



Umbilical Anomalies and Exomphalos: A Review of Current Practice

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Abstract

Objective: To review current practice in the diagnosis and management of umbilical anomalies, with emphasis on exomphalos.

Design: Narrative review of published literature and clinical guidelines.

Results: Umbilical anomalies range from minor hernia of the cord to giant exomphalos. Antenatal diagnosis is feasible with ultrasound and MRI. Management strategies vary from primary closure to staged silo reduction and conservative epithelisation. Prognosis depends on defect size, liver involvement, and associated anomalies.

Conclusions: Exomphalos remains a major neonatal surgical challenge. Current practice balances surgical innovation with conservative approaches, but consensus guidelines are lacking.

Keywords: Umbilical Anomalies; Exomphalos; Omphalocele; Congenital Abdominal Wall Defects; Neonatal Surgery

Summary Box

What is already known on this topic

- Exomphalos is a congenital abdominal wall defect with significant morbidity and mortality.
- Management strategies vary widely, with no universal consensus.

What this study adds

- Synthesises current evidence on classification, diagnosis, and management.
- Highlights evolving practices such as compression dressing and conservative minimal access external silo.
- Identifies gaps in consensus guidelines and future research needs.

Introduction

The umbilicus is the remnant of the vital connection between mother and fetus, its proper development is essential. The umbilicus is a central landmark of the anterior abdominal wall and a site of embryological convergence. Anomalies of the umbilicus are relatively common in pediatric surgical practice and range from common minor benign self-limiting lesions to critical conditions requiring urgent surgical intervention. Understanding their sound embryology, clinical spectrum, and management is essential for pediatric surgeons and ultimately improving perinatal outcomes.

Umbilical anomalies represent a spectrum of congenital conditions, ranging from benign variants to life-threatening abdominal wall defects. Exomphalos (omphalocele) is the most clinically significant anomaly, with an incidence of 1 in 4,000–6,000 live births. It is frequently associated with chromosomal anomalies (trisomy 13, 18) and syndromes such as Beckwith–Wiedemann. This review synthesises current practice in diagnosis and management, highlighting controversies and

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Table 1 Classification of Umbilical anomalies by timing, type			
Timing	Type	Anomaly	Clinical relevance
Vascular	Vascular	Single umbilical artery (SUA) Fetal echo	May indicate cardiac, renal, or chromosomal anomalies: Fetal check
	Mechanical	Velamentous cord insertion (VCI)	Risk of growth restriction, preterm risk
Structural	Structural	Omphalocele	Life-threatening fetal hemorrhage risk during labor
	Abnormal cord length	Umbilical cord length	Long, predisposing to knot Short movement restriction, abrasion
Structural	Structural	Omphalocele	Midgut fails to return; sac covered herniation
	Postnatal	Umbilical hernia risk Observe/repair	Common, responds to silver nitrate Genetic counsel

Timing schematic

Antenatal vigilance cues

Risk-to-action pathways

SUA → Cardiac risk
Vasa previa Fetal echo
Omphalocele Surgical counsel
Omphalitis Observe/repair

Vascular

Mechanical

Healing

Infective

Immunologic

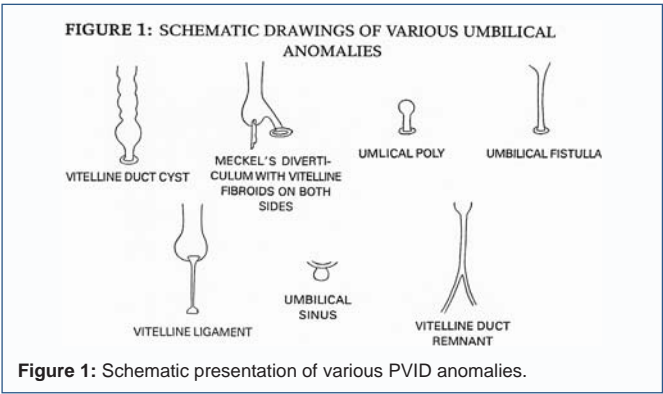
Table 1: Composite classification of umbilical anomalies by timing, type, clinical relevance, and management.

Panel A presents a structured matrix categorizing umbilical anomalies by timing (antenatal vs postnatal) and type (vascular, mechanical, structural, healing, infective, immunologic), with associated clinical relevance and first-line management cues.

Panel B illustrates timing-based detection triggers: antenatal anomalies are typically identified via prenatal ultrasound, while postnatal anomalies require clinical vigilance for signs such as discharge, mass, or delayed cord separation.

