Top 60 Paeds Multisystem Concepts

EXTRACTED FROM BLOCK Q PREPROFFS 2024 **METABOLIC • GENETICS • RHEUMATOLOGY**

1. METABOLIC & STORAGE DISORDERS

Concept	Question Bank Fact
Glycogen Storage Disease Type 1	Diagnosis confirmed by **Enzyme assay of cultured skin fibroblasts** (or gene mutation analysis).
Pompe Disease (GSD II)	Infant with **Enlarged Heart** (Cardiomegaly), macroglossia, and hypotonia.
Von Gierke's Disease (GSD I)	Doll-like facies, protuberant abdomen, **Hypoglycemia**, Lactic Acidosis, Hypertriglyceridemia.
Galactosemia Enzyme	Primary cause is deficiency of **Galactose-1-phosphate uridyltransferase (GALT)**.
Galactosemia Presentation	Newborn with jaundice, vomiting, hepatomegaly, and **E. Coli Sepsis** risk.
Galactosemia Diet	Management requires **Lactose-free formula** (Soy based). No breast milk.
PKU Damage	Elevated Phenylalanine primarily damages the **Brain** (Intellectual disability).
PKU Screening	Positive newborn screen in asymptomatic infant -> Start **Low-phenylalanine diet** immediately.
PKU Diet	Avoid high protein. Use **Tyrosine supplementation** (becomes essential).
McArdle Disease (GSD V)	Muscle cramps and fatigue with exercise. Myoglobinuria.

2. GENETICS & SYNDROMES

Concept	Question Bank Fact
Down Syndrome (Trisomy 21)	Karyotype shows Trisomy 21. Assoc with **Duodenal Atresia** ("Double Bubble") and AV Canal defects.
Edward Syndrome (Trisomy 18)	Microcephaly, **Rocker bottom feet**, overlapping fingers, cardiac defects.
Patau Syndrome (Trisomy 13)	**Cleft lip/palate**, microcephaly, polydactyly, scalp defects.
Marfan Syndrome Heart	Most common cardiac abnormality is **Aortic Dilation** (Risk of dissection) or MVP.
Osteogenesis Imperfecta	Frequent fractures, **Blue Sclerae**, hearing loss. Family history positive.
Ehlers-Danlos Syndrome	**Hypermobile joints**, skin hyperextensibility, easy bruising.
Fragile X Syndrome	Most common inherited cause of intellectual disability. Macroorchidism.
Turner Syndrome	45 XO. Short stature, webbed neck, Coarctation of Aorta.
Klinefelter Syndrome	47 XXY. Tall stature, gynecomastia, infertility.
Delayed Tooth Eruption	Causes: Familial (Most common), Hypothyroidism, Down Syndrome. (Not Hyperthyroidism).

3. MUCOPOLYSACCHARIDOSIS (MPS)

Concept	Question Bank Fact
Hurler Syndrome (MPS I)	**Corneal Clouding**, coarse facies, hepatosplenomegaly. Deficiency of Alpha-L-Iduronidase.
Hunter Syndrome (MPS II)	Deficiency of **Iduronate Sulfatase**. No corneal clouding. X-Linked Recessive.
Morquio Syndrome (MPS IV)	Deficiency of **N-acetylgalactosamine-6-sulfate sulfatase**. Skeletal dysplasia prominent.
Sanfilippo (MPS III)	Severe **Neurodegeneration** and behavioral problems. Mild somatic features.
Maroteaux-Lamy (MPS VI)	Deficiency of **Arylsulfatase B**. Normal intelligence but severe skeletal issues.

4. PORPHYRIA & TOXICOLOGY

Concept	Question Bank Fact
Acute Intermittent Porphyria	**Abdominal Pain** + Vomiting + Confusion + Hypertension. **Dark Urine** on standing.
AIP Management	Acute attack treatment: **Intravenous Hematin** (Heme Arginate) and Glucose.
Porphyria Cutanea Tarda	**Blistering rash** on sun-exposed areas. Urine darkens in sunlight.
Erythropoietic Protoporphyria	Painful photosensitivity (screaming child) but **no blisters** initially.
Porphyria Trigger	Attacks precipitated by: Stress, Alcohol, Sulfonamides, Barbiturates.
Kawasaki Disease	Fever > 5 days + **CRASH** (Conjunctivitis, Rash, Adenopathy, Strawberry tongue, Hand/Foot changes).
Kawasaki vs Toxic Shock	Kawasaki lacks **Diffuse Myalgia** and hypotension compared to TSS.
Kawasaki Complication	Highest risk of **Coronary Artery Aneurysms** in the subacute phase.
Heat Stroke	Temp > 104°F + **Altered Mental Status**. Rx: Rapid cooling (Ice water immersion).
Hypoglycemia Definition	In older infants/children: Whole blood glucose **< 60 mg/dL** (Some sources say <45, check local cutoff).

5. NUTRITION & GI MANIFESTATIONS

Concept	Question Bank Fact
Kwashiorkor	Edema + **Flaky Paint Dermatosis** + Sparse hair. Protein deficiency.
Marasmus	Severe wasting + No edema + "Old man facies". Calorie deficiency.
Celiac Disease	Chronic diarrhea, wasted buttocks, **Iron deficiency anemia**. Diagnosis: Small bowel biopsy.
Toddler's Diarrhea	Chronic diarrhea with **undigested food** particles. Normal growth. Reassurance needed.
Cow Milk Allergy	Vomiting/Diarrhea/Eczema after milk. Rx: Hydrolyzed formula.
Vitamin A Deficiency	Night blindness, Bitot's spots, Corneal xerosis.
Vitamin D Deficiency	Rickets: Bow legs, Rachitic rosary, Wide wrists. Low Ca/PO4, High ALP.
Zinc Deficiency	Acrodermatitis Enteropathica (Rash around mouth/anus) + Diarrhea.
Refeeding Syndrome	Risk of **Hypophosphatemia** when feeding malnourished child too fast.
Hirschsprung Disease	Delayed meconium, abdominal distension. Dx: Rectal biopsy (No ganglion cells).

6. RHEUMATOLOGY & MISC

Concept	Question Bank Fact
SLE in Children	Malar rash, fever, joint pain. **ANA positive**.
Lupus Nephritis	Common complication of SLE in children. Hematuria/Proteinuria.
Henoch-Schonlein Purpura	**Palpable Purpura** on buttocks/legs + Abd pain + Joint pain. IgA vasculitis.
Juvenile Dermatomyositis	Heliotrope rash (eyelids) + Gottron papules (knuckles) + Proximal weakness.
Scarlet Fever	"Sandpaper" rash sparing mouth (circumoral pallor) + Strawberry tongue.
Mitochondrial Inheritance	Affected mother transmits to **ALL** offspring. Affected father transmits to **NONE**.
X-Linked Recessive	Affected mother -> 50% sons affected. Affected father -> All daughters carriers, no sons affected.
UTI Screening	**Positive Nitrites** and Leukocyte Esterase on dipstick are specific.
UTI Confirmation	**Urine Culture** is the gold standard (Single organism growth).
Nephrotic Syndrome	Edema + Massive Proteinuria + Hypoalbuminemia. Assoc with Hyperlipidemia.

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