

[Newborn Care Manual: Contents](#)

### 29-1 WHAT IS A CONGENITAL ABNORMALITY?

A congenital abnormality (birth defect) is an abnormality in body structure or function that is present at birth. While most congenital abnormalities can be recognised at birth, unfortunately some internal abnormalities (e.g. of the heart) or functions (e.g. haemophilia) can sometimes only be diagnosed weeks or months after birth. About 3% of all infants have a congenital abnormality. These may be minor and not important, or serious enough to make the infant appear abnormal or to be the cause of the infant's death.

<b>ABOUT 3% OF INFANTS HAVE A CONGENITAL ABNORMALITY</b>
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Congenital abnormalities are discussed in detail in the Birth Defects manual of the Perinatal Education Programme.

### 29-2 WHAT ARE THE CAUSES OF CONGENITAL ABNORMALITIES?

There are many different causes of congenital abnormalities. The main causes are:

1. Chromosomal abnormalities, e.g. Down syndrome, in which there is an extra chromosome and Turner syndrome where there is a missing chromosome.
2. Gene abnormalities. These are often inherited from either one parent or both parents (e.g. autosomal dominant, autosomal recessive, X-linked recessive).
3. Teratogens. These are substances in the environment which can damage the fetus, e.g. alcohol (fetal alcohol syndrome) and rubella (German measles).
4. Multifactorial causes (interaction of an environmental and a genetic factor), e.g. neural tube defects.
5. Maternal diabetes. The high blood glucose concentration damages the fetus.
6. Compression of the fetus due to oligohydramnios.

Factors that may alter the intra-uterine environment, such as infections, teratogens and maternal diabetes, have a far greater chance of causing congenital abnormalities if they are present during the first trimester when the fetal organs are still forming.

Unfortunately the cause of many congenital abnormalities is not known.

*\*\*\* Congenital abnormalities may be due to failure of the normal development of one or more parts of the body in early pregnancy (malformation) or pressure on part of the body during later pregnancy (deformity). With autosomal dominant inheritance the risk is 50% while the risk is 25% with autosomal recessive inheritance. X-linked recessive inheritance affects males only with a risk of 50%.*

<b>ALL INFANTS SHOULD BE CAREFULLY EXAMINED AFTER DELIVERY FOR CONGENITAL ABNORMALITIES</b>
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### 29-3 WHEN SHOULD YOU ANTICIPATE A CONGENITAL ABNORMALITY?

1. If there is a family history of congenital abnormalities.
2. Maternal illness in the first trimester, e.g. rubella (German measles).
3. Maternal diabetes. With poorly controlled diabetes during the first trimester the risk of congenital abnormality in the fetus increases 10 times.
4. If the pregnant woman drinks excessive alcohol.
5. Maternal drugs in the first trimester, e.g. Warfarin or anticonvulsants.
6. Maternal age 35 years or above. In these older mothers the risk of Down syndrome is increased.
7. If there has been polyhydramnios or oligohydramnios, look for congenital abnormalities. With polyhydramnios think of oesophageal atresia or neural tube defects (the fetus does not swallow) while with oligohydramnios think of renal abnormalities (the fetus passes very little urine).
8. Persistent breech presentation.
9. Twins, especially if they are identical.

10. Underweight for gestational age infants, especially if no obvious maternal cause.

Many pregnant women are now being screened for major congenital abnormalities by having an ultrasound examination of the fetus and screening blood tests at 12-20 weeks.

#### **29-4 WHAT SHOULD YOU DO IF AN INFANT HAS EXTRA FINGERS?**

It is not uncommon for an infant to be born with extra fingers (or toes). Extra fingers are usually attached to the side of the hand with a narrow thread of skin. Often the mother or father also had extra fingers at birth. After getting the parents' consent, these extra fingers can be tied off with a piece of cotton or surgical silk. An assistant must gently pull the finger away from the hand so that you can tie the thread as close to the skin as possible.

