UNIT 3 CONGENITAL MALFORMATIONS

Structure

3.0 INTRODUCTION

We have all seen a baby with cleft lip or palate, or a baby with club feet. These are birth defects that are obvious to anyone. Many may manifest in the newborn period, such as oesophageal atresia, imperforate anus, meningomyelocele or exomphalos major. Others may not manifest early, and present later as failure to thrive, recurrent chest infections or poor feeding as in congenital heart disease, or poor urinary stream and repeated urinary infections in a male child with posterior urethral valves.

Birth defects or anomalies can occur among 6–7 of every 100 live births. They could occur because of Genetic facors, Environmental factors or a mix of both (this is more common). Some of them may be minor, but many cause significant problems, including causing premature deaths and significant disability. We shall learn about the common and important congenital malfunctions in this unit.

3.1 OBJECTIVES

After completing this unit, you should be able to:

- recognise some of the common and serious birth defects in the newborn period and early childhood;
- provide immediate care where feasible and prior to referral in order to prevent further damage;
- know briefly about their broad management to facilitate early referral to appropriate facility; and
- provide follow up care required after the definitive procedure has been done which should be ensured for the child.

3.2 GENERAL CARE FOR NEWBORNS WITH BIRTH DEFECTS

The newborn with birth defects who needs referral, must be transported while ensuring that the baby is kept warm. This is because they do not shiver to bring up body temperature. Also preterm babies are at additional risk of hypothermia. Also the babies who loose heat due to exposed internal viscera such as exomphalos, Birth defect of abdominal wall, meningomyelocele, and exstrophy bladder, require additional care to prevent hypothermia. They should be wrapped using foil or cotton and cling film around each limb and the scalp; and then wrapped in a small blanket Fig. 3.1. Any exposure to cold draughts of air should be avoided.

Fig. 3.1: Newborn Wrapped with foil

3.3 CLEFT LIP AND CLEFT PALATE

This anomaly may be one sided (more common) or involve both sides. The palate may or may not be cleft with cleft in the lip. Palatal clefts can also be isolated. They may involve both the anterior and posterior parts of the palate (complete cleft palate) or may be incomplete, usually involving only the posterior or secondary palate (Fig. 3.2 and Fig. 3.3).

Fig. 3.2: Cleft lip and Palate, Unilateral

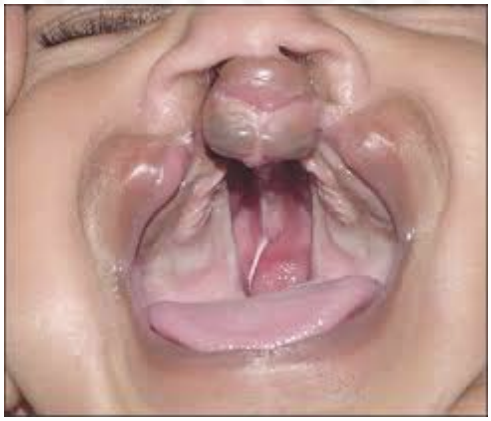

Fig. 3.3: Cleft lip and Palate, Bilateral

Besides the obvious visible defect, the baby may have difficulty in feeding due to poor sucking, nasal regurgitation of milk, often leading to poor weight gain and undernutrition. The mother should be instructed to provide expressed breast milk using a katori-spoon. Often a pallada spoon is useful to deposit the milk into the posterior half of the oral cavity.

Another common presentation is with recurrent ear discharge and poor/unclear speech. This is due to nasal escape of air while attempting to speak, as also eustachian tube blockage.

Definitive treatment requires surgical intervention, preferably by a special surgeon (plastic or paediatric surgeon). The lip should be operated upon early at 3 months while repair of the palate must wait until the baby is atleast 18 months and adequately nourished (10 kg).

Following palate repair, speech therapy must be started three to four weeks later and the parents should be encouraged to follow up with the treating team.

3.4 ANORECTAL MALFORMATIONS/ IMPERFORATE ANUS

Anorectal malformations or Imperforate anus is usually obvious especially among male newborns. However females have defects with a fistula commonly opening into the vestibule, or vagina from which they decompress stools and flatus and thus do not become obstructed. The defect may only be brought to the notice of the mother on closer examination later (Fig. 3.4 and 3.5).

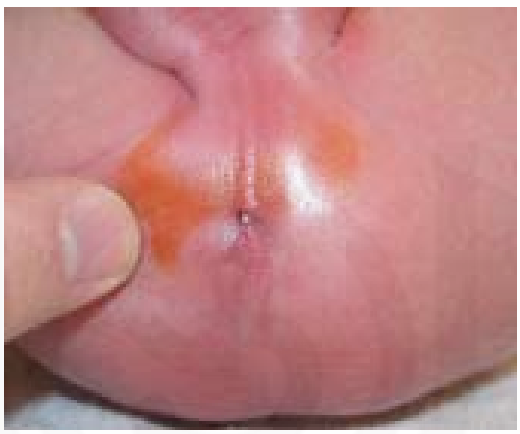

Fig. 3.4: Male with imperforate anus

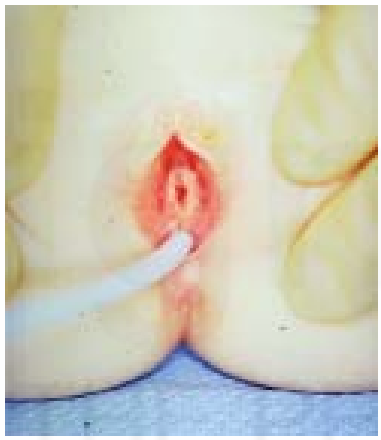

Fig. 3.5: Female with Vestibular opening

Male defects with obstruction require immediate referral for surgery, where a colostomy is usually performed to decompress the bowel. Female defects should be seen by a paediatric surgeon early so that their treatment plan can be made by the end of the first month. Definitive surgery can usually be performed after 3 months. After the new anal opening has been formed, it needs to be calibrated daily for the initial 3–6 months to prevent stenosis. The treating doctor usually would provide a dilator that must be used everyday after lubricating it with xylocaine jelly. The colostomy is generally closed 3 months after the anorectoplasty.

3.5 NEONATAL INTESTINAL OBSTRUCTION

Any newborn or infant who has bilious (yellow-green) vomiting is likely to have intestinal obstruction. The vomiting is after feeds as well as between feeds. The baby is often dehydrated, with the anterior fontanelle sunken. The abdomen may be minimally distended in cases with proximal obstruction, but could be massively distended when the obstruction is in the distal bowel.

The baby should be started on IV fluids (N/5 or N/4 normal saline with dextrose at a rate of 4 ml/kg/hour) and a nasogastric tube inserted (6F for a newborn). The baby should be placed under a radiant warmer while this is being done, and then wrapped well before referral to a hospital where pediatric surgical care is possible.

Delayed presentation or failure to recognise the condition may lead to sepsis and even perforation of the bowel proximal to the obstruction as seen in the picture below (Fig. 3.6). It will be very difficult to save such a delayed presenting baby.

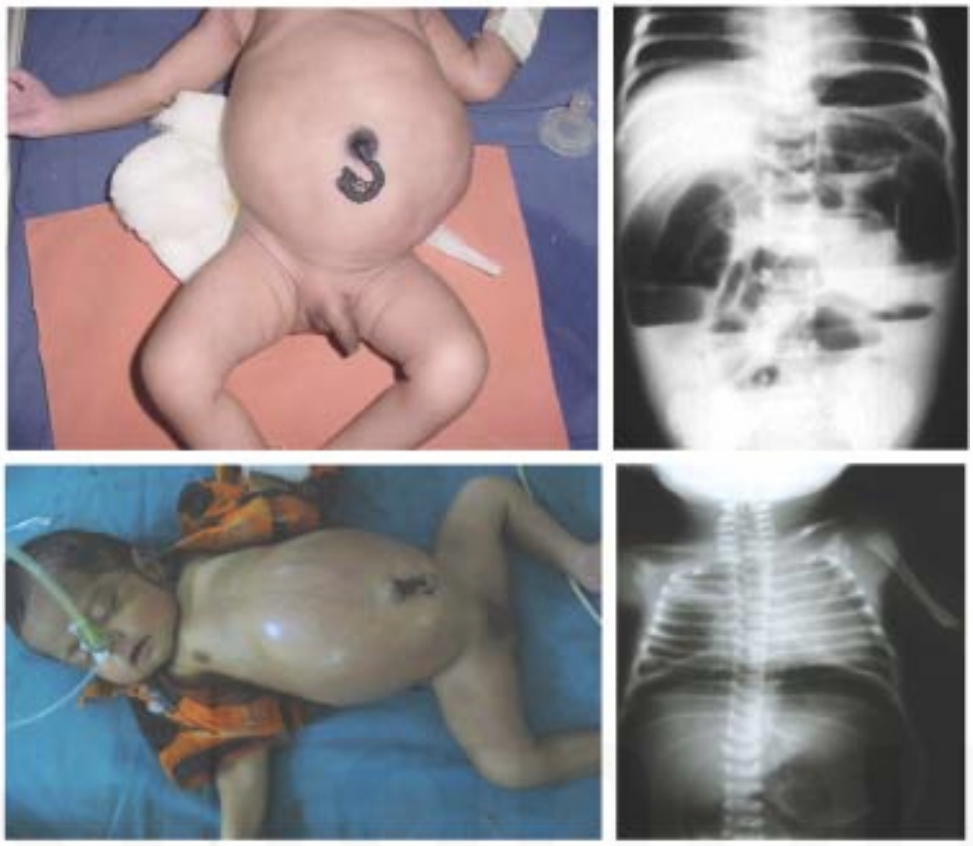

Fig. 3.6: Free air under diaphragm after perforation Late presentation with perforation and sepsis

Check Your Progress 1

1) What is the ideal age to operate on babies with Cleft lip and Cleft palate?

