Complement deficiency

- all are Autosomal recessive EXCEPT c1 inhibitor deficiency (AD) & Properdin deficiency (X-linked)
- Screening: CH50 test
- Treatment: nothing except for purified C1
  - C1r, C1q, C2 and C4 deficiency
    - Autoimmunity, SLE-like
  - C5-C9 deficiency
    - Recurrent N.meningitis sepsis
  - Factor D & properdin deficiency
    - Recurrent N.meningitis sepsis
  - C3 deficiency
    - Membrane-Proliferative glomerulonephritis & vasculitis
  - Factor H deficiency
    - Familial relapsing thrombocytopenic purpura
eic syndrome
  - Mannose binding lectin deficiency
    - Recurrent infections

Wiskott-Aldrich Syndrome

- **Triad:** Eczema, Thrombocytopenia and Immunodeficiency
- X-linked recessive - Defective gene encoding WASP
- **Diagnosis:** bleeding in young age + low IgM High IgA & IgE
- **Management:** IVIG + Splenectomy + platelet transfusion

Ataxia Telangiectasia

- **Autosomal Recessive**
• Recurrent sinopulmonary infection - Telangiectasia - progressive ataxia

• **Combined immunodeficiency of** IgA and IgG2

• **management:** supportive, IVIG, transplant

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**Chronic Granulomatous Disease (CGD)**

• Recurrent deep seated abscess + pyogenic infection

• dysfunction of NADPH

• MC infection is lymphadenitis

• Granuloma formation is common

• Nitrate Blue Tetrazolium Test (NBT)

• **Management:** prophylactic and antibiotic treatment of infections - recombinant human interferon G - bone marrow transplant.

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**Leukocytes Adhesion Defect (LAD)**

• kills but can’t create inflammation

• The hallmark of the disease is neutrophilia without PMNs in the infected tissue or pus

• History of: Poor wound healing - Delayed umbilical cord separation - Sepsis

• **Diagnosis:** decrease or absence of CD18

• **Management:** Continuous antimicrobial treatment - good oral hygiene - WBC transfusion - bone marrow transplant

• Omphalitis in LAD I

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**Bare Lymphocyte Syndrome**

• MHC class 2 deficiency

• recurrent infections + FTT + Chronic diarrhea

• Bone marrow transplantation
• Poly-Articular (5 or more joints) (smaller joints) (Symmetrical) (+ RF = more severe)
• Systemic (Stills Disease) = (- RF) (negativ anti nuclear antibodies) (CVD, Liver, spleen, lymph, GI problems) (remain into adulthood)

- **Diagnosis:** mostly by finding features.
- **Management:** Dec pain and swelling / inc ROM / prevent joint damage
  - Excercise can help
  - NSAIDs
  - DMARD
  - Biological modifiers
  - Corticosteroids (LAST RESORT)

14- Child Maltreatment

intro

- **Red Flags:**
  1- history not consistent with story
  2- long time to seek treatment
  3- multiple injuries
  4- injuries at different ages
- **Risk Factors:**
  - **Parents:** substance abuse. anger problems
  - **Families:** Crowded house, early separation
  - **Child:** behavior problems, difficult
  - **Parent child relationship:** lack of emotional connection
  - **Environment:** poverty - low education
- **When taking history** be as detailed as possible so they don't fabricate the story
- Always check for: Bruises / fractures / skeletal survey / ophthalmic exam / head imaging / coagulation profile
- Bruises in non-ambulating children are unusual
- Skeletal survey: multiple healing lateral rib fractures
- Excessive concern w/ body weight and shape
- fear of weight gain
- obsession with food
- Excessive weight loss or failure to gain weight
- Amnorrehea

- **Signs and symptoms**: starvation and malnutrition, and can affect nearly every organ system, thin build, tachycardia, hypotension, dry skin, brittle hair,

• **Bulimia Nervosa**
  - Repeated episodes of binge eating
  - often purging
  - **Signs and Symptoms**: binge eating and purging, dental erosion, abrasions on dorsum of hand

The cause of eating disorders is multifactorial (3). Genetic predisposition, neurochemical factors, psychological factors and sociocultural influences

21- **Conjunctivitis**

  intro

• **Ophthalmia neonatorum**: MC ocular disease in newborns

• Causes of neonatal conjunctivitis: (are chemical, chlamydial and bacterial)

