PEDIATRICS

4 PEDIATRICS

GROWTH

00:00:40

- Growth is assessed by Anthropometric parameters
- Major Anthropometric parameters are
  - Weight
  - Height
  - Head Circumference

Other important Anthropometric Parameters

- MUAC (Mid – Upper Arm Circumference)
  - Measured by SHAKIR'S TAPE.
  - 3 Colour Coded zones: red, yellow & green
- Skin fold thickness
  - Measured by Harpenden Calipers
  - Usual site
    - → Bicep's area
    - → Triceps area
    - → Supra scapular area
    - → Sub scapular area
    - → Measures to nearest mm

Important Information

- For normal values, WHO charts used (Earlier used → TANNER'S CHARTS)

Harpenden Calipers

- BMI (Body Mass Index)

$$BMI = \frac{\text{wt. (kg)}}{\text{Ht. (m)}2}$$

- Chest Circumference (CC)
  - At Birth - HC > CC (HC = Head Circumference)
  - 9 m – 1 yr. - HC = CC
  - > 1 yr. - CC > HC

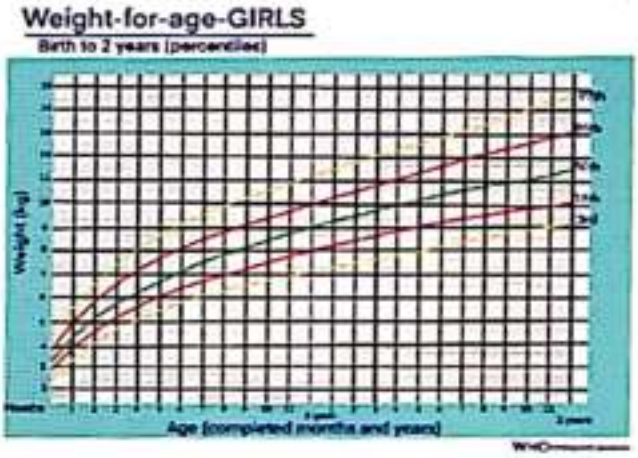
Important Information

- If HC is still more than CC, beyond 1 yr it indicates malnutrition

- Preferred Growth chart for under 5 all over world: WHO Growth Charts

WHO Growth Charts

00:03:30

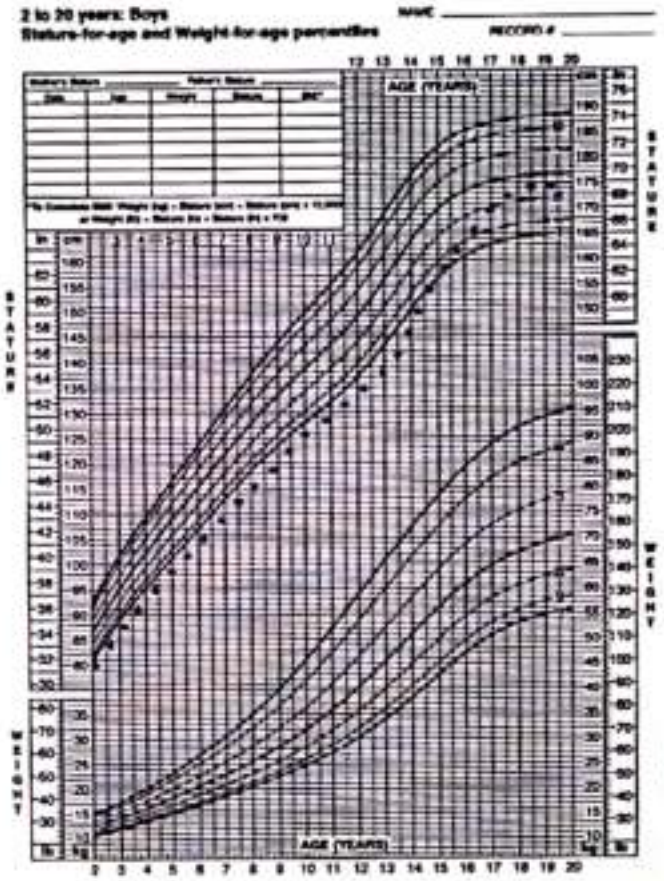


- First published in 2006
- Based on MGRS (Multicentre Growth reference study) 6 countries including India.

Important Information

- Exclusively breast-fed babies
- Maternal factors like alcohol & smoking have been excluded

- All anthropometric parameters covered
- Available in various formats (S.D. based, Z-score based, percentile based)
  - For children > 5 yrs. in India: Indian Growth Charts are preferred
  - Examples of Indian growth charts:



  - IAP (Indian academy of Paediatrics) Charts
  - K.N Aggarwal charts
  - Khadilkar charts

Weight

00:05:19

Infantometer

- Relation of weight with age
  - At birth: W (birth weight)
  - 5 months age: 2w (Birth weight doubles at 5 m)
  - 1 yr. - 3W (Birth weight triples by 1 year age)
  - 2 yr. - 4W
  - 3 yr. - 5W
  - 5 yr. - 6W
  - 7 yr. - 7W
  - 10 yr. - 10W

To Calculate Expected Weight of Child

- $< 1\,yr = \frac{x+9}{2}$ (x is age in months)
- 2 - 6 yrs. = 2x + 8, (x is age in yrs.)
- $7 - 12\,yrs = \frac{7x-5}{2}$, (x is age in yrs)

STADIOMETER

Height

00:07:53

- < 2 yrs. - Length (measured by Infantometer)
- >2 yrs. - Height (measured by stadiometer)
  - Standing Height is 0.7 cm less than recumbent Length

Important Information

- Average birth Weight of an Indian Baby – 2.9 kg

Previous Year's Questions

Q. A mother of a 5-year-old boy feels that he is too tall for his age & she brought him to hospital for evaluation. O/e his height was 108 cm, arm span of 106 cm, upper segment to lower segment ratio 1.2:1. What would be your advice to the mother?

(JIPMER - DEC - 2013)

A. Order for karyotyping
B. Reassure parents
C. Echocardiography to rule out Marfan syndrome
D. Ophthalmological examination & homocysteine levels

Relation of Length or Height with Age

- At birth: 50 cm
- At 1 yr.: 75 cm
- At 2 yr.: 90 cm
- At 4 – 4 $\frac{1}{2}$ yr.: 100 cm (Height doubles/ increases by 100% in 4- 4½ Yrs)

Important Information

- Length increased by 50% at 1 year
- Max. growth takes place in 1st year f/b puberty

- Formula for calculating expected Height According to Age
- Expected Height = (6 x + 77) cm (x → age in yrs.)

Upper Segment to Lower Segment Ratio

Age	Normal US: LS Ratio
At birth	1.7-1.9:1
3 yr	1.3:1
7 – 10 yr	1:1

Disorders of Height 00:13:05

Important Information

Short Stature

- Def: Height of a child < 3rd percentile or < -2 S.D of expected (acc. to age & sex)

2 Types of short stature

1. Proportionate: US: LS ratio remains Normal
2. Disproportionate: US: LS change

Refer Table 1.1

In GH Deficiency, US: LS Remains Normal

- BA < CA (Bone Age < Chronological Age)
  - CDGP
  - GH deficiency
  - Severe Malnutrition
  - Congenital Hypothyroidism

00:20:05

Disproportionate Short Stature	
Short Trunk Dwarfism (US: LS Decreases)	Short Limb Dwarfism (US: LS Increases)

Mnemonic: " Short Man May Climb High"

- Spondylo-Epiphyseal Dysplasia
- Mucopolysaccharidosis
- Mucolipidosis
- Caries Spine (Pott's disease)
- Vertebral defects like Hemivertebrae, Butterfly Vertebrae etc.
- Seen in Alagille syndrome
  - Triangular facies (Also seen in Russell Silver syndrome)
  - Butterfly vertebra
  - Pulmonary Stenosis
  - Cholestatic Jaundice
- Rickets
- Achondroplasia
- Osteogenesis Imperfecta
- Congenital Hypothyroidism

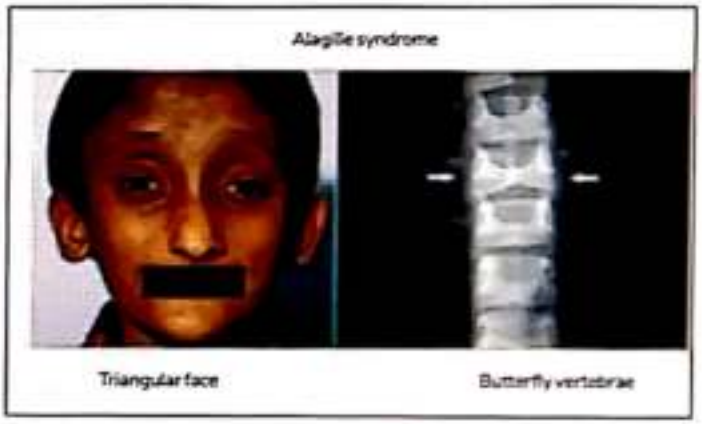
Head Circumference (HC) 00:23:50

(aka Occipitofrontal Circumference)

Important Information

- Head circumference at birth 33-35 cm

- Normal rate of increase in HC
- 1st 3 months: 2 cm / month
- Next 3 months: 1 cm / month
- Next 6 months: 0.5 / month
- Next 2 years: 0.2 cm / mont



Microcephaly 00:25:28

- Def: HC < -3 S.D or Z- score of expected (acc. To age & sex of child)

Refer Table 1.2

Macrocephaly 00:32:20

- Def: Macrocephaly (HC > + 2 SD of Expected, ACC to Age & Sex of Child)

Etiology

- Increased Thickness of Cranial Bones e.g. Osteogenesis Imperfecta, Chronic hemolytic Anemia like Thalassemia;
- Subdural Fluid Collection e.g. subdural empyema, Subdural effusion (complication of meningitis)
- Megalocephaly (increases in size of brain) causes
  - Mnemonic: "Balwan SINGH"
    - → Benign familial megalocephaly
    - → Amino acid disorders - MSUD, Glutaric aciduria
    - → Lysosomal storage diseases - Mucopolysaccharidosis, GM 1 Gangliosidosis
    - → Weaver syndrome
    - → Achondroplasia
    - → Neurofibromatosis
    - → Sotos Syndrome (aka Cerebral Gigantism)
    - → Neuro - degenerative disorders- Alexander's disease, Canavan's disease
    - → Galactosemia

Important Information

- Hydranencephaly (Cerebral hemispheres are absent and replaced with fluid filled sacs. So Transillumination is positive
- Hydrocephalus: increase in size of the ventricles inside the brain.

Abnormal Head Shape 00:35:32

Previous Year's Questions

Q. Most common cause of craniosynostosis is?
(JIPMER - Nov - 2018)

A. Plagiocephaly
B. Brachycephaly
C. Scaphocephaly
D. Trigonocephaly

- Craniosynostosis → Premature fusion of cranial sutures

Important Information

- M / C Type of abnormality - Dolichocephaly? Due to Premature fusion of Sagittal suture.
  - Trigonocephaly: Due to Premature fusion of Metopic suture
  - Turricephaly: Due to premature fusion of Coronal, Spheno - frontal & fronto - ethmoid suture.
  - Syndromes associated with Craniosynostosis

1. Carpenter Syndrome
2. Apert Syndrome
3. Pfeiffer syndrome
4. Crouzon Syndrome
   - → Brachycephaly (d / t premature fusion of coronal suture)
   - → Bulging eyes, mid - face hypoplasia.
   - → Prognathism (protruding jaw)

Normal Puberty 00:37:38

Important Information

First Sign of Puberty

- **Girls: Thelarche (Breast development)**
- **Boys: Testicular Enlargement (testicular volume 2 - 4 ml)**

Device used to assess testicular size - Orchidometer

Orchidometer

- Pubertal changes assessed by TANNER'S Staging OR SMR (Sexual Maturing Rating)
- Stages 1 (Pre - pubertal) to 5 (Mature Adult)
- Parameters for assessing SMR
  - Female: Breast, Pubic hair
  - Male: Penis, Scrotum, Pubic Hair

- Growth spurt in seen in
  - Stage 3: in Female
  - Stage 4: in Male (growth spurt occurs later & lasts longer in males)

Dentition

00:39:50

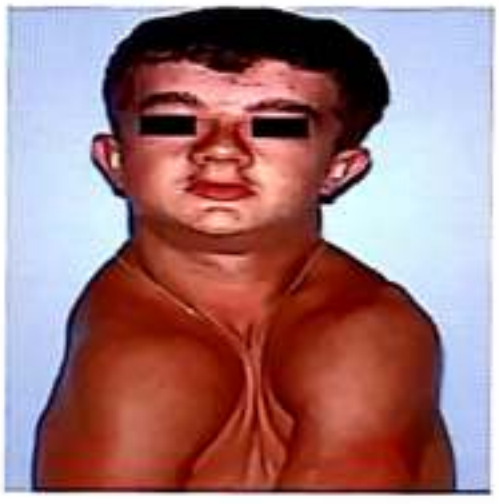
	Primary Dentition (Temporary Teeth)	Secondary Dentition (Permanent Teeth)
Begins At	6 months	6 year
1st Tooth	Lower central incisor	1st Molar
last Tooth	2nd Molar	3rd molar
Completes By	2 ½ year - 3 year	12 yrs. except 3rd molar (18-25 year; may not erupt also)
Total No. Of Teeth	20	28 - 32

Delayed Dentition

- Def: When no Teeth Erupts By 13 Months of Age

Important Causes

- Mnemonic: "FRIED Chop"
  - Familial
  - Rickets
  - Idiopathic
  - Endocrine (3 "hypos")
    - → Hypo - pituitarism
    - → Hypo - thyroidism
    - → Hypo - parathyroidism
  - Down Syndrome
- Cleidocranial dysostosis (Clavicles absent, large anterior fontanelle, supernumerary teeth)



Cleidocranial Dysostos

Important Information

Hutchinson's Teeth

- Notched incisors
- Seen in congenital syphilis

Hutchinson's Triad (seen in congenital syphilis)

- Hutchinson's teeth
- Sensorineural hearing loss
- Interstitial keratitis

Table 1.1 Important Causes of Proportionate Short Stature

Normal Variants

- Familial
- CDGP (Constitutional Delay in growth & Puberty)
  - ↓
  - M / C cause of short stature during childhood
  - Delayed Puberty
  - Family h/o delayed puberty
  - Bone Age < Chronological Age

Intrauterine

- IUGR
- IU Infections (Particularly TORCH)
- Genetic Syndromes
  - Down's Syndrome
  - Turner's Syndrome

Postnatal or Acquired

- Severe Long-standing Malnutrition
- Any Chronic Systemic
- Illness (e.g.,CKD)
- Celiac Disease
- Endocrine Causes
  - GH deficiency
  - Cushing syndrome
- Psychosocial (e.g. Maternal deprivation)

