



Common Neonatal Problems

objectives:

- Recognize the uniqueness of neonatal pathophysiology affecting illness presentation
- Mention some of the most common neonatal problems encountered in well-baby nursery and their management.

Done by: Hamad Almousa

Revised by: Saud Alrsheed

Team Leader: Saud Alrsheed

Special thanks to team 438 & Faisal alsaif



Lecture outline:

- History and examination.
- Fetal maturation.
- Neonatal physiology
- Normal variations in newborn.
- Minor trauma in newborn.

Note: This lecture ends at slide 11 What comes after is "Jaundice", and jaundice was not give in a separate lecture like the previous years, but it's in the objectives.

Also, what comes after jaundice are extra stuff from 437 slides but related to the main lecture.

معلیش اذا کثرنا علیکم

History and examination. Neonatal period: the first 4 weeks of life

Maternal History:

- Age
- Medical / surgical history E.g. Seizure, hypothyroidism, HTN, and DM
- Medications E.g. Anticonvulsant, and warfarin
- Pregnancy history E.g. Natural conception or IVF, and Hx of trauma
- Labour and delivery history E.g. premature rupture of membrane
- Family history Syndromes and genetic

• Neonatal History

• Physical examination:

- Head large
- Face rounder

Note:

DM can cause:

- First Trimester Hyperglycemia: congenital malformations
- Last Trimester Hypoglycemia: macrosomia
- At Birth Hypoglycemia In Baby, as the beta cells are hyeractive
- Small mandible And grow wider and bigger with time
- Prominent abdomen (But it's normal, as the anterior abdominal muscles are weak
- Mid point umbilicus (Upper segment = lower segment, In neonates is the umbilicus (adult = symphysis pubis) As we grow the midpoint gets lower)
- Liver and spleen easily felt, kidneys palpable
- Flexion posture.

• Other specific neonatal examinations:

- Neonatal reflexes.
 Born with it and disappear at 12 weeks
- Gestational assessment
 Two parts: Neurological and physical
- Congenital malformations



Neonatal physiology

Fetal Maturation

- Second Trimester If the mother had any problem e.g. infection the baby is affected
 - CVS attains Final Form 12 wk
 - Respiratory movement as early as **18 wk.**
 - Sufficient alveolar structures **24 wk.**
 - Surfactant production by 20 wk, (34 wk ; Enough surfactants to survive and breath) Surfactant is a phospholipid that prevent the collapse of lungs; alveoli
 - Tidal flow of amniotic fluid, out of lungs
 - Hemoglobin is fetal (HB A 30% at birth) Thus thalassemia won't bother us at the first few days of life. Remember Hb F is not good as it has high affinity for oxygen, Hb A will become predominant at the age of 3 month
 - Coordinated suck 34 wk (Suck, swallow, and breath at the same time)
 - Normal sucking (26 28 wk). The preterm baby are not fed due to the risk of aspiration and apnea

Respiratory system:

MCQ:

at what age the baby have enough maturity to survive on its own? Minimum is 34 wks

- Established at delivery
- PR 50 60/min.
- Wet lung syndrome. aka transient tachypnea of the newborn, which causes the baby to have RDS usually manifest by sudden shortness of breath, low blood oxygen levels, and fluid in the lungs

CVS

- HR 120-160/min
- Transient murmur.
- Fetal neonatal circulation
- R ventricle predominant.
- On ecg there is right axis deviation

Note:

And continue for 3-6 months of life, because the right ventricle is the strong and big one due to the fetal circulation where it pumps against high pressure. After delivery the left ventricle start pumping against the aorta and becomes predominant and turn to left axis deviation.

The combined ventricular output is 400-500 ml/kg/min. 65% originates from the right ventricle

Fetal pulmonary flow is 40 ml/kg/min is less than 10 % of the combined ventricular output

During fetal life the ductus arteriosus provides a conduit for meeting systemic circulatory requirements



Neonatal physiology

Gastrointestinal system

- Activity usually addressed toward meeting nutritional needs (crying hungry, active reflexes)
- End of first wk. feeds regular 2-5 hours.
- First stool passed within the first 24 hours. Meconium is a newborn's first poop. This sticky, thick, dark green poop is made up of cells, protein, fats, and intestinal secretions, like bile

Thermal regulation & metabolism

- At delivery same temp. as mother.
- End of first wk. 110 kcal/kg/day.
- Extracellular fluid compartment 35%
- Body wt lost in the first 10 days
 First 10 days of life = weight loss (water not lean mass), start gaining after 10 days.
- End of first wk 120-150 m/kg/d.

Renal System

- They reach the GFR of the adult at end of first year.
- Proteinuria common and urate crystals Turns urine pink. Tubules are not mature enough for absorption NOT nephrotic syndrome
- Urea clearance law and concentrating ability limited.

Hematology

- Hb 17 19 g/dl ¹
- WBC 10 30 x 10³
 Normal, unless they have signs of infections
- Plat. 150 750 x 10^3
- Coagulation (acquisition of gut flora).²

1- High HB as the baby in utero are relatively hypoxic and the bone marrow react and produce RBC, and then when they are born RBC hemolysis causing jaundice Normal, unless they have signs of infections

2- Coagulation disorder of newborn due to the deficiency of vitamin K (factor 2,5,7,9,10) as there is no normal flora to produce it. Hence, The babies are given vitamin K injection soon after birth. if u didnt give it you will increase the risk of having brain hemorrhages

When is it abnormal not to pass urine? 24 hrs and stool ? 48 hrs

MCQ: ★



Neonatal physiology

Immunity

- High IgG levels (can cross, materno-fetal transfer).
- IgM, IgE & IgA do not cross placenta. To diagnose congenital infection in newborn measure the IgM (IgG is maternal)
- Maternal IgG disappear by 3 months.

Metabolic

- Placental transfer of maternal hormone
- Diminished capacity of liver to conjugate

Common encounters



Hyperextend neck, cause the mother had fibroid during pregnancy which caused this abnormal posture and need physical therapy to fix it. Maternal Hx is important



Flat head

• Caused by C-section or breech delivery



Salmon patches

- Birthmarks that are caused by dilations of capillaries
- On the face called an angel kiss (Grows and becomes prominent but disappear at the age of one with no treatment), on the back of the neck, it is known as a stork bite (Persist and sometimes remains or disappear)



- It is common
- It usually disappears within the first 3 years of life.
- Not hemangioma (abnormal growth of the blood vessels)











Hemangioma Natural History A. At 1 month B. At 2 years C. At 7 years



Port-wine stain (Nevus flammeus)

- Early port-wine stains are usually flat and pink in appearance. May deepen to a dark red or purplish color with age.
- They occur most often on the face.
- Consisting of deep dilated capillaries in the skin
- Doesn't disappear easily

Strawberry hemangiomas

- Also called, nevus vascularis, capillary hemangioma, hemangioma simplex.
- Common type of vascular birthmark
- It is usually painless and harmless.
- Unknown cause
- Consist of small, closely packed blood vessels Abnormal growth of blood vessels not dilation
- May be absent at birth, and **develop at several weeks**.
- They disappear by the time a child is 9 years old The bigger the less likely to disappear
- No need for treatment unless it's huge or in a sensitive area such as the eyelid / B-blockers or Laser

Mongolian spots

- Congenital dermal melanocytosis
- Flat, blue, or blue-gray skin markings with wavy borders and irregular shape.
- Near or around the buttocks
- Commonly appear at birth or shortly thereafter.
- It disappears within 1-5 years from birth.
- Doesn't disappear easily
- Misdiagnosed as abuse





Harlequin phenomenon

- Transient in approximately 10% of healthy newborns.
- This presents as a well-demarcated colour change, with one half of the body displaying erythema and the other half pallor. Vasomotor instability of the blood vessels area constrict and the other dilates
- Usually occurring between two and five days of age, but can be seen as late as three weeks of age
- The condition is benign, and the change of colour fades away in 30 seconds to 20 minutes Common with trisomy 21

Cutis Marmorata

- A reticulated mottling of the skin that symmetrically involves the trunk and extremities.
- It is caused by a vascular response to cold and generally resolves when the skin is warmed.
- A tendency to cutis marmorata may persist for several weeks or months, or sometimes into early childhood.
- No treatment is indicated. It is also commonly found in babies with Down's syndrome

Miliaria crystallina

- Superficial sweat ducts obstruction Under epidermal layer
- It consists of 1- to 2-mm vesicles without surrounding erythema
- Common on the head, neck, and trunk
- Rupture followed by desquamation
- Persist for hours to days.

Erythema toxicum neonatorum





- Common benign self-limited rash Sebaceous gland obstructed with surrounding erythema
- Usually appear between day 2-5 after birth
- Characterized by blotchy red spots on the skin with overlying white or yellow papules or pustules
- May be few or numerous that typically resolves within a few days. Not seen on the palm and sole
- Benign no treatment is needed
- Under microscope: full of eosinophils. Because it allergenic and cause the release of eosinophils









Neonatal pustular melanosis

- Transient, a benign idiopathic skin condition
- Mainly seen in newborns with skin of color Born with the rash
- Distinctive features characterized by vesicles, superficial pustules, and pigmented macules Seen on palms and soles
- The vesicles and pustules rupture easily and resolve within 48 hours
- Under microscope: neutrophils but no infection
- Benign no need for treatment

MCQ:



Sucking blisters

- Erosions, or calluses result from vigorous sucking by the infant during fetal life.
 - The lesions are present at birth and resolve without specific treatment within days to weeks.



Tonic neck reflex

- The face is turned to one side, the arm and the leg on that side stretch out and the opposite arm and leg bend up.
- It is called the "fencing" position
- The tonic neck reflex lasts about six to seven months
- Present at 6 wks (not from birth)
- it should disappear by 6 month



Ranula

- Cystic lesions in the floor of the mouth
- Retention cysts of the excretory duct of the sublingual gland
- Benign and causing no discomfort
- It has the tendency to rupture or resolve spontaneously within the first few months of life.
- Treatment: only if it's large and interfere with breathing or feeding











Natal - Neonatal teeth

- Teeth that are already present at the time of birth.
- Neonatal teeth, which grow in during the first 30 days after birth.
- In both cases, once erupted, teeth has to be extracted due to the danger of getting aspirated or may cause discomfort.
- removed only if its loose because there is risk of aspiration, or if the baby is biting the mother

Neonatal gynecomastia Can happen in both boys and girls

- Is caused by the passage of maternal hormones across the placenta during pregnancy leading to the enlargement of the breasts Some babies tissues are very sensitive to hormone
- Usually progressing over the first 2 months of life.
- Breast discharge is also sometimes seen which is commonly called "witch's milk".Don't squeeze it, risk of infection

Neonatal Menstruation

Only in girls

- Is caused by the passage of maternal hormones across the placenta during pregnancy. Stimulating the endometrium
- Withdrawal bleeding takes place at the end of first week
- Usually progressing over the first 2 months of life.
- It is benign and needs no treatment





Extra

Tongue tie: Delay the treatment unless it's interfere with feeding or breathing.

Subconjunctival hemorrhage



- Bleeding underneath the conjunctiva.
- Appears as bright red patch in the sclera
- It is a painless and harmless.Normal and doesn't affect the baby vision
- As a consequence of elevated venous pressure in the head and neck. Common when the shoulder is stuck during delivery, or cord around the neck
- It usually resolves within 1-2 weeks

Caput succedaneum, and Cephalhematoma

Caput succedaneum	Cephalhematoma
Diffuse scalp swelling that extends across the	localized effusion of blood beneath the periosteum
midline and over suture lines.	of the skull, due to disruption of the vessels during
It is commonly associated with head moulding.	birth.
Serous edema under the skin, And can come to	It does not cross cranial suture lines. Common on
the eyes	the parietal area
Occurring due to pressure from uterine or vaginal	It is firmer to the touch than an edematous area.
wall during vertex vaginal delivery.	It usually appears on the second or third day after
Does not usually cause complications and	birth and disappears within weeks or months.
resolves over the first few days	
Salital suture Skull bone Cross the Suture	Scolp Periosteum Skull bone Doesn't Cross the suture



Subgaleal hematoma:bleeding between glea and periosteum, usually it's localized to one bone but can involve the entire head,baby can be in shock it should be treated immediately (emergency) you can't push the injury to stop the bleed, the aponeurosis space can take all you the blood of body, you have to take him to ICU.

