Omphalocele and Gastroschisis

Background:

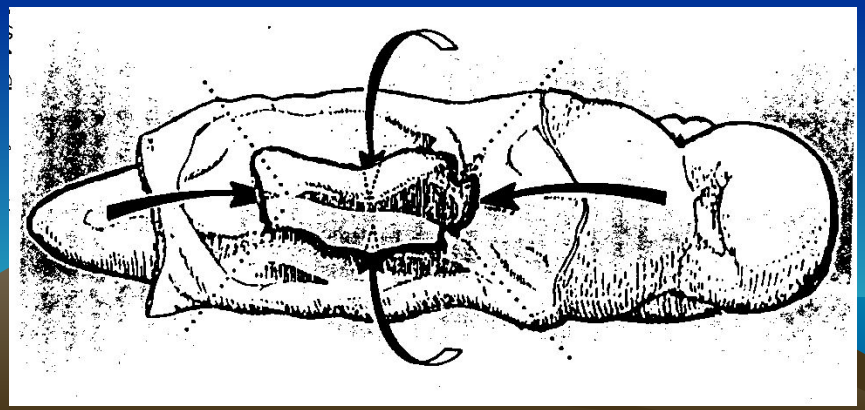
- Gastroschisis and omphalocele are among the most frequently encountered congenital anomalies in pediatric surgery.
- Combined incidence of these anomalies is **1 in 2000** births, which means, for example, that a pediatric surgeon will see 2 such babies for every 1 born with esophageal atresia or tracheoesophageal fistula.

Background:

- Many babies have correctable lesions and simply require routine pediatric care.
- For others, the abdominal wall defect is part of a larger constellation of unresolved problems, and further care by specialists is necessary.
- All of these children, however, require general management by pediatricians who have knowledge of their particular anomalies and their past surgical histories.
- For example, physicians should know if an associated malrotation was corrected (to prevent midgut volvulus) and whether an abnormally located appendix was removed (to prevent occurrence of atypical appendicitis).

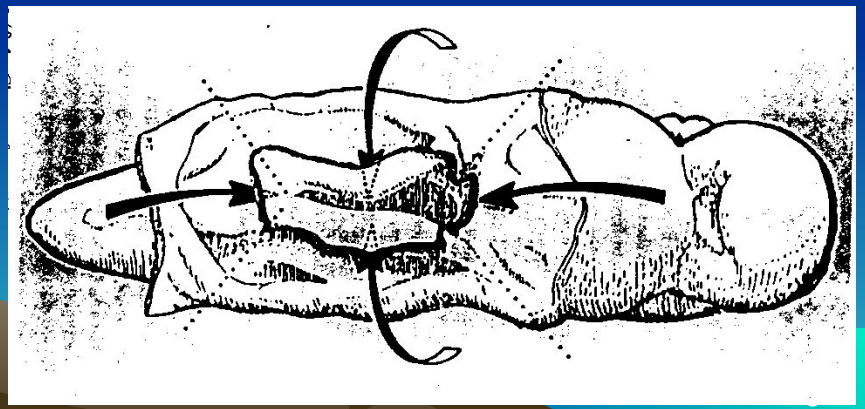
Pathophysiology: Embryology

- the human embryo initially is disc-shaped and composed of 2 cell layers.
- It acquires a third cell layer as it grows above the umbilical ring and becomes cylindrical by elongation and inward folding.
- The body folds (cephalic, caudal, lateral) meet in the center of the embryo where the amnion invests the yolk sac.
- Defective development at this critical location results in a spectrum of abdominal wall defects.



Pathophysiology: Embryology

- By the **sixth week**, rapid growth of the midgut causes a physiologic hernia of the intestine through the umbilical ring.
- The intestine returns to the abdominal cavity during the **tenth week**, and rotation and fixation of the midgut occur.
- This process does not occur in babies with gastroschisis or omphalocele, resulting in an increased risk of midgut volvulus.



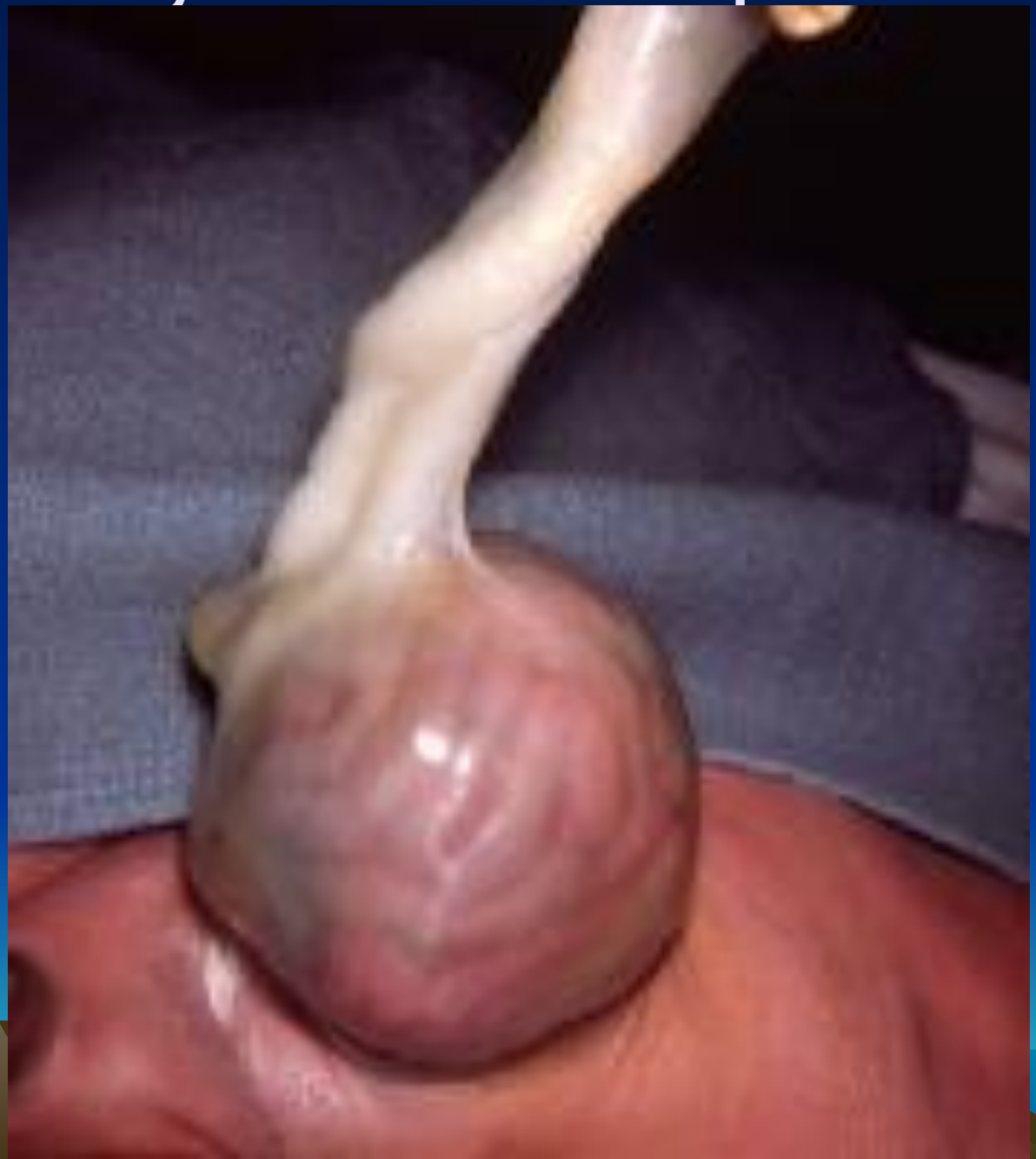
Pathogenesis of omphalocele and gastroschisis

- Abdominal wall defects occur as a result of failure of the mesoderm to replace the body stalk,
- Embryonic dysplasia causes insufficient outgrowth at the umbilical ring. Decreased apoptotic cell death and underdevelopment of the mesodermal cell compartment cause enlargement of the umbilical ring's diameter.
- The amnion does not apply itself to the yolk sac or connecting stalk but remains at the margin of the body wall defect, causing faulty development of the umbilical cord and a persistent communication between the intraembryonic body cavity and the extraembryonic coelom.