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2) If a male baby with absent anal opening has a bead of meconium at his urethral meatus, what does it indicate?

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3) What would you do if you see a newborn at day 2 of life with bilious vomiting, and mild upper abdominal distension?

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3.6 ESOPHAGEAL ATRESIA/TRACHEO ESOPHAGEAL FISTULA (TEF)

A newborn who has excessive salivation and frothing from the mouth is likely to have esophageal atresia. Attempts at feeding are likely to result in choking. When

presenting late, there may be features of pneumonia due to aspiration of gastric contents from the distal fistula. The diagnosis is confirmed by attempting to pass a No. 8 infant feeding tube through his oral cavity down his throat. The tube encounters resistance 7 cm from his lips and then coils back into the mouth. Suck out secretions/ saliva from the oral cavity as frequently as required. Keep the baby in a 30 degree head up position to prevent aspiration of refluxed gastric contents through the distal esophageal fistula. Keep the baby under a radiant warmer and start an I/V line and I/V fluids (10% dextrose @ 3 ml/kg/hr). Check the oxygen saturation if the baby is tachypneic or cyanosed and start oxygen therapy. The baby must never be fed. Refer the baby to a hospital with paediatric surgery facilities (Fig. 3.7).

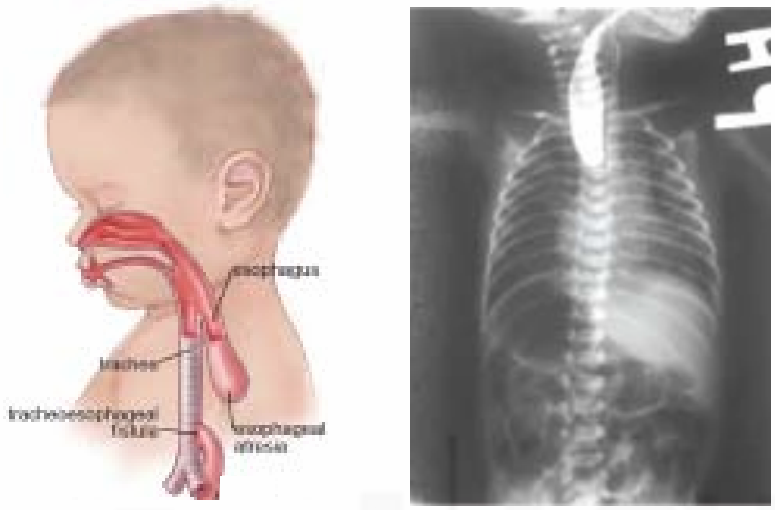


Fig. 3.7: Contrast study shows the blind ending esophagus
Typical Defect of Esophageal Atresia with Distal TEF

3.7 MENINGOCELE AND MENINGOMYELOCELE

Broadly known as spina bifida, these birth defects occur because of a failure of the posterior spinal elements to fuse, thereby exposing variable amounts of meninges and neural elements (cord and nerve roots) at the back. This can happen anywhere from cervical to sacral spine (Fig. 3.8 a and b). The defect is usually caused by a nutritional deficiency of Folic acid in the mother. Hence for future pregnancies, the mother and father should be advised to take Folic acid before the next baby is conceived, and to be continued into the end of first trimester.

As the neural elements may be exposed and dysplastic, there may be associated weakness (partial or complete) of the muscles of the lower limbs. Also, the anal canal opening may be patulous or loose with meconium or stools frequently released. Similarly there may be urinary incontinence. The defect is also associated with a malformation leading to hydrocephalus and thus a large head with a prominent anterior fontanelle.

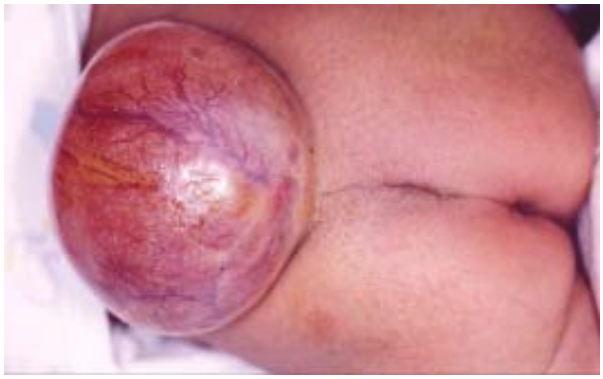


Fig. 3.8(a): Thin sac of Meningomyelocele

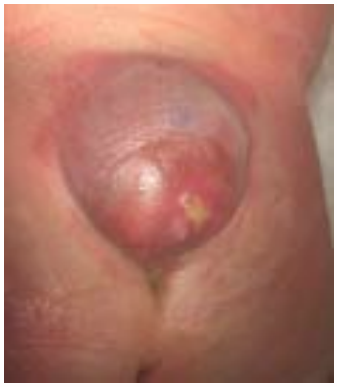


Fig. 3.8(b): Skin covered defect

3.8 HYDROCEPHALUS

This anomaly is caused by an obstruction to the flow of cerebrospinal fluid within and around the brain of a baby (Fig. 3.9). Among the common causes are aqueductal stenosis, Arnold Chiari Malformation and posterior fossa cysts. This obstruction leads to fluid accumulation and enlargement of the ventricular size in the brain, causing thinning of the surrounding developing brain parenchyma, as well as increase in size of the head. A normal term newborn has a head circumference of 35 cm at birth which increases to 40 cm by 3 months and 45 cm at 1 year. Thus serial head circumference is important to assess the rate of increase in the head size. When the intracranial pressure is high, it is reflected as

1) Widely open anterior fontanelle which is full or bulging
2) Separation of sutures, especially the squamoparietal suture above the ears
3) Sunset like appearance of the eyes
4) Prominent veins on the scalp
5) Increased tone and jerks of the lower limbs

Persistently raised pressure leads to optic atrophy and loss of vision along with thinning of the developing brain parenchyma causing developmental retardation.

The anomaly should be picked up early as surgical intervention may improve outcome. This may include placing ventriculo-peritoneal shunts or doing third ventriculostomy.

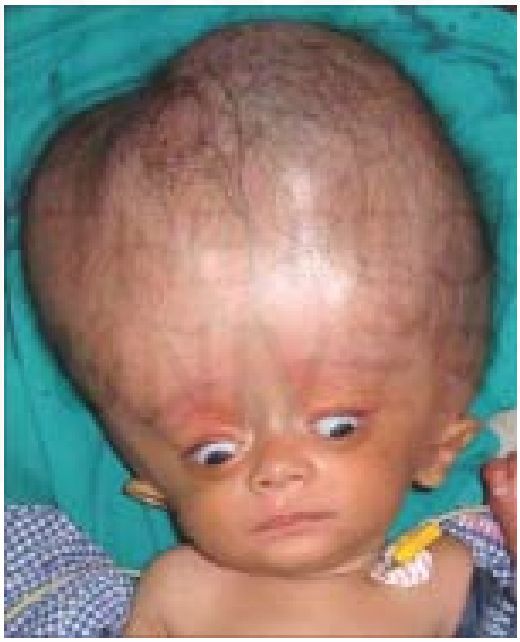


Fig. 3.9: Huge Head size with prominent veins and 'sunset' eyes

3.9 EXOMPHALOS MAJOR AND MINOR, GASTROSCHISIS

This is a defect in the formation of the anterior abdominal wall so that internal viscera protrude out of the abdominal cavity. The herniated contents are covered by a sac consisting of peritoneum and a jelly like substance that gradually dries up to form an eschar. The cord hangs from the summit of the sac. The important thing to remember when such a baby is delivered is not to cut the cord too close to the sac. The contents of the sac may include the liver, spleen, small and large bowel, as well as the stomach. Sometimes there may be no sac and the bowel is

lying exposed on the skin. The defect is usually to one side of the umbilicus and tight, so that the bowel and its vascularity may gradually be compromised (Fig. 3.10 and 3.11). The baby's exposed bowel or the sac must be cleaned with sterile normal saline and then wrapped in paraffin gauze to prevent drying or exposure. The baby should not be fed and a nasogastric tube (No. 6 F) should be placed and aspirated. I/V fluids must be started - 10% dextrose@ 3 ml/kg/hr on day 1 or N/5 dextrose saline @4 ml/ kg/hr on day 2 or later. The baby must be referred at the earliest to a paediatric surgery facility. Any Gastroschisis cancer exposes bowel due to lack of anterior abdominal wall.