Erb's Palsy

- C5-C6 Injury
- Paralysis of deltoid, supraspinatus, biceps and teres major.
- Loss of sensation over deltoid, lateral forearm and hand
- The elbow is extended and the forearm is pronated (the "waiter's tip position).
- The Moro reflex is absent but grasp reflex of the hand is present
- Occurs with breech delivery
- Injury is more common on the nerve sheath and resolve with time, It can be accompanied with phrenic nerve injury causing elevated diaphragm
- Management:observe for two weeks most of the infant will recover if not>physiotherapist for 2 weeks then referral to neurosurgeon.

Klumpke Palsy

- lower plexus injury to the lower roots of the brachial plexus
- Involving C8 and T1 roots.
- There is loss of grasp reflex. The hand is supinated, the wrist extended, and the fingers clawed .

Facial Palsy

- Lower motor neuron lesion
- Both upper and lower parts are affected
- Conservative treatment Protect and close the eye to prevent keratitis
- During the forceps delivery or compression

Asymetric crying facies

- A congenital deficiency or absence of the depressor anguli oris muscle which controls the downward motion of the lip.
- The eye and forehead muscles are unaffected

Other Injuries:











Swelling in the left side when compare to the normal dipping on the other side Clavicle fracture management:observe,family instruction.







Soft-tissue injuries

- chignon oedema and bruising from Ventouse delivery
 bruising to the face after a face presentation and to the genitalia and buttocks after breech delivery. Preterm infants bruise readily from even mild trauma
- abrasions to the skin from scalp electrodes applied during labour or from accidental scalpel incision at caesarean section
- forceps marks to face from pressure of blades transient
 subaponeurotic haemorrhage diffuse, boggy swelling of scalp on examination, blood loss may be severe and can lead to hypovolaemic shock and coagulopathy.



Some significant abnormalities detected on routine examination

Box 10.8 Some significant abnormalities detected on routine examination

- Port-wine stain (naevus flammeus). Present from birth and usually grows with the infant (Fig. 10.15a). It is due to a vascular malformation of the capillaries in the dermis. Rarely, if along the distribution of the trigeminal nerve, it may be associated with intracranial vascular anomalies (Sturge-Weber syndrome), or severe lesions on the limbs with bone hypertrophy (Klippel-Trenaunay syndrome). Disfiguring lesions can now be improved with laser therapy.
- Strawberry naevus (cavernous haemangioma). Usually not present at birth, but appear in the 1st month of life and may be multiple (Fig. 10.15b). They are more common in preterm infants. Increase in size until 3-15-months of age, then gradually regress. No treatment is indicated for small lesions, but topical propranolol may speed regression. Large lesions or if interferes with vision or the airway are treated with oral propranolol. Ulceration or haemorrhage may occur.
- Natal teeth consisting of the front lower incisors - may be present at birth. If loose, they should be removed to avoid the risk of aspiration.
- Extra digits are sometimes connected by a thin skin tag but may be completely attached containing bone (Fig. 10.15c) and should ideally

be removed by a plastic surgeon or else tied off at its base. Skin tags anterior to the ear and accessory auricles should be removed by a plastic surgeon.

- Heart murmur poses a difficult problem, as most murmurs audible in the first few days of life resolve shortly afterwards. However, some are caused by congenital heart disease. If there are any features of a significant murmur (see Ch. 18), upper and lower limb blood pressures, and pre-ductal and post-ductal pulse oximetry should be checked followed by an echocardiogram. Otherwise, a follow-up examination is arranged and the parents warned to seek medical assistance if their baby feeds poorly, develops laboured breathing, or becomes cyanosed.
- Midline abnormality over the spine or skull, such as a tuft of hair, swelling, or naevus requires further evaluation as it may indicate an underlying abnormality of the vertebrae, spinal cord, or brain.
- Palpable and large bladder if there is urinary outflow obstruction, particularly in boys with posterior urethral valves. Requires prompt evaluation with ultrasound. Usually diagnosed on antenatal ultrasound.
- Talipes equinovarus cannot be corrected as in positional talipes.