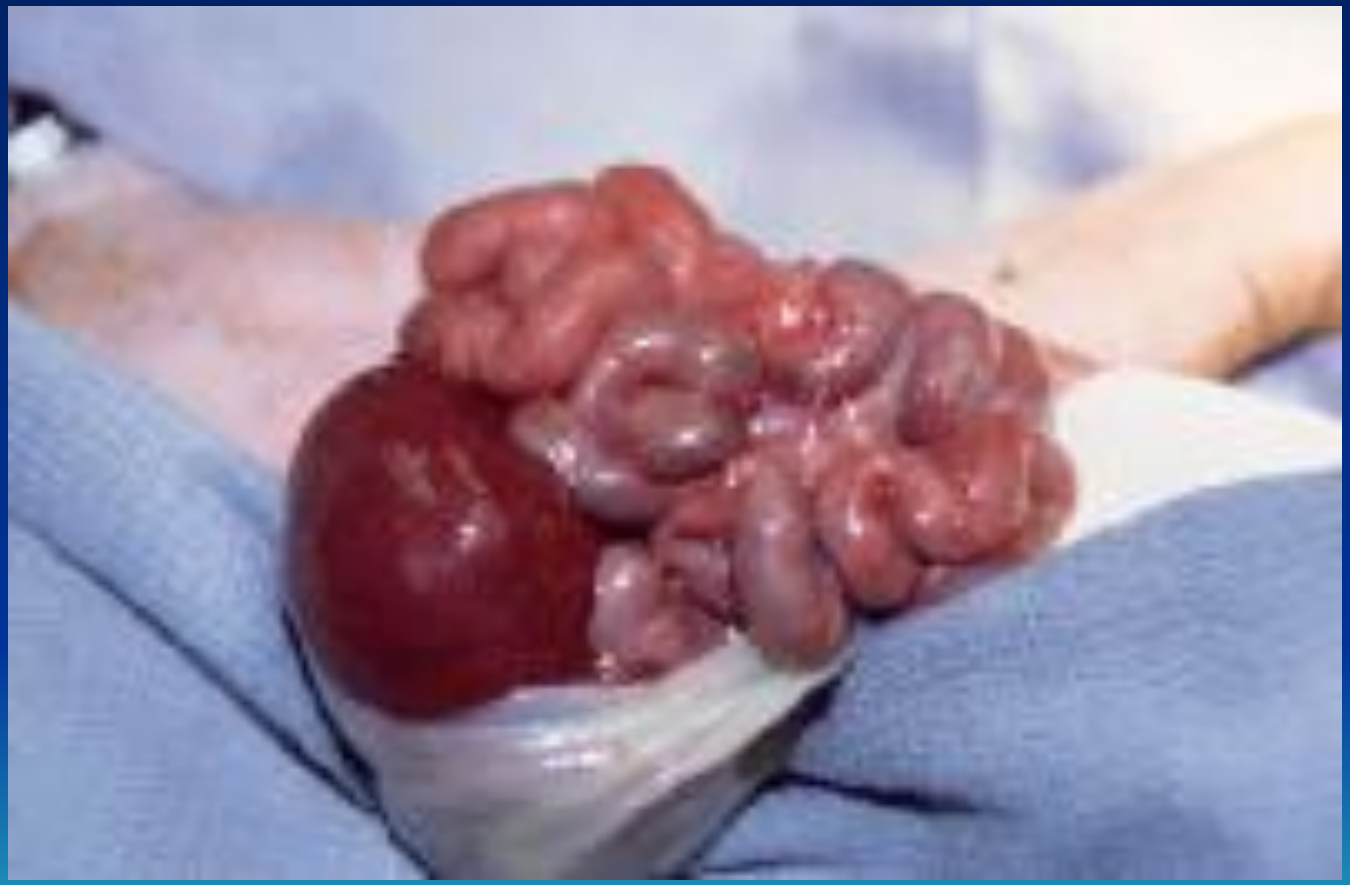
Pathogenesis of Omphalocele

- In babies with omphalocele failure of central fusion at the umbilical ring by growth of the mesoderm causes defective abdominal wall closure and persistent herniation of the midgut.
- The abdominal viscera are contained within a translucent sac, which is composed of amnion, Wharton jelly, and peritoneum.
- The umbilical vessels radiate onto the wall of the sac. In 50% of cases, the liver, spleen, and ovaries or testes accompany the extruded midgut.

Baby with an omphalocele.



Baby with a ruptured omphalocele.

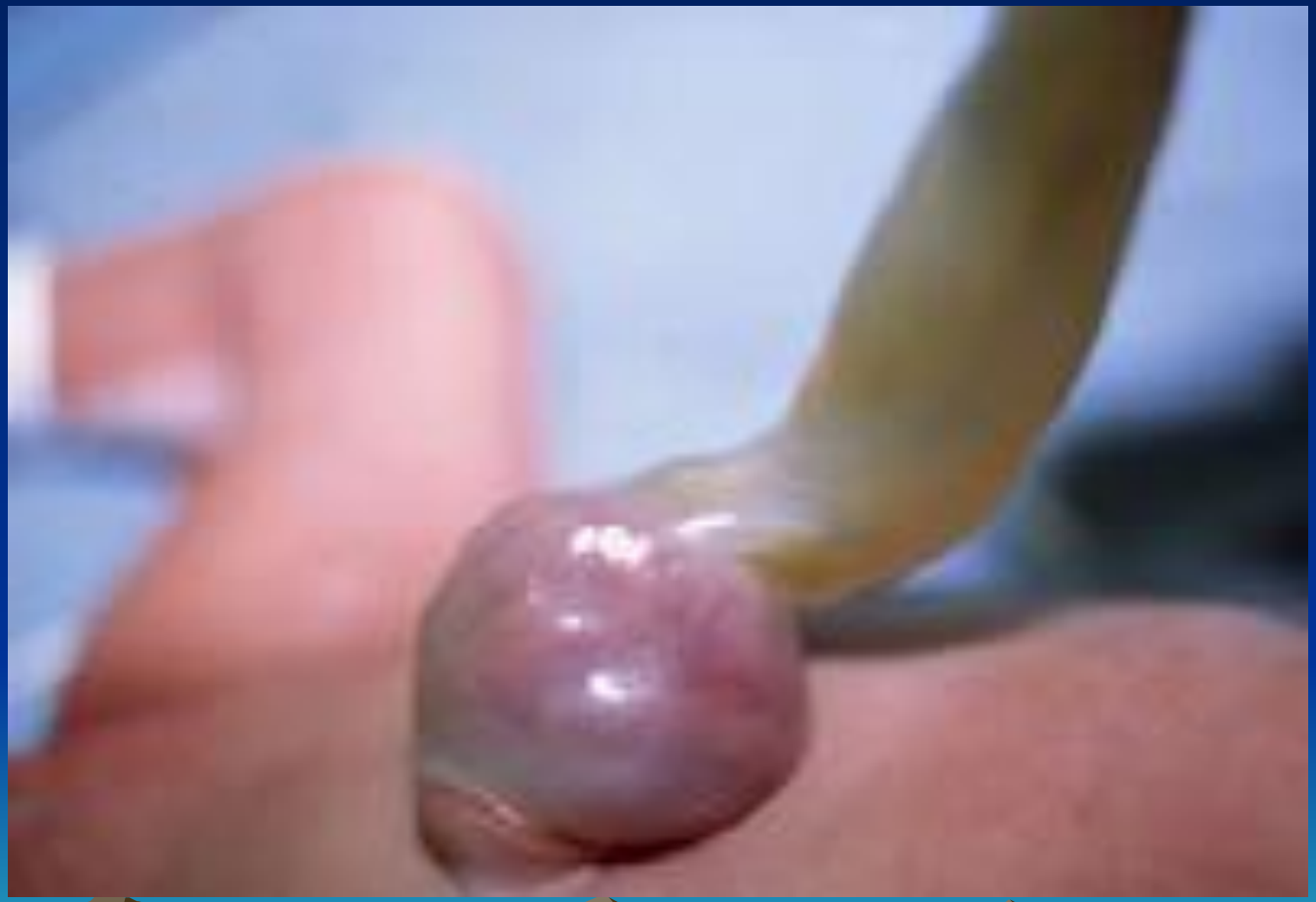


Pathogenesis of gastroschisis

- Possible explanations of the embryology of abdominal wall defect in gastroschisis include the following:

1. **Defective mesenchymal development** at the body stalk-abdominal wall junction results in a dysplastic abdominal wall that may rupture with increased abdominal pressure.
2. **Abnormal involution of the right umbilical vein or a vascular accident** involving the omphalomesenteric artery causes localized abdominal wall weakness that subsequently ruptures.
3. **Rupture of a small omphalocele** with absorption of the sac and growth of a skin bridge between the abdominal wall defect and the umbilical cord has been chronicled on prenatal ultrasound.