Table 1.2 Important cause of microcephaly

Primary (Genetic)

Mnemonic: "Cannot See PEFR in Children "

- Cri - du - Chat Syndrome (5p-) (Cat - like cry, CHD, Microcephaly)
- Smith - Lemli - Opitz Syndrome
- Patau Syndrome (Trisomy 13)
- Edward Syndrome (Trisomy 18)
- Familial
- Rubinstein Taybi Syndrome (Heart defects, Thumb abnormality, Microcephaly) (aka Broad Thumb - Hallux syndrome)
- Cornelia de Lange Syndrome (Microcephaly, CNS manifestations, Long eye lashes)

Secondary

1. Maternal Causes
- Maternal alcohol intake
  - ↓
  - Fetal Alcohol Syndrome
  - Facial dysmorphism (small palpebral fissures, thin lips)
  - Congenital Heart Disease
  - Microcephaly
- Maternal smoking
- Maternal infections like TORCH
- Maternal exposure to Radiation
- Maternal drug intake like phenytoin
- Maternal Phenylketonuria

2. Meningoencephalitis (CNS infections during infancy)
3. Severe Malnutrition
4. Birth Asphyxia (HIE): Cerebral Palsy
5. Acquired Microcephaly ("R-A- S")
- Rett syndrome
  - HC Normal at birth & microcephaly develops later
  - X - linked dominant inheritance.
  - MECP2 gene m/c involved
  - Developmental delay
  - Stereotypic hand wringing movements
  - A/w seizures, breathing irregularities, behavioral problems.
- Angelmann syndrome (Happy Puppet Syndrome)
- Bouts of laughter, CNS abnormalities, developmental delay (Due to genetic imprinting)
- Seckel syndrome ('Bird headed ' dwarfism)

DEVELOPMENT

⏱ 00:44:07

A. GROSS MOTOR MILESTONES

- **In Ventral Suspension**
  - 1 month: Head below the plane of rest of body
  - 2 Months: Head in the plane of rest of body
  - 3 months: Head above the plane of rest of body (Neck control develops)

- **In Prone Position**
  - At birth: High pelvis, knees under the abdomen, head turned to one side
  - At 1-2 months: Raises head & chin off table at 45°
  - At 3 months: Supports weight on forearms
  - At 6 months: Supports weight on extended forearms or hands

- **Other Gross Motor Milestones**

6 Months	Sitting with Support (Tripod Position); Rolls Over (Prone To Supine)
8 Months	Sitting without Support
9 Months	Standing with Support
10-11. months	Pivots & cruising
12 months	Standing without support; Walking with support
15 months	Walks without support; Creeps upstairs
18 months	Goes upstairs & downstairs holding side rails, RUNS;
2 yrs.	Upstairs & downstairs (2 feet/step); Kicks a ball
3 yrs.	Goes upstairs with alternate feet but downstairs with 2. feet/step, Rides a Tricycle
4 yrs.	Goes both upstairs & downstairs with 1 foot/step (alternate steps); Hops on 1 foot

Previous Year's Questions

Q. A 6 years old child with developmental delay, can ride a tricycle, can climb upstairs with alternate feet, but downstairs with 2 feet per step, can tell his name, knows his own sex, but cannot narrate a story. What is his developmental age?
(AIIMS MAY 2019)

A. 3 years
B. 4 years
C. 5 years
D. 2 years

B. FINE MOTOR MILESTONES

3 months	Hand regard appears; Holds an object when placed in hand (Palmar grasp reflex is lost)
4 months	Tries to reach for object
5 months	Bidextrous Grasp
6 months	Unidextrous Grasp (Palmar Grasp)
7 months	Transfers Objects
9 months	Immature /Assisted Pincer Grasp
12 months	Mature / Unassisted Pincer Grasp, Casting.
15 months	Spontaneous scribbling; Tower of 2 cubes; Drinks from cup
18 months	Feeds self with spoon
2 yrs	Tower of 6-7 cubes; Copies a straight line; Turns a door knob/Unscrews a lid
3 yrs	Tower of 9-10 cubes; Handedness get established; Copies a circle
4 yrs	Copies a rectangle or cross; Makes a bridge with cubes; buttons /unbuttons
5 yrs	Copies a triangle or tilted cross; Ties shoes laces; Makes a 'gate'/'door' with cubes

Previous Year's Questions

Q. Bidextrous grasp is seen at what age?
(NEET - JAN - 2019)

A. 4 months
B. 5 months
C. 6 months
D. 7 months

Previous Year's Questions

Q. A child transfers objects from one hand to other. What does it imply?
(AIIMS - JUNE - 2020)

A. Visual motor co-ordination
B. Explores small object
C. Object release
D. Comparison of objects

C. SOCIAL MILESTONES

2 Months	Social Smile
6 months	Mirror Play
7 months	Stranger anxiety
8 months	Object permanence
9 Months	Waves Bye-Bye
10 months	Plays peek-a-boo
12 months	Kisses on request; Releases objects; Plays a simple ball game.
15 months	Points to objects; Indicates wet pants.
18 months	Domestic mimicry; Dry during day time.
2yrs	Parallel Play.
3yrs	Joins in Play, Knows name, age & gender, Dry at night (Night - time continence).

Previous Year's Questions

Q. An 8-year-old male child presented with a history of bed wetting. There are no other associated symptoms, apart from the discomfort due to bedwetting. What is the initial and most effective therapy?
(INICET - Nov - 2020)

A. Pharmacological therapy with Imipramine
B. Bladder training with holding urine for longer periods during daytime
C. Classical conditioning with alarm & pad at night
D. Psychodynamic therapy

Important Information

- Nocturnal Enuresis → Involuntary urination @ night beyond 5 yrs. age.
- Rx of Nocturnal enuresis:
  - 1st line: Lifestyle measures & motivational therapy
  - 2nd line: Bed and Alarms
  - 3rd line: Pharmacotherapy with drugs like Imipramine, Desmopressin

D. LANGUAGE MILESTONES

6 months	Speak monosyllables
9 months	Bi - syllables e.g.- mama, papa but without meaning
12 months	2-3 words with meaning
18 months	Vocabulary of 10 words
2 yrs.	2-word sentences; Vocabulary of 100 words; uses Pronouns
3 yrs.	3-word sentences; uses plurals and past tense
4 yrs.	Sings a song; Tells rhymes / story (Normal child has dysfluency of speech → 2 - 5 years of age)

DQ (Developmental Quotient) 00:58:23

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

Development delay

- Performance in 1 or more domains is significantly below average.

RED flag signs of developmental delay:

Gross motor		Fine motor	
Sitting with support	9 months	Pincer grasp	12 months
Standing with support	12 months		
Language		**Social**	
Speaks Single word	16 months	Social smile	6 months
		Waving bye bye	12 months

IQ (intelligence Quotient) $IQ = \frac{\text{Mental Age}}{\text{Chronological Age}} \times 100$

- Normal IQ: 90–110
- Borderline: 70–89
- Moron: 50–70
- Imbecile: 20–49

Important Information

Idiot/ profound ID: <20

- **ID = intellectual disability: previously known as Mental retardation (MR)**

NEONATOLOGY

01:02:03

CLASSIFICATION OF NEONATES

1. **According to Gestational Age (Irrespective of Birth weight)**
   - Term neonate: born between 37 completed wks. to < 42 wks.
   - Preterm: born at < 37 completed weeks of gestation
   - Post Term: born after > 42 weeks gestation
2. **According to Birth wight: (Irrespective of Gestational Age)**
   - Low Birth weight (LBW): < 2500 g
   - Very Low Birth weight (VLBW): < 1500 g
   - Extremely low Birth weight (ELBW): < 1000 g
3. **According to gestational Age & Birth Weight.**
   - Large For Date (LFD) or Large for Gestational Age (LGA): Birth weight > 90th Percentile of expected, according to gestational age
   - SFD (Small for Date) or SGA (Small for Gestational Age): Birth weight < 10th percentile of expected, according to gestational age.

CHARACTERISTIC OF A NORMAL NEONATE

- HR: 110-160/min
- RR: 40 - 60/min
- Peripheral Cyanosis: presence is normal

Important Information

- **Soft systolic murmur: may be present (Normal)**

- Pupils Constricted at Birth
- Gestational age of a neonate is assessed by ENBS: Expanded new Ballard score (Used over Gestational age of 20 - 44 weeks)

01:05:58

Characteristics of Pre-Team Neonate	
General	**Head to Toe**
• Small & emaciated appearance • Skin is fragile, thin, translucent	• Head appears relatively large • Ant. Fontanelle is large & wide open
• Generalized hypotonia (Extended posture) • Abundant lanugo (but little vernix caseosa)	• Ears are crumpled or poorly formed • Breast buds < 5 mm • Genitals o Male: Undescended testis, poorly formed (smooth, hypopigmented) scrotum o Female: Labia minora visible, labia major widely separated • Absent deep creases on sole

Mongolian Spots

Epstein pearls Absent

Deep Creases on sole

Milia

Erythema toxicum

01:10:15

Conditions in Neonates Not Requiring Rx	
Skin & Mucosa	**Others**
• Milia (Colourless papules d / t plugging of sweat ducts) • Erythema Toxicum Neonatorum (red coloured rashes, mainly on trunk during 1st week for life d / t some immune phenomenon) • Mongolian Spots (Bluish - black areas of discoloration on lower back & buttocks) • Stork Bites / Salmon Patches (capillary hemangiomas) • Epstein Pearls (pearl like white lesions on palate, which are epithelial inclusion cysts) • Subconjunctival Hemorrhage	• Mastitis Neonatorum: B / L Breast Engorgement (in both female and male due to maternal estrogens) • Vaginal Bleeding in female Neonates (No Rx reqd.) • Hymenal Tags • Physiological wt. Loss (in term babies, up to 10 % of birth wt. loss is physiological)

Previous Year's Questions

Q. A term neonate, with a birth weight of 2700 g, who is otherwise well, and is exclusively breastfed, presents for routine evaluation. His total serum bilirubin is found to be 14mg/dl on day 5. What is the management?

(NEET - JAN - 2020)

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

Cephalhematoma	Caput Succedaneum
• Sub periosteal haemorrhage involving cranial bones • Does not cross sutures • Takes 24 - 48 hrs to appear completely • Takes 5-7 weeks to disappear • Predisposes to jaundice (d / t collection of blood)	• Due to edema in the layers of scalp • May cross sutures • Present at birth in its max size • Disappear in 48 - 72 hrs. • Does not predispose to jaundice.

Primitive Neonatal Reflexes	
Present at Birth	**Appear After Birth**
Rooting Reflex • Earliest primitive neonatal reflex to disappear • Helps mother in breast feeding the baby • Appears: 32 weeks gestation • Disappears: Starts disappearing at 1-month postnatal age	STNR (Symmetric Tonic Neck Reflex) • Appears after birth • Appears at 4 - 6 months • Disappears at 8 - 12 months
MORO'S Reflex • Persistence > 6 months: indicates cerebral damage • Absent in Down syndrome & Stage 3 HIE • Starts Appearing: 28 weeks gestation • Completely Appears: 37 weeks gestation • Disappears: 5 - 6 months postnatal age	Parachute Reflex • Appears at 7 - 8 months • Persists Throughout Life (Never Disappears)

Palmar Grasp Reflex
- Appears: 28 weeks
- Disappears: 3 months
- Voluntary palmar grasp appears, only when palmar grasp reflex disappears

ATNR (Asymmetric Tonic Neck Reflex)
- Disappearance of ATNR Baby learns to roll over
- Appears: 35 weeks gestation
- Disappears: 5-6 months postnatal age

Asymmetric Tonic Neck Reflex (ATNR)

Causes of asymmetric MORO's Reflex	
Nerve Related Cause	**Bone Related Cause**
• Erb's Palsy • Congenital Hemiplegia	• Fracture clavicle • Shoulder joint dislocation

- Clavicle Is the most Common Bone to Fracture in a Neonate
- Neonatal Resuscitation (NT)
  - Temp of Delivery Room ~ 25 C (22 - 28 C)
- Suction (Not all newborns require suction)
- Order of suction
  - Mouth f/b Nose (Tracheal very rarely reqd.)
  - Pressure required - 80-100 mmHg

Previous Year's Questions

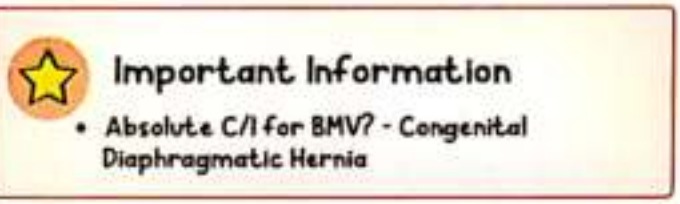
Q. Correct order of suctioning during neonatal resuscitation is?

