Abdominal birth defects

Summary

- Three types of birth defects that affect the abdomen include diaphragmatic hernia (organs protrude into the chest cavity), exomphalos (organs protrude through the navel) and gastroschisis (organs protrude through the abdominal wall).
- These conditions may be detected during pregnancy with ultrasound scans.
- The causes are unknown, so prevention is not possible.

The organs of the digestive system - intestines, stomach, pancreas and liver - are located in the abdomen, separated from the chest cavity by the diaphragm. During fetal development, the diaphragm or abdominal wall may fail to properly fuse, allowing the abdominal organs to protrude. Three types of birth defects that affect the abdomen include diaphragmatic hernia (organs protrude into the chest cavity), exomphalos (organs protrude through the navel) and gastroschisis (organs protrude through the abdominal wall). These conditions may be picked up during pregnancy with ultrasound scans. The presence of too much amniotic fluid surrounding the baby (polyhydramnios) may also be symptomatic of these defects. The causes are unknown, so prevention is not possible.

Diaphragmatic hernia

The diaphragm is a sheet of muscle slung beneath the lungs. When we breathe, the diaphragm contracts and relaxes. This change in air pressure sucks air into the lungs on inhalation and pushes air out on exhalation. Normally, the abdominal organs are located beneath the diaphragm. A diaphragmatic hernia means there is an abnormal hole in the diaphragm and the abdominal organs develop inside the ribcage. The lack of space within the chest cavity and the structural defects of the diaphragm mean that breathing may be difficult. The crowded lung may also have failed to develop properly. At birth, the baby looks blue (cyanotic) because of the lack of oxygen, and may breathe erratically or not at all. The abdomen also feels flatter than it should. The incidence of diaphragmatic hernia in Victoria is around one in every 2,500 births.

Treatment for diaphragmatic hernia

Diaphragmatic hernia is diagnosed by physical examination and chest x-rays. This defect is potentially fatal and the baby needs immediate surgery to relocate the abdominal organs and repair the diaphragm. In most cases, the baby must have respiratory support and a prolonged hospital stay after surgery. The outlook depends on how severely the lungs were affected by the overcrowding. The child may be prone to lung infections.

Exomphalos

The incidence of exomphalos in Victoria is around one in every 2,500 births. Exomphalos (omphalocele) is an abnormal hole in the abdominal wall that allows the intestines and other organs to protrude. The organs are covered by the abdominal membrane (peritoneum) instead of skin. In most cases, the hernia occurs at the navel (umbilicus). The navel of the newborn baby may have a diameter of 10 cm in severe cases, and their organs may have herniated into the umbilical cord. Estimates vary, but it is thought that around one third to half of all babies born with exomphalos have other birth defects as well, including congenital heart disorders.

Treatment for exomphalos

Physical examination alone is usually enough to diagnose exomphalos. Following delivery, the baby needs surgery in the first days of life. If there is insufficient room to repair the hernia straight away, a special sack is stitched around it. This sack shrinks over time, gently pressing the intestines back into the abdomen. The hole can then be sutured closed. The baby may need a prolonged hospital stay.

Gastroschisis

The incidence of gastroschisis in Victoria is around one in every 4,000 births. Gastroschisis is another condition...
involving herniated abdominal organs, but the defect differs from exomphalos in a number of ways: it is less likely that there will be other organ problems; the hernia is through the abdominal wall instead of the navel; and the protruding organs aren’t sheathed with the protective peritoneum. The exposed organs can dry out or become infected, and so are covered in moist, sterile dressings as soon as possible after birth. In severe cases, the entire contents of the abdomen are located outside of the body.

**Treatment for gastroschisis**

Physical examination alone is usually enough to diagnose gastroschisis. Following delivery, the baby requires surgery in the first days of life. Treatment depends on the degree of the hernia and whether there is sufficient room inside the abdominal cavity to accept the herniated organs. If the baby’s abdomen is large enough, the organs are relocated inside the body and the hernia sewn shut. In severe cases, a mesh sack is sewn around the hernia and the repair operation performed at a later stage. If the intestines are damaged, the child may have digestion problems.

**Where to get help**
- Your doctor
- Paediatrician

---

**This page has been produced in consultation with and approved by:**

Murdoch Children's Research Institute