Baby with an umbilical cord hernia.



Frequency:

- **In the US:** Combined incidence of omphalocele and gastroschisis is 1 in 2000 births.
- Incidence of **omphalocele** has remained constant and is associated with increased maternal age. There is an inherited predilection
- Incidence of **gastroschisis** is increasing, and it is associated with young maternal age and low gravity.
- Prematurity and low birth weights, secondary to in utero growth retardation, are more common in babies with gastroschisis.

Mortality/Morbidity:

- Over the past 30 years, the survival rate of babies with gastroschisis and omphalocele has steadily improved, from approximately **60% in the 1960s to more than 90% currently:**

1. Improvements in the care of low birth weight and premature babies.
2. Better anesthetic management and surgical techniques.
3. Availability of excellent parenteral nutrition

Mortality/Morbidity:

- Long-term morbidity from **gastroschisis** is related to intestinal dysfunction and wound problems.
- **Short gut syndrome** may be caused by a number of factors.

1. An antenatal mesenteric vascular accident
2. Constriction of the extruded intestine's mesentery by a small abdominal wall defect may cause an obstructed, shortened intestine with diminished absorptive capacity.
3. Gut necrosis may complicate excessively tight closure of the abdominal wall defect by impeding splanchnic blood flow with resultant intestinal ischemia and necrotizing enterocolitis (NEC),
4. or it may occur consequent to **closed loop obstruction** caused by adhesions or midgut volvulus.
5. Loss of intestinal length exacerbates the dysfunction consequent to antenatal exposure of the intestine to amniotic fluid.

Mortality/Morbidity:

- **Management** of babies with short gut syndrome also has improved significantly as a result of providing nutrition by **parenteral and enteral routes**.
- Obtaining venous access and treating catheter sepsis, and optimizing gut adaptation with innovative surgical procedures and aggressive treatment of bacterial overgrowth within stagnant intestinal loops.
- Even so, babies with short gut as a consequence of gastroschisis comprise a large percentage of children undergoing intestinal transplantation.

Mortality/Morbidity:

- **Poor healing** of the abdominal wound usually results in a ventral hernia, which may require secondary surgical repair.
- Paradoxically, babies with small (unimpressive) omphaloceles are most likely to have associated abnormalities, including intestinal problems (Meckel diverticulum, atresia), genetic syndromes (Beckwith-Wiedemann), and congenital heart disease.
- Babies with giant omphaloceles usually have small, bell-shaped, thoracic cavities and minimal pulmonary reserve; reduction and repair of the omphalocele frequently precipitates respiratory failure, which may be chronic and require a tracheotomy and long-term ventilator support.

Mortality/Morbidity:

- Even with successful repair, which usually requires a synthetic patch, and good clinical outcome, the location of the child's liver is central, directly beneath the patch, rendering it more vulnerable to trauma.
- **Race:** No geographic or racial predilection exists for omphalocele or gastroschisis.
- **Sex:** The male-to-female ratio is 1.5:1.

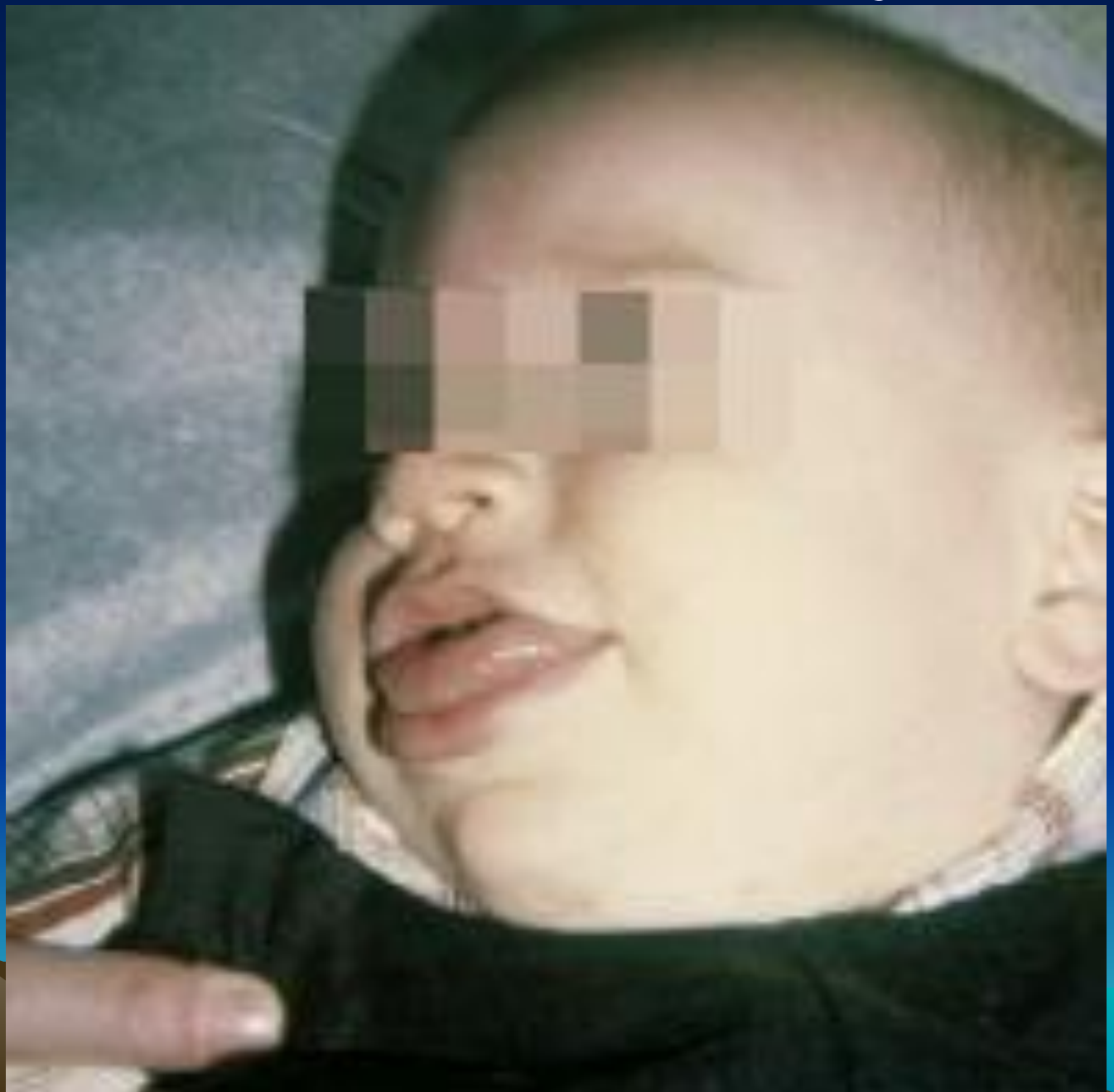
CLINICAL

- **Physical:**
- **Omphalocele**
  - In babies with **omphaloceles,** the size of the abdominal wall defect ranges from 4-12 cm, and the location of the defect may be central, epigastric, or hypogastric.
  - Although the ease of surgical reduction and repair correlate with the size of the abdominal wall defect, a small omphalocele is no guarantee of an uncomplicated clinical course. Associated genetic syndromes involving multiple organ systems, or abnormalities of the intestine, such as an atresia or a patent omphalomesenteric duct, are potential problems.
  - With a large omphalocele, dystocia may occur and result in injury to the baby's liver; hence, a **cesarean section** may be indicated.
  - The omphalocele sac is usually intact, although it may be **ruptured in 10-20%** of cases. Rupture may occur in utero or during or after delivery.

CLINICAL

- Babies with the Beckwith-Wiedemann syndrome (ie, macroglossia, gigantism) have large, rounded facial features, hypoglycemia from hyperplasia of the pancreatic islet cells, and visceromegaly. They may have genitourinary abnormalities, and they are at risk for development of Wilms tumors, liver tumors (hepatoblastoma), and adrenocortical neoplasms.
- Pentalogy of Cantrell describes an epigastric omphalocele associated with a cleft sternum and anterior diaphragmatic hernia (Morgagni), cardiac defects (eg, ectopia cordis, ventricular septal defect [VSD]) and an absent pericardium.
- Giant omphaloceles have large central or epigastric defects. The liver is centrally located and entirely contained within the omphalocele sac. The abdominal cavity is small and undeveloped, and operative closure is very difficult. The thoracic cavity is also small. Associated pulmonary hypoplasia or restrictive lung disease may be present.