(AIIMS - MAY - 2018)

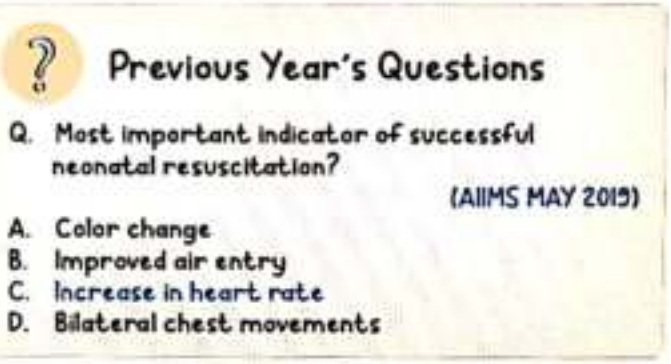
A. Trachea - nose - mouth
B. Nose - mouth
C. Mouth - nose – Trachea
D. Mouth – nose

- **Indications of Positive Pressure ventilation (PPV)**
  - Apnea
  - Gasping (Severe respiratory distress)
  - HR < 100 / min (use of ECG is now recommended for knowing heart rate in neonatal resuscitation

- **Pressure required for positive pressure ventilation**
  - 1st breath: 30-40 cm H2O
  - Subsequent breaths: 15-20 cm H2O
- PPV is done using a self - inflating Bag & Mask (may be attached to reservoir to increase 02 concentration delivered to baby)



- Most sensitive indicator of successful Resuscitation: Improvement in Heart Rate
- Indication to start chest compression: When HR < 60/ min, persistently even after BMV.
- Always start BMV with Room Air (fio2 - 21%) and start O2, only if baby not improving.
  - CC: PPV → 3: 1



  - During chest compressions → Use 100% O_2
  - No improvement despite chest compressions
  - Injection Adrenaline
    Dose: 0.01 mg/kg/dose
    OR
    0.1 ml/kg/dose of 1: 10,000 Adr.
    } x Upto 3 times
- Preferred Route: I/V (Through UVC)
  - Umbilical venous catheter (UVC) (Preferred vascular access in Neonatal resuscitation)
  - If not able to secure vascular access → then Adrenaline may be given intra-tracheally

Resuscitation of Baby Born Through MSL

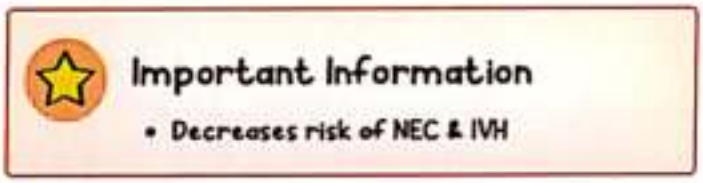
- Intrapartum suction of mouth & nose not recommended
- Routine ET intubation & tracheal suction of non - vigorous neonates, born through MSL is not recommended (However 1 person skilled in ET intubation should available at the time of resuscitation)

Delayed Cord Clamping (Now included in Neonatal resuscitation)

- Wait for at least 30 seconds after birth, before clamping cord.
- Should be done in all stable term & preterm neonates
- Done in Vigorous Neonates (don't wait in c / o sick neonates)
  - HR > 100
  - Good muscle tone
  - Strong Respiratory efforts

Advantages of delayed cord clamping

- Higher Hb level
- Lesser anemia - less need for blood transfusion
- Higher BP - less chances of shock



Feeding of Preterm

- < 28 weeks gestation: I/v fluids with or without TPN
- 28 - 31 weeks: OG tube feeding OR GAVAGE feeding.
- 32 - 34 weeks: Katori spoon feeding or Paladai feeding
- > 34 weeks: Direct breast feeding

IUGR (Intrauterine Growth Restriction)	
Symmetric	**Asymmetric**
• Weight, length, HC equally affected • Ponderal index (PI) >or= 2	• Brain growth spared PI < 2

$$\text{PONDERAL INDEX} = \frac{\text{Wt (g)}}{\text{Length (cm)}^3} \times 100$$

NEONATAL SEPSIS

01:27:04

- MC organism responsible for neonatal sepsis:
  - In India: Acinetobacter > Klebsiella
  - In hospitals in India: Acinetobacter
  - In hospital across world: E. coli
  - Overall, throughout world: Group B streptococcus
  - Early onset sepsis: Group B streptococcus
- Most important method to prevent neonatal sepsis: Proper hand washing of caregivers. (For around 2 min following 6 steps technique).

Important Information

- Earliest indicator of Neonatal sepsis: Poor feeding

6 Steps of hand washing

Sepsis Screen

- Total leukocyte count
- Absolute Neutrophil count
- I - T Ratio
- CRP
- Micro ESR
  - If 2 out 5 suggestive then sepsis screen 6 Steps of hand wash
  - Confirmatory Test - Blood culture

Important Information

- For all babies with late onset neonatal sepsis: Do LP (to r / o meningitis)

  - Rx of choice: I/V broad spectrum empirical antibiotics (No role of oral antibiotics)

NEONATAL HYPOTHERMIA

- Definition: Axillary temp < 36.5 C in newborn (thermometer should be kept for 3 – 5 min in axilla)

Classification	Axillary Temp
Cold stress	36 – 36.4 °C
Moderate hypothermia	32 – 35.9 °C
Severe hypothermia	< 32 °C

Important Information

- Maximum heat loss is from head of baby (due to more surface area)

- Shivering is absent in neonates
- Non shivering Thermogenesis (most imp mechanism for defense against hypothermia) seen d/t presence of brown fat (mitochondria rich)
- Brown Fat is present in
  - Axilla
  - Groin
  - Inter-scapular area
  - Nape of neck

"Warm Chain" (series of step to keep baby warm)

- Components
  - Warm Room
  - Warm Resuscitation
  - Early initiation of Breast feeding
  - Skin to skin contact with mother
  - Rooming in
  - Room in draught free area
  - Warm transportation

Kangaroo Mother Care (KMC)

Kangaroo Mother Care

- Recommended for all stable LBW babies
- Components
  - Kangaroo position (skin to skin contact)
  - Kangaroo Nutrition (exclusive breast feeding)
  - Kangaroo discharge & follow up

Advantages

- Higher exclusive breast-feeding rates
- Decreases risk of
  - Hypothermia
  - Neonatal sepsis
  - Neonatal mortality
  - Shortens Hospital stay & Early discharge
- Devices to keep baby warm

Devices	(Most Important) Mode of Heating
Incubator (closed box)	Convection
Radiant warmer (open system)	Radiation

NEONATAL HYPOGLYCEMIA

01:34:30

- Def: Blood Glucose < 40 mg/dl
  or
  Plasma glucose < 45 mg/dl
  (N. hyperglycemia → Blood glucose > 125 mg/dl
  or
  Plasma Glucose > 150 mg/dl)
- M/C C/F: Jitteriness or Tremors
- Rx: I/V 10 % Dextrose

Important Information

- MC cause of persistent Hypoglycemia during infancy: PHHI (Persistent Hyperinsulinemic Hypoglycemia of Infancy) (earlier k/a Nesidioblastosis)

Infant of Diabetic Mother (IDM)

Infant of Diabetic Mother

LFD baby (macrosomia) with Hairy Pinna

- **Problems**
  - Large for Date (LFD)
  - Birth Injury
  - Perinatal Asphyxia
- **Metabolic derangements**
  - Hypo – glycemia
  - Hypo – calcemia
  - Hypo – magnesemia
  - Polycythemia
  - Neonatal jaundice
- **Respiratory System:** More Chances of RDS d / t delayed maturation of surfactant even in term babies.
- **CVS**
  - MC congenital abℕ in IDM: Congenital Heart disease
  - MC CHD in IDM: VSD

Important Information

- **Most specific CHD in IDM: TGA (Transposition of great arteries)**

- **CNS**
  - MC congenital neurological abℕ in IDM → Neural tube Defects

Important Information

- **Most specific CNS congenital abnormality in IDM - Sacral Agenesis or Caudal Regression syndrome**

- **Long Term Problems:** (Mnemonic: "BOND")
  - Blindness
  - Obesity
  - Non Ketotic hypoglycemia
  - Diabetes Mellitus

PERINATAL ASPHYXIA

01:40:27

- CNS Changes d/t Perinatal Asphyxia is known as HIE (Hypoxic Ischemic Encephalopathy)
- In severe Birth Asphyxia, APGAR score: 0-3

Previous Year's Questions

Q. APGAR score 3 at 1 minute indicates?
(NEET - Jan - 2019)

A. Mildly depressed
B. Further resuscitation not needed
C. Severely depressed
D. Normal

- Part of brain most commonly involved in HIE
  - In Term Neonates: Para - sagittal Area
  - In Preterm Neonates: Peri - ventricular Area

  } Leading to Cerebral Palsy

Staging of HIE

	Stage 1	Stage 2	Stage 3
Moros Reflex	Exaggerated	N / ↓	Absent
Seizures	-	Present	-
Prognosis	99% Normal Outcome	80% Normal Outcome	50% - Mortality 50% - severe neurological sequelae

- Upcoming / Latest Rx for moderate to severe HIE in neonates: Therapeutic Hypothermia

NEONATAL SEIZURES

01:42:55

- Mc type: Subtle Seizure
- Mc cause: Hypoxia
- Best prognosis: Focal Clonic Seizure
- Worst Prognosis: Myoclonic Seizure
- DOC for neonatal Seizures: Phenobarbitone

Important Information

- Preferred CNS imaging in Neonates? Transcranial USG through anterior fontanelle

APGAR score			
	0	1	2
Appearance	Completely blue or pale	Body pink but extremities blue	Completely pink
Pulse rate	Absent	< 100 / min	> 100 /min
Grimace	No response	Grimaces only	Cough/sneezes
Activity	Limp/Flaccid	Some Flexion	Posture with movement
Respiratory Effort	None	Slow & irregular	Strong

- Min^m Score: 0
- Max^m Score: 10
- Score > 7: Normal
- Score < 3: Severe birth Asphyxia
- APGAR score has no role in NR only has prognostic value

Scores used to assess Respiratory distress

- Preterm neonates: Silverman Score
- Term neonates: Downe's Score

Silverman Score 01:47:43

- Used to quantify respiratory distress in preterm neonates
- Has 5 components

	0	1	2
Upper chest retraction	Chest & abdomen moving equally, in same direction	Chest movement lags behind abdomen	See saw movements or paradoxical breathing
Lower chest Retractions	None	Minimal	Marked
Xiphisternal Retractions	None	Minimal	Marked
Nasal Flare	None	Minimal	Marked
Grunt	None	Audible only with stethoscope	Audible without stethoscope

Previous Year's Questions

Q. Which of the following scoring is done to assess respiratory distress in Neonates?
(JIPMER - May - 2019)

A. CRIB score
B. Silverman- Anderson score
C. APGAR score
D. SNAP Score

- Max score: 10
- Min score: 0
- More score: More Respiratory Distress
- Score > 7: Severe Respiratory Distress

RESPIRATORY DISTRESS SYNDROME (RDS) OR HYALINE MEMBRANE DISEASE (HMD)

Previous Year's Questions

Q. In a preterm baby with respiratory distress syndrome, which type of cell is deficient?
(NEET - Jan - 2020)

A. Type I alveolar cell
B. Type 2 alveolar cell
C. Alveolar capillary endothelial cells
D. Bronchial mucosal epithelial cell

- Due to deficiency of Mature Surfactant
- Surfactant

Important Information

- Synthesis of surfactant begins in fetal lungs at 20 weeks of gestation.

  - Begins to appear in Amniotic fluid 28 -32 weeks
  - Mature surfactant in adequate amount > 35 weeks
  - Most imp component DPPC (Dipalmitoyl phosphatidyl Choline) or lecithin.
  - Most important surfactant protein Type B
  - RDS is mc cause of Respiratory distress in a preterm neonate.

- CXR

Ground glass haziness in RDS

  - Ground glass haziness of lungs
  - Air bronchogram
  - Reticulogranular or reticulonodular appearance

- **Ways to Detect Adequacy of Surfactant in Amniotic Fluid**
  - L: S > 2:1 - Mature surfactant (Lecithin: Sphingomyelin)
  - Nile Blue sulphatase test
  - Shake test
- **Rx of RDS**
  - Mild: CPAP only
  - Moderate to Severe: intra-tracheal Surfactant + Respiratory Support

Antenatal Corticosteroids for prevention of RDS	
Inj Betamethasone	**Inj Dexamethasone**
• 12 mg IM, 2 does, 24 hr apart (12 x 2 = 24) • Preferred • More Neuroprotective	• 6 mg, 4 dozes, 12 hr. apart (6 x 4 = 24) • Recommended by government of India because cheaper, readily available and efficacy almost same

Advantages of Antenatal Steroids in Preterm

- Decreases Risk of
  - RDS
  - NEC
  - IVH
  - Neonatal Mortality (doesn't decrease risk of Neonatal jaundice)

Neonatal Pulmonary Alveolar Proteinosis 01:56:04

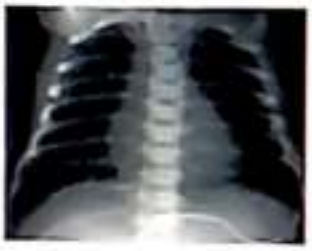
- Intra - alveolar accumulation of surfactant d/t surfactant dysfunction.

Important Information

- Fatal form of neonatal pulmonary alveolar proteinosis is due to Deficiency of Surfactant protein Type B.

- CXR & lung biopsy finding: same as RDS
- Differentiating features
  - Term Neonate
  - Family History
  - Fatal disease

MECONIUM ASPIRATION SYNDROME



CXR in Meconium aspiration syndrome

- A term SGA / IUGR baby, born through MSL, develops respiratory distress soon after birth.

Important Information

- O / E in meconium aspiration syndrome - AP diameter of chest increased.

- **Typical CXR findings**
  - Flattening of domes of diaphragm
  - Hyper inflated Lungs (↑radiolucency)
  - Pulmonary infiltrates
  - Segmental collapse
- **TTNB (Transient Tachypnea of New Born)**
  - Due to delayed clearance of lung fluid

  Delivery by caesarean - section is a risk factor
  - Mild & Self Limiting
  - No Rx required.
- **Neonatal Apnea** 01:59:40
  - Def: Cessation of breathing for at least 20 seconds or for any duration in presence of Bradycardia or Cyanosis.

Important Information

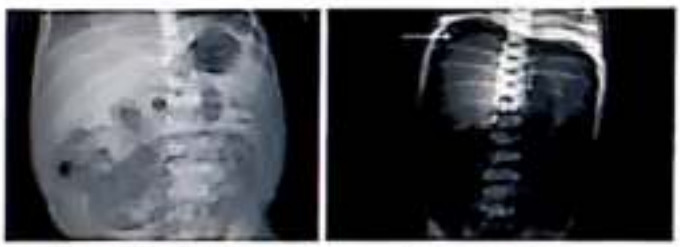
- DOC for Apnea of Prematurity?
  Inj. caffeine citrate > Inj. Aminophylline

NEC (NECROTIZING ENTEROCOLITIS)

02:01:03

- Most important Risk Factor: Prematurity
- Clinical features: Feed intolerance, recurrent vomiting, blood in stools, and distension

Staging of NEC



Pneumatosis intestinalis | Pneumoperitoneum

- Stage
  - Ia: Occult blood in stool
  - Ib: Fresh blood in stool
  - IIa: Pneumatosis intestinalis (gas in intestinal wall)
  - IIb: Portal vein Gas shadows
  - III: In Blood investigations: Metabolic acidosis, Thrombocytopenia, Hyponatremia seen
  - IIIa: Peritonitis
  - IIIb: Pneumoperitoneum (intestinal perforation)
- Rx in all stage: NPO + IV fluid + IV antibiotic
- Stage IIIb: S, required
- Prognosis: 10 – 30% risk of mortality despite best supportive care.

