

Exomphalos and gastroschisis

Synonyms: ventral hernia

Ventral body wall defects include ectopia cordis, bladder exstrophy, and the abdominal wall malformations gastroschisis and exomphalos.

What is exomphalos?

Exomphalos literally translated from the Greek means 'outside the navel'. It is also called an omphalocele. It is a congenital abnormality in which the contents of the abdomen herniate into the umbilical cord through the umbilical ring. The viscera, which often includes the liver, is covered by a thin membrane consisting of peritoneum and amnion.

What is gastroschisis?

Gastroschisis means 'stomach cleft'. It is a congenital defect of the abdominal wall, usually to the right of the umbilical cord insertion. Abdominal contents herniate into the amniotic sac, usually just involving the small intestine but sometimes also the stomach, colon and ovaries. Unlike exomphalos, there is no covering membrane.

How common are exomphalos and gastroschisis? (Epidemiology)

Estimating the overall frequency of abdominal wall defects is complicated by the fact that the pregnancy may be ended by elective termination. Estimates of the birth prevalence for abdominal wall defects are 1.85–1.96 per 10,000 births.^[1]

The incidence of gastroschisis has increased markedly over a period of 25 years, to a level of 4-5 per 10,000 live births in the USA.^[2] The increase in prevalence has been described as a 'pandemic' and raises interesting possibilities about interaction between genes and environment. Gastroschisis often occurs as a single defect so (after surgical repair) is associated with a good prognosis.

However, fetuses with exomphalos have an increased risk for chromosomal abnormalities and so are more likely to have severe comorbidities and long-term problems.^[3] Both exomphalos and gastroschisis have been linked to conditions affecting placental insufficiency, including maternal illness and infection, drug dependency and smoking. Known risk factors include young maternal age, use of substances during pregnancy, and maternal hypothyroidism.^[4]

Presentation of exomphalos and gastroschisis

Exomphalos and gastroschisis may both present prenatally as a rise in alpha-fetoprotein in the second trimester or as an abnormality on ultrasound scan.^[5]

Exomphalos results in a 4-12 cm abdominal wall defect which may be central, epigastric or hypogastric. A large defect may be associated with underdevelopment of the abdominal and thoracic cavities. Associated congenital defects may be evident (see 'Differential diagnosis', below). One study found that 74.4% of patients with exomphalos had associated defects, as opposed to 16.6% of patients with gastroschisis.^[6]

Gastroschisis presents as an opening usually less than 5 cm in length, to the right of the umbilical cord.^[2] There is much less variation in size than there is in exomphalos. Other abnormalities are not usually present.

Differential diagnosis^[5]

Exomphalos and gastroschisis need to be differentiated from each other and from other causes of an abdominal wall mass. Exomphalos is more centrally placed than gastroschisis, is covered with a membrane and is more likely to be associated with other congenital defects. The hernial sac in the two conditions also feels different. An exomphalic sac feels firm and may contain liver and tightly packed bowel, whereas in gastroschisis the sac contains only free loops of bowel and feels softer.

Other causes of abdominal wall mass include:

- **Physiological bowel herniation.** This occurs at 10–13 weeks of gestation and may be detected on ultrasound scan. It can be differentiated from exomphalos, as a scan at 15 weeks will show that it has disappeared. A large defect which contains liver indicates exomphalos at any stage.
- **Umbilical hernia.** These are frequent in premature infants. An ultrasound scan will help to diagnose them.
- **Amniotic band syndrome.** This is a common cause of abdominal wall defects. This starts with an accidental break in the inner amniotic membrane which results in the fetus being exposed to fibrous amniotic bands. The appearance may be similar to gastroschisis but a scan may demonstrate the atypical location of the defect and contiguity with amniotic membranes.
- **Extrophy of the urinary bladder.** This appears on a scan, in female babies, as a mass superior to the fetal genitalia. Serial scans subsequently fail to demonstrate the presence of a bladder, and other abnormalities of the urogenital system may be detected.
- **Limb-body wall complex.** There is a severe and fatal anterior abdominal wall defect lateral to the umbilical cord insertion point. Other congenital defects involving the heart, limbs, brain and spine may also be detected.
- **Multiple cavernous haemangiomas.** These are most frequently found over the lower body. Other surface masses may be found that cause limb hypertrophy. This is called [Klippel-Trénaunay syndrome](#).
- **Pseudo-omphalocele.** This term is used to describe a structurally normal abdomen which has been changed in shape by external pressure. This has been known to occur as a result of pressure from an ultrasound transducer. It can also occur in oligohydramnios and other causes of compression of the lower thoracic wall.