Beckwith-Wiedemann syndrome

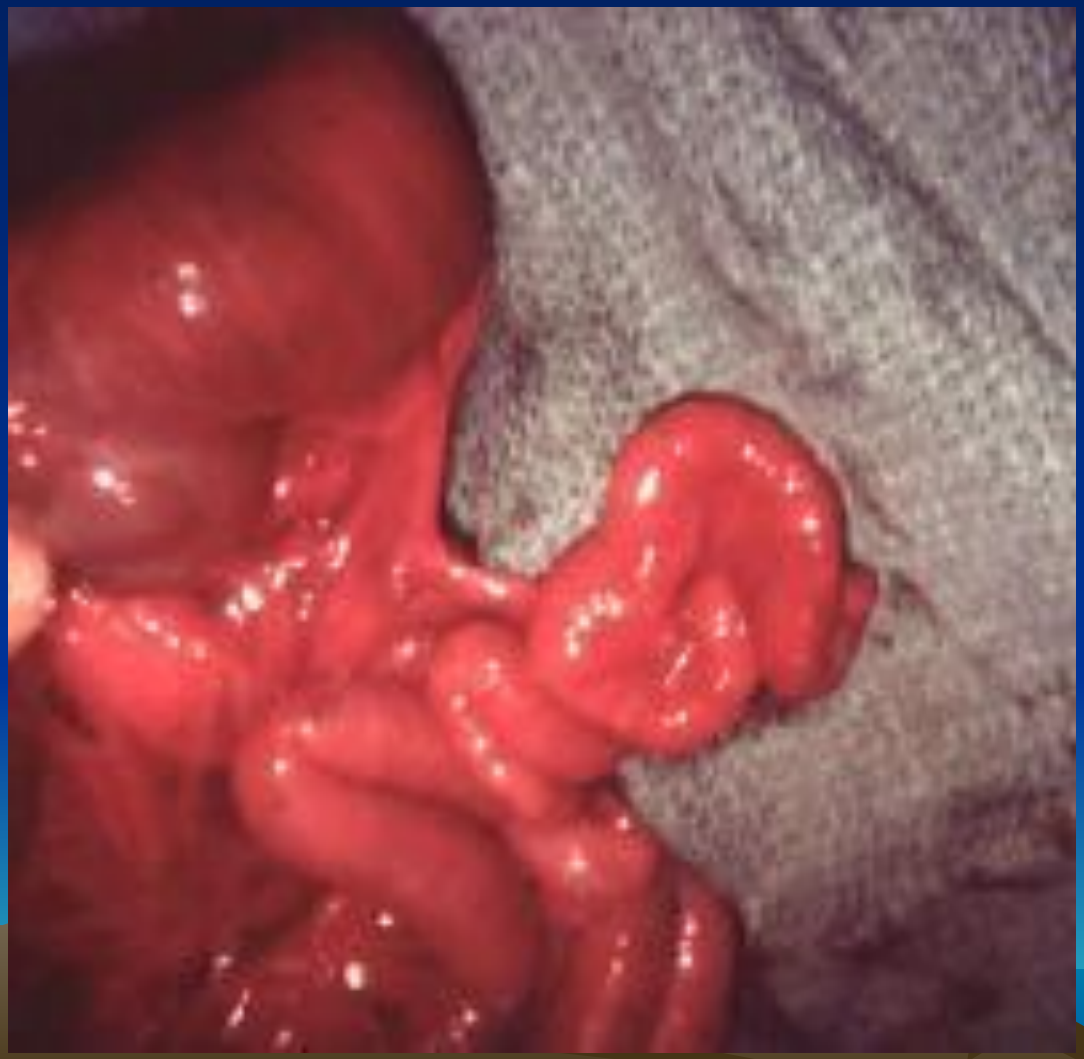


Baby with pentalogy of Cantrell

CLINICAL- Gastroschisis

- The defect is fairly uniform in size and location; a 5-cm vertical opening to the left of the umbilical cord.
- However, the extent of intestinal inflammation and resultant edema and turgor greatly affect reduction and closure of the abdomen. Inflammation so distorts the bowel's appearance that it may be difficult to determine if associated intestinal atresia is present.
- Once reduction and closure is obtained, inflammation resolves, and the intestine softens and regains a normal appearance.
- Correction of associated intestinal atresia is best left until this time, usually 3 weeks after the first operative procedure.
- Intestinal dysfunction takes longer to normalize, from 6 weeks to several months.
- If gastroschisis is identified, perform serial examinations to assess intestinal integrity and amniocentesis to monitor lung maturity.

Baby with gastroschisis and associated intestinal atresia.



baby with gastroschisis and colon atresia.

Causes:

- Factors associated with **high-risk pregnancies**, such as maternal illness and infection, drug use, smoking, and genetic abnormalities, also are associated with the birth of babies with omphalocele and gastroschisis.
- These factors contribute to **placental insufficiency** and the birth of small for gestational age (SGA) or premature babies, among whom gastroschisis and omphalocele most commonly occur.
- **Folic acid deficiency, hypoxia, and salicylates** have caused laboratory rats to develop abdominal wall defects, but the clinical significance of these experiments is conjectural.
- Certainly, **elevation of maternal serum alpha-fetoprotein** (MSAFP) warrants investigation by high-resolution sonography to determine if any structural abnormalities are present in the fetus.
- If such abnormalities are present and associated with an omphalocele, perform **amniocentesis** to check for a genetic abnormality.
- **Polyhydramnios** suggests fetal intestinal atresia, and this possibility should be investigated by ultrasound. Ideally, such information will prompt referral to a tertiary care facility, where the infant can receive expeditious specialty care.

DIFFERENTIALS

- **Other Problems to be Considered:**
- In babies with omphalocele, a **35-80%** incidence of other clinical problems is seen.
- These include **congenital heart disease**, **cleft palate**, and **musculoskeletal and dental occlusion abnormalities**.
- **Patent omphalomesenteric duct** and **small bowel atresias** may occur in babies with umbilical cord hernias where the size of the defect is smaller than 4 cm.

  Incidence of associated **chromosomal abnormalities** is 10-40%. These include trisomies 12, 13, 15, 18, and 21.

  Babies with gastroschisis, in which the incidence of chromosomal anomalies is less than 5 percent, may have **gastroesophageal reflux** disease or **Hirschsprung disease** in addition to abnormal intestinal absorption and motility.

WORKUP

- **Lab Studies:**

- Maternal serum alpha-fetoprotein
  - Prenatal diagnosis of abdominal wall defects can be made by detection of an elevation in MSAPF.
  - MSAPF levels are greater in gastroschisis than in omphalocele.
  - MSAPF also is increased in spina bifida, which additionally demonstrates an increased ratio of acetylcholinesterase and pseudocholinesterase.

WORKUP

- **Imaging Studies:**

- Fetal sonography may detect a genetic abnormality, with identification of a structural marker of the karyotypic abnormality.
- Fetal echocardiography also may identify a cardiac abnormality.
- Confirm positive findings suggestive of a genetic abnormality by amniocentesis.
- If serial ultrasounds show dilatation and thickening of the intestine in a baby with gastroschisis, and if lung maturity can be verified by amniocentesis, delivery is induced.

TREATMENT

- **Medical Care:**
- Intestinal inflammation
  - Intestinal inflammation may occur with either gastroschisis or ruptured omphalocele.
  - The eviscerated intestine may be either normal or abnormal in structure and function. The degree of abnormality depends upon the extent of the inflammatory and ischemic injury, manifested by shortened length and surface exudate (peel), which is related to the composition and duration of the intestine's exposure to the amniotic fluid and fetal urine.
  - Inflamed intestine is thick and edematous, the loops of bowel are matted together, and the mesentery is congested and foreshortened.
  - Histologically, atrophy of the myenteric ganglion cells is seen.
  - The intestine is dysmotile, with prolonged transit time and decreased absorption of carbohydrate, fat, and protein. These deleterious effects remit as the inflammation resolves, usually in 4-6 weeks. During this time, total parenteral nutrition (TPN) is required.

TREATMENT

- **Intact omphalocele**
  - Usually, neonates with intact omphalocele are in no distress, unless associated pulmonary hypoplasia is present.
  - Examine the baby carefully to detect any associated problems, such as Beckwith-Wiedemann syndrome, chromosomal abnormalities, congenital heart disease, or other associated malformations. Give nothing by mouth (NPO) pending operative repair.
  - Administer maintenance IV fluids, and cover the omphalocele sac with sterile saline-soaked gauze and with plastic wrap, using sterile technique. As an alternative, the baby's lower torso may be placed in a bowel bag.