Previous Year's Questions

Q. Which among the following is not included in triad of necrotizing enterocolitis?

(NEET - Jan - 2019)

A. Thrombocytopenia
B. Metabolic acidosis
C. Hyponatremia
D. Hypokalemia

NEONATAL JAUNDICE

02:03:23

- Clinical jaundice in Neonates: Bilirubin > or=5 mg / dl

Physiological Jaundice	Pathological Jaundice
• Clinical jaundice never appears in 1st 24 hrs of life. • Always unconjugated; so, urine does not stain diaper & no pale stools • Icterus does not involve palms and soles • Clinical jaundice does not persist beyond 3 weeks	• May appear in 1st 24 hrs of life • Conjugated or unconjugated; high colored urine & / or pale stools may be seen • Palms and soles may be stained yellow • May persist for longer time

Breast Feeding Jaundice

- d / t decreased breast feeding
- Rx: ↑Breast feeding

Breast Milk Jaundice

- D / t inhibitors present in breast milk like pregnanediol, FFA that inhibit conjugation of bilirubin
- Breast milk increases enterohepatic circulation
- R$_x$: Continue breast feed unless bilirubin level > 20 mg / dl

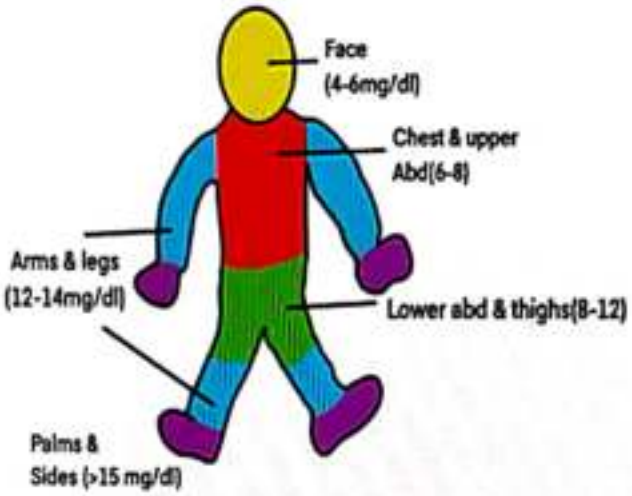
Important Information

- Mc cause of Neonatal jaundice within 1st 24 hrs. of life – erythroblastosis fetalis

Unconjugated Hyperbilirubinemia	
Increased Production	**Decreased Conjugation**
○ Erythroblastosis fetalis ○ Hereditary spherocytosis ○ G6PD deficiency	○ Crigler Najjar syndrome (deficiency of UDPGT enzyme)

Refer Table 3.1

Modified Kramer's Rule for Estimation of Bilirubinemia in Children



- Face: 4-6 mg/dl
- Chest & upper abdomen: 8-10 mg/dl
- Lower abdomen & thighs: 12-14 mg/dl
- Arms & Legs: 15-18 mg/dl
- Palms & soles: 20 mg/dl

Previous Year's Questions

Q. Greenish black stool in neonate is due to?
(NEET - Jan - 2020)

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

KERNICTERUS (CNS Manifestation of Hyperbilirubinemia) 02:10:16

- Part of brain mc involved: Basal ganglia
- Type of cerebral Palsy: Extrapyramidal or dyskinetic type

Acute Bilirubin Encephalopathy

Phototherapy

- Early - Hypotonia, poor feeding, loss of Moro's reflex
- Late - Hypertonia, seizures, coma, opisthotonos, Death

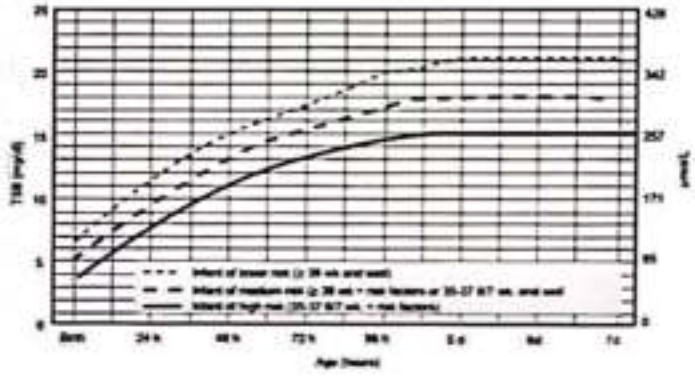
Chronic Bilirubin Encephalopathy (Long Term Sequelae)

- Mnemonic: "SADMUM"
  - ○ Sensorineural Hearing Loss (SNHL)
  - ○ Athetosis
  - ○ Dental changes
  - ○ Mental retardation
  - ○ Upward Gaze limitation

R, of NNJ

- Phototherapy
- Exchange transfusion } Used mainly in Rh incompatibility (Hydrops Fetalis)
- Drugs E.g. IVIg

Phototherapy



AAP nomogram for phototherapy

Important Information

- Most effective wavelength in phototherapy: 450-460 nm
- Most important mechanism: Structural isomerization

Factors Affecting Effectiveness of Phototherapy

- Exposed surface area of baby
- Type of Lamp (LED more effective)
- Distance b/w baby & Phototherapy unit
- Does not depend on skin pigmentation

Previous Year's Questions

Q. In a neonate on Phototherapy, Bilirubin is converted into?

(AIIMS - Jun - 2020)

A. Biliverdin
B. Lumirubin
C. Urobilin
D. Stercobilin

Cut – Off levels (in an otherwise healthy neonate)

Age	Phototherapy	Exchange Transfusion
24 – 48 hrs	> 15 mg/dl	> 20 mg/dl
48 – 72 hrs	> 18 mg/dl	> 25 mg/dl
> 72 hrs	> 20 mg/dl	> 25 mg/dl

Important Information

- **Mean normal Hb of neonate? 16.5 g/dl**

Adverse effects of phototherapy

- Bronze baby syndrome (in c/o conjugated hyperbilirubinemia)
- Watery diarrhea
- Dehydration
- Hypocalcemia
- Retinal toxicity
- Gonadal toxicity

- Indication of Exchange Transfusion in Rh incompatibility
  - Cord blood Bilirubin > 5mg/dl or
  - Cord blood Hb < 10g/dl

Table 3.1

Conjugated Hyperbilirubinemia (Conjugated bilirubin > 2mg / dl or 20 % of Total Bilirubin)

Non-Obstructive

- **Infections**
- **Metabolic**
  - α1Anti trypsin def (PAS +ve globules on liver biopsy)
  - Galactosemia
  - Tyrosinemia
  - Cystic Fibrosis
- **Idiopathic Neonatal Hepatitis:** mc cause of conjugated Hyper bilirubinemia in Neonates

Obstructive causes

Intrahepatic

- Congenital hepatic fibrosis
- Caroli's disease
- Alagille syndrome
- Dubin johnson syndrome
- Rotor syndrome

Extra hepatic

- Biliary Atresia
  - Screening: HIDA scan or hepatic scintigraphy
  - Sx: Kasai procedure
  - MC indication of liver transplantation in children is Biliary atresia

NUTRITION

BREAST FEEDING (BF)

02:15:30

Q. Should BF be initiated?

- As soon as possible (best answer)
- Within 1 hr. of child birth (if a time frame is asked)

Important Information

Exclusive Breast Feeding Is Recommended for How Long?

- 6 months
- After 6 Months: Complementary feeding initiated

Q. If Food Grain Introduced at Earlier Age

- More chances of food allergy.

Q. Storage of Expressed Breast Milk Can Be Done for How Long?

- At room temp: 8 - 10 hrs
- In refrigerator 24 hrs
- Deep freezer (-20 °C) 3 months

Q. Signs of Good Attachment While Breast Feeding

- Mouth wide open
- Entire areola in baby's mouth except small upper part that may be visible
- Lower lip everted
- Chin should touch Mother's breast

Previous Year's Questions

Q. Breast feeding contraindicated in?

(JIPMER - Dec - 2019)

A. MDR Tuberculosis
B. Zika virus infection
C. Hepatitis B infection
D. Mastitis with abscess

Q. C/I of Breast Feeding

- Baby factor: Confirmed c/o Galactosemia

Maternal Factors	
Absolute	**Relative**
o Chemotherapy or Radiotherapy	o Maternal HIV o Active TB o Active varicella / Herpes o Breast Abscess

Composition of Breast Milk

02:19:50

Previous Year's Questions

Q. Amount of Protein present in 100 ml of breast milk is?

(JIPMER - May - 2019)

A. 2.2 g
B. 1.1 g
C. 0.55 g
D. 3.35

- Carbohydrate: Lactose concentration in BM (7 g / dl) > Cow Milk (CM) (4.5 g / dl)
- Proteins
  - BM (1 g / dl) < CM (3.5 g / dl)
  - BM richer in whey protein (lactalbumin) which is more easily digestible than casein in CM.
  - BM contains adequate amount of Cysteine, Taurine, Methionine - Vital for CNS development
- Lipids
  - BM is richer in PUFA & DHA (Docosahexaenoic Acid) → helps in CNS development of baby
- Minerals
  - Iron in BM: more easily absorbable
  - Ca: P ratio in BM Favours Calcium absorption
  - (CM → more phosphate; leads to less calcium absorption; therefore, predominantly CM fed baby → risk of hypocalcemia & scurvy)
- Vitamins
  - BM contains adequate amount of all vitamins except
  - Vit D → (Supplement 400 IU / day throughout infancy)

Previous Year's Questions

Q. Fat content of breastmilk?

(JIPMER - Dec - 2019)

A. 2.4 percent
B. 3.4 percent
C. 4.4 percent
D. 5.4 percent

- Vit K: (1 mg IM to all babies at birth to prevent hemorrhagic disease of new born)
- Vit B_{12}: (In strictly vegan mothers)

- Anti - infective Substances Present in BM Are
  - Mnemonic: "Teach for PLAB"
    - → TGF - β
    - → Phagocytic macrophages
    - → PABA (Para Amino Benzoic Acid)
    - → Lactoferrin
    - → Lysozyme
    - → Antibodies especially IgA
    - → Anti - staph factor
    - → Bifidus factor
    - → Bile stimulated lipase
  - Colostrum
    - → Seen in 1st 72 hrs. after birth
    - → Thick & yellowish
    - → Scanty in amount

Important Information

- **Colostrum is Rich in proteins, Ig, Macrophages**

→ Poorer in lactose
→ "1st immunization of baby"
→ Any Pre lacteal feed is strictly contraindicated

MARASMUS

Kwashiorkor

MALNUTRITION

- Best indicator of
  - Acute malnutrition: ↓ in wt for Ht (wasting)
  - Chronic malnutrition: ↓ in Ht for age (stunting)

	Kwashiorkor	Marasmus
• Edema	Present	Absent
• Appetite	Poor	Voracious
• CNS	Apathy, Lethargy	Active & Alert
• Hepatomegaly	Seen	Not seen
• Skin & hair changes	More common (Skin: Flaky Paint Dermatosis) (Hair: Flag sign)	Less common

Previous Year's Questions

Q. Severe acute malnutrition as per WHO criteria?

(NEET - JAN - 2019)

A. Weight for age - 2 SD less than median
B. Weight for height - 2 SD less than median
C. Weight for age - 3 SD less than median
D. Weight for height - 3 SD less than median

WHO classification of Malnutrition in based on

- Weight for Height
- Height for age
- Edema

Severe Acute Malnutrition (SAM) 02:27:43

- Def: In a child of 6 m - 5 yrs. of age, presence of 1 or more of the following

- Weight for Height < -3 z score or < 70 % of expected (severe wasting)

or

- Mid upper arm circumference < 11.5 cm

or

- Symmetric bipedal edema of nutritional origin.

Previous Year's Questions

Q. All are diagnostic criteria's for 'Severe acute malnutrition' (SAM) except?

(JIPMER - Nov - 2018)

A. Mid-upper arm circumference (MUAC) < 115 mm
B. Weight for age < -3 Z score
C. Presence of bipedal edema
D. Presence of visible severe wasting

Complications of SAM: (With Treatment)

- Mnemonic: "SHIELDED"
  - Sugar deficiency (Hypoglycemia) (BG < 54 mg/dl): 10 % Dextrose
  - Hypothermia (Rectal temp < 35.5C): Warm up
  - Infections: Antibiotics
  - Electrolyte imbalance (Hypokalemia / Hypophosphatemia): Supplement K, Phosphate
  - Dehydration: WHO ORS (in double dilution) / Resomal - Rehydration solution for malnourished child (↓Na, ↑K)
  - Deficiency of Micronutrients
    - → Supplementation
    - → Iron should be started 1 - 2 weeks later

Rx of SAM

- Hospitalization
- Rx of complications (as mentioned above)
- Nutritional Rehabilitation

Important Information

Nutritional Rehabilitation

- **Should be gradual to prevent "Nutritional recovery syndrome" or "Re - feeding syndrome"**
  - Start with low calories & proteins & gradually build up
    - → Initially F75: 75 kcal / 100 ml
    - → Then F100: 100 kcal / 100 ml
    - → Later RUTF: 543 kcal / 100g (Ready to Use Therapeutic Food).

Fluids & Electrolytes 02:32:39

- Total body Water (TBW)
  - → 75 % of body weight at birth
  - → 60 % of body wt. by 2 years of age & remains so till puberty
- Calculation of 24 hrs. maintenance fluid requirement
  - → 1ST 10 kg: 100 ml / kg
  - → Next 10 kg: 50 ml / kg
  - → Beyond 20 kg: 20 ml / kg

Important Information

- **Fluid of choice: 1\2 DNS with added Potassium.**

- In neonates
  - → Birth weight > or = 1500 g: start with 60 ml / kg / day
  - → BW < 1500 g: start with 80 ml / kg / day
  - → By Day 7 of life: 150 ml / kg / day
  - → 1st 48 hours IV fluid of choice: 10 % Dextrose with no electrolytes

Fluid of Choice for Shock in Children - Normal Saline (NS)

- NS 20 ml / kg upto 3 times (bolus) + O_2 + IV Antibiotics (suspected septic shock)
- If No improvement: Inotropes (e.g.: Dopamine, Dobutamine, Epinephrine, norepinephrine)

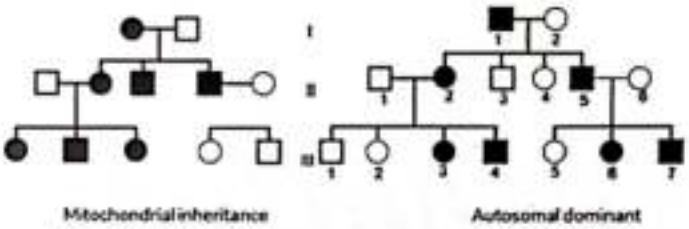
GENETICS

Pedigree Analysis ⏱ 02:36:26

- If all children of an affected female have the disease - Mitochondrial inheritance
- If at least 1 of the parent is affected by disease - Dominant Inheritance

Important Information

- Father to Son transmission rules out X - linked disease



Mitochondrial inheritance | Autosomal dominant

- E.g. Fragile X Syndrome
  - Trinucleotide repeat disorder: increase in no. of CGG repeats

Previous Year's Questions

Q. True about Fragile X syndrome is?

(NEET - Jan - 2019)

A. Triple nucleotide CAG Sequence mutation
B. 10% Female carriers mentally retarded
C. Males have IQ 20-40
D. Gain of function mutation

  - Intellectual Disability
  - Anticipation: worsening with each successive generation seen
  - Genomic Imprinting
    - → E.g. Prader: Willi syndrome
    - → Beckwith: Wiedemann syndrome
    - → Russel: Silver Syndrome
    - → Angelman syndrome
- Examples of Mitochondrial Inheritance

Previous Year's Questions

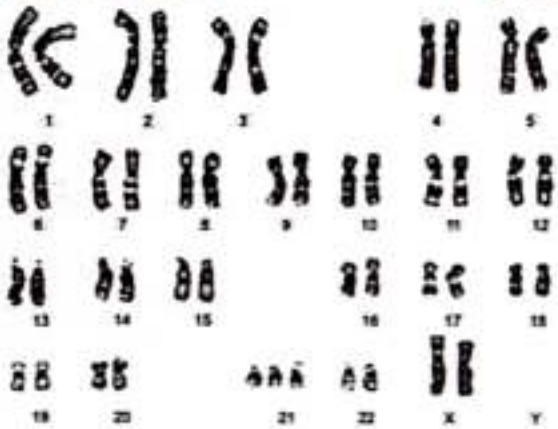
Q. An affected male does not have affected children but affected female always has affected children. Type of inheritance?