Figure 10.15a Port-wine stain in an infant.



Figure 10.15b Strawberry naevus.



Figure 10.15c Extra digits.

Lesions in newborn infants that resolve spontaneously

Box 10.7 Lesions in newborn infants that resolve spontaneously

- Peripheral cyanosis of the hands and feet - common in the 1st day.
- Traumatic cyanosis from a cord around the baby's neck or from a face or brow presentation – causes blue discolouration of the skin, petechiae over the head and neck or affected part but not the tongue.
- Swollen eyelids and distortion of shape of the head from the delivery.
- Subconjunctival haemorrhages occur during delivery but should be documented to avoid confusion with non-accidental injury when older.
- Small white pearls along the midline of the palate (Epstein pearls).
- Cysts of the gums (epulis) or floor of the mouth (ranula).
- Breast enlargement may occur in newborn babies of either sex (Fig. 10.14a). A small amount of milk may be discharged.
- White vaginal discharge or small withdrawal bleed in girls. There may be a prolapse of a ring of vaginal mucosa.
- Capillary haemangioma or 'stork bites' pink macules on the upper eyelids, midforehead, and nape of the neck are common and arise from distension of the dermal capillaries. Those on the eyelids gradually fade over the 1st year; those on the neck become covered with hair.

- Neonatal urticaria (erythema toxicum) a common rash appearing at 2–3-days of age, consisting of white pinpoint papules at the centre of an erythematous base (Fig. 10.14b). The fluid contains eosinophils. The lesions are concentrated on the trunk; they come and go at different sites.
- Milia white pimples on the nose and cheeks, from retention of keratin and sebaceous material in the pilosebaceous follicles (Fig. 10.14c).
- Mongolian blue spots blue/black macular discolouration at the base of the spine and on the buttocks (Fig. 10.14d); occasionally occur on the legs and other parts of the body. Usually but not invariably in Afro-Caribbean or Asian infants. They fade slowly over the first few years. They are of no significance unless misdiagnosed as bruises.
- Umbilical hernia common, particularly in Afro-Caribbean infants. No treatment is indicated as it usually resolves within the first 2–3-years.
- Positional talipes the feet often remain in their in utero position. Unlike true talipes equinovarus, the foot can be fully dorsiflexed to touch the front of the lower leg (Fig. 10.14e and f).
- Caput succedaneum (see Fig. 11.6) and cephalhaematoma (Figs. 11.6 and 11.7).



Figure 10.14a Breast enlargement in a newborn infant.



Figure 10.14b Erythema toxicum (neonatal urticaria) often has a raised pale centre (Courtesy of Nim Subhedar.)



Figure 10.14d Mongolian blue spot.



Rodney Rivers.)

Figure 10.14c Milia (Courtesy of

Figure 10.14e Positional talipes. Appearance at birth. Figure 10.14f The foot can be fully dorsiflexed to touch the front of the lower leg. In true talipes equinovarus this is not possible.

EXTRA

Neonatal Jaundice

Over 50% of all newborn infants become visibly jaundiced (Fig. 11.15). This is because:

- There is marked physiological release of haemoglobin from the breakdown of red cells because of the high haemoglobin concentration at birth
- The red cell lifespan of newborn infants (70 days) is markedly shorter than that of adults (120 days)
- hepatic bilirubin metabolism is less efficient in the first few days of life.

Neonatal jaundice is important as:

- it maybe a sign of another disorder,
 - e.g.haemolytic anaemia, infection, inborn error of metabolism, liver disease
- unconjugated bilirubin can be deposited in the brain, particularly in the basal ganglia, causing brain damage from kernicterus

Kernicterus

- This is the **encephalopathy** resulting from the deposition of unconjugated bilirubin in the basal ganglia and brainstem nuclei .
- It may occur when the level of unconjugated bilirubin exceeds the albumin-binding capacity of bilirubin of the blood. As this free bilirubin is fat soluble, it can cross the blood-brain barrier. The neurotoxic effects vary in severity from transient disturbance to severe damage and death.
- Acute manifestations are lethargy and poor feeding. In severe cases, there is irritability, increased muscle tone causing the baby to lie with an arched back (opisthotonos), seizures and coma. Infants who survive may develop choreoathetoid cerebral palsy

Jaundice <24 hours of age (Haemolytic disorders)

Rhesus haemolytic disease

• Usually identified antenatally and monitored and treated if necessary.