TREATMENT

- The omphalocele should be supported to avoid excessive traction to the mesentery.
- Give prophylactic antibiotics preoperatively, because of the possibility of an associated intestinal anomaly.
- Closure of a small or moderate size omphalocele usually is accomplished without difficulty.
- A ruptured omphalocele is treated like gastroschisis.
- Closure of a giant omphalocele that contains the liver can be very challenging.

TREATMENT

- **Gastroschisis**
  - Respiratory distress in neonates with gastroschisis may respond to gastric decompression, although endotracheal intubation may still be needed.
  - Fluid, electrolyte, and heat losses must be minimized and corrected. Administer an intravenous fluid bolus (20 mL/kg LR), followed by 5% dextrose ¼ NS at 2-3 times the baby's maintenance fluid rate.
  - The baby should be placed under a radiant heater, and the exposed intestines should be covered with plastic wrap and supported to avoid excessive traction on the mesentery. As an alternative, the baby's lower torso may be placed in a bowel bag.
  - Insert a urinary catheter to monitor urine output and facilitate reduction of the herniated viscera by avoiding bladder distention.
  - Administer antibiotics to prevent infection, since neonates have low levels of circulating immunoglobulin G (IgG).
  - Place a central venous line to provide parenteral nutrition, thereby minimizing protein loss during the period of gastrointestinal dysfunction.

TREATMENT-Surgical Care:

- **Omphalocele**
  - Ambroise Pare, the 17th-century French surgeon, accurately described omphalocele and the dire consequences of opening the sac to attempt surgical closure. Certainly, his admonition encouraged conservative treatment, ie, squeezing the sac to effect reduction of the herniated viscera or painting the sac with escharotic agents to promote epithelization.
  - The problem with this approach is that it is slow. During this time the sac may rupture, resulting in a wound infection. Even if complications do not occur, the healing of such a large wound exacts a significant metabolic and nutritional toll.

TREATMENT-Surgical Care:

- Healing may be hastened by surgically mobilizing skin flaps sufficient to cover the omphalocele sac, thereby obtaining closure of the abdominal wall defect in a way comparable to closing a burn wound with skin grafts (Gross technique). This, however, results in the creation of a ventral hernia.
- In 1967, Schuster developed a technique that may be used in the initial treatment of a baby with a giant omphalocele or in correcting the ventral hernia created by skin flap closure. An incision is made along the skin-sac junction of the abdominal wall defect, which is enlarged in the midline. The anterior rectus fascia is exposed from the xiphoid to the pubis, and Teflon sheets are sutured to its medial edge. The Teflon sheets are then closed over the omphalocele sac and gradually tightened, approximating the rectus muscles over the abdominal viscera.

TREATMENT-Surgical Care

- **Gastroschisis**
  - In 1969, Allen and Wrenn adapted Schuster's technique for treatment of gastroschisis.
  - Silastic sheets are sutured to the full thickness of the enlarged abdominal wall defect and closed over the eviscerated intestine, whose reduction is facilitated by stretching the abdominal musculature, emptying the stomach and bladder, and manually evacuating the colon.
  - A major factor in the reduction of the extruded viscera is resolution of intestinal inflammation, which results in a change from a rigid, congealed mass of bowel to soft, pliable loops of intestine, which squeeze into the abdominal cavity.
  - Too tight a closure of the abdominal wall must be avoided, for this limits excursion of the diaphragm and necessitates increased inspiratory pressure to compensate for the increase in airway resistance. In general, peak inspiratory pressures (PIPs) higher than 25 mmHg should be avoided. High-frequency oscillatory ventilation may be an alternative to conventional ventilation if intraabdominal pressures are markedly increased.

TREATMENT-Surgical Care

- In addition, tight closure of the abdominal cavity impedes venous return to the heart, compromising cardiac output and decreasing renal blood flow and glomerular filtration rate. Renal vein thrombosis and renal failure may ensue.
- Diminished mesenteric blood flow may facilitate the development of necrotizing enterocolitis.
- In order to avoid these problems, techniques have been developed to monitor central venous pressure (CVP), intraabdominal pressure, intravesicular pressure, and intragastric pressure (which should not exceed 20 cm of water).

- **Consultations:**
- Neonatologists and pediatric surgeons usually care for babies with these anomalies.
- Consult with cardiology, pulmonology, gastroenterology, and genetics, as indicated.

Diet:

- Babies with omphalocele usually do not require special formulas; their intestines are typically normal, with the exception of occasional atresias, which, in the author's experience, are located in the distal ileum and are not associated with short gut.
- Babies with **gastroschisis**, on the other hand, typically require special elemental, crystalline amino acid, or protein hydrolysate formulas with nonlactose carbohydrate and medium-chain triglycerides because of the associated gut inflammation and resultant tendency towards substrate malabsorption and allergy.
- Babies with short gut syndrome absorb medium-chain triglycerides more readily than long-chain triglycerides; however, the latter are more valuable with regard to gut adaptation.

Activity:

- A child with a repaired giant omphalocele has an epigastric liver. In this location, the liver is more vulnerable to trauma. Avoidance of contact sports is prudent.

FOLLOW-UP

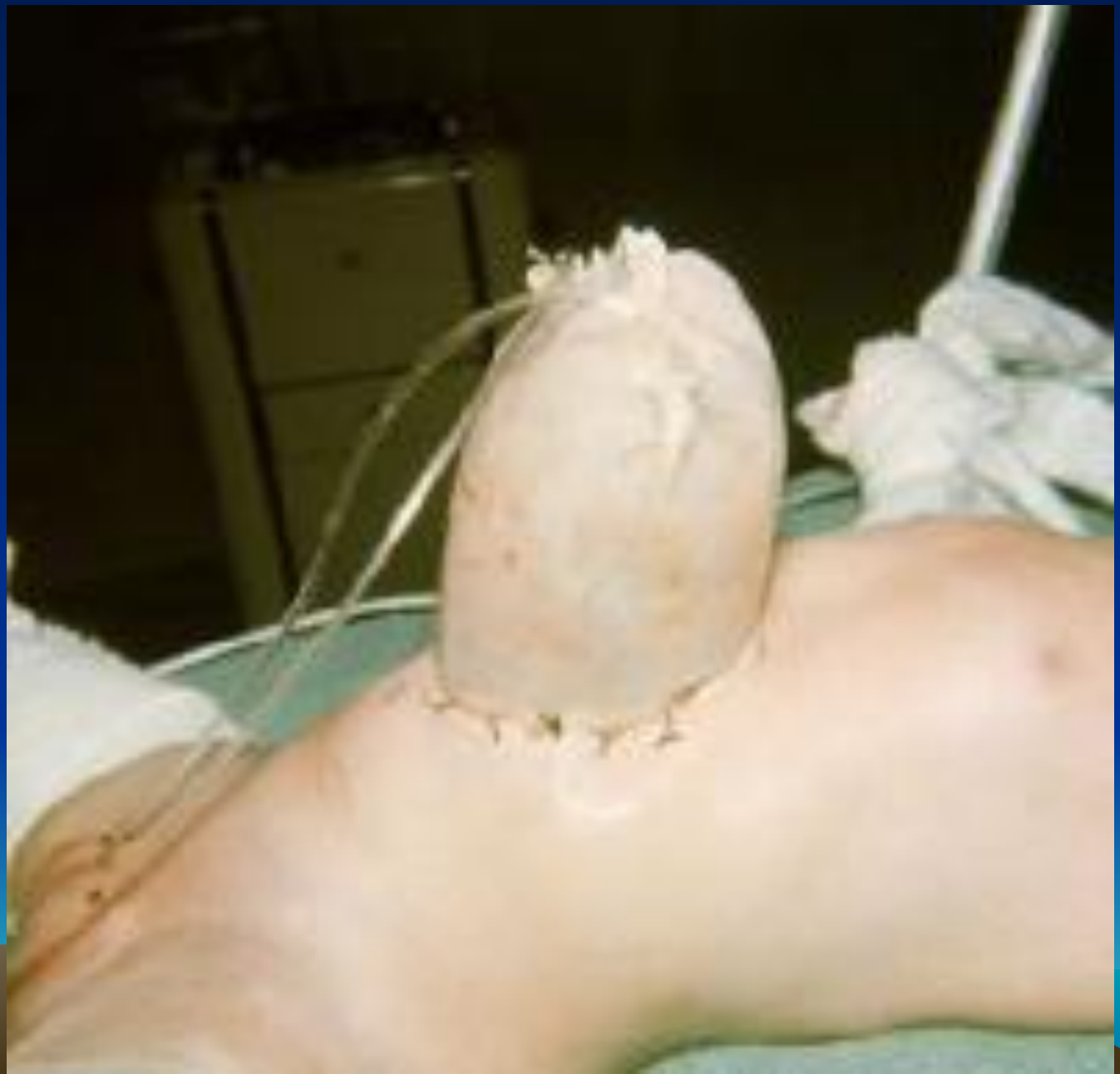
- **Further Inpatient Care:**

- **Omphalocele**
  - Babies with omphalocele usually have rapid return of intestinal function after surgical repair, even if intestinal atresia occurs concomitantly, because no associated gut inflammation is present.
  - Babies with giant omphaloceles usually have a protracted hospital course; and overall morbidity and mortality is higher for these patients. Multiple procedures are necessary to obtain closure of the abdominal wall defect.
  - Respiratory compromise may complicate the repair and require prolonged support and possibly a tracheotomy. Ventilator management, tracheotomy care, and, ultimately, decannulation require close cooperation by the neonatologist, pulmonologist, and pediatric surgeon.