Panel C maps risk-to-action pathways, linking each anomaly to its potential complications and recommended initial intervention.

Panel D provides an icon glossary for anomaly types, enhancing visual decoding and accessibility.



- Umbilical sinus: Blind tract from umbilicus toward ileum.
- Umbilical cyst: Cystic remnant along VID tract.
- Meckel’s diverticulum with umbilical connection as latent VID or separate lesion (Figure 1).

B. Umbilical Lesions

- Umbilical granuloma: Overgrowth of granulation tissue after cord separation.
- Umbilical polyp: Remnant of intestinal or gastric mucosa.
- Infected umbilicus (omphalitis).

C. Urachal Anomalies

- Patent urachus: Urine discharge from umbilicus.
- Urachal sinus: Blind tract toward umbilicus.
- Urachal cyst: Midline cyst between umbilicus and bladder.
- Urachal diverticulum: Outpouching from bladder dome.

D. Other Rare Anomalies

- Umbilical hernia/exomphalos.
- Supernumerary umbilicus.
- Umbilical endometriosis (rare in adolescents).

Clinical Presentation

- Discharge: feculent (VID), mucoid (polyp), purulent (granuloma/omphalitis), or urinary (urachus).
- Mass: cystic swelling, polyp, granuloma.
- Pain/fever: infection or abscess formation.
- Associated anomalies: Meckel’s diverticulum, bladder anomalies.

Diagnostic Evaluation

- Clinical examination: nature of discharge, presence of mass.
- Ultrasound abdomen: cysts, sinus tracts, urachal anomalies.
- Fistulography/sinogram: delineates patent tracts.
- CT/MRI: complex or recurrent cases.
- Histopathology: differentiates granuloma vs polyp (Figure 2).

Management Strategies

- Umbilical granuloma: topical silver nitrate cauterization; surgical excision if resistant.

evolving approaches.

Methods

This narrative review was conducted using PubMed, Embase, and guideline repositories (NHS Scotland, RCPCH, AAP). Search terms included umbilical anomalies, exomphalos, omphalocele, neonatal surgery, and current practice. Articles published between 2000–2025 were included. Case series, guidelines, and systematic reviews were prioritised.

Embryology of the Umbilicus and Exomphalos

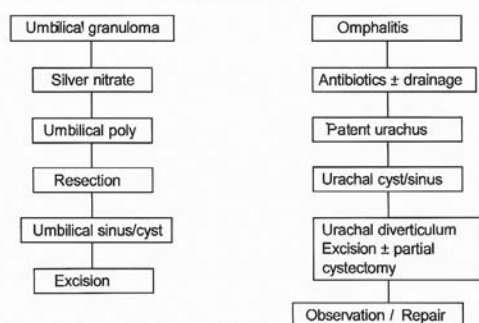
- Formed by the convergence of the vitellointestinal duct, urachus, and umbilical vessels.
- Normally obliterates by the 7th week of gestation.
- Persistence or abnormal involution leads to congenital anomalies.

Classification of Umbilical Anomalies

See Table 1.

A. Vitellointestinal Duct (VID) Anomalies

- Patent VID: Persistent communication between ileum and umbilicus → feculent discharge. Or persistence of VID as a obliterated fibrous cord->intestinal obstruction.

FIGURE 2: SCHEMATIC DIAGRAM OF MANAGEMENT PATHWAYS**Figure 2:** Management pathways flowchart for Umbilical Anomalies.

This flowchart maps each anomaly to its standard treatment:

- Granuloma → Silver nitrate (or excision if resistant)
- Umbilical polyp → Surgical excision
- Patent VID → Segmental resection
- Umbilical cyst/sinus → Excision
- Urachal cyst → Excision
- Omphalitis → Antibiotics ± drainage
- Umbilical hernia → Observation or repair.

- Omphalitis: antibiotics, drainage if abscess.
- Umbilical hernia: observation (most resolve by 2–3 years); repair if persistent or large (Figure 3).

Complications and Prognosis

- Infection: recurrent omphalitis, abscess.
- Persistent discharge: social stigma, delayed diagnosis.
- Malignancy risk: rare, but urachal remnants may predispose to adenocarcinoma in adulthood.
- Prognosis: excellent with timely diagnosis and complete excision.