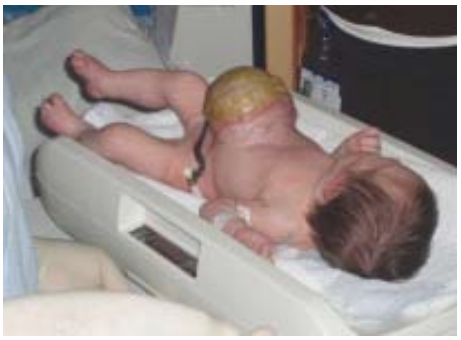


Fig. 3.10: Exomphalos major

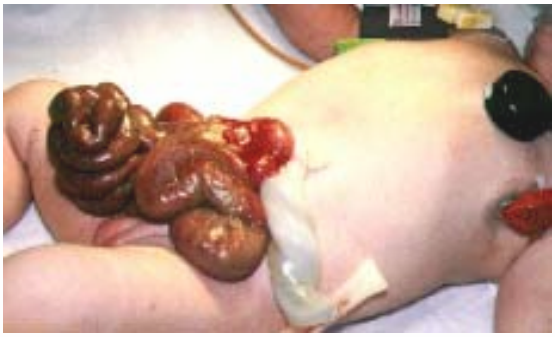


Fig. 3.11: Gastroschisis with dusky bowel

3.10 TALIPES EQUINUS/CLUB FOOT

A child may sometimes be born with shortening of the soft tissues of the flexor aspect of the leg and medial side of the foot, so that the foot is pointing downward (equinus), and inwards (varus), while his forefoot is adducted at the tarso-metatarsal joints (Fig. 3.12). This may happen in one or both feet. Such feet are called club feet. Most of them can be managed by early diagnosis and early proper manipulation.

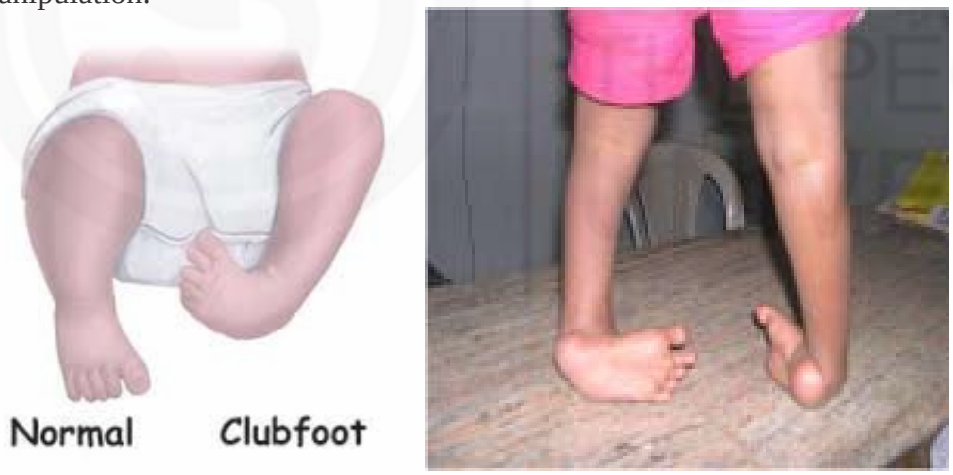


Fig. 3.12: Older child, abnormal gait

By the time they are 6 weeks, they require Ponsetti's technique of manipulation and repeated casts by a person trained to do this (Fig. 3.13). Such casts need to be revised every one to two weeks and subsequent advice after the final cast has been completed depends on the chances of the foot springing back to the original position. If these are significant the treating doctor advises braces to be worn as special shoes (Fig. 3.14). Special footwear is also provided which should be worn by the baby when she starts walking. Regular follow up visits to the treating doctor and physiotherapist must be encouraged, and use of braces or strapping as advised by them must be ensured.