(AIIMS - May - 2019)

A. X linked recessive
B. Autosomal recessive
C. X linked dominant
D. Mitochondrial

  - Kearns Sayre syndrome
  - Leber's hereditary optic Neuropathy (LHON)
  - MELAS syndrome (Mitochondrial encephalomyopathy, Lactic Acidosis and stroke like episodes)
- Examples of Multifactorial Inheritance
  - Neural Tube Defects
  - Cleft Palate
  - Hypertension

DOWN SYNDROME ⏱ 02:41:39

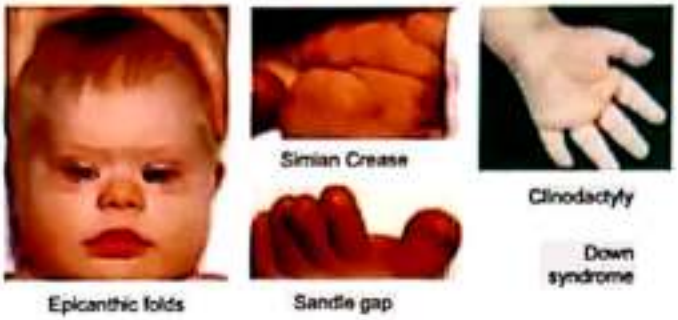


- Trisomy 21
- MC: d/t maternal meiotic non - disjunction
- Risk increases with increase in mother's age

Important Information

- MC heart disease in Down syndrome? AVSD (Atrioventricular septal defect) aka Endocardial cushion defect

Antenatal Screening for Down's Syndrome



USG

- Nuchal Thickness > 3 mm @ 11 - 14 weeks of gestation
- Absence of nasal bones
- Short femur
- Duodenal Atresia

Biochemical Tests

- 1st Trimester
  - β - HCG, PAPP - A (Pregnancy associated Plasma Protein - A) (Dual Test)
- 2nd Trimester
  - Triple Test: AFP (α - Fetoprotein) + β - HCG + Unconjugated Estriol
  - Quadruple Test: Triple Test + Inhibin
    → Mnemonic - HI High (↑↑)
    → HCG & Inhibin levels increase in Down syndrome

Integrated Test

- Best test for screening Down's syndrome (Sensitivity 95 - 97%)
- Maternal age + 1st Trimester (NT + PAPP - A) + Quadruple Test (2nd Trimester).
- Confirmatory test for antenatal Dx of Down syndrome Fetal karyotype, for which genetic material may be obtained by
  - Chorionic Villous Sampling (CVS)
  - Amniocentesis
  - Cordocentesis (Percutaneous Umbilical cord Blood sampling) But these are invasive procedures that carry risk of abortion.

TURNER SYNDROME (45, XO) 02:47:28

- Always seen in Females, as no Y chromosome is present
- Mnemonic - "See A Baby CLOWN"
  - S: Short Stature
  - A: Amenorrhea (1)
  - B: Barr Body Absent
  - C: Cardiac abnormalities, Cystic hygroma
- MC congenital cardiac Abnormality in Turner's Bicuspid Aortic Valve > Coarctation of Aorta
  - L: Lymphedema, Low thyroid
  - O: Ovaries underdeveloped – Infertility
- Turner syndrome patients with webbed neck have 3 times more chances of having CHD, than those without webbed neck
  - W: Webbed neck
  - N: Nipples widely spaced (shield shaped chest)

Previous Year's Questions

Q. Which of the following is true statement regarding Turner's syndrome?
(AIIMS - Nov - 2018)

A. Turner syndrome with webbed neck is 10 times more likely to develop CVS defects than non-webbed neck
B. Coarctation of aorta is more likely in non-webbed neck
C. Male turner (Noonan syndrome) is much more likely to have CVS defects
D. Turner syndrome with webbed fingers and toes is likely to be associated with visceral anomalies

NOONAN SYNDROME 02:50:47

Noonan Syndrome

- AD inheritance, mc gene PTPN11 gene (deletion)

Similarities with Turner's	Differences with Turner Syndrome
○ Short stature ○ Webbed Neck ○ Cubitus valgus ○ Shield shaped chest with widely spaced nipples	○ Can be seen in both boys and girls ○ Karyotype Normal ○ Anti - mongoloid slant of eyes ○ Intellectual disability ○ Delayed puberty but fertility is normal

- 80 % of Pts with Noonan syndrome have Congenital Heart Diseases (CHD).

Important Information

- MC CHD in Noonan: Pulmonary stenosis (also seen in hypertrophic obstructive cardiomyopathy)

INBORN ERRORS OF METABOLISM

02:53:38

A. DISORDERS OF CARBOHYDRATES

- **Glycogen Storage Disease**

i. Predominantly Liver Involvement (Liver Glycogenosis)

Type	Name	Enzyme Deficiency
I	Von Gierke's Disease	Glucose 6 phosphatase
III	Cori's disease	Debranching enzyme
IV	Anderson's Disease	Branching Enzyme
VI	Hers Disease	Hepatic Phosphorylase

Important Information

- **Von Gierke's Disease: MC GSD in Children**

ii. Predominantly Muscle Involvement (Muscle Glycogenosis)

Type	Name	Enzyme Deficiency
II	Pompe diseases	Acid maltase
V	McArdle's disease	Muscle Phosphorylase
VII	Tarui's Disease	Phosphofructokinase

- Mnemonic: 2 + 5 = 7 (Types II, V & VII are muscle glycogenosis)

Important Information

- **MC GSD in Adolescents & Adults → MC Ardle's Disease**

Von Gierke's Disease 02:57:37

- Autosomal recessive
- Recurrent hypoglycemia
- Doll like facies
- Hepatomegaly
- Easy bruising

Investigations

- Hypo - glycemia
- Hyper - uricemia
- Hyper - lipidemia
- Lactic acidosis
- Treatment: Corn Starch Diet

Hypoglycemia with Hepatomegaly with Hyperlipidemia seen in Both

	Type I GSD (Von Gierke's)	Type III GSD (Cori's)
Muscle involvement	Not Involved	May be present
CPK levels	Normal	Elevated
Response to glycogen	No increase in blood glucose but lactic acid increases	Increase in blood glucose in fed state, but not in fasting state, lactic acid levels are normal

Pompe Disease 03:01:20

- Cardiac muscle also involved
- Presentation: Child + Hypotonia + Myocardial dysfunction or cardiomegaly

1. Galactosemia Deficiency of
   - GALT (mc) (Galactose - 1 - Phosphate uridyl transferase)
   - Galactokinase
   - Epimerase
   - Duarte variant: mild - usually asymptomatic
   - Clinical features
     - → Jaundice
     - → Diarrhea
     - → Vomiting
     - → Hepatomegaly
     - → Hypoglycemia
     - → Cataracts
     - → CNS abnormalities

Important Information

- Cataract can be the sole manifestation of Galactokinase deficiency d/t accumulation of Galactitol

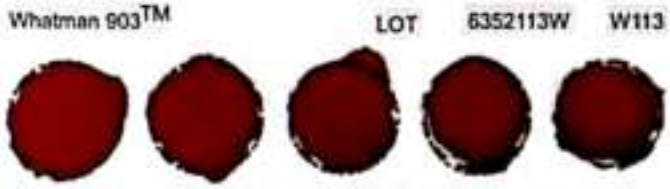
- Sepsis with E. coli is most common
- **Treatment**
  - → Eliminate milk & milk products from diet
  - → Breast feeding contraindicated in confirmed cases.

2. Hereditary Fructose intolerance (HFI)
   - Deficiency of Aldolase B
   - Aversion to sweet food (Sucrose = Glucose + fructose)
   - Reducing substance in urine present.
   - Rx: eliminate fructose from diet

B. DISORDERS OF AMINO ACID METABOLISM

03:06:19

- Screening Tests for Metabolic Disorders



Dried blood spots for TMS

- TMS (Tandem Mass spectrometry) - Using dried blood spots
- GCMS (Gas chromatography mass spectroscopy) → Urine sample
- Electrophoresis of Plasma / Urine
- HPLC (High performance Liquid Chromatography) of Plasma / Urine

Phenylketonuria (PKU)

- Enzyme deficiency: Phenylalanine Hydroxylase

  ↓

  X
- Phenylalanine: Tyrosine
- In PKU, tyrosine becomes an essential amino acid
- Tyrosine gives rise to DOPA & melanin

Clinical features

- Hypopigmentation (fair skin, blond hair, blue iris)
- Hypertonia
- Intellectual disability

Investigations

- Plasma HPLC / TMS: Elevated phenylalanine & its metabolites

Important Information

- Guthrie test → Biological assay for PKU

- Ferric chloride test
- Rx
  - Low phenylalanine diet with Tyrosine supplementation

Alkaptonuria

- Deficiency of Homogentisic Acid Oxidase
- Clinical features
  - Dark spots on sclera & ear cartilage (ochronosis)
  - Darkening of urine on standing

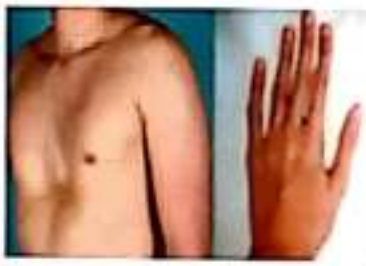
Previous Year's Questions

Q. A patient complains of knee pain. Routine investigations are unremarkable and still, the patient is unsatisfied. Urine turns black on standing. what is the enzyme involved?
(NEET - Jan - 2020)

A. Homogentisate oxidase
B. Xanthine oxidase
C. Methyl malonate oxidase
D. Phenyl pyruvate oxidase

Homocystinuria

- Classical Type: Deficiency of cystathionine β - synthase
- Clinical features
  - Skeletal manifestations like Marfan syndrome (tall stature, arachnodactyly, Pectus excavatum)
  - Subluxation of lens of eye: Infero – medial
  - Recurrent Strokes



Marfanoid habitus

Subluxation of lens

- Treatment
  - Vit B6 & vit B12 have some role
  - In Marfan's: Supero - lateral (Mnemonic: MSL)

Multiple Carboxylase Deficiency 03:14:04

Important Information

- **Cofactor for carboxylase enzyme: Biotin (Biotin Skin & / or Hair involvement)**

- Presentation
  - Intellectual disability
  - Alopecia
  - Eczema
  - Tom Cat Urine Odour
- Treatment
  - Biotin

Hartnup Disorder

- Mutation of SLC6A19 gene: Problem with transport of neutral amino acids

Previous Year's Questions

Q. False about hartnup's disease?
(JIPMER - May - 2019)

A. Defect in neutral amino acid transport
B. Mental retardation is the common presentation
C. Most children are asymptomatic
D. Photosensitivity

Pellagra like rash

- Mostly asymptomatic
- Pellagra like rash (Casal's necklace) & Photosensitivity
- Rx: Nicotinic Acid

Tyrosinemia

- Type 1: Deficiency of fumarate acetoacetate hydrolase (FAH)
- Clinical features: Infant presenting with
  - Hepatomegaly
  - Jaundice
  - Bleeding manifestations

Investigations

- Prolonged prothrombin Time (PT)
- Elevated Urine succinyl acetone: used for screening
- Rx: Nitisinone

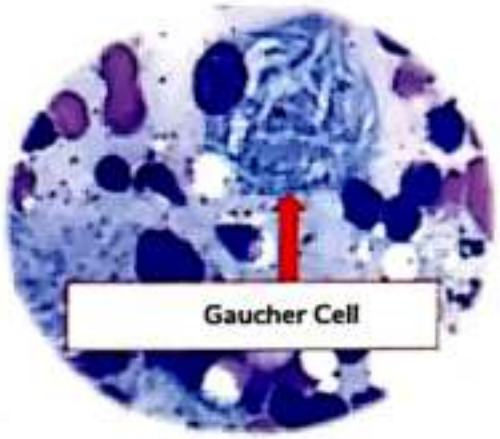
Maple Syrup Urine Disease (MSUD)

- Metabolism of Branched chain amino acids affected (Isoleucine, Leucine & Valine)
- Deficiency of α - ketoacid dehydrogenase
- Burnt sugar / maple syrup odour from body fluids

C. LYSOSOMAL STORAGE DISEASES (LSD)

03:18:33

- **Gaucher Disease**



- It is the most common LSD in children

Important Information

- **Gaucher disease: Deficiency of Glucocerebrosidase enzyme**

- Accumulation of Glucocerebrosides in cells

↓

"Gaucher cells" in Bone marrow, liver etc
(Crumpled tissue paper/ wrinkled paper appearance of cytoplasm)

Clinical features

- Splenohepatomegaly (Splenic enlargements >> Liver)
- Pancytopenia (D/ t bone marrow infiltration)
- Anemia: easy fatigability
- Thrombocytopenia: bleeding
- Neutropenia: recurrent infections
- Bone pains
- Neurological features +/-

Treatment

- Enzyme Replacement Therapy
- Enzyme Replacement Therapy is Available for
  - Gaucher's disease
  - Hurler's disease (Type I MPS)
  - Maroteaux - lamy disease (Type VI MPS)
  - Pompe's disease
  - Fabry's disease (α galactosidase deficiency) (angiokeratomas seen)

Niemann Pick Disease

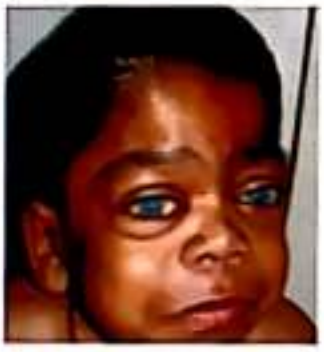
- Deficiency of sphingomyelinase
- Features like Gaucher's disease

- Cherry Red spot seen (Usually not seen in Gaucher's disease)

> **Important Information**
>
> Cherry Red spot seen in
> - Niemann pick disease
> - Tay sachs disease
> - Gm1 gangliosidosis