Investigations

Laboratory tests

- Maternal serum alpha-fetoprotein (MSAFP) levels. These are raised in exomphalos and gastroschisis. The levels are higher in gastroschisis than in omphalocele.
- Karyotyping. The strong link between exomphalos and chromosomal abnormalities makes karyotyping an essential part of the investigation of this condition.

Imaging

- Ultrasonography. This is the mainstay of diagnosis. Differentiation of exomphalos from gastroschisis and other causes of abdominal wall mass can be made from the size and site of the defect and any associated features (see 'Differential diagnosis', above).
- MRI. This may be helpful as an adjunct to ultrasonography, particularly in identifying abnormalities in the position of the liver.
- Other imaging. In infants with exomphalos, renal ultrasound, echocardiogram and plain films of the sacrum may be useful to exclude associated abnormalities.

Procedures

- Amniocentesis is indicated if a raised alpha-fetoprotein is combined with appearances on ultrasound suggestive of exomphalos.

Associated diseases

Exomphalos

This may be associated with a number of other congenital abnormalities, including:

- **Thoraco-abdominal pentalogy of Cantrell.** In this condition, exomphalos is associated with a cleft sternum, anterior diaphragmatic hernia, heart defects (ventricular septal defect and ectopia cordis) and an absent pericardium.
- [Beckwith-Wiedemann syndrome](#). This is a congenital overgrowth condition characterised by exomphalos, macroglossia and gigantism.
- **Genetic disorders.** Trisomy 13, 18 and 21 can all be associated, highlighting the theory that some cases can have a genetic cause.

- **Cloacal extrophy.** This is a low exomphalos combined with bladder or cloacal extrophy. It may also be associated with other caudal anomalies, such as meningomyelocele, anal atresia and lower-limb anomalies.
- **Dental malocclusion and musculoskeletal abnormalities.** These can also occur.

Gastroschisis

Although associated abnormalities are less common in gastroschisis than in exomphalos, they do occur in 7-30% of babies. Deformities include anencephaly, cleft lip and palate, ectopia cordis, atrial septal defect, diaphragmatic hernia, scoliosis, syndactyly and amniotic band. Intestinal atresia, malabsorption, atypical appendicitis, midgut volvulus, gastro-oesophageal reflux and Hirschsprung's disease also occur. A prenatal association with polyhydramnios has been noted and the latter finding on ultrasound should raise suspicion of the diagnosis.

Management of exomphalos and gastroschisis^[7] ^[8]

Exomphalos

Mode of delivery depends on size of defect. Small defects may be able to deliver vaginally.^[9]

- A baby with an intact sac is usually medically stable and may not require much in the way of pre-operative medical care. If the sac ruptures, the medical treatment should be the same as for gastroschisis.
- Fluid intake should be maintained intravenously and the sac covered with non-adherent gauze.
- Replacement of the contents within the abdominal cavity and surgical closure are usually achieved without much difficulty with small- to moderate-sized sacs. Large exomphalos containing the liver may be more challenging.

Similarly, timing and method of repair also depend on size of defect.^[9] Small defects are often able to be repaired as a primary closure.

- Closure of a giant omphalocele containing the liver is always challenging.^[10] A temporary artificial holding sac (a silo) may need to be constructed. Non-adhesive dressing and Saran® wrap are often used for this purpose. Closure of a large abdominal wall defect may require an artificial patch. A variety of rigid and non-rigid patches has been used. Non-rigid patches have the advantage of revascularisation from liver blood vessels and hence lower chance of sepsis; however, the incidence of subsequent secondary herniation is higher. Optimal outcomes are obtained by utilising a multidisciplinary approach in both the antenatal and postnatal period.

Gastroschisis

Delivery decisions are not affected by the presence of gastroschisis alone. Other factors may necessitate preterm delivery, such as intrauterine growth restriction (IUGR).

Some patients can be managed by plastic closure (vigorous stretching of the abdominal wall with gradual decompression of the abdominal contents from a temporary silo into the abdomen). In the majority of patients, however, primary closure of the defect is the main objective. This may need to be delayed if the intestines are too inflamed and hence too enlarged to be replaced in the abdominal cavity. Too tight closure must be avoided in order to prevent respiratory problems and a reduction in cardiac output. Recent advances in surgery have resulted in improved cosmetic appearance.

