first 2 years, after which the growth is stagnant, which signifies brain growth



Q6.2: How does lymphoid growth look on a graph?

Answer:

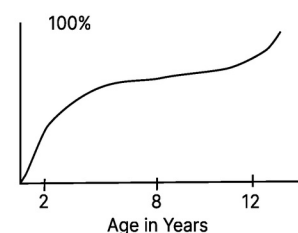
- In the above graph, maximum growth is taking place between 4 to 9 years, which signifies lymphoid growth; there is physiologic lymphoid hyperplasia between 4-9 years



Q6.3: How does somatic growth look on a graph?

Answer:

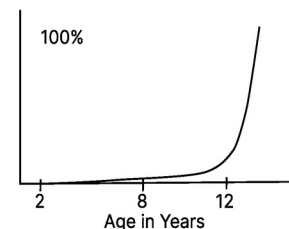
- In the above graph, a sigmoid curve is seen, an accelerated growth phase from birth to 2 years, followed by slowing of growth, after which the rate of growth picks up again, which signifies somatic growth showing two periods of accelerated growth, i.e., infancy and puberty



Q6.4: How does gonadal growth look on a graph?

Answer:

- In the above graph, the rate of growth is minimal in the first 10 years, after which the rate of growth picks up, which signifies gonadal growth



BIG QUESTION 7: What are the two types of dentition in humans?

Broad Answer:

- Primary dentition- Milk teeth
- Secondary dentition- Permanent teeth

DETAILED QUESTIONS

Q7.1: What are the key differences between primary and secondary dentition in terms of timing, sequence, and dental formula?

PYQ: NEET PG 2021

Answer:

	Primary	Secondary
Begins at	6-7 months of life	6 years
1 st tooth	Lower central incisor	1 st molar

Last tooth	2 nd molar	3 rd molar
Completes by	2.5-3 years	12 years except 3 rd molars
Dental formula	I C P M 2 1 0 2 = $5 \times 4 = 20$	I C P M 2 1 2 3 = $8 \times 4 = 32$

Q7.2: Between which ages does mixed dentition occur?

Answer:

- The period of mixed dentition occurs between 6 to 12 years

Q7.3: What is delayed dentition?

Answer:

- When no teeth erupt by 13 months of age, it is called delayed dentition

Q7.4: What are the etiologies of delayed dentition?

Answer:

- Familial
- Rickets
- Idiopathic
- Endocrine: Hypopituitarism/Hypothyroidism/Hypoparathyroidism
- Down's syndrome
- Cleidocranial dysostosis

Q7.5: What are the key clinical features and mode of inheritance of cleidocranial dysostosis?

Answer:

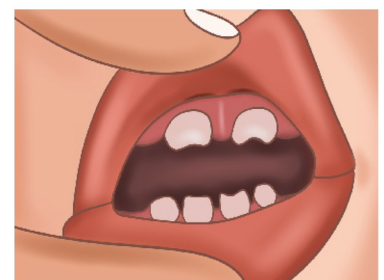
- Complete or partial absence of clavicles
- Autosomal dominant inheritance
- Large anterior fontanelle
- Supernumerary teeth



Q7.6: What is Hutchinson's triad, and which condition is it a late manifestation of?

Answer:

- Hutchinson's triad is a late manifestation of congenital syphilis
- The triad includes:
 - o Hutchinson's teeth (notching of incisors)
 - o SNHL
 - o Interstitial keratitis



Q7.7: What are the other features of Congenital syphilis?

Answer:

- Saddle nose
- Mulberry molars
- Running nose
- Clutton's joints

PYQ: FMGE 2020

BIG QUESTION 8: How is development assessed in a child?

Broad Answer:

- Developmental milestones help in assessing development in a child.
- They include:
 - o Gross Motor Milestones
 - o Fine Motor Milestones
 - o Social Milestones
 - o Language Milestones

DETAILED QUESTIONS

Q8.1: What are the major motor milestones?

 PYQ: INICET 2022, 2023

Answer:

Motor milestone	Age of development
Neck holding	• Partial: 3 months • Complete: 5 months
Rolls over	• 5-6 months
Sitting	• With support (Tripod position): 6 months • Without support: 8 months
Pivotting and cruising	• 10-11 months
Standing	• With support: 9 months • Without support: 12 months
Walking without support	• 13 months
Creeps upstairs	• 13 months
Upstairs and downstairs	• 2 years
Upstairs with alternate feet and downstairs with two feet per step	• 3 years
Alternate feet up and down?	• 4 years

Q8.2: What is the below image suggestive of?

Answer:

- In the image, the child is looking at his hands intently, suggestive of hand regard, which develops at 3 months



Q8.3: What is the below image suggestive of?

Answer:

- In the image, the child is seen lifting his head above the rest of his body, suggestive of having partial neck control, which develops between 3 to 4 months

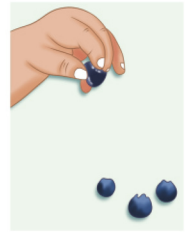


Q8.4: What does the image below indicate?

Answer:

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- The image depicts pincer grasp, where the child is holding small objects using the thumb and the index finger while taking support from other fingers, which develops at 9 months



Q8.5: What does the image below suggest?



Answer:

- In the image, a child is seen riding a tricycle, which develops at 3 years old

Q8.6: At what age does a child reach out for objects?

Answer:

- 4 months

Q8.7: At what age does the child develop bidextrous, unidextrous, and palmar grasp?

Answer:

- Bidextrous grasp: 5 months
- Unidextrous/ Palmar grasp: 6 months

Q8.8: Transfer of objects develop at which age?

Answer:

- Transfer of objects develops at 6 months of age, indicating the child can compare objects

Q8.9: At what age does pincer grasp develop?

Answer:

- Immature pincer grasp: 9 months
- Mature pincer grasp: 12 months

Q8.10: At what age does casting develop?

Answer:

- Casting, which means the child drops everything down, develops at 13 months of age

Q8.11: How does block play (e.g., tower, train, bridge) progress with age in young children, and what does it indicate about their fine motor development?

Answer:

- Tower of Cubes: Number of cubes put on top of each other = Age $\times$ 3
- 2 years: Tower of 6 cubes
- 3 years: Tower of 9-10 cubes
- Train without chimney: 2 years
- Train with chimney: 2.5 years
- Bridge: 3 years

- Gate: 4 years
- Steps: 5-6 years

Q8.12: What are the fine motor milestones developing at 2 years of age?

Answer:

- Copies a straight line
- Turns door knob
- Unscrews a lid

Q8.13: What fine motor milestones develop at 3 years of age?

Answer:

- Copies a circle
- Handedness gets established
- Dress and undress by self - Except buttons

Q8.14: What fine motor milestones develop at 4 years of age?

Answer:

- Copies a rectangle or a cross
- Buttons and unbuttons

Q8.15: What fine motor milestones develop at 5 years of age?

Answer:

- Copies a triangle/ a tilted cross
- Tie shoe laces

Q8.16: Name the social milestones and the age at which they develop.

Answer:

 PYQ: INICET 2022
  PYQ: NEET PG 2022
  PYQ: FMGE 2021

Social milestone	Age of development
Social smile	2 months
Mirror play	6 months
Stager anxiety	7 months
Waving bye-bye	9 months
Plays peek-a-boo	10 months
Kisses/ plays a simple ball game	12 months
Points to objects	15 months
Domestic mimicry and dry during the daytime	18 months
Parallel play	2 years
Joins in play and nighttime continence	3 years

Q8.17: What is the below image suggestive of?

Answer:

- In the image, a child displays mirror play, which is seen at 6 months



Q8.18: What is the below image suggestive of?

Answer:

- The image depicts a child playing peek-a-boo, which develops at 10 months old



Q8.19: What is the below image suggestive of?

Answer:

- In the image, a child kissing his mother, which develops at 12 months



Q8.20: What is the below image suggestive of?

Answer:

- In the image, the child is tying shoe lace, which develops at 5 years of age



Q8.21: What is nocturnal enuresis?

Answer:

- Nocturnal enuresis is involuntary urination at night beyond 5 years of age

Q8.22: What is the 1st line management of nocturnal enuresis?

Answer:

- Lifestyle and behavioral modifications like early dinner, limitation of fluids in the evening
- Positive reinforcement/ motivational therapy/ Star chart

Q8.23: What is the 2nd line management of nocturnal enuresis?

Answer:

- Conditioning: Bell and alarm technique
- A moisture-sensing alarm is placed in the underwear of the child. As soon as the child voids, the alarm rings, the child wakes up, and goes to the washroom, and voids

Q8.24: What is the 3rd line management of nocturnal enuresis?

Answer:

- Pharmacological:
 - o Oral Desmopressin
 - o Imipramine
- Relapse rates are high

Q8.25: Elaborate on what ages important language milestones develop.

Answer:

Language milestone	Age of development
Vocalizes	2 months

Cooing	3 months
Monosyllables	6 months
Bisyllables	9 months
Two to three words with meaning	1 year
Jargon speech	15 months
Vocabulary of 10 words	18 months
Vocabulary of 50-100 words	2 years
Two-word sentences and the use of pronouns	2 years
Three-word sentences and repeats three digits	3 years
Tells a poem/ story/ sings a song	4 years

Q8.26: How is the Developmental Quotient (DQ) calculated?

Answer:

- $DQ = \frac{\text{Development age}}{\text{Chronological age}} \times 100$

Q8.27: If a 6-year-old child can do milestones only of a 3-year-old child and not beyond that, what is the DQ?

Answer:

- $DQ = \frac{\text{Development age}}{\text{Chronological age}} \times 100$

$$= \frac{3}{6} \times 100$$

$$= 50$$

Q8.28: Elaborate on the red flag signs of developmental delay.

Answer:

	Normal	Red flag
Sitting with support	6 months	9 months
Walking with support	12 months	15 months
Social smile	2 months	3 months
Waves bye bye	9 months	12 months
Pincer grasp	9 months	12 months
Meaningful words	1 year	16 months

Q8.29: What are the developmental implications of behaviors like hand regard, reaching for objects, and pretending to drink from a cup in infants and toddlers?

Answer:

- Hand regard: Self-discovery of hands
- Reaches for objects: Visuomotor coordination
- Transfers objects: Comparison of objects
- Points to objects: Interactive communication

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- Uncovers hidden toys: Object permanence
- Pretends to drink from a cup: Symbolic thought

BIG QUESTION 9: What is puberty, and at what age does it typically begin in girls and boys?

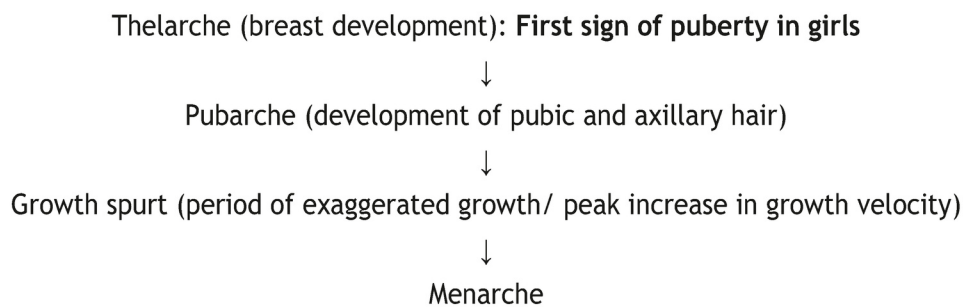
Broad Answer:

- Puberty is the period of physical and hormonal changes during which a child's body matures into an adult body capable of sexual reproduction. It typically occurs between ages 8-13 in girls and 9-14 in boys

DETAILED QUESTIONS

Q9.1: What is the typical sequence of pubertal events in girls?

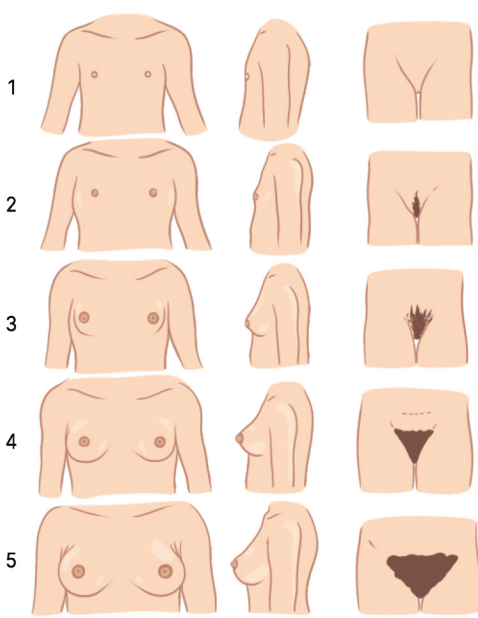
Answer:



Q9.2: How is Tanner staging (Sexual Maturity Rating) in females classified based on pubic hair development?

Answer:

 PYQ: FMGE 2019

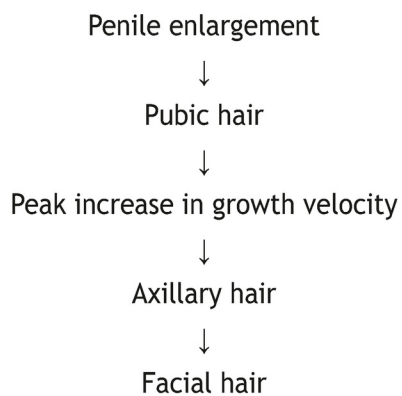
Stage 1 • Prepubertal	
Stage 2 • Pubic hair is sparse and located along the medial border of the labia	
Stage 3 • Pubic hair is thicker and curled	
Stage 4 • Pubic hair is coarse and spreads along the mons pubis	
Stage 5 • Mature adult, Pubic hair extends to the medial part of the thigh	

Q9.3: What is the typical sequence of pubertal events in boys?

Answer:

Testicular enlargement (assessed using Orchidometer): **First sign of puberty in boys**





- Deepening of the voice takes place in the later part of puberty

Q9.4: How is Tanner staging in males assessed, and what are the key features at each stage of sexual maturity?

Answer:

Stage 1 • Prepubertal • Testicular volume <4 CC • Long axis <2.5 cms	
Stage 2 • Testicular volume >4 CC • Minimal sparse hair at the root penis and scrotum	
Stage 3 • Scrotum reddens • Penis increases in length • Coarser pubic hair	
Stage 4 • Penis increases in length and breadth • Testicular enlargement continues • Pubic hair spreads mons pubis	
Stage 5 • Mature adult • Pubic hair is coarse and curly in the male pattern • Spreads across the medial thigh	

Q9.5: According to WHO, what is the age range for adolescence, and how is it subdivided?

Answer:

- Adolescent age group according to WHO: 10-19 years of age
 - o Early: 10-13 years
 - o Mid: 14-16 years
 - o Late: 17-19 years

Q9.6: At what stage of SMR does growth spurt occur in males and females?**Answer:**

- Females- SMR stage 3
- Males- SMR stage 4
 - Growth spurt occurs later and lasts longer in males.

Q9.7: What is pubertal gynecomastia in males, and does it require treatment?**Answer:**

- During puberty, males can sometimes develop pubertal gynecomastia, which is usually self-limiting, and no management is required

Q9.8: What is the HEAD-SS assessment, and how does it help in evaluating adolescents?**Answer:**

- It is a non-judgmental assessment
- Assess about
 - Home
 - Education
 - Activities/alcohol
 - Diet/drugs
 - Self-esteem/sexuality
 - Safety/suicidal tendencies
- Helps to identify adolescent problems



2. NEONATOLOGY

BIG QUESTION 1: Define neonates.

Broad Answer: A neonate refers to a newborn infant during the **first 28 days of life** (0-28 days) after birth, regardless of gestational age at delivery.

DETAILED QUESTIONS

Q1.1 Define and classify neonates based on birth weight and gestational age.

Answer:

Gestational age :

- Preterm: <37 completed weeks of gestation
- Term: 37-42 weeks of gestation
- Post-term/mature neonates: ≥ 42 weeks of gestation

Birth weight:

- Low birth weight neonate (LBW): <2500 g
- Very low birth weight neonate (VLBW): <1500 g
- Extremely low birth weight neonates (ELBW): <1000 g

PYQ: INICET 2022

Q 1.2 Differentiate between SGA and LGA neonates with examples.

Answer:

- **Small for gestational age (SGA):** Birth weight is <10th percentile of expected gestational age.
 - o Seen in:
 - Intrauterine growth restriction
 - Maternal malnutrition
 - Placental insufficiency.
- **Large for gestational age (LGA):** Birth weight is >90th percentile of expected gestational age
 - o Seen in:
 - Infants of diabetic mothers
 - Congenital hypothyroidism
 - Beckwith-Wiedemann syndrome.

Q1.3 What is the normal heart rate and respiratory rate of a neonate?

Answer:

- Heart rate: 120-140 bpm.
- Respiratory rate: 40-60/min

Q1.4 Why is a short systolic murmur considered normal in neonates?

Answer: It is due to the sudden increase in pulmonary blood flow as pulmonary vascular resistance drops.

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Q1.5 What is acrocyanosis, and is it normal in neonates?

Answer: Peripheral cyanosis of hands and feet; yes, it is normal at birth.

Q1.6 Describe the Expanded New Ballard Score and its components.

Answer:

Expanded new Ballard score (ENBS):

- Used for the assessment of the gestational age in a neonate.
- Between 20-44 weeks
- Parameters of physical maturity and neurological maturity

Neurological maturity

- Upper limb:
 - o Square window
 - o Scarf sign
- Lower limb:
 - o Popliteal angle
 - o Heel to ear

Q1.7 What are the signs of physical maturity in a preterm neonate?

Answer:

General features:

- Small/ emaciated
- Lesser subcutaneous fat- more chances of developing hypothermia
- Skin- thin, pink and fragile- more chances of infections and neonatal sepsis
- Generalised hypotonia- extended posture
- Abundant lanugo but little vernix caseosa

Head to toe:

- The head appears relatively large
- Anterior fontanelle (diamond-shaped area on the skull) - large and wide open (other causes: rickets, cleidocranial dysostosis, osteogenesis imperfecta, congenital infections, hydrocephalus)
- Ear cartilage is poorly formed - ear recoil is poor
- Breast buds <5mm
- Male: Undescended testis, poorly formed smooth scrotum
- Female: Labia majora is widely separated, and labia minora is clearly visible
- Absent deep creases on the sole

BIG QUESTION 2: What are common neonatal conditions that do not require treatment, and how can they be differentiated from pathological conditions?

Broad Answer: Certain neonatal conditions like erythema toxicum, milia, Mongolian spots, stork bites, subconjunctival haemorrhage, mastitis neonatorum, hymenal tags, vaginal bleeding, and physiological phimosis or weight loss are self-limiting and benign. They are often due to maternal hormone withdrawal, minor vascular or skin changes, or immature skin structures. These conditions resolve spontaneously, requiring no specific treatment except reassurance.

DETAILED QUESTIONS

Q2.1 Describe the benign skin conditions of the skin and mucosa seen in neonates that do not require treatment.

Answer:

- **Erythema toxicum:**
 - o Reddish maculopapular lesions present mainly on the trunk
 - Usually present in 1st week of life
 - Sterile eosinophil-filled lesions
- **Milia:** Colourless papules due to the plugging of sweat ducts
- **Mongolian spots:** Bluish black areas of discolouration mainly on the lower back, buttocks, and back of the thigh
- **Stork bites:** Capillary hemangiomas- seen in between the brows, nape of the neck
- Subconjunctival hemorrhage

Q2.2 Explain mastitis neonatorum and its clinical significance.

Answer:

- B/l breast engorgement in both males and females due to the effect of maternal hormones
 - o Milk output can also sometimes be seen
 - o Vaginal bleeding in females: Due to the withdrawal of maternal hormones
 - o Hymenal tags in females
 - o Physiological phimosis in males
 - o Physiological weight loss

Q2.3 What is physiological weight loss in neonates, and when is it regained?

Answer:

	Weight loss	Regained by
Term neonates	10% of their birth weight in 3-5 days	Day 10 of life
Preterm neonates	15% of their birth weight in 7-10 days	Day 15 of life

Q2.4 Differentiate between Cephalhematoma and Caput Succedaneum

🔍 PYQ: FMGE 2021 🔍 PYQ: NEET PG 2022

Answer:

Cephalhematoma	Caput succedaneum
• Subperiosteal hemorrhage involving cranial bones • Collection of blood between bone and periosteum • Swelling will not cross sutures/ midline • Takes 24-48 hours to appear completely • May take upto 5-7 weeks to disappear • Predisposes to neonatal jaundice	• Due to edema in the layers of scalp • Swelling can cross sutures/ midline • Present at birth in its maximum size • Disappears in 48-72 hours • Does not predispose to neonatal jaundice (as there is no collection of blood)

Q2.5 What are the various neonatal primitive reflexes, and when they appear and disappear?

Answer:

🔍 PYQ: INICET 2022, 23 🔍 PYQ: NEET PG 2023 🔍 PYQ: FMGE 2022, 23

	Appears	Disappears
Rooting reflex	32 weeks of gestation	1 month post natally
Moro's reflex	-	-
Palmar grasp reflex	28 weeks of gestation	3 months post natally
ATNR	35 weeks of gestation	3-5 months post natally

Q2.6 At what gestational age does the rooting reflex appear?**Answer:** 32 weeks GA**Q2.7 Which primitive reflex persists throughout life?****Answer:** Parachute reflex**Q2.8 Which reflex, if absent at birth, suggests severe HIE?****Answer:** Moro's reflex**Q2.9 Describe the asymmetric tonic neck reflex****Answer:** When the head of the neonate is turned to one side, the upper and lower limbs on the same side are extended, and those on the opposite side are flexed**Q2.10 Which neonatal reflexes appear after birth?****Answer:**

	Appears	Disappears
STNR (Symmetric Tonic Neck Reflex)	4-6 months post natally	8-12 months
Parachute reflex	7-8 months post natally	Persists throughout life

Q2.11 What is Moro's reflex? Explain its timing and clinical implications.**Answer:**

- Begins to appear: 28 weeks of gestation
- Completely appears: 37 weeks of gestation
- Disappears: 5-6 months post-natally (if it does not disappear beyond 6 months- Cerebral damage)
- Absent Moro's reflex: Stage 3 HIE (Hypoxic ischaemic encephalopathy), Down's syndrome

Q2.12 How is asymmetric Moro's reflex clinically interpreted?**Answer:**

- Erb's palsy
- Congenital hemiplegia
- Fracture of the clavicle
- Unilateral shoulder joint dislocation

BIG QUESTION 3: Describe the essential steps and key interventions in neonatal resuscitation.**Broad Answer:**

- Neonatal resuscitation begins with maintaining an ideal delivery room temperature of 25°C, assessing term status, tone, and breathing. Routine care includes providing warmth (radiant warmer/skin contact), clearing the airway (mouth > nose), drying the baby, and ongoing evaluation for respiratory distress.
- Positive Pressure Ventilation (PPV) is indicated for apnea, gasping, or HR <100 bpm (rate: 30-40 breaths/min, FiO₂: 21% for term, 30% for preterm). Chest compressions are started if HR <60/min despite PPV, with a 3:1 ratio of compressions to breaths. Adrenaline (0.02 mg/kg of 1:10,000) is given IV (via UVC) if HR <60/min persists. Special considerations include avoidance of bag-mask ventilation in congenital diaphragmatic hernia, and delayed cord clamping for ≥30 sec in stable babies.

DETAILED QUESTIONS

Q3.1 What is the ideal delivery room temperature for neonatal resuscitation?

Answer: 25°C

Q3.2 Is routine suctioning done for all babies during resuscitation?

Answer: No, only when excessive secretions are present.

Q3.3 Why is mouth suction done before nose suction?

Answer: To prevent aspiration and avoid stimulating a breath with a blocked airway.

Q3.4 When is routine care provided to a newborn?

Answer: When the baby is term, has good tone, and is breathing/crying normally.

Q3.5 What is the first step in routine care?

Answer: Provide warmth via a radiant warmer or skin-to-skin contact.

Q3.6 Is airway clearing always required?

Answer: No, only if secretions or obstruction are present.

Q3.7 What are the components of routine care?

Answer:

- Provide warmth: Radiant warmer / skin contact
- Clear the airway if required
- Dry the baby
- Ongoing evaluation - for respiratory distress

Q3.8 What ongoing assessment is done in routine care?

Answer: Monitor for respiratory distress and effective transition.

Q3.9 What are the Indications of PPV during neonatal resuscitation and how is it given?

Answer:

- Indications:
 - o Apnea
 - o Gasping
 - o HR < 100 bpm
- Given using a self-inflating bag and mask
- Oxygen may or may not be connected
- Reservoir: Increases the FiO₂ delivered to the baby
- In term babies - start with room air (FiO₂ - 21%)
- In preterm babies, start with 30% oxygen using a blender
- Self-inflating bag and mask are given at the rate of 30-40 breaths/ min

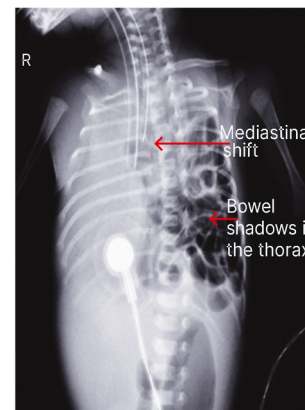


Q3.10 What are the Absolute contraindications to bag and mask ventilation?

Answer:

🔍 PYQ: INICET 2023, 24 🔍 PYQ: NEET PG 2023 🔍 PYQ: FMGE 2021

- Congenital diaphragmatic hernia
- Bowel gas shadows are present in the thorax
- The diaphragmatic outline is not clearly visible
- Heart shadow is not visualised due to mediastinal shift
- Pulmonary hypoplasia
- If there is severe respiratory distress - Intubation and bag and tube ventilation need to be done
- CDH can be diagnosed antenatally or after birth if the baby has a scaphoid abdomen, respiratory distress and features of mediastinal shift



Q3.11 When are chest compressions indicated during neonatal resuscitation, and how are they performed?

Answer:

- Indication: HR < 60/min, despite effective PPV
- **Ratio of CC: PPV - 3:1**
- Site: Lower 1/3 of the body of the sternum / at the midline, just below the line joining the 2 nipples
- Use of O₂: 100% O₂

Q3.12 When is adrenaline indicated in neonatal resuscitation, and how is it administered?

Answer:

- **Indication:** HR < 60/min, despite 60 seconds of chest compressions and PPV
- **Dose:** 0.02 mg/kg/dose or 0.2ml/kg/dose of 1:10,000 or (can be repeated 3 times)
- **Preferred route:** IV through the Umbilical venous catheterisation (UVC)
- Intratracheal (only if vascular access cannot be secured)

Q3.13 What is the minimum time for delayed cord clamping?

Answer: For at least 30 sec in all stable term and preterm neonates

Q3.14 What are the advantages of delayed cord clamping?

Answer:

- Higher Hb level
- Lesser need for blood transfusion
- Lesser risk of hypotension
- There is a slightly higher risk of neonatal jaundice in delayed cord clamping, but the advantages outweigh the risk

Q3.15 What are the latest recommendations for the resuscitation of a baby with a history of meconium-stained liquor (MSL)?

Answer:

Latest recommendations

- Continue resuscitation as per the neonatal resuscitation protocol
- Routine endotracheal intubation and tracheal suction of all non-vigorous neonates is not required
- At least 1 person skilled in endotracheal intubation should be available during resuscitation

BIG QUESTION 4: Explain the major complications seen in infants of diabetic mothers (IDM) and their underlying pathophysiology.

Broad Answer: Infants of diabetic mothers are often large for date (macrosomia) due to maternal hyperglycemia, causing fetal hyperinsulinemia, which promotes fat and glycogen deposition. This increases the risk of birth trauma and asphyxia. Metabolically, they commonly develop hypoglycemia, hypocalcemia, polycythemia, and hyperbilirubinemia due to increased insulin activity and stress responses post-delivery. Respiratory distress syndrome (RDS) may occur even in term neonates because hyperinsulinemia delays surfactant maturation. Cardiovascular anomalies like VSD (most common) and transposition of great arteries (most specific) are frequent. Neurologically, caudal regression syndrome (sacral agenesis) is highly specific for IDM.

DETAILED QUESTIONS

Q4.1 What are the components of essential newborn care?

Answer:

- Keep baby warm and dry
- Resuscitation, if required
- Care of eyes (two separate sterile gauze swiped from medial to lateral)
- Care of cord (use of sterile cord clamp and cord tie; nothing to be applied to the umbilical cord) ·
- Initiation of breastfeeding

Q4.2 What are the recommended initial modes of feeding in different gestational ages?

Answer:

Gestational age	Recommended initial method of feeding
<28 weeks	IV fluids +/- TPN
28-31 weeks	Orogastric or Gavage feeding
32-34 weeks	Katori spoon or Paladai feeding
>34 weeks	Direct breastfeeding

Q4.3 Which is the most common organisms responsible for neonatal sepsis?

Answer:

- 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 streptococcus

Q4.4 What are the risk factors for early onset sepsis?

Answer:

- Maternal fever
- Foul-smelling liquor
- Multiple PV examinations during pregnancy
- Low birth weight
- Prematurity

Q4.5 What are the Risk factors for late-onset sepsis?

Answer:

- Lack of breastfeeding
- Skin infection of the neonate
- Infection of the umbilical cord stump
- Multiple interventions

Q4.6 Which is the most common method to prevent neonatal sepsis?

Answer: Proper hand washing of the caregivers (for 2 minutes)

Q4.7 What are the components of the sepsis screen?

Answer:

- TLC (total leukocyte count)
- ANC (absolute neutrophil count)
- IT ratio (immature: total neutrophil ratio)
- CRP
- Micro-ESR

Q4.8 When is the sepsis screen said to be positive?

Answer:

- When any 2 out of the 5 parameters are abnormal
 - o Confirmatory test to diagnose neonatal sepsis: Blood culture
 - o Treatment of choice for neonatal sepsis: IV broad-spectrum empirical antibiotics
 - o Duration of antibiotic therapy:

Sepsis screen	Blood c/s	CSF s/o meningitis	Duration
+	-	-	1 week
+/-	+	-	2 weeks
+/-	+/-	+	3 weeks

Q4.9 Define and classify neonatal hypothermia.

Answer:

- Definition: Axillary temperature $<35.5^{\circ}\text{C}$
- Classification
 - o Cold stress: $36-36.4^{\circ}\text{C}$
 - o Moderate hypothermia: $32-35.9^{\circ}\text{C}$
 - o Severe hypothermia: $<32^{\circ}\text{C}$

Q4.10 What is cold stress?

Answer: Clinically, when the abdomen is warm to the touch, but the soles are cold, it is cold stress

Q4.11 What is the most important mechanism for protection against hypothermia in neonates?

Answer: Non-shivering thermogenesis- due to the presence of brown fat (lipid deposits rich in mitochondria).

Q4.12 Why is it recommended to keep the head of the baby covered?

Answer: Maximum heat loss- via the head of the baby

Q4.13 Which areas in neonates are rich in brown fat?

Answer:

- Axilla
- Groin
- Nape of neck
- Inter scapular area

Q4.14 What are the indications and components of Kangaroo mother care?

Answer:

Indication

- All low birth weight neonates are stable

Components

- Kangaroo position: Skin-to-skin contact
- Kangaroo nutrition: Exclusive breastfeeding, no supplementation
- Early discharge from the hospital

Q4.15 What are the advantages of Kangaroo mother care?

Answer:

- Lesser chances of: Sepsis, Hypothermia & Neonatal mortality
- Higher weight gain (better gain of anthropometric parameters)
- Earlier discharge from the hospital
- Higher breastfeeding rates

Q4.16 What are the devices used to keep neonates warm?

Answer:

Radiant warmer

- Open care system
- Method by which the neonate is kept warm: Radiation

Incubator

- The baby is kept inside a closed box
- Method by which the neonate is kept warm: Convection
- Preferred for preterm neonates as it helps to maintain humidity to prevent further losses due to evaporation



Incubator

Radiant warmer

Q4.17 Define Neonatal hypoglycemia and specify its treatment

Answer:

Definition:

- Blood glucose < 40 mg/dl or plasma glucose < 45 mg
- M.C.: Jitteriness/ tremors (when limbs are held, jitteriness stops and is not associated with uprolling of eyes; whereas in seizures, movements do not stop when limb is held)
- Others: Apnea, seizures, increased sweating, lethargy

Treatment

- If asymptomatic: Offer a feed and regular monitoring of blood glucose
- Frequent feeding
- If symptomatic: IV 10% dextrose 2 ml/kg
- Start continuous IV fluid GIR- glucose infusion rate (6 mg/kg/min)

Q4.18 What are the Important causes of persistent hypoglycemia during infancy?**Answer:**

Endocrine causes	Metabolic disorders
• Congenital hypopituitarism • Congenital adrenal insufficiency. • Congenital hyperinsulinemia (earlier known as Nesidioblastosis and currently it is known as PHHI- persistent hyperinsulinemic hypoglycemia of infancy) - most common cause	• Glycogen storage disease • Galactosemia • Hereditary fructose intolerance • Mitochondrial disorders • Fatty acid oxidation defects

Q4.19 Discuss the pathophysiology behind macrosomia and hypoglycemia in infants of diabetic mothers.**Answer:**

Large for date/ macrosomia: Increased chances of obstructed/prolonged labour



Use of instruments



Increased chances of birth trauma



Increased chances of birth/perinatal asphyxia

Q4.20 Describe the major complications observed in infants of diabetic mothers (IDM) with emphasis on metabolic, cardiovascular, and neurological effects**Answer:**

Metabolic	• Hypoglycemia • Hypocalcemia • Polycythemia • Hyperbilirubinemia
Respiratory system	• Increased risk of respiratory distress syndrome/ Hyaline membrane disease- due to delayed maturation of surfactant
Cardiovascular system	• Most common CHD: VSD • Most specific: Transposition of great arteries
Neurologic system	• Most common: Neural tube defects • Most specific: Sacral agenesis/ Caudal regression syndrome

BIG QUESTION 5: What is perinatal asphyxia, and how is it diagnosed and managed?**Broad Answer:**

- Perinatal asphyxia is defined as the failure of a newborn to initiate or sustain breathing at birth, resulting in decreased oxygen delivery and blood flow to vital organs, which can cause hypoxic-ischemic encephalopathy (HIE). In term neonates, the parasagittal cortex is commonly affected, whereas in preterm babies the periventricular white matter is more vulnerable.
- The diagnosis is suggested by an APGAR score of 0-3 persisting for more than 5 minutes, umbilical cord blood pH less than 7, clinical evidence of central nervous system dysfunction such as seizures or abnormal tone, and the presence of multi-organ involvement.

- Management is mainly supportive and includes prompt neonatal resuscitation, ensuring adequate oxygenation and ventilation, correction of metabolic abnormalities, and maintaining circulatory stability. In infants with moderate HIE, therapeutic hypothermia started within 6 hours of birth has been shown to improve neurological outcomes.
- The prognosis depends on the severity and stage of HIE, with mild cases often recovering well while severe asphyxia carries a high risk of long-term neurodevelopmental impairment or death.

DETAILED QUESTIONS

Q5.1 Define Perinatal asphyxia, and what are the Parts of the brain most commonly affected due to asphyxia?

Answer:

- Definition: Inability to initiate or sustain breathing → damage to the brain (HIE)
- Parts of the brain most commonly affected due to asphyxia:
 - o In term neonate: Parasagittal area → Spastic quadriplegia
 - o In preterm neonates: Periventricular area → Spastic diplegia

Q5.2 How is Perinatal asphyxia diagnosed?

Answer:

- APGAR score: 0-3 for > 5 mins
- Cord blood pH: pH < 7
- CNS dysfunction: Seizures, tone abnormalities, abnormal Moro's
- Dysfunction of other organs: Acute kidney injury, liver dysfunction

Q5.3 What are the components of the APGAR score, and how is it interpreted?

