

Birth defects - digestive tract

The digestive tract begins at the mouth and ends at the anus. The oesophagus is the muscular tube that connects the back of the mouth to the stomach. Waste (faeces) is temporarily stored in the rectum before being passed out of the body through the anus. During fetal development, the digestive tract may fail to develop properly. Birth defects of the digestive tract include oesophageal atresia (obstruction of the oesophagus) and imperforate anus (malformations of the anus). These defects may occur together. Most babies born with oesophageal atresia also have tracheo-oesophageal fistula, which means the trachea (windpipe) leading to the lungs is connected to the oesophagus. The causes of these malformations are unknown, so prevention is not possible. Too much amniotic fluid surrounding the baby during pregnancy (polyhydramnios) may indicate the presence of these defects.

Oesophageal atresia

Oesophageal atresia is a group of malformations that block the oesophagus. Instead of attaching to the stomach, the oesophagus may end in a closed sac, or join with the trachea (windpipe) leading to the lungs. Around one third of affected babies will also have other birth defects, including congenital heart disorders and imperforate anus. Symptoms of oesophageal atresia include excessive dribbling, the inability to feed properly and vomiting. The incidence of oesophageal atresia or stenosis (abnormally narrow oesophagus) in Victoria is around one in every 1,800 births.

Treatment for oesophageal atresia

Diagnosis methods include threading a slender feeding tube down the baby's oesophagus to see if the stomach can be reached, and the use of x-rays. Oesophageal atresia requires immediate surgery. (If the baby also has a tracheo-oesophageal fistula, this will also need prompt repair. Preferably, the two operations are done at the same time.) Oesophageal atresia is repaired by sewing together the two halves of the tube. If the gap is too large to bridge, the baby may have to be tube fed for some time until the gap lessens. A prolonged hospital stay follows surgery.

Long term difficulties for the child

In the majority of cases, the child will experience feeding difficulties, even if the operation is deemed successful. These difficulties can include:

- Peristalsis (the muscular motions of the oesophagus that massage food down to the stomach) is not as coordinated as normal.
- There may be problems with swallowing.
- Swallowed food may occasionally lodge in the oesophagus instead of continuing into the stomach.
- The child is susceptible to reflux or heartburn.

Tracheo-oesophageal fistula

Most babies born with oesophageal atresia also have tracheo-oesophageal fistula, which means the oesophagus is connected to the trachea. This allows swallowed food into the lungs and air into the oesophagus. Symptoms include coughing and choking when feeding, and a blue tinge to the skin due to lack of oxygen (cyanosis).

Treatment for tracheo-oesophageal fistula

An x-ray will reveal air inside the oesophagus. Tracheo-oesophageal fistula requires immediate surgery. The fistula is sewn closed, generally during the same operation to repair the oesophageal atresia.

Long term difficulties for the child

Long term problems of a repaired tracheo-oesophageal fistula can include:

- A characteristic cough, caused by the flaccidity of the trachea.
- Increased susceptibility to respiratory infections.
- The trachea may temporarily collapse during episodes of hard breathing, such as crying, leading to noisy breathing and (sometimes) respiratory difficulties.

Imperforate anus

Instead of the rectum attaching normally to the anal opening, several malformations may occur:

- The rectum may end in a closed sac without connecting to the anus at all.
- The rectum may lead to other areas of the body, such as the vagina or urethra.
- The anus itself may be missing.
- In many cases, the rectum may also be abnormally narrow, which further limits its function.

The incidence of anorectal atresia (obstruction) and stenosis (narrowing) in Victoria is around one in every 1,800 births. Symptoms include an absent or unusually located anus, no faeces passed within two days of birth, or faeces exiting the body via the vagina or urethra. Other problems sometimes seen with imperforate anus include exomphalos (herniated intestines and other abdominal organs through a hole in the abdominal wall), oesophageal atresia and tracheo-oesophageal fistula.

Treatment for imperforate anus

The condition is diagnosed by physical examination and x-rays. Imperforate anus is corrected with surgery, and techniques differ depending on the nature of the defect. Usually, initial treatment involves sewing the rectum to a surgical hole in the abdominal wall (stoma) and fitting a colostomy bag so that faeces can leave the body. Any connections to the vagina or urethra are closed. Anal repair surgery is carried out at a later stage. The rectum is attached to the anus and an opening in the anus may need to be made.

Long term difficulties for the child

Depending on the availability and functioning of the surrounding nerves and muscles, the child may have problems with bowel continence. Preparations to ensure soft faeces and a special diet may be needed for several years.

Where to get help

- Your doctor
- Paediatrician.

Things to remember

- Birth defects of the digestive tract include oesophageal atresia (obstruction of the oesophagus) and imperforate anus (malformations of the anus).
- Most babies born with oesophageal atresia also have tracheo-oesophageal fistula, which means the trachea and oesophagus are connected.
- Treatment options include surgery.

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