The baby may furthermore require medical stabilisation before surgery. Respiratory distress may require gastric decompression and sometimes endotracheal intubation. Fluid loss must be corrected intravenously. The baby should be placed under a radiant heater to minimise heat loss. A broad-spectrum antibiotic should be administered to prevent infection.

Parenteral nutrition should be provided via a central venous line, the baby should be catheterised to measure urine output and digital examination should be performed to dilate the rectum.

Gastroschisis repair may be performed immediately or within five days with equivalent outcomes.^[11]

Complications of exomphalos and gastroschisis^[5] ^[12]

- Exomphalos can act as a metabolic drain affecting nitrogen balance and leading to [failure to thrive](#).
- Poor nutritional status can result from the omphalocele acting as a metabolic drain.
- [Hepatomegaly](#) and [cholestasis](#) can result from prolonged parenteral feeding.
- Sepsis is highly likely if nutritional status is poor and this can be further complicated by respiratory dysfunction due to increased intrathoracic pressure.
- A large exomphalos can require several surgical procedures to correct the defect. A prolonged stay in hospital is common and may be complicated by respiratory compromise requiring intubation.
- Trauma to the liver can be a late complication if it is sited beneath the area normally protected by the ribcage.
- The main complication of gastroschisis is intestinal atresia which can occur in 10–20% of patients. In this condition, the mucosa and submucosa of the intestine form a web or diaphragm, partly obstructing the lumen. Malabsorption syndromes are common sequelae.
- Postoperatively, several weeks of intestinal dysfunction are common and prolonged parenteral feeding may be required. Too tight closure of the defect can result in compromise of pulmonary function, cardiac output and blood circulation to the kidneys.
- Significant loss of viable bowel can result in [short bowel syndrome](#) in both exomphalos and gastroschisis.

Prognosis^[5] ^[12]

The prognosis of both conditions has improved considerably over 35 years, due to the improvement in ventilatory care and total parenteral nutrition. In gastroschisis in particular, such improvements have increased the incidence of primary closure, with lessening of the complications of delayed surgery, such as sepsis.

Significant morbidity, however, still occurs. In both conditions, [intrauterine growth restriction](#) and exteriorisation of the liver have been found to be factors predictive of poor outcome.

Exomphalos

Prognosis depends on the size of the defect, whether any associated chromosomal abnormalities are present and whether complications such as pulmonary dysfunction or sepsis develop. Despite improvements in nutritional and respiratory support, mortality and morbidity still occur. The mortality rate of giant exomphalos without chromosomal anomaly or major malformations is low when treated by gradual reduction of the contents. The mortality rate is much higher in the presence of chromosomal abnormalities with severe associated defects.

Gastroschisis

Most deaths are the result of sepsis, [premature delivery](#) or bowel infarction. Poor prognosis is more likely with bowel-related complications, such as necrosis, atresia, severe dilation or thickening of the bowel, or difficulty in closing the defect. Compared with infants with simple gastroschisis (intact, uncompromised, continuous bowel), those with complex gastroschisis (bowel perforation, necrosis or atresia) take longer to reach full enteral feeding, require a longer duration of parenteral nutrition, require a longer stay in hospital, are more likely to develop intestinal failure and liver disease associated with intestinal failure and are more likely to require unplanned re-operation. ^[13] ^[14]

Adverse prognostic factors include prematurity, gastrointestinal complications and non-gastrointestinal anomalies.

Prevention

[Pre-pregnancy counselling](#) should include advice about a healthy lifestyle with adequate nutrition and the avoidance of smoking and recreational drugs.

The role of folic acid supplementation, whilst established for the prevention of spina bifida, is less supported for the prevention of ventral hernias, although animal experiments suggest it may play a part.

Termination of pregnancy may be considered if there is a large exomphalos, especially if there are associated congenital abnormalities.

Further reading

- [Gledina A, Singh S](#); Gastroschisis, a congenital anterior abdominal wall defect: a rare clinical image. *Pan Afr Med J.* 2022 Aug 22;42:298. doi: 10.11604/pamj.2022.42.298.36378. eCollection 2022.

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Authored by:	Peer Reviewed by: Dr Colin Tidy, MRCP	
Originally Published: 20/11/2023	Next review date: 13/03/2023	Document ID: doc_1023

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