Key Points

- Differentiate granuloma vs polyp (histology).
- Patent VID → feculent discharge; Patent urachus → urinary discharge.
- Silver nitrate is first-line for granuloma.
- Always excise urachal anomalies due to infection/malignancy risk.
- Meckel's diverticulum association should be remembered.

Exomphalos

Introduction

Umbilical abdominal wall defect anomalies include hernia of the umbilical cord, exomphalos minor, intermediate, major, and giant. Exomphalos is the most clinically significant anomaly. It is frequently associated with chromosomal anomalies (trisomy 13, 18) and syndromes such as Beckwith–Wiedemann. It results from a failure of the normal physiological return of the midgut to the abdominal cavity during the 6th to 10th week of gestation.

Definition

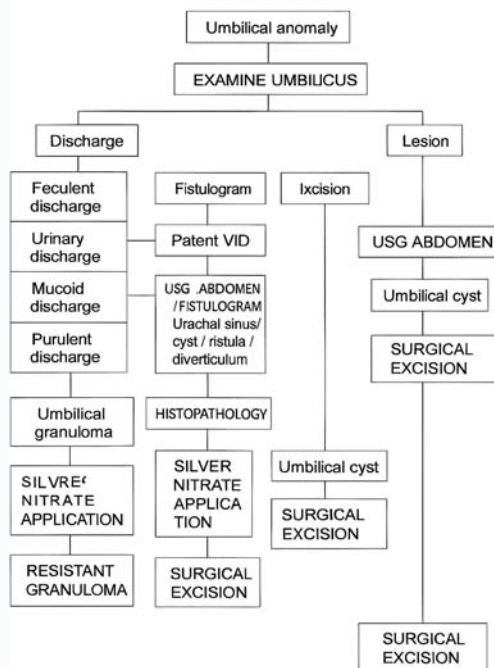
Exomphalos, also known as omphalocele, is a congenital anterior abdominal wall defect resulting in the herniation of abdominal viscera into the base of the umbilical cord. The herniated contents are covered by a translucent avascular membrane, derived from the amnion and peritoneum, which may contain intestines, liver, spleen, and ovaries (Figure 4 and 5).

Aetiology and Pathophysiology

The exact cause is unknown, but it is thought to be due to a failure of the lateral embryonic folds to fuse at the midline or a defect in the development of the umbilical ring musculature.

Key pathophysiological features include:

- **Viscero-abdominal disproportion (VAD):** The abdominal cavity is often underdeveloped (hypoplastic) because the viscera grew outside it, making immediate reduction challenging, especially in large defects.
- **Intestinal anomalies:** The intestine typically has non-rotation or malrotation.
- **Pulmonary hypoplasia:** This is a major concern, particularly in large exomphalos cases associated with other anomalies, contributing significantly to morbidity and mortality.
- **Sac rupture:** While the sac is usually intact, rupture can occur,

FIGURE 3: SCHEMATIC DIAGRAM OF MANAGEMENT ALGORITHM**Figure 3:** Management Algorithm for Umbilical Anomalies.

Decision tree guiding diagnosis and treatment based on clinical presentation. Discharge types (feculent, urinary, mucoid, purulent) and lesion types (granuloma, cyst) are mapped to appropriate investigations and interventions.

- Umbilical polyp: surgical excision; histopathology mandatory.
- Patent VID: segmental ileal resection with end-to-end anastomosis.
- Umbilical sinus/cyst: complete excision.
- Patent urachus: excision of tract with bladder cuff.
- Urachal cyst/sinus/diverticulum: excision ± partial cystectomy.

Figure 4:
SCHEMATIC COMPARISON OF
ABDOMINAL WALL DEFECTS

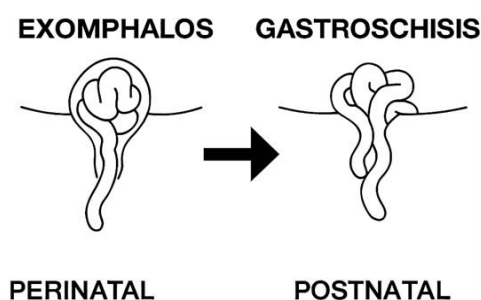


Figure 5. Exomphalos vs Gastroschisis.

Side-by-side comparison of two congenital abdominal wall defects. Exomphalos presents as a central defect at the umbilicus with sac-covered herniation, often associated with syndromes. Gastroschisis is a paraumbilical defect with exposed bowel and no covering sac, typically isolated.