*\*\*\* This is an example of autosomal dominant inheritance where a parent and infant have the same abnormality.*

Less commonly the extra finger or toe contains bone or cartilage. This is often associated with other major congenital abnormalities. These fingers or toes cannot be simply tied off and the infant must be referred to a level 2 or 3 hospital for further investigation. The fingers or toes will later be removed surgically.

#### **29-5 WHAT ARE CLUBBED FEET AND HOW SHOULD THEY BE MANAGED?**

Many infants have feet that are slightly twisted inward due to the position of the fetus. These feet are not abnormal as they can easily be turned into a normal position by gentle pressure.

Some infants have one or both feet which are twisted inward and cannot be turned into a normal position. These are clubbed feet. The cause may be familial or due to oligohydramnios (pressure on feet during pregnancy). Often the cause is unknown. These infants must be referred to an orthopaedic clinic within a few days of delivery, as early treatment with strapping or serial plaster of Paris casts can correct the abnormality. They may also need a minor operation later. Without correct treatment, clubbed feet result in permanent deformity and crippling.

#### **29-6 HOW SHOULD YOU DIAGNOSE AND TREAT DISLOCATED HIPS?**

At birth the upper end of an infant's femur (the femoral head) is normally in the hip joint and cannot be dislocated. However, occasionally one or both hips are dislocated or are dislocatable. If they are dislocated, the femoral head is not in the hip joint. If the hip is dislocatable then the femoral head can easily be moved out of the joint. The hips of all infants should be examined at birth (Barlow's test) to detect either a dislocated or dislocatable joint. If the early diagnosis is missed, the infant may start to walk late and will have an abnormal waddling gait. The surgical results are poor with late treatment.

Examination the hips is described in skills workshop 18 of this PEP manual.

If a hip is dislocated, then the infant must be referred to an orthopaedic clinic at a level 2 or 3 hospital for treatment within a few days of delivery. Once the clinical diagnosis is confirmed with an X-ray or by ultrasonography, the infant's legs should be placed in a plaster of Paris splint.

If the hip is only dislocatable, the infant should be examined again after 2 weeks. If the hip remains dislocatable, the infant must be referred as above. However most dislocatable hips return to normal within 2 weeks and need no further treatment.

<b>THE HIPS OF ALL INFANTS SHOULD BE EXAMINED AFTER BIRTH</b>
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#### **29-7 SHOULD AN UNDESCENDED TESTIS BE TREATED?**

By term, both testes should have descended normally into the scrotum. If a testis is not in the scrotum and cannot be gently pushed into the scrotum, then it is undescended. Many undescended testes will move into the scrotum spontaneously during the first 3 months. Thereafter, surgery is usually needed to bring down the undescended testis. The operation is usually done at about 1 year. With bilateral

undescended testes, an operation is important to reduce the risk of infertility. All undescended testes have an increased risk of malignancy in adulthood even if they were corrected in infancy.

### **29-8 WHAT IS HYPOSPADIAS AND HOW SHOULD IT BE MANAGED?**

Normally the urethral opening in a male infant is at the end of the penis. If the opening is on the underside of the penis or at the base of the scrotum, then the infant has hypospadias. These infants also have a curved rather than a straight penis and at birth appear to have been partially circumcised.

It is important to refer these infants to a urological clinic within a few weeks of birth. The hypospadias can be corrected surgically when the infant is a few months old. These infants must NOT be circumcised as the foreskin may be used to correct the urethra. It is important to reassure the parents that the abnormality can be corrected and that the infant's sexual function will be normal when he grows up.