Answer:

 PYQ: NEET PG 2020, 23  PYQ: FMGE 2021

	0	1	2
A - Appearance	Completely blue or pale	Body pink but extremities blue	Completely pink.
P - Pulse rate	Absent	< 100/ min	> 100/ min
G - Grimace	No response	Grimaces only	Coughs/ sneezes
A - Activity	Limp and flaccid	Some flexion present	Flexed posture with actively moving limbs
R-Respiratory effort	None	Slow and irregular	Normal/ strong

- Total score: 10
- Minimum score: 0
- APGAR score > 7- normal
- APGAR score 0-3 - severely depressed neonate

Q5.4 Describe the Staging of HIE (Hypoxic Ischemic Encephalopathy).

Answer:

- Treatment: Mainly supportive care
- Therapeutic hypothermia may be tried in moderate HIE

Parameters	Stage I (mild)	Stage II (moderate)	Stage III
Level of consciousness	Hyper alert	Normal/ depressed	Comatosed
Tone	Normal	Mild hypotonia	Severe hypotonia
Moro's reflex	Exaggerated	Normal/ depressed	Absent
Seizures	-	Present	-
Prognosis	99% normal	80% normal	• 50% death • 50% severe neurologic sequelae

Q5.5 What are the common types and causes of neonatal seizures?

Answer:

- M.C. type: Subtle seizures
- Most common cause: Hypoxia
- Type with worst prognosis: Myoclonic seizures
- Best outcome: Focal clonic seizures

Q5.6 What is the preferred first-line neuroimaging in neonates for Neonatal seizures?

Answer: Transcranial USG through the open anterior fontanelle

Q5.7 What is the DOC for Neonatal seizures?

 PYQ: INICET 2022

Answer:

- Phenobarbitone
- Levetiracetam (fewer side effects)

 PYQ: NEET PG 2019  PYQ: FMGE 2021, 22  PYQ: INICET 2021, 22

BIG QUESTION 6: What is Respiratory Distress Syndrome (RDS) in neonates, and how is it managed?

Broad Answer: RDS, also known as hyaline membrane disease, is the most common cause of respiratory distress in preterm neonates due to a deficiency of mature surfactant. Surfactant (mainly DPPC and surfactant protein B) is produced by type II alveolar cells, matures by 35 weeks, and prevents alveolar collapse by reducing surface tension. Deficiency leads to atelectasis, interstitial oedema, and ground-glass opacity with air bronchograms on X-ray. Diagnosis includes tests like L:S ratio >2:1, phosphatidylglycerol estimation, and the shake test. Management includes CPAP, intratracheal surfactant (INSURE or LISA), and mechanical ventilation if severe. Antenatal corticosteroids (Betamethasone or Dexamethasone) prevent RDS by accelerating lung maturity.

Q6.1 What is the basic defect in respiratory distress?

Answer: Defect: deficiency of mature surfactant

Q6.2 What is the significance of pulmonary surfactant in neonatal lung function, and how does its deficiency lead to RDS?

Answer:

- Produced by type II alveolar cells
- Synthesis begins in fetal lungs at 20 weeks
- Begins to appear in amniotic fluid- 28 weeks
- Mature and in adequate amount- >35 weeks
- Most important component of surfactant: DPPC (Dipalmitoyl Phosphatidyl Choline or Lecithin)
- Most important surfactant protein: Surfactant Protein B

- Function of surfactant: to decrease the surface tension of alveoli
 - o Prevents alveoli from collapsing
 - o Increases lung compliance

When mature surfactant is deficient

- Alveolar collapse
- Interstitial edema
- Alveolar damage
- Decreased lung compliance
- Deposition of fibrin-hyaline membrane disease

Q6.3 What are the radiological findings of RDS?

Answer:

- Small-volume lungs with ground-glass haziness of lungs are seen
- Sometimes air bronchogram is seen- radiolucent areas

Q6.4 What are the Ways to detect the adequacy of mature surfactant in amniotic fluid?

Answer:

- L:S ratio (Lecithin: Sphingomyelin ratio): > 2:1
- Estimation of phosphatidylglycerol level
- Nile blue sulphatase test
- **Shake test:** Equal volumes of amniotic fluid and ethyl alcohol are taken and shaken together and let to stand for a few hours- if one or more complete rings of bubbles are present on top indicates that mature surfactant is present

Q6.5 Describe Silverman's score and its parameters for assessing respiratory distress in neonates.

Answer:

 PYQ: INICET 2022, 23
  PYQ: NEET PG 2022
  PYQ: FMGE 2022

	0	1	2
Upper chest retraction	Chest and abdomen move equally with respiration	Chest wall lags behind abdomen	See-saw pattern (paradoxical breathing)
Lower chest retraction	Absent	Minimal	Marked
Xiphisternal retractions	Absent	Minimal	Marked
Nasal flare	Absent	Minimal	Marked
Grunt	None	Audible only with stethoscope	Loud, audible without stethoscope

Q6.6 Define grunt.

Answer: Grunt is a noise due to expiration against a partially closed glottis

Q6.7 Explain Downe's score and its clinical significance in term neonates.

Answer:

- For the assessment of respiratory distress in a term neonate
- Maximum score: 10

YOURWISH

- Minimum score: 0
- Score <3 - Normal
- Score >7 - Severe respiratory distress

	0	1	2
Cyanosis	No cyanosis	Present on room air	Present at FiO ₂ >40%
Air entry	Normal	Decreased	Barely audible
Respiratory rate	<60 mins	60-80/ min	>80/ min
Grunt	Absent	Audible only with stethoscope	Audible without stethoscope
Retractions	None	Mild	Marked

Q6.8 Describe the stepwise management of mild, moderate, and severe respiratory distress syndrome in neonates.

Answer:

- Mild: CPAP (continuous positive airway pressure- prevents alveoli from collapsing during expiration) +/- O₂
- Moderate to severe:
 - o Intratracheal surfactant:
 - INSURE: Intubate → give Surfactant → Extubate
 - LISA: Less Invasive Surfactant Administration
 - o Respiratory support: CPAP or Mechanical ventilation

Q6.9 Discuss the role of antenatal corticosteroids in the prevention of respiratory distress syndrome (RDS).

Answer:

Betamethasone:	• 12 mg • 2 doses 24 hours apart • Total dose: 24 mg • IM • Slightly more neuroprotective	? PYQ: INICET 2024 ? PYQ: FMGE 2020, 21 ? PYQ: NEET PG 2022, 23
Dexamethasone:	• 6mg • 4 doses 12 hours apart • Total dose: 24mg • Cheaper and easily available	

Q6.10 What are the benefits and contraindications of Antenatal corticosteroids?

Answer:

Benefits	• Antenatal corticosteroids decrease the risk of: - o RDS - o IVH - o Neonatal mortality - o Necrotising enterocolitis • Does not decrease the risk of neonatal jaundice • Administered to all pregnant women between 24 to 34 weeks of gestation
Contraindication	• Clinical chorioamnionitis

Q6.11 What are the clinical features, radiological findings and treatment of Meconium aspiration syndrome?

Answer:

 PYQ: FMGE 2019

- C/F: SGA/IUGR baby born through meconium-stained liquor with respiratory distress soon after birth
- On examination: Increased anteroposterior diameter of the chest due to hyperinflation
- CXR:
 - o Hyperinflated lungs (appear more black, flattening of the diaphragm)
 - o Chemical pneumonitis
 - o Segmental collapse
- Treatment
 - o Supportive

Q6.12 What is transient tachypnea of the newborn (TTN), what are its chest ray features and how is it managed?

Answer:

- Also called delayed adaptation
- Due to the delayed clearance of lung fluid
- M.C. in babies born through - C-section

CXR

- Fluid in the interlobar fissure
- Perihilar streaking/ prominent bronchovascular markings
- Pleural effusion

Management:

- Self-limiting

Q6.13 Discuss the risk factors, clinical staging, and management of necrotising enterocolitis.

Answer:

Important risk factors

- Prematurity
- Lack of breastfeeding/ use of formula feeding
- 10% cases can occur in term neonates

Stages of NEC

- Ia: Occult blood in stool
- Ib: Fresh blood in stool
- IIa: Pneumatosis intestinalis
- IIb: Portal vein gas
- IIIa: Peritonitis
- IIIb: Gas under diaphragm/ pneumoperitoneum

Treatment

- Mainly supportive
- NPO
- IV fluids
- IV antibiotics
- Surgery (stage IIIb)

BIG QUESTION 7: Differentiate physiological from pathological jaundice, and outline key causes and treatment of neonatal jaundice.

Broad Answer: Physiological jaundice appears after 24 hours, is always unconjugated, and resolves within 3 weeks, whereas pathological jaundice may appear in the first 24 hours, involve conjugated bilirubin, dark urine, pale stools, and prolonged duration. Causes of unconjugated jaundice include hemolysis (ABO/Rh incompatibility, G6PD deficiency) and reduced conjugation (Crigler-Najjar). Conjugated jaundice arises from infections, metabolic disorders, or biliary atresia. Treatment involves phototherapy (460-490 nm, structural isomerisation to lumirubin), exchange transfusion for severe cases, and IVIg for hemolytic disease

DETAILED QUESTIONS

Q7.1 Discuss the differences between physiological and pathological jaundice with clinical features.

Answer:

🔍 PYQ: INICET 2021, 22 🔍 PYQ: NEET PG 2023 🔍 PYQ: FMGE 2020, 21

Physiological jaundice	Pathological jaundice
Clinical jaundice never appears in 24 hours	May appear in first 24 hours
- Always unconjugated - No dark urine/ pale stools	- May be conjugated or unconjugated - High coloured urine/ pale stools+
Palms and soles are not yellow	Palms and soles may be yellow
Does not last beyond 3 weeks	May last longer

Q7.2 What are the differences between breast milk jaundice and breastfeeding jaundice

Answer:

Breastfeeding jaundice	Breastmilk jaundice
Due to inadequate breastfeeding	Due to pregnanediol and free fatty acids ↓ Interfere with the conjugation of bilirubin
Rx: Frequent breastfeeding	Rx: Continue breastfeeding, unless bilirubin >20 mg/dl

Q7.3 Describe the aetiology of unconjugated hyperbilirubinemia in neonates.

Answer:

- Increased production of bilirubin
 - Erythroblastosis fetalis / Hemolytic disease of newborn (ABO/ Rh incompatibility)
 - Hereditary spherocytosis
 - G6PD deficiency
 - Cephalhematoma
- Decreased conjugation of bilirubin
- Crigler-Najjar syndrome (deficiency of UDP-glucuronyl transferase)

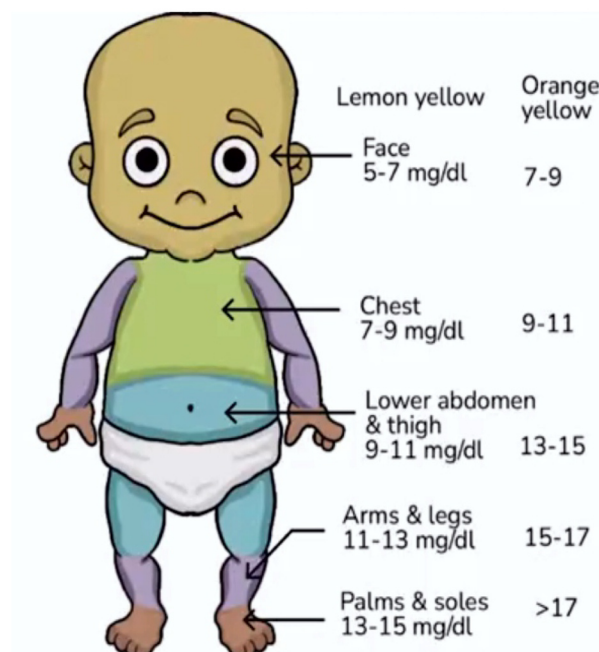
Q7.4 Describe the aetiology of conjugated hyperbilirubinemia in neonates.

Answer: Conjugated hyperbilirubinemia: > 2 mg/dl or >20% of total bilirubin

Non obstructive:	• Neonatal sepsis: Bacterial, viral, parasitic • UTIs • Viral infections • Metabolic disorders: HFI, alpha 1 antitrypsin deficiency, galactosemia • Idiopathic neonatal hepatitis
Obstructive:	Extrahepatic: • Extrahepatic biliary atresia: is done for screening HIDA scan • Surgery: Kasai procedure Intrahepatic: • Congenital hepatic fibrosis • Progressive familial intrahepatic cholestasis • Alagille syndrome • Dubin-Johnson syndrome • Rotors syndrome

Q7.5 Describe modified Krammer's rule.

Answer:



Q7.6 Which part of the brain mc involved in hyperbilirubinemia?

Answer: Basal ganglia (kernicterus, yellowish discolouration of the basal ganglia)

Q7.7 Name the type of cerebral palsy seen in kernicterus.

Answer: Extrapyraxidal

Q7.8 What are the Features of acute bilirubin encephalopathy?

Answer:

- Early: Hypotonia, lethargy, poor feeding
- Late: Fever, seizures, hypertonia, opisthotonia

YOURWISH

Q7.9 What are the features of chronic bilirubin encephalopathy?

Answer:

- SNHL
- Athetosis
- Dental enamel changes
- Upward gaze palsy
- Intellectual disability

Q7.10 What is the treatment of neonatal jaundice?

PYQ: NEET PG 2023

Answer:

- First line: Phototherapy
- Exchange transfusion (severe cases)
- IVIg (ABO or Rh incompatibility)

Q7.11 What is the principle of phototherapy in neonatal jaundice, and how can its effectiveness be enhanced?

Answer:

- Most effective wavelength: **460-490 nm**
- Most important mechanism: Structural isomerisation; bilirubin is converted to Lumirubin, which is excreted through the kidney, Lumirubin without conjugation
- Ways to increase the effectiveness of phototherapy
 - o Decrease the distance between the baby and the phototherapy unit
 - o Increased exposed surface area
 - o Use of LED lamps

Q7.12 What are the adverse effects of phototherapy?

Answer:

- **Bronze baby syndrome** - colour of the baby becomes dark- transient
- Watery diarrhea
- Dehydration
- Hypocalcemia
- **Retinal toxicity**
- Gonadal toxicity/ mutations
- Impaired maternal-child bonding

Q7.13 When to start phototherapy?

Answer: In an otherwise well-term neonate

PYQ: INICET 2023 PYQ: NEET PG 2023

	Phototherapy cut-off (mg/dl)	Exchange transfusion cut-off (mg/dl)
24-48 hours	> 15	> 20
48-72 hours	> 18	> 25
>72 hours	> 20	> 25

- In high-risk and preterm babies, cut-offs will be lower
- Indication of exchange transfusion in a baby with Rh incompatibility:
 - o Cord blood bilirubin: **> 5 mg/dl**
 - o Cord blood Hb: **< 10 g/dl** or **PCV < 30**



3. NUTRITION AND MALNUTRITION

BIG QUESTION 1: Write briefly regarding breastfeeding and breast milk.

Broad Answer: Breast milk is the optimal source of nutrition for infants, offering a unique blend of nutrients and protective factors. The initiation of breastfeeding is crucial, with the WHO recommending it within one hour of childbirth. Exclusive breastfeeding for the first six months provides all necessary nourishment, followed by the introduction of complementary feeding, given in addition to continued breastfeeding.

DETAILED QUESTIONS

Q1.1: When is breastfeeding initiated?

Answer:

- Breastfeeding is initiated as soon as possible
- WHO recommends initiating within 1 hour of childbirth (irrespective of vaginal delivery or cesarean section)

Q1.2: What is exclusive breastfeeding?

PYQ: FMGE 2020

Answer:

- Baby should be fed only breast milk and nothing else, not even sips of water, unless medically indicated
- Recommended for the first 6 months of life exclusively

Q1.3: What is complementary feeding, and when is it initiated?

Answer:

- Complementary feeding is defined as semi-solid, energy-dense food, given in addition to breast milk
- Initiated at 6 months

Q1.4: What are the characteristics of food items used for complementary feeding?

Answer:

Mnemonic: **AFASS**

- Acceptable
- Feasible
- Affordable
- Sustainable
- Safe

Q1.5: What are the recommended storage durations for expressed breast milk (EBM)?

Answer:

- At room temperature (25°C): 8-10 hrs
- In a refrigerator (2-8°C): 24 hrs
- In a deep freezer (-20°C): 3 months

Q1.6: When is the breast milk output maximum?**Answer:**

- At 5-6 months of lactation
- Approx. 730 ml / day

Q1.7: What are the factors affecting breast milk output?**Answer:**

Milk Output Increased by	Milk Output Decreased by
• Thought of baby • Sight of baby • Sound of baby • Nighttime feeds • Frequent feeding/Emptying of breasts	• Maternal Stress, Anxiety • Use of Top feeds/ formula feeds • Feeding bottle/pacifiers • Maternal pain

Q1.8: What are the signs of good positioning while breastfeeding?**Answer:**

- The body of the baby should be well supported
- The occiput, shoulders, and buttocks of the baby should be in a straight line
- The entire body of the baby should be turned towards the mother
- The abdomen of the baby should touch the abdomen of the mother

Q1.9: What are the signs of good attachment while breastfeeding?**Answer:**

- The mouth of the baby should be wide open
- The entire areola should be in the baby's mouth, except for a small upper part that may be visible
- The lower lip of the baby should be everted/ turned out
- The chin of the baby should touch the mother's breast

Q1.10: Name the contraindications to breastfeeding related to the baby.**Answer:**

- Galactosemia
- Congenital lactose intolerance

 PYQ: FMGE 2023
 PYQ: NEET PG 2021

Q1.11: Name the contraindications to breastfeeding related to the mother.**Answer:**

- Absolute contraindications
 - o Mother on chemotherapy or radiotherapy
- Relative contraindications
 - o Maternal HIV
 - o Maternal varicella involving the areola and nipple area
 - o Maternal untreated tuberculosis
 - At least 3 weeks for the disease to become paucibacillary
 - o Breast abscess

Q1.12: What are the key nutritional differences between breast milk and cow milk?

PYQ: FMGE 2023

Answer:

	Breast milk	Cow milk
Lactose	7 g/dl • More energy as carbohydrate • It helps in the formation of galactose and Lactobacillus in the intestine	4.5 g/dl
Proteins	1 g/dl • Lesser solute load on kidneys • Richer in whey protein like Lactoglobulin which are easily digestible as compared to Casein in Cow milk • Richer in cysteine, Methionine that are required for CNS development	3.5 g/dl
Lipids	• Breast Milk is richer in PUFA (poly unsaturated fatty acids) and DHA (Docosahexaenoic acid): Required for CNS development of the baby	
Minerals	• Calcium: Phosphate ratio in Breast Milk is such that it favors calcium absorption. (2:1 ratio) • Iron present in Breast Milk is much more easily absorbable than the one present in cow's milk.	• Cow's Milk is richer in phosphate which hinders calcium absorption so increasing the chances of hypocalcemia in the baby
Vitamins	• Breast Milk contains adequate amounts of all vitamins except vitamin D, vitamin K & vitamin B12 in strictly vegan mothers.	

Q1.13: Why is vitamin D supplementation recommended for infants, and what is the suggested dose?**Answer:** Vitamin D (400 IU/day) is recommended for all babies till 1 year of life since Vitamin D is deficient in breast milk**Q1.14: Why is vitamin K given intramuscularly at birth, and what condition does it help prevent in newborns?****Answer:**

- Vitamin K - 1 mg Intramuscularly at birth is given to all babies since Vitamin K is deficient in breast milk
- It prevents hemorrhagic disease in newborns

Q1.15: What are babies predominantly fed on cow's milk at higher risk for, and why?**Answer:**

- Babies predominantly fed cow milk are at a higher risk of developing hypocalcemia and tetany, seizures, and even higher chances of Scurvy (Vitamin C is deficient in Cow's milk)
 - o Vitamin C is heat-labile (destroyed when boiled)

Q1.16: What are the health benefits of breast milk for the baby in both the neonatal period and later in life?**Answer:**

- Breast milk protects against diseases like
 - o Neonatal period: NEC, neonatal sepsis
 - o Later in life: Obesity, HTN, diabetes, allergies, Bronchial asthma, Lymphoma
 - o Breast-fed babies have higher IQs
 - o Breast milk helps in maternal-child bonding
 - o Breast milk is safe, free from contamination, and easily available even in resource-limited settings

YOURWISH

Q1.17: What are the anti-infective substances present in breast milk?**Answer:**

- Mnemonic- Teach for **PLAB**
 - Transforming growth factor β
 - Phagocytic macrophages
 - Lactoferrin
 - Lysozyme
 - Antibodies, especially IgA
 - Bifidus factor
 - Bile-stimulated lipase

Q1.18: What is colostrum, and why is it important for newborns?**Answer:**

- Any form of prelacteal feeding is contraindicated, and all babies should be fed Colostrum
- Colostrum is thick, yellowish colored milk produced in small quantities during the 1st 72 hours after the birth of the baby
- It is rich in IgA, phagocytic macrophages, and low in lactose
- It is known as 1st immunisation of the baby

Q1.19: What is preterm breast milk rich in?**Answer:**

- Mnemonic- **SIP** For Intelligent CNS
 - Sodium
 - Immunoglobulins like IgA
 - Proteins
 - Fats
 - Iron
 - Calories

Q1.20: Why is complete emptying of each breast important during feeding?

Answer: Complete emptying of each breast during a feeding session is important because the composition of foremilk and hindmilk is different

Q1.21: Elaborate on the composition of foremilk and hind milk.**Answer:**

Fore Milk	Hind Milk
● At the beginning of a feed ● More thin & watery ● Satisfies mainly the thirst of the baby	● At the end of a feed ● More thicker and calorie dense ● Richer in fat ● Satisfies the hunger of the baby

Q1.22: 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 to top feed her 2-month-old baby. How will you know whether breastfeeds are adequate for a baby?

Answer:

- The feeds are adequate when
 - o Baby sleeps for 1-2 hours after a feed
 - o Baby should pass urine at least 6-8 times/day
 - o The baby should be gaining weight

BIG QUESTION 2: What are micronutrients, and what quantity of them are required?**Broad Answer:**

- Micronutrients are indeed a vital category of nutrients that the human body requires in relatively small quantities, in contrast to macronutrients (carbohydrates, fats, and proteins), which are needed in much larger amounts. Despite being required in lesser quantities, their impact on overall health and proper bodily function is profound
- The main micronutrients are:
 - o Vitamins
 - o Minerals

DETAILED QUESTIONS**Q2.1: What is the earliest symptom and sign of vitamin A deficiency?****Answer:**

- Earliest symptom: Night blindness
- Earliest sign: Conjunctival Xerosis (Drying of conjunctiva)

Q2.2: What is the WHO classification of eye lesions due to Vitamin A deficiency?**Answer:**

XN	Night blindness	
X1A	Conjunctival Xerosis/Dryness	
X1B	• Bitot's spot • Bitot's spot are the triangular spots which are pearly white in colour	
X2	Corneal Xerosis	
X3	• Corneal ulceration/Keratomalacia • X3A: Involvement of $< 1/3^{\text{rd}}$ cornea • X3B: Involvement of $> 1/3^{\text{rd}}$ of cornea	
XS	Corneal Scars	

Q2.3: What is the treatment for vitamin A deficiency?**Answer:**

Age	Oral Vitamin A dose
< 6 months	50,000 IU
6 months-1 year or Weight < 8 kg	1,00,000 IU
>1 year or > 8 kg	2 lakh IU

Q2.4: What is the therapeutic dose of vitamin A?**Answer:**

- Therapeutic dose: 3 doses are given
 - o Dose 0 (at first contact)
 - o Dose at 24 hrs
 - o Dose at 2-4 weeks later

Q2.5: What is the recommended prophylaxis schedule for Vitamin A in children?**Answer:**

- A total of 9 mega doses is given.
- 1st dose: At 9 months with measles-rubella vaccine
- 2nd dose: At 16-24 months
- Subsequent doses: Every 6 months thereafter

Q2.6: What are the types of Beri-Beri caused by Thiamine deficiency and their features?**Answer:**

- Wet Beri-Beri: There can be features of congestive cardiac failure, like oedema, Tachypnea, and Tachycardia
- Dry Beri-Beri: Neurological features. It is not very common in children

Q2.7: What is the main cause of Thiamine deficiency?

Answer: Thiamine deficiency is predominantly caused by the consumption of polished rice

Q2.8: What are the clinical features of Riboflavin deficiency?**Answer:**

- Glossitis: The Tongue becomes shiny, pink, and devoid of papillae
- Angular Cheilitis
- Corneal involvement
- Oral ulcers

Q2.9: Name the disease caused by Niacin deficiency, its clinical features, and its treatment.**Answer:**

- Niacin deficiency causes Pellagra
- Pellagra is a disease of the 4D's
 - o Diarrhoea
 - o Dermatitis: Photosensitivity, rashes on sun-exposed areas like Casal's necklace (hyperpigmented rash in the neck area)
 - o Dementia (early onset)
 - o Death
- Treatment: Niacin or Nicotinamide

**Q2.10: Which metabolic disorder can present with a Pellagra-like rash and why?****Answer:**

- Pellagra-like rash can be seen in metabolic disorders like Hartnup disease
- It occurs due to impaired transport of neutral amino acids

Q2.11: What are the clinical features of Pyridoxine deficiency in children?**Answer:**

- **Infants:** Refractory seizures not controlled with multiple anti-epileptics
- **Older children:** Neuropathies

Q2.12: What are the main features of Biotin deficiency?**Answer:**

- Neurological manifestations: Developmental delay, Seizures
- Skin and hair changes: Skin rash, Alopecia, Abnormal hair

Q2.13: What are the main features of vitamin B12 deficiency?**Answer:**

- CNS manifestation: Subacute combined degeneration of the spinal cord
- Haematological manifestation: Megaloblastic anaemia

Q2.14: What are the main features of Folic acid deficiency?**Answer:**

- Causes Megaloblastic anaemia
- Neurological manifestations are not exclusive to isolated folic acid deficiency

Q2.15: Which coagulation factors require Vitamin K for their synthesis, and what process is involved?**Answer:**

- Vitamin K-dependent coagulation factors: II, VII, IX, and X
- They require γ -carboxylation (post-translational carboxylation) for activation

Q2.16: What are the clinical manifestations of vitamin K deficiency, and how is it treated?**Answer:**

- Vitamin K deficiency leads to bleeding manifestations known as Hemorrhagic disease of the newborn
- In older children, it leads to bleeding manifestations known as Vitamin K deficiency bleeding
- Treatment:
 - o By administration of Vitamin K
 - o In severe bleeding, Fresh frozen plasma

Q2.17: What are the features of Zinc deficiency, and what is the treatment?
 **PYQ: FMGE 2020**
Answer:

- Clinical features:
 - o Diarrhoea
 - o Delayed wound healing
 - o Hypogonadism
 - o Skin rashes around the oral cavity, genital area
 - o Acrodermatitis Enteropathica
- Treatment: High-dose Zinc

YOURWISH

Q2.18: What is the cause and clinical presentation of Acrodermatitis Enteropathica?

Answer:

- Acrodermatitis Enteropathica is caused by a defect in the transporter that helps in the absorption of Zinc
- Clinical presentation: A child with extensive rashes over the oral cavity, genital region



BIG QUESTION 3: Write briefly about malnutrition and its causes.

PYQ: FMGE 2022

PYQ: INICET 2022

Broad Answer:

- Malnutrition is an imbalance in energy or nutrient intake, leading to **undernutrition or overnutrition**
- **Causes:**
 - o Inadequate dietary intake
 - o Poor absorption or increased loss
 - o Increased requirements
 - o Chronic illnesses
 - o Overeating or an unhealthy diet

DETAILED QUESTIONS

Q3.1: What are the best indicators for acute and chronic malnutrition?

Answer:

- The best indicator of Acute Malnutrition: Decrease in weight for height (Wasting)
- The best indicator of Chronic Malnutrition: Decrease in Height for age (Stunting)

Q3.2: According to the WHO classification, how are wasting and stunting defined in children?

PYQ: INICET 2023

Answer:

- WHO classification is based on weight for height, height for age & oedema
- If oedema is present, add 'edematous' to the category

Weight for Height	Height for Age
Between -2 to -3 Z score or 70-79% of expected called as Wasting	Between -2 to -3 Z score or 85-89% of expected called as Stunting
< -3 Z score or <70% of expected called as Severe Wasting	< -3 Z score or < 85% of expected Severe Stunting

Q3.3: Elaborate on the differences between Kwashiorkor & Marasmus.**Answer:**

	Kwashiorkor	Marasmus
Edema	Present	Absent
Appetite	Poor	Good
Apathy	Present	Absent
Liver involvement	Fatty hepatomegaly	Usually absent
Skin & hair changes	More common	Less common

Q3.4: What are the findings noted in the images below?**Answer:**

Severely malnourished without oedema - Marasmus	Flaky paint dermatosis	Flag sign: Alternate band of pigmented and hypopigmented hairs
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Q3.5: What is the WHO definition of Severe Acute Malnutrition (SAM), and what are the findings in it?**Answer:**

PYQ: NEET PG 2021

- Definition: In a child between 6 months to 5 years of age, the presence of any 1 or more of the following
- Findings:
 - o Weight for height < -3 Z score or $< 70\%$ of expected or
 - o Mid arm circumference < 11.5 cm or
 - o Symmetrical bipedal oedema of nutritional origin

Q3.6: What are the complications of Severe Acute Malnutrition?**Answer:**Mnemonic: **SHIELDED**

- Sugar deficiency (hypoglycemia): Blood Glucose < 45 mg/dl
- Hypothermia: Rectal temperature $< 35.5^{\circ}\text{C}$
- Infections
- Electrolyte imbalance, especially Hypokalemia and Hypophosphatemia
- Dehydration
- Deficiency of Micronutrients

Q3.7: How is SAM treated?

Answer:

- Initial- Immediate hospitalisation, especially for those with complications
- Identify complications and treat them
- Nutritional rehabilitation.

Q3.8: How are the complications treated in SAM?

Answer:

Hypoglycemia	• 10% dextrose orally or by NG tube • If the child is very sick, it can be given through the IV route
Hypothermia	• Remove wet clothing • Cover the child properly • Use warmer
Infections	• Antibiotics (Oral/IV) usually include Amoxiclav, Broad-spectrum-Ampicillin, and Gentamycin
Electrolyte imbalance	• Supplement K⁺
Dehydration	• Oral: Resomal (rehydration solution for a malnourished child) • It has less Sodium and more Potassium than the WHO ORS
Deficiency of micronutrients	• Supplement micronutrients, Iron started later

Q3.9: How is nutritional rehabilitation done in children with Severe Acute Malnutrition?

Answer:

- Start with low calories and proteins and build it up gradually
- While feeding is introduced again, the F-75 diet is started (75 kcal and 0.75 g protein/100 ml)
- Slowly switched to F-100 diet (100 kcal and 3 g protein/100 ml)
- Discharge the child with RUTF

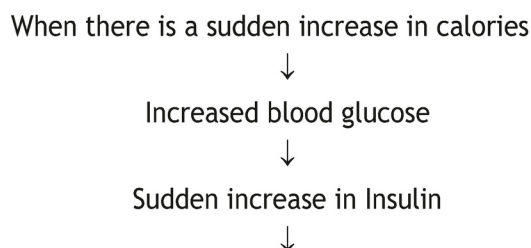
Q3.10: What is Ready-to-Use Therapeutic Food (RUTF), and why is it important in SAM management?

Answer:

- RUTF (Ready-to-Use Therapeutic Food) is a calorie-dense, paste-like, and ready-to-use food
- RUTF has 543 kcal of energy/100 grams
- Proteins and calories are increased gradually to prevent Refeeding syndrome/Nutritional recovery syndrome

Q3.11: What is the pathophysiology behind Refeeding Syndrome in malnourished children?

Answer:



Increased regeneration of tissues (A Lot of Phosphate will get used up from the body)



Abrupt decrease in Serum Phosphate



A lot of Thiamine will be used up & there is increased uptake of Potassium and Magnesium by the cells

Q3.12: What complications occur in Refeeding Syndrome due to hypophosphatemia, thiamine deficiency, and potassium & magnesium deficiency?

Answer:

- Edema
- Heart failure
- Worsening of respiratory distress
- Tachypnea, Tachycardia



4. FLUID AND ELECTROLYTES/GENETIC DISORDERS

BIG QUESTION 1: How are maintenance fluid requirements calculated in children, and what fluids are preferred?

PYQ: FMGE 2019, 20

Broad Answer: Maintenance fluids are calculated using the Holliday-Segar method: 100 ml/kg for the first 10 kg, 50 ml/kg for the next 10 kg, and 20 ml/kg for weight beyond 20 kg. For neonates, fluid needs depend on birth weight and day of life (60 ml/kg on Day 1, progressing to 150 ml/kg by Day 7). Preferred fluids include N/2 in 5% dextrose or DNS + KCl (20 mEq/L) after 48 hours of life, while 10% dextrose alone is used for the first 48 hours in neonates.

Body Weight	Fluid
For 1 st 10 kg	100 ml/kg
Next 10 kg	50 ml/kg
Beyond 20 kg	20 ml/kg

DETAILED QUESTIONS

Q1.1 How are fluid requirements in neonates calculated, and which fluid is given?

Answer: Based on birth Weight & day of life (in ml/kg/day)

Example:

- Neonate of weight 3 kg:
Day 1 → $3 \times 60 = 180 \text{ ml}$ → 12 feeds → 15 ml/ feed
- By the end of 1 week → 150 ml/kg/day

Fluid given:

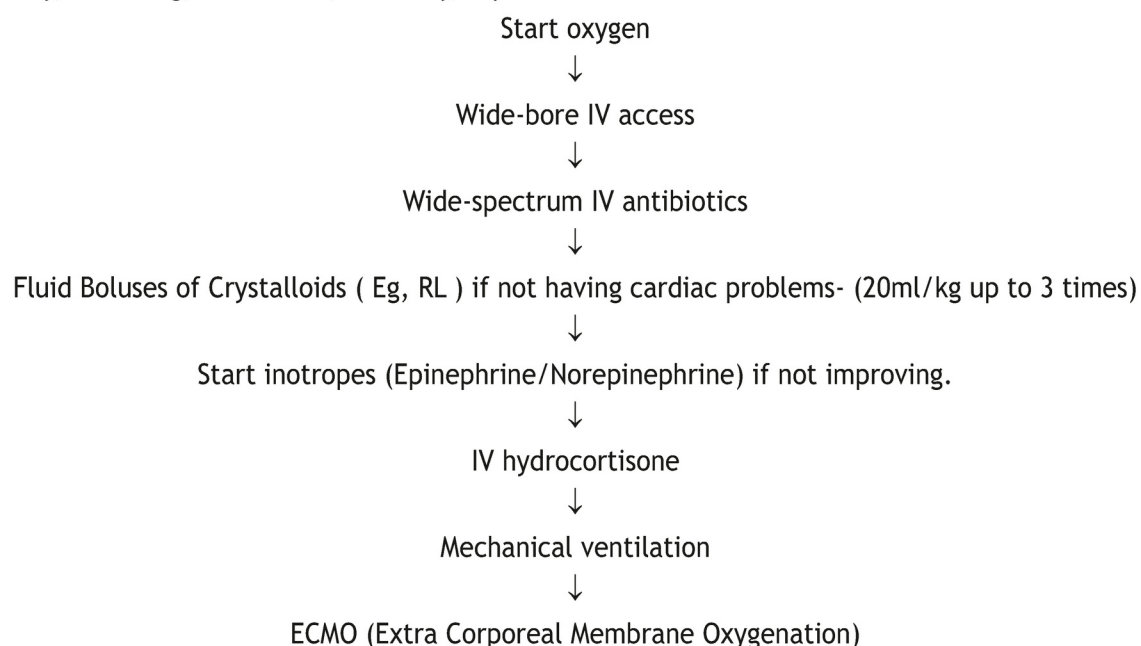
- 1st 48 hrs. of life: 10% dextrose alone
- After 48 hrs.: N/5 in 5% dextrose with 1ml KCL/100 ml

Birth Weight	D1
< 1500 gm	80/kg/day
> 1500 gm	60ml/kg/day

Q1.2 How is shock managed in children?

Answer: Airway, Breathing, Circulation, Disability, Exposure

PYQ: FMGE 2023



Q1.3 What are the recent updates in pediatric fluid and electrolyte therapy, particularly in critically ill children?**Answer:**

- A protocolised approach is not preferred.
- Individualised approach to suit patient physiology & settings.
- Crystalloids Are Preferred
- Deresuscitation
 - Restricting Maintenance fluid after initial resuscitation
 - Use of diuretics
- Advantage:
 - More ventilation-free days & shorter ICU stays (Faster recovery)
 - Decreases complications of fluid overload

Q1.4 How is fluid resuscitation calculated in a child with burns?

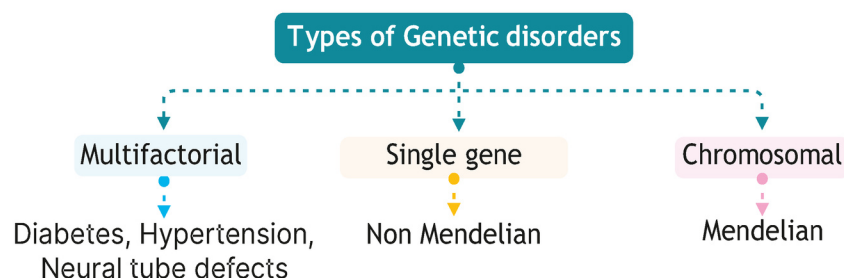
Answer: The Parkland formula is used: 4 ml/kg/% BSA burned over 24 hours. Half the volume is administered in the first 8 hours, and the remaining half is administered over the next 16 hours. Ringer's Lactate (RL) is the fluid of choice. In the next 24 hours, fluid shifts occur with reabsorption of oedema fluid and diuresis, during which RL with 5% dextrose is preferred.

BIG QUESTION 2: What is the genetic basis, screening, and clinical profile of Down syndrome?

Broad Answer: Down syndrome is the most common chromosomal abnormality in children, caused by trisomy 21 due to maternal meiotic nondisjunction (95%), Robertsonian translocation (3%), or mosaicism (1-2%). Antenatal screening includes nuchal translucency (>3 mm), absent nasal bone, and biochemical markers (β-HCG, Inhibin A). Clinical features can be remembered by "I C A PROBLEMS" - intellectual disability, congenital heart defects (VSD), Atlantoaxial instability, protruding tongue, round face, Brushfield spots, low tone, epicanthic folds, mental retardation, and increased risk of leukaemia and early-onset Alzheimer's.

DETAILED QUESTIONS**Q2.1 What are the types of genetic disorders?**

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Answer:**Q2.2 Describe the genetic causes of Down syndrome and their incidence.****Answer:**

PYQ: FMGE 2020, 23

PYQ: NEET PG 2021

- The most common chromosomal abnormality seen in children.
- Basic defect: Presence of 3 copies of chromosome 21 instead of 2, also known as **Trisomy 21**.

95% cases	• Down syndrome caused by Trisomy 21 which is due to maternal meiotic nondisjunction (most common) i.e., separation of chromosomes does not take place properly during maternal meiosis
3% cases	• Robertsonian Translocation i.e., translocation of chromosome 21 with chromosome 21 or translocation of chromosome 21 with another chromosome like chromosome 14 or 15
1-2% cases	• Mosaicism- Presence of >1 different cell line in the same individual (E.g. Trisomy 21, monosomy 21) • Individuals with mosaicism have less severe clinical features

Q2.3 Discuss the clinical features of Down syndrome

Answer: Mnemonic: I C A PROBLEMS

I	- Intellectual disability/ mental retardation, Incurved little finger/ Clinodactyly - Developmental delay	Simian crease Clinodactyly Sandal gap Epicanthic folds
C	Congenital heart diseases, Congenital hypothyroidism, Congenital diaphragmatic hernia, Duodenal atresia	
A	- Atlantoaxial subluxation - should be examined before participating in contact sports or surgeries requiring general anesthesia requiring neck extension - Acute leukemia, especially Myeloid leukemia (M7 variety) - Alzheimer's disease (Early onset) - Atresia of duodenum - Absent Moro's reflex - Due to hypotonia of limbs	
P	Protruding tongue	
R	Round face, refractive errors	
O	Occiput flat; Open; large sized anterior fontanelle	
B	Brushfield spots on iris, Brachycephaly	
L	Low tone, Low/ depressed nasal bridge, Low set ears/ Dysplastic ears, deafness	
E	Epicanthic folds/ Fold of skin over medial canthus	
M	Mongoloid slant of eyes	
S	- Sandal gap - The increased gap between the great toe and the rest of the toes - Simian crease - Short stature	

Q2.4 What are the Radiological markers for Antenatal Screening of Down syndrome?

Answer:

- In 1st trimester, increased nuchal fold thickness (>3mm) and absent Nasal bone are considered very high risk.
- For nuchal thickness, a scan is done at 11 weeks of gestation in 1st trimester.
- Soft markers: Shortened femur, absent/hypoplastic nasal bones (in 2nd trimester)
- Cardiac anomalies
- Duodenal atresia

Q2.5 What are the biochemical markers and integrated tests used for Antenatal Screening of Down syndrome?

Answer:

Biochemical markers	1st trimester (T₁): B-HCG, PAPP-A (Pregnancy Associated Plasma Protein A) 2nd trimester (T₂) - Triple test: B-HCG, Unconjugated estriol, α-fetoprotein - Quadruple test: B-HCG, Unconjugated estriol, α-fetoprotein, Inhibin A - Biochemical Markers that increase in Down syndrome: b-HCG and Inhibin A
Integrated test	- Maternal age + T₁ (Nuchal thickness + PAPP-A levels) + T₂ (Quadruple test) - All these values are used for calculated risk or composite risk in Down syndrome - Best screening test for Down syndrome in the antenatal period - It has pick up rate of > 95% cases for Downs syndrome

Q2.6 What is NIPS/NIPT?

Answer:

NIPS (Non-Invasive Prenatal Screening Test)/ NIPT (Non-Invasive Prenatal Test)

- Cell-free fetal DNA circulating in the mother's blood is used to diagnose aneuploidies like Down syndrome
- syndrome
- Major chromosomal abnormalities can be easily diagnosed or screened for by this test.
- The limiting factor for this test was its cost, and it was not easily available.

Q2.7 Which are the Confirmatory tests for prenatal diagnosis of Down Syndrome?

Answer:

- Fetal karyotype
 - o 11-13 weeks of gestation: Chorionic Villous Sampling
 - o At 14-16 weeks of gestation, Amniocentesis can be done.
 - o At 17-20 weeks of gestation, Cordocentesis can be done.
 - The above procedures are invasive and carry a risk of abortion.
 - Offered to mothers at high risk based on screening tests or previous children with Down syndrome.

Q. 2.8 What is the karyotype in Turner syndrome?

Answer: Basic defect: 45, XO

Q2.9 Which is the most common congenital heart disease in Turner syndrome?

Answer: Bicuspid aortic valve > Coarctation of the aorta

Q 2.10 Describe the clinical features of Turner syndrome

Answer: Turner syndrome has **Normal intelligence**

- | |
|--|
| <ul style="list-style-type: none"> • Short stature • Short 4th metacarpal • Sensorineural hearing loss |
| <ul style="list-style-type: none"> • Amenorrhea (Primary) |
| <ul style="list-style-type: none"> • Barr body absent |
| <ul style="list-style-type: none"> • Cardiac abnormalities • Cystic hygroma • Cubitus valgus |
| <ul style="list-style-type: none"> • Low posterior hairline • Lymphedema of hands and feet • Low thyroid |
| <ul style="list-style-type: none"> • Ovaries streak shaped/ underdeveloped • Rudimentary nucleus |
| <ul style="list-style-type: none"> • Webbed neck |
| <ul style="list-style-type: none"> • Nipples are widely spaced on a shield-shaped chest • Normal intelligence |

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Q.2.11 What are Barr Bodies?

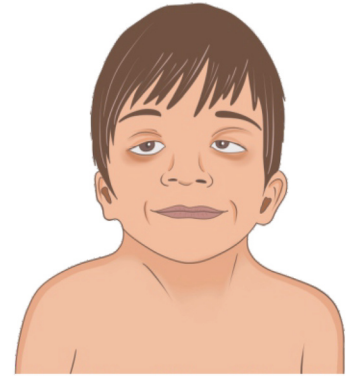
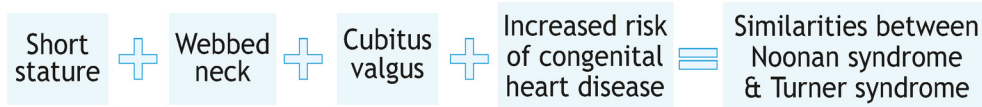
Answer:

- Number of Barr bodies in any individual = Number of X chromosomes - 1
- A sample from the buccal Mucosa under a microscope shows a dark or dense nucleus, which is a Barr body
- Normal males: $1 - 1 = 0$
- Normal females: $2 - 1 = 1$
- Turner syndrome females: $1 - 1 = 0$
 - o Female with absent Barr body

Q2.12 What are the salient features of Noonan syndrome?

Answer:

- Autosomal dominant inheritance
- The most common gene involved is the PTPN11 gene.
- It can be seen in both boys and girls.



Q2.13 Differentiate between Noonan syndrome and Turner syndrome.

Answer:

Noonan Syndrome	Turner Syndrome
• Autosomal dominant inheritance • Karyotype - Normal (microscopic deletion) • Seen in both boys and girls • Intellectual disability present • Delayed puberty, but fertility is preserved • Antimongoloid slant and low-set ears	• Do not follow any inheritance pattern • Karyotype - 45, XO • Seen only in girls • Normal intelligence • Infertile (Streak ovaries and rudimentary uterus)

- Children with Noonan syndrome have more congenital heart diseases more commonly than Turner syndrome.
- Children with webbed neck in Turner syndrome have a higher chance of having congenital heart disease.

Q.2.14 Which is the most common congenital heart disease in Noonan syndrome?

Answer: Pulmonary stenosis (supravalvular) or Pulmonary tract outflow obstruction

Q2.15 Name one syndrome other than Noonan where antimongoloid slant is seen

Answer: Treacher-Collins syndrome.

BIG QUESTION 3: How does understanding different patterns of genetic inheritance assist in diagnosing and differentiating between key genetic disorders?

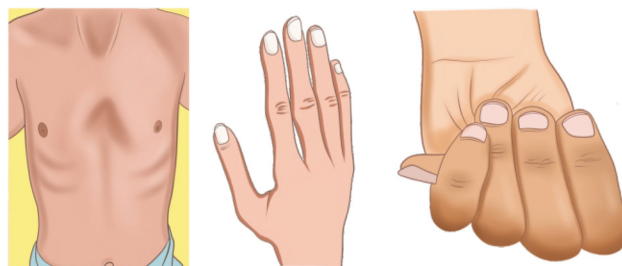
Broad Answer: Inheritance patterns reveal whether a disorder is dominant, recessive, X-linked, or mitochondrial, helping predict who in the family is at risk. For example, autosomal dominant diseases like Marfan syndrome show vertical transmission and affect both sexes. X-linked recessive diseases like Duchenne muscular dystrophy mostly affect males and skip generations. Disorders like Fragile X syndrome are X-linked dominant with anticipation, and mitochondrial disorders only pass through the maternal line. Pedigree analysis aids in clinical decision-making and genetic counselling.

DETAILED QUESTIONS

Q3.1 Explain Autosomal Dominant Inheritance

Answer:

- The disease will manifest even if one allele is abnormal.
- Equally common in boys and girls.
- Examples :
 - o Ehler-Danlos syndrome
 - o Osteogenesis imperfecta
 - o Marfan syndrome
 - o Intermittent Porphyrria
 - o Noonan syndrome
 - o Achondroplasia
 - o Neurofibromatosis
 - o Tuberous sclerosis
- Osteogenesis imperfecta- blue sclera, deafness, recurrent fractures
- Marfan syndrome: Fibrillin-1 gene is defective
 - o Tall stature, Pectus excavatum, Supero-Temporal Dislocation of lens, Arachnodactyly (spider-like fingers), Thumb sign positive
 - o Similar features are seen in Homocystinuria.
 - o A hypercoagulable state is seen in Homocystinuria and Inferonasal lens dislocation.



Q3.2 Give examples of Autosomal Recessive Inheritance

Answer:

- Alkaptonuria
- Beta Thalassemia
- Cystic fibrosis, Congenital Adrenal Hyperplasia
- Gaucher disease
- Homocystinuria
- Inborn errors of metabolism

Q3.3 Give examples of X-linked Recessive and X-linked Dominant

Answer:

X-linked Recessive	X-linked Dominant
• Glucose 6-phosphate dehydrogenase def • Duchenne Muscular Dystrophy • Colour blindness • Fabry disease • Chronic Granulomatous Disease • Haemophilia A & B • Agammaglobulinemia • Wiskott-Aldrich syndrome • Albinism • Lesch Nyhan syndrome	• Hypophosphatemic Rickets • Alport syndrome • Rett syndrome

Q3.4 What is Fragile X syndrome, and how is it inherited?

Answer:

- The FMR1 gene on the X chromosome
- Increase in CGG repeats > 200
- Anticipation → clinical features worsen with successive generations.
- Mothers are usually carriers.

Clinical features:

- Large head
- Large face
- Large ears
- Large jaw
- Large testes after puberty
- 10% of females have intellectual disability and premature ovarian insufficiency.

Q.3.5 What is genomic imprinting, and how does it relate to Prader-Willi and Angelman syndromes?

Answer:

Prader-Willi syndrome	Angelman syndrome
• The paternal copy of the gene is deleted • Infantile feeding problems, obesity, hypogonadism, and facial dysmorphism	• The maternal copy of the gene is deleted • Happy puppets, microcephaly, seizures

Q3.6 What inheritance pattern is suggested if all children of an affected mother are affected?

Answer: Mitochondrial inheritance

Q3.7 What pattern suggests autosomal dominant inheritance?

Answer: At least one parent of an affected individual is also affected

Q3.8 Which finding rules out X-linked inheritance in pedigree analysis?

Answer: Father-to-son transmission

Q3.9 Describe the pedigree analysis of Autosomal Dominant

Answer:

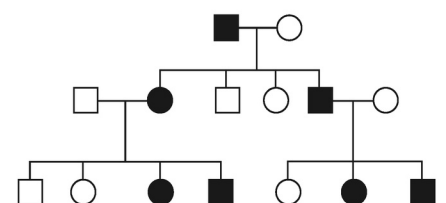
- Dominant type of inheritance (Male is affected)
 - o Father-to-son transmission
 - o Rules out the X-linked inheritance
- Inheritance- Autosomal dominant

Q3.10 Describe the pedigree analysis of Mitochondrial inheritance

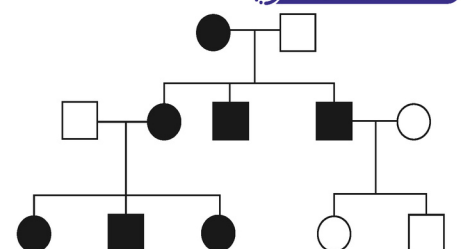
Answer:

- All the children of females are affected
 - o Due to heteroplasmy
- Mitochondrial inheritance pattern
 - o Kearns-Sayre syndrome
 - o Leigh disease
 - o MELAS

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🔍 PYQ: FMGE 2021

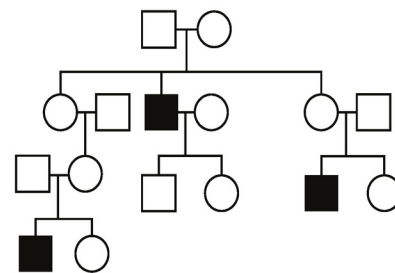


- o MERRF
- o NARP
- o Optic neuropathy

Q3.11 Describe the pedigree analysis of X-linked Recessive

Answer:

- Males are most commonly affected
- No father-to-son transmission
- Parents are not affected
- Recessive mode of inheritance
- X-linked recessive inheritance





5. INBORN ERRORS OF METABOLISM

BIG QUESTION 1: What are the important disorders of carbohydrate metabolism, and what are their types, clinical features, and significance?

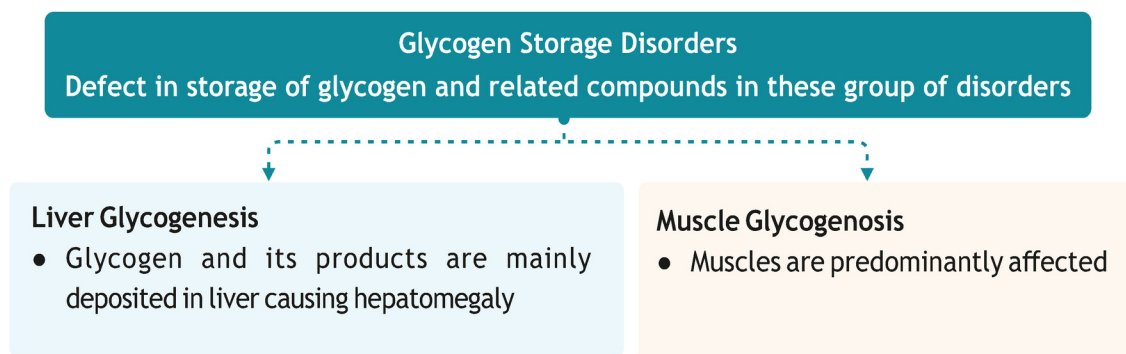
Broad Answer:

- Glycogen Storage Disorders (GSDs)
 - o Diseases due to defective liver glycogenesis
 - o Diseases due to defective muscle glycogenesis
- Galactosemia
- Hereditary fructose intolerance

DETAILED QUESTIONS

Q1.1: How are Glycogen Storage Disorders (GSDs) classified based on the main site of involvement?

Answer:



Q1.2: What are the important types of liver glycogen storage diseases and their enzyme defects?

Answer:

Type	Name of the Disease	Enzyme Deficient
I	Von Gierke disease (most common in younger age groups)	Glucose 6 Phosphatase
III	Cori disease	Debranching enzyme
IV	Anderson disease	Branching enzyme
VI	Her's disease	Hepatic phosphorylase

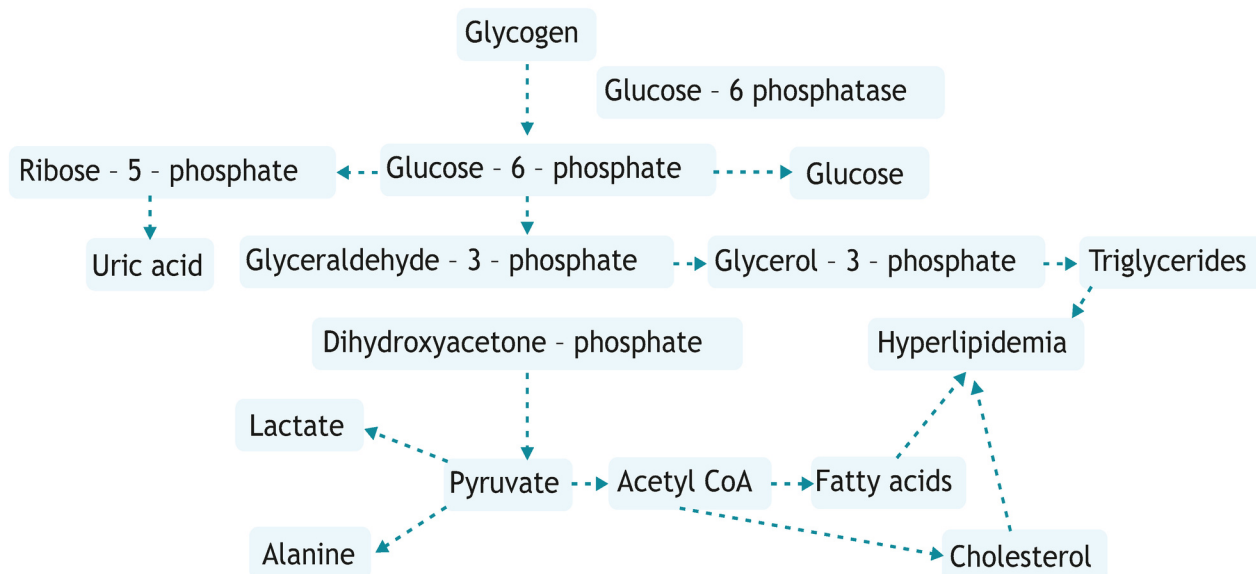
Q1.3: What is the most common glycogen storage disease in children, and its inheritance pattern?

Answer:

- Von Gierke Disease (Type I GSD) is the most common GSD among children
- Inheritance - Autosomal recessive

Q1.4: What are the metabolic pathways of Von Gierke disease?

Answer: Type I Glycogen Storage Disorder.



🔍 PYQ: FMGE 2021

🔍 PYQ: NEET PG 2021

Q1.5: What are the complications of Von Gierke disease?

Answer:

- Due to a deficiency of Glucose-6-Phosphatase, Glucose-6-Phosphate cannot be converted back into Glucose, leading to **Hypoglycemia**
- Glucose-6-Phosphate accumulated gets shunted to the Pentose Phosphate Pathway, causing excess Ribose 5-phosphate and elevated uric acid levels, causing **Hyperuricemia**
 - Hyperuricemia can cause renal shutdown, renal stones, and arthritis
- Excessive Glucose-6-P leads to increased levels of Glyceraldehyde-3-P & Dihydroxyacetone-P, causing the following:
 - Increased pyruvate levels and lactate levels cause elevated **Lactic Acid** levels and **Metabolic Acidosis**
 - Increased AcetylCoA leading to increased fatty acids and cholesterol, contributing to **Hyperlipidemia**
 - Increased levels of Glycerol3-P leading to elevated Triglycerides contributing to **Hyperlipidemia**

Q1.6: What are the clinical features of Von Gierke's Disease?

Answer:

- Present in infancy or early childhood
- Recurrent hypoglycemia, Early morning hypoglycemia (due to less feeding at night), leading to lethargy or seizures
- Hypertriglyceridemia
 - Doll-like facies: Chubby cheeks
- Abdominal distension (hepatomegaly)
- Uric acid stones in the kidney

Q1.7: What is the treatment of Von Gierke's Disease?

Answer:

- Corn starch diet
 - Give a slow, sustained source of glycogen or glucose to the body
 - Use uncooked corn starch, like corn flour, dissolved in water, which can be fed to the baby
- **Frequent feeding:** To prevent hypoglycemia

Q1.8: What are the important types of muscle glycogen storage diseases and their enzyme defects?**Answer:**

Type	Name of the Disease	Enzyme Deficient
II	Pompe disease	Acid maltase/ α -1,4-glucosidase
V	McArdle disease (The most common GSD present in adolescents)	Muscle phosphorylase
VII	Tarui disease	Phosphofructokinase

Q1.9: What are the clinical features and treatment of Pompe's disease?**Answer:**

- Clinical features
 - o Hypotonia (Skeletal muscle involvement)
 - o Cardiomegaly (Hypertrophic cardiomyopathy) - Elevated CPK level, ECG changes - Shortened P-R interval, Tall QRS complexes
 - o Coarse-looking facies
 - o Hepatomegaly
- Treatment: Enzyme replacement therapy

Q1.10: Which enzyme deficiencies are responsible for causing galactosemia? PYQ: FMGE 2021**Answer:**

- Galactose-1-Phosphate-Uridyltransferase (GALT): Classical-type
- Galactokinase
- Epimerase

Q1.11: Describe the pathophysiology of Galactosemia, highlighting the role of different enzyme deficiencies.**Answer:**

- Milk contains lactose, which is converted into glucose and galactose
- In Galactosemia, Galactose cannot be digested, and Galactose and its toxic metabolites accumulate in the body, causing various manifestations
- Even breastfeeding is contraindicated
- In **GALT deficiency**, there is an accumulation of Galactose 1-phosphate (toxic product) - Toxic to the liver, Eye, Kidney, and brain.
- In **Galactokinase deficiency**, there is an accumulation of galactose and galactitol (toxic to the eye)
 - o Congenital cataract is the most important manifestation of galactokinase deficiency.
 - o Galactokinase deficiency can present with bilateral cataracts as its sole manifestation.

Q1.12: What are the clinical features of Galactosemia?**Answer:**

- Hepatic involvement- Jaundice, Hepatomegaly
- GI manifestations- Diarrhoea, Vomiting
- Failure to thrive
- CNS manifestations- Seizures, Intellectual disability
- Bilateral congenital cataract (in galactokinase deficiency)
- Renal tubular manifestations
- Sepsis with E.coli (very common)

Q1.13: How is Galactosemia diagnosed?**Answer:**

- Urine test: Reducing substance in urine → Positive Benedict's test
- Enzyme assay: Direct measurement of GALT activity
- Genetic testing: Demonstration of genetic mutation (prenatal diagnosis possible)

Q1.14: What are the management modalities of Galactosemia?**Answer:**

- Lactose-free diet
- All milk and milk products are avoided
- Soya-based formulas can be given (lactose-free infant feeding formulas)
- Breastfeeding is contraindicated in confirmed cases of Galactosemia

Q1.15: Which enzyme deficiency causes Hereditary Fructose Intolerance, and how does it differ from Galactosemia?**Answer:**

- Enzyme Deficient: Aldolase B or Fructose 1,6-bisphosphate aldolase
 - Sugar → Sucrose → Glucose and fructose (inability to digest fructose)
- Similar to galactosemia, except that cataract is not seen
- Ingestion of Sweets can cause disease manifestation

Q1.16: How does Hereditary Fructose Intolerance present?**Answer:**

- Present beyond 6 months of age
- Aversion to sweet foods in children is due to their inability to digest sweet foods, which results in adverse manifestations:
 - Jaundice
 - Diarrhoea
 - Hepatomegaly
 - Failure to thrive
- Deposition of fructose 21-phosphate is toxic to the liver (mainly)

Q1.17: How is Hereditary Fructose Intolerance treated?**Answer:**

- Ensure a fructose-free diet
- Complete elimination of sweet food (sucrose and fructose) items

BIG QUESTION 2: What are the important disorders of amino acid metabolism, and what are their biochemical basis, clinical features, and significance?

Broad Answer: Disorders of amino acid metabolism are inherited or acquired conditions caused by enzyme defects affecting amino acid synthesis, degradation, or transport.

- Phenylketonuria
- Alkaptonuria
- Homocystinuria
- Multiple Carboxylase Deficiency
- Hartnup disease
- Maple syrup urine disease

These disorders lead to developmental delays, neurological deficits, or organ damage.

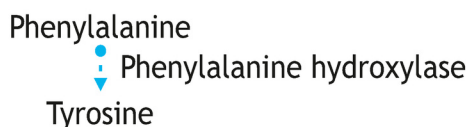
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DETAILED QUESTIONS

Q2.1: What is the etiopathogenesis of phenylketonuria?

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Answer:



- Caused due to the deficiency of **phenylalanine hydroxylase**, causing accumulation of phenylalanine in the body
- Accumulation of phenylalanine will cause brain damage, CNS manifestations, and is converted into metabolites like **phenyl acetate & phenylpyruvate**, which can be detected in urine and blood.
- Therefore, Tyrosine becomes an essential amino acid in phenylketonuria.

Q2.2: What are the clinical features of Phenylketonuria?

Answer:

- Hypopigmentation: Blond hair, fair skin, blue iris (because of no melanin)
- CNS manifestation: Intellectual disability, seizures, microcephaly, hypotonia, irritability
- Mousy/musty body odor
- Growth restriction
- Dental enamel changes

Q2.3: How is Phenylketonuria managed?

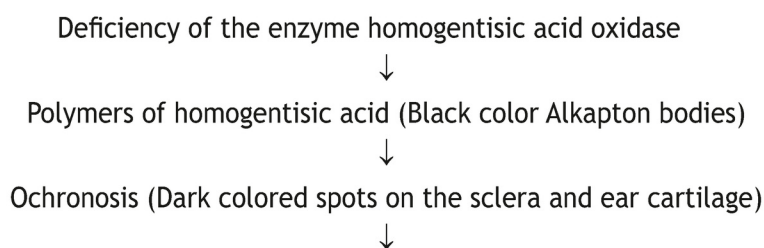
Answer: Low phenylalanine diet

Q2.4: What is the inheritance pattern of Alkaptonuria?

Answer: Alkaptonuria is an autosomal recessive disorder

Q2.5: What is the etiopathogenesis of Alkaptonuria?

Answer:



Excessive polymers of homogentisic acid are secreted in urine, which, on oxidation, give black- colored urine or darkening of urine on standing.

Q2.6: What is the clinical presentation of Alkaptonuria?

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Answer:

- Darkening of urine on standing, or the mother complains of a grey-coloured diaper (the only manifestation)
- Ochronosis: Dark spots on the sclera or ear cartilage
- Arthritis: Due to deposition of homogentisic acid polymer, usually involving the spine and large joints like the shoulder, knees
 - o **On X-ray:** Osteoarthritis-like changes are seen, including calcification of the intervertebral disc with occasional flares
- Cardiac involvement: Mitral or Aortic valvulitis

Q2.7: What are the causes of Homocystinuria?**Answer:**

- The classical type of homocystinuria is caused by a deficiency of **cystathionine β -synthase**.
- **Pyridoxine** is a cofactor for cystathionine β -synthase.

Q2.8: What are the clinical features of Homocystinuria?**Answer:**

- Failure to thrive
- Developmental delay, Intellectual disability
- Seizures, behavioral disorders
- Skeletal abnormalities resembling Marfan syndrome, like tall stature, arachnodactyly, pectus excavatum, and pectus carinatum
- Ectopia lentis/Subluxation of the lens of the eye in the inferomedial direction (in Marfan syndrome, the direction is superolateral)
- Recurrent episodes of stroke due to a hypercoagulable state

Q2.9: How is Homocystinuria treated?**Answer:**

- High doses of pyridoxine (vitamin B6) and folic acid
- Restrict methionine intake
- Cysteine supplementation

Q2.10: What is Multiple Carboxylase Deficiency, and how does it present clinically?**Answer:**

- Multiple Carboxylase Deficiency is an organic acidemia, with biotin is a cofactor
- Clinical features:
 - o Skin and hair changes are seen, like alopecia, skin rashes due to biotin involvement
 - o Global developmental delay and intellectual disability can occur
 - o There is a **Tomcat urine odor**
- Treatment: Biotin

Q2.11: What is the inheritance pattern of Hartnup disease?

Answer: Hartnup disease is an autosomal recessive disorder.

Q2.12: What is the genetic defect and pathogenesis of Hartnup disease?**Answer:**

- Defect in the **SLC6A19 gene on Chr.5**
- Defect in the transport of mono-amine, mono-carboxylic, or neutral amino acids by renal tubules and intestinal mucosa

Q2.13: What are the clinical features of Hartnup disease?**Answer:**

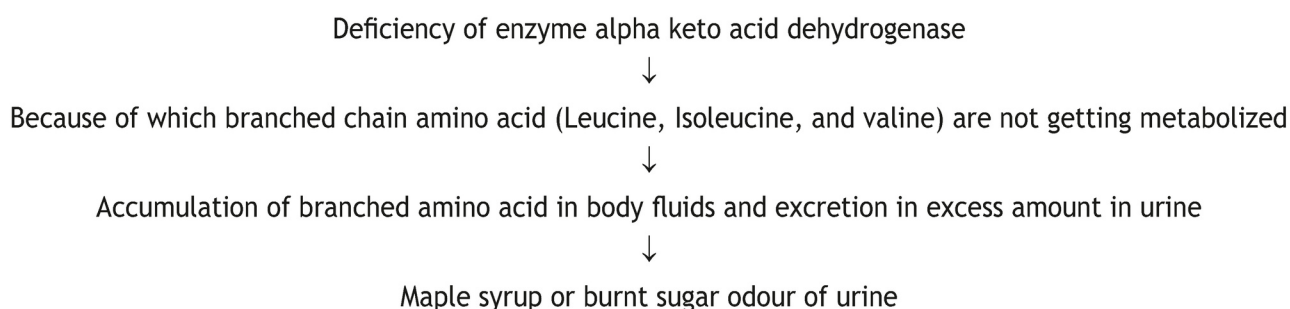
- Most remain asymptomatic
- Cutaneous photosensitivity and pellagra-like rash involving the neck area (Casals necklace) are seen in symptomatic cases.

Q2.14: What investigations are used in diagnosing Hartnup disease?**Answer:**

- Urine and plasma HPLC: Amino aciduria restricted to neutral amino acids like Valine, Leucine, Phenylalanine, Tyrosine, and Tryptophan
- Demonstrate the defect in the SLC6A19 gene.

Q2.15: How is Hartnup disease treated?**Answer:**

- Nicotinic acid or Niacin or Nicotinamide
- High protein diet

Q2.16: What enzyme deficiency causes Maple Syrup Urine Disease (MSUD), and what is its characteristic odor?**Answer:****Q2.17: What is the main clinical feature of patients with Maple syrup urine disease?**

Answer: Global developmental delay.

Q2.18: How is Maple Syrup Urine Disease treated?

Answer: Maple Syrup Urine Disease is treated by restricting the use of branched-chain amino acids.

Q2.19: What are the screening tests for inborn errors of metabolism?**Answer:**

- Tandem mass spectrometry- Dried blood spots
- Gas chromatography-mass spectroscopy- Urine
- High-performance liquid chromatography/ electrophoresis of plasma and urine

BIG QUESTION 3: What are lysosomal storage disorders, and what are their types, enzyme defects, clinical features, and significance?**Broad Answer:**

- LSDs are inherited metabolic disorders caused by a deficiency of specific lysosomal enzymes, leading to the accumulation of undigested macromolecules inside lysosomes.
- Some disorders include
 - o Gaucher disease
 - o Niemann-Pick disease
 - o GM₁ gangliosidosis
 - o Tay-Sachs disease

DETAILED QUESTIONS

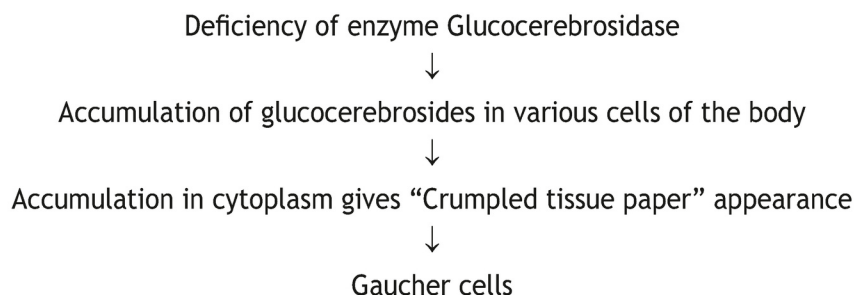
Q3.1: Which is the most common Lysosomal storage disorder seen in children?

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Answer: Gaucher disease

Q3.2: What is the pathogenesis of Gaucher disease?

Answer:



Q3.3: What are the clinical manifestations due to the accumulation of Gaucher cells?

Answer:

Gaucher cells accumulate in various parts of the body	Accumulation in spleen: Hepatosplenomegaly
	Accumulation in bone: Pancytopenia causing bone pains and pathological fractures.
	Accumulation in nervous system: Neurological involvement.

Q3.4: What are the clinical features of Gaucher's disease?

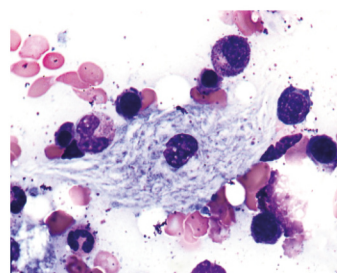
Answer:

- Some children may remain asymptomatic
- Hepatosplenomegaly: One of the causes of massive splenomegaly is that it can even cross the umbilicus.
- Pancytopenia
 - Anemia: easy fatigability, lassitude, lethargy
 - Leukopenia: Recurrent infections
 - Thrombocytopenia: Bleeding manifestations
- Bone pains and pathological fractures
- Neuronal involvement may or may not be present
 - **Neurological involvement is absent in type 1**
 - Types 2 & 3 can have some neurological involvement

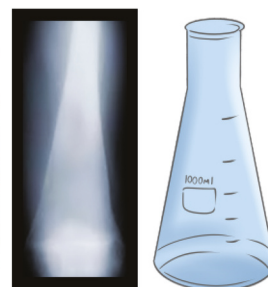
Q3.5: What are the diagnostic findings in Gaucher's disease?

Answer:

- Characteristic clinical presentation
- Bone marrow aspiration ± biopsy: Gaucher cells with characteristic "**crumpled tissue paper**" appearance of the cytoplasm
- Demonstrate the **deficient glucocerebrosidase enzyme** activity in the leucocytes or skin fibroblasts.
- X-ray of long bones: **Erlenmeyer flask deformity**



Gaucher cells



Erlenmeyer flask deformity

YOURWISH

Q3.6: How is Gaucher's disease managed?**Answer:**

- Enzyme replacement therapy is available
 - Can have a near-normal life, but it's expensive
- Supportive care
 - Regular blood transfusion
 - Treatment of infections
- Hematopoietic stem cell transplantation plays a potential role.

Q3.7: What are the diseases for which Enzyme Replacement therapy is available?

PYQ: INICET 2023

Answer:

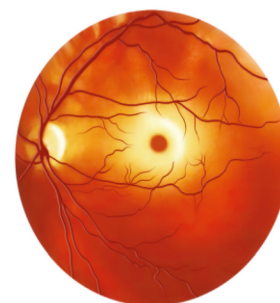
- Gaucher disease
- Type II glycogen storage disorder /Pompe disease
- Mucopolysaccharidosis Type I (Hurler disease), Type II (Hunter disease), and Type IV (Maroteaux-Lamy syndrome)
- Fabry disease
- Niemann-Pick disease (Recently available)
- Morquio disease

Q3.8: What is a cherry-red spot, and in which conditions is it typically observed?

PYQ: INICET 2022

Answer:

- Cherry Red Spot is a red spot in the macula
- Cherry red spot- Seen in inborn errors of metabolism
 - GM1 gangliosidosis
 - Niemann-Pick
 - Tay-Sach disease
- Not a usual feature of Gaucher disease

**BIG QUESTION 4: What are the different types of mucopolysaccharidoses, and what are their enzyme defects, clinical features, and management?**

Broad Answer: Mucopolysaccharidoses (MPS) are a group of inherited lysosomal storage disorders caused by deficiencies of enzymes that degrade glycosaminoglycans. They lead to progressive multisystem involvement, including skeletal abnormalities, organ enlargement, and developmental delays.

- Type I: Hurler disease
- Type II: Hunter disease
- Type III: Sanfilippo disease
- Type IV: Morquio disease
- Type VI: Maroteaux-Lamy disease

DETAILED QUESTIONS**Q4.1: Which enzyme deficiencies are responsible for the different types of mucopolysaccharidoses (MPS)?****Answer:**

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Type	Name	Enzyme deficient
I	Hurler disease (more severe)/Scheie disease (milder severe)	α -L-Iduronidase
II	Hunter disease	Iduronate 2-sulfatase enzyme

III	Sanfilippo disease	Heparin-S-Sulfamidase
IV	Morquio disease	N-acetyl-galactosamine Sulfatase
VI	Maroteaux-Lamy disease	Aryl Sulfatase B

Q4.2: What are the clinical manifestations of Hurler disease?

Answer:

- Coarse facies, i.e., puffy-looking facies, periorbital puffiness, depressed nose bridge, prominent philtrum, protruding tongue
- Intellectual disability/mental retardation
- Corneal clouding
- Hepatosplenomegaly
- Copious nasal discharge, airway problems
- Bony abnormalities: **Dysostosis multiplex** (proximal tapering of metacarpals or bullet-shaped metacarpals, anterior beaking of the vertebral body, J-shaped Sella turcica)
- Cardiac abnormalities

Q4.3: What are the clinical manifestations of other mucopolysaccharidoses?

Answer:

Disease	Clinical Features
Hunter disease	Same clinical features as Hurler disease, but there is no corneal opacity
San Filippo disease	Mainly presents with Intellectual disability/mental retardation, behavioral problems
Morquio disease	Bony abnormalities are most prominent
Maroteaux-Lamy disease	Same as Morquio disease + Coarse facies + Visceromegaly

Q4.4: What are the general inheritance patterns for Mucopolysaccharidoses (MPS) and lysosomal storage disorders?

Answer:

- All types of **MPS** have **autosomal recessive inheritance** except Type II MPS (**Hunter disease**), which has **X-linked recessive inheritance**.
- Most **lysosomal storage disorders** have **autosomal recessive inheritance**, except **Fabry's disease**, which has **X-linked recessive inheritance**.

Q4.5: What are the characteristic features of "Coarse facies"?

Answer:

- Puffiness around the eyes
- Protrusion of the tongue



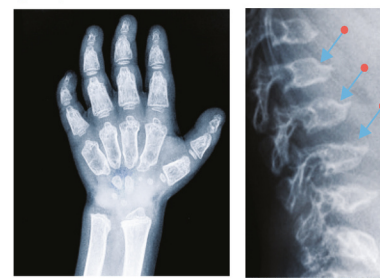
Q4.6: What is a distinguishing clinical feature of Hurler disease?

Answer: Hurler's disease presents with corneal opacity

Q4.7: What are the characteristic skeletal manifestations observed in Type IV Mucopolysaccharidosis?

Answer:

- Proximal ends of metacarpals- Tapering
 - Bullet or peg-shaped metacarpals
- Anterior beaking of vertebral bodies
- Skeletal manifestations seen in mucopolysaccharidosis- Dysostosis multiplex
 - Seen in type IV mucopolysaccharidosis



Bullet or peg-shaped metacarpals

Beaking

BIG QUESTION 5: What are the key features and characteristics of Menkes Disease?

PYQ: FMGE 2019

Broad Answer: Menkes Disease is a genetic disorder caused by a mutation in the ATP7A gene, leading to impaired copper metabolism. It is characterized by progressive cerebral degeneration, seizures, feeding difficulties, failure to thrive, and distinctive hair abnormalities, with a poor prognosis and often fatal outcome by early childhood.

Q5.1: What is the gene involved in Menkes disease, and what is its pathophysiology?

Answer:

- Menke disease is caused by a mutation in the **ATP7A gene**
- The ATP7A gene is a gene encoding copper-transporting ATPase
- **Pathophysiology:** Impaired copper metabolism and lower copper levels are found in the liver and brain, but high copper levels are found in fibroblasts and enterocytes

Q5.2: What are the clinical features of Menke disease?

Answer:

- Progressive cerebral degeneration or neurodegeneration
- Seizures
- Feeding difficulties
- Failure to thrive
- Hypothermia
- Apnea
- Characteristic hair abnormalities: **Woolly or Fuzzy/Kinky hair**

Q5.3: What are the microscopic findings in Menke disease?

Answer:

- On hair microscopy
 - **Trichorrhexis nodosa**, i.e., breakages in the hair shaft
 - **Pilli torti**, i.e., twisting of hairs at different places

Q5.4: What is the prognosis of Menke disease?

Answer: Prognosis is poor, and in the classical form of the disease, death usually occurs by 3 years of age.

BIG QUESTION 6: What are the key features, clinical manifestations, and management strategies for Wilson's Disease?

PYQ: NEET PG 2022

Broad Answer: Wilson's Disease is a genetic disorder caused by a defect in the ATP7B gene, leading to impaired copper metabolism and accumulation in various organs. It presents with liver disease, neurological symptoms, and Kayser-Fleischer rings in the eyes. Management involves chelation therapy with penicillamine or trientine, zinc supplementation, and a low-copper diet.

Q6.1: What are the genetic and biochemical defects associated with Wilson's disease, and what is its primary clinical manifestation?

Answer:

- Defect in the **ATP7B gene on chromosome 13q**
- Defective incorporation of Copper in Apo-ceruloplasmin
- Hepatolenticular degeneration

Q6.2: What are the clinical features of Wilson's disease?

Answer:

- Liver: Chronic/acute hepatitis
- Eye: KF ring (Kayser Fleischer ring), sunflower cataract
- Endocrine: Hypogonadism, Diabetes
- Neurology: Dystonia, gait abnormalities

Q6.3: What are the screening and confirmatory tests in the diagnosis of Wilson's disease?

Answer:

- **Best screening test**
 - o 24-hour urinary copper with or without d-penicillamine challenge
- **Confirmatory test**
 - o Estimation of copper in Liver Biopsy $>250 \mu\text{g/kg}$
 - o Genetic mutation diagnosis
 - o Screening of family members

Q6.4: What is the management of Wilson's disease?

Answer:

- Low copper diet: d-penicillamine, Trientine
- Chelators of copper
- Zinc supplementation

BIG QUESTION 7: What are the key features, clinical manifestations, and management strategies for Lesch-Nyhan syndrome?

Broad Answer: Lesch-Nyhan syndrome is a genetic disorder caused by a deficiency of the enzyme hypoxanthine-guanine phosphoribosyltransferase (HGPRT), leading to excessive uric acid production and neurological symptoms like developmental delay, dystonia, and self-injury. Management involves controlling uric acid levels with medications like allopurinol and addressing behavioral and developmental issues.

Q7.1: What is the basic defect and inheritance pattern of Lesch-Nyhan syndrome?

Answer:

- **Basic defect:**
 - o Deficiency of HGPRT (Hypoxanthine Guanine Phosphoribosyl Transferase) enzyme
 - o This enzyme helps in the recycling of the building blocks of DNA and RNA.
- **Inheritance:** X-linked recessive inheritance

YOURWISH

Q7.2: What are the clinical features and diagnostic tests of Lesch-Nyhan syndrome?**Answer:****Clinical features:**

- Asymptomatic at birth
- Subsequently, Developmental delay and other neurological features like dystonia, dysarthria, and spasticity
- Self-Injury/Self-Mutilation

Diagnosis:

- Increased level of uric acid
- Deficiency of the HGPRT enzyme is present

Q7.3: What are the treatment options for Lesch-Nyhan syndrome?**Answer :****Treatment:**

Due to high uric acid levels, use

- Allopurinol
- Alkalinization of urine
- High fluid intake



6. VASCULITIS DISORDERS IN CHILDREN/ IMMUNIZATION

BIG QUESTION 1: What are the features, diagnostic criteria, and management of Henoch-Schönlein purpura?

Broad Answer: Henoch-Schönlein purpura (HSP) is a small-vessel vasculitis characterized by palpable purpura, joint pain, abdominal pain, and renal involvement. It is primarily diagnosed clinically and managed with supportive care, corticosteroids, and treatment of complications.

DETAILED QUESTIONS

Q1.1: A child presents with abdominal pain, hematuria, and palpable purpura over the leg, as given in the image. His skin lesions show IgA deposition on biopsy. What could be the likely diagnosis?

Answer:

- Henoch-Schönlein Purpura (HSP)

🔍 PYQ: NEET PG 2019, 2021, 2022 🔍 PYQ: FMGE 2022



Q1.2: What are the diagnostic criteria for Henoch-Schönlein Purpura (HSP)?

Answer:

- Palpable purpura with the presence of 1 or more of:
 - o Diffuse abdominal pain
 - o Arthritis or arthralgia
 - o Any biopsy showing IgA deposition
 - o Renal involvement

Q1.3: How is Henoch-Schönlein Purpura (HSP) treated?

Answer:

- Most of the cases are self-limiting
- Severe cases - Corticosteroids

Q1.4: What renal complications can occur in Henoch-Schönlein purpura?

Answer:

- Glomerulonephritis is seen in 1/3 of patients
 - o Thrombocytopenia is Absent

BIG QUESTION 2: What are the clinical presentation, diagnostic criteria, investigations, and management of Kawasaki disease?

Broad Answer: Kawasaki disease is an acute vasculitis of childhood affecting medium-sized arteries, especially the coronary arteries, presenting with fever, rash, conjunctivitis, mucosal changes, and lymphadenopathy. Diagnosis is clinical, supported by laboratory and echocardiographic findings, and management includes intravenous immunoglobulin and aspirin to prevent cardiac complications.

DETAILED QUESTIONS

Q2.1: A 4-year-old child presents with fever and rash for 6 days, along with red eyes and tongue, and swelling of the hands. What could be the likely diagnosis?

Answer:

- Kawasaki disease

Q2.2: What are the diagnostic criteria for Kawasaki disease?

 PYQ: INICET 2023

Answer:

- Fever for >5 days with any 4 out of 5- “CREAM” features
 - o C- Conjunctivitis, B/L non-purulent
 - o R- Rash involving trunk
 - o E- Erythema & Edema of palms & soles along with desquamation
 - o A- Adenopathy (usually unilateral, cervical)
 - o M- Mucositis (Strawberry Tongue)



Conjunctivitis



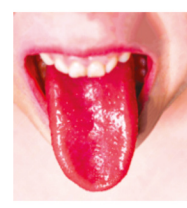
Polymorphism rash in the trunk



Edema and erythema in palm



Lymphadenopathy



Mucosal involvement in the tongue

Q2.3: What are the diagnostic findings in Kawasaki disease?

Answer:

- No single diagnostic test
- Leukocytosis, thrombocytosis, elevated ESR, or CRP
- Coronary artery aneurysm (**Giant >8mm in diameter**) is an important complication, but the incidence has decreased due to early diagnosis and treatment.

Q2.4: What are the treatment modalities of Kawasaki disease?

Answer:

- IVIg: Early treatment following diagnosis prevents complications
- Aspirin: High dose f/b low dose
- Corticosteroids if persistent fever despite IVIg

Q2.5: What is the major complication of Kawasaki disease?

Answer:

- Coronary artery aneurysm

BIG QUESTION 3: What are the causative agent, pathogenesis, clinical features, complications, and management of measles?

Broad Answer:

- Measles is a highly contagious viral infection caused by the Measles virus, a member of the genus **Morbillivirus**, family **Paramyxoviridae**. Transmitted via respiratory droplets and airborne spread.

DETAILED QUESTIONS

Q3.1: What is the incubation period of Measles?

Answer:

- Incubation period: 8-10 days
- Remains communicable from 3 days before to 4-6 days after the onset of rash

Q3.2: What are the receptors for the Measles virus?

Answer:

- CD 150
- PVRL 4

Q3.3: What is the pathognomic feature of Measles?

Answer:

- Presence of multinucleated **Warthin-Finkeldey giant cells**

Q3.4: What are the clinical features of Measles infection?

Answer:

- **Prodromal phase** (Child has a wet kind of look): Fever, conjunctivitis, coryza
- **Koplik spot** (Reddish spots with a white centre): It is present on the inner side of the cheek on the buccal mucosa
 - It can also be present in the conjunctiva and vaginal mucosa
- A rash of measles typically begins from behind the ear and then spreads to the rest of the body
 - Rash is an erythematous maculopapular rash that is confluent in places
 - Rash usually appears on day 4 of fever, and the rash fades in 7 days in the same progression as it appears, leaving behind a brownish discolouration

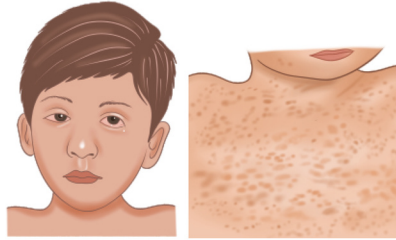
Q3.5: What are the complications of Measles?

Answer:

- **The most common complication:** Otitis media
- **The most common cause of death:** Pneumonia
 - After measles, the cell-mediated immunity of a child decreases, and the child is prone to infections like respiratory infections, pneumonia, and diarrhoea.
 - So, **Vitamin A** is often used in the treatment of measles, as it helps in mucosal immunity.
- Rare, fatal, long-term complication: **Subacute Sclerosing Pan Encephalitis**

Q3.6: An 18-month-old unimmunised 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?

YOURWISH

**Answer:**

- Vitamin A

Q3.7: What is Subacute Sclerosing Panencephalitis (SSPE), and how does it present clinically?**Answer:**

- SSPE is a rare, fatal, long-term complication of measles that develops **7-10 years** after 1^o measles infection
- Initially, it is characterized by decreasing school performance, then there is the appearance of myoclonic jerks, sudden falls, choreoathetosis, dystonia, and rigidity.
- Neurodegenerative disorder with a progressive downhill course
- No good treatment is available to date
- **Diagnosis:** CSF and Blood anti-measles antibody is done

BIG QUESTION 4: What is Varicella, and what are its key clinical features?**Broad Answer:**

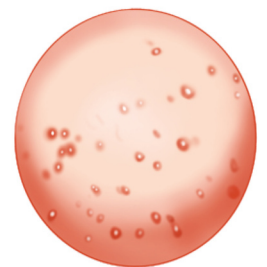
- Acute, highly contagious viral infection caused by **Varicella-Zoster Virus (VZV)**, a Herpesvirus
- Characterized by a generalized vesicular rash and fever
- Primary infection → Varicella (chickenpox)
- Reactivation later in life → Herpes Zoster (shingles)

DETAILED QUESTIONS**Q4.1: What is the incubation period of Varicella?****Answer:**

- Incubation period: 10-21 days

Q4.2: What type of rash is seen in Varicella (chickenpox), and how long does the child remain infectious?**Answer:**

- Characteristics of the Rashes
 - o Macules- Not raised
 - o Papules- Raised
 - o Vesicles- Fluid-filled
 - o Pustules- Pus-filled
- Hence, the Rash is pleomorphic
- Pleomorphic skin Rash + fever: Indications of chicken pox or varicella infection
- A child with varicella remains infectious till all vesicles have crusted.

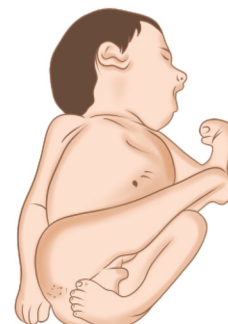
**Q4.3: What is the prevalence of Congenital varicella?****Answer:**

- Seen in 0.4% of babies of mothers who get infected in 1st trimester
- 2% of those are infected in the 2nd trimester

Q4.4: What are the clinical features of Congenital varicella?

Answer:

- **Cicatricial scarring** in zoster-like or dermatomal distribution around the umbilicus
- Limb hypoplasia
- Low birth weight
- CNS involvement: **Microcephaly**, developmental delay, intellectual disability, seizures
- Eye involvement: **Chorioretinitis**, **cataract**, microphthalmia (small eye)
- Renal involvement: Hydronephrosis
- Autonomic dysfunction: Swallowing dysfunction, neurogenic bladder



Q4.5: How is congenital varicella infection diagnosed?

Answer:

- Mother with a history of varicella infection during pregnancy
- Suggestive clinical features in a baby
- Demonstrate Anti-varicella IgM in a baby

BIG QUESTION 5: What are the causative agent, clinical features, and pregnancy-related complications of rubella?

Broad Answer:

- Rubella is a mild exanthematous viral infection that is usually self-limiting; however, it is of greatest concern in pregnancy due to the risk of Congenital Rubella Syndrome (CRS).
- Transmitted via respiratory droplets.

DETAILED QUESTIONS

Q5.1: What is the causative agent of Rubella?

Answer:

- Rubella virus (RNA) belongs to the Togaviridae family.

Q5.2: What is the incubation period of Rubella infection?

Answer:

- Incubation period: 14-21 days
- Becomes communicable from 5 days before to 6 days after the onset of rash

Q5.3: What are the clinical features of Rubella infection?

Answer:

- Prodrôme phase: Low-grade fever, sore throat, malaise, and headache
- Rash begins on the face and spreads centrifugally- **measles-like rash**
- **Forcheimer spots** on the soft palate and uvula
- Posterior auricular lymphadenopathy
- Rubella is a mild and self-limiting illness that typically resolves within 3-5 days.

Q5.4: During which period of pregnancy is the risk of transmission and frequency of congenital Rubella infection highest?

Answer:

- The risk of transmission of infection and the frequency of congenital infection are highest before 11 weeks of gestation.

Q5.5: What are the clinical features of Congenital Rubella?

 PYQ: FMGE 2020

Answer:

Clinical features	Other clinical features
• Congenital bilateral cataract • Deafness (Sensorineural hearing loss) • Congenital heart diseases ◦ Most common: Patent Ductus Arteriosus ◦ Least common: Atrial Septal Defect ◦ Other like VSD, pulmonary stenosis	• Microcephaly, IUGR • Glaucoma • Salt and pepper retinopathy • Hepatosplenomegaly • Jaundice • Thrombocytopenia • Blueberry muffin lesion: Bluish red nodular lesions is a characteristic of congenital rubella

BIG QUESTION 6: What are the causative agent, features, and transmission of Congenital CMV infection?

Broad Answer:

- Caused by Cytomegalovirus, a Herpesvirus (HHV-5)
- **Transmission:** Transplacental (most important), Perinatal (breast milk, vaginal secretions), and Postnatal (blood transfusions, organ transplantation)

DETAILED QUESTIONS

Q6.1: Why is routine screening for congenital CMV infection not recommended, and what is the risk of transmission in primary vs. secondary maternal infection?

Answer:

- 90% of infected babies with CMV (cytomegalovirus) are asymptomatic - No use of routine screening
- Risk of transmission during pregnancy
 - 1° CMV infection → 30%
 - 2° CMV infection → 1-2% (Developing countries like India)

Q6.2: What are the clinical features of symptomatic Congenital CMV infection?

Answer:

- Microcephaly
- Chorioretinitis
- Hepatosplenomegaly
- Jaundice
- Petechiae
- Periventricular calcifications
- The most important long-term sequelae of congenital CMV infection: **Sensorineural hearing loss**
- Congenital CMV infection is one of the most common nonsyndromic causes of hearing loss.

Q6.3: How is congenital CMV infection diagnosed, and what is the best sample for testing?**Answer:**

- Demonstrate replicating the virus in any of the samples from the baby within the first 3 weeks.
 - PCR/Viral culture: Best sample- **urine sample**
 - It can be done using a saliva or blood sample
- CMV IgM: It can also provide some indirect clue about whether the baby is infected or not.

Q6.4: What is the drug of choice in treating symptomatic congenital CMV infection?**Answer:**

- Ganciclovir

BIG QUESTION 7: What are the modes of transmission, clinical features, prevention, and management of congenital toxoplasmosis?**Broad Answer:**

- Congenital toxoplasmosis is seen in a baby if a mother acquires primary toxoplasma infection during pregnancy.
- The severity of fetal infection is greater if the fetus is affected earlier in pregnancy.

DETAILED QUESTIONS**Q7.1: What is the clinical presentation of Congenital toxoplasmosis?****Answer:**

Clinical features	Other features
• Chorioretinitis • Hydrocephalus • Cerebral calcification (detection on Non contrast CT head)	• Macrocephaly due to hydrocephalus • Small for gestational age • Prematurity • Hydrops fetalis, i.e., generalized anasarca • Persistence jaundice • Thrombocytopenia

Q7.2: Which group of organisms is most commonly responsible for causing congenital infections in babies?**Answer:**

- TORCH group of organisms

Q7.3: What are the key measures for prevention and treatment of Congenital toxoplasmosis?**Answer:**

- Early diagnosis of infection and treatment of the mother treatment of mothers is by Sulphadiazine.

BIG QUESTION 8: What is erythema infectiosum? What is its causative agent, and what are its characteristic features and complications?**Broad Answer:**

- Erythema infectiosum (fifth disease) is a childhood viral illness caused by Parvovirus B19, spread mainly by respiratory droplets. It presents with a “slapped-cheek” rash followed by a lacy, reticular rash on the body. The illness is usually mild, but it can cause aplastic crisis in hemolytic anemias and hydrops fetalis in pregnancy.

YOURWISH

DETAILED QUESTIONS

Q8.1: What is the incubation period of Erythema infectiosum?

Answer:

- Incubation period: 4-28 days

Q8.2: What are the clinical features of Erythema infectiosum?

Answer:

- Low-grade fever, headache, malaise, upper respiratory tract infection
- Characteristic feature: "Slapped cheek" appearance of the rash



Q8.3: What are the other diseases caused by Parvovirus?

Answer:

- Transient aplastic crisis: Can be caused spontaneously or in some children with underlying diseases like sickle cell disease
- Papular purpural gloves and socks syndrome: There is an involvement of the distal part of the extremities
- Pure red cell aplasia

BIG QUESTION 9: What are the clinical presentation, complications, and management of mumps?

Broad Answer:

- Mumps is a viral infection caused by the mumps virus, presenting with fever, malaise, and painful parotid gland swelling. Complications include orchitis, meningitis, and pancreatitis, and management is mainly supportive.

DETAILED QUESTIONS

Q9.1: 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 the child in the same village. What will be the likely diagnosis?

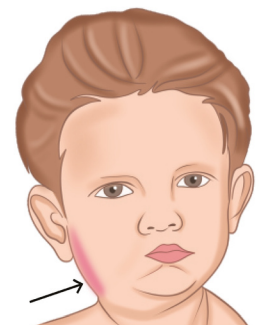
Answer:

- Mumps- It is a very infectious viral infection; hence, various children in the locality could suffer from it

Q9.2: What is the characteristic presentation of Mumps?

Answer:

- In children, an acute onset with unilateral or bilateral parotid swelling along with fever
- In adolescent males with mumps, orchitis may be seen.



Q9.3: What is the most common complication of Mumps?

Answer:

- Aseptic meningitis

Q9.4: How is Mumps treated?

Answer:

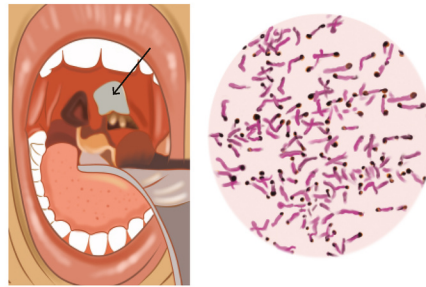
- Mumps is a self-limiting illness, and no specific treatment is required.

BIG QUESTION 10: What are the causative agents, clinical features, complications, management, and prevention of diphtheria?

Broad Answer: Diphtheria is an acute bacterial infection caused by *Corynebacterium diphtheriae*, producing a toxin that leads to pharyngeal pseudomembrane, sore throat, and lymphadenopathy. Complications include myocarditis and neuropathy, and prevention is through DPT vaccination with treatment by antitoxin and antibiotics.

DETAILED QUESTIONS

Q10.1: An unimmunized 8-year-old girl presented with fever, sore throat, and swelling of the neck. The examination of the throat revealed the 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?



- Diphtheria

Q10.2: What is the causative agent for Diphtheria?

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Answer:

- *Corynebacterium diphtheria*

Q10.3: What is the incubation period of Diphtheria?

Answer:

- Incubation period: 2-4 days

Q10.4: What are the clinical features of Diphtheria?

Answer:

- H/o unimmunized child
- Universal early symptom: **Sore throat**
- Fever (only in 50%), malaise, headache
- Dysphagia, hoarseness
- Stridor & cough
- Bull neck appearance
- Drooling of saliva

Q10.5: What are the findings observed in the throat of a diphtheria-affected patient?

Answer:

- **Pseudomembrane** is observed on throat examination
 - o Removal causes bleeding
 - o Can extend to the tonsils, uvula, soft palate, and posterior oropharynx wall

Q10.6: What is the treatment of diphtheria, and how should the contacts of an affected child be managed?**Answer:**

Treatment	• Supportive care • Mainstay of therapy- Anti-diphtheria serum • Antibiotic- Azithromycin/ 3rd generation Cephalosporins
Management of Contacts of a Child	Isolation and monitoring • All the household and intimate contacts must be isolated and closely monitored for at least 7 days. Chemoprophylaxis • Should be given regardless of the immunization status of contacts Vaccination • Given to all individuals who have not received a booster dose in the last 5 years (< 7 years: DPT and > 7 years: Tdap)

Q10.7: What are the complications of Diphtheria?**Answer:**

- Early - Myocarditis, Airway obstruction
- Long-term- Neurological (Multiple cranial nerve palsy, Quadriplegia)

BIG QUESTION 11: What are the features, diagnosis, and prophylaxis of pediatric tuberculosis?**Broad Answer:**

- Paediatric Tuberculosis (TB) in children caused by the bacterium *Mycobacterium tuberculosis* primarily affects the lungs but can spread to other parts of the body, including the lymph nodes, bones, and brain. Due to their developing immune systems, children are particularly vulnerable to severe forms of TB. Diagnosis can be challenging in children, as symptoms are often nonspecific and the disease can be difficult to confirm microbiologically.

DETAILED QUESTIONS**Q11.1: Where is Ghon's focus located in primary tuberculosis?****Answer:**

- Childhood TB- Lungs (Ghon's focus + Hilar LN- Ghons complex)
- Congenital TB- Liver

Q11.2: What are the main diagnostic methods for confirming tuberculosis in children?**Answer:**

- Demonstrate bacteriological evidence for TB from the early morning Gastric aspirate.
- CBNAAT (cartridge-based nucleic acid amplification test)
- Microscopy with ZN stain

Q11.3: What is the treatment approach for pediatric tuberculosis?**Answer:**

- Daily treatment is given to all pediatric patients
- Only pyrazinamide is stopped

Q11.4: What is the recommended prophylaxis for pediatric tuberculosis?**Answer:**

INH prophylaxis	- Recommended for all children less than 5 years of age who have H/o contact with an active TB patient in the last 2 years should be evaluated for TB. - If positive, ATT is started. - If negative, INH prophylaxis 10 mg/kg/day
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BIG QUESTION 12: What is the National Immunization Schedule (NIS) of India, and what does it aim to provide?**Broad Answer:**

- The National Immunization Schedule (NIS) of India provides free vaccination to all children and pregnant women. It includes vaccines against major diseases such as tuberculosis, polio, diphtheria, pertussis, tetanus, measles, hepatitis B, and rotavirus, given at specific ages to ensure maximum protection and reduce childhood morbidity and mortality.

DETAILED QUESTIONS**Q12.1: What is the latest National Immunization Schedule (NIS) of India?****Answer:**

At birth	OPV-0, BCG, Hepatitis B birth dose
6 weeks	Pentavalent-1, OPV-1, Rota-1, f-IPV, PCV-1
10 weeks	Pentavalent-2, OPV-2, Rota-2
14 weeks	Pentavalent-3, OPV-3, Rota -3, f-IPV-2, PCV- 2
9 months	MR-1, PCV booster, JE-1, f - IPV -3, Vitamin A- 1 st megadose
16-24 months	MR-2, DPT-booster-1, OPV-booster, JE-2, Vitamin A- 2 nd megadose
2 years	Typhoid vaccine (not given in all states)
5-6 years	DPT booster - 2
10-16 years	Td

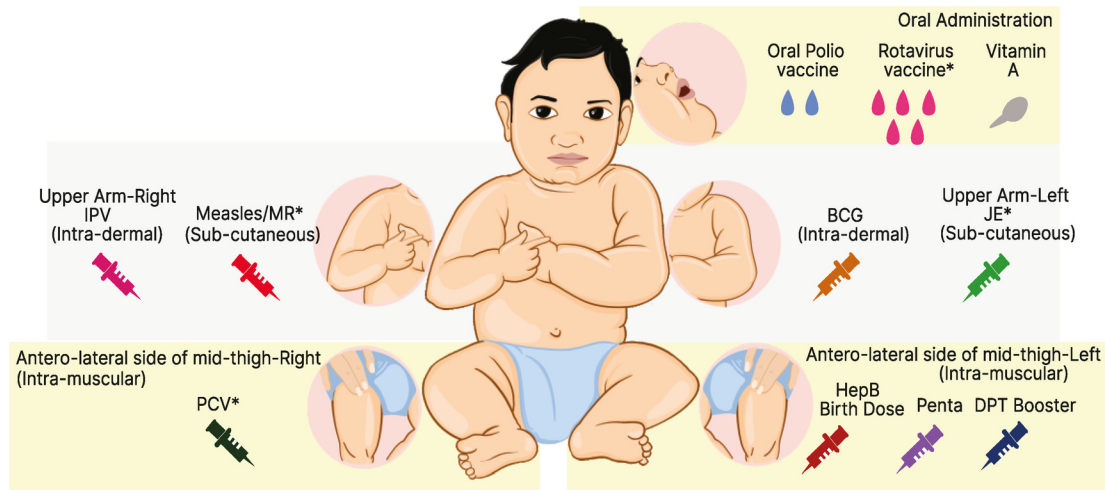
Q12.2: What is the latest recommendation for administering the Hepatitis B vaccine?**Answer:**

- Hepatitis B is now recommended to be given along with Vitamin K in the delivery room immediately after birth.

Q12.3: What does the pentavalent vaccine contain?**Answer:**

- Pentavalent vaccine - A Combination of 5 vaccines
 - DPT + Hep-B + Hib

Q12.4: What are the recommended injection sites and routes of administration for different vaccines in the National Immunization Schedule?**Answer:**



Oral	• Polio vaccine • Rotavirus vaccine • Vitamin A
Left upper arm	• BCG (intradermal) • Japanese encephalitis (Subcutaneous)
Anterolateral side of thigh - Left	• Hepatitis B (At birth) • Pentavalent • DPT booster
Right upper arm	• IPV (Intradermal) • Measles or MR (Subcutaneous)
Anterolateral side of thigh - Right	• PCV vaccine - o Given at: 6weeks, 14weeks & 9months

Q12.5: What is the recommended approach when multiple vaccines need to be administered to a child during the same visit?

Answer:

- When more vaccines are to be given at the same visit, start with the least painful one.

Q12.6: What is the recommended catch-up vaccination schedule for an unvaccinated child at their first visit?

Answer:

- Catch-up vaccination in an unvaccinated child, at the 1st visit
 - o BCG- <1 year age
 - o Penta valent vaccine: <1 year
 - o OPV- <5 years age
 - o DPT- <7 years age
 - o Tdap (for 1st dose) - >7 yrs
 - o Td, Td (for 2nd & 3rd dose)
 - o MMR do 6-monthly intervals
 - o Hib:
 - 6-12m- 2 doses + 1B
 - 12-15m- 1 dose +1B
 - >15m - 1 dose

Q12.7: What are the important adverse effects associated with common childhood vaccines?**Answer:**

Vaccines	Adverse effects
BCG	Osteitis, BCG lymphadenitis, Disseminated BCG
DPT	Persistent cry, HHE (Hypotonic hyporesponsive episodes)
MMR	Thrombocytopenia, Mild measles-like illness
OPV	VAPP, contraindicated in immunodeficiency
Rotavirus	Intussusception

Q12.8: 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 immunizations according to schedule
- B. Siblings should not be given OPV
- C. Give killed vaccines
- D. Pneumococcal vaccine should ideally be given before commencing the treatment

Answer:

- A. Give all immunizations according to schedule
- Live vaccines (e.g., OPV, MMR, Varicella) are contraindicated during steroid therapy.

Q12.9: Which vaccines can only be given beyond 2 years of age?**Answer:**

- Polysaccharide vaccine

Q12.10: Which vaccines should be given beyond 9 years of age?**Answer:**

- HPV vaccine

Q12.11: Which are the vaccines that can be given to adolescents?**Answer:**

- TT, Tdap, Td
- Hep B
- Influenza
- JE and Rabies

Q12.12: Which vaccines are contraindicated in children?**Answer:**

- Influenza vaccine
- Yellow fever vaccine

Q12.13: Which vaccine causes thrombocytopenia?**Answer:**

- Measles vaccine

Q12.14: Which vaccine acts faster than the incubation period of the disease it prevents?**Answer:**

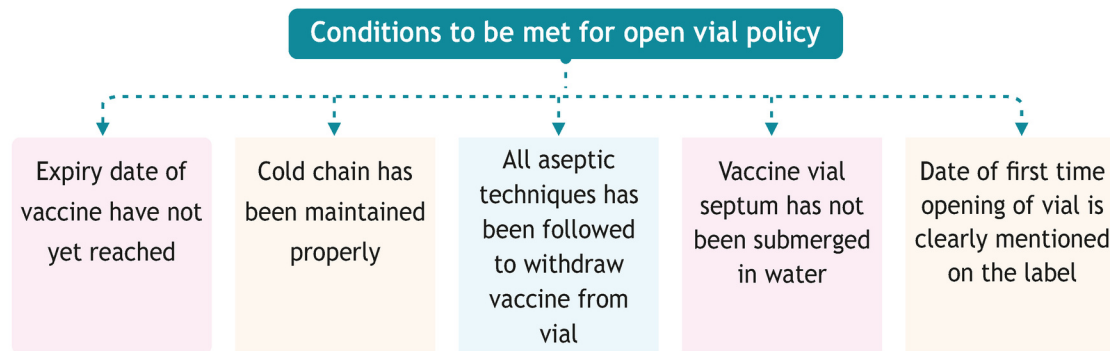
- Varicella vaccine

BIG QUESTION 13: What is the Open Vial Policy, and what is its primary goal in immunization programs?**Broad Answer:**

- The Open Vial Policy refers to guidelines for the safe and effective use of multi-dose vaccine vials once they have been opened and reconstituted (if necessary). The primary goal of this policy is to reduce vaccine wastage while ensuring the potency and sterility of the vaccine. Certain vaccine vials can be reused for up to 4 weeks, provided certain conditions are met.

DETAILED QUESTIONS**Q13.1: What are the conditions to be met for the open vial policy?**

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Answer:**Q13.2: Which vaccines are covered under the Open Vial Policy, and which are not?****Answer:**

Applicable for: PPT H	Not applicable for: BMR J
• Polio vaccine • PCV • Tetanus containing vaccine - o Pentavalent - o DPT - o TT - o Td • Hepatitis B	• BCG • Measles/Measles containing vaccine (MR) • Rotavirus (some types) • JE (some types)

Q13.3: What is the vaccine vial monitor?

Answer: A vaccine vial monitor is a heat-sensitive label that indicates the cumulative heat exposure over time to that vaccine vial.

Q13.4: How is the Vaccine Vial Monitor (VVM) interpreted to decide whether a vaccine should be used or discarded?

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Answer:

	Inner square lighter than outer ring. If the expiry date has not been passed, USE the vaccine.
	At a later time, inner square still lighter than outer ring. If the expiry date has not been passed, USE the vaccine.
	Discard point: Inner square matches colour of outer ring. DO NOT use the vaccine.
	Beyond the discard point: Inner square darker than outer ring. DO NOT use the vaccine.

Q13.5: What is the shake test, and how is it used to detect damage in vaccines?

Answer:

- Test done to check for cold damage to the vaccine due to freezing
 - o Shake the vial and let it stand for some time
 - o If the sediments settle down, do not use the vaccine



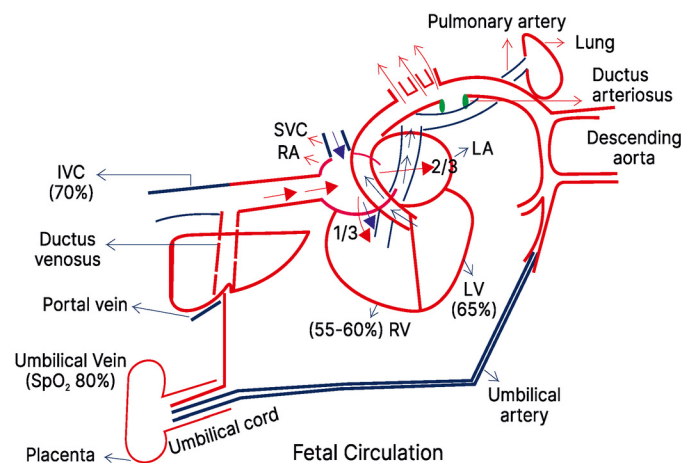
7. PEDIATRIC CARDIOLOGY

BIG QUESTION 1: Describe the key components and characteristics of fetal circulation

Broad Answer:

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- The placenta characterizes fetal circulation as the primary source of oxygen for the fetus.
- The umbilical cord contains **one umbilical vein and two umbilical arteries**.
- Blood flows through: ductus venosus, foramen ovale, and ductus arteriosus
- At birth, the lungs become the oxygen source, leading to decreased pulmonary vascular resistance and increased systemic vascular resistance.
- This triggers the closure of fetal shunts: the foramen ovale closes functionally immediately; the ductus venosus forms the ligamentum venosum; and the ductus arteriosus closes functionally at 10-15 hours.



DETAILED QUESTIONS

Q1.1: What is the primary source of oxygen in fetal circulation?

Answer:

- Placenta.

Q1.2: What are the components found within the umbilical cord?

Answer:

- 1 vein & 2 arteries.

Q1.3: Which vessel in the umbilical cord carries oxygenated blood, and what is its typical oxygen saturation?

Answer:

- The umbilical vein carries oxygenated blood (SpO₂ - 80%).

Q1.4: What is the clinical significance of a single umbilical artery?

Answer:

- It occurs in 5-10/1000 live births and is associated with an **increased risk of a renal anomaly** in the baby.

Q1.5: What are the structures in fetal circulation in decreasing order of oxygen saturation?

Answer:

- Umbilical vein (80% - Maximum) > Ductus venosus > Inferior vena cava (IVC: 70%) > LV (65%) > RV (55-60%) > Umbilical artery.
- The Superior vena cava has the lowest O₂ concentration.

Q1.6: What becomes the source of oxygen at or after birth, replacing the placenta?**Answer:**

- Lungs.

Q1.7: What two significant vascular resistance changes occur at birth, and what is their effect on pulmonary blood flow?**Answer:**

- Pulmonary vascular resistance decreases, leading to increased blood flow to the lungs (which can produce a systolic murmur).
- Systemic vascular resistance increases.

Q1.8: What are the main fetal shunts that close at or after birth?**Answer:**

- Foramen ovale, Ductus venosus, Ductus arteriosus.

Q1.9: What does the ductus venosus become after closure?**Answer:**

- Ligamentum venosus.

Q1.10: What is the postnatal remnant of the ductus arteriosus, and which substance is primarily responsible for maintaining its patency in the fetal circulation?**Answer:**

- After closure, the ductus arteriosus becomes the ligamentum arteriosum.
- Prostaglandins, particularly **Prostaglandin E₂ (PGE₂)**, play a key role in maintaining ductal patency during fetal life.

Q1.11: What is the difference between the functional and anatomical closure of the ductus arteriosus?**Answer:**

- **Functional closure:** Immediately after birth, at 10-15 hours (due to smooth muscle contraction).
- **Anatomical closure:** Day 10 - Day 21, up to 3 months of life (due to proliferation of cells of the intima of the ductus arteriosus).

Q1.12: What type of medications are used to maintain a patent ductus arteriosus?**Answer:**

- PGE₁ analogues / Alprostadil.

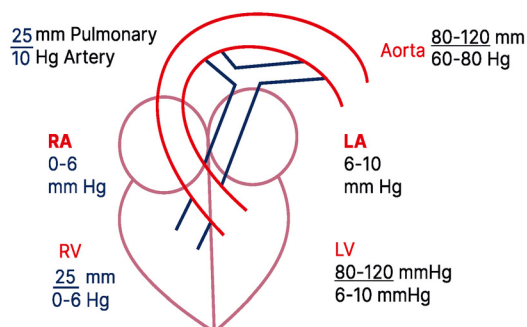
Q1.13: By what age do the pressures in different heart chambers normally become established?**Answer:**

- By 2-3 weeks of age.

Q1.14: In the presence of congenital heart diseases like VSD or PDA, how is the timeline for the establishment of normal cardiac chamber pressures affected?

Answer:

- In the presence of heart diseases (VSD or PDA), these pressures get established later, by 6-10 weeks of age.



BIG QUESTION 2: How are the congenital heart diseases classified, and what are their features?

Broad Answer:

- Nada's Criteria** are used to assess the possibility of congenital heart disease, requiring 1 major or 2 minor criteria for suspicion.
- Congenital heart diseases are broadly classified by their hemodynamics:

Classification of Congenital Heart Diseases				
L → R shunt	Complete mixing of blood of L & R side	Parallel Circulation	Ductus Arteriosus Dependent Congenital Heart disease	
- VSD - ASD - PDA (Note: R to L Eisenmenger syndrome)	- Truncus arteriosus - TAPVC (Total anomalous pulmonary venous connection)	TGA (transposition of great arteries)	Systemic circulation - Critical AS - Severe coarctation of aorta - Interrupted aortic arch - HLHS (Hypoplastic Left Heart Syndrome) (CHD MC causes mortality in 1st week of life)	Pulmonary circulation - Severe PS - Severe TOF - Pulm. atresia - Tricuspid atresia - Ebstein anomaly

DETAILED QUESTIONS

Q2.1: What are the major and minor criteria in Nada's Criteria?

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Answer:

- 1 major or 2 minor criteria indicate the possibility of congenital heart disease.

Major criteria	Minor criteria
- Systolic murmur $\geq$ Grade 3 - Any Diastolic murmur - Central Cyanosis - Congestive heart failure	- Systolic murmur $<$ Grade 2 - Abnormal S_2 heart sound - Abnormal BP - Abnormal ECG - Abnormal Chest X-ray

Q2.2: What are the three main types of Left to Right (L → R) shunt congenital heart diseases?**Answer:**

- Ventricular Septal Defect, Atrial Septal Defect, Patent Ductus Arteriosus

Q2.3: Which condition represents a reversal of shunt in L → R shunt diseases?**Answer:**

- Eisenmenger syndrome (R to L shunt).

Q2.4: Which CHDs are characterized by complete mixing of blood from the left and right sides of the heart?**Answer:**

- Truncus arteriosus, TAPVC (Total anomalous pulmonary venous connection).

Q2.5: Which congenital heart disease is characterized by parallel circulation?**Answer:**

- TGA (transposition of great arteries).

Q2.6: What is the significance of "Ductus Arteriosus Dependent Congenital Heart disease"?**Answer:**

- These conditions require a patent ductus arteriosus for either systemic or pulmonary blood flow and survival.

Q2.7: What are the congenital heart diseases that are "Ductus Arteriosus Dependent" for systemic circulation?**Answer:**

- Critical AS
- Severe coarctation of the aorta
- Interrupted aortic arch
- HLHS (Hypoplastic Left Heart Syndrome).

Q2.8: Which congenital heart disease is the most common cause of mortality in the 1st week of life?**Answer:**

- HLHS (Hypoplastic Left Heart Syndrome).

Q2.9: What are the congenital heart diseases that are "Ductus Arteriosus Dependent" for pulmonary circulation?**Answer:**

- Severe PS
- Severe TOF
- Pulmonary atresia
- Tricuspid atresia
- Ebstein anomaly.

BIG QUESTION 3: Describe the characteristic findings associated with Total Anomalous Pulmonary Venous Connection (TAPVC), Transposition of Great Arteries (TGA), Coarctation of Aorta, and Ebstein Anomaly.**Broad Answer:**

- Total Anomalous Pulmonary Venous Connection (TAPVC) shows a "figure of 8" or "Snowman" appearance on Chest X-ray and equal oxygen saturation in all four heart chambers.

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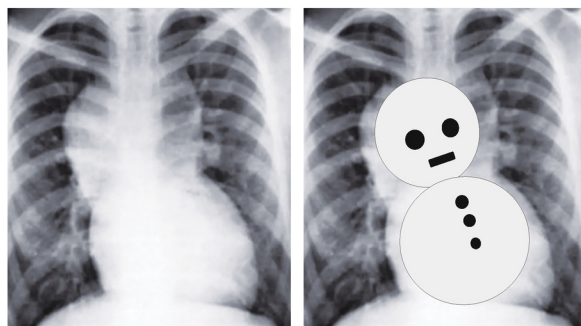
- Transposition of the Great Arteries (TGA) presents with an "Egg on the side" cardiac silhouette and increased pulmonary blood flow if associated with VSD/PDA.
- Coarctation of Aorta: Chest X-ray shows a "figure of 3" appearance and notching of the inferior margin of the 3rd-9th ribs.
- Ebstein Anomaly displays a "Box-shaped heart."

DETAILED QUESTIONS

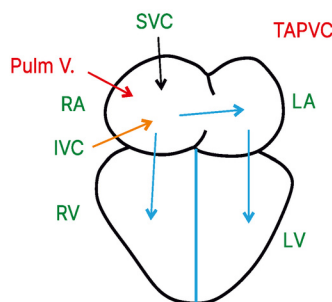
Q3.1: Which congenital heart disease shows a "figure of 8" or "Snowman" appearance on Chest X-ray?

Answer:

- Total anomalous pulmonary venous connection (TAPVC).



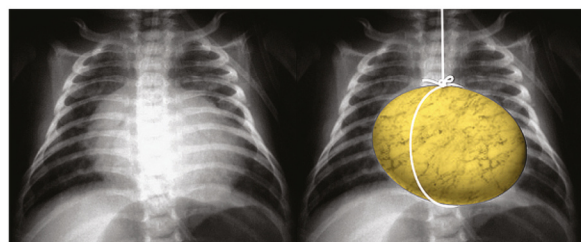
- All 4 chambers of the heart have equal oxygen saturation.



Q3.2: What is the characteristic cardiac silhouette seen in Transposition of Great Arteries (TGA)?

Answer:

- The cardiac silhouette shows an "Egg on the side" appearance.



Q3.3: In TGA, what happens to pulmonary blood flow if it is associated with a VSD or PDA?

Answer:

- There is an increase in pulmonary blood flow if TGA is associated with a shunt (like VSD or PDA).

Q3.4: What is the clinical presentation of severe Coarctation of Aorta in neonates?

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Answer:

- Heart failure in neonates with B/L feeble or impalpable femoral pulses.

Q3.5: How does moderate Coarctation of Aorta present clinically?**Answer:**

- Weak femoral pulses and hypertension.

Q3.6: What is a typical clinical symptom of milder Coarctation of Aorta?**Answer:**

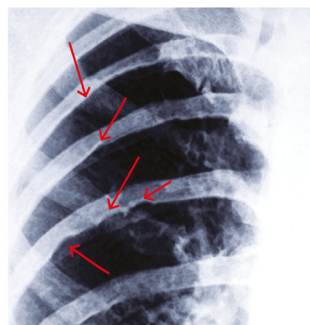
- Intermittent claudication of lower limbs.

Q3.7: Where is the murmur typically heard on auscultation in Coarctation of Aorta, and what causes it?**Answer:**

- Murmur in the shoulder region due to the collaterals.

Q3.8: What characteristic X-ray finding is associated with Coarctation of Aorta?**Answer:**

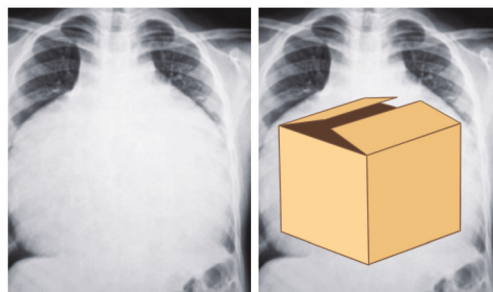
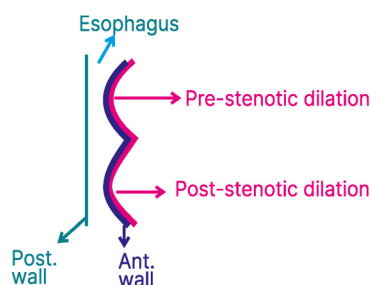
- A "figure of 3" appearance.

**Q3.9: What specific rib abnormality is seen on Chest X-ray in Coarctation of the Aorta, and at what age is it usually observed?****Answer:**

- Notching of the inferior margin of the 3rd-9th ribs, usually seen >3 years of age.

Q3.10: What is the characteristic X-ray finding for Ebstein Anomaly?**Answer:**

- The X-rays show cardiac shadow typically covering the entire thorax, appearing as a "Box-shaped heart."

**Q3.11: What specific ECG finding is seen in Ebstein Anomaly?****Answer:**

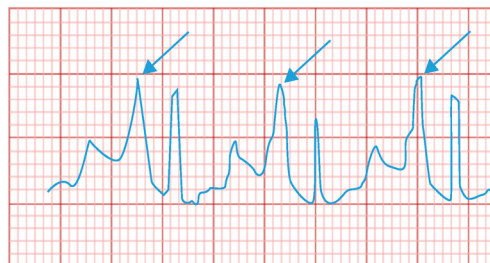
- ECG shows Himalayan 'P' waves.

Downward displacement of tricuspid valve

Atrialization of the right ventricle

+

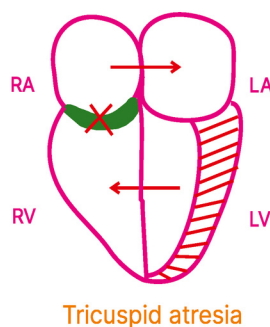
Functionally abnormal tricuspid valve



Q3.12: Which congenital cyanotic heart disease is characterized by left axis deviation on ECG?

Answer:

- Tricuspid atresia.



BIG QUESTION 4: Briefly describe Ventricular Septal Defect (VSD)

Broad Answer:

- Ventricular Septal Defect (VSD) is the most common congenital heart disease overall, and the most common congenital acyanotic heart disease in children, also the most affected by infective endocarditis.
- Clinical presentation in infants (6-10 weeks) includes tachypnea, tachycardia, heart failure, recurrent pneumonia, and failure to thrive.
- ECG shows left axis deviation and LV hypertrophy
- Complications include Infective endocarditis and Eisenmenger syndrome

DETAILED QUESTIONS

Q4.1: What is the most common congenital heart disease in children?

Answer:

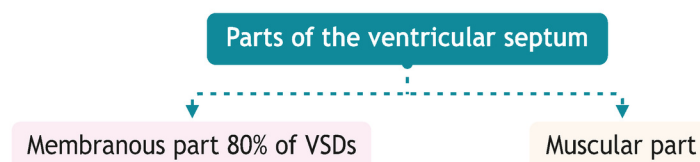
- Ventricular Septal Defect (VSD).

Q4.2: What are the parts of the ventricular septum?

Answer:

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Q4.3: Which congenital heart disease is most commonly affected by infective endocarditis in children?

Answer:

- Ventricular Septal Defect (VSD).

Q4.4: What are the typical pressure ranges in the Right Ventricle (RV) and Left Ventricle (LV)?

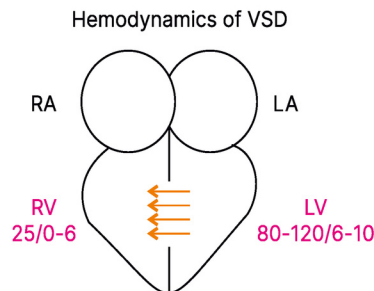
Answer:

- RV: 25/0-6 mmHg.
- LV: 80-120/6-10 mmHg.

Q4.5: What is the Hemodynamics in VSD during systole and diastole?

Answer:

- Normally, blood flows from the LV to the RV with a gush throughout systole.
- During diastole, the blood also flows from the LV to the RV, but the pressure gradient is less.



Q4.6: What is the classic auscultatory finding in VSD, and when does it occur during the cardiac cycle?

Answer:

- Pansystolic murmur - begins at the first heart sound and extends up to the second heart sound.

Q4.7: What is the pathology feature that leads to the auscultatory findings in VSD?

Answer:

- Due to the excessive turbulence of the blood flow in a case of VSD.

Q4.8: At what age do clinical presentations of VSD typically become apparent?

Answer:

- Age of presentation is 6-10 weeks.

Q4.9: What are the common clinical presentations of VSD in infants?

Answer:

- Tachypnea
- Tachycardia
- Features of heart failure
- Recurrent episodes of pneumonia
- Failure to thrive.

Q4.10: What are the characteristic ECG findings in VSD?

Answer:

- Left axis deviation (normally, all babies at birth have right axis deviation)
- Left ventricular hypertrophy.

Q4.11: What are the Chest X-ray findings typically seen in VSD?

Answer:

- Cardiomegaly with LV apex (moves down & out)
- Pulmonary plethora due to excess blood going to the lungs.

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Q4.12: What are the differences between Infective Endocarditis and Eisenmenger Syndrome as complications of VSD based on clinical features?

Answer:

- Infective endocarditis: VSD + fever + Clubbing without cyanosis.
- Eisenmenger syndrome: VSD + Clubbing + Cyanosis [No fever].

Q4.13: What are the main components of medical management for VSD?

Answer:

- Treatment of Heart Failure (Diuretics like Furosemide, Digoxin)
- Treatment of pneumonia, Nutritional rehabilitation.

Q4.14: What is the surgical management for VSD?

Answer:

- Closure of VSD by Dacron Patch.

Q4.15: What are the indications for surgical intervention in VSD?

Answer:

- Heart failure refractory to medical management.
- If pulmonary blood flow is more than double of systemic blood flow

Q4.16: What are the contraindications for surgical intervention in VSD?

Answer:

- Eisenmenger syndrome, Irreversible pulmonary HTN.

BIG QUESTION 5: Briefly describe Atrial Septal Defect (ASD)

Broad Answer:

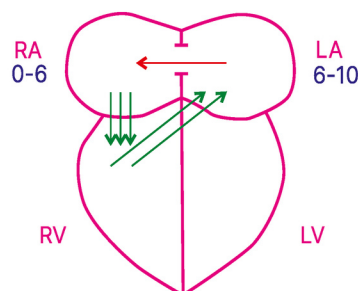
- Atrial Septal Defect (ASD) is a congenital heart disease where the shunt remains silent without a murmur.
- The most common type of ASD is Ostium Secundum, involving the upper part of the atrial septum, while Ostium Primum involves the lower part.

DETAILED QUESTIONS

Q5.1: What is the typical auscultatory characteristic of the shunt in ASD?

Answer:

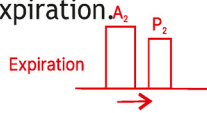
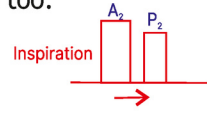
- The shunt remains silent in ASD (No Murmur).



Q5.2: What are the components of a normal second heart sound (S2)?**Answer:**

- Normal S2 consists of A₂ (due to the closure of the aortic valve) and P₂ (due to the closure of the pulmonary valve).

Q5.3: What is the difference between the normal physiological splitting of S2 and the "wide, fixed split of S2" seen in ASD during expiration and inspiration?**Answer:**

Normal second heart sounds	ASD
• During expiration, the aortic and pulmonary close almost together. S, A₂, and P₂ are heard as single heart sounds.  Expiration	• Irrespective of inspiration or expiration, RA receives excess blood from SVC, IVC, and LA. Here, P₂ occurs later even during expiration.  Expiration
• During inspiration, due to excess blood inflow into RA, the pulmonary valve closes late. Here, the physiological splitting of the heart sound occurs.  Inspiration Physiological Split of S₂	• During inspiration, RA cannot take in any more blood due to the presence of blood during expiration. Hence, P₂ occurs later here too. • Wide, Fixed Split of S2 is seen in ASD. (Also seen in TAPVR and RBBB)  Inspiration

Q5.4: What are the two other conditions besides ASD where a wide, fixed split of S2 is observed?**Answer:**

- TAPVR (Total anomalous pulmonary venous return)
- RBBB (Right Bundle Branch Block).

Q5.5: What are the types of ASD?

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Answer:

Types	
Ostium Primum • Involves the lower part of the atrial septum	Ostium Secundum • Involves the upper part of the atrial septum • Most common type

BIG QUESTION 6: Describe Patent Ductus Arteriosus (PDA)**Broad Answer:**

- Patent Ductus Arteriosus (PDA) is a structure connecting the pulmonary artery to the aorta, just distal to the origin of the left subclavian artery.
- Hemodynamically, a large pressure difference between the aorta and pulmonary artery results in a characteristic continuous machinery murmur on auscultation.
- In preterm neonates, PDA presents with hyperdynamic circulation, bounding pulses, apnea, tachypnea, and tachycardia.
- In older infants (6-10 weeks), it can lead to failure to thrive and features of heart failure.

DETAILED QUESTIONS

Q6.1: What anatomical connection does the ductus arteriosus form?

Answer:

- It is a structure that connects the pulmonary artery to the aorta, just distal to the origin of the left subclavian artery.

Q6.2: What are the two main factors that predispose to the patency of the ductus arteriosus?

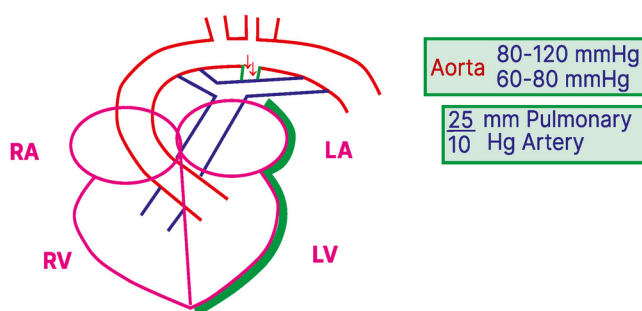
Answer:

- Hypoxia and Prematurity.

Q6.3: What is the classic auscultatory finding in PDA, and what causes it?

Answer:

- Continuous machinery murmur - due to a huge difference between pressures in the aorta & pulmonary artery.



Q6.4: What are the typical clinical presentations of PDA in preterm neonates?

Answer:

- Seen in preterm neonates, hyperdynamic circulation, bounding pulses, apnea, tachypnea, and tachycardia.

Q6.5: What are the typical clinical presentations of PDA

Answer:

Neonates	Older infants (6-10 weeks)
• Seen in Preterm neonates • Hyperdynamic circulation • Bounding pulses • Apnea	• Tachypnea • Tachycardia • Failure to thrive • Features of heart failure

Q6.6: In which specific congenital heart disease and associated syndrome is Differential Cyanosis seen?

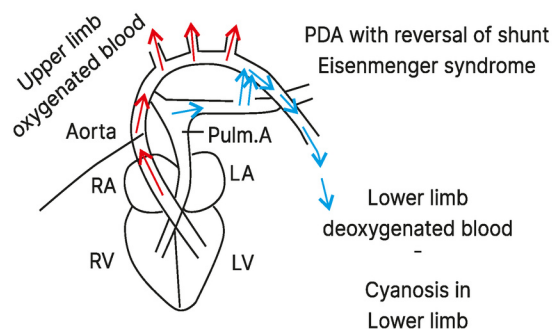
Answer:

- Seen in PDA with reversal of shunt [Eisenmenger Syndrome].

Q6.7: How does Differential Cyanosis manifest clinically in terms of upper and lower limb oxygenation?

Answer:

- No cyanosis in the upper limbs (as oxygenated blood goes there), but cyanosis in the lower limbs (mixing of blood with deoxygenated blood).



Q6.8: What is the pathophysiology that leads to Differential Cyanosis in PDA with Eisenmenger Syndrome?

Answer:

- In PDA with Eisenmenger Syndrome, the deoxygenated blood moves from the pulmonary artery to the aorta.
- The upper limbs receive oxygenated blood from the ascending aorta (proximal to the PDA), whereas the lower limbs receive deoxygenated blood entering the aorta from the pulmonary artery (distal to the left subclavian artery).

BIG QUESTION 7: Describe Tetralogy of Fallot (TOF) and its prevalence

Broad Answer:

- Tetralogy of Fallot (TOF) is the most common congenital cyanotic heart disease in children and beyond infancy.
- Its four components are a large non-restrictive VSD, pulmonary infundibular stenosis, overriding of the aorta, and right ventricular hypertrophy.

DETAILED QUESTIONS

Q7.1: What is the most common congenital cyanotic heart disease in children?

Answer: Tetralogy of Fallot (TOF).

Q7.2: What is the most common congenital cyanotic heart disease beyond infancy?

Answer: Tetralogy of Fallot (TOF).

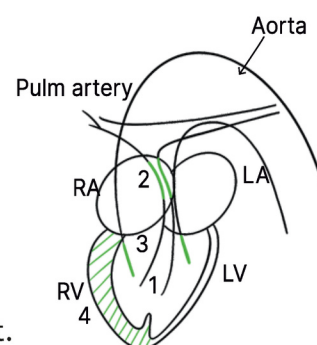
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Q7.3: What are the classic components of Tetralogy of Fallot (TOF)?

Answer:

1. Large non-restrictive VSD
 2. Pulmonary infundibular stenosis
 3. Overriding of the aorta
 4. Right ventricular hypertrophy
- ASD is a component of the trilog and pentalogy of Fallot, not the tetralogy of Fallot.



Q7.4: What is the primary pathophysiological mechanism that causes the symptoms in TOF?

Answer:

- The primary pathophysiological mechanism in TOF is right ventricular outflow tract obstruction (usually due to pulmonary stenosis), which leads to reduced pulmonary blood flow.
- This causes right-to-left shunting of deoxygenated blood through the ventricular septal defect (VSD) into the systemic circulation, resulting in hypoxemia and cyanosis.
- Reversal of the shunt is never seen in TOF.

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Q7.5: What are the common clinical features of TOF related to hypoxia?

Answer:

- Cyanosis (central) due to hypoxia ($\text{SpO}_2 = 75\text{-}85\%$), Clubbing, Polycythemia, Cyanotic spells.
- Heart failure is not seen in TOF unless it is complicated by anemia, infective endocarditis, myocarditis, or systemic hypertension.

Q7.6: What are the characteristic auscultatory findings in TOF?

Answer:

- Ejection systolic murmur in the pulmonary area, Single S2 (P2 is soft and inaudible).

Q7.7: What are the key Chest X-ray findings in TOF?

Answer:

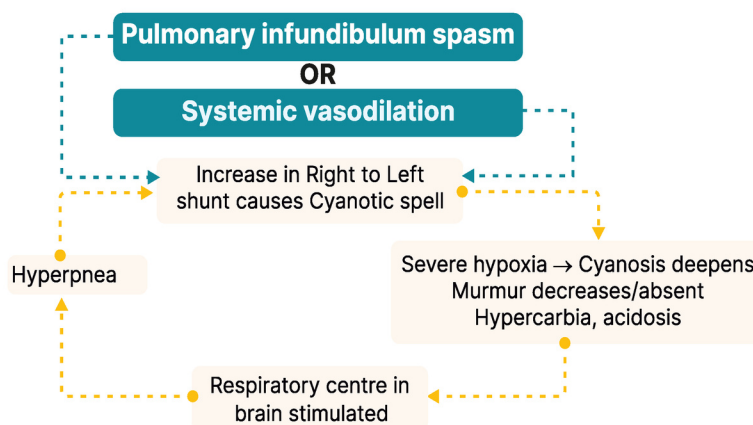
- Boot-Shaped Heart (or) 'Cor En Sabot' Appearance + Pulmonary Oligemia.



Q7.8: What is another name for a Cyanotic Spell in TOF, and explain its pathophysiology?

Answer:

- Tet Spell.



Q7.9: What is the immediate treatment for a cyanotic spell in TOF?

Answer:

- Moist O_2 inhalation (to reduce hypoxia)
- Inj. Sodium Bicarbonate (to neutralize acidosis)
- Morphine
- Ketamine (increases systemic vascular resistance)
- Alpha agonists (phenylephrine)
- Beta-blockers (Propranolol): ↓ pulmonary infundibular spasm
- Squatting or knee-chest position
- PRBC transfusion.

Q7.10: What is the definitive (corrective) surgical treatment for TOF?**Answer:**

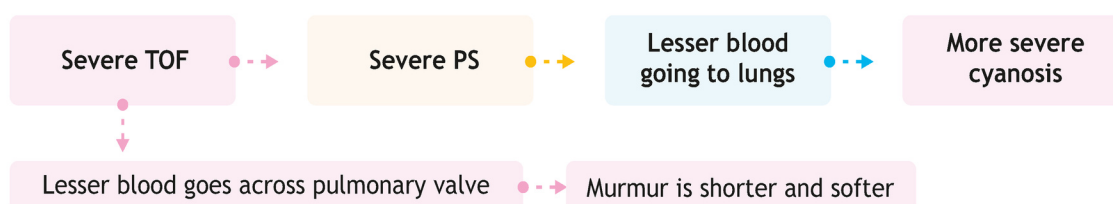
- VSD closure + Repair of pulmonary stenosis.

Q7.11: What is the purpose of palliative shunt surgery in TOF?**Answer:**

- To connect the pulmonary artery & aorta, or their branches, so that more blood is directed to the lungs, thereby improving oxygenation.

Q7.12: What are the types of palliative shunts?**Answer:**

Name of the shunt	The pulmonary artery connected to
Blalock Taussig shunt	Subclavian artery
Waterston's Shunt	Ascending Aorta
Pott's Shunt	Descending Aorta

Q7.13: What are the effects of severe TOF?**Answer:****BIG QUESTION 8: Detail the diagnostic criteria for Acute Rheumatic Fever (ARF) according to the Jones criteria (major and minor).****Broad Answer:**

- Acute Rheumatic Fever (ARF) is diagnosed using Jones Criteria, which include major and minor criteria, along with essential evidence of a preceding streptococcal infection.

Major Criteria	Minor Criteria
1. Carditis (clinical or subclinical): 50-60% 2. Arthritis: Polyarthritis/ monoarthritis/ polyarthralgia (moderate to high-risk population) - 75% 3. Chorea (Sydenham's chorea): 10-15% 4. Erythema marginatum 5. Subcutaneous nodules	1. Monoarthralgia 2. Fever 3. Increased ESR/CRP 4. Prolonged PR interval

DETAILED QUESTIONS**Q8.1: What are the two main categories of diagnostic criteria for Acute Rheumatic Fever (ARF)?****Answer:**

- Major Criteria and Minor Criteria.

Q8.2: What are the major criteria for diagnosing ARF?**Answer:**

- Carditis (clinical or subclinical): 50-60%
- Arthritis: **Polyarthritis/monoarthritis/polyarthralgia** (moderate to high-risk population): 75%
- Chorea (Sydenham's chorea): 10-15%
- Erythema marginatum
- Subcutaneous nodules

Q8.3: What are the minor criteria for diagnosing ARF?

Answer:

- Monoarthralgia
- Fever
- Increased ESR/CRP
- Prolonged PR interval

Q8.4: What is the essential prerequisite for diagnosing Rheumatic Fever, besides the major and minor criteria?

Answer:

- Evidence of streptococcal infection (in the form of throat swab culture, rapid antigen detection test, or **elevated ASO titers**).

Q8.5: What criteria are required to diagnose an initial episode of ARF?

Answer:

- 2 major or 1 major + 2 minor criteria.

Q8.6: What criteria are required to diagnose a recurrence of ARF?

Answer:

- 2 major or 1 major + 2 minor or 3 minor criteria.

Q8.7: What is the antibiotic treatment for ARF?

Answer:

- A course of antibiotics with penicillin.
- Erythromycin or Azithromycin in penicillin allergy.

Q8.8: What is the use of Aspirin in the management of ARF?

Answer:

- High dose (50-70 mg/kg/dose) followed by low dose.

Q8.9: When are corticosteroids indicated in the treatment of ARF?

Answer:

- For Carditis + features of CCF.

Q8.10: What is the primary medication used for secondary prophylaxis of ARF?

Answer:

- **Inj. Benzathine penicillin** is given every 3-4 weeks IM.
 - o 6 lakh IU in children ≤ 27 kg
 - o 1.2 million IU in children > 27 kg

Q8.11: What is the duration of secondary prophylaxis for ARF?**Answer:**

- The duration is whichever is later for the given age criteria:

Clinical features	For next	Till the age of
Without carditis	5 years	21 years
With carditis but without residual heart disease	10 years	21 years
Residual heart disease	10 years	40 years or life

Q8.12: What are the main maternal drugs that can cause specific congenital defects in a child?**Answer:**

Maternal Drug	Defect In Child
Lithium	Ebstein anomaly
ACE inhibitors	IUGR, Potter sequence
Carbamazepine	Neural tube defects
Carbimazole	Scalp defects, Choanal atresia, esophageal atresia
Warfarin	Nasal bone hypoplasia, Bleeding manifestations
Phenytoin	Neuroblastoma, Vit K deficiency, Bleeding, microcephaly, and NTD

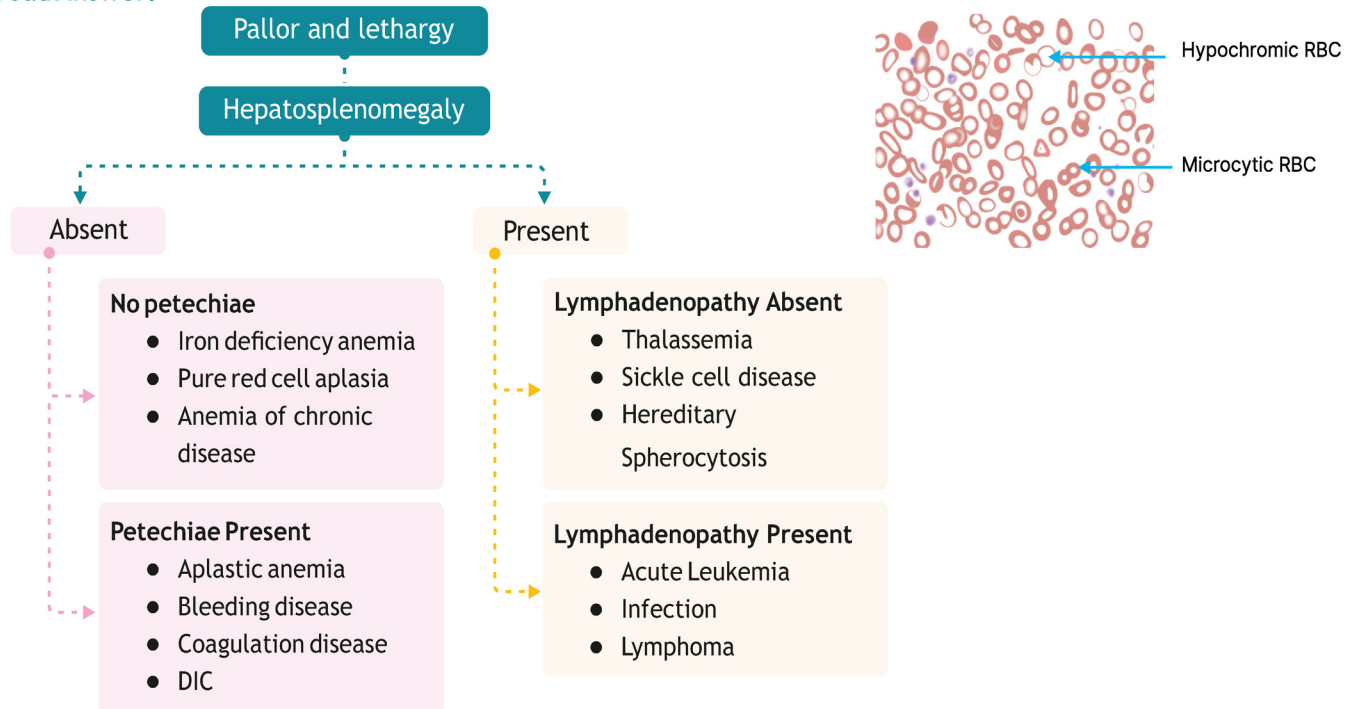


8. PEDIATRIC HEMATO-ONCOLOGY

BIG QUESTION 1: Briefly describe the approach to anemia in children

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Broad Answer:



DETAILED QUESTIONS

Q1.1: What is the WHO definition of anemia for children between 6 months and 5 years?

Answer:

- Hb < 11 g/dl.

Q1.2: What is the WHO definition of anemia for children between 6 and 14 years?

Answer:

- Hb < 12 g/dl.

Q1.3: What is the most common cause of nutritional disorders globally?

Answer:

- Iron Deficiency Anemia.

Q1.4: What is the most common cause of anemia worldwide?

Answer:

- Iron Deficiency.

Q1.5: Which vitamin helps in the absorption of iron?

Answer:

- Vitamin C.

Q1.6: Iron Deficiency Anemia is associated with which gastrointestinal condition?**Answer:**

- Celiac disease.

Q1.7: What are the differential diagnoses of Microcytic, Hypochromic Anemia?**Answer:**

Parameters	Iron deficiency anemia	B Thalassemia trait	Anaemia of Chronic disease
RDW	Increased	Normal	Normal/Increased
S. Iron	Low	Normal/Increased	Low
S. Ferritin	Low	Normal/Increased	Increased
TIBC	Increased	Normal	Decreased
Mentzer Index	>14	<13	

Q1.8. What is the formula for calculating the Mentzer index?**Answer:**

$$\text{Mentzer Index} = \frac{\text{MCV (fl)}}{\text{RBC count (million)} \frac{\mu\text{l}}{\mu\text{l}}}$$

Q1.9: What type of anemia is Megaloblastic Anemia, and what is the typical MCV range?**Answer:**

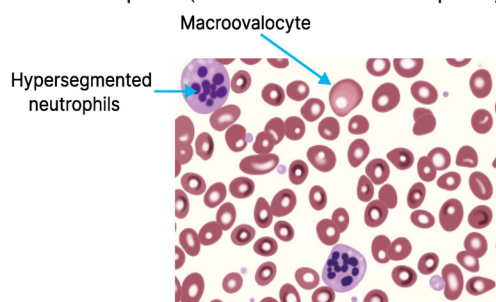
- Macrocytic anemia with MCV >90 fL.

Q1.10: What are the underlying deficiencies that cause Megaloblastic Anemia?**Answer:**

- Due to Vit B12 or folic acid deficiency.

Q1.11: What are the characteristic findings on a peripheral smear in Megaloblastic Anemia?**Answer:**

- Macroovalocytes, Hypersegmented neutrophils (>5 lobes in 5% neutrophils).

**Q1.12: What is the treatment for Megaloblastic Anemia?****Answer:**

- Supplementation of Vitamin B12 and Folic acid.

BIG QUESTION 2: What are the types, pathogenesis, clinical manifestations, microscopic examination, and management of Thalassemia?

Broad Answer:

- Beta-Thalassemia is an autosomal recessive disorder with decreased beta-globin chain production
- Clinical features include severe anemia, regular transfusion requirement, hepatosplenomegaly, hemolytic jaundice, short stature, features of iron toxicity, and Hemolytic Facies
- The definitive diagnosis is globin gene mutation analysis
- Treatment involves regular blood transfusions
 - o Hematopoietic Stem Cell Transplantation (HSCT)
 - o Iron chelation therapy

DETAILED QUESTIONS**Q2.1: What is the basic genetic defect in Beta-Thalassemia?**

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Answer:

- Decreased production of Beta globin chains.

Q2.2: What is the inheritance pattern of Beta-Thalassemia?**Answer:**

- Autosomal recessive.

Q2.3: What are the common Beta-Thalassemia mutations found in India?**Answer:**

- IVS 1-5 G → C, IVS 1-1 G → T, Codon 41/42.

Q2.4: What is the characteristic "Hemolytic Facies" seen in Beta-Thalassemia?**Answer:**

- Frontal prominence, Depressed bridge of the nose, Maxillary prominence.

Q2.5: What are the clinical features of Beta-Thalassemia?**Answer:**

- Severe anemia, regular transfusion requirement, Hepatosplenomegaly, Hemolytic jaundice, Short stature, Features of iron toxicity.

Q2.6: What specific radiological finding is seen on a lateral X-ray skull in severe Beta-Thalassemia?**Answer:**

- 'Hair on End' or 'Crew Cut appearance'.



Q2.7: What are the typical findings of hemolytic anemia on investigation for Beta-Thalassemia?**Answer:**

- Low Hb, low MCV, low MCH (thalassemia major)
- Microcytic-hypochromic anemia
- Target cells
- Howel jolly bodies
- Poikilocytosis
- Increased LDH
- Increased unconjugated bilirubin

Q2.8: How does osmolar fragility typically present in Beta-Thalassemia, and what specific test is used?**Answer:**

- Decreased (NESTROFT - Naked Eye Single Red Tube Cell Osmotic Fragility Test).

Q2.9: What is the result of the Coombs test in Beta-Thalassemia, and why is it significant?**Answer:**

- Negative (+ve in immune hemolytic anemias), indicating it's not immune-mediated hemolysis.

Q2.10: What are the expected findings on Hb HPLC (High-performance liquid chromatography) or Hb electrophoresis in Beta-Thalassemia?**Answer:**

- HbA decreased, HbA2 increased, and HbF increased.
- Beta thalassemia trait: HbA2:3.5%-7%.

Q2.11: What is the definitive diagnostic method for Beta-Thalassemia?**Answer:**

- Globin gene mutation analysis - helps in prenatal diagnosis in the next pregnancy.

Q2.12: What is the primary management strategy for Beta-Thalassemia to maintain Hb levels?**Answer:**

- Regular Blood Transfusions (PRBC): To maintain pre-transfusion Hb level between 9 - 10.5 g/dL.

Q2.13: What is the only curative treatment for Beta-Thalassemia?**Answer:**

- Hematopoietic stem cell transplantation (HSCT).

Q2.14: What are the common iron chelation therapy drugs used in Thalassemia?**Answer:**

- Deferoxamine, Deferiprone, Deferasirox.

BIG QUESTION 3: What are the types, pathogenesis, clinical manifestations, microscopic examination, and management of sickle cell anemia?**Broad Answer:**

- Sickle Cell Anemia is the most common Structural Hemoglobinopathy.

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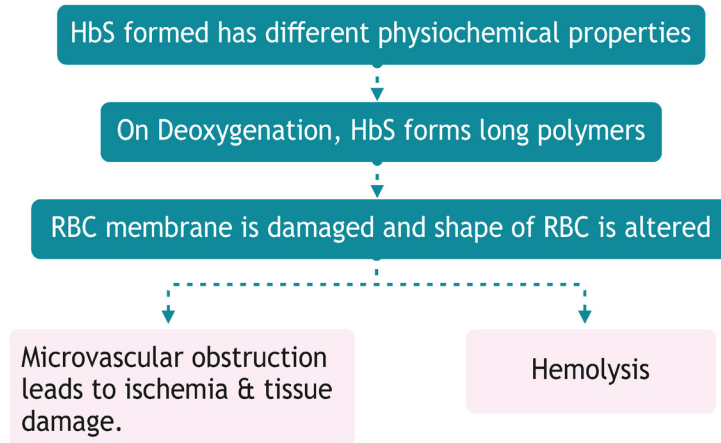
- Autosomal recessive inheritance
- The basic defect is a point mutation in the 6th codon of the β -globin gene, replacing glutamate with valine, forming HbS.

DETAILED QUESTIONS

Q3.1: What is the pathophysiology of Sickle Cell Anemia?

Answer:

- A point mutation, resulting in the replacement of glutamate with valine, forms HbS.



Q3.2: What is the typical age of presentation for clinical features of Sickle Cell Anemia?

Answer:

- It usually presents after 6 months of age.

Q3.3: What are the common clinical features of Sickle Cell Anemia?

Answer:

- Anemia: Pallor, generalized weakness.
- Ischemia affecting CNS (Stroke), Retinopathy, Hand (Dactylitis), Priapism in males, Auto splenectomy due to repeated splenic infarcts, and Renal papillary necrosis.

Q3.4: What is the most common cause of death in patients with Sickle Cell Disease?

Answer:

- Infections.

Q3.5: What is the most common cause of osteomyelitis in Sickle Cell Disease?

Answer:

- Salmonella.

Q3.6: What are the types of crises seen in Sickle Cell Disease?

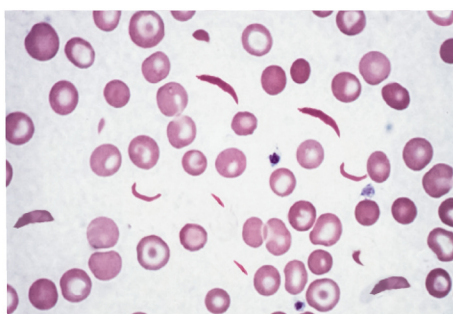
Answer:

- Vaso-occlusive crisis: Painful.
- Aplastic crisis: Due to Parvovirus B19.
- Hemolytic crisis.
- Splenic sequestration.
- Acute chest syndrome.

Q3.7: What are the key diagnostic findings on a peripheral smear for Sickle Cell Anemia?

Answer:

- Evidence of hemolysis, Sick cells, and Howell-Jolly bodies.



Q3.8: How is a Sickling Test performed, and what does it detect?

Answer:

- Mixing a blood sample with metabisulfite or dithionate induces sickling of RBCs if HbS is present.

Q3.9: What specific finding is seen on a spleen biopsy in Sickle Cell Disease?

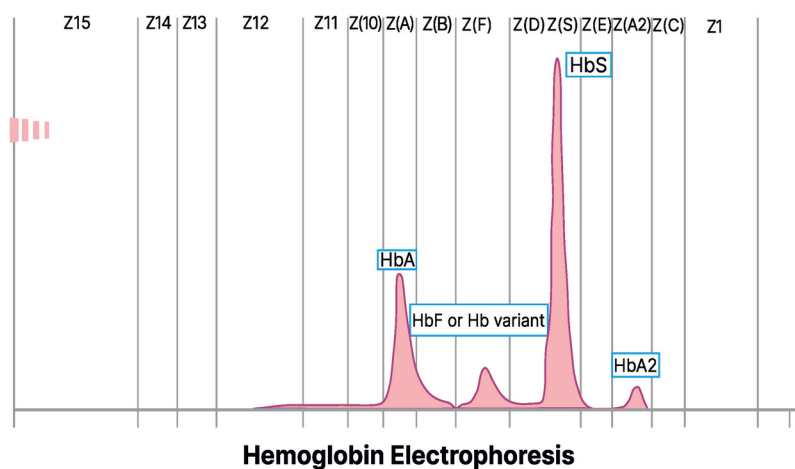
Answer:

- Gamma Gandy bodies.

Q3.10: What definitive laboratory test is used to detect the HbS peak?

Answer:

- Hb Electrophoresis.



Q3.11: What is the role of Hydroxyurea in the treatment of Sickle Cell Anemia?

Answer:

- Hydroxyurea increases HbF levels and decreases the requirement for blood transfusions.

Q3.12: What is considered the mainstay of treatment for Sickle Cell Anemia?

Answer:

- Regular blood transfusion.

Q3.13: What are the curative treatment options for Sickle Cell Anemia?

Answer:

- Hematopoietic stem cell transplant and gene therapy.

Q3.14: What are the newer drugs used in the treatment of Sickle Cell Anemia?

Answer:

- Voxelotor: to increase oxygen affinity and reduce HbS polymerization.
- L-glutamine: to reduce vaso-occlusion.
- Crizanlizumab: also reduces vaso-occlusion.

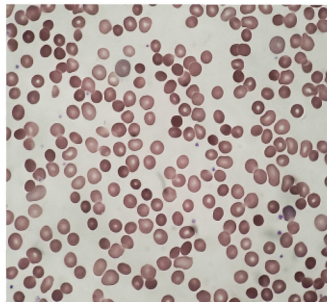
BIG QUESTION 4: Briefly describe the pathogenesis, features of Aplastic Anemia, ITP, Wiskott-Aldrich, and Hemophilia A.

Broad Answer:

- **Aplastic Anemia** is an inherited/acquired condition characterized by pancytopenia with hypo-/acellular bone marrow.
- **ITP (Immune Thrombocytopenic Purpura)** presents with generalized petechiae and gum bleeding, often following a viral URI.
- **Wiskott-Aldrich Syndrome** is an X-linked recessive disorder (WASP gene), most common in males, characterized by a triad of recurrent infections, eczema, and thrombocytopenia.
- **Haemophilia A** is an X-linked recessive disorder (more common in males) due to Factor VIII deficiency.

DETAILED QUESTIONS

Q4.1: A 2-year-old boy is brought in with episodes of icterus and pallor. On examination, Splenomegaly is positive, and a family history of similar illness in the father is observed. PS is shown below. What is the diagnosis?



Answer:

- Hereditary Spherocytosis
 - o Autosomal dominant inheritance
 - o Spherocytosis in the peripheral smear

Q4.2: A child with severe pallor presents with bleeding manifestations. On examination, no hepatosplenomegaly. BM biopsy shows fat globules. What is the diagnosis?

Answer:

- Aplastic Anemia.

Q4.3: What are the bone marrow and peripheral blood findings in aplastic anemia?

Answer:

- Inherited/acquired condition with pancytopenia with hypo / acellular bone marrow.

Q4.4: What are the clinical features of Aplastic Anemia?

Answer:

- Anemia: Pallor, easy fatigability.
- Thrombocytopenia: Bleeding manifestations.
- Leukopenia: Fever, recurrent infections.

Q4.5: What physical examination findings are typically absent in Aplastic Anemia?

Answer:

- No lymphadenopathy, no hepatosplenomegaly.

Q4.6: A child with generalized petechiae and gum bleeding presents with a history of viral URI 2 weeks back. What is the diagnosis?

Answer:

- ITP (Idiopathic/Immune Thrombocytopenic Purpura).

Q4.7: What is the initial treatment approach for a first episode of ITP with no significant bleeding?

Answer:

- Wait and watch.

Q4.8: What is the first-line treatment for ITP?

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Answer:

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- A short course of oral Prednisolone 2 mg/kg/day for 5-7 days.
- If no response, IV Ig or Anti-D.

Q4.9: When are platelet transfusions typically indicated in ITP?

Answer:

- Usually avoided unless life-threatening bleeding is present.

Q4.10: What are the TOP (Thrombopoietin) agonists used in ITP treatment?

Answer:

- Eltrombopag, Romiplostim.

Q4.11: What medication is considered if there is no response to initial ITP treatments?

Answer:

- Rituximab.

Q4.12: What intervention might be considered in refractory ITP cases?

Answer:

- Splenectomy.

Q4.13: What is the inheritance pattern of Wiskott-Aldrich Syndrome, and what gene is involved?

Answer:

- X-linked recessive:
- WASP gene.

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Q4.14: What is the classic triad of Wiskott-Aldrich Syndrome?**Answer:**

- Recurrent infections, Eczema, Thrombocytopenia.

Q4.15: What are the typical immunoglobulin findings in Wiskott-Aldrich Syndrome?**Answer:**

- IgM: Low; IgE and IgA: elevated.

Q4.16: A male 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?**Answer:**

- Haemophilia A.

Q4.17: What is the basic defect in Haemophilia A?**Answer:**

- Deficiency of factor VIII.

Q4.18: What is the inheritance pattern of Haemophilia A, and what is its prevalence regarding gender and family history?**Answer:**

- X-linked recessive: more common in males.
- Family h/o maternal uncles being affected.

Q4.19: How is Haemophilia A classified by factor VIII levels?**Answer:**

- Mild (factor VIII >5%)
- Moderate (factor VIII: 1-5%)
- Severe (factor VIII <1% factor level)

Q4.20: What are the common clinical features of Haemophilia A?**Answer:**

- A male child with or without a family history, H/o joint swelling following trauma (Ankle > knee joint), and deep bleeding.

Q4.21: What are the characteristic laboratory findings in Haemophilia A?**Answer:**

- Prolonged aPTT but normal PT, BT, and platelet counts.
- Low factor VIII levels.

Q4.22: What is the treatment of choice for Haemophilia A?**Answer:**

- Factor VIII replacement is the treatment of choice.
- If not available, use fresh frozen plasma (FFP) or Cryoprecipitate.
- Avoid contact sports

Q4.23: What precautions should be taken for children with Haemophilia A?**Answer:**

- Precautions during immunization - Compress the site of injection to prevent the formation of a hematoma.

BIG QUESTION 5: What are the types, pathogenesis, clinical manifestations, diagnosis, and management of paediatric leukemias?**Broad Answer:**

- Acute Leukemia is the most common malignancy and the most common hematological malignancy in children.
- The most common leukemia in children is ALL (B-cell ALL > T-cell ALL)
- Superior mediastinal syndrome is more common in adolescent boys with T-cell ALL.

DETAILED QUESTIONS**Q5.1: What is the most common type of leukemia in children? Which subtype is more prevalent?****Answer:**

- ALL (Acute Lymphoblastic Leukemia), with B-cell ALL > T-cell ALL.

Q5.2: Which specific syndrome is more common in adolescent boys with T-cell ALL?**Answer:**

- Superior mediastinal syndrome.

Q5.3: What are the clinical features of leukemia related to pancytopenia?**Answer:**

- Anemia: Easy fatigability, Pallor.
- Thrombocytopenia: Bleeding.
- Leukopenia: Fever, recurrent infections.

Q5.4: What are the initial non-specific clinical features of leukemia in children?**Answer:**

- Anorexia, irritability, and fever.

Q5.5: What are the common types of pain in leukemia?**Answer:**

- Severe bone/ joint pains.

Q5.6: What are the common organs that can be involved in leukemia?**Answer:**

- Lymphadenopathy, hepatosplenomegaly, testicular involvement, CNS involvement, and Respiratory involvement.

Q5.7: What are the differences between AML and ALL?
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🔍 PYQ: FMGE 2021
Answer:

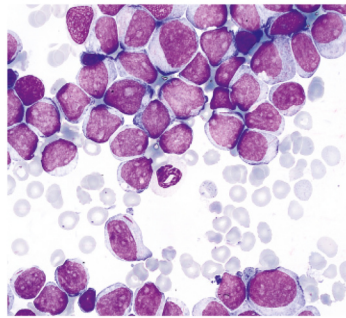
	ALL	AML
CNS infiltration	++	+
Testicular infiltration	++	+

Mediastinal LN	++	+
Chloromas	-	++ (M2> M5)
Gum hypertrophy	-	++ (M5)
DIC	-	++ (M3 variety)

Q5.8: What percentage of blasts in peripheral blood or bone marrow is generally essential to diagnose ALL & AML?

Answer:

- $\geq 20\%$ blasts.



Q5.9: What are the genetic exceptions in AML that allow for diagnosis irrespective of the blast percentage?

Answer:

- t(8;21), t(15;17), inv 16.

Q5.10: What are the characteristic cytochemistry findings that differentiate ALL from AML?

Answer:

- ALL: PAS +.
- AML: MPO & SBB (SUDAN BLACK B + STAIN) +.

Q5.11: What specific cytoplasmic inclusions are typically seen in AML?

Answer:

- Auer rods.

Q5.12: What is the general immunophenotyping marker for blasts?

Answer:

- Blasts (CD 34+).

Q5.13: How are myeloid lineage blast cells identified by immunophenotyping?

Answer:

- Blast cells of myeloid lineage show MPO +.

Q5.14: How are lymphoid lineage B-cells, T-cells, and NK cells identified by immunophenotyping?

Answer:

- B cells show CD19+.
- T cells show CD3+.
- NK cells show CD16+.

Q5.15: What are the drugs used in the induction phase of ALL treatment?**Answer:**

- Vincristine, Prednisolone, L-asparaginase, Anthracycline.

Q5.16: What is used for CNS prophylaxis in ALL treatment?**Answer:**

- Intrathecal Methotrexate with or without CNS radiation.

Q5.17: What are the drugs used in the consolidation/intensification phase of ALL treatment?**Answer:**

- Methotrexate, Cyclophosphamide, L-asparaginase, Anthracycline.

Q5.18: What is the purpose of the maintenance phase in ALL treatment, and what drugs are typically used?**Answer:**

- To prevent relapse.
- Drugs: Methotrexate, 6-Mercaptopurine.

Q5.19: What specific drugs are used in the treatment of AML M3?**Answer:**

- Arsenic, ATRA (All Trans Retinoic Acid)
- Ara-C (cytosine arabinoside)
- Anthracycline (doxorubicin).

Q5.20: What are the prognostic markers of ALL?**Answer:**

Characteristic	Good	Bad
Age	2-9 years	≤ 1 years or ≥ 10 years
Race	Whites	Blacks
Gender	Females	Males
TLC	< 10,000/micro-L	>2 Lac/micro-L
FAB	L1	L3
Cytogenetics	• t (12;21) • Hyperploidy	• t (5;14), t (9;22) • Hypoploidy
Phenotypically	Early Pre-B-cells	Mature B cells, T cells
Remission	≤14 days	≥28 days
MRD (Minimal Residual Disease)	< 0.01 %	>0.01%

BIG QUESTION 6: Describe Hodgkin's Disease, including its key clinical features, characteristic histological findings, and standard treatment regimen.**Broad Answer:**

- Hodgkin's Disease presents with clinical features such as fever for >10 days, generalized/cervical lymphadenopathy, and hepatosplenomegaly.
- The characteristic histological finding on biopsy is the Reed-Sternberg cell.
- The standard treatment regimen includes chemotherapy

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DETAILED QUESTIONS

Q6.1: What are the key clinical features of Hodgkin's Disease?

Answer:

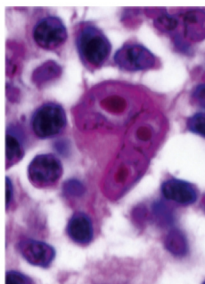
- Fever for >10 days
- Generalized/cervical lymphadenopathy, Hepatosplenomegaly on examination.

Q6.2: What is the characteristic histological finding on biopsy in Hodgkin's Disease?

Answer:

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- Reed-Sternberg cell.



Q6.3: What is the common chemotherapy regimen (ABVD) used for Hodgkin's Disease?

Answer:

- Adriamycin (Doxorubicin), Bleomycin, Vinblastine, Dacarbazine.

BIG QUESTION 7: What is the pathogenesis, clinical features, diagnosis, and treatment of Langerhans cell histiocytosis?

Broad Answer:

- Langerhans Cell Histiocytosis (LCH) is caused by the clonal proliferation of CD1a or CD207 cells.
- Diagnosis involves biopsy showing CD1a or S-100, CD207 markers, and electron microscopy revealing tennis racket-shaped Birbeck granules.
- Clinical features are diverse and depend on organ involvement
- Treatment for localized LCH includes steroids, radiation, and curettage.
- Multifocal LCH is treated with chemotherapy

DETAILED QUESTIONS

Q7.1: What is the underlying cause of Langerhans Cell Histiocytosis (LCH)?

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Answer:

- Due to clonal proliferation of **CD1a** or **CD207**.

Q7.2: What key cell markers are identified on biopsy for diagnosing LCH?

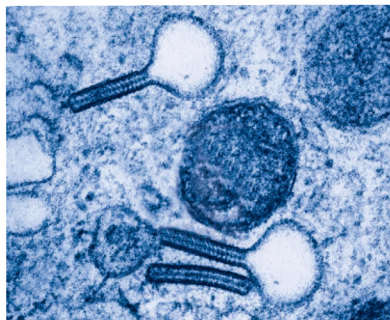
Answer:

- CD1a or S-100, CD207.

Q7.3: What characteristic finding is seen on electron microscopy for LCH?

Answer:

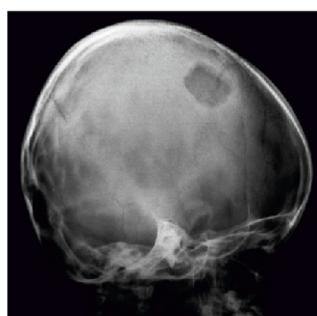
- Tennis racket-shaped **Birbeck granules**.



Q7.4: What are the typical clinical features of LCH related to bone involvement?

Answer:

- Osteolytic bone lesions ("punched out lesions"), especially skull & mastoid involvement, causing chronic ear discharge.



Q7.5: What skin manifestations are seen in LCH?

Answer:

- Seborrhea, Skin rash.

Q7.6: How does LCH manifest hematologically, and what other organs are involved?

Answer:

- Pancytopenia (Anemia, bleeding, fever), extramedullary hematopoiesis leading to hepatosplenomegaly.

Q7.7: What are the possible lung involvements in LCH?

Answer:

- Pneumothorax, pleural effusion.

Q7.8: What endocrine manifestations can occur with pituitary involvement in LCH?

Answer:

- Diabetes insipidus & GH deficiency.

Q7.9: What are the treatment options for localized LCH?

Answer:

- Steroids, Radiation, Curettage.

Q7.10: What chemotherapy agents are used for multifocal LCH?

Answer:

- Vinblastine, Etoposide, Prednisolone.



9. PEDIATRIC GASTROENTEROLOGY

BIG QUESTION 1: What are the key features and management of acute diarrhea in children?

Broad Answer:

- Acute diarrhea is defined as the passage of **≥3 loose stools in 24 hours**. The most common cause in children is rotavirus. Dehydration assessment guides management: ORS (5-10 ml/kg per stool) for ongoing losses, 75 ml/kg over 4 hours for some dehydration, and 100 ml/kg IV fluids (preferably Ringer's lactate + 5% dextrose) for severe dehydration. Zinc supplementation (10-20 mg/day) for 10-14 days reduces duration and severity. Antibiotics are not routinely indicated. Continue a normal diet to prevent nutritional compromise.

DETAILED QUESTIONS

Q1.1 What is acute diarrhea?

Answer:

- Passage of 3 or more loose liquid or watery stools in a day.

Q1.2 What is the most common cause of diarrhea in children?

Answer:

- Rotavirus.

Q1.3 What is the most common cause of constipation in children?

Answer:

- Functional or habitual due to improper toilet training

Q1.4 Which is the most common metabolic abnormality in diarrhea?

Answer:

- Isonatremic dehydration with metabolic acidosis and hypokalemia.

Q1.5 How is dehydration assessed in a child with diarrhea?

Answer:

Parameters	No Dehydration	Some Dehydration	Severe Dehydration
Sensorium	Active and Alert	Restless, Irritable	Lethargic, Floppy
Eyes	Normal	Sunken	Very sunken & dry
Tears	Present	Absent	Absent
Mucosa	Moist	Dry	Very dry
Thirst	Drinks normally, not thirsty	Thirsty, drinks eagerly	Drinks poorly/ not able to drink
Skin pinch	Goes back quickly	Goes back slowly (< 2sec)	Goes back very slowly (> 2sec)

- If the child has any 2 or more signs of some dehydration or severe dehydration, including at least one of the signs of sensorium, thirst, or skin pinch.

Q1.6 How is dehydration in a child with diarrhea managed?

Answer:

- Replacement of ongoing losses by WHO ORS [5-10 ml/kg 1 loose stool]
- Some dehydration: 75 ml/kg over 4 hours [if cannot accept orally, give IV fluid]
- Severe dehydration: 100 ml/kg fluid

Q1.7 What is the composition of New WHO ORS [in mOsm/L]?

 PYQ: FMGE 2020

Answer:

Glucose	75
Sodium	75
Potassium	20
Chloride	65
Citrate	10
Total Osmolarity	245 mOsm/L

Note:

- Resomal is a rehydration solution for a malnourished child. It has more potassium and less sodium, and some minerals.

Q1.8 How is fluid treatment given in severe dehydration?

Answer:

Age	1 st 30ml / kg	Next 70 ml/kg	Total (100 ml/kg)
< 1 year	Over 1 hour	Over 5 hour	6 hour
> 1 year	Over ½ hour	Over 2 ½ hour	3 hour

- IV fluid of choice: Ringer's Lactate in 5% Dextrose > Ringer's Lactate / Normal Saline

Q1.9 What is the treatment of acute diarrhea in children?

Answer:

Treatment or prevention of dehydration

- Maintain Normal Diet
- No role of antibiotics
- Zinc: Reduces the duration and severity of diarrhea in children from developing countries.
 - o Dose:
 - 2-6-month age: 10 mg/day
 - ≥ 6-month age: 20 mg/day
 - Duration is 10-14 days.

Q1.10 What is persistent diarrhea in children?

Answer:

- Diarrhea starts as an acute episode and lasts for at least 14 days.

YOURWISH

Q1.11 What is the management of persistent diarrhea in children?

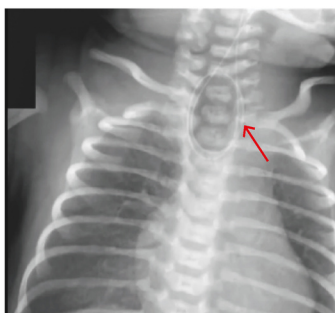
Answer:

- Correction of dehydration
- Supplement Zinc, Vitamin A
- Dietary modification
 - o Diet A (Reduced lactose diet)
 - o Diet B (Lactose-free diet with reduced starch)
 - o Diet C (Monosaccharide-based diet)

Q1.12 What is Osmotic diarrhea?

Answer: Diarrhea improves when the child is not fed for 24 hours.

Q1.13 A neonate presents with excessive frothing from the mouth and difficulty in feeding. There is also a history of polyhydramnios in the antenatal period. What is the diagnosis?



Coiled tube in the upper esophagus

Answer:

- Esophageal Atresia (EA) & Tracheoesophageal Fistula (TEF)
- Types
 - o 5 types based on the presence & location of EA & TEF
 - o The most common type is type C: The upper end of the esophagus is blind, and the lower end is connected to the Trachea

PYQ: FMGE 2023

BIG QUESTION 2: Describe Congenital Hypertrophic Pyloric Stenosis (CHPS), including its clinical features, risk factors, diagnosis, and management.

Broad Answer: Congenital Hypertrophic Pyloric Stenosis typically presents in infants between 2-6 weeks of age with projectile, non-bilious vomiting and features of dehydration. On examination, visible gastric peristalsis and a palpable “olive-shaped” mass in the epigastrium are characteristic.

Risk factors include maternal azithromycin use during pregnancy. Diagnosis is supported by **hypokalemic metabolic alkalosis with paradoxical aciduria** and confirmed by ultrasound showing a thickened pylorus. Initial management involves fluid and electrolyte correction with isotonic saline and potassium. The definitive treatment is **Ramstedt's pyloromyotomy**, which relieves the obstruction.

DETAILED QUESTIONS

Q2.1 What is the clinical presentation of congenital hypertrophic pyloric stenosis?

Answer:

Clinical Feature:

- Infants with forceful, projectile, non-bilious vomiting presenting between 2-6 weeks of age.
- Can present with dehydration.

On Examination:

- Visible peristalsis in the upper abdomen from left to right.
- Visible and palpable olive-shaped mass best appreciated in the Mid-Gastric Area.
- Easiest to palpate just after an episode of vomiting.

Q2.2 What is the risk factor for congenital hypertrophic pyloric stenosis?

Answer:

- Maternal Azithromycin intake during pregnancy is a risk factor.

Q2.3 What are the findings that lead to the diagnosis of congenital hypertrophic pyloric stenosis?