FOLLOW-UP

- **Gastroschisis**
  - Even if primary closure of the abdominal wall defect is obtained, a period of several weeks of intestinal dysfunction (ileus) usually follows, as a result of associated gut inflammation. In this situation, parenteral nutrition is essential, followed by the gradual introduction of enteral feedings. Continuous drip feedings usually are tolerated optimally.
  - If reduction of the herniated intestine requires the use of a **silo**, it usually is removed within 5-7 days. The period of ileus follows, during which the baby requires parenteral nutrition until the gradual return of intestinal function. If this expected recovery does not occur within 3-4 weeks, intestinal obstruction is presumed, and a contrast study is obtained to document intestinal transit.
  - If intestinal obstruction is present, a laparotomy must be performed.

Silo closure of a baby with gastroschisis.



FOLLOW-UP

- **Further Outpatient Care:**

- After hospital discharge, babies require close follow-up care to assess growth and weight gain.
- Patients usually have gastroesophageal reflux and may require medical therapy, but fundoplication should not be necessary.
- Hirschsprung disease (aganglionic megacolon) also may occur. Physicians should be alert to a history of constipation.

FOLLOW-UP

- Transfer:

- The best way to treat the exposed intestines of a baby with gastroschisis who is being transported to a tertiary center includes the application of a moist lap pad. The moist lap pad is placed over the intestines and held directly over the abdominal wall defect with dry Kerlix wrap applied around the baby's torso including the extruded intestine. This prevents traction upon the mesentery. A warm, wet, lap pad placed in a bowel bag with the eviscerated intestine soon becomes a cold, wet, lap pad.

- The patient’s condition improved dramatically once closure of the abdominal cavity was achieved. Again, the author tried to wean him from the ventilator, but his copious secretions and episodes of high fever and drenching sweats prevented this. Finally, it was determined that the patient was experiencing narcotic withdrawal. He had been postoperative for so long, and narcotics had been used liberally to provide postoperative pain relief.

Prognosis:

- **Omphalocele**
  - Prognosis is dependent upon the severity of the associated problems. Babies with omphalocele are considerably complex, with involvement of many other organ systems.
  - Even giant omphaloceles can be closed, although multiple procedures may be necessary.
  - The limiting factor for many of these babies, however, is their diminutive thoracic cavities and associated pulmonary hypoplasia and resultant chronic respiratory failure. Even so, lung growth and development continue well into childhood, encouraging optimism regarding the ultimate prognosis.

Prognosis

- **Gastroschisis**
  - Prognosis is dependent mainly upon severity of associated problems, including prematurity, intestinal atresia, short gut, and intestinal inflammatory dysfunction.
  - Many pediatric surgeons believe that prognosis has improved because of maternal ultrasound diagnosis and monitoring, which leads to expeditious delivery of babies at tertiary centers.
  - Years ago, obtaining primary closure of a baby with gastroschisis was unusual. Usually, it was necessary to use a silo. Now, primary closure is commonly attained.

Patient Education:

Instruct parents regarding the significance of bilious (green) vomiting, since these babies may develop adhesive small bowel obstruction or midgut volvulus.

- Inform parents that their child's appendix is probably in an unusual location and that a CT scan may be the most reliable way to diagnose acute appendicitis.

Special Concerns:

- Prenatal care and planning
  - With increased availability of sonography, prenatal diagnosis is more frequent.
  - Diagnosis of omphalocele mandates further workup to determine if an associated genetic abnormality is present, in which case appropriate counseling is necessary.
  - When gastroschisis is diagnosed, perform serial examinations to detect signs of intestinal injury (decreased peristalsis or distension).
  - Provide the baby's parents with information concerning the anomaly before delivery. Also, optimal management requires that the obstetrician understands the particular needs of these babies and ensures that they are delivered in a facility where neonatal, pediatric anesthesia, and pediatric surgery services are available.

Comparison of Gastroschisis and Omphalocele

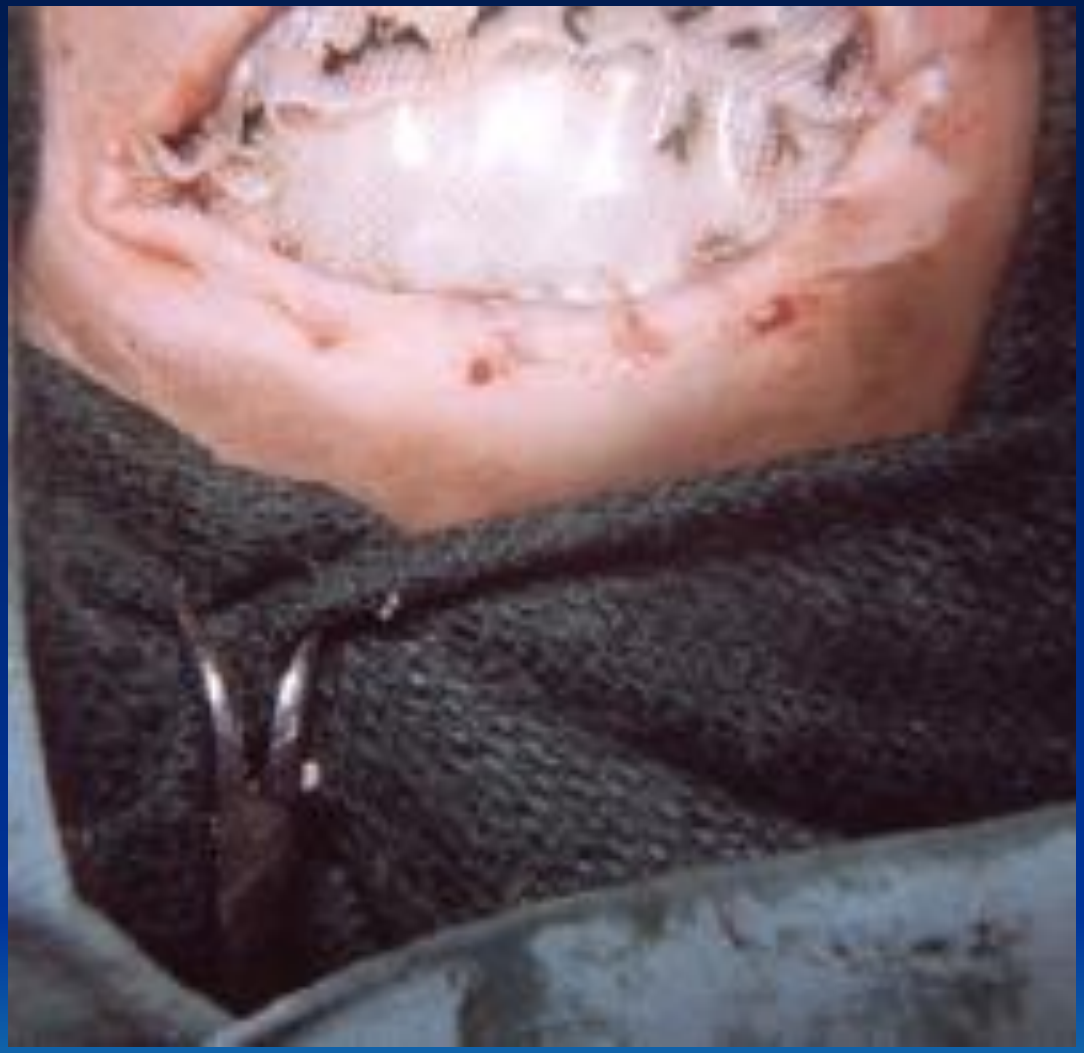
	Omphalocele	Gastroschisis
Incidence	1:6,000-10,000	1:20,000-30,000
Prenatal Diagnosis	Ultrasound (should be followed up with fetal echocardiography and amniocentesis)	Ultrasound
Delivery	Vaginal or C-section	C-section (debated)
Covering Sac	Present, may be torn	Absent
Fascial Defect	Small or large	Small (may lead to vascular compromise of herniated gut)
Cord Attachment	Onto the sac	Onto the abdominal wall