• Viral = herpes simplex virus (occurs infrequently) mode of infectious transmission is during passage through a colonized or infected birth canal

• infection with Neisseria gonorrhoeae is felt to be one of the most serious because of its potential to damage vision and cause blindness

• **Chlamydia trachomatis** is now the most common infectious agent causing neonatal conjunctivitis

• **silver nitrate** for prophylaxis (not for chlamydia)
• **Nevi:**
  - **Macular Hemangioma** = true vascular nevus - on eyes
  - **Port-wine stain** = at birth - doesn't blanch with pressure
  - **Mongolian Spot** = Blue/purple bruise like spots on back - disappear after 4 years
    - **MOST COMMON BIRTHMARK**
  - **Strawberry Hemangioma** = Flat red sharply demarcated lesions on face - disappear after 7 years

• **Head:**
  - check for cuts or bruises & fontanels
  - Ant. Closes at **9-12 months** / Post. closes at **2-4 months**
  - **Molding:** temporary asymmetry of the skull
  - **Caput Succedaneum** = Edematous swelling - **EXTENDS BEYOND SUTURE LINES**
  - **Cephalhematoma** = Subperiosteal Hemorrhage - **DOES NOT EXTEND**
  - **Subgaleal Hematoma** = bleeding below epicranial aponeurosis - can extend - DANGEROUS
  - **Craniosynstosis** = premature closure

• **Eyes:**
  - **Subconjunctival Hemorrhage**
  - **Congenital Cataract**

• **Ear:**
  - **Ear Tag**

• **Mouth:**
  - **Cleft Lip**
  - **Micro gnathia**
- Causes = Parainfluenza - influenza
- Treatment = Supportive + Steroids (Dexamethasone) - Epinephrine

**Epiglottis**
- can be life threatening
- Sick looking - fever - drooling - not immunized (opposite to croup)
- mc = h.influenza or S. Pneumoniae
- Diagnosis = Clinical + see epiglottis
  - (X-Ray = Thumb sign)

**Anaphylactic**
- More than 2 systems
- IM Epinephrine

**Hypoglycemia**

**Clavicle Fracture** = just give pain killer
**Pulled elbow** = pop it back in place

questions:
- Shock
- Seizure
- Croup
- Epiglottis
- Asthma
- Anaphylaxis
• **Big baby** Hyperinsulinemia (Infant of diabetic mother)

• **Short stature, cleft palate, or micro penis if boy** Growth hormone deficiency.

• **Macrosomia, Macroglossia, hepatosplenomegaly, ear creases & hernia** Beckwith-Wiedemann

• **Hyperpigmentation** May be a clue to adrenal insufficiency (Addison’s disease).

• **Hepatomegaly** One of the metabolic diseases can be involved.

- **Treatment:**
  - not able to feed = 10% Dextrose
  - if not working = IV Glucose

• Renal or Pre-Renal (General Look)

• How to differentiate acute VS chronic

• Not much into management

Vaccination / Contraindication of Vaccinations

Viral Exanthemas very important.

Paracetamol / Iron poisoning

- **37- Common Viral Exanthems**

  intro

  • skin eruption after acute infection

- **Measles (Rubeola)**

  • caused by **Paramyxovirus** (spread by resp. droplet)

  • Exanthema: on skin

  • Enanthem: on mucous membrane
40- Respiratory Distress Syndrome

intro

- Grunting + Nasal Flare + Pale skin + Pink + Tachypnea + Preterm + Decreased urine output
- Chest retractions
- Mild Hypotonia
- Most common cause is **Transient Tachypnea of Newborn**
- **Chest X-Ray** = Diffuse reticulogranular pattern (ground-glass appearance)
- **Treatment** = Exogenous Surfactant

41- Foreign body ingestion/aspiration

intro

- Ingestion VS Aspiration
  - For ingestion = Endoscopy is diagnostic and therapeutic
  - For Aspiration = Bronchoscopy is diagnostic and therapeutic

42- Obstructive Sleep Apnea

intro

- Episodes of complete or partial upper airway obstruction during sleep
- gas exchange abnormalities and disturbed sleep
- **Risk factors:**
  - Adenotonsillar hypertrophy
  - Obesity
- **Diagnosis:**
  - overnight polysomnographic evaluation in the sleep laboratory