

Cerebral palsy

History

Complaint

- Delayed developmental motor and mental milestones
- Convulsions
- Chest infection

Present history

- Complaint analysis
 - Onset, course (progressive), duration
- Antenatal history maternal fever, rash, drugs or exposure to radiation
- Natal history: obstructed or prolonged delivery (hypoxia)
- Neonatal history :
 - Onset of crying after birth (delayed with hypoxia)
 - Cyanosis
 - Jaundice
 - CNS infections (fever-convulsions-admission to fever hospital)
- Developmental history: significant delay in motor and mental aspects
- Neurological symptoms e.g. convulsions - blindness- paraparesis
- Aspiration or dysphagia (bulbar manifestations)

Past history

a) Perinatal history:

Prenatal.....
.....

- Natal-Postnatal

b) Developmental history

Motor development:

Mental development:

c) Nutritional history

d) Vaccination history

e) Previous infections, significant illness

Family history

Parents: consanguinity

Similar condition in the family.....

Examination

General

Measure.....

Head circumference: microcephaly with early closure of fontanelle

2- Systems

Neurological

- Motor system

- Inspection: disuse atrophy
- Power: paralysis or paresis (hemiplegia, diplegia or quadriplegia)
- Tone: spasticity (may be with scissoring)
- Abnormal movement e.g. chorea or athetosis or both in cases of extrapyramidal cerebral palsy e.g. post kernicteric
- Reflexes :

Deep reflexes: hyperreflexia - may be with clonus (ankle clonus)

Superficial reflexes:

Positive planter reflex (dorsiflexion)

Lost abdominal reflexes

Abnormal persistence of neonatal reflexes as: Moro and grasp

- Sensation:

- Not affected - cerebral palsy is pure motor disease

- Cranial nerves:

- Only motor nerves are affected and it is upper motor neuron lesion

Pseudo bulbar palsy (9, 10) is the commonest

Weakness of muscles of palate and pharynx

Exaggerated palatal and pharyngeal reflexes (Gag reflex)

Nasal regurgitate

Other cranial nerve affection

- Ocular nerves (3,4,6) > squint
- 5th nerve affection) exaggerated jaw reflex
- 7th nerve affection → facial muscle weakness

What is your diagnosis?

A case of cerebral palsy post hypoxic, quadriplegic, infection

- What are the associations of C.P.?

What are the investigations that may be done in cerebral palsy?

Laboratory

- Karyotyping
- TORCH screening
- Metabolic screening

Imaging

- CT or MRI
- EEG

Others

- Auditory and hearing assessment

What is the differential diagnosis of cerebral palsy?

Spinal cord lesion

Degenerative brain disease

Atonic cerebral palsy from

Causes of floppy infant e.g. Werdnig Hoffman disease

- What is the difference between bulbar and pseudobulbar palsy?

Bulbar palsy is lower motor neuron lesion (damage in the nuclei)
Pseudobulbar palsy is upper motor neuron lesion (damage is in the pyramidal tract supplying the nuclei)

What are the sites of CNS damage in cerebral palsy?

What are the causes of inability to walk?

What are the causes of MENTAL RETARDATION?

What are the treatment lines in C.P.?

Ventricular septal defect (VSD)

History

Complaint

- Recurrent chest infections

- S.O.B.

Present history

- Complaint analysis
Onset, course (progressive), duration
- Manifestation of pulmonary congestion
- Manifestations of low cardiac output
- Failure to grow
- Recurrent chest infection: cough, expectoration

Other symptoms of same system.....

Other symptoms of other systems.....

- Associated symptoms-investigations -treatment

Past history

a) Perinatal history:

Prenatal... maternal drugs or infection

.....
- *Natal-Postnatal*

b) Developmental history

Motor development (usually delayed motor development)

Mental development

c) Nutritional history

d) Vaccination history

e) Previous infections, significant illness

Family history

Parents: consanguinity

Similar condition in the family.....

Examination

1- General.....

Usually under weight

2- Systems

Heart

Inspection and palpation

- Biventricular enlargement (normal in small defect)

- Apex is shifted downward and outward
- Left parasternal pulsation → 3rd and 4th space
- Systolic thrill over the parasternal area
- Palpable 2nd heart sound due to pulmonary hypertension and
- Pulsation on the 2nd left space, if pulmonary dilatation

Auscultation :

- Murmurs (VSD)

Site: 3rd and 4th intercostal space at parasternal line

Area of maximum propagation: all over the precordium

Character: harsh

Timing: pansystolic

- Sounds (pulmonary hypertension)

- Accentuation of the pulmonary component of the second heart sound

Chest

If bronchitis is present, auscultation may reveal:

Medium sized crepitations and wheezes scattered on both lung fields

What is your diagnosis?

