CONDITION

Omphalocele

Omphalocele is a birth condition in which abdominal organs develop outside the belly within a thin sac.

WHAT IT IS
Membrane-covered abdominal organs outside the body at birth

BODY SYSTEM
Abdomen and digestive system

MAIN SYMPTOMS
Abdominal sac, breathing difficulty, feeding problems

WHAT CAUSES IT
Abnormal fetal development, usually unknown

HOW IT IS DIAGNOSED
Prenatal ultrasound or physical examination after birth

MAIN TREATMENT
Newborn intensive care and surgical closure

CAN IT BE CURED?
Yes - surgery can close the abdominal wall

IS IT AN EMERGENCY?
Yes - newborns need immediate specialist care

LONG-TERM OUTLOOK
Often good, shaped by size and associated conditions

Overview

Omphalocele is a condition present from birth in which some abdominal organs develop outside the baby's body. The organs push through an opening where the umbilical cord joins the abdomen. They are usually covered by a thin, transparent sac made from layers of tissue.

A small omphalocele may contain only part of the intestine. A large one may also contain the liver and other organs. Very large defects are sometimes called giant omphaloceles.

During early pregnancy, a developing baby's intestines temporarily extend into the umbilical cord as part of normal growth. They usually move back into the abdomen as development continues. In omphalocele, this process and the closing of the abdominal wall do not happen normally.

Omphalocele is different from gastroschisis. In gastroschisis, the opening is usually beside the umbilical cord, and the bowel is not covered by a sac. In omphalocele, the opening is at the base of the cord and the organs are usually covered.

The condition matters because the exposed sac needs protection after birth. The baby may also have difficulty breathing or feeding. Omphalocele is often linked with differences affecting the heart, chromosomes, or other organs, so care involves more than closing the abdominal wall.

Symptoms

Omphalocele does not usually cause symptoms that a pregnant person can feel. It is often found during a routine pregnancy ultrasound.

After birth, the main sign is immediately visible. A sac containing bowel, liver, or other abdominal organs projects from the belly at the umbilical cord.

Other problems depend on the defect's size and whether the baby has associated conditions. They may include:

The covering sac may occasionally tear before or during birth. If that happens, the organs lose their protective covering and need urgent protection and assessment.

Omphalocele itself is usually not painful in the way an injury is. However, newborns may need procedures, breathing support, and surgery, so the care team will provide appropriate comfort and pain relief.

- Difficulty breathing, especially with a large omphalocele
- Problems beginning or tolerating feeds
- A small chest or underdeveloped lungs
- Signs related to an associated heart or genetic condition

Causes and risk factors

Omphalocele forms early in pregnancy as the abdominal wall and digestive organs develop. The exact reason is usually not known. It is not caused by something a parent did or did not do during pregnancy.

Most cases are not inherited as an isolated condition. However, omphalocele can occur as part of a genetic or chromosome condition. Chromosomes are packages of DNA that carry genetic

information.

Associated conditions can include chromosome differences such as trisomy 13, trisomy 18, or trisomy 21. Omphalocele can also occur with Beckwith-Wiedemann syndrome, which affects growth, or with patterns of birth differences involving the heart, chest, spine, or lower body.

Heart differences are among the more common associated concerns. Some babies also have differences affecting the kidneys, brain, digestive tract, or skeleton.

These links do not mean that every baby with omphalocele has another condition. Testing is used to understand each baby's anatomy and likely care needs.

Diagnosis

Omphalocele is often diagnosed before birth with ultrasound. The scan shows abdominal organs extending into the base of the umbilical cord within a sac. Ultrasound can also help estimate the defect's size and identify which organs it contains.

A raised level of alpha-fetoprotein, or AFP, on pregnancy blood screening may lead to further testing. AFP screening alone does not confirm the diagnosis.

After prenatal diagnosis, further assessment may include:

These tests help with planning but cannot predict every issue before birth.

If the condition was not found during pregnancy, the appearance of the sac at birth usually confirms it. The newborn team may then use heart scans, X-rays, ultrasound, blood tests, or genetic testing to look for associated conditions and prepare treatment.

Birth is generally planned at a hospital with newborn intensive care and pediatric surgical services. The safest timing and method of delivery are based on obstetric factors, the sac's size, its contents, and the baby's overall condition. An omphalocele does not automatically require a cesarean birth in every case.

- Detailed ultrasound of the baby's organs and growth
- Fetal echocardiogram, a detailed ultrasound of the heart
- Diagnostic genetic testing, if chosen after counselling
- Magnetic resonance imaging, or MRI, in selected cases

Treatment and management

Immediately after birth, the priority is to protect the sac and keep the baby stable. The care

team covers the sac with sterile material to reduce heat and fluid loss and lower the risk of injury. The baby may receive fluids through a vein and a tube into the stomach to remove swallowed air and fluid.

Some babies breathe on their own. Others need oxygen or mechanical ventilation, particularly when a large omphalocele has limited lung development or leaves little room in the abdomen.

The timing and type of closure depend on the defect's size, the organs involved, the space inside the abdomen, and the baby's other health needs.

A small omphalocele may be closed during one operation. The surgeon places the organs into the abdomen and repairs the opening.

With a larger defect, forcing all the organs into the abdomen at once could place too much pressure on the lungs, heart, intestines, and major blood vessels. Closure may therefore happen in stages. The organs are moved inward gradually before the abdominal wall is repaired.

For some giant omphaloceles, the sac is allowed to develop a skin covering over time. The remaining abdominal wall hernia can then be repaired months or sometimes years later. This approach is often described as delayed closure or "paint and wait," although the exact materials and method are chosen by the specialist team.