Answer:

- Hypokalemic metabolic alkalosis with paradoxical aciduria
- USG s/o Thickened Pylorus

Q2.4 What is the treatment for congenital hypertrophic pyloric stenosis?

 PYQ: FMGE 2021

Answer:

Treatment:

- Isotonic saline with Potassium to correct dehydration and hypokalemia.

Definitive treatment:

- RAMSTED'S pyloromyotomy

Q2.5 A 5-year-old baby presents with recurrent episodes of non-bilious, projectile vomiting and hard pellet stools. On examination, 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?

Answer:

- Hypokalemic metabolic alkalosis with paradoxical aciduria, since the diagnosis is congenital hypertrophic pyloric stenosis

BIG QUESTION 3: Describe Celiac Disease, including its definition, diagnosis, role of biopsy, and treatment.

Broad Answer:

- Celiac disease, also known as gluten-sensitive enteropathy, is a T-cell-mediated autoimmune disorder triggered by ingestion of gluten-containing grains such as wheat, rye, and barley. The 2019 ESPGHAN guidelines recommend initial screening with anti-tTG IgA and total IgA levels, while **HLA-DQ2 or DQ8 typing** and symptoms are not mandatory for a biopsy-free diagnosis. Duodenal biopsy is still essential in some cases and is graded using the **Marsh classification** system. The mainstay of treatment is a lifelong gluten-free diet, which leads to clinical and histological recovery.

DETAILED QUESTIONS

Q3.1 What is celiac disease?

Answer:

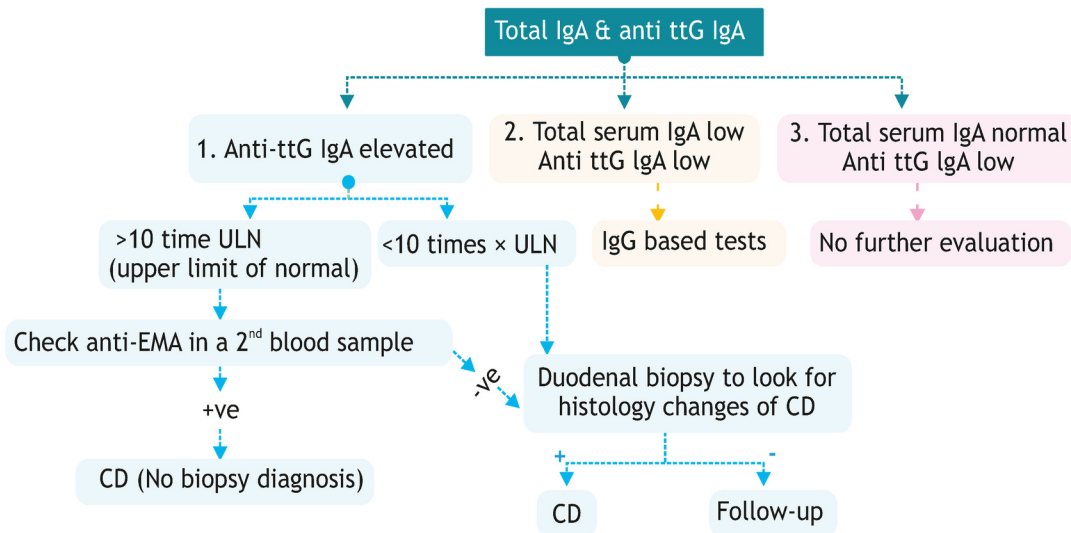
- Also known as Gluten Sensitive Enteropathy
- AT-Cell mediated autoimmune disorder in which intolerance to wheat/rye/barley containing gluten occurs.

Q3.2 How is celiac disease diagnosed?

PYQ: NEET PG 2021 PYQ: FMGE 2020

Answer:

- Latest ESPGHAN (European Society for Pediatric Gastroenterology, Hepatology & Nutrition) guidelines 2019
- For initial screening for CD: Combination of anti-tTG IgA and Total TgA

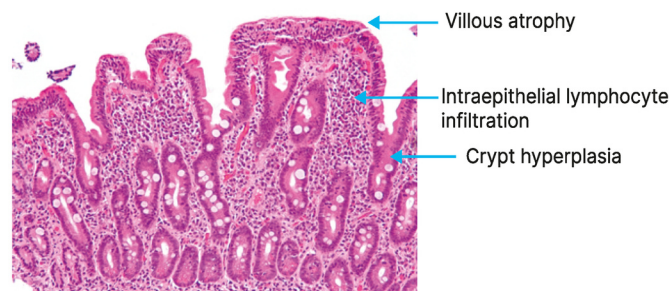


- HLA DQ2 or DQ8 testing & presence of symptoms are not obligatory criteria for “NO-Biopsy” diagnosis of CD

Q3.3 What is the role of Duodenal Biopsy in celiac disease?

Answer:

- It is a part of the Components of Marsh grading



Q3.4 What is the treatment for celiac disease?

Answer:

- Lifelong gluten-free diet

BIG QUESTION 4: Describe Intussusception, including its definition, clinical features, diagnosis, and treatment.

Broad Answer:

- Intussusception is the telescoping of one part of the gastrointestinal tract into an adjacent segment, most

commonly the ileum into the cecum. Clinically, it presents with severe colicky abdominal pain, passage of red currant jelly stools, and a sausage-shaped abdominal mass. Diagnosis is best established by ultrasonography, which is highly sensitive, while barium enema may show the characteristic “coiled spring” or “claw sign.” Initial management includes hydrostatic reduction under fluoroscopic or ultrasonographic guidance, while surgery is indicated in cases of shock, intestinal perforation, necrosis, peritonitis, or multiple recurrences.

DETAILED QUESTIONS

Q4.1 A 11-month-old baby was brought in with a history of incessant crying and abnormal stool with blood and mucus. A tender mass is palpable in the right abdominal region. What is the diagnosis?

Answer:

- Intussusception

Q4.2 What is intussusception? What are its clinical features?

Answer:

- When 1 portion of the GI tract is telescoped into another.

Clinical features:

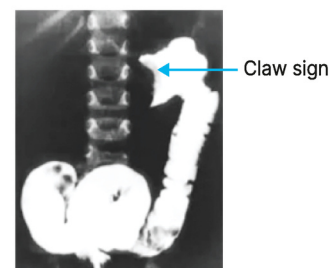
- Severe colicky pain in the abdomen + Red currant jelly stools
- Sausage-shaped palpable mass in the abdomen

Q4.3 How is intussusception diagnosed?

Answer:

- USG has good sensitivity in diagnosis.
- Barium enema shows a COILED Spring Sign or Claw sign.

 PYQ: FMGE 2021



Q4.4 What is the treatment for intussusception?

Answer:

- Spontaneous resolution occurs in 10% of the patients.
- Hydrostatic reduction under Fluoroscopic or USG guidance.
- Surgical reduction may be required in:
 - o Refractory shock
 - o Suspected intestinal perforation or necrosis.
 - o Multiple recurrences or peritonitis

Q4.5 What are the Common Childhood Poisonings?

Answer:

- The most common type is accidental poisoning
- Most common age affected- 1-5 years (Males > females)
- The most common route is ingestion > inhalation

 PYQ: FMGE 2021, 2022, 2023

 PYQ: NEET PG 2021, 2023

YOURWISH

Poison	Antidote
Organophosphate	Atropine + Pralidoxime
Paracetamol	N-Acetyl Cysteine
Benzodiazepine	Flumazenil
Opioids	Naloxone
Iron	Desferrioxamine
Heparin	Protamine Sulphate



10. PEDIATRIC RESPIRATORY DISORDERS

BIG QUESTION 1: What are the most common causes of key pediatric respiratory illnesses, and how are they presented, diagnosed, and managed?

Broad Answer: Common pediatric respiratory illnesses, such as pneumonia, bronchiolitis, asthma, and croup, are typically caused by infections (viral or bacterial) or airway hyperreactivity. They present with symptoms like cough, fever, and breathing difficulty, and are diagnosed clinically with support from imaging or lab tests. Management includes supportive care, antibiotics or antivirals when indicated, and bronchodilators or steroids for obstructive conditions.

DETAILED QUESTIONS

Q1.1 What is the most common cause of bacterial pneumonia in children?

Answer: *Streptococcus pneumoniae*

Q1.2 Which virus is the leading cause of bronchiolitis in infants?

Answer: RSV (Respiratory Syncytial Virus)

Q1.3 What is the most common cause of pneumonia in HIV-positive infants?

Answer: *Pneumocystis jirovecii*

Q1.4 What is the most common cause of stridor in infants?

Answer: Laryngomalacia

Q1.5 Which virus most frequently causes the common cold in children?

Answer: Rhinovirus

Q1.6 What is the most common causative organism responsible for croup?

Answer: Parainfluenza virus

Q1.7 What are the common causes of acute epiglottitis in immunized vs. unimmunized children?

Answer:

- Immunized: *Streptococcus* species
- Unimmunized: *Haemophilus influenzae* type b

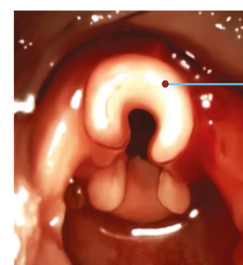
Q1.8 What are the features of laryngomalacia?

Answer:

- Omega-shaped epiglottis
- Loose and floppy
- Curled upon itself

🔍 PYQ: NEET PG 2021

🔍 PYQ: FMGE 2021

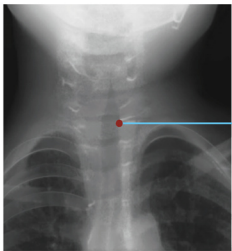
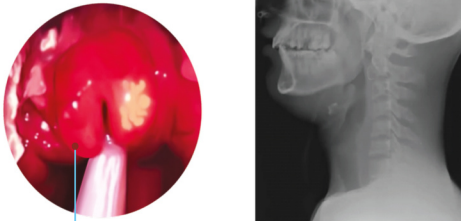


Omega shaped epiglottis

- Stridor may develop from 2 to 4 weeks
- Self-limiting disorder
- The most common cause of stridor in infants

Q1.9 What are the differences between croup and acute epiglottitis?

Answer:

	Croup/Acute laryngotracheobronchitis	Acute Epiglottitis
Clinical Features	• Low-grade fever, coryza, malaise • Stridor: Only on crying or at rest in severe cases • Breathing difficulty • Hypoxia may or may not be present	• High-grade fever • Sick or toxic-looking child • Stridor • Drooling of saliva • Severe respiratory distress • Hypoxia • Poor oral intake
Treatment	• No role of antibiotics • Supportive care, including O₂ • Mild cases: Single dose dexamethasone • Treatment of choice - Moderate to Severe cases: Single dose dexamethasone + Nebulized Epinephrine is used	• Supportive care: Oxygen, IV fluids • Treatment of choice: IV 3rd generation cephalosporin (Ceftriaxone)
	 Steeple sign	 Red, swollen congested epiglottis Thumb sign seen in acute epiglottitis

BIG QUESTION 2: What is the clinical approach to a child presenting with a cough or difficulty in breathing?

Broad Answer: The approach to a child with cough or breathing difficulty involves assessing severity (danger signs, respiratory rate, oxygen saturation), identifying probable causes (infections, asthma, foreign body, congenital anomalies), and initiating timely management, including supportive care, medications, or referral as needed.

DETAILED QUESTIONS

Q2.1: What parameters are assessed in a child with difficulty in breathing?

Answer:

- Count breaths in 1 minute
- Look for chest indrawing
- Look for stridor & wheeze
- Look for any danger signs

Q2.2: What are the criteria for fast breathing based on age?**Answer:**

- < 2 months: > 60/ min
- 2-12 months: > 50/ min
- 1-5 years: > 40/ min

Q2.3: What are the General Danger Signs in a child with a cough or difficulty in breathing?**Answer:**

- Convulsions
- Lethargy
- Inability to drink or breastfeed
- Persistent vomiting
- Unconsciousness
- Stridor in a calm child

Q2.4 What are the latest IMNCI guidelines for the Approach to a Child with Cough or Difficulty in Breathing?**Answer:**

Signs	Classify as	Treatment
Presence of General danger sign	• Severe Pneumonia or Very Severe Disease	• Give 1st dose of Inj. Ampicillin and Gentamycin and refer urgently to hospital
Presence of either chest indrawing &/or fast breathing or both but no general danger sign	• Pneumonia	• Start oral Amoxycillin 5 days • The child is sent home and assessed after 48 hours. • If the child has improved continue the same treatment. • If the child has worsened, refer to a higher center.
No Fast breathing, no chest indrawing, no general danger signs	• No Pneumonia; cough or cold only	• Home-based supportive care

Q2.5: How is wheezing managed?

Answer: Give a trial of bronchodilators (e.g., nebulized salbutamol)

Q2.6: When is a child referred for TB evaluation?

Answer: Cough >2 weeks

Q2.7: What is the indication for urgent referral to a higher center?

Answer: SpO₂ < 90%

BIG QUESTION 3: What are the diagnostic criteria and stepwise management of bronchial asthma in children, and how is status asthmaticus managed?

Broad Answer: Bronchial asthma is a chronic airway inflammation presenting with recurrent wheeze, cough, chest tightness, and dyspnea, often triggered by allergens, infections, or cold air. Management depends on severity, ranging from inhaled steroids with bronchodilators to emergency care for status asthmaticus with oxygen, systemic drugs, and advanced therapies.

YOURWISH

DETAILED QUESTIONS

Q3.1 What is bronchial asthma?

Answer: A chronic inflammatory disorder of airways, characterized by hyperresponsiveness of airways, leading to recurrent episodes of wheezing, cough, shortness of breath, and chest tightness.

Q3.2 What are the clinical features and examination findings of bronchial asthma?

Answer:

Clinical features:

Classical symptoms:	Severe disease:
• Cough, shortness of breath, wheeze, chest tightness ◦ More at night or early morning ◦ Exacerbated by allergens, exercise, and cold air ◦ Triggered by viral resp. infections	• Cyanosis, altered sensorium, and Respiratory distress

Q3.3 What are the key examination findings seen in asthma?

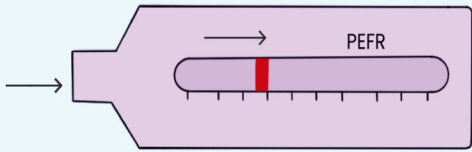
Answer:

- Increased work on breathing
 - Retractions
 - The child sits in the Tripod position
 - Grunting
- Hyperinflated chest
- Wheezing sound
- Percussion
 - Hyper-resonant chest
- Auscultation
 - Bilateral wheeze/Ronchi

Q3.4 What are the diagnostic tests available for asthma?

Answer:

 PYQ: INICET 2020

Pulmonary function tests (PFTs) or spirometry	• FEV_1/FVC: low(80%) • Bronchodilator responsiveness or reversibility: FEV_1 increases by 12% of baseline value following inhalation of short-acting β agonists. • Positive exercise challenge test: FEV_1 decreases by $\geq 12\%$.
Peak expiratory flow meters 	• Portable, hand-held devices • Used at home for monitoring PEFR • Increase diurnal variability of PEFR (peak expiratory flow rate) $> 13\%$

Q3.5 How is bronchial asthma managed in children?

Answer: As per the latest GINA (Global Initiative for Asthma), all children with Bronchial Asthma should receive inhaled corticosteroids along with SABA, either symptom-driven or daily.

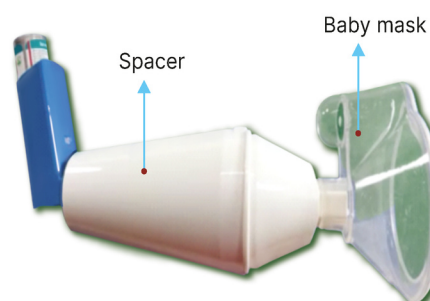
Q3.6 Write a short note on a metered dose inhaler.

Answer:

⌚ PYQ: NEET PG 2021 ⌚ PYQ: INICET 2022

Metered dose inhaler

- Used in drug delivery in children with bronchial asthma
- Parts
 - o Blue colored structure:
 - Metered dose inhaler
 - Contains bronchodilator
 - o White-colored structure:
 - Spacer
 - Must since it improves in drug delivery
 - o Baby mask: Required in < 4 years age children.



Q3.7 How is status asthmaticus in a child managed?

Answer:

- Also known as acute severe asthma
 - The pulmonary index score is used for the assessment of the severity
- Primary management of airway, breathing, and circulation (Chest x-ray and ABG to look for ventilation therapy)



Start oxygen therapy and admission.



Continuous nebulization with levosalbutamol/MDI



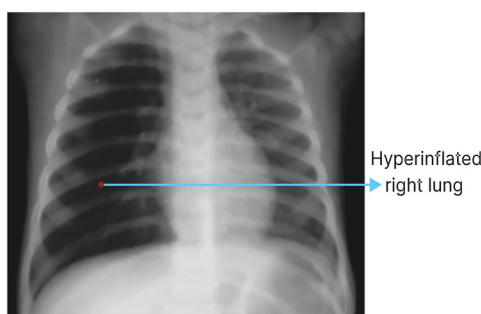
Start systemic steroids (Inj. hydrocortisone and later oral prednisolone). Ipratropium nebulization can be used.



In case of no improvement: Inj. Magnesium sulfate, theophylline, and ketamine are used.

Q3.8 A 2-year-old girl is brought in with a sudden onset of cough and difficulty in breathing. There is no history of fever. On probing, there was a history of choking while feeding. The X-ray is shown below. What is the diagnosis?

⌚ PYQ: FMGE 2022



Answer: Foreign body aspiration

Explanation

- Right lung
 - o More radiolucent
 - o Hyperinflation
- Indicates foreign body aspiration
- Method of choice for foreign body removal: Rigid bronchoscopy (Flexible bronchoscope is used nowadays)

Q.3.9 A 20-month-old girl presents with foul-smelling, bulky, greasy stools, intermittent rectal prolapse, and recurrent episodes of pneumonia. What is the probable diagnosis?

Answer: Cystic fibrosis

Q3.10 What are the diagnostic criteria and treatment of cystic fibrosis?

 PYQ: INICET 2023

Answer:

Any One of:

- Clinical features characteristic of CF
 - or
- History of CF in a sibling
 - or
- +ve NB screening for CF [IRT- Immuno-Reactive Trypsinogen]

And Any One of

- Sweat chloride (>60 mmol/L) on 2 or more occasions
 - or
- 2 mutations detected in the CFTR gene (Delta F508 is the most common mutation)
 - or
- Abnormal nasal transepithelial Potential difference

Q3.11: What are the treatment options available for cystic fibrosis?

Answer:

- Treat the infections, Enzyme replacement therapy, and potentiators like IVACFTOR can be used.



11. PEDIATRIC NEPHROLOGY

BIG QUESTION 1: What are the key pediatric kidney disorders including nephrotic and nephritic syndromes, and how are they differentiated and managed?

Broad Answer: Nephrotic Syndrome is marked by massive proteinuria, hypoalbuminemia, edema, and hyperlipidemia – most commonly caused by Minimal Change Disease in children. Treated with prednisolone.

- Nephritic Syndrome presents with hematuria, hypertension, mild edema, and proteinuria in nephritic range, most commonly due to PSGN.
- HUS is a classic triad of MAHA, thrombocytopenia, and AKI, often post-diarrhea due to Shiga toxin-producing *E. coli*.
- AKI is staged by KDIGO, and PUJ Obstruction and Posterior Urethral Valves are key congenital causes of hydronephrosis and obstructive uropathy in children.

DETAILED QUESTIONS

Q1.1 What is the most common cause of Nephrotic syndrome in children?

Answer: MC cause of Nephrotic syndrome in children is Minimal change disease.

Q1.2 What is kidney biopsy finding in nephrotic syndrome?

Answer: Normal on light microscopy and flattened footplate of podocytes on electron microscopy)

Q1.3 What are the components of nephrotic syndrome?

Answer:

- Nephrotic range/massive proteinuria (Up : Uc ratio ≥ 2)
- Generalized edema
- Hypoalbuminemia
- Hyperlipidemia

Q1.4 What is the drug of choice and dose for nephrotic syndrome?

Answer:

- DOC: Prednisolone
- Dose: 2mg/kg/day daily for 6 weeks, followed by 1.5 mg/kg/day every alternate day for the next 6 weeks (80%-90% of children respond within 2-3 weeks)

Q1.5 Based on the classification of steroid response patterns in childhood nephrotic syndrome, what are the various treatment options available?

Answer:

IFRNS	• Treat each relapse • Prednisolone 2mg/kg/day till remission or 60 mg/m²/day daily till remission followed by 4 weeks of alternate day steroids (1.5 mg/kg)
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YOURWISH

FRNS (or) SDNS	• Tapering doses of alternate-day steroids Steroid threshold <0.5 mg/kg alternate day • Continue long-term alternate dose alternate day prednisolone for 9-18 months Steroid threshold >0.5 mg/kg alternate day (or) features of steroid Toxicity • Levamisole • Oral Cyclophosphamide • Mycophenolate Mofetil
SRNS	• Drug of choice: Calcineurin inhibitors (Cyclosporine/Tacrolimus)
Refractory cases	• Rituximab (Anti CD20 Ab)

Q1.6 What is the most common cause of nephritic syndrome in children?

Answer: PSGN (Post-streptococcal Glomerulonephritis)

Q1.7 Define nephritic syndrome.

Answer:

Sudden onset of	Gross hematuria - Cola-colored urine
	Proteinuria - Nephritic range (Up: Uc:- 0.2-2)
	Mild Edema
	Hypertension
	With or without renal dysfunction

Q1.8 How is nephritic syndrome diagnosed?

Answer:

- Urine examination - Proteinuria, RBC, RBC casts, and Active sediments.
- C level decreased in 90% of patients, but normalized in 6-8 weeks of onset
- Increased ASO (Anti Streptolysin O) titer following pharyngitis
- Increased Anti DNase B the following pyoderma

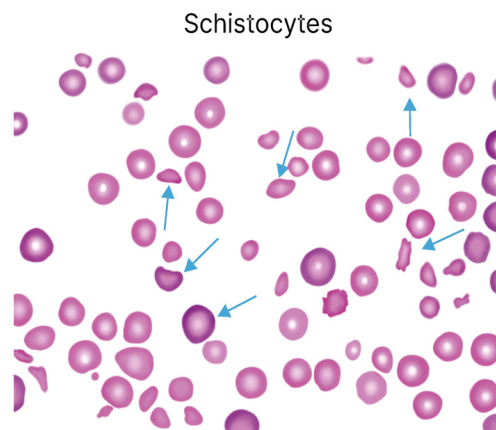
Q1.9 What are the probable differential diagnoses in a child with hematuria?

Answer:

Hematuria in a child	
Number of days after URI	Probable underlying diagnosis
• 2-3 days • 2-3 weeks	• IgA nephropathy • PSGN

Q1.10 A 3-year-old child with diarrhoea presents with a history of epistaxis and not passing urine for the last 12 hrs. 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 5mg/dl, TLC is 10,800/mm³, and platelet count is 36,000/mm³. The peripheral smear shows the following picture. What is the probable diagnosis?

Answer: Hemolytic Uremic Syndrome (HUS)



Q1.11 Describe Hemolytic Uremic Syndrome (HUS)

Answer:

- Endothelial injury triggers the other events in HUS leading to a triad of injuries.
 - o Microangiopathic Hemolytic Anemia
 - o Thrombocytopenia
 - o Acute kidney injury

Q1.12 What are the types of HUS and specify the treatment for HUS

Answer:

Types of HUS	
Classical (D+ HUS) • Secondary to infection by Shiga toxin-producing E. coli or Shigella dysentery. • Secondary to infection by Streptococcal infection or Malaria.	Atypical (D- HUS) • Excess of activation or insufficient inhibition of alternative complement pathway • E.g.: Factor H deficiency or factor I deficiency or the presence of Anti-factor HAb

Treatment of HUS

- Plasmapheresis/Therapeutic plasma exchange
- Eculizumab: Useful in cases of refractory atypical HUS

Q1.13 Describe the KDIGO Staging of AKI

Answer: KDIGO: Kidney Disease Improving Global Outcomes

PYQ: FMGE 2020

Stage	Serum creatinine	Urine output
1	≥ 0.3 mg/dl increase or 1.5-1.9 times baseline	< 0.5 ml/kg/hr for 6-12 hrs
2	2-2.9 times baseline	< 0.5 ml/kg/hr for >12 hrs
3	≥ 3 times baseline (or) serum creatinine ≥ 4 mg/dl (or) indication of Renal replacement therapy (or) eGFR < 35 ml/min/1.73m ²	< 0.3 ml/kg/ml for ≥ 24 hrs (or) Anuria for ≥12 hrs

Q1.14 How is the Bedside assessment of GFR done?

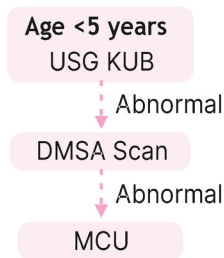
Answer:

- Schwartz formula

$$eGFR = \frac{(k \times Ht \text{ (CM)})}{\text{Serum creatinine (mg/dl)}} \quad \text{where } k = 0.413 \text{ in children}$$

Q1.15 Which is the most common cause of hydronephrosis in children?**Answer:** Pelviureteric Junction Obstruction (PUJD)**Q1.16 Which is the most common cause of obstructive uropathy in boys, and how is it diagnosed?****Answer:**

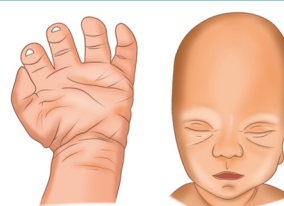
- Posterior Urethral Valves (PUV)
- Diagnosed by
 - o Strain during urination
 - o Improper urinary stream
 - o Palpable urinary bladder

Q1.17 Which is the most common cause of renal scarring in children?**Answer:** Reflux-induced nephropathy**Q1.18 Which is the most common cause of UTI in children?****Answer:** E-coli > Klebsiella & Proteus**Q1.19 What is the Recommended Imaging in Children with UTI?****Answer:****Q1.20 What is Potter Syndrome/Sequence?****Answer:** B/L renal agenesis (incompatible with life)**Q1.21 Which is the most common cause of mortality in Potter syndrome?****Answer:** Pulmonary hypoplasia**Q1.22 What are the clinical features of Potter syndrome?****Answer:**

- Extremities anomaly
- Wrinkled skin
- Facial dysmorphism
- Renal agenesis B/L (primary anomaly)
- Oligohydramnios

Q1.23 Explain Potter facies.**Answer:**

- Eyes widely separated.
- Epicanthic folds
- Flat facies
- High forehead
- Receding chin
- Washer man's hand



Potter facies



12. PEDIATRIC NEUROLOGY

BIG QUESTION 1: Name some of the neural tube defects.

Broad Answer:

- Anencephaly
- Iniencephaly
- Craniorachischisis
- Myelomeningocele
- Meningocele

DETAILED QUESTIONS

Q1.1: What is the most common congenital abnormality of the neurologic system in children?

Answer: Neural Tube Defects

Q1.2: How are Neural tube defects diagnosed?

Answer:

- Antenatal USG: Can pick up large NT defects
- Maternal serum or Amniotic fluid markers
- α -fetoprotein levels or Acetylcholinesterase levels
- Neuroimaging to know the exact extent of the malformation

Q1.3: What is its underlying developmental cause of Anencephaly?

PYQ: FMGE 2020

Answer: Anencephaly occurs due to the failure of closure of the rostral neuropore (the opening of the anterior neural tube)



Q1.4: What is Iniencephaly, and what are its key clinical features?

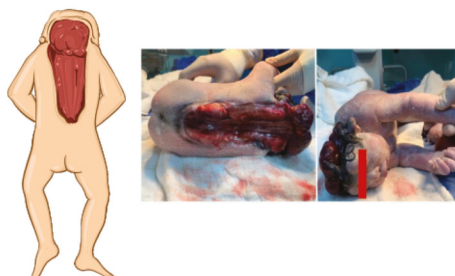
Answer:

- In Iniencephaly, the brain and upper spinal cord are exposed to the outside
- Retroflexed head with absent neck



Q1.5: What is Craniorachischisis, and how does it present?

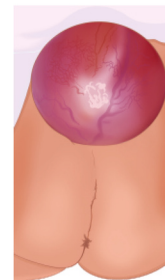
Answer: In Craniorachischisis, the cranium and entire neuraxis are exposed



YOURWISH

Q1.6: What is the basic defect in and most common location of Myelomeningocele?**Answer:**

- Basic defect: Protrusion of neural components along with a sac formed by meninges through a midline vertebral defect
- Most common location: Lumbosacral area

**Q1.7: What is the treatment approach for Myelomeningocele in a newborn?****Answer:**

- Surgery
- Ruptured myelomeningocele
 - o Swabs should be collected to check for any bacterial growth, and IV antibiotics should be started
 - o Emergency surgery
 - o The swelling can be covered with saline-soaked gauze to prevent it from drying

Q1.8: What is a meningocele, and what are its distinguishing features?**Answer:**

- In a Meningocele, there is a protrusion of a meningeal sac from a vertebral defect containing only clear fluid and no neural components
- Transillumination is positive

Q1.9: What measures are taken to prevent Neural tube defects?

PYQ: INICET 2022

PYQ: FMGE 2022

Answer:

Folic Acid Supplementation	
400 µg/day in all women of childbearing age.	4000 µg/day in high-risk women.
Started at least one month before conception.	Decrease the risk of neural tube defects by 70%

BIG QUESTION 2: How do you define febrile seizures?

Broad Answer: Seizure with significant fever ($\geq 100.4^{\circ}\text{F}$) without any evidence of CNS infection in the age group of 6 months - 6 years

DETAILED QUESTIONS**Q2.1: What is the most common cause of seizures in children under 5 years of age?**

Answer: Febrile seizures are the most common cause of seizures in children <5 years of age.

Q2.2: What are the types of febrile seizures?**Answer:**

Types	
Simple Febrile Seizures • Generalized seizures • Lasts <15 minutes • No recurrence within 24 hrs	Complex Febrile Seizures • Focal seizures • Fifteen minutes or longer • Frequent (recurrence may be seen)

Q2.3: What is the management approach for febrile seizures in children?**Answer:**

- Make the child lie down in the left lateral (recovery position)- Do not give anything orally
- In cases where the seizure lasts > 5 minutes
 - o Home Management: Rectal/Buccal/Nasal Midazolam
 - o Hospital Management: IV lorazepam or Midazolam
- No long-term anti-epileptics are recommended in the management of simple-type febrile seizure
- Recurrent episodes- Intermittent prophylaxis with Clobazam
- Paracetamol to control fever and make the child more comfortable
- Complex febrile seizure- Long-term anti-epileptics like Valproate

Q2.4: What are the factors increasing recurrence risk in a child with febrile seizures?**Answer:**

- Family history of Febrile seizures
- Complex febrile seizures
- Lower serum Na⁺ level at the time of presentation
- Age <1 yr at onset
- Temperature 100.4° -102.2° F
- Duration of fever <24 hrs

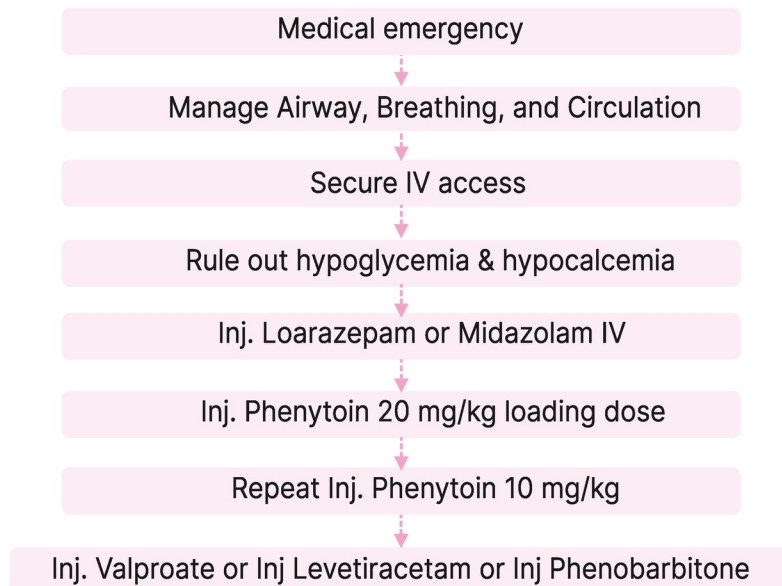
Q2.5: What are the risk factors for developing epilepsy in a child with febrile seizures, and how do they affect the risk percentage?**Answer:**

Risk factors	Risk for epilepsy (%)
Simple febrile seizures	1%
Complex febrile seizures	6%
Fever of < 1 hr before febrile seizure	11%
Family H/O epilepsy	18%
Focal febrile seizure	29%
Pre-existing neurodevelopment abnormalities	33%

BIG QUESTION 3: What is the operational definition of Status epilepticus in children?

Broad Answer: Any seizure lasting for >5 min or multiple episodes of seizures without regaining consciousness in between episodes, or a child brought with ongoing seizures to a medical facility.

DETAILED QUESTIONS
 **PYQ: INICET 2021**
Q3.1: How is Status epilepticus in children managed?**Answer:**



Q3.2: What are the drugs of choice for various pediatric neurological conditions?

Answer:

- Absence seizures: Valproate/Ethosuximide
- Juvenile myoclonic epilepsy: Valproate
- West syndrome: Inj. ACTH
- Infantile spasms in Tuberous Sclerosis: Vigabatrine

Q3.3: What are the classical features of West syndrome in infants?

🔍 PYQ: NEET PG 2022

🔍 PYQ: INICET 2022

Answer:

- Infantile spasms (Salaam Attacks/flexor spasms)
- Global developmental delay
- Hypsarrhythmia (on EEG)

BIG QUESTION 4: A premorbidly developmentally normal, unimmunized child presents with gradually deteriorating performance in school and frequent falls. There is a history of measles during childhood. On examination, the child has myoclonic jerks, and the EEG shows burst suppression. What is the diagnosis?

Broad Answer:

- Subacute Sclerosing Panencephalitis (SSPE).
- History of measles infection in childhood, Gradual cognitive decline (poor school performance), Frequent falls and myoclonic jerks, and EEG: Burst suppression pattern (classically seen in SSPE)

DETAILED QUESTIONS

Q4.1: What is SSPE, and when does it typically develop after primary measles infection?

Answer:

- Subacute Sclerosing Panencephalitis is a rare, fatal, long-term complication of measles
- Develops 7-10 years after 1° measles infection.

Q4.2: What are the clinical features and diagnostic methods of Subacute Sclerosing Panencephalitis?

Answer:

- Initially, it is characterized by decreasing school performance, then there is the appearance of myoclonic jerks, sudden falls, choreoathetosis, dystonia, and rigidity
- Neurodegenerative disorder with a progressive downhill course
- Diagnosis: CSF and Blood anti-measles antibody is done

BIG QUESTION 5: A 7-month-old boy presents with a history of fever for 3 days along with irritability, poor feeding, and 2 episodes of seizures. On examination, the baby has a tense, bulging fontanelle. What is the probable diagnosis?