### **29-9 WHAT ARE AMBIGUOUS GENITALIA?**

Ambiguous genitalia means that the external sex organs are not typically male or female. It is, therefore, difficult to decide on the sex of the infant. There are many causes of ambiguous genitalia. Some of these infants are male and others female. They should all be referred urgently to a level 3 hospital for investigation, as one of the common causes of ambiguous genitalia results in a lack of important adrenal hormones that may cause dangerous changes in the serum sodium and potassium concentrations. This can be fatal in the first few days of life if not correctly treated. It is also important to determine the correct sex of the infant and to tell the parents as soon as possible whether the infant should be brought up as a boy or girl. This may be a very difficult decision and take some time. These infants will need corrective surgery later during childhood.

### **29-10 WHAT IS AN INGUINAL HERNIA AND HOW SHOULD IT BE MANAGED?**

Normally the inguinal canal closes after the testes have descended into the scrotum at about 36 weeks of gestation. However, if the canal does not close normally, bowel will herniate into the scrotum resulting in an inguinal hernia. This presents as an oval shaped mass in one or both sides of the scrotum. The mass may be firm or soft, often changes in size as the bowel moves in and out of the scrotum, and usually becomes bigger when the infant cries. Peristalsis may be felt in the hernia. The hernia does not transilluminate. Inguinal hernias are very common in infants who were born preterm.

The danger of an inguinal hernia is that the bowel may become trapped (incarcerated) in the scrotum. This will cut off the blood supply to that portion of the gut resulting in bowel obstruction, death of the bowel wall (gangrene) and perforation. A trapped hernia presents as a hard, red, tender and tense mass in the scrotum. The abdomen may also become distended and the infant may vomit repeatedly. This is a surgical emergency and requires urgent referral.

To prevent this complication, inguinal hernias should be repaired when the infant is well enough to have a general anaesthetic and weighs more than 2500 g. Usually inguinal hernias are repaired before the infant is discharged home.

\*\*\* *Torsion of a testis can also present as a red, tender and swollen scrotum.*

### **29-11 WHAT IS A FLUID HERNIA?**

This is an inguinal hernia where the opening from the abdominal cavity into the scrotum is only big enough to allow through fluid but not bowel. It also presents as a one sided or bilateral scrotal swelling. The scrotum transilluminates very well, and this sign is used to differentiate between inguinal and fluid hernias.

Most fluid hernias (hydrocoeles) disappear after a few months and need no treatment. However, some fluid hernias, especially if they are very big, do not disappear and require surgical correction at about 3 months.

**29-12 WHAT IS A BIRTH MARK?**

A birth mark (a naevus) is a mark on the skin caused by increased pigment or an abnormal collection of blood vessels. There are 3 important types of birth mark:

1. About 10% of infants have 1 or more raised, red marks on the skin which appear in the first few weeks of life. They are never present at birth. These are known as "strawberry marks" and are formed by an abnormal collection of large veins. They become bigger for a few months then gradually fade in 1-5 years. Unless they become very big they usually do not need any treatment.
2. Far less commonly, infants are born with a large pink mark, usually on the face. This is known as a "port wine stain" and is formed by an abnormal collection of small veins. These marks are always present at birth and do not fade. They become worse with age. The pigmented area can be covered with cosmetic cream. Laser treatment can remove the mark.
3. Some infants are born with a large dark brown birth mark, often on the back. It is due to excessive pigment cells and, therefore, does not change colour when pressed. It becomes more marked with age and may become hairy. When these children are older the area of affected skin should be removed by a plastic surgeon as this birth mark can become malignant in adulthood.

**29-13 IS IT ABNORMAL FOR AN INFANT TO HAVE ONLY ONE UMBILICAL ARTERY?**

Yes. Most infants have 2 umbilical arteries and 1 umbilical vein. If the infant has a single umbilical artery, then there is a much higher than normal chance that the infant also has other congenital abnormalities. These infants, therefore, must be carefully examined after delivery.