• The birth of a severely affected infant, with anaemia, hydrops and hepatosplenomegaly with rapidly developing severe jaundice has become rare.

• Antibodies may develop to rhesus antigens other than D and to the Kell and Duffy blood groups.

Table 11.3 Causes of neonatal jaundice

Jaundice starting at <24 h of age	Haemolytic disorders: Rh (rhesus) incompatibility ABO incompatibility G6PD deficiency Spherocytosis, pyruvate kinase deficiency Congenital infection
Jaundice at 24 h to 2 weeks of age	Physiological jaundice Breast milk jaundice Infection, e.g. urinary tract infection Haemolysis, e.g. G6PD deficiency, ABO incompatibility Bruising Polycythaemia Crigler–Najjar syndrome
Jaundice at >2 weeks of age	Unconjugated: Physiological or breast milk jaundice Infection (particularly urinary tract) Hypothyroidism Haemolytic anaemia, e.g. G6PD deficiency High gastrointestinal obstruction, e.g. pyloric stenosis Conjugated (>25 µmol/l): Bile duct obstruction Neonatal hepatitis

EXTRA

Neonatal Jaundice

Jaundice <24 hours of age (Haemolytic disorders)

ABO incompatibility

• Most ABO antibodies are IgM and do not cross the placenta, but some group O women have an IgG anti- A-haemolysin in their blood, which can cross the placenta and haemolyse the red cells of a group A infant. Occasionally, group B infants are affected by anti-B haemolysins.

• Haemolysis Can Cause Severe Jaundice

• The infant's haemoglobin level is usually normal or only slightly reduced and, in contrast to rhesus disease, hepatosplenomegaly is absent.

• The direct antibody test (Coombs' test), which demonstrates antibody on the surface of red cells, is positive.

Jaundice at 2 days–2 weeks of age

Physiological jaundice

• Most babies who become mildly or moderately jaundiced during this period have no underlying cause and the bilirubin has risen as the infant is adapting to the transition from fetal life. The term 'physiological jaundice' can only be used after other causes have been considered.

Breast milk jaundice

- Jaundice is more common and more prolonged in breastfed infants.
- The hyperbilirubinemia is unconjugated. The cause is multifactorial but may involve increased enterohepatic circulation of bilirubin.

Dehydration

• In some infants, the jaundice is exacerbated if milk intake is poor from a delay in establishing breastfeeding and the infant becomes dehydrated (>10% weight loss from birth weight).

• Breastfeeding should be continued, although supplemental feeding is sometimes needed to reverse the dehydration.

Infection

• An infected baby may develop an unconjugated hyperbilirubinemia from poor fluid intake, haemolysis, reduced hepatic function and an increase in the enterohepatic circulation.

• If infection is suspected, appropriate investigations and treatment should be instigated. In particular, urinary tract infection may present in this way.

EXTRA



Neonatal Jaundice

Management

Phototherapy

• Light (wavelength 450 nm) from the blue–green band of the visible spectrum converts unconjugated bilirubin into a harmless water-soluble pigment excreted predominantly in the urine.

• It is delivered with an overhead light source placed at an optimal distance above the infant to achieve high irradiance.

• The infant's eyes are covered, as bright light is uncomfortable.

• Phototherapy can result in temperature instability as the infant is undressed, a macular rash, and bronze discoloration of the skin if the jaundice is conjugated.

Exchange transfusion

• Exchange transfusion is required if the bilirubin rises to levels that are considered potentially dangerous.

• Usually, twice the infant's blood volume (2 × 90 ml/kg) is exchanged.

Jaundice at >2 weeks of age

• Jaundice in babies >2 weeks old (3 weeks if preterm) is called persistent or prolonged neonatal jaundice, and needs to be evaluated differently from jaundice at an earlier age

• The key feature is that it may be caused by biliary atresia, and it is important to diagnose biliary atresia promptly, as delay in surgical treatment adversely affects outcome. However, in most infants with persistent neonatal jaundice, the hyperbilirubinemia is unconjugated, but this needs to be confirmed on laboratory testing.