Broad Answer:
 **PYQ: FMGE 2020**

- Acute Bacterial Meningitis.**
- Infant presenting with fever, irritability, poor feeding, seizures, along with a tense, bulging fontanelle → indicates raised intracranial pressure are constellation of findings for meningitis in infants

DETAILED QUESTIONS**Q5.1: What is the clinical presentation of Acute bacterial meningitis?****Answer:**

Non-specific features	- Fever, anorexia, poor feeding, lethargy, irritability with shrill cry, petechiae, purpura - Older children: headache, photophobia, myalgia & arthralgia
Signs of meningeal irritation are beyond 12-18 months of age	- Neck rigidity - Meningeal signs - Kernig's sign - Brudzinski's sign - Older children- Headache, blurring of vision, diplopia - Younger infants- Bulging fontanelle, Widening of sutures - Fundus examination: Papilledema - Focal neurological signs may be seen - Seizures - Altered sensorium/ encephalopathy

Q5.2: How is Acute bacterial meningitis treated?**Answer:**

- Prompt initiation of empirical antibiotics
 - IV 3rd generation Cephalosporin (Ceftriaxone) ± Vancomycin

BIG QUESTION 6: A 7-day-old male baby presents with tense bulging fontanelle, bounding pulses, and features of heart failure. On auscultation over the anterior fontanelle, a cranial bruit is heard. What is the probable diagnosis?

Broad Answer:

- Vein of Galen Malformation (VOGM).**
- Neonate with tense, bulging fontanelle, bounding pulses + features of heart failure (due to high-output cardiac failure from arteriovenous shunt) and Cranial bruit heard over anterior fontanelle → highly suggestive of a vascular malformation
- These are classical causes of a Vein of Galen arteriovenous malformation

YOURWISH

DETAILED QUESTIONS

Q6.1: What is the basic defect in Vein of Galen Malformation, and how can a neonate present clinically?

Answer:

- Basic defect: Due to a precursor of the vein of Galen, which is the medial prosencephalic vein of Markowsky (tortuous and dilated)
- The neonate could present with features of heart failure

BIG QUESTION 7: What is the definition of brain death in children?

Broad Answer: Brain death is an irreversible cessation of all functions of the entire brain, including the brain stem

DETAILED QUESTIONS

Q7.1: What are the common causes of brain death in children?

Answer: Brain Death in children is usually due to trauma or asphyxial brain injury

Q7.2: What are the key diagnostic components of brain death in children?

Answer:

Irreversible coma with a known cause	
Apnea: Absence of respiratory efforts in response to an adequate stimulus ($\text{PCO}_2 > 60$ mm Hg)	Absence of brain stem reflexes (light reflex, corneal reflex, gag reflex)

- All these Findings must remain consistent for 2 observations separated by an observation period of 24 hours - 48 hours

Q7.3: Which clinical features are incompatible with the diagnosis of brain death?

Answer:

- Decerebrate/Decorticate posturing
- Presence of seizures
- Extensor or flexor response to a painful stimulus

BIG QUESTION 8: Name some of the Neurocutaneous disorders in children.

Broad Answer:

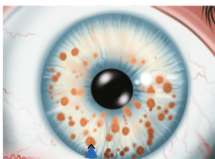
- Neurofibromatosis 1
- Tuberous Sclerosis
- Sturge-Weber syndrome

DETAILED QUESTIONS

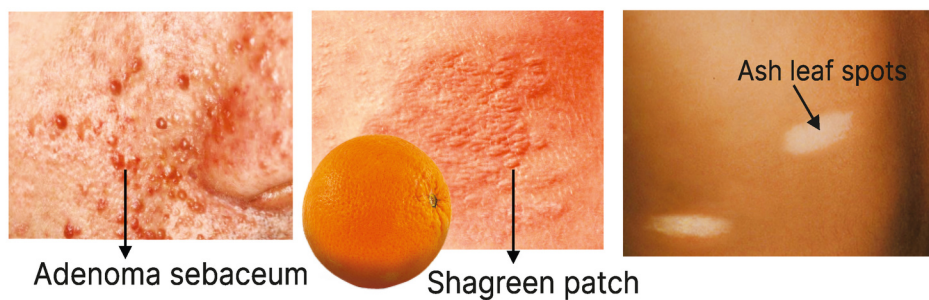
Q8.1: What is the inheritance pattern and distinguishing features of Neurofibromatosis 1?

PYQ: NEET PG 2019

Answer:

Inheritance	Autosomal dominance
 Cafe au lait spots	 Lisch nodules

Q8.2: A child with Intellectual disability and seizures has the following features shown in the images below. What is the diagnosis?



Answer: Tuberous Sclerosis

Q8.3: What are the clinical features of Sturge-Weber syndrome?

Answer:

- Unilateral Port wine stain of the face (presence of Hemangiomas) usually in the areas innervated by the ophthalmic and Trigeminal nerves
- Seizures

Pigmentation involving the
Ophthalmic and maxillary
area of trigeminal nerve





13. MUSCULOSKELETAL DISORDERS IN CHILDREN

BIG QUESTION 1: A 4-year-old boy presents with difficulty in climbing stairs. There is a family history of a similar illness in the child's maternal uncle. The Child's CPK levels are 12,400 units/L. What is the probable diagnosis?

Broad Answer:

PYQ: NEET PG 2020, 2021

- Duchenne Muscular Dystrophy (DMD).
- Difficulty in climbing stairs shows proximal muscle weakness
- Family history of maternal uncle indicates X-linked recessive inheritance
- Very high CPK (12,400 U/L) → typical of DMD

DETAILED QUESTIONS

Q1.1: Which gene is affected in Duchenne Muscular Dystrophy, and what is its mode of inheritance?

Answer:

- Gene affected: Dystrophin gene; one of the largest genes found in humans, and it is present on Chr Xp21
- Mode of inheritance: X-linked recessive inheritance

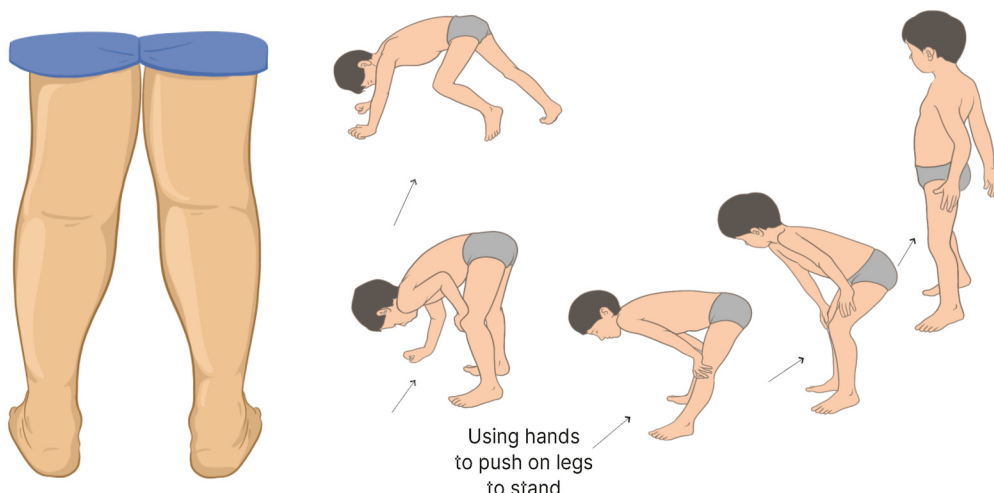
Q1.2: What are the clinical features of Duchenne Muscular Dystrophy?

Answer: Progressive proximal muscle weakness i.e., weakness involving thigh muscles, arm muscles

Q1.3: What are the clinical signs of Duchenne Muscular Dystrophy?

Answer:

- On examination, Pseudohypertrophy of calf muscles is seen, giving an inverted bottle appearance
- **Valley sign positive:** It is a groove-like depressed area between pseudo-hypertrophied deltoid and infraspinatus muscles
- **Gower sign positive:** If a child with DMD is asked to get up from a sitting position, he will first take the help of a hand and put them on the ground and take support from his legs, and slowly, taking help from himself, the child will get up



Q1.4: What are the investigative findings in Duchenne Muscular Dystrophy?**Answer:**

- Serum CPK levels (Normal - < 160 U/L): Markedly elevated; > 10,000 U/L
 - This is often used as a screening and first-line investigation
- Definitive diagnosis: Demonstration of mutation in the dystrophin gene
 - It can be done by multiplex PCR or MLPA (Multiplex Ligation-dependent Probe Amplification)
- Chest X-ray: Cardiomegaly

Q1.5: What is the treatment approach for Duchenne Muscular Dystrophy (DMD)?**Answer:**

- Supportive care, including physiotherapy, taking care of cardio-respiratory issues
- Corticosteroids: They decrease the progression of the disease, but they can not cure the disease
 - It will improve muscle strength and prolong ambulation.
 - Prednisolone or Deflazacort can be used
- Newer drugs: Anti-sense oligonucleotides
- Eteplirsen (exon-skipping drug)

BIG QUESTION 2: What is rickets, and in which group of children does it occur?
 **PYQ: FMGE 2020**

Broad Answer: Rickets is a disease of growing bones due to defective mineralization of the bone matrix at the growth plate in children before the fusion of the epiphysis.

DETAILED QUESTIONS**Q2.1: What is the most common cause of Rickets?**

Answer: The most common cause of Rickets is a nutritional deficiency of Vitamin D.

Q2.2: What are the causes of Nutritional Rickets?**Answer:**

- Vitamin D deficiency
 - Congenital deficiency
 - Inadequate dietary intake
 - Malabsorption
 - Liver / Kidney Disease
- Calcium deficiency
- Phosphate deficiency

Q2.3: What is Refractory Rickets, and its types?**Answer:**

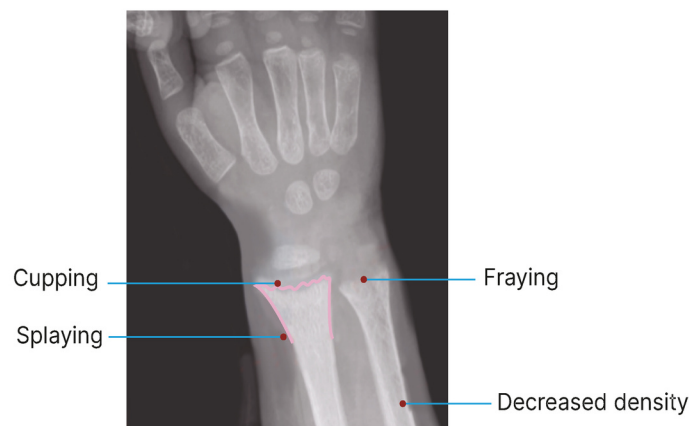
- Rickets that does not respond to the usual treatment of nutritional Rickets
 - VDDR types I and II
 - Hypophosphatemic Rickets(XLD): PHEX gene
 - Chronic kidney disease (CKD): Increased Phosphate level
 - Renal tubular acidosis (proximal / distal)
 - Oncogenic / Tumor-induced
 - Some benign mesenchymal tumors: Secrete FGF-23 → Phosphaturia & hypophosphatemia

YOURWISH

Q2.4: Explain in detail the clinical features of Rickets.

Answer:

General • Failure to thrive. • Protruded abdomen • Listlessness • Increased risk of respiratory infections: softening of ribs, impairs air movement during respiration • Increased risk of fractures	Head and face • (Earliest)Craniotabes: Due to softening of cranial bones; also seen in osteogenesis imperfecta, congenital syphilis, hydrocephalus, and prematurity. • Large AF with delayed closure • Frontal and parietal bossing • Delayed dentition and dental caries • Delayed motor milestones
Chest • Rachitic rosary: Beading of costochondral junctions • Harrison sulcus: Due to the pulling of softened ribs by the diaphragm	Limbs • Wrist widening • Genu varum or valgum deformity • Bowing of tibia • Double malleoli
Hypocalcaemia • Tetany • Seizures • Stridor • Trousseau sign (more specific) • Chvostek sign	



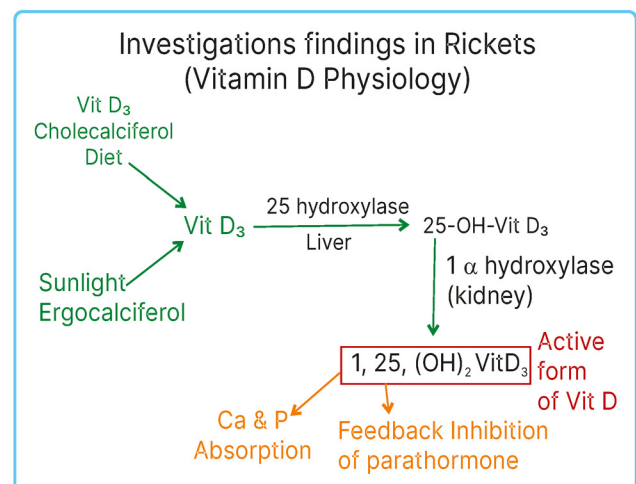
PYQ: INICET 2020

PYQ: NEET PG 2021

Q2.5: What are the investigative findings in Rickets?

Answer:

- Serum Calcium Normal or low
- Serum Phosphate: Normal or Low
- PTH
- Alkaline phosphatase: High
- 25 (OH) Vit D₃: Low (< 10 ng/ml or < 30 nmol/L)
 - o Normal: > 20 ng/ml or > 50 nmol/L



Q2.6: How can different types of rickets be differentiated based on biochemical findings?

Answer:

S. Ca	S.PO4	Alk Phos	25(OH) Vit D ₃	1,25, (OH) Vit D ₃	PTH	
N/↓	N/↓	↑	↓	↓	↑	Nutritional Rickets
↓	↓/N	↑	(N)	↓	↑	VDDR Type 1
↓	↓/N	↑	(N)	(↑)	↑	VDDR Type 2
N	↓	↑	N	N	N/↑	Hypophosphatemia Rickets
N/↓	↑	↑	N	N/↓	N/↑	Chronic Kidney Disease

BIG QUESTION 3: A 1-year-old exclusively cow milk-fed boy presented with crying with touching of limbs, gum bleeding, and inability to move the right lower limb. What is the probable diagnosis?

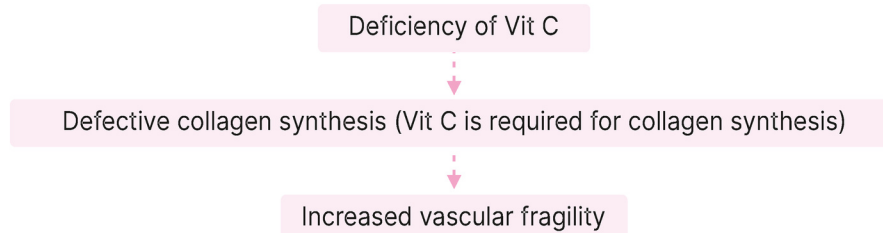
Broad Answer:

- Scurvy (Vitamin C deficiency)
- Exclusively cow's milk-fed (cow's milk is deficient in Vitamin C), Subperiosteal hemorrhage, Gum bleeding (classic for scurvy), and Pseudoparalysis (inability to move limb) due to painful hemorrhages all indicate a diagnosis of Scurvy

DETAILED QUESTIONS

Q3.1: What is the basic defect in Scurvy?

Answer:

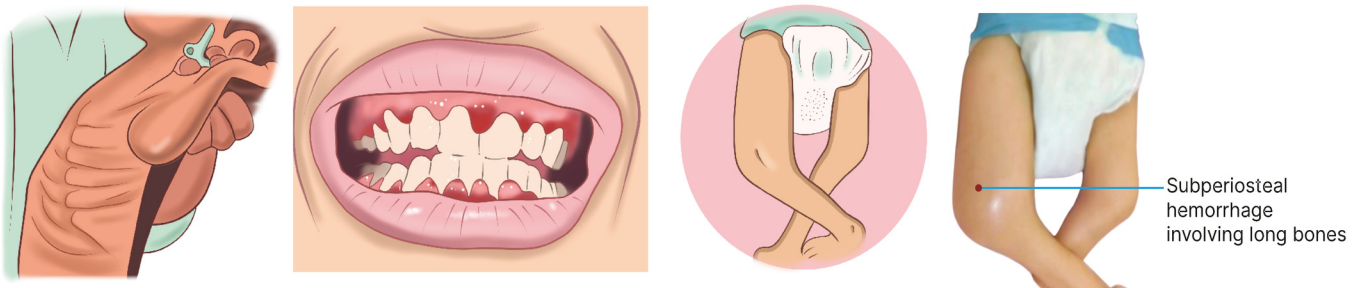


Q3.2: Who is at major risk of developing Scurvy?

Answer: Children predominantly fed on cow's milk

Q3.3: What are the clinical features of Scurvy?

Answer:

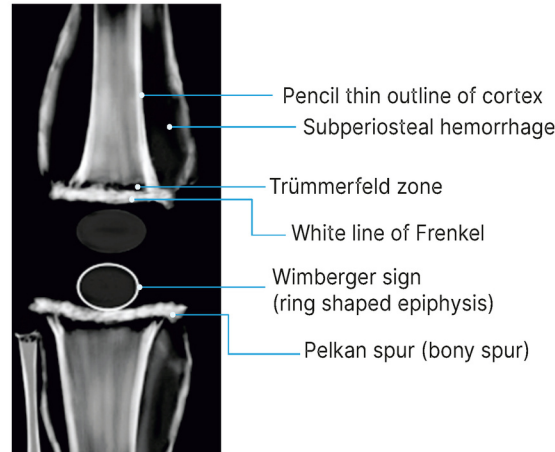


- Gum bleeding
- Petechiae

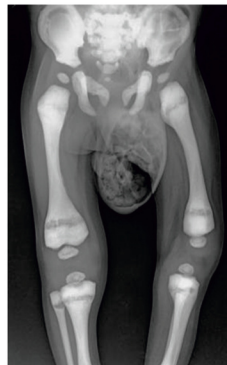
- Subperiosteal haemorrhage involving long bones
 - o Painful pseudoparalysis
 - o Crying on touch
- Scorbutic Rosary

Q3.4: What are the features noted on X-ray in a Scurvy patient?

Answer:



BIG QUESTION 4: An 11-month-old girl presented with pancytopenia and hepatosplenomegaly. An X-ray of her limbs showed the following picture. What is the diagnosis?



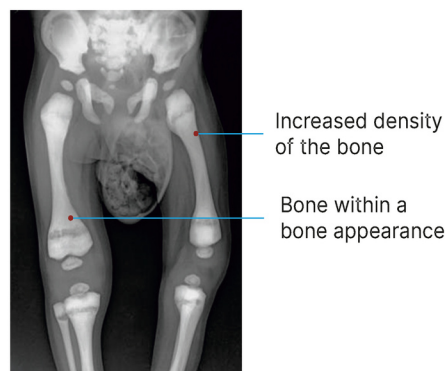
Broad Answer:

- Osteopetrosis (Marble Bone Disease)
- Pancytopenia + Hepatosplenomegaly (due to extramedullary hematopoiesis) along with X-ray showing “bone within bone” appearance

DETAILED QUESTIONS

Q4.1: What are the X-ray findings noted in Osteopetrosis (Marble Bone Disease)?

Answer:



BIG QUESTION 5: A baby was born with the limb defects shown in the picture below. There is a history of thalidomide ingestion by the mother during her pregnancy. The probable diagnosis is?

Broad Answer:

- Phocomelia
- Phocomelia is a classic thalidomide-induced congenital malformation

DETAILED QUESTIONS

Q5.1: What is Phocomelia, and what is its common risk factor?

Answer:

- Limbs resemble the flippers of a seal
- Maternal intake of thalidomide during pregnancy is a risk factor
- Short proximal segments of limbs



BIG QUESTION 6: A 15-month-old baby presented with multiple bony deformities and deafness. On enquiring, there was a recurrent history of limb fractures following trivial trauma in the child. On close examination, the child has a blue sclera. What is the probable diagnosis?

Broad Answer:

- Osteogenesis Imperfecta
- Blue sclera, along with recurrent limb fractures, is indicative of Osteogenesis Imperfecta

DETAILED QUESTIONS

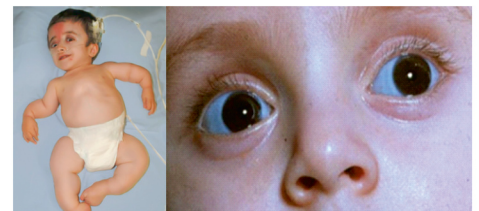
Q6.1: What is the inheritance pattern of Osteogenesis Imperfecta?

Answer: The most common mode of inheritance in osteogenesis imperfecta: AD

Q6.2: What is the clinical triad of Osteogenesis Imperfecta?

Answer:

- Blue sclera
- Limb deformities due to recurrent fractures
- Deafness



Brittle Bone Disease

Q6.3: How is Osteogenesis imperfecta treated?

Answer: Treated with Bisphosphonates like pamidronate

BIG QUESTION 7: A 5-year-old girl presented with pain and swelling in the right knee for 3 months. There is also a history of pain in the right eye, on and off. What is the diagnosis?

Broad Answer:

- Juvenile Idiopathic Arthritis
- Chronic monoarthritis, along with Eye involvement, is suggestive of Juvenile Idiopathic Arthritis

DETAILED QUESTIONS

Q7.1: What is Juvenile Idiopathic Arthritis?

Answer: Juvenile Idiopathic Arthritis is an arthritis of ≥ 1 joint, lasting for at least 6 weeks, in a child < 16 years of age

YOURWISH

Q7.2: What are the types of Juvenile Idiopathic Arthritis?**Answer:**

Types		
Oligo articular • 4 or less joints involved • The most common type associated with Uveitis/iridocyclitis • More common in girls	Poly articular • > 4 joints involved	Systemic onset • Fever, rash hepatosplenomegaly

Q7.3: How is Juvenile Idiopathic Arthritis treated?**Answer:**

- Disease-modifying agents like weekly Methotrexate
- In severe cases: Steroid pulse therapy

BIG QUESTION 8: A 4-year-old 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 bloody stools 4 weeks ago. What is the probable diagnosis?

Broad Answer:

- **Reactive Arthritis**
- Joint swelling with pain and restrictive movements, along with a history of infection, is indicative of Reactive Arthritis

DETAILED QUESTIONS**Q8.1: What is Reactive Arthritis?**

Answer: Reactive arthritis is joint inflammation caused by a sterile inflammatory reaction following a recent enteropathic or urogenital infection

Q8.2: How is Reactive arthritis treated?**Answer:**
 **PYQ: NEET PG 2019**

- Physical therapy and rehabilitation
- NSAIDs
- Intra-articular steroid injection in severe cases

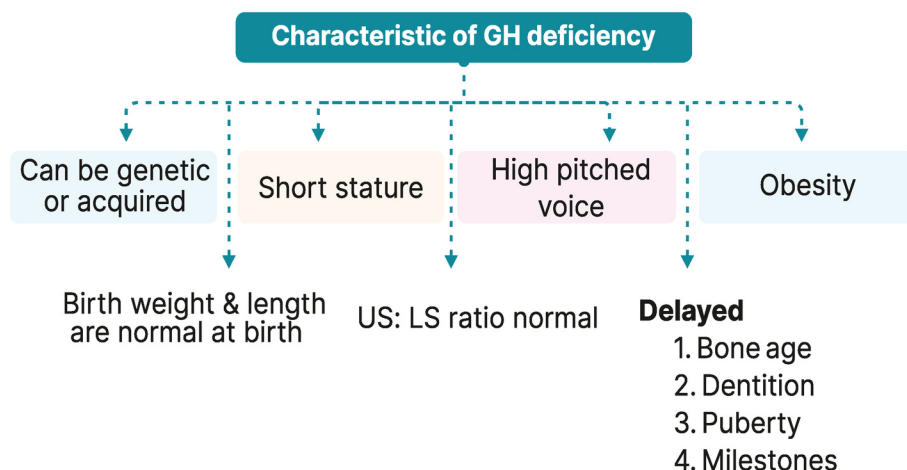


14. PEDIATRICS ENDOCRINOLOGY

BIG QUESTION 1: Briefly describe the characteristics of GH deficiency

PYQ: NEET PG 2021

Broad Answer:



DETAILED QUESTIONS

Q1.1: A 5-year-old baby presents with recurrent hypoglycemia, short stature, and micropenis. His bone age is found to be 2.5 years. What is the diagnosis?

Answer: Congenital hypopituitarism

Q1.2: Does Growth Hormone (GH) play a role in the growth of the fetus?

Answer: GH does not play a role in the growth of the fetus

Q1.3: What is the primary diagnostic test for Isolated GH Deficiency?

Answer:

- GH stimulation test
- Done using arginine/clonidine/insulin glucagon, levodopa

Q1.4: What peak GH level in a stimulation test indicates GH deficiency?

Answer: Peak GH level <10 ng/ml

Q1.5: How does bone age compare to chronological age in GH deficiency?

Answer: Bone age < Chronological age.

Q1.6: How is recombinant GH administered, and what is the typical frequency?

Answer: Subcutaneously for 5 days /week with a gap of 2 days

Q1.7: What is an important consideration regarding the timing of GH treatment initiation?

Answer: Should be given before the fusion of the epiphysis

Q1.8: How long should GH treatment be given for results to become apparent?

Answer: For at least 1-2 years.

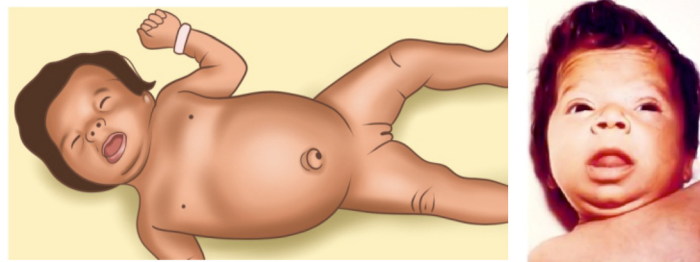
Q1.9: List the potential adverse effects of GH therapy.

Answer: Pseudotumor cerebri, Gynecomastia, Impaired glucose tolerance

Q1.10: A 4-month-old male baby presents with a puffy-looking face, abdominal distension, umbilical hernia, constipation, and prolonged neonatal jaundice. There is also a hoarse cry and hypotonia. What is the diagnosis?

PYQ: FMGE 2019

Answer: Congenital Hypothyroidism



Q1.11: What is the most common preventable/treatable cause of mental retardation/intellectual disability in children?

Answer: Congenital Hypothyroidism

PYQ: FMGE 2020

Q1.12: What is the most common cause of congenital hypothyroidism?

PYQ: INICET 2021

Answer: Thyroid dysgenesis

PYQ: FMGE 2020

Q1.13: What is the most common cause of congenital hypothyroidism in a child with goitre?

Answer: Thyroid dysmorphogenesis

Q1.14: What screening program is in place for congenital hypothyroidism?

Answer:

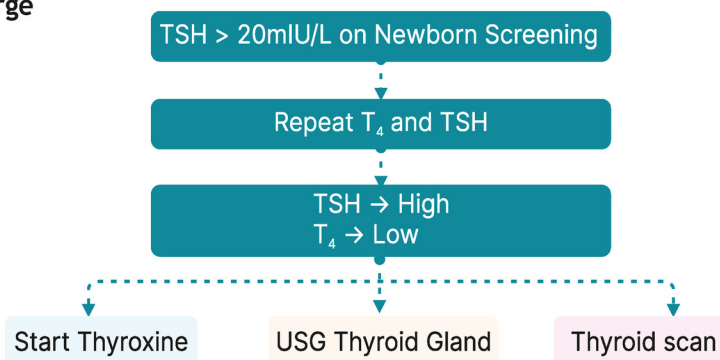
- Universal newborn screening

Q1.15: What are the common methods for collecting samples for newborn screening for congenital hypothyroidism?

Answer: At birth, with umbilical cord blood, or heel prick: dried blood spots, sample collected beyond 48 hrs or 48-72 hrs

Q1.16: Why should newborn screening for congenital hypothyroidism not be done in the first 1-2 days of life?

Answer: To avoid a TSH surge



Q1.17: What is the most sensitive approach for diagnosing congenital hypothyroidism?

Answer: Check for T4 & TSH both.

Q1.18: What is the treatment for congenital hypothyroidism, and how is it typically administered?

Answer: Oral Levothyroxine (early morning with empty stomach)

Q1.19: How does the dose of Levothyroxine change with the baby's age?

Answer: The dose is higher in the earlier age group and as the babies grow the dose reduces.

BIG QUESTION 2: Describe Adrenal disorders in children

Broad Answer:

- **Cushing Syndrome** in Children: Iatrogenic causes (exogenous corticosteroids) are the most common, leading to symptoms like obesity, short stature, and hypertension, with moon facies and buffalo hump being rare.
- **Congenital Adrenal Hyperplasia (CAH)** in Children: Most commonly due to 21-hydroxylase deficiency, presenting with ambiguous genitalia in females, salt-wasting crises (dehydration, hyperkalemia, hyponatremia), and requiring lifelong hormone replacement.
- **Diabetic Ketoacidosis (DKA)** in Children: A life-threatening complication of diabetes characterized by hyperglycemia, metabolic acidosis, and ketosis, requiring prompt fluid resuscitation and insulin infusion.

DETAILED QUESTIONS

Q2.1: What does the adrenal cortex of the fetus release in early pregnancy?

Answer: Cortisol

Q2.2: What does the adrenal cortex of the fetus release in the 2nd trimester?

Answer: DHEA

Q2.3: What is the most common cause of Cushing syndrome in children?

Answer: Iatrogenic

Q2.4: List the common clinical features of Cushing Syndrome in children.

Answer:

- Obesity, striae
- Short stature
- Hypertension
- Bone pains
- Muscle weakness
- Cataract
- Hyperglycemia
- Neuropsychiatric changes
- Behavioral problems
- Hirsutism

Q2.5: Which classic Cushingoid features are rare in children?

Answer: Moon facies and buffalo hump is rare

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Q2.6: A 10-day-old baby is brought with severe dehydration. On examination, the baby has ambiguous genitalia and feeble peripheral pulses. On investigation, there is metabolic acidosis, serum sodium levels are 131 meq/L and serum potassium level is 7.2 meq/L. What is the probable diagnosis?

Answer: Congenital adrenal hyperplasia

Q2.7: What is the screening test for 21-Hydroxylase Deficiency?

Answer: Increased 17-hydroxy progesterone mutation levels

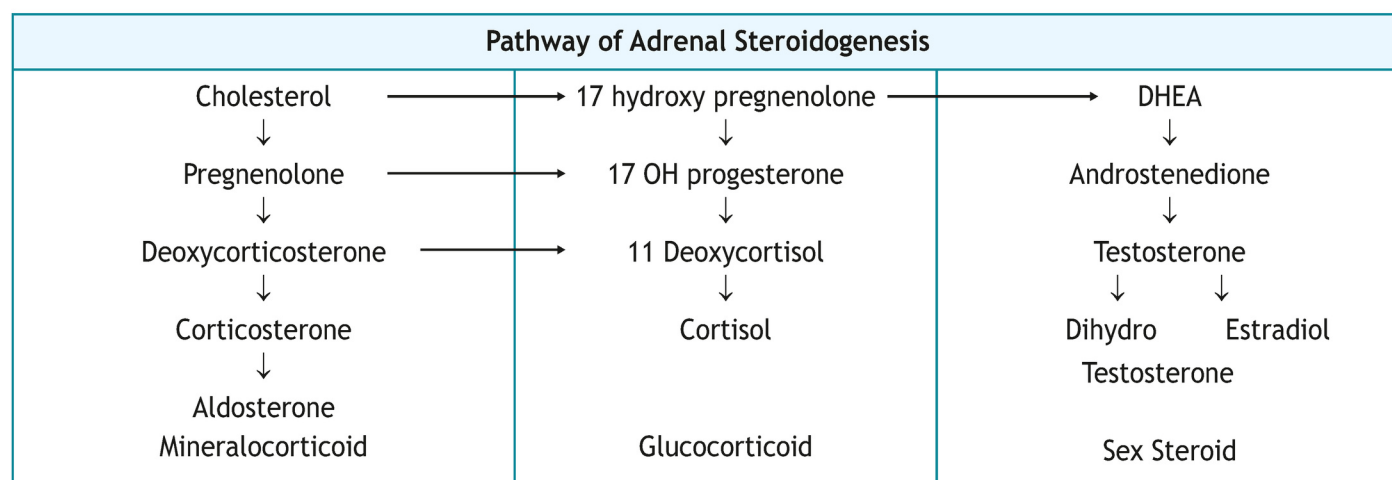
Q2.8: What are the two lifelong replacement treatments for 21-Hydroxylase Deficiency?

Answer: Hydrocortisone (Life long replacement), Fludrocortisone (Life long replacement)

Q2.9: Describe the adrenal steroidogenesis pathway?

Answer:

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Q2.10: What is the drug of choice for antenatal treatment of CAH?

Answer: Dexamethasone.

Q2.11: When should Dexamethasone be started in a couple with previous cases of CAH neonate, and what is its purpose?

Answer: Start dexamethasone as soon as the next pregnancy is diagnosed to prevent virilisation of the female.

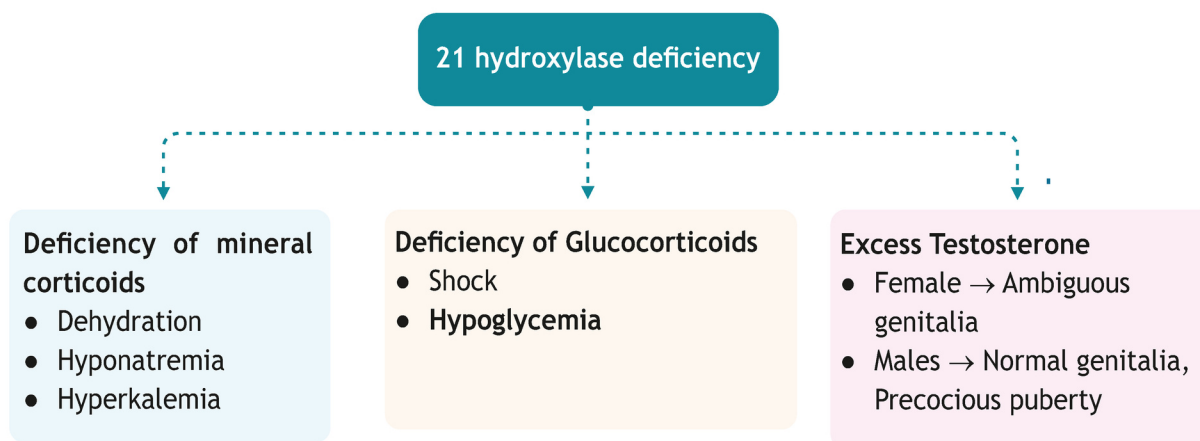
Q2.12: What are the types of CAH

Answer:

	Female virilization	Under virilization in Males
With salt wasting & Hyperkalemia	21 hydroxylase deficiency	3 B hydroxy steroid dehydrogenase deficiency
With Hypertension	11hydroxylase deficiency	17 α Hydroxylase deficiency

Q2.13: What are the clinical consequences of 21-hydroxylase deficiency?

Answer:



Q2.14: A 10-year-old child presented with tachypnea and severe dehydration. His random blood glucose was 524 mg/dl, ABG showed a pH of 7.0, HCO₃⁻ of 5 mmol/L, and urine showed moderate ketones. How do you manage the case?

Answer: The diagnosis is Severe Diabetic Ketoacidosis.

Q2.15: What are the three main diagnostic criteria for Diabetic Ketoacidosis (DKA) in children regarding blood glucose, acid-base status, and ketones?

Answer:

- Blood glucose >200 mg/dl.
- Along with metabolic acidosis (pH <7.3 or HCO₃⁻ <18 mEq/L).
- Along with ketonemia or moderate/large ketonuria.

Q2.16: Explain the management of DKA?

Answer:

In the 1st hour	- Normal saline - Mild- 10 ml/kg - Moderate to severe- 20 ml/kg - The amount of fluid is decreased from the total fluid correction in dehydration correction - Shock- rapid bolus of normal saline
After 1st hour	- Start regular insulin infusion @ 0.05-0.1 U/kg/hr continued till acidosis has improved - Dehydration correction over 48 hrs - Slow transition of subcutaneous insulin and diabetic diet
Other measures	- Treat infections - Fever