A case of congenital cyanotic heart disease in the form of VSD with:

- Biventricular enlargement and pulmonary hypertension
- Compensated heart
- With or without chest infection as a complication

What are the causes of congenital acyanotic heart disease?

What are the investigations of VSD

- What are the complications of VSD?

- complications of surgery
- delayed motor development
- Recurrent chest infections
- Congestive heart failure
- Infective endocarditis
- Shunt reversal: Eisenminger syndrome

- What is the treatment of VSD?

- About 60 % of VSD close spontaneously within 2- 4 years (mild cases and some moderate cases)

- Large defect: surgical closure

Fallot tetralogy

History

Complaint

BLUISH DISCOLORATION of the skin & M.M.

Present history

- Complaint analysis
Onset of cyanosis (not at birth), course (progressive), duration
Exertional dyspnea
Squatting
Hypercyanotic spells
- Failure to grow

Other symptoms of same system.....

Other symptoms of other systems.....

.....

- **Associated symptoms-investigations -treatment**

Past history

a) Perinatal history:

Prenatal... maternal drugs or infection

.....
- *Natal-Postnatal*

b) Developmental history

Motor development (usually delayed motor development)

Mental development

c) Nutritional history

d) Vaccination history

e) Previous infections, significant illness

Family history

Parents: consanguinity

Similar condition in the family.....

Onset

Past history

Antenatal history (maternal drugs or infection)

Developmental history (may be delayed motor aspect)

Examination

1- General

- Central Cyanosis
- Clubbing of the fingers and toes
- Under weight

2- Heart examination

- Inspection and palpation
Normal or parasternal pulsation (mild right ventricular hypertrophy)
- Auscultation
Ejection systolic murmurs heard at the pulmonary area

What is your diagnosis?

A case of congenital cyanotic heart disease in the form of Fallot tetralogy.
No cardiomegaly or heart failure

What to do for him?

What are the complications of Fallot tetralogy? 4

- Cerebral thrombosis
- Brain abscess
- Bacterial endocarditis
- Heart failure is rare

What is the treatment of Fallot tetralogy?

Medical

- Treatment of cyanotic spells
 - Placement of the infant in the knee chest position
 - Administration
 - Morphine
 - Sodium bicarbonate to correct metabolic acidosis
 - Propranolol
- Iron therapy-for prevention of iron deficiency anemia

Surgical

- Blalock Taussing operation :Anastomotic procedures (subclavian and left pulmonary artery)
- Total correction at elective age of 1 years

What is the value of in Squatting Fallot tetralogy?

What is Fallot triology? 4

What are the other causes of congenital cyanotic heart disease?

1- With increased pulmonary blood flow

- TGA : Transposition of great arteries (cyanosis at birth)
- Truncus arteriosus
- Hypoplastic left heart
- Single ventricle
- Total anomalous pulmonary venous return.

2- With decreased pulmonary blood flow

- Pulmonary atresia
- Tricuspid atresia

Nephrotic syndrome

History

Personal history

Age: minimal lesion type: 2-6 years

Complaint

Swelling of lower limbs or puffiness of the eye lids

Present history

- Analysis of the complaint
 - Edema: - characters specific for renal edema (**March**)
- Urinary symptoms e.g. hematuria (in non minimal lesion)
- Symptoms of hypertension e.g. headache (in non minimal lesion)
- Symptoms of complications e.g. chest infection
- Exclude other causes of edema e.g.
 - Cardiac: absence of cardiac symptoms e.g. dyspnea, orthopnea .
 - Hepatic: absence of hepatic symptoms e.g. jaundice

Nutritional: by age and good nutritional history

Other symptoms of same system.....

Other symptoms of other systems.....

- Associated symptoms-investigations -treatment

Past history

a) Perinatal history:

Prenatal- Natal-Postnatal

b) Developmental history

Motor development -Mental development

c) Nutritional history

d) Vaccination history

e) Previous infections, significant illness

Family history

Parents: consanguinity

Similar condition in the family

Examination

1- General

- Vital signs

RR (MAY BE INCREASED).....