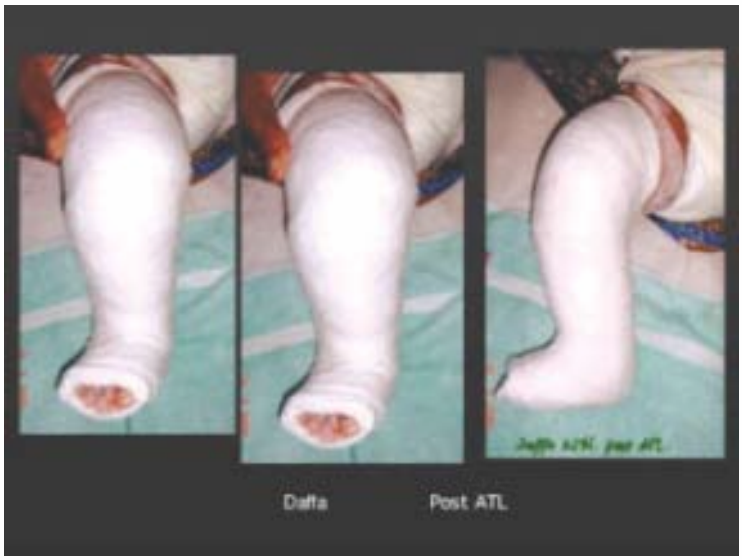


Fig. 3.13: Serial Ponsetti's casts

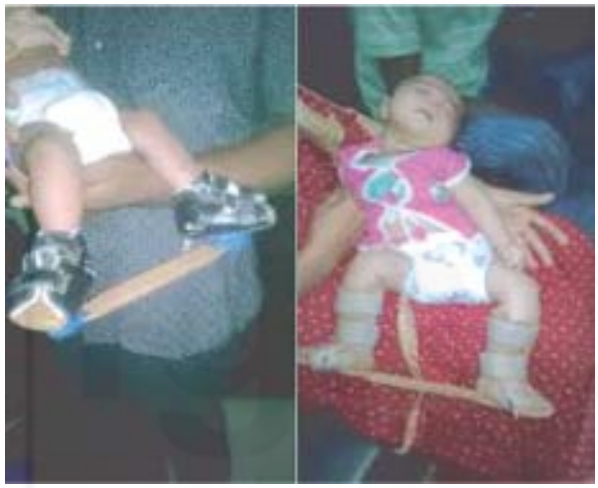


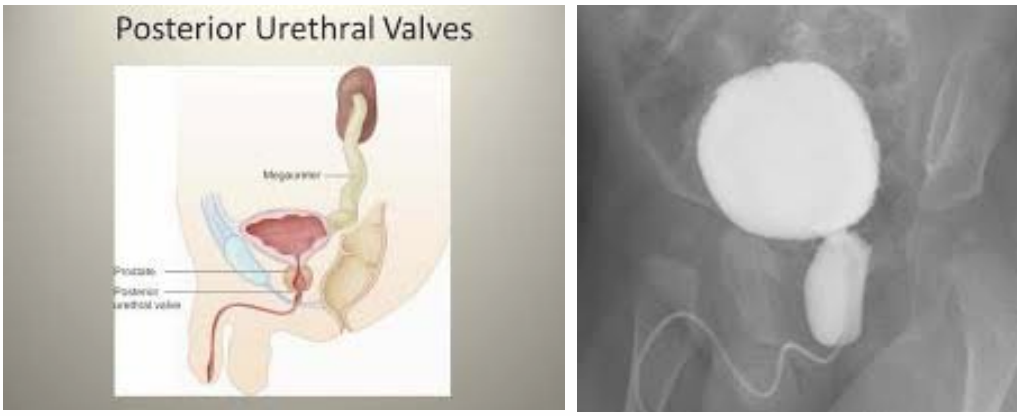
Fig. 3.14: Special shoes with splints worn after complete correction

Some children may require surgical correction which usually is done after one year age.

3.11 OBSTRUCTIVE MICTURITION IN NEWBORN PERIOD/INFANCY

The baby passes urine in a poor stream, often straining to pass few drops. There may be episodes of repeated urinary tract infection with fever, foul smelling and visibly turbid urine. The child may be thriving poorly and the abdomen may appear distended due to a partially full, thick walled bladder and often ballooned kidneys. The cause of obstruction in male babies is usually a birth defect where valves are present in the posterior urethra that obstructs the antegrade flow of urine. The baby must be referred early to a paediatric surgical facility as the kidneys get increasingly damaged with each episode of UTI.

Diagnosis is established by doing radiologic studies (Ultrasound examination and Micturating cystourethrogram) and urethroscopy. Treatment involves valve ablation using fulguration. Some babies may require temporary urinary diversion and then valve ablation. All of this requires a dedicated set up to handle these problems.



(A) (B)

Fig. 3.15: Obstructive micturation (Radiofogic Studis)

Hypospadias, Epispadias, Exstrophy bladder:

The position of the urethra may be abnormal due to developmental defects. When located on the ventral aspect, it is called Hypospadias (Fig. 3.16A). Whereas, abnormal opening on the dorsal surface of the phallus is called epispadias. In its extreme form, the entire urethra and anterior bladder wall may be incompletely closed and display on the surface of the lower abdomen. Such a defect is called an Exstrophy-epispadias complex (Fig. 3.16B). The ureters open on the posterior open bladder wall and keep leaking urine.

All these defects require surgical correction, often complex, and multistage. Appropriate referral is necessary.