Feature	Exomphalos	Gastroschisis
Defect location	Central, at umbilicus	Paraumbilical (usually right side)
Covering sac	Present (peritoneum + amnion)	Absent – bowel exposed
Herniated contents	Bowel ± liver	Bowel only (liver usually not involved)
Associated anomalies	Common (trisomy 13/18, BWS, cardiac)	Rare
Umbilical cord	Inserts into sac	Separate from defect
Diagnosis	Antenatal ultrasound	Antenatal ultrasound
Management	Protect sac → staged or primary repair	Cover bowel → silo or primary closure

Figure 4: Congenital central and paracentral abdominal wall defects-Schematic presentation of exomphalos and gastroschisis.

Figure 5: Principal differential features exomphalos vs Gastroschisis.

Expanded comparison table of two congenital abdominal wall defects. Exomphalos presents as a central defect at the umbilicus with sac-covered herniation, commonly associated with syndromes. Gastroschisis is a paraumbilical defect with exposed bowel and no covering sac, typically isolated. The table contrasts location, sac presence, herniated contents, associated anomalies, umbilical cord insertion, diagnosis, and management strategies.

increasing the risk of infection and requiring immediate surgical intervention.

Embryology

It results from failure of the midgut to return to the abdominal cavity during embryogenesis (typically around the 10th week of gestation). The defect is central and covered by a membranous sac.

Epidemiology

- **Incidence:** ~1 in 4,000 live births.
- Slight male predominance.
- High association with chromosomal and syndromic anomalies.

Classification: Exomphalos can be classified based on defect size and liver involvement:

See Table 2.

Classification of Exomphalos

Type	Liver in Sac	Defect Size	Severity (VAD)	Associated Malformations
Hernia of Umbilical Cord	None	<2.5 cm	None	None
Exomphalos Minor	None	2.5–5 cm	None	Infrequent
Exomphalos Intermediate	Edge of liver (<25%)	5–7.5 cm	Mild	Few, minor
Exomphalos Major	25–50%	7.5–10 cm	Moderate	Many, major
Giant Exomphalos	>50%	>10 cm	Severe	Several, severe

Table 2: Classification of Exomphalos.

Progressive schematic illustrating the severity spectrum of exomphalos:

- Progressive schematic illustrating the severity spectrum of exomphalos.
- Hernia of the umbilical cord (small, benign defects).
- Minor: No liver, 2.5–5 cm defect, insignificant anomalies.
- Intermediate: Edge of liver (<25%), 5–7.5 cm defect, mild anomalies.
- Major: 25–50% liver, 7.5–10 cm defect, moderate anomalies.
- Giant: >50% liver, >10 cm defect, severe anomalies.

Associated Anomalies

- **Chromosomal:** Trisomy 13, 18, 21.
- **Syndromic:** Beckwith-Wiedemann Syndrome, Pentalogy of Cantrell.
- **Structural:** Cardiac, genitourinary, neural tube defects.

Antenatal and Postnatal Diagnosis

- **Antenatal ultrasound:** Detects sac and herniated contents
Detectable from 12 weeks.
- **MRI:** Defines anatomy and associated anomalies.
- **Amniocentesis:** For karyotyping.
- **ECG and Echocardiogram:** For those with associated cardiac anomalies.
- **Genetic testing:** Essential due to high syndromic association.

Presentation and Associated Anomalies

Exomphalos is typically diagnosed antenatally via ultrasound during the mid-pregnancy scan. Postnatally, it presents as a membrane-covered protrusion from the umbilicus. A thorough search for associated anomalies is crucial, as they significantly impact prognosis. Approximately 50-75% of cases have other congenital anomalies.

- **Chromosomal Abnormalities:** Common, especially Trisomy 13 (Patau's syndrome), Trisomy 18 (Edwards' syndrome), and Beckwith-Wiedemann syndrome (BWS). The recurrence risk for BWS can be up to 50% depending on the underlying genetic cause.
- **Cardiac Defects:** The most frequent associated structural anomalies, including Tetralogy of Fallot, VSD, and atrial septal defects.
- **Gastrointestinal Anomalies:** Intestinal malrotation is common.
- **Midline Syndromes:** Associations with Pentalogy of Cantrell

and OEIS complex (omphalocele, exstrophy, imperforate anus, spinal defects) are rare but important to identify.