Comparison of Gastroschisis and Omphalocele

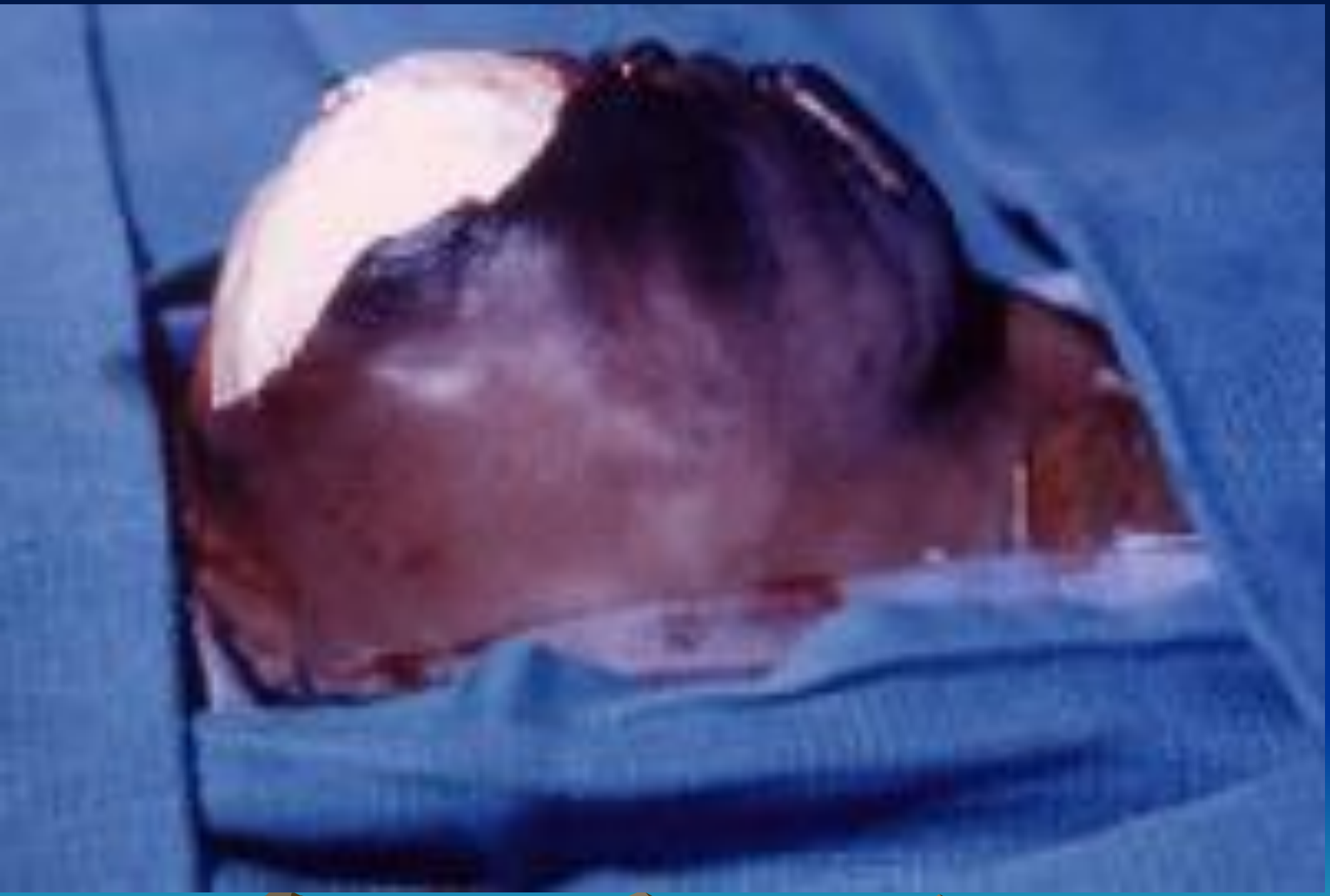
	Omphalocele	Gastroschisis
Herniated Bowel	Normal	Edematous, matted
Other Organs	Liver often out in the sac	Never out
Prematurity (%)	10-20%	50-60%
IUGR	Less common	Common
Necrotizing Enterocolitis (%)	Only if sac is ruptured	18%

Comparison of Gastroschisis and Omphalocele

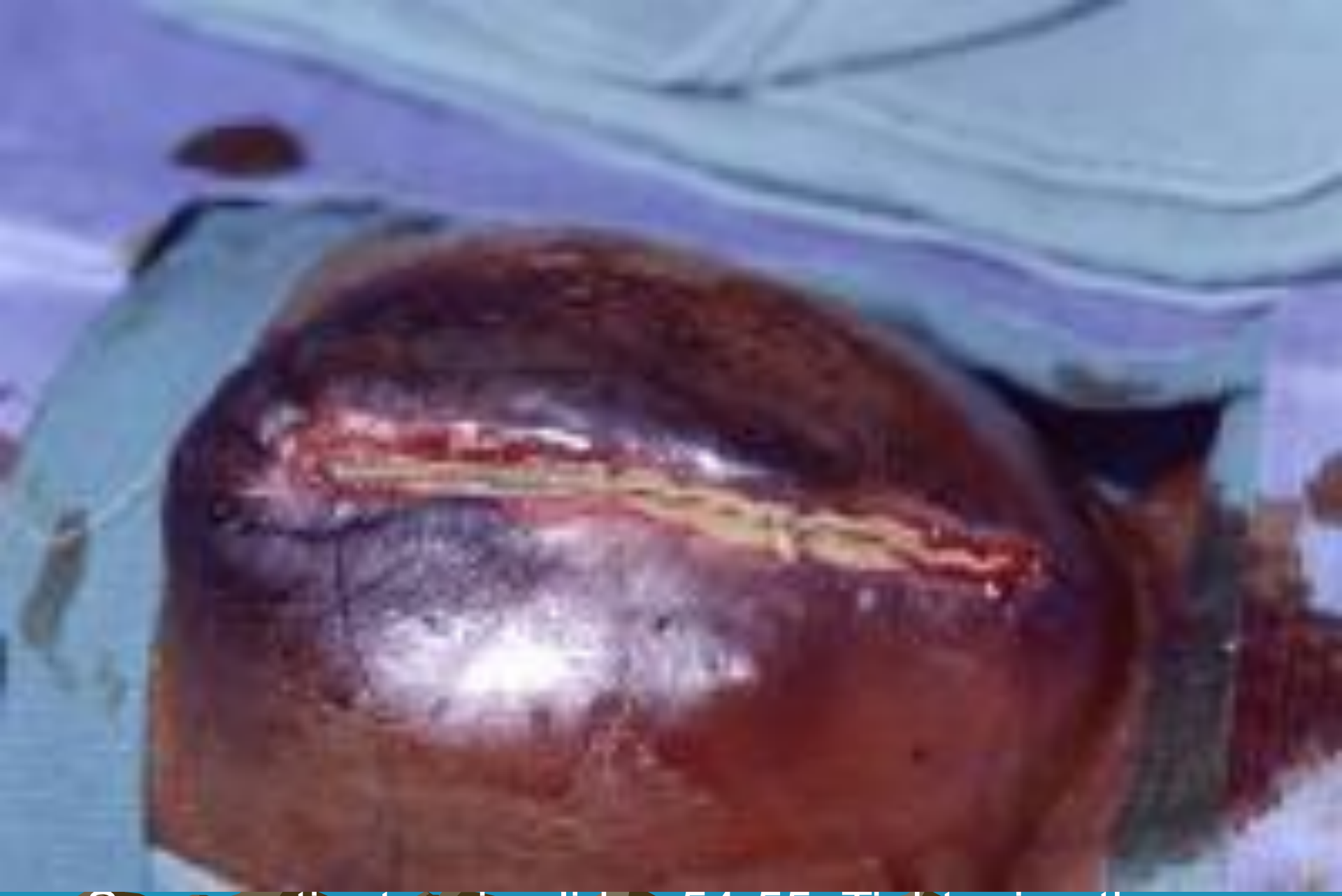
	Omphalocele	Gastroschisis
Malabsorption	Only if sac is ruptured	Common
Associated Anomalies (%)	45-55% (Taeusch), 67% (Avery)	10-15% (Taeusch)
-- GI	37% (midgut volvulus, Meckel's diverticulum, intestinal atresia, duplication)	18% (typically malrotation, atresias)



