Omphalocele

Omphalocele is a herniation of abdominal organs via the umbilicus in infants which is covered by the peritoneum. It occurs due to a defect of lateral fold closure during embryological development. It is associated with certain disorders such as trisomy syndromes and Beckwith-Wiedemann syndrome. Increased AFP can be detected in maternal serum. Omphalocele is treated with surgery.

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Characteristics

Herniation of Abdominal Organs via Umbilicus

Hermit-crab on the Baby Abdominal standing on O-full-of-eels and holding an Umbrella

Omphalocele is characterized by a persistent herniation of abdominal organs via the umbilicus. Physiologically, herniation is a part of organogenesis during the sixth week of development due to increased abdominal content. The hernia will shrink by 12 weeks of gestation. If the hernia persists, omphalocele can occur.

Covered by Peritoneum

Peritoneum-curtain

Omphalocele is covered by the peritoneum and can be filled with intestines, stomach, liver, and other abdominal contents. This characteristic makes it different from gastroschisis, which is not covered by peritoneum.

Defect of Lateral Fold Closure

Broken Folding Ladders

Omphalocele is characterized by an anterior wall defect found in infants. It occurs due to a failure of lateral fold closure.

Associations

Trisomy Syndromes

Tricycle

Omphalocele has been associated with trisomy syndromes (25-50% of cases), such as Patau syndrome (trisomy 13) and Edward syndrome (trisomy 18).

Beckwith-Wiedemann Syndrome

Beckham-with-Wide-man

Omphalocele is known to be associated with Beckwith-Wiedemann syndrome. This is an overgrowth syndrome characterized by macroglossia, organomegaly, hemihyperplasia (WT2 mutation), omphalocele, and other structural abnormalities. Children with Beckwith-Wiedemann syndrome have a higher risk of developing Wilms tumor.

Diagnosis

Increased AFP

Up-arrow Air Force Pilot

Increased AFP (alpha fetoprotein) in maternal serum has been linked to omphalocele. Increased AFP can also be seen in other abdominal defects (e.g., gastroschisis), neural tube defects, and other malformations.

Management

Surgery

Surgeon

Small omphaloceles can be repaired within 72 hours after delivery with primary closure of the skin and fascia. A large omphalocele can be approached with staged surgery or the "paint and wait" technique. In this technique, an antibiotic-laden cream is applied to the external surface of the omphalocele ("painting") then covered by gauze. Over time, the neonate's skin will grow over the omphalocele ("waiting").