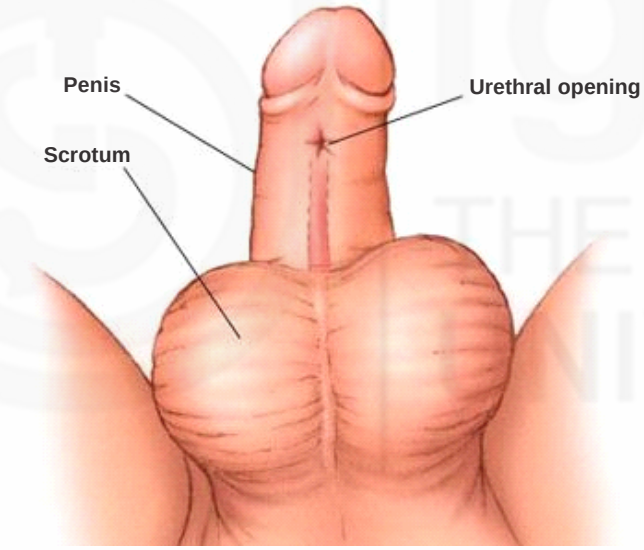


Fig. 3.16A: Hyporpadias

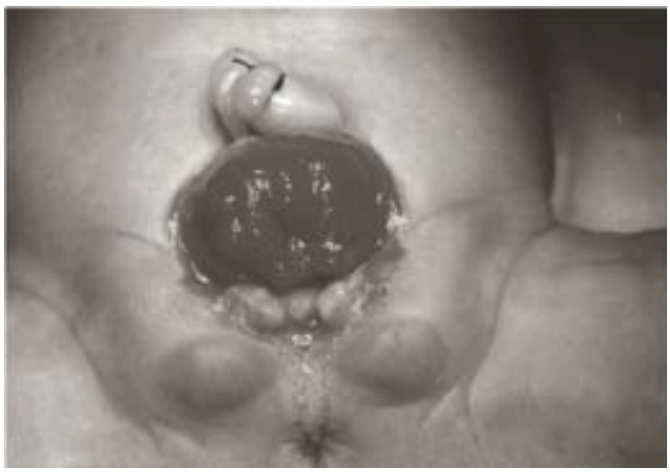


Fig. 3.16B: Extrophy Bladder

3.12 BLUE BABY (CYANOTIC CONGENITAL HEART DISEASE)

Congenital heart disease (CHD) refers to a defect in the structure of the heart which is present since birth. It is the most common type of birth defect. CHD can be of different types depending upon the type of defect and the symptoms vary with the type of disease. The cause of CHD is often unknown. It is often associated with maternal infections during pregnancy and genetic defects such as Down's syndrome. It is also associated with other malformations, which is called the VACTERL association.

V- Vertebral anomalies

A- Anal malformation

C- Cardiac abnormalities

T- Tracheo-esophageal fistula

E- Esophageal atresia

R- Renal anomalies

L- Limb anomalies

Symptoms of congenital heart disease may vary from none to severe, depending upon the type of birth defect. Common symptoms include

a) Rapid breathing

b) Tiredness on feeding

c) Cyanosis or bluish discolouration of the lips and nails (Fig. 3.17)

d) Poor weight gain

e) Cyanotic spells where the baby becomes deeply blue and unconscious after crying

Congenital heart defects are broadly divided into-

Acyanotic- which are not associated with cyanosis, e.g. ASD, VSD

Cyanotic- which are associated with cyanosis, e.g. TOF

The most common heart defects include:

1) VSD or ventricular septal defect which is a hole in the wall between the right and left ventricle.

2) ASD or atrial septal defect which is a hole in the wall between the two atria.

3) TOF or tetralogy of Fallot in which the baby appears cyanosed and may have cyanotic spells on crying.

Other heart defects include complex conditions like Transposition of Great Arteries (TGA), pulmonary atresia, tricuspid atresia etc.

Some of the heart defects are life threatening if not diagnosed and treated soon after birth whereas some may be picked when the child is much older.

A health worker should suspect presence of a heart defect if any of the above symptoms are present. On examination one may find a rapid heart rate and presence of a murmur on auscultation of the heart. Pulse oximetry

should be done. Presence of cyanosis which does not improve with oxygen suggests cyanotic heart disease. Such a child should be referred to a paediatrician for further evaluation and treatment. Newborns and infants may present with failure to thrive, poor feeding, recurrent lower respiratory tract infections and cyanosis. Auscultation over the precordium will often elicit a murmur.

Conclusive diagnosis requires a chest xray, ECG, Echocardiography and sometimes more sophisticated tests. These are available at large centres.