**IF YOU FIND ONE ABNORMALITY, ALWAYS LOOK FOR OTHER ABNORMALITIES**

**29-14 WHAT IS THE MANAGEMENT OF A CLEFT LIP?**

A cleft lip may occur alone or together with a cleft palate. These infants look very abnormal and therefore the parents must be reassured that the abnormality can be repaired. Infants with a cleft lip must be referred to a plastic surgery clinic at a level 2 or 3 hospital. The lip is usually repaired at about 3 months. These infants usually feed and gain weight well.

**29-15 WHAT IS THE MANAGEMENT OF A CLEFT PALATE?**

This may be on one or both sides and is usually seen together with a cleft lip. The infant may need cup or nasogastric feeds and must be referred within a day or 2 to a plastic surgery clinic at a level 3 hospital. A plastic plate may be fitted against the palate to help correct the position of the gums and the sides of the palate. With a plate, most infants will be able to feed well. The cleft lip is usually repaired at 3 months but the cleft palate is repaired later, possibly after a few years. The infant will need to have the plate replaced a number of times as the palate grows.

**29-16 WHAT IS THE PRESENTATION AND EMERGENCY MANAGEMENT OF OESOPHAGEAL ATRESIA?**

Oesophageal atresia is an obstruction of the oesophagus. It is usually associated with a connection (fistula) between the lower oesophagus and the bronchi of the lungs. Polyhydramnios is almost always present during pregnancy and may be severe. After birth these infants cannot swallow as the oesophagus ends in a blind pouch. They dribble saliva. Feeds cause choking, cyanosis and collapse as the feed cannot be swallowed and is inhaled into the lungs. Gastric acid passes from the stomach into the bronchi, via a fistula, especially when the infants lie down. Both inhaled feeds and the reflux of gastric acid result in respiratory distress.

Do NOT feed any infant that you suspect of having an oesophageal atresia. The diagnosis is confirmed by the inability to pass a nasogastric tube. Any aspirate will test alkaline with litmus paper as the tube is not in the stomach. Whenever polyhydramnios is diagnosed, a nasogastric tube must be passed at birth to exclude oesophageal atresia BEFORE the first feed is given.

Infants with oesophageal atresia must be nursed head up, they must not be fed and the mouth should be repeatedly suctioned. They must be urgently referred to a level 3 hospital and this emergency treatment should be continued during the transfer. As the infant is kept nil per mouth, an intravenous infusion of maintenance fluid (e.g. Neonatalyte) may be needed.

*\*\*\* Barium must not be injected down the oesophagus in an attempt to confirm the diagnosis as it may enter and damage the lungs. A straight antero-posterior chest X-ray usually shows the air filled upper oesophageal pouch which contains the coiled up nasogastric tube.*

## POLYHYDRAMNIOS ALWAYS SUGGESTS OESOPHAGEAL ATRESIA

### **29-17 HOW DO YOU DIAGNOSE AND MANAGE DUODENAL ATRESIA?**

Duodenal atresia is an obstruction of the duodenum. Polyhydramnios may have been present. The liquor may also be bile stained due to the fetus vomiting. The infant may have Down syndrome. Soon after delivery the infant starts vomiting. The vomitus is often bile stained. The diagnosis is easily confirmed by an abdominal X-ray that shows 2 bubbles of air only in the bowel. These infants must be kept nil per mouth, the stomach should be emptied via a nasogastric tube, and they should be referred urgently to a level 3 hospital for surgery.

Other forms of small bowel obstruction may present in a similar way.

### **29-18 WHAT SHOULD YOU DO IF NO ANUS IS PRESENT?**

It is important to examine all newborn infants to make sure that an anus is present. The anus may simply be covered with skin or the absent anus may indicate a major abnormality of the large bowel. Some of these infants can pass meconium via a fistula into the vagina or bladder, but soon they develop abdominal distension due to bowel obstruction. They should be kept nil per mouth and referred urgently to a level 3 hospital for investigation. A covered anus can be corrected with a simple operation. Major defects of the large bowel require a colostomy followed later by complicated surgical correction.