Feeding usually begins when the baby's condition and bowel function allow it. Nutrition may initially be given through a vein. Milk can then be introduced through a feeding tube or by mouth. Feeding therapy may help babies who have trouble coordinating sucking and swallowing or who have been unable to feed by mouth for a long period.

Treatment may also be needed for heart differences, genetic conditions, reflux, lung problems, or other associated concerns.

Complications

The risks differ greatly between a small isolated omphalocele and a giant omphalocele with other medical conditions.

Possible early complications include:

Pressure inside the abdomen is watched closely during and after closure. Excess pressure can interfere with breathing, circulation, kidney function, and blood flow to the intestines.

Later complications can include an abdominal wall hernia, reflux, feeding difficulties, or bowel obstruction. An obstruction may develop from bands of internal scar tissue called adhesions. It can cause vomiting, abdominal swelling, and difficulty passing stool.

For many babies, associated heart, lung, chromosome, or genetic conditions have a greater effect on health than the abdominal wall defect itself.

- Tearing of the protective sac
- Infection or injury involving the exposed organs
- Breathing problems from small lungs or raised abdominal pressure
- Reduced blood flow to organs after closure
- Feeding intolerance and a need for intravenous nutrition
- Wound problems after surgery

Prevention

There is no established way to prevent most omphaloceles. The condition forms early in fetal development, and its cause is usually unknown.

When omphalocele is connected with a chromosome or genetic condition, genetic counselling can explain test results and discuss the chance of the condition occurring in another pregnancy. This information is specific to the diagnosis and family history.

Routine pre-pregnancy and prenatal care supports overall health, but it cannot guarantee that an omphalocele will not occur.

Living with it and what to expect

The course over months and years is shaped mainly by the defect's size, lung development, feeding progress, and associated conditions.

A baby with a small, isolated omphalocele may have one operation and begin feeding after the bowel recovers. Hospital follow-up then focuses on growth, the surgical scar, and abdominal wall strength.

A baby with a giant omphalocele may remain in hospital longer and need breathing support, intravenous nutrition, tube feeding, or several procedures. If closure is delayed, families learn how to protect and care for the developing skin covering with guidance from the surgical team.

Feeding can take time. Some children need higher-calorie nutrition, tube feeding, or help from feeding and speech therapists. Progress is often gradual rather than steady.

After discharge, routine follow-up may involve pediatric surgery, primary care, nutrition, cardiology, genetics, lung specialists, and developmental services. Not every child needs every

specialist.

As children grow, doctors monitor for hernias, bowel obstruction, breathing concerns, reflux, and delayed growth or development. Further abdominal wall surgery may be considered if closure was delayed or a hernia remains.

Many children with an isolated, successfully repaired omphalocele take part in ordinary play, school, and family life. A scar and a belly button that looks different are common. Long-term health is less predictable when there are major heart, lung, chromosome, or genetic conditions.

When to get medical help

Omphalocele identified before birth needs planned specialist care, but a stable pregnancy diagnosis is not usually an emergency for the pregnant person. Keep scheduled appointments with the fetal medicine, obstetric, newborn, and surgical teams.

At birth, omphalocele requires immediate hospital care. If a baby is unexpectedly born outside a hospital with organs visible at the umbilical area, call emergency services. Do not push the organs inward or place dry material directly on them.

After discharge, contact the child's healthcare team promptly for:

Seek urgent or emergency care for:

These symptoms can indicate bowel obstruction, breathing trouble, infection, or another serious complication.

- New feeding difficulty or repeated vomiting
- Poor weight gain
- Increasing redness, swelling, drainage, or pain near the repair
- A new or enlarging abdominal bulge
- Ongoing constipation, reflux, or abdominal discomfort
- Green vomit, which may contain bile
- A swollen, firm, or very tender abdomen
- Repeated vomiting with inability to pass stool or gas
- Sudden breathing difficulty, blue or grey skin, or unusual limpness
- Heavy bleeding, an opened wound, or a torn omphalocele covering
- Signs of severe dehydration, such as very few wet diapers and marked sleepiness

Common questions

Can an omphalocele go away before birth?

A confirmed omphalocele generally does not disappear before birth. Occasionally, an early ultrasound finding may later be reclassified as normal temporary bowel development or another condition. Follow-up imaging helps confirm the diagnosis.

Is omphalocele the same as gastroschisis?

No. An omphalocele is centered at the umbilical cord and usually has a covering sac. Gastroschisis is usually beside the cord, and the exposed bowel has no sac. Omphalocele is also more often associated with chromosome, heart, and other organ differences.

Will my baby need surgery right after birth?

Most babies need surgery or a planned method of delayed closure, but not all have immediate final closure. A small defect may be repaired early. A large defect may need gradual or delayed treatment to avoid dangerous pressure inside the abdomen.

Can a baby with omphalocele eat normally?

Many eventually feed by mouth, but feeding may be delayed after birth. Some need intravenous nutrition or tube feeding while the bowel recovers and breathing becomes stable. Long-term feeding problems are more likely after a large defect or a prolonged hospital stay.

Does omphalocele mean the baby has a genetic condition?

Not always. Some babies have an isolated omphalocele, while others have a chromosome or genetic condition. Detailed imaging and optional diagnostic genetic testing can provide more information.

Can someone with a repaired omphalocele have a normal life?

Many people with an isolated, repaired omphalocele live active lives. They may have an abdominal scar, a different-looking belly button, or later hernia treatment. The outlook is influenced most by lung development and any associated heart or genetic conditions.

Sources

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