

Introduction to congenital disorders

What is a congenital disorder?

A congenital disorder (also known as birth defect or congenital abnormality) is an abnormality in body structure or function that is present at birth. While most structural (physical) congenital disorders can be recognised at birth, unfortunately some internal abnormalities (e.g. of the heart) or functions (e.g. haemophilia, single gene defects and other metabolic disorders) can sometimes only be diagnosed weeks or months after birth. About 3% of all infants have a congenital disorder. 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.

About 3% of infants have a congenital disorder.

NOTE

Intrauterine infections which do not cause structural defects (e.g. congenital syphilis) and recent acute insults (e.g. intrapartum hypoxia) are usually not regarded as congenital disorders.

What are the causes of congenital disorders?

There are many different causes of congenital disorders. 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 (single gene defects). These are often inherited from either one parent or both parents (e.g. autosomal dominant, autosomal recessive and 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 structural congenital disorders if they are present during the first trimester when the fetal organs are still forming.

Unfortunately the cause of many congenital disorders is not known.

NOTE

Congenital disorders 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 of a congenital disorder is 50% while the risk is 25% with autosomal recessive inheritance. X-linked recessive inheritance affects males only with a risk of 50%.

All infants should be carefully examined after delivery for congenital disorders.

When should you anticipate a congenital disorder?

1. If there is a family history of a congenital disorder.
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 structural defects 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 structural congenital disorders. 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 structural congenital disorders by having an ultrasound examination of the fetus and screening blood tests at 12–20 weeks.

Common structural congenital disorders

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. Less commonly the extra finger or toe contains bone or cartilage. This is often associated with other major congenital abnormalities.

NOTE

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

Management: Tying off of extra digits is no longer recommended. Surgical removal once the baby is older may be indicated. Contact your referral hospital for guidance on further management.

What are clubbed feet and how should they be managed?

Many infants have feet that are slightly twisted inward due to the position the fetus lies in during pregnancy. 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 serial plaster of Paris casts can correct the abnormality. They may also need a minor operation later. The result of treatment is good and these children can walk normally. Without correct treatment, clubbed feet result in permanent deformity and crippling.

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 pushed out (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.

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. With the correct, early treatment most children with a dislocated hip will walk normally although arthritis in adult life is common.

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.

The hips of all infants should be examined after birth.

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 earlier 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.

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 needed 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.

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 hypoglycaemia and 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 may take some time. These infants will need corrective surgery later during childhood.

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 push (herniated) 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.

NOTE

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

What is a birth mark?

A birth mark (a naevus) is a mark on the skin at or soon after birth 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 to 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.

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, there is a much higher than normal chance that the infant also has other structural congenital disorders. These infants, therefore, must be carefully examined after delivery.

If you find one structural congenital disorder, always look for another.

Serious structural congenital disorders

What is the management of a cleft lip?

A cleft lip may occur alone or together with a cleft palate. Infants with a cleft lip look very abnormal and therefore the parents must be reassured that the cleft can be repaired. They 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. It is very helpful to show the parents a photo of a child with a repaired cleft lip.

What is the management of a cleft palate?

This may be on one or both sides of the mouth and is usually seen together with a cleft lip. These infants have difficulty sucking. They must be referred within a day or 2 to a plastic surgery clinic at a level 3 hospital. Sometimes a plastic plate is fitted against the palate to help correct the position of the gums and the sides of the palate. A plate also makes feeding easier. Some infants with a cleft palate breastfeed well. Otherwise cup feeding or bottle feeding with a large hole in the teat helps. The cleft lip is usually repaired at 3 months but the cleft palate is repaired later, possibly after a few years. Speech and hearing problems are common. A multidisciplinary team at a combined assessment clinic is needed for the best results (plastic surgeon, dentist, audiologist and paediatrician).

What is the presentation and emergency management of oesophageal atresia?

Oesophageal atresia is an obstruction of the oesophagus due to a section of the oesophagus which is missing. 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 as the fetus cannot swallow. After birth these infants also cannot swallow as the oesophagus ends in a blind pouch. They dribble saliva. Feeds cause choking, cyanosis and collapse as the feed, which cannot be swallowed, 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 to prevent acid reflux, 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.

NOTE

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.

How do you diagnose and manage duodenal atresia?

Duodenal atresia is an obstruction of the duodenum. Polyhydramnios may have been present and the amniotic fluid 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.

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.

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 birth the cord should be clamped well away from the exomphalos. The abnormality should be covered with sterile gauze or plastic wrapping. 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.

NOTE

A gastroschisis is similar to an exomphalos 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 as it is exposed to the amniotic fluid during pregnancy. Other congenital disorders are uncommon. Urgent surgery is needed.

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.

Major neurological defects

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.

What is a meningomyelocele 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 sterile occlusive dressing 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.

Can these neurological defects be prevented?

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. Maize meal and wheat flour should be fortified with folic acid. This is very important in women who have previously had a child with either anencephaly or meningomyelocele as both these congenital disorders are more common in some families.

Folic acid supplements reduce the risk of major neural defects.

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.

Important syndromes

What is a syndrome?

This is a collection of abnormalities that form a clinical pattern which can be recognised. Therefore all children with the same syndrome look alike. Most experienced doctors and nurses can recognise an infant with Down syndrome or fetal alcohol syndrome soon after birth.

What is Down syndrome?

Down syndrome is caused by an extra number 21 chromosome (trisomy 21) and presents at birth with a number of recognisable 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 palmar crease is, however, not uncommon in normal infants.
6. The feet are also short and wide, often with a wide 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 abnormalities, especially heart defects and duodenal atresia. They are all mentally retarded and, therefore, develop slowly.

The diagnosis must always be confirmed by a genetics laboratory where the extra chromosome 21 in white cells, obtained from a sample of blood, can be identified.

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. Ideally all pregnant women, especially women of 35 years or more, should be screened. An amniocentesis at 16 weeks of pregnancy should be offered to women who are identified at high risk with the screening tests.

Chromosome analysis on the cells of the amniotic fluid 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 most women at high risk of having a fetus with Down syndrome.

NOTE

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.

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 be 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.

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 eyelids). 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. Unfortunately fetal alcohol syndrome is common in South Africa.

Pregnant women should not drink alcohol.

Managing parents of infants with a congenital disorder

14-30 How should you manage parents of an infant with a congenital disorder?

When telling parents that their infant has a congenital disorder, 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.