BP (hypertension in non minimal lesion)

- Head

Eye: Puffy eyelids

Mouth: pallor (lips)

Cushioned features in long-standing treatment with steroids

- Limbs

Edema: bilateral pitting not tender edema reaching below knee or above ankle.

- Genitalia

- Scrotal edema

2- Systems

-Abdominal: Complete abdominal examinations

- Inspection

• Shape and contour

• Generalized distention → marked ascites

Or bulging flanks → moderate ascites

- Skin : stretched-may be striae from steroids or distension
- Umbilicus : shifted downward and everted
- Subcutaneous tissues: visible veins
- Muscles : diverication of recti
- Bone : subcostal angle (wide in long standing distension)

- Palpation

- Superficial : tenderness and rigidity (if present suspect peritonitis)
- Deep: no organomegaly - renal angles are free in bimanual examination

- Percussion

- There is moderate amount of ascites detected by shifting dullness or there is
- Huge amount of ascites detected by transmitted thrill

What is your diagnosis?

What is dif. Diagnosis of edema?

What to do for him?

Investigations

- Urine
 - Protein in 24 hours urine above 2 gram/ m2/ 24 hours
 - Selective proteinuria
- Serum protein
- Serum cholesterol
- Renal function
- C3
- Renal biopsy in selected cases

What are the indications of renal biopsy in nephritic syndrome ?

- Clinically

- Age: less than 1 year or more than 8 years
- Gross hematuria or hypertension

- Laboratory

- Non selective proteinuria
- Impaired renal functions
- Decreased serum complement

- Therapy

- Before start of Immunosuppressive drugs (steroid resistant- steroid dependent)

What are the complication of nephritic syndrome?

What are the types of nephrotic syndrome?

What are the diagnostic criteria of minimal lesion nephrotic syndrome?

- Age incidence: 2-6 years
- No hypertension or gross hematuria
- Selective proteinuria
- Normal renal function tests
- Normal C3
- Biopsy findings
- Light microscope:- no abnormalities
- Electron microscope:- fusion of foot process of podocytes
- Excellent response to steroids
- Relapse without impairing renal functions

What are the causes of nephrotic syndrome in the first year of life?

- Primary causes
 - Congenital nephrotic syndrome
 - Nephrotic syndrome with focal glomerulosclerosis
 - Membranous and membranoproliferative nephritis
- Secondary causes
 - Infections: TORCH infections
 - Neoplasm: Wilms
 - Drugs: heavy metals

What are the differences between minimal lesion nephrotic syndrome and nephritis?

- Age
- Hypertension and hematuria are constant findings
- Proteinuria if present, it is mild
- Renal functions are impaired
- C3 is reduced in post-streptococcal nephritis
- Biopsy findings are different
- Treatment is directed towards hypertension and its complications

- Complications are due to hypertension and impaired renal functions

What is meant by steroid resistant and steroid dependant?

What to do for steroid resistant and steroid dependant cases?

What are the side effects of prolonged steroid use?

- Hypertension
- Cushinoid features
- Growth retardation
- Mood alteration
- Impaired immunity

What are the side effects of cyclophosphamide?

- Hemorrhagic cystitis
- Alopecia

What is the prognosis?

Congenital hypothyroidism

History

Complaint

Delayed motor and mental development

Present history

- Analysis of the complaint
- Delayed motor development (delayed sitting, sanding and waking) and delayed mental (delayed mother recognition and speech) in details.
- Constipation
- Decreased activity
- Associated symptoms-investigations -treatment

Past history

a)Perinatal

- Antenatal history: maternal intake of antithyroid drugs during pregnancy

- Neonatal history: -Jaundice, constipation, feeding difficulties and much sleep
- Developmental history (delayed motor and mental development)

b) Developmental history

-Motor development.....

-Mental development.....

c) Nutritional history

d) Vaccination history

e) Previous infections, significant illness

Family history

Parents: consanguinity

Similar condition in the familial.

Examination

- Measurements

Short stature (**inappropriate upper segment/lower segment ratio**)

- Head → Widely opened anterior and posterior, fontanel.

- Hair → Coarse - hair lines reaches far down on forehead.

- Forehead → Short and wrinkled.

- Eyes → Swollen eye lids

- Depressed broad nasal bridge

- Mouth

- Kept opened with large thick broad tongue protruded from it.
- Delayed dentition.
- Lips: pallor.

- Neck → Short and thick.