### **29-19 WHAT IS EXOMPHALOS AND HOW SHOULD IT BE MANAGED?**

An infant with exomphalos (omphalocele) has no abdominal wall muscle around the base of the umbilical cord. The normal abdominal wall is replaced by a thin membrane through which the bowel may be seen. In a large exomphalos the bowel bulges into the umbilical cord. The covering membrane may burst at delivery. After delivery the cord should be clamped well away from the exomphalos. The abnormality should be covered with sterile gauze. Whether the exomphalos is big or small, all these infants must be transferred urgently to a level 2 or 3 hospital for management. Infants with exomphalos often have other major abnormalities. An exomphalos is not the same as an umbilical hernia which is covered with skin and does not need to be treated.

*\*\*\* A gastroschisis is similar to an omphalocele but the defect in the abdominal wall is not central but to the side of the umbilical cord. Loops of bowel are not covered by a membrane and fall out of the gastroschisis. The bowel is usually abnormal. Other congenital abnormalities are uncommon. Urgent surgery is needed.*

### **29-20 WHAT CLINICAL SIGNS WOULD SUGGEST A CONGENITAL HEART ABNORMALITY?**

1. Central cyanosis, especially if there is little or no respiratory distress and the cyanosis is not corrected by 100% oxygen.
2. A heart murmur.
3. Absent femoral pulses.
4. Signs of heart failure: hepatomegaly, excessive weight gain, oedema, respiratory distress.

There are many different types of congenital heart abnormality. Any infant with any of the above signs should be urgently referred to a level 2 or 3 hospital for further investigation.

**29-21 WHAT IS ANENCEPHALY?**

In these infants the top of the skull is absent, exposing a poorly formed brain. They all die in a few hours or days. These infants should be kept warm and comfortable in the nursery until they die. They can be fed if necessary.

**29-22 WHAT IS A MENINGOMYELOCOELE AND HOW IS IT MANAGED?**

A meningomyelocele is a major abnormality of the spine, usually in the lumbar area. A flat area of the spinal cord is exposed on the skin. Sometimes a thin walled sac is also present and this may rupture with delivery. The legs are usually paralysed and hydrocephalus is common. The infants also dribble urine due to a paralysed bladder. Polyhydramnios is common with anencephaly or meningomyelocele.

The meningomyelocele should be covered with a piece of sterile gauze and the infant referred urgently to a level 3 hospital for possible closure of the area. Many of these infants die and most of the survivors have major orthopaedic and urological problems. They often also have other major abnormalities.

Most cases of anencephaly and meningomyelocele (also called neural tube defects) can be prevented if the mother takes 0,5 mg folic acid daily for a few weeks before and after falling pregnant. This is very important in women who have previously had a child with either anencephaly or meningomyelocele.

Neural tube defects are discussed in detail in the Birth Defects manual of the Perinatal Education Programme.

**29-23 WHAT IS HYDROCEPHALUS?**

Hydrocephalus is an excessive amount of cerebrospinal fluid in the ventricles of the brain. Hydrocephalus may be mild or severe and has many causes. The prognosis depends on the cause rather than the severity. Marked hydrocephalus should be operated on (shunted) to relieve the pressure in the brain. All infants with hydrocephalus must be referred to a level 3 hospital for further investigation.

Ultrasonography during pregnancy can diagnose hydrocephalus, anencephaly and meningomyelocele.

**29-24 WHAT IS DOWN SYNDROME?**

Down syndrome is caused by an extra number 21 chromosome (trisomy 21) and presents at birth with a number of recognizable signs:

1. A typical flat face with downward slanting eyes and a wide nasal bridge.
2. The head is round and the back of the head (occiput) is flat.
3. The tongue appears big and frequently sticks out.
4. The ears are small.
5. The hands are short and wide, often with a single crease on the palm. A single palmer crease is, however, not uncommon in normal infants.
6. The feet are also short and wide, often with a gap between the big and second toe (a sandal gap).
7. The infant is floppy (hypotonic) when handled.
8. The infant feeds poorly.