Investigation

- **Antenatal:** Serial ultrasound scans to monitor fetal growth and anatomy. Invasive procedures like chorionic villus sampling or amniocentesis are offered to assess for chromosomal anomalies.
- **Postnatal:**
 - o **Clinical examination:** Comprehensive assessment for dysmorphic features and other visible anomalies.
 - o **Echocardiography:** To screen for cardiac defects.
 - o **Renal Ultrasound:** To check for renal anomalies.
 - o **Genetic testing/counselling:** Essential, especially if anomalies are detected antenatally or postnatally.

Discussion

Exomphalos remains a major neonatal surgical challenge. While primary closure is feasible for small defects, larger anomalies require staged or conservative approaches. The debate between early closure and conservative epithelialisation continues, with compression dressing offering promising results. Multidisciplinary antenatal counselling is essential. Future research should focus on standardising management protocols and long-term outcomes.

Current Management Practices

Management is multidisciplinary, involving fetal medicine, neonatology, and paediatric surgery.

Antenatal Care

- Delivery is planned at a specialist centre with a co-located neonatal surgical unit around 38 weeks gestation.
- Mode of delivery is decided based on obstetric factors; Caesarean section is considered if the exomphalos is very large (e.g., >75% of liver in sac) to minimise sac rupture risk.

Postnatal Stabilisation

- The baby is kept warm, and the sac is protected with warm, saline-soaked gauze covered in plastic wrap to prevent hypothermia and fluid loss.
- A nasogastric tube is inserted for decompression.
- Intravenous access is established for fluid resuscitation and Total Parenteral Nutrition (TPN).
- Urgent transfer to a surgical NICU if not delivered at one.

Definitive Management

Management depends on size and the presence of VAD.

Exomphalos Minor

- Primary surgical closure is the standard approach, usually performed within the first few days of life. The sac is excised, contents reduced into the abdomen, and the fascial defect closed primarily.

Exomphalos Major (Giant)

Management is controversial, with no national consensus. Two main strategies exist:

Primary Surgical Closure:

1. Attempted if viscerio-abdominal disproportion is not prohibitive and intra-abdominal pressure can be kept below critical levels (e.g., <20 cmH₂O intravesical pressure).
2. May involve the use of a synthetic patch (e.g., GORE-TEX mesh) if the fascia cannot be closed primarily, with subsequent staged closure.

Staged/Conservative Management:

1. The sac is left intact and treated topically with antiseptic/astringent agents (e.g., silver sulfadiazine, iodine solution, or chlorhexidine) to encourage desiccation and epithelialisation.
2. This non-operative approach allows the abdominal cavity to grow over several months (median 14 months).
3. Definitive repair of the resulting ventral hernia is performed later, typically around 1 year of age.
4. Topical management with silver sulfadiazine is associated with lower mortality and shorter TPN duration compared to staged surgical repair in some series.
5. **Silo technique:** A pre-formed or custom silo (pouch) can be sutured to the fascial edges and the contents gradually reduced into the abdomen over days to a week, either at the bedside or in theatre, followed by formal closure.

Perinatal Management

- **Delivery** in tertiary center.
- **Vaginal delivery** acceptable unless obstetric contraindications.
- **Immediate care at birth:** NICU stabilisation with the protection of the sac, prevention of hypothermia, IV fluids, analgesia +/- antibiotics.

Surgical Management

- **Primary Closure:** Minor and some intermediate cases small defects.
- **Staged Silo Reduction or Repair:** Major and giant cases using silo or tissue expanders.
- **Delayed Closure:** In cases with pulmonary hypoplasia or large abdominal domain mismatch with the conservative epithelialisation: Topical agents applied until sac epithelializes, followed by delayed closure.
- **Graduated compression dressing:** Emerging technique allowing bedside reduction without general anaesthesia.

Postoperative Care

- **Ventilatory support.**
- **Parenteral nutrition.**
- **Monitor for infection,** wound issues, compartment syndrome.

Prognosis and Long-Term Outcomes

- **Prognosis** is primarily determined by defect size, liver involvement, and associated anomalies, particularly cardiac defects and chromosomal abnormalities.
- **One-year survival** for isolated exomphalos is excellent

(around 91%), but drops significantly with multiple anomalies (81%) and is poor with chromosomal abnormalities (27%).

- **Long-term outcomes** for survivors are generally good. Patients may experience feeding issues, prolonged TPN needs, and the need for further operations for ventral hernia repair.
- **Some children require long-term respiratory** support due to pulmonary hypoplasia.
- **Giant exomphalos with anomalies:** High morbidity and mortality.