- Trunk

- Distended Abdomen and umbilical hernia.
- Skin: Yellow discoloration (hypercarotenemia) - dry and scaly.

- Limbs

- Short limbs.
- Hands: Broad and short

What is your diagnosis?

A case of congenital hypothyroidism

What are the investigations required ?

- Laboratory

Thyroid profile: T3, T4, and T.S.H.

- Decreased T4 Increased T.S.H. in primary hypothyroidism (the most sensitive test)
 - Decreased T.S.H. in secondary hypothyroidism (pituitary or hypothalamic)
- Imaging
- Thyroid scanning (I^{125}) → For diagnosis of developmental anomalies of the thyroid
 - Plain x-ray
 - Retarded bone age
 - Diminished number of ossific centers
 - Epiphyseal dysgenesis: -fragmentation of the Epiphyseal ends

What is the treatment of congenital hypothyroidism?

Life long therapy with thyroxin

- Preparation: Sodium-L thyroxin tab.
- Dose: - Neonates 10 microgram /kg/day.

- Children 100 microgram/nr/day.

Response: signs of therapeutic response are:

- Increase activity and alertness.
- Normal TSH
- Reversal of all symptoms and signs

Assessment of height, growth velocity and I.Q every 3 months

Bone age every 6 months.

Chronic hemolytic anemia

History

Complaint

- Progressive pallor (very early in life - above 6 months)
- Yellow discoloration of skin and mucus membranes
- Progressive abdominal distention

Present history

- Complaint analysis

Onset of **pallor** (not at birth), Last part of the first year → course (progressive), duration.....

- History of repeated blood transfusion
- History of crises

Sudden pallor and red urine (hemolytic crisis)

Pain on the hand and feet (vasoocclusive crisis)

Other symptoms of same system.....

Other symptoms of other systems.....

- Associated symptoms-investigations -treatment(History of splenectomy)

Past history

a) Perinatal history:

Prenatal... maternal drugs or infection

.....
- *Natal-Postnatal*

b) Developmental history

Motor development (usually delayed motor development)

Mental development

c) Nutritional history

d) Vaccination history

e) Previous infections, significant illness

Family history

Parents: consanguinity

Similar condition in the family.....Family history

- Consanguinity

- History of similar conditions in the family

Examination

1- General

- Measurements
 - Stunted growth
 - Large head
- Vital Signs
 - Manifestations of hyperdynamic circulation
- Head
 - Pallor
 - Jaundice: mild
 - Mongoloid features
- Depressed nasal bridges
- Prominent maxilla
- Protruding upper central incisors
- Neck
 - Lymphadenopathy
- Trunk
 - Skin color is greenish brown (pallor + jaundice + hemosidrosis)
- Limbs
 - Short and broad hand

2- Systems

- Abdominal

Inspection

- Shape and contour → Generalized distention if huge spleen and liver
- Bone: sub costal angle is wide
- Free respiratory movement
- No visible peristalsis.
- Diverticulation of recti.
- Umbilicus: shifted downward and everted
- Normal genitalia.
- Back → no mass or spina bifida.
- No striae or pigmentation.
- Skin: stretched-may be striae from distension

- No hernia.....
- Subcutaneous tissues: visible veins
- **Palpation**
 - Superficial: for superficial tenderness and rigidity
 - Deep palpation:
 - hepatosplenomegaly
 - free renal angle.

Hepatomegaly

Lower border of the liver is felt

In midline cm below costal margin

In mid-clavicular..... cm below costal margin

, firm in consistency with rounded border, smooth surface.

It is not tender, not pulsating

Splenomegaly

Lower border of the spleen is felt.... cm below left costal margin. It is not tender, firm in consistency with rounded border, a notch may be felt

- Percussion

- Upper border of the liver is on the 5th intercostal space
- No ascites

- Heart

- Hemic murmur: short systolic murmur at the heart base (pulmonary area and aortic area)

What is your diagnosis?

A case of chronic hemolytic anemia most probably **Thalassemia major**

- Why Thalassemia ?

- It is the commonest type of chronic hemolytic anemia in Egypt.
- It is the most severe type: The case present with: - very huge spleen-very evident mongoloid features

However, confirmation needs laboratory investigations

- **Why the presentation of beta thalassemia does not start immediately at birth?**

What are the investigations to be done to this case ?