Infants with Down syndrome often have major congenital abnormalities, especially heart defects and duodenal atresia. They are all mentally retarded and, therefore, develop slowly.

The diagnosis must be confirmed by a genetics laboratory where the number of chromosomes in white cells, obtained from a sample of blood, will be counted.

Down syndrome is discussed in detail in the Birth Defects manual of the Perinatal Education Programme.

**29-25 CAN DOWN SYNDROME BE PREVENTED?**

The risk of Down syndrome in the general population is about 1 in 600. However the risk increases to about 1 in 200 for mothers at 35 years and 1 in 100 at 40 years. The older the mother the higher is the risk. All pregnant women of 35 years or more should, therefore, be offered an amniocentesis at 16 weeks of pregnancy. Chromosome analysis on the cells of the liquor will diagnose Down syndrome. A termination of pregnancy can then be offered to the parents. Ultrasonography and a blood test early in pregnancy can identify many but not all women at high risk of having a fetus with Down syndrome.

*\*\*\* In South Africa a termination of pregnancy is legal if there is a substantial risk of severe damage to the fetus. Terminations should not be done after 24 weeks as the infant may be viable.*

**29-26 HOW SHOULD YOU MANAGE AN INFANT WITH DOWN SYNDROME?**

It is important to make the diagnosis and tell the parents as soon as possible after birth. The parents should be told what it means to have Down syndrome. These infants must carefully examined for signs of major congenital abnormalities, especially heart abnormalities and duodenal atresia. If these are present, they must be referred to a level 3 hospital for further investigation. Infants with Down syndrome must be followed up to monitor their development. If possible the parents should be put in contact with other families with a Down syndrome infant. The Down Syndrome Association or other groups of parents of Down syndrome infants are very helpful. With a caring, stimulating home many children with Down syndrome are progressing far better than before.

**29-27 WHAT IS THE FETAL ALCOHOL SYNDROME?**

Infants with this syndrome have been damaged by excessive alcohol intake by the mother during pregnancy. They have typical faces with a long, smooth upper lip. The eyes appear small due to a narrow palpebral fissure (opening between the eye lids). In addition, they are growth retarded with small heads and are often born preterm. Many also have abnormalities of the heart or limbs. They remain small for their age after birth and are mentally retarded. All pregnant women should be advised not to drink alcohol at all.

**PREGNANT WOMEN SHOULD NOT DRINK ALCOHOL**

Fetal alcohol syndrome is discussed in detail in the Birth Defects manual of the Perinatal Education Programme.

**29-28 HOW SHOULD YOU MANAGE PARENTS OF AN INFANT WITH CONGENITAL ABNORMALITIES?**

When telling parents that their infant has a congenital abnormality, there are a number of important points to remember:

1. If possible speak to the parents together.
2. The sooner they are told of the abnormality the better.
3. Always be honest with parents, although all the details of the abnormality and the full implications of the prognosis need not be told immediately. Do not try to give all the details at once.
4. Be kind and tell the parents that you care.
5. Be understanding. Parents are often angry with the staff and family when told that their infant is not normal.
6. Be patient, as parents often need to be told again and again. Explain the problem in simple, easy to understand language. If needed, get an interpreter to help you. Shocked parents often forget what they have been told.
7. Do not make the parents feel that it is their fault that the infant is abnormal. Many parents of an abnormal infant feel very guilty.
8. Allow the parents to see and hold their infant. Point out the normal as well as the abnormal parts of the infant. By the way you handle the infant, indicate that you accept the infant and do not reject the infant as a "monster".
9. If possible, try to be optimistic and encouraging about the prognosis.