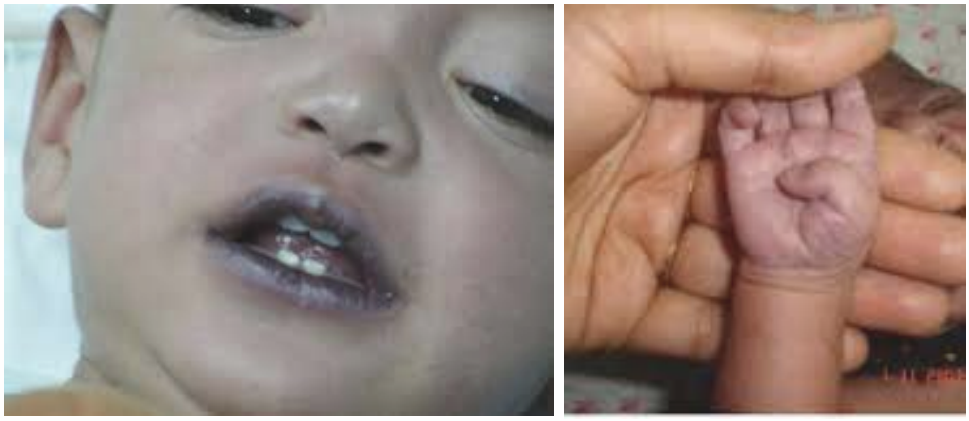


Fig. 3.17: Cyanosis of Lips and Nails

Check Your Progress 2

1) How would you position a one day old baby with frothing at the mouth?

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2) Deficiency of which vitamin can cause spina bifida?

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3) What is the head circumference of a term baby at birth, at 3 months and 12 months?

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4) How would you manage a newborn delivered at your sub-centre who has her bowel loops exposed and coming out of a defect in the abdominal wall?

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5) The common symptoms of congenital heart disease are____?

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3.13 IMPORTANT POINTS TO REMEMBER

1) Many congenital malformations (birth defects) are obvious and can be picked up by careful observation at birth or newborn period.
2) Babies with Cleft palate require special care with feeding as they may require a long spoon (Pallada's) to feed them.
3) Imperforate anus is apparent at birth; male defects require urgent attention. Female defects most often decompress well through a fistula in the perineum or vagina.
4) Bilious vomiting in a newborn is often indicative of some bowel pathology. They require further investigations to rule out bowel obstruction.
5) A newborn who is frothing saliva from the mouth is likely to have esophageal atresia. Failure to pass a stiff red rubber catheter 7–8 cm beyond the lips from the mouth almost confirms the diagnosis.
6) Meningo-myelocele is a serious birth defect caused by nutritional deficiency of folic acid.
7) Hydrocephalus manifests as large head in newborn and infancy. Head circumference must be measured when suspected.
8) Exomphalos major babies are likely to loose heat rapidly and become hypothermic due to the large exposed viscera.
9) CTEV or Club foot is easily correctible by early manipulation and later casts (Ponsetti's technique).
10) Poor urine stream, straining at micturition and frequent episodes of turbid urine with fever in a male baby, point towards obstructed urethra often due to Posterior Urethral Valves.
11) Early surgery for Bladder Exstrophy newborns makes the procedure simpler and helps improve outcomes.
12) Congenital heart disease may manifest with cyanosis in newborns or later as failure to thrive, recurrent lower respiratory tract infections.

3.14 LET US SUM UP

In this unit we have focused on congenital malformations such as cleft lip, cleft palate, anorectal malformations or imperforate anus, neonatal intestinal obstruction, esophageal atresia tracheo-esophageal fistula, meningocele and meningomyelocele, hydrocephalus, exomphalos major and minor, gastroschisis, talipes equinus or club foot, obstructive micturition in newborn period or infancy and blue baby.

3.15 MODEL ANSWERS

Check Your Progress 1

1) Cleft lip at 3 months and palate at 18 months.
2) It indicates a Recto-urethral fistula.
3) Consider Neonatal Intestinal Obstruction. Start IV fluids (Kidrolyte @ 4ml/kg/hr), pass a nasogastric tube and aspirate, keep the baby warm and transfer securely to a higher facility with possibility of surgical intervention.

Check Your Progress 2

1) Suspect Esophageal atresia. Position 30 degrees head up in supine position.

2) Folic acid.

3) It is 35 cm, 40 cm and 45 cm respectively.

4) How would you manage a newborn delivered at your sub-centre who has her bowel loops exposed and coming out of a defect in the abdominal wall?

A. Suspect Gastroschisis. Clean the loops gently with sterile saline, apply paraffin gauze and wrap the bowel in sterile dressing material. Keep the baby warm, start IV 10% Dextrose @ 3 ml/kg/hr and transfer to a higher facility.

5) a) Rapid breathing

   b) Tiredness on feeding

   c) Cyanosis or bluish discolouration of the lips and nails

   d) Poor weight gain

   e) Cyanotic spells where the baby becomes deeply blue and unconscious after crying