D. MUCOPOLYSACCHARIDOSIS (MPS)

03:22:02



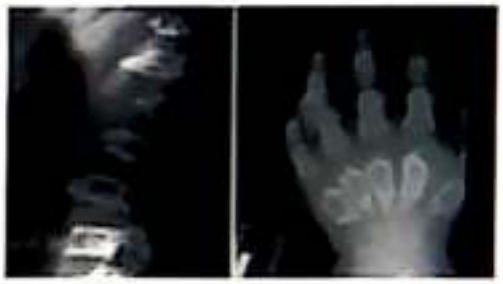
Hurler's Disease

> **Important Information**
>
> - Type I - HURLER's Disease → α - L - Iduronidase deficiency → AR

- Clinical features
  - Coarse Facies
  - Hepatosplenomegaly
  - Corneal opacity
  - Copious Nasal discharge
  - Airway problems
  - Intellectual disability
- Type II - HUNTER'S → X Linked Recessive Inheritance
- All MPS Are AR Except Type II
  - In Type II all features of Type 1 MPS seen except corneal opacity

> **Previous Year's Questions**
>
> Q. A 5-year-old boy has peculiar facial features, enlarged head, hepatosplenomegaly, protuberant abdomen, breathing difficulty with obstructive sleep apnea and cardiac valve thickening. What is the likely diagnosis?
> (AIIMS - Nov - 2018)
>
> A. Hurler's disease
> B. Hunter's disease
> C. Fragile X syndrome
> D. Tay-Sachs disease

Bony Abnormalities in MPS: Dysostosis Multiplex



Dysostosis multiplex in Mucopolysaccharidoses

- Most severe in Type IV MPS (Morquio's disease)
- Anterior beaking of vertebrae
- Egg shaped / Bullet shaped metacarpals
- Coarse facies

Menke's Disease

> **Important Information**
>
> - ATP7A gene involved in Menke's Disease

- Disorder of copper metabolism
- Clinical features

- Seizures
- Developmental delay
- Abnormal kinky hair (Microscopically: Trichorrhexis nodosa & Pili torti)

Hair microscopy in Menke disease

- Hypopigmentation

F. Lesch Nyhan Disease 032506

- Deficiency of HGPRT enzyme (Hypoxanthine Guanine Phospho - ribosyl transferase)
- Clinical features
  - Developmental delay
  - Self-injury
- Investigation: ↑ Uric Acid
- Treatment
  - High fluid intake
  - Allopurinol

DISEASE OF IMMUNE SYSTEM

VASCULITIS

Henoch Schonlein Purpura 03:26:19

- Mc vasculitis in children.
- MC age group involved 3-10 years
- Diagnostic Criteria: Palpable purpura with at least 1 of the following
  - Arthritis/ arthralgia
  - Abdominal pain
  - Renal involvement
  - Any biopsy showing IgA deposition

Important Information

- Platelet counts N. Purpura d / t vasculitis & not thrombocytopenia

- 1/3rd of cases have Glomerulonephritis

Kawasaki Disease 03:27:52

- Diagnostic Criteria: Fever for > 5 days with any 4 out of following
- Mnemonic: "CREAM"
  - Conjunctivitis (non - purulent)
  - Rash (involving trunk)
  - Edema & Erythema of Extremities
  - Adenopathy (Lymphadenopathy)
  - Mucosal involvement (Strawberry tongue)
- Rx of Choice: IV - Ig

Important Information

- Complication: Coronary Artery Aneurysm

Kawasaki disease

Coronary angiogram in Kawasaki

INFECTIOUS DISEASES IN CHILDREN

1. VIRAL DISEASES

03:29:51

a. Varicella (Chicken Pox)

- Incubation period: 10 - 21 days

> **Important Information**
>
> - **Characteristic feature of varicella: pleomorphic rash (simultaneous presence of different types of skin lesions)**

- Remains infected till all vesicles are crusted
- Congenital Varicella Syndrome

Chicken pox

  - H/O Rash & fever in mother
  - Baby has cicatricial skin scars in zoster like distribution (dermatomal distribution)
  - Limb abnormalities
  - Low birth weight.

b. Rubella

- Mild exanthematous illness + lymphadenopathy

> **Important Information**
>
> - **Forchheimer spots seen in oropharynx**

- Congenital Rubella Syndrome
  - Risk of congenital defects is maximum before 11 weeks of gestation.
- Clinical features (Mnemonic "CDC")
  - **Cataract**
  - **Deafness**
  - **Congenital heart diseases (Most common is PDA)**
  - IUGR
  - Microcephaly
  - Blue berry muffin lesions
  - Retinopathy

> **Important Information**
>
> - **Most common manifestation of CRS: Sensorineural deafness**

  - Late onset manifestations of CRS → Diabetes mellitus, Thyroid dysfunction, Rubella Panencephalitis.

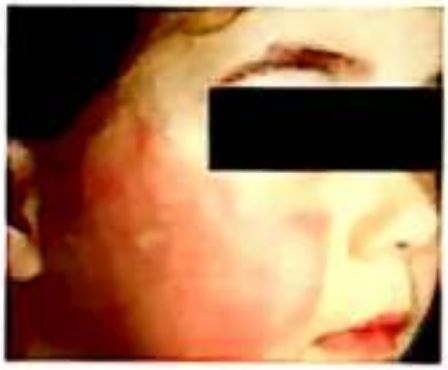
c. Measles

- Incubation period: 8 - 10 days

> **Important Information**
>
> - **Koplik Spots (Pathognomonic) - Buccal mucosa, conjunctiva, vagina**
> - **Warthin Finkeldey giant cells on biopsy of skin lesion.**

- Receptor for Measle Virus: CD150 & PVRL4
- Mc complication: Otitis media
- Mc cause of death: Pneumonia
- Long standing complication: SSPE (Subacute Sclerosing Pan Encephalitis) (usually fatal)

d. Erythema Infectiosum



Erythema infectiosum

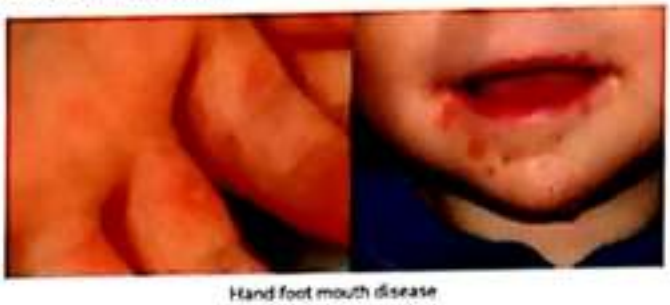
- Caused by Parvovirus B
- "Slapped cheek" Appearance of child

e. Hand Foot Mouth Disease

- Coxsackie Virus A16
- Plantar & Palmar pustules
- Oral ulcers

- Mild illness, self - limiting

f. Roseola Infantum



Hand foot mouth disease

- HHV 6A & 6B

Important Information

- **In Roseola Infantum, Nagayama spots are seen**

- Rash appears when fever subsides

g. HIV in Children

- Preferred diagnostic test in neonate & infants
  - HIV DNA PCR
  - HIV RNA PCR
  - HIV culture (Not Serology based tests)

Important Information

- **Prophylaxis to Infants born to mother with HIV → Nevirapine +/- Zidovudine**

- Pregnant lady / Mother should receive ART
- In developed countries: In babies born to mothers with HIV, breast feeding is contraindicated
- In developing countries: Continue breast feeding, as the benefits outweigh the risks.

h. H1N1

- DOC: Oseltamivir
- Dose:
  - Infants: 3 mg/kg
  - < 15 kg: 30 mg
  - 15 - 23 kg: 45 mg
  - 24 - 40 kg: 60 mg
  - > 40 kg: 75 mg (Adult dose)

} B.D. X 5 days

i. CMV

Important Information

- **MC cause of non-syndromic hearing loss in children → Congenital CMVrn to mother with HIV → Nevirapine +/- Zidovudine**

- 90 % cases asymptomatic
- Best sample for CMV PCR: urine

j. MUMPS

- Acute U/L or B/L Parotid gland enlargement along with fever
- Orchitis in common in adolescent males
- Mc complication: Aseptic meningitis

Previous Year's Questions

Q. An 18-month baby with poor feeding & fever for 3 days is brought with lower limb weakness. On examination, he has Lethargy, abnormal movement of lower limbs & anterior fontanelle is bulging. diagnosis?

(AIIMS - May - 2019)

A. Intra cranial hemorrhage
B. Pseudotumor cerebri
C. Meningitis
D. Cerebral palsy

2. PEDIATRIC TB

- In Primary TB, Ghon focus is seen in: Lungs
- In congenital TB, Ghon focus is seen in: Liver

Important Information

- **MC route of infection of baby born by C- section: Airborne**

- INH Prophylaxis recommended for all infants with exposure to TB
- Dose: Isoniazid 10mg day for 6 months
- Treatment
  - Daily treatment, instead of thrice weekly for all Pediatric TB patient.
  - Only Pyrazinamide should be stopped in continuation phase.

3. CONGENITAL TOXOPLASMOSIS

- Risk of fetal infection increases with each trimester
- T1 - 15 %
- T2 - 25 %
- T3 - 60 %
- But severity of infection is greater, if infected early in pregnancy.
- Clinical features
  - Chorioretinitis
  - Hydrocephalus
  - Cerebral calcifications
  - Treatment: Sulphadiazine with Pyrimethamine & leucovorin
- Immunization
  - Vaccines that must be given beyond
    - → 2 years: Polysaccharide vaccines
    - → 9 years: HPV vaccine
  - Vaccines recommended in adolescents
    - → Tdap, TT, Td, Hep - B
    - → HPV, influenza
    - → Japanese encephalitis (In endemic area)
    - → Pneumococcal polysaccharide vaccine
    - → Rabies vaccine

    } If indicated
  - In an unvaccinated child ~ 18 m age
  - 1st visit: BCG (Catch up till → 1 years (acc to NIS)
    - → 5 year (acc to (AP)
    - → OPV (till 5 year of age)
    - → Hep B (any age)
    - → DTP (up to 7 year) 0,1,6
- Hib: Catchup age
  - 6-12 m: 2 doses + 1 booster
  - 12 - 15 m: 1 dose + 1 booster
  - > 15 m: 1 dose
- MMR vaccine (< 1 year's measles) 2 to 3 doses
- Vaccines C/I in Egg Allergy: Influenza vaccine & yellow fever vaccine

Previous Year's Questions

Q. All of the following will be benefitted by the 23-valent pneumococcal vaccine EXCEPT?

(AIIMS - Nov - 2019)

A. Recurrent otitis media
B. Cystic fibrosis
C. Sickle cell anemia
D. Less than 2 yrs. age
E. Lupus nephritis

Important Information

- **Vaccine That May Cause Thrombocytopenia?**
  Measles vaccine
- **Strains Covered in Meningococcal Vaccine?**
  A, C, Y, W-135

- Maximum efficacy after single dose: MR > TT

BCG

Previous Year's Questions

Q. BCG is maximally protective against:

(AIIMS - Nov - 2018)

A. Pulmonary TB
B. Pulmonary and CNS TB
C. CNS and Disseminated TB
D. Extra pulmonary TB
D. Lupus nephritis

- Route: intra dermal
- Protection against CNS & disseminated TB (not Pulmonary TB)
- S/E: Osteitis, BCG adenitis

DPT

- Adverse effects: Persistent crying (mc)
- Hypotonic Hyporesponsive episodes
- Fever
- Seizures
- Encephalopathy

Previous Year's Questions

Q. A 5yr old unimmunized child developed Diphtheria. He has a 3yr old immunized sibling contact, who received last booster 18 months back. What to do with the contact?

(NEET - Jan - 2020)

A. Two doses of polysaccharide vaccine
B. Three doses of conjugate vaccine
C. Single dose of toxoid vaccine
D. No vaccine needed

Clinical features

- H/o anaphylaxis to previous dose
- Any progressive neurological illness (static illness like CP

not a c/l)
- Encephalopathy within 7 days of vaccination

Polio
- Live vaccine → Oral → Sabin
- Killed vaccine → Injectable → Salk
- Serotypes → P1 & P3 (Bivalent)

VAPP
- Vaccine associated Paralytic Poliomyelitis
- Definition of VAPP: Those case of AFP, who have residual weakness 60 days after the onset, and from whose stool sample vaccine related virus is isolated. (not wild virus)

MMR
- Live vaccine
- 3 doses
- 9 m, 15 m, 4 - 6 years
- National Immunization Schedule → Salk
- MR vaccine at 9 m & 16 - 24 m (special vaccination drive → To catch up → extra dose given)
- Strain: Edmonston Zagreb strain

Previous Year's Questions

Q. An un-immunized 13 months old child comes to you in OPD, according to the latest immunization schedule, what vaccines will you advise?
(AIIMS - May - 2018)

A. OPV 3 doses, IPV, 3 Pentavalent,
B. BCG, OPV 3 doses, 3 IPV, 3 Pentavalent and 1 measles
C. OPV 3 doses, 1 IPV, 3 Pentavalent and 2 measles
D. OPV 3 doses, 3 IPV, 3 DPT, 3 Hep. B

Rotavirus Vaccine
- 116 E strain
- Age of initiation – 6 weeks - 15 weeks
- Oral live attenuated vaccine
- Clinical features
  - H/o Intussusceptions
  - SCID
  - Hypersensitivity to any vaccine components.

Hep A
- Killed / inactivated: 2 doses
- Live - single dose: 12 - 33 months

Important Information

- **Vaccine for cholera?**
  Dukoral
- **Strain of Yellow fever vaccine?**
  17 - D strain (live attenuated)
- **New Dengue vaccine?**
  Dengvaxia

PEDIATRIC CARDIOLOGY

03:57:59

- Oxygenated blood in fetus is carried by Umbilical Vein (O_2 Saturation ~ 80 %)

Important Information

Innocent Murmur
- Heard till 7 years of age
- Best heard at left lower sternal border.