10. Allow the parents to speak and ask questions.
11. Speak about the risk of an abnormal infant in following pregnancies.
12. Give details of the future management of the infant.
13. Always keep the infant comfortable in the nursery even if the infant is going to die. Never let parents feel that the staff have abandoned their infant.
14. Consent for operation may be needed.

Counselling parents of a child with a congenital abnormality is discussed in detail in the Birth Defects manual of the Perinatal Education Programme.

## **CASE PROBLEMS**

### **CASE 1**

A patient delivers an infant at term after a pregnancy complicated by polyhydramnios. The infant appears normal but dribbles a lot of saliva.

**1. What should you suspect if the mother has polyhydramnios during pregnancy?**

The infant may have a congenital abnormality, especially oesophageal atresia, anencephaly or meningomyelocele.

**2. What congenital abnormality may present with excess saliva and dribbling.**

Oesophageal atresia. These infants cannot swallow because the oesophagus ends in a blind pouch.

**3. How can you confirm the diagnosis of oesophageal atresia?**

Attempt to pass a nasogastric tube. Usually the tube will curl back into the mouth if oesophageal atresia is present. The aspirate will be alkaline when tested with litmus paper as the nasogastric tube is not in the stomach. A chest X-ray will usually show the air filled, blind oesophageal pouch containing the coiled up feeding tube.

**4. What is the emergency treatment of this infant?**

Keep the infant's head raised to prevent gastric acid refluxing up the fistula into the lungs, do not feed the infant by mouth, keep the mouth well suctioned to prevent the saliva being inhaled into the lungs.

**5. What further management is needed?**

The infant must be transferred to a level 3 hospital for surgical correction of the abnormality. The emergency treatment must be continued while the infant is being transported. As the infant is kept nil per mouth, an intravenous infusion may be needed. Speak to the parents and obtain consent for operation.

### **CASE 2**

An unbooked patient of 42 years old delivers an unusual looking infant which is very floppy. The hands and feet appear wider than usual. The infant's tongue appears large and cyanosed. After the second feed the infant vomits green fluid.

**1. What is the probable diagnosis?**

Down syndrome. These infants typically have an abnormal looking face, broad hands and feet, and hypotonia.

**2. How is this diagnosis confirmed?**

Phone the nearest genetics laboratory and arrange for a sample of blood to be sent to them for chromosome analysis.

**3. What chromosomal abnormality will confirm the clinical diagnosis of Down syndrome?**

An extra chromosome 21.

**4. Why was this mother at high risk of delivering an infant with Down syndrome?**

Because she is 42 years old (older than 35). She should have booked early and been offered an amniocentesis.

**5. What associated congenital abnormalities does this infant probably have?**

A congenital heart abnormality, as suggested by the central cyanosis, and duodenal atresia, as suggested by the bile stained vomit. Both these abnormalities are common in infants with Down syndrome.

**6. What long term supportive care should be offered to these parents?**

They should be referred to the Down Syndrome Association or similar group in your area.

**CASE 3**

An infant is born at term with an abnormal foot that is twisted inward and cannot be turned back into a normal position. On careful examination it is noticed that the infant also has a swelling in the left side of the scrotum.

**1. What is wrong with this infant's foot?**

The infant has a clubbed foot. A foot that is simply twisted inward, due to the position before delivery, is easily turned back into a normal position.

**2. What management is needed to correct this foot?**

The infant should be referred as soon as possible to an orthopaedic clinic where the foot will be placed in serial plaster of Paris casts until it is straight.

**3. What are the 2 common causes of swelling of the scrotum in a newborn infant?**

Inguinal hernia and fluid hernia (hydrocoele).

**4. How can you differentiate between these conditions?**

A fluid hernia transilluminates very well as it contains clear fluid while an inguinal hernia does not transilluminate because it contains bowel.

**5. What is the correct management of this infant's inguinal hernia?**

It should be surgically corrected within the first few days if the infant is well. A delay in surgery may result in the bowel becoming trapped with resultant gangrene.