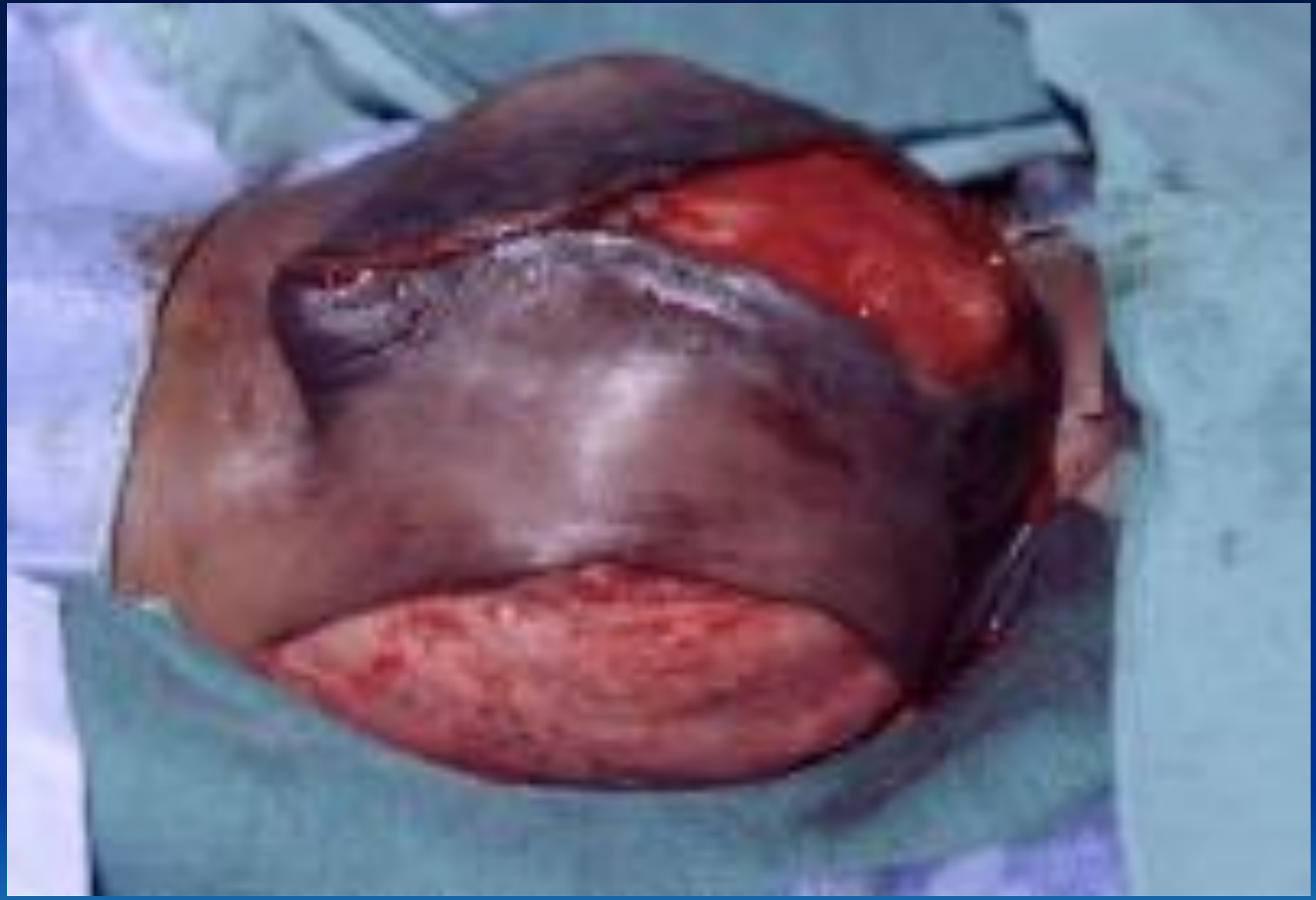
- Completed reduction of the bowel contained within the silo; the silo is about to be removed and the abdominal wall closed.



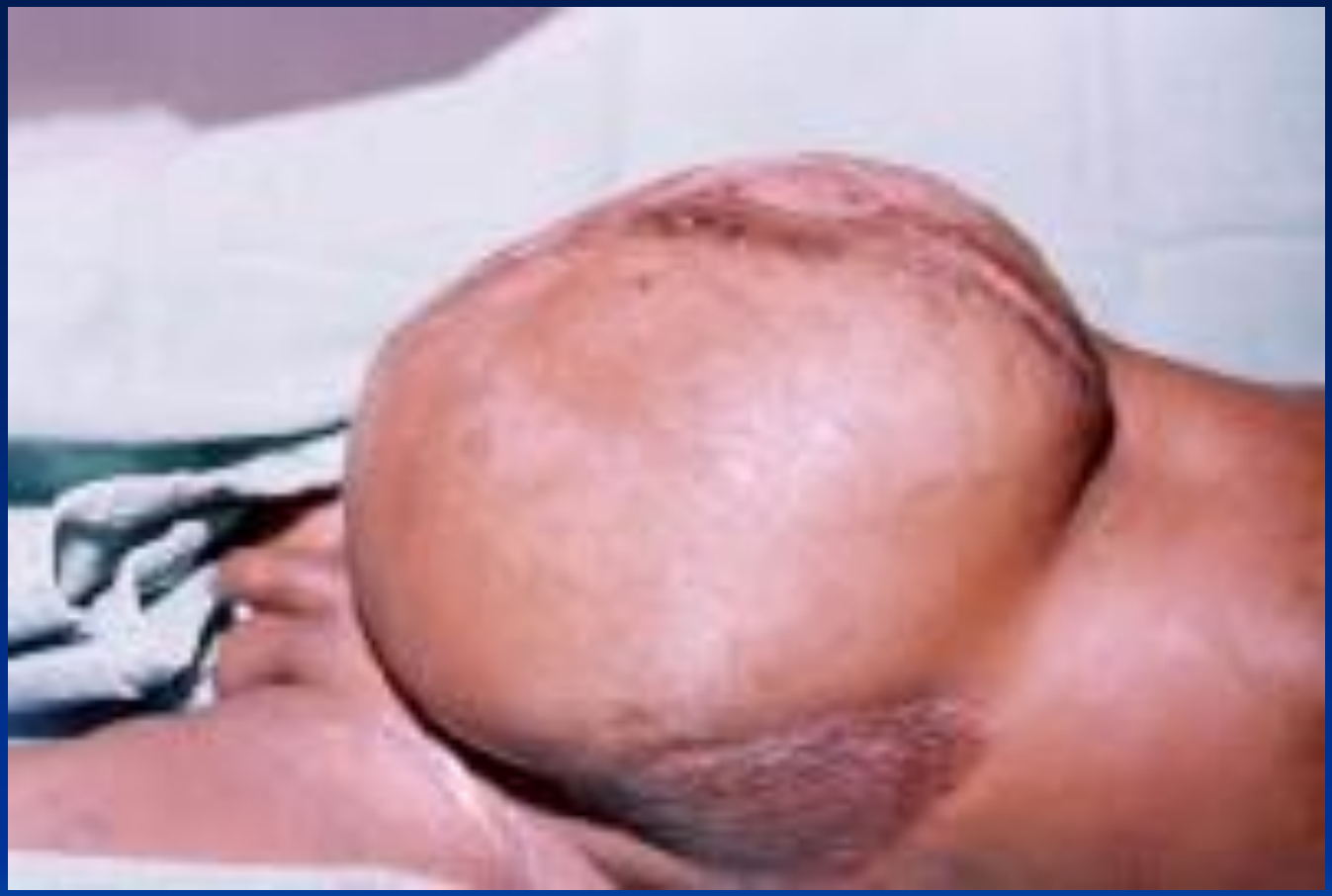
- Same patient as in slide 54. Closure of the giant omphalocele using a synthetic patch.



- Same patient as in slides 54-55. Tightening the abdominal wall closure



- Same patient as in slides 54-56. Flank flaps were used to close the giant omphalocele in the baby whose patch became infected.



- Omphalocele and gastroschisis. Same patient as in slides 54-57. The flank wounds were skin grafted and closure of the giant omphalocele obtained