In prolonged unconjugated hyperbilirubinemia

- breast milk jaundice is the most common cause
- infection, particularly of the urinary tract
- congenital hypothyroidism may cause prolonged jaundice





EXTRA Common encounters





Normal red reflex

Leukocoria Can be retinoblastoma or cataract interrupting light





Bilateral Large iris and hazy eyes > congenital glaucoma



The Ears

Low set ear: less than 1/3 of ear is above an imaginary line between eye and occiput. Can be associated with trisomy 21 or 18...



Preauricular sinus and tag can be associated with renal anomalies



Normal ear



Can come with rubella; test hearing

EXTRA Common encounters

- Congenital : the defect is present at birth Not necessary genetic
- Major (2% to 3% of live born infants) Less common but it affect the function
 - Medical and social consequences (e.g. cleft palate and neural tube defects)
- Minor (Up to 15%)
 - No significant health or social burden (e.g. a single palmar crease)
- Normal phenotypic variants Like the differences in eye shape
 - Physical differences occurring in 4% or more of a general population

Malformations

- Abnormal processes during the initial formation of a structure
- May result in:
 - Faulty configuration (e.g. TGA)
 - Incomplete formation (e.g. cleft palate)
 - Agenesis (e.g. absence of radius)

Causes

- Genetic
 - chromosomal(10%), or single gene (4%)
- Environmental insults (teratogens)
 - Drugs thalidomide
 - Congenitally acquired viruses Rubella
- Multifactorial (25%)
- Unknown (40%-45%)

Disruptions

Breakdown of normal tissue after formation

Causes

 Mechanical compressive forces, hemorrhage, thrombosis, and other vascular impairments

Manifestations

• Alterations of configuration, division of parts not usually divided, fusion of parts not usually fused, and the loss of previously present parts



Major



Minor



Normal variant







The limb Was normally formed previously

EXTRA Common encounters

The nose

- Shape & Size,
 - Race and family determined
- Patency
 - Infant is an "obligate nasal breather"
 - Shut infant's mouth to look for choanal atresia
- Deformities
 - Trauma or syndromic





Here the base is widened and deviated. High risk of perforation (emergency)call ENT

The base is in the mid but the septum is deviated only. Can be secondary to oligohydramnios. It will be corrected with time but make sure that baby is breathing and feeding

Sacral dimples



Which sacral dimple to investigate? When to suspect other issues and you should investigate

- >5 mm in size
- >25 mm cephalad to the anal orifice.
- Associated with overlying cutaneous markers:
 - True hypertrichosis, or hairs within the dimple
 - Skin tags
 - Telangiectasia or hemangioma
 - Subcutaneous mass or lump.
 - Abnormal pigmentation.
 - Bifurcation (fork) or asymmetry of the superior gluteal crease

Ultrasonography is the screening modality of choice

EXTRA Congenital Anomalies

Can be non inherited

Deformations

- Unusual and prolonged mechanical forces acting on normal tissue
- External (uterine constraint) vs. intrinsic (edema)
- Mostly Musculoskeletal tissues
 - Tibial bowing and hip dislocation associated with breech presentation
 - Webbing of the neck upon the involution of a giant cystic hygroma)
- Typically, deformations improve postnatally
- Their resolution depends on the duration of the abnormal forces and the extent of subsequent growth. Early closure of the sutures can lead to abnormal skull shape

Dysplasia

- Abnormal cellular organization or function
- Typically, affects a single tissue type

Examples

- Ectodermal dysplasia abnormal teeth and abnormal fingernails
- Skeletal dysplasia and hamartoma

Multiple Anomalies



Genu recurvatum





Ectodermal dysplasia

Ectodermal dysplasia

EXTRA Congenital Anomalies



The hips



It's not dislocation cause it's not caused by trauma

The left is shorter

Barlow:guiding the hips into mild adduction and applying a slight downward pressure with the thumb. If the hip is unstable, will produce a palpable sensation of subluxation or dislocation. Means you are trying to dislocate the hip but the ortolani is the opposite you are trying to reduce it.

The genitals

Inguinal hernia vs.



Inguinoscrotal swelling with negative transillumination test. Evaluate and it should be treated



Hydrocele ?

Scrotal swelling with a positive transillumination test Reassure the family and follow up





Hypospadias No circumcision before referring to urologists.

11. Ambiguous genitalia

Most important thing is to identify the cause then to determine the gender of baby, investigate thoroughly.









1- Natasha, a female infant, is delivered by caesarean section at 32 weeks' gestation because of maternal pre-eclampsia. Her birth weight is 1.9 kg. No resuscitation is required. At 2 hours of age she develops respiratory distress, with a respiratory rate of 70 breaths/min, grunting respirations, and indrawing of her rib cage. Respiratory support with CPAP (continuous positive airway pressure) and 45% oxygen is required. A chest X-ray is taken 4 hours of age, and is shown (Fig. 11.1).

What is the most likely reason that this baby needs oxygen therapy and respiratory support?

- A. Aspiration of meconium has resulted in lung collapse
- B. Blood is still flowing from the pulmonary artery to the aorta as in the fetal circulation
- C. The alveoli still contain fluid
- D. The chest wall and ribs are too compliant
- E. There is ventilation-perfusion mismatch from surfactant deficiency

2- Robert is a full term male infant, born 10 hours ago. His mother is blood group O rhesus positive and her membranes ruptured 2 days before delivery. He is breastfeeding well but the midwife noticed he looks jaundiced. On examination the baby is clinically well but markedly jaundiced.

What investigation should be performed first?

- A. Bilirubin level
- B. Blood culture
- C. Blood group
- D. Congenital infection screen
- E. Direct antibody test

3- You are asked to review a newborn baby who is only 24 hours old and has developed very swollen eyelids with a purulent discharge.

What management is required?

- A. Clean with cool boiled water
- B. Intravenous antibiotics
- C. Oral antibiotics
- D. Reassure that it will resolve spontaneously
- E. Topical antibiotic therapy

4- Zak, a full-term male infant, with a birth weight of 3.7 kg, is born by elective caesarean section. His mother was well during pregnancy and had a normal blood glucose screen. Zak becomes tachypnoeic with indrawing between his ribs at 2 hours of age. Examination is otherwise normal. A chest X-ray looks normal.

Select the most likely diagnosis.

- A-Pneumonia
- **B- Pneumothorax**
- C- Respiratory distress syndrome
- D- Tracheo-oesphageal fistula
- E- Transient tachypnoea of the newborn

5- George, a 2-day-old baby boy, is reviewed by the paediatric doctor at the request of the midwife. He was born by forceps delivery and has been feeding well. There were no risk factors for sepsis and mother was well during her pregnancy. When examined, he appears very well but has the rash shown in Fig. 10.1. His mother reports the rash keeps moving around his body.

- A. Erythema toxicum (neonatal urticarial)
- B. Group B streptococcal infection
- C. Milia
- D. Mongolian blue spots
- E. Neonatal varicella zoster





Answers

1- E. There is ventilation-perfusion mismatch from surfactant deficiency Correct. Surfactant deficiency is common in preterm infants. This results in alveolar collapse which in turn results in areas of lung being perfused but not ventilated. The CXR appearance here is typical with a diffuse granular or 'ground glass' appearance.

2- A. Bilirubin level

Correct. Jaundice starting at less than 24 hours of age is most likely due to haemolysis and may rapidly rise to dangerously high levels. It needs urgent assessment and close monitoring The most urgent investigation is to measure the bilirubin level, as this will determine the management required.

3- B. Intravenous antibiotics

Correct. Purulent discharge and eyelid swelling in the first 48 hours of life must be taken seriously. This should be treated promptly, e.g. with a third-generation cephalosporin intravenously, as permanent loss of vision can occur. The most common cause of severe neonatal purulent conjunctivitis is Chlamydia trachomatis, but gonococcus may also be the cause.

4- E. Transient tachypnoea of the newborn The most common cause of respiratory distress in a term infant. It is due to a delay in the resorption of lung liquid – an increased risk following caesarean section, as the babies have not had to undergo the same physical and physiological stressors as those who pass through the birth canal.

5- A.