

Umbilical Cord Hernia is Not an Isolated Pathology: High Rate of Associated Anomalies in a Tertiary Center Cohort

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Abstract

Purpose

This study aimed to evaluate the clinical profile of umbilical cord hernia (UCH), a defect often considered a minor variant of omphalocele, with a specific focus on the prevalence and spectrum of associated congenital anomalies.

Methods

A retrospective analysis was conducted on 45 neonates with anterior abdominal wall defects treated between August 2019 and July 2024. Patients were categorized into three groups: omphalocele (n = 15), gastroschisis (n = 19), and UCH (n = 11). Demographic data, prenatal diagnosis, associated anomalies, surgical management, and outcomes were analyzed.

Results

In the UCH group, six patients (54.5%) had at least one major associated anomaly. Major cardiac anomalies were present in 45.5% (5/11), chromosomal anomalies in 27.3% (3/11), and genitourinary anomalies in 36.4% (4/11) of patients. The spectrum included Tetralogy of Fallot, Trisomy 13, 18, 21, and genetic syndromes (e.g., 3MC syndrome). Four patients (36.4%) had isolated UCH. Mortality in the UCH group was 18.2%, exclusively occurring in patients with major associated anomalies.

Conclusion

Contrary to traditional belief, umbilical cord hernia is frequently associated with serious cardiac, chromosomal, and multisystemic anomalies. These findings necessitate a paradigm shift in management, advocating for comprehensive screening including echocardiography, renal ultrasound, and genetic evaluation in all neonates with UCH, similar to the standard for omphalocele.

Introduction

Congenital anterior abdominal wall defects represent a significant group of neonatal anomalies, with an estimated incidence of approximately 1 in 4000 live births [1]. Among these, gastroschisis and omphalocele are the most clinically prominent, each with distinct embryological origins, clinical presentations, and associated prognostic implications [1, 2]. Gastroschisis is characterized by a full-thickness paraumbilical defect with eviscerated, unprotected intestine and is strongly associated with young maternