CONGENITAL ABNORMALITIES

This section only deals with a few of the more common congenital abnormalities which you may see. There are thousands of less common congenital abnormalities not covered here. If you are not sure whether or not a baby is abnormal, or what to do with the baby, **consult a paediatrician**.

LEARNING OBJECTIVES

In this section you will learn:
- To recognise some major congenital abnormalities
- To provide emergency care
- What plans must be implemented to manage the baby
- The principles of counselling parents about the condition of the baby

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CONGENITAL ABNORMALITIES

Look to see if the baby has any major abnormalities and minor abnormalities. Common major abnormalities include the conditions we have dealt with such as neural tube defect, cleft lip or palate, club foot, microcephaly, omphaolocoele.
Minor abnormalities include extra digits, abnormal size, shape or position of the eyes, ears, nose, chin and digits.

If a baby has one major abnormality and two minor abnormalities or 3 minor abnormalities the baby is classified as having a major congenital abnormality and needs to have blood taken for genetic analysis and referred to a paediatrician for assessment.

A baby with only one or two abnormalities (dysmorphisms) is classified as having a minor abnormality. Consult a paediatricians, genetic nurse or textbook for management.

The management of the most common major and minor congenital abnormalities are discussed in turn in this section.

NEURAL TUBE DEFECTS

What is a neural tube defect?

- A neural tube defect is a lesion in which the spine has not yet closed completely posteriorly. Meninges and spinal nerve tissue may bulge through this gap.
- The gap in the spine is known as **spina bifida**. This may not be visible.

- A **meningocoele** is when the meninges have bulged out through the gap, so that there is a lump over the midline of the spine. This may be covered by a membrane.
- A **myelomeningocoele** is when the meninges and spinal nerve tissue have come out in the sac. This often results in paralysis of the lower limbs and involvement of the bladder and bowel. The outflow of cerebrospinal fluid may be blocked so that the baby may also develop hydrocephalus.
- The lesion may be covered with skin, meninges, or be completely open and draining cerebrospinal fluid.

**With neural tube defects, the higher the lesion in the spine,
the more likely there is to be significant neurological damage**

Clinical features of neural tube defects

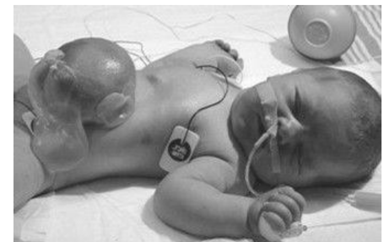
- The swelling over the midline of the back
- Poor motor functioning of the lower limbs. Check for deformities and whether the baby is moving the legs normally?
- Poor bladder function. Urine tends to dribble when the bladder is paralysed, and the bladder may be palpable.
- Poor bowel function. Look at the anus. If there is paralysis the tone of the anal muscles will be poor, and meconium may be leaking out.
- Measure, record, and chart the size of the head on a growth chart. Hydrocephalus is commonly associated with neural tube defects.

Management of neural tube defects

- If the lesion is not covered by skin, cover it with sterile “Opsite” or cling film. The purpose of this is to prevent further damage, infection and cerebrospinal fluid drainage.
- If there is no neurological deficit, refer the baby urgently to a neurosurgical service for immediate closure.
- All these babies will need to be referred and it is better to do this sooner rather than later. Consult with a neurosurgeon first.
- Measure and record the head circumference daily while in hospital and weekly thereafter. Refer to the neurosurgical service urgently if hydrocephalus develops.
- Refer and follow up at a special clinic that will monitor development, provide rehabilitation and bladder and bowel care.
- Counsel the parents
    - About the plans and probable outcomes for the baby.
    - About the need for planning the next pregnancies and to take folic acid before and during the pregnancy. The mother will need a letter to take to the clinic to get the folic acid when she is planning her next pregnancy.

MAJOR GASTROINTESTINAL ABNORMALITY

The following lesions need to be treated urgently



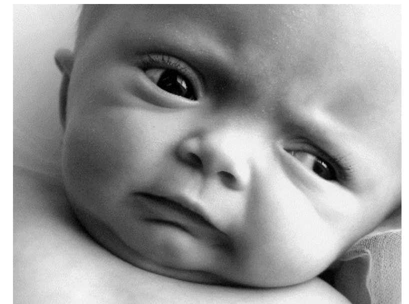
- **Omphalocoele:** This is a defect at the base of umbilicus with protrusion of abdominal contents **covered by peritoneum**. The baby may have other abnormalities.
- **Gastroschisis**: This is a defect usually to the right of the umbilicus with protrusion of the abdominal contents **without peritoneal covering**.
- **Imperforate anus**: There is no clear anal canal on examination. These babies will not be able to pass meconium normally. However: there may be a recto-vaginal fistula in a girl or a recto-urethral fistula in a boy, so that meconium may be seen on the nappy.
- **Congenital bowel obstructions**
    - If this is proximal in the bowel, the baby presents with bile stained vomiting with little abdominal distension.
    - If the lesion is in the distal bowel, the baby will present with abdominal distension and the bile stained vomiting only starts later.
    - In both cases the baby may pass meconium.

Remember: Always look at the baby's anus as part of the newborn examination

Management of major gastrointestinal abnormality

- Keep the baby nil per mouth
- Give IV fluids (Neonatalyte) at the appropriate rate for weight and age.
- Pass a nasogastric tube, and leave it open to drain.
- Cover the open lesions with sterile Opsite or cling film or a plastic bag
- Keep the baby warm
- Refer the baby urgently to a tertiary paediatric surgical service.

HYDROCEPHALUS



- A baby's head normally grows because the brain is growing. If the head grows too rapidly it is usually because there is excessive fluid in or around the brain.
- Hydrocephalus is one of the causes of macrocephaly (a large head, greater than the 97th centile). All babies with macrocephaly need to be referred for assessment and investigation at a level 3 service.
- If the head circumference is increasing rapidly, this means that there is almost certainly hydrocephalus. These babies need to be referred for neurosurgical drainage.
- **Do not delay the referral** to a tertiary centre for neuro-imaging. Surgery for hydrocephalus is an emergency and should not be delayed.

The head circumference of a newborn baby normally increases at about 1 mm / day (0.7 cm / week)

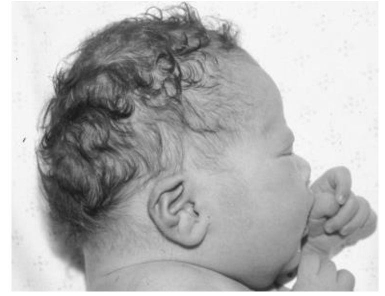
AMBIGUOUS GENITALIA

Ambiguous genitalia is when it is difficult to decide on the gender of the newborn by looking at the external genitalia.

The parents must be counselled and advised that the gender is not clear. The management of the newborn is:

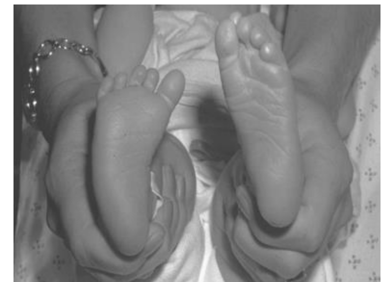
- Check the sodium, potassium and urea of the newborn immediately for hypoaldosteronism
- Refer the baby as soon as possible to a paediatrician for investigations and gender assignment

MICROCEPHALY



- This means that the baby has a small head – the head circumference is less than the 3rd centile
- The head does not grow because the brain does not grow. It is often associated with other abnormalities
- Management of microcephaly is::
  - Compare the weight and head circumference centile
  - Investigated for other abnormalities such as congenital infection, a structural abnormality of the brain or be part of a genetic syndrome.
  - Counsel the parents
  - Refer for a paediatric assessment (not urgent)
- They usually have varying degrees of developmental delay and mental impairment
- The parents need to be counselled about the condition and prognosis for the baby

CLUB FOOT



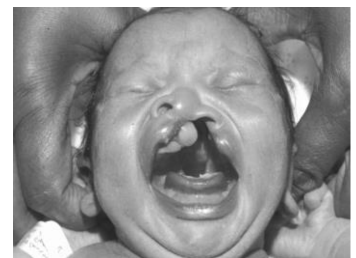
The commonest form is Talipes equinovarus in which the foot is plantar flexed at the ankle(bending of the foot downwards), with the sole turned inwards and the fore foot also bent inwards. The opposite deformity sometimes occurs.

It may be the result of the position of the foot in the uterus, a developmental abnormality in the bone or cartilage, a neuromuscular problem or a spinal cord problem.

Management of club foot

- Assess the baby for other problems of the bone, spine or central nervous system. If these are present the baby needs to be referred to a paediatrician in a level 3 hospital
- If the baby is otherwise normal refer as soon as possible for orthopaedic correction, usually serial splinting or plaster of Paris casts.
- If these measures do not work surgical correction must be planned at 10 weeks. Delay in referral and starting treatment of these babies may result in the above measures not working and the babies being permanently disabled. .

CLEFT LIP AND / OR PALATE

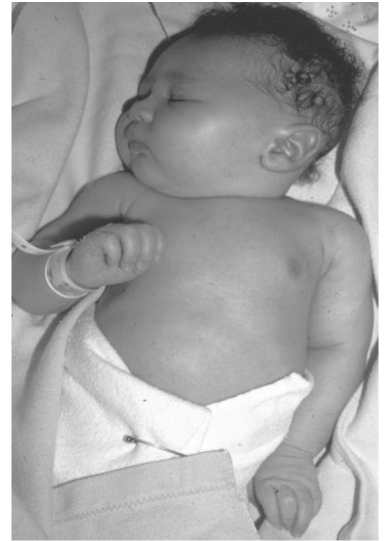


There is a gap in the lip, gum margin and / or palate as the result of incomplete closure of the skin, bones and muscles of the face and mouth. The gap may be on one side(unilateral), both sides(bilateral), in the midline, and may involve the whole of the palate, mandible and lip(complete), or only some parts(incomplete). There may be other associated abnormalities, and sometimes it occurs in families.

Management of cleft lip and palate

- Examine the baby carefully to exclude other abnormalities or syndromes. If these are found or suspected, refer to the tertiary unit for further assessment.
- Counsel the parents
- Assist the mother with feeding as these babies do not always feed easily. Sometimes they can breast feed normally.
- The baby needs to be referred to a cleft lip / palate, or maxillo-facial clinic at a level 3 hospital.
- A plate is often fitted to assist the feeding, and the cleft lip is usually closed at about 3 months of age. The palate is usually repaired at about 9 months of age.

LIMB INJURIES



These are often associated with a difficult or assisted delivery of the baby.

- Shoulder dystocia:There may be a fracture of the clavicle, or humerus, or a brachial plexus injury (Erb's palsy)
- Breech delivery: There may be a fracture of the femur or humerus

Clinical features of limb injuries

- There is an abnormal position of the arm or leg
- The limb does not move well
- There may be pain on movement of the limb

Management of limb injuries

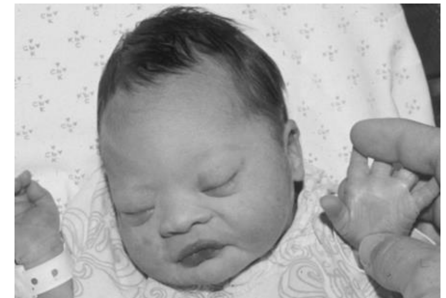
- Counsel the parents
- Handle the baby gently
- X-ray the affected limb if a fracture is suspected
- Exclude congenital syphilis as a cause of the painful limb
- If there is a fracture, immobilise the limb and treat with advice from orthopaedic doctors.
- If the arm is not moving, and flaccid, and no fracture is present a brachial plexus palsy is likely. Show the mother how to move the arm gently and refer to physiotherapy. The palsy normally recovers spontaneously, but if the arm is not better, the baby should be referred to an orthopaedic surgeon.

MAJOR CONGENITAL ABNORMLAITY

DOWN SYNDROME

This is a chromosome abnormality with an additional chromosome 21: i.e. trisomy 21

Clinical features of Down syndrome



- The babies are usually less than 2.8kg at birth
- The babies have a characteristic appearance
- The face is rather flat and the eyes slant downwards. The bridge of the nose is quite wide.
- The ears are small and might be low set
- The occiput (back of the head) is rather flat
- The baby has rather low muscle tone – slightly "floppy"
- The hands and feet are short and wide. The hands typically, but not always have a single palmar crease, and the feet a gap between the 1st and 2nd toes

The babies may also have:

- Congenital heart lesions
- Duodenal atresia

Management of Down syndrome

- Arrange for a paediatrician and genetic nurse to see the baby and ensure appropriate follow up.
- The diagnosis should ideally be confirmed with chromosome analysis or Quantitative Flouresecent (QF) PCR for Aneuploidy, that detects Trisomy 13, 18, 21. If you are certain of the clinical features and the parents are not going to have more children this can be omitted. Chromosomal analysis and QF PCR for Aneuploidy can be done at level 2 and 3 facilities.
- Counsel the parents

MINOR CONGENITAL ABNORMALITY

EXTRA DIGITS

This is a common abnormality. There is usually a sixth digit on the hand or foot. It is attached to the fifth digit usually by a thin pedicle. They are often familial.

Management of extra digits

- If the pedicle is **thin** (less then 1mm), the digit can be tied off tightly close to the finger (while the baby is breastfeeding or has another form of analgesia).
- If the pedicle is **not thin**, the baby should be discussed with a paediatrician