- NADA's criteria is used for assessment of CHD

Acyanotic Congenital Heart Disease
- MC congenital heart disease: VSD (mc type of VSD → membranous)
- MC CHD most affected by IE: VSD
- MC ASD type: Ostium secundum (Least affected by IE)

Previous Year's Questions

Q. On repair of VSD, the patient will show improvement in which of the following?
(AIIMS - Jun - 2020)

A. Arrhythmia
B. Heart block
C. Respiratory alkalosis
D. Failure to thrive

Important Information

Innocent Murmur
- DOC for closure of PDA in Preterm neonates Indomethacin > Ibuprofen
- DOC for keeping PDA patent? PGEI Analogue or Alprostadil

Imp Auscultation Findings in CHD
- VSD: Pansystolic murmur
- ASD: Wide fixed split of second heart sound
- PDA: Continuous machinery murmur
- TOF: Ejection systolic murmur in pulmonary area and single S2 (P. soft and inaudible)

Previous Year's Questions

Q. Where to look for pre-ductal O2 saturation in PDA in a 3-minute born infant?
(NEET - Jan - 2019)

A. Left upper limb
B. Left lower limb
C. Right upper limb
D. Right lower limb

Cyanotic CHD
- MC congenital cyanotic heart disease in
  - Children: TOF
  - Neonates: TGA
  - Causing death in 1st week: Hypoplastic left heart syndrome (HLHS)

Tetralogy of Fallot (TOF)

'Boot Shaped' heart in Tetralogy of Fallot

- 4 Components
  - Large VSD
  - Pulmonary infundibular stenosis
  - Overriding of aorta
  - Right ventricular Hypertrophy
- Heart failure is not seen in TOF
- Cyanotic spells seen: During spell, murmur decreases or becomes absent

Treatment Of Cyanotic Spells
- Squatting / knee chest position
- O2
- Injection sodium bicarbonate
- Injection morphine / Ketamine
- α agonist (E.g. phenylephrine)
- β blockers (E.g. propranolol)
- Surgical Rx of TOF: Shunt Surgeries

Name	Pulmonary Artery Connected To
Blalock taussig shunt	Subclavian Artery
Waterston's shunt	Ascending Aorta
Pott's shunt	Descending aorta

Previous Year's Questions

Q. Administration of PGEI infusion will deteriorate condition in which of the following?
(JIPMER - Nov - 2018)

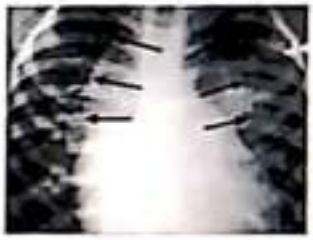
A. TGA with intact ventricular septum and restrictive foramen ovale
B. HLHS with restrictive foramen ovale
C. Mitral atresia with restrictive patent foramen ovale
D. Supracardiac total anomalous pulmonary venous connection

CXR Findings in CHD

- Boot shaped Heart, or 'Cor-en-sabot appearance': TOF
- Notching of inferior margins of Ribs or figure of '3' or 'E' sign: Coarctation of aorta
- Egg on side: TGA
- Snowman or Figure of '8' appearance: Supracardiac TAPVC
- Box shaped heart or large shadow of heart: Ebstein anomaly (also seen are ' Himalayan' P waves on ECG)

'Egg on side' in transposition of great arteries (TGA)

Coarctation of aorta



Snowman or figure of 8 in Supracardiac TAPVC

Previous Year's Questions

Q. Pulmonary plethora in a child presenting with cyanosis, is seen in?
(NEET - Jan - 2020)

A. Tetralogy of Fallot
B. Total anomalous pulmonary venous connection
C. Coarctation of aorta
D. Tricuspid Atresia

Recent Changes in Diagnostic Criteria of RHD

- Separated criteria for low risk & moderate to high-risk population
- Dx of recurrent Acute RF can be made by presence of 3 minor criteria
- Definition of carditis expanded to include subclinical evidence also (Like MR on ECHO)

BP In Children

- Age appropriate cuff size should be used
- Smaller cuff size → BP recorded is higher than Normal

PEDIATRIC GASTROENTEROLOGY

04:11:00

Q. A Neonate presents with excessive frothing from mouth & difficulty in feeding. There is a H/O Polyhydramnios in the antenatal period.

- Diagnosis: Esophageal Atresia with TEF (Tracheo - Esophageal Fistula)
- Mc type: Type C (Proximal end → blind; Distal end - connected to trachea)

Q. A 2–3-year-old child with recurrent diarrhea, abdominal distension, short stature, anemia, Failure to thrive

- Celiac disease

Previous Year's Questions

Q.A patient presented with diarrhea, poor appetite and malabsorption. His duodenal biopsy was taken which showed crypt hyperplasia, villi atrophy and infiltration of CD8+ T cells in the epithelium. What is the likely diagnosis of the patient?

(NEET - Jan - 2020)

A. Environmental enteropathy
B. Celiac disease
C. Whipple disease
D. Pancreatitis

Q. A 2-3 – year - old child with recurrent pneumonia, foul smelling bulky stool with oil in stools → Cystic fibrosis

Diarrhea

- MC cause of Diarrhea: Rotavirus (Worldwide), (All pediatric age group)

Important Information

- MC finding in diarrhea: Isotonic dehydration with acidosis

Treatment

1. WHO ORS (Reduced osmolality ORS)

- Na: 75 mEq/L
- Glucose: 75 mEq/L
- Osmolality: 245 mEq/L
- Treatment Plan A: No Dehydration
- Treatment Plan A: No Dehydration
- Replacement for ongoing cases: 5-10 ml/kg/loose stool
- Plan B: Some dehydration → 75 ml/kg over 4 hrs.
- Plan C: Severe dehydration → iv fluids → 100 ml/kg
- IV Fluid of choice for dehydration due to diarrhea: RL (with 5 % dextrose) (dextrose alone should not be used)

Previous Year's Questions

Q. Baby with diarrhea presented with restlessness. but was able to drink water. Skin turgor goes back in 2 sec. Best management is?

(NEET - Jan - 2018)

A. Plan A
B. Plan B
C. Plan C
D. Plan D

2. Zinc
   - < 6 months age - 10mg/day
   - > 6 m age - 20mg/day

   } 10 - 14 days
3. Continue normal diet
4. No antibiotics (except in c / o dysentery, cholera, SAM, Very sick child)

Previous Year's Questions

A: Antibiotics are the mainstay of treatment in neonatal diarrhea

B: Majority of neonatal diarrhea are viral. In this question. Statement (A) is the Assertion and Statement (R) is the Reason that explains the Assertion (Statement A)?

(AIIMS - May - 2019)

A. Both assertion and reason are correct and reason is correct explanation of assertion.
B. Both assertion and reason are correct but reason is not a correct explanation of assertion.
C. Assertion correct but reason wrong.
D. Reason correct, but assertion wrong.
E. Both assertion and reason are wrong.

Persistent Diarrhea

- Diarrhea of acute onset but lasting for > 14 days

Treatment

- Diet modification: Reduced lactose / lactose free diet / Elemental Diet.
- Vit A + zinc supplementation

PEDIATRIC RESPIRATORY SYSTEM

Most Common Causes ⏱ 04:18:42

- Stridor in infants: Laryngomalacia (Omega shaped epiglottis)
- Common cold / Coryza: Rhinovirus
- Croup / Laryngotracheobronchitis (LTB): Parainfluenza virus
- Acute epiglottitis (in vaccinated children): Streptococcus (Previously H. influenzae)
- Bronchiolitis: Respiratory syncytial virus (RSV)

Previous Year's Questions

Q. In exhausted child with severe bronchiolitis, for every 10 mm Hg increase in PCO2, how many milli eq of bicarbonate will increase? (JIPMER - Nov - 2018)

A. 2
B. 4
C. 8
D. 1

Important Information

- MC cause of Bacterial Pneumonia: Streptococcal pneumoniae

CROUP

- Clinical features
  - 3–4-year child / infant with low grade fever with barking cough & Stridor
- Treatment
  - Mild cases: Single dose dexamethasone
  - Moderate -severe cases: Single dose dexamethasone Add nebulized epinephrine.
  - No role of antibiotics, as it is a viral illness;

Previous Year's Questions

Q. Steeple sign is seen in? (JIPMER - Nov - 2018)

A. Influenza infection
B. Croup
C. Laryngomalacia
D. Acute epiglottitis

Approach to child with Cough and Difficulty Breathing

Fast Breathing	Chest Indrawing	General Danger Signs	Category	Rx
⊖	⊖	⊖	No pneumonia; only cough & cold	Home care advice
⊕/⊖	⊕/⊖	⊖	Pneumonia	Oral antibiotics + Homecare advice
⊕/⊖	⊕/⊖		Severe pneumonia or very severe disease	1st dose antibiotics and refer for admission

(IMNCI Color coding Green, yellow, Pink)
(General danger signs Persistent vomiting, unconsciousness, stridor in a calm child, severe malnutrition, Inability to drink or breast feed)

Previous Year's Questions

Q. A child presents with high grade fever, stridor and develops swallowing difficulty with drooling saliva. Along with airway management, which of following to be given? (AIIMS - Jun - 2020)

A. IV antibiotics
B. Steroids
C. Nebulized racemic epinephrine
D. Diphtheria anti toxin.

MDI + Spacer + Mask

BRONCHIAL ASTHMA

- Device used for inhalational therapy
- < 4 years: MDI + spacer + Baby Mask
- 4-12 years: MDI + spacer
- >12 years: MDI directly
- MDI (Metered Dose Inhaler)

Previous Year's Questions

Q. All are indicative of Pediatric Asthma EXCEPT?
(AIIMS - Jun - 2020)

A. Increase in FEV1 more than 15% after bronchodilator
B. AM PM Variation in FEV1 more than 15%
C. FEV1 Decreases more than 15% after exercise
D. FEV1/FVC less than 80%

FOREIGN BODY ASPIRATION

- Case scenario: H/o sudden choking & respiratory distress
- O/E: New onset wheezing (U/L)
- Treatment: Rigid Bronchoscopy & removal

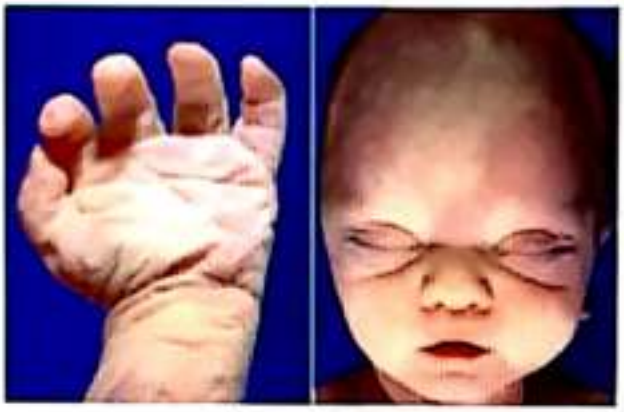
PEDIATRIC NEPHROLOGY

Potter's Sequence (Congenital Defect) 04:26:55

- Basic defect: B/L Renal Agenesis (other features – Facial dysmorphism, Oligohydramnios, limb abnormalities)

Important Information

- MC cause of death: Pulmonary Hypoplasia



Potter sequence

ARPKD (Autosomal Recessive Polycystic kidney disease)

- Antenatal USG: B/L enlarged, hyperechogenic kidneys
- Gene Involved: PKHD 1 gene on chromosome 6
- Neonate with B/L flank masses
- 50% will progress to end stage kidney disease by 10 years of age.

Previous Year's Questions

Q. A 1-year-old child was brought with sudden onset, multiple spasms: on examination he had shagreen patch and 4 hypomelanotic macules on extremities. What is the drug of choice for the seizures?

(JIPMER - Dec - 2019)

A. Carbamazepine
B. Phenytoin
C. Vigabatrin
D. Steroids

NEPHROTIC SYNDROME (NS) 04:28:54

- Generalized edema
- Massive proteinuria (U_{prot} U_{creat} > 2)
- Hypo albuminemia
- Hyperlipidemia
- Mc cause: Minimal change disease
- **Treatment**
  - Drug of choice for
    - → 1° episode NS - Prednisolone
    - → Relapse of NS - Prednisolone
    - → Steroid Dependent Nephrotic syndrome (SDNS) - Levamisole (In India) cyclophosphamide (In western countries)
    - → Steroid Resistant Nephrotic syndrome (SRNS) - Calcineurin inhibitors (cyclosporine & Tacrolimus)

Previous Year's Questions

Q. A 3-yr girl presents with generalized edema, shortly after recovery from an upper respiratory infection. Lab studies reveal marked albuminuria, hypoalbuminemia & hyperlipidemia. Prior similar episodes responded to steroid medication. The most likely diagnosis is?

(NEET - Jan - 2020)

A. Focal segmental glomerulosclerosis
B. Membranous glomerulonephritis
C. Minimal change disease
D. Post-streptococcal glomerulonephritis

NEPHRITIC SYNDROME 04:30:51

- Most common cause in children PSGN
- Clinical features
  - Hematuria, hypertension, mild edema, mild proteinuria
  - H/o respiratory or skin infection 2-3 weeks prior (Antecedent streptococcal infection)
  - Mc age group: 5-12 years
- **Hematuria**
  - If begins 2 - 3 weeks after respiratory infection: PSGN
  - If begins 2 - 3 days after resp infection: IgA Nephropathy or Berger's disease

Previous Year's Questions

Q. A Child with sensorineural hearing loss presented with hematuria. There is a history of chronic kidney disease in the grandfather, who was on dialysis. Diagnosis?

(AIIMS - Nov - 2019)

A. Alport syndrome
B. IgA nephropathy
C. Nephrotic syndrome

- PSGN
  - Investigation Findings
    - → Elevated ASO titers
    - → Low C levels: return to Ⓝ in 6 - 8 weeks.

HUS (HEMOLYTIC UREMIC SYNDROME)

04:32:54

Q. A child presents with oliguria / Petechiae (acute Onset) with h/o Diarrhea may or may not be present. Peripheral smear - schistocytes

- Diagnosis: Hemolytic Uremic Syndrome (HUS)
- Triad
  - Microangiopathic hemolytic anemia
  - Thrombocytopenia
  - Renal dysfunction
- Other Important Points

Important Information

- **Mc cause of hydronephrosis in children: PUJ obstruction**
- **Mc cause of obstructive uropathy in boy: PUV (posterior urethral valve)**
- **Mc cause of Renal scarring in children: VUR induced Pyelonephritis**

  - Mc cause of UTI in children: E. coli
  - Formula used to assess GFR in children with Renal failure: Schwartz Formula

$$eGFR = \frac{K \times HT\ (cm)}{\text{serum creatinine (mg / dl)}}$$

  - Value of 'k' varies with age; generally, for children → 0.4

PEDIATRIC NEUROLOGY

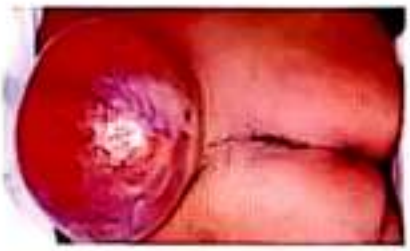
NEURAL TUBE DEFECTS

04:36:04

- Mc congenital abnormality in neurological system of children

> **Important Information**
>
> - **Antenatal markers: Maternal serum AFP, maternal Acetyl cholinesterase**

- Antenatal USG to detect severe malformations
- Myelomeningocele M/C involves lumbosacral region
- Prevention: Folic Acid to be given to all women of child bearing age. Dose: 400 mcg / day
- For high risk women, dose in 10 times - 4000 mcg/day (4 mg / day) Should be started at least 1 month before conception. (High risk: Previous Wo birth with NTD)



Myelomeningocele

Hydrocephalus

HYDROCEPHALUS

- MC cause of obstructive hydrocephalus in Children: Aqueductal stenosis
- Types of Hydrocephalus: communicating and non-communicating

Causes of Communicating Hydrocephalus

- **Mnemonic - "CAMP"**
  - Choroid Plexus papilloma
  - Achondroplasia
  - Meningeal Malignancy/ Metastasis
  - Post Hemorrhagic
- For babies with open Ant. Fontanelle with hydrocephalus - Bulging Anterior fontanelle.
- For closed fontanelle: Fundus examination can show Papilledema (indicator of raised ICT)

Rx of Raised ICT

- Acute Mannitol: Hypertonic Saline (3 % NaCl)
- Long term: Oral Acetazolamide / Glycerol

> **Important Information**
>
> - **Shunt Sx. of choice for Rx of hydrocephalus is children: VP shunt (Ventriculoperitoneal shunt)**

FEBRILE SEIZURES

04:43:30

- Def: Seizures + significant fever (104 °F) Without any evidence of CNS infection in age 6 m - 5 year
  - Mc cause of Seizure in children < 5 years

2 Types	
Simple	**Complex**
GTCS	Focal sz
Last < 15 min	Lasts ≥ 15 Min.
Single episode	Multiple episodes

- Treatment: Required only if seizure lasting > 5 min
  At Home: Buccal/Nasal Midazolam spray
  or
  Per-rectal Diazepam
- In hospital: For ongoing Sz treat like status epilepticus.