1- Investigation to prove anemia is hemolytic

Laboratory

- Blood picture

- Hemoglobin: low
- RBCs count: low
- Reticulocyte: reticulocytosis
- Normoblast

- Blood chemistry

- Increased serum iron
- Decreased iron binding capacity
- Increased unconjugated bilirubin

Imaging: X-ray

- Skull

- Widening of diploid spaces
- Hair on end appearance

- Long bones

- Generalized rarefaction
- Thinning of cortex
- Widening of medulla with reticular appearance

1- Investigations to diagnose the etiology of hemolysis

- Hb electrophoresis: -increased level of hemoglobin F-hemoglobin A is very low or may be absent
- Other investigations may needed to exclude other causes of chronic hemolysis e.g. osmotic fragility test- coombs test

What are the causes of chronic hemolysis?

What is the pathogenesis of hemolysis on beta thalassemia?

What are the causes of death in thalassemia?

What is the clinical picture of (hemosidrosis?)

What is the aim of blood transfusion?

What are the types of crises that may occur in thalassemia?

- Sequestration crises
- Hyper hemolytic crises

- Aplastic crises

What are the treatment options of thalassemia?

What are the hazards of blood transfusion

Purpura

History

Age 2-6 years

- Complaint

Reddish spots with or without mucous membrane bleeding

- Present history

- Complaint analysis

- History of fever 2 weeks before the onset of Purpura.
- onset → acutely
- Purpura as regard: - site (all over the body together with mucous membrane bleeding)
- size.....
- effect of pressure.....
- not elevated.
- Bleeding → Site: bleeding gum, epistaxis, rectal, subcutaneous.

Amount & Need for blood transfusion:

Exclude

- Preceding fever in immune thrombocytopenic purpura (ITP)
- no history of associated prolonged fever
- Drugs: cytotoxic drugs with aplastic
- No history of blood transfusion
- Arthralgia, marked weight loss, continuous fever with malignancy
- Course and duration (regressive and short with ITP) prolonged with aplastic anemia, progressive with malignancy (important)
 - History of blood transfusion (important) In aplastic anemia blood transfusion is so frequent
 - In ITP if blood transfusion was given it usually once
- Medical consultation- investigation- treatment

Past history

a) Perinatal history:

Prenatal- Natal-Postnatal

b) Developmental history

Motor development -Mental development

c) Nutritional history

d) Vaccination history

e) Previous infections, significant illness

Family history

Similar condition in the family

History of similar conditions in the family

Examination

General:

Observation: - *Level of consciousness (LOC) and activity:* Conscious, lethargic or comatose

- *Appearance:* Pallor, cyanosis, jaundice

2. Vital signs:

a) Temperature: 37.0 - 37.5°C.

b) Heart rate:

c) Respiratory rate

d) Blood pressure

All measures are within normal- child general condition is good.

- Head

Eye: subconjunctival hemorrhage

Lips: +/- pallor-bleeding gums

- Neck

Lymph nodes to exclude malignancy

- Limbs and trunk

- Petechial hemorrhage less commonly ecchymosis
- Not raised and do not blanch on pressure
- Red when fresh, then → violet → blue → green → fade.

- Tender bone with malignancy

2- Systems

- Abdominal: the presence of splenomegaly

What is your diagnosis?

A case of purpura most probably immune thrombocytopenic purpura or most probably Aplastic anemia

What are the investigations to be done ?

What are the causes thrombocytopenic purpura?

1- Increased destruction (normal megakaryocyte)

Immune

- Immune thrombocytopenic purpura
- Other autoimmune disorders e.g. systemic lupus
- Congenital (neonatal isoimmune or maternal immune)

Non immune

- Disseminated intravascular coagulopathy
- Thrombotic thrombocytopenic purpura
- Infections
- Drug induced
- Hypersplenism

2- Decreased production (low megakaryocytes)

Bone marrow depression :

- Aplastic anemia (idiopathic-drugs-toxins- irradiation)
- Megakaryocytic aplasia (idiopathic-drugs)
- Congenital: Fanconi anemia, thrombocytopenia with absent radius

Bone marrow replacement

- Leukemia - neuroblastoma
- Congenital leukemia

Deficiency

- B 12-folic acid deficiency
- Congenital: deficiency of thrombopoietin

What are the causes of non thrombocytopenic purpura?

- Vascular defect
 - Hereditary
 - Acquired:
 - Henoch Schonlein purpura
 - Infections e.g. meningococemia
 - Scurvy