Controversies and Evolving Practice

The primary controversy in exomphalos management centres on the optimal approach for exomphalos major with the debate between early closure vs conservative delayed closure. There is no national or international consensus on whether primary staged surgical closure (with silo/patch) or conservative non-operative management is superior. Recent systematic reviews suggest that non-operative delayed management might be associated with lower mortality and shorter duration of enteric feeding, but the evidence base remains limited, and management strategies vary between centres. The choice of management often depends on local expertise and the severity of VAD. Compression dressing techniques show promise in reducing morbidity. Multidisciplinary antenatal counselling remains essential.

Key Pearls

1. Differentiate from gastroschisis: location, sac, liver involvement.
2. Always assess for associated anomalies.
3. Surgical strategy depends on size and abdominal domain.
4. Genetic counselling is essential.

Rationale and Contribution

Exomphalos remains a major neonatal surgical challenge, with management strategies ranging from primary closure to conservative epithelisation. Despite advances, there is no universal consensus on best practice. Our review highlights evolving approaches, including compression dressing techniques, and identifies gaps in standardised guidelines. We believe this article will be of interest to clinicians, researchers, and policymakers engaged in neonatal surgery and congenital anomaly care.

Limitations

This review has several limitations that should be acknowledged:

- **Narrative design:** As a narrative review, the synthesis is subject to selection bias. We prioritised guideline documents, systematic reviews, and case series, but did not perform a formal meta-analysis.
- **Heterogeneity of evidence:** The literature on exomphalos management is heterogeneous, with variable definitions of “major” and “giant” defects, differing surgical techniques, and inconsistent reporting of outcomes. This limits direct comparison across studies.
- **Lack of randomised trials:** Most available data derive from retrospective case series or institutional protocols. Randomised controlled trials are lacking, which constrains

the strength of evidence supporting one management strategy over another.

- **Evolving practice:** Emerging techniques such as compression dressing and conservative epithelisation are reported in small series, often without long-term follow-up. Their generalisability remains uncertain.
- **Associated anomalies:** Prognosis is heavily influenced by associated chromosomal and syndromic anomalies. Many studies do not stratify outcomes by these factors, which may confound survival statistics.
- **Publication bias:** Positive outcomes are more likely to be reported, potentially skewing the apparent success of certain interventions.

Despite these limitations, the review provides a comprehensive synthesis of current practice and highlights areas where consensus guidelines and prospective studies are urgently needed.

Future Directions

Several areas warrant further exploration to improve outcomes for infants with umbilical anomalies and exomphalos:

- **Consensus guidelines:** Development of international, evidence-based protocols for classification and management would reduce variability in care and improve comparability of outcomes.
- **Prospective registries:** Establishing multicentre registries could capture long-term survival, neurodevelopmental outcomes, and quality of life, providing a more comprehensive evidence base.
- **Innovative surgical techniques:** Emerging approaches such as graduated compression dressing and minimally invasive silo placement should be evaluated in controlled studies to determine safety, efficacy, and cost-effectiveness.
- **Genomic integration:** Advances in prenatal genetic testing and whole-exome sequencing may allow earlier identification of syndromic associations, enabling tailored counselling and management.
- **Multidisciplinary antenatal counselling:** Structured pathways involving fetal medicine specialists, neonatologists, surgeons, and geneticists can optimise parental decision-making and perinatal planning.
- **Global equity:** Research should address disparities in access to neonatal surgery and intensive care, particularly in low-resource settings where conservative management may be the only feasible option.
- **Long-term follow-up:** Studies focusing on growth, abdominal wall function, and psychosocial outcomes into adolescence and adulthood are needed to guide holistic care.

Conclusion

Umbilical anomalies, particularly exomphalos, represent a spectrum of congenital abdominal wall defects with variable severity. Current practice balances surgical innovation with conservative approaches, tailored to defect size and associated anomalies. Future consensus guidelines are needed to standardise care and improve outcomes.

Funding and Ethics/Compliance Statements Block

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Competing Interests

The authors declare that they have no competing interests.

Ethics Approval

Not applicable. This study is a narrative review of published literature and did not involve human participants or animal subjects.

Patient and Public Involvement

No patients or members of the public were directly involved in the design, conduct, reporting, or dissemination of this review.

Data Availability Statement

All data relevant to the study are included in the article or uploaded as supplementary information.

Author Contributions

All authors conceived the study, conducted the literature search and drafted the manuscript. All authors contributed to revisions, approved the final version, and agree to be accountable for all aspects of the work.

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