Risk factors of Recurrence of Febrile Sz	
Major	**Minor**
Age < 1years	Family h/o febrile sz
Duration of fever < 24 hrs.	Complex febrile sz
38 - 39 °C (lower temperature)	Male
	Low Na^+ on presentation

- Treatment
  - For simple Febrile Sz - No long-term anti-epileptic
  - Intermittent prophylaxis with Clobazam / Diazepam
  - Antipyretic like PCM: makes child comfortable but not prevent recurrence

JUVENILE MYOCLONIC EPILEPSY

- Adolescent with dropping objects from hand, more in morning and during stressful condition
- EEG showing epileptic spikes.
- DOC: Valproate

Previous Year's Questions

Q. About Juvenile Myoclonic epilepsy, all are true EXCEPT?

(AIIMS - Nov - 2019)

A. Valproate is contraindicated
B. Lamotrigine can be given
C. Phenytoin is not the preferred drug
D. Polygenic inheritance

LENNOX GASTAUT SYNDROME

- Multiple Seizure types
- Refractory difficult to control
- EEG - 1 - 2 Hz spike and slow waves

Previous Year's Questions

Q. Which of the following epileptic syndromes will not present during infancy?

(JIPMER - Dec - 2019)

A. Ohtahara syndrome
B. West syndrome
C. Lennox - Gastaut syndrome
D. Dravet Syndrome

ABSENCE SEIZURES

- 5 - 8 years
- Day dreaming / fall in school performance
- Usually lasts for a few seconds only
- Eye movements are seen
- EEG: 3 Hz spike & wave pattern
- DOC: Ethosuximide (not in India), India - Valproate

Previous Year's Questions

Q. Drug of choice for absence seizures?

(INICET - Nov - 2020)

A. Ethosuximide
B. Valproate
C. Carbamazepine
D. Phenytoin

Rx of Status Epilepticus

- Def: Seizure lasting > 5 min or Any child brought with ongoing seizure brought to medical facility
- Rx: ABC

IV Access > 10 Access > Buccal or nasal midazolam to abort seizure

↓

Lorazepam > Midazolam

↓

IV Phenytoin

↓

Repeat IV phenytoin (half does)

↓

IV valproate / Levetiracetam / phenobarbitone

CNS INFECTIONS

- Infants with bulging fontanelles with h/o fever, irritability, poor feeding ± seizures → Meningitis

Mc cause of meningitis

- Neonates
  - India: E. coli
  - Worldwide: Group B Streptococci > E. coli > Listeria
- Infants / children: streptococcus pneumoniae

Previous Year's Questions

Q. A video of sick intubated neonate having bilateral jerks of both right and upper limbs with some occasional twitching of neck as well. Likely seizures?

(AIIMS - Nov - 2018)

A. Focal clonic
B. Multifocal clonic
C. Multifocal tonic clonic
D. Focal tonic

Mc cause of meningoencephalitis: Enterovirus

Case → Neonate with bulging fontanelle, heart failure & cranial bruit heard on auscultation over Anterior Fontanelle

↓

'Vein of Galen Malformation'

↓

(Misnomer Vein involved in Median Prosencephalic vein (precursor of vein of Galen)

Important Information

- Mc cause of syncope in children – Neuro cardiogenic / vasovagal

Brain Death in Children

- Definition: Irreversible cessation of function of entire brain including brain stem
- Diagnosis (3 components)
  a. Demonstration of irreversible coma with a known cause
  b. Absence of brainstem reflexes
  c. Apnea
- Finding should remain consistent: Examination done at least 2 time 12 – 24 hrs. apart
- Incompatible with Diagnosis of Brain Deaths
  - Seizure
  - Decerebrate/Decorticate posturing
  - Motor responses to painful stimuli

PEDIATRIC ENDOCRINOLOGY

⏱ 04:54:25

- Case: 3-year-old, recurrent hypoglycemia, short stature, micropenis → Congenital Hypopituitarism

THYROID

Congenital Hypothyroidism

- Mc cause: Thyroid dysgenesis
- Mc cause in a child with goiter - Thyroid dyshormonogenesis

Previous Year's Questions

Q. Most sensitive test for thyroid dysfunction in newborn?

(JIPMER - Dec - 2015)

A. Total T3
B. Total T4
C. TSH
D. Free T3

Important Information

- Best time for screening Congenital Hypothyroidism → D2 - D4 of life → Heel prick sample

- Clinical features
  - Myxedematous facies
  - Hoarse cry
  - Hypotonia
  - Lethargy
  - Umbilical hernia
  - Prolonged Jaundice
  - Constipation

ADRENALS

- Adrenal cortex of fetus releases
  - In early pregnancy: Cortisol
  - 2nd trimester: DHEA

Previous Year's Questions

Q. A 3-week neonate with ambiguous genitalia presented with Na+ 127 meq/L, K+ 6 meq/L with BP 52/24 mm Hg and he was managed with IV fluids. What is the next step of management?

(AIIMS - May - 2015)

A. Spironolactone
B. Hydrocortisone administration
C. Broad spectrum antibiotics
D. Calcium gluconate

Congenital Adrenal Hypoplasia (CAH)

Mc enzyme deficiency → 21 hydroxylase
↓
Mineralocorticoid deficiency (Salt wasting, hyperkalemia)
&
Ambiguous genitalia in female

	Female Virilization	Male Undervirilization
Salt wasting present	21 hydroxylase deficiency	3 β hydroxy steroid dehydrogenase deficiency
Hypertension present	11 β hydroxylase deficiency	17 α hydroxylase deficiency

- Drugs used for Rx of 21 hydroxylase deficiency: Hydrocortisone + Fludrocortisone
- For antenatal Rx of CAH: Dexamethasone

Important Information

- Mc cause of Cushing's syndrome in children → Iatrogenic

PUBERTY

Definition	Girls	Boys
Precocious puberty (secondary sexual characters)	< 8 years	< 9 years
Delayed puberty	> 13 years (No breast budding)	> 14 years (No testicular enlargement)

- Mc cause of central precocious puberty
  - In Boys: Structural CNS abnormalities e.g., hypothalamic hamartoma
  - In Girls: Idiopathic

DIABETES MELLITUS

MC → Type 1 Diabetes

↓

DOC → Insulin

Mixed split regime | Basal bolus regime

- Screening for Nephropathy should begin:
- In Pre-pubertal age: 5 years after onset
- Pubertal age: 2 years after onset

Previous Year's Questions

Q. After the delivery of an infant of diabetic mother, glucose of the infant was 60 mg/dL Which other investigation does the sister expects that the physician would ask her to do?
(AIIMS - May - 2018)

A. Serum potassium
B. CBC
C. Serum calcium
D. Serum chloride

Obesity in Children

- Def: BMI > 95th Percentile

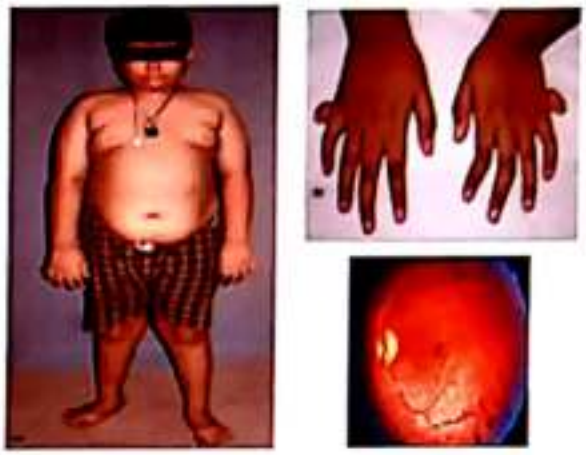
Leptin keeps 'thin'

↓

Secreted by adipose cells → decrease food intake & increases energy expenditure

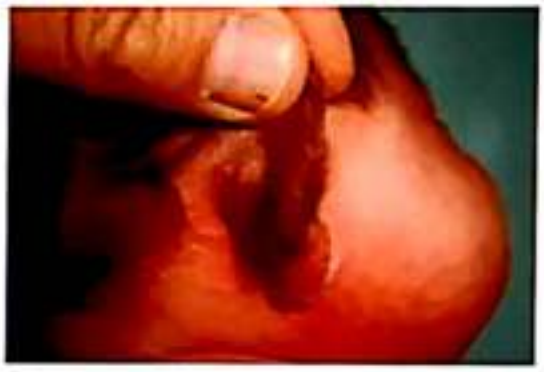
↓

Syndromes Associated with Obesity in children



Laurence Moon Bardet Biedel Syndrome

- Laurence moon Bardet Biedl syndrome (Polydactyly, Retinal pigment changes)
- Prader Willi syndrome
- Cushing's syndrome



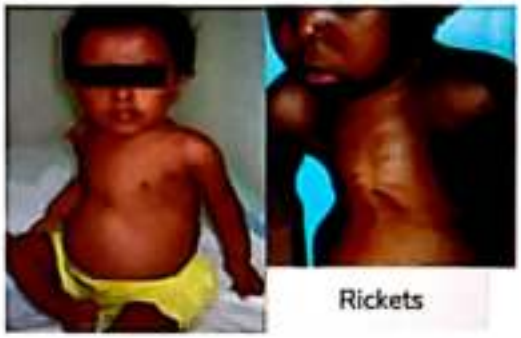
Infant of diabetic mother

DISORDERS OF BONE

RICKETS

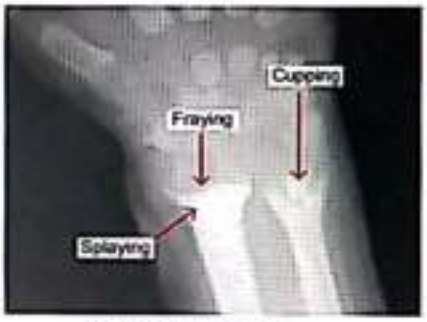
05:05:09

- Disease of growing bones d / t defective mineralization of bony matrix
- Not seen in SAM
- Important Causes of Rickets



Rickets

  - Vit D deficiency
  - Calcium deficiency
  - Phosphate deficiency
  - Renal losses (like hypophosphatemia, Renal Tubular Acidosis)
- Clinical features
  - Frontal & Parietal bossing
  - Rachitic Rosary
  - Wrist widening
  - Pot belly
- Investigations



X-ray finding in Rickets

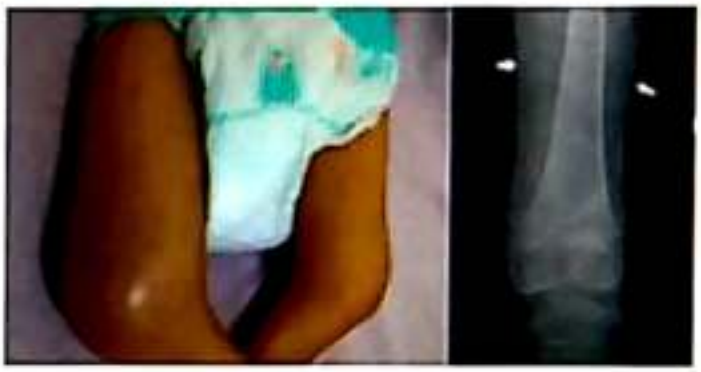
Important Information

- Elevated serum alkaline phosphatase levels are seen in rickets

  - X-Rays: fraying, cupping, splaying of extremities
  - Ca, PO Vit D levels vary with cause of Rickets
  - CKD: Phosphate elevated (other causes - Po4 Normal or low)
  - VDDR -1: Low levels of 1, 25 dihydroxy vit D3 (deficiency of 1 - alpha hydroxylase enzyme)
  - VDDR - 2: High levels of 1, 25 dihydroxy vit D3 (end organ resistance at level of receptor)
  - In Hypophosphatemic Rickets: Po_4 low
- Treatment
  - Vit D_3: 3 lac – 6 lac IU

SCURVY

05:08:10

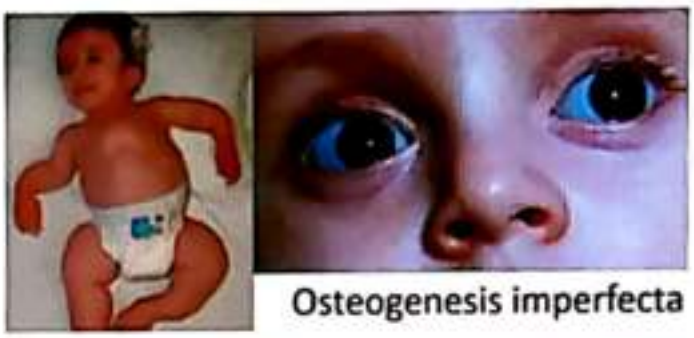


Subperiosteal hemorrhage in scurvy

- Vit C deficiency: Collagen synthesis impaired
- Predominantly cow milk fed babies: more at risk
- Clinical features
  - Bleeding
    - → Petechiae
    - → Gum bleeding
    - → Painful pseudo paralysis / cry on touch (Due to Subperiosteal hemorrhage involving long bones)
- Treatment: Vit C

OSTEOGENESIS IMPERFECTA (BRITTLE BONE DISEASE)

- Triad
  - Blue Sclera
  - Deafness
  - Recurrent fractures
- Treatment: I/V Bisphosphonates E.g. pamidronate



Osteogenesis imperfecta

ACHONDROPLASIA 05:10:34

- Autosomal Dominant inheritance
- Clinical features
  (Mnemonic - 'ACHONDROPLASIA')
  - Champagne Glass Pelvis
  - Hand ab[illegible]: Trident hand (oblique fingers)
  - Obesity
  - Neurological problem
  - Delayed motor milestones
  - Recognized at birth
  - Bowings leg
  - Proximal limb shortening
  - Large head
  - Short stature
  - Interpedicular distance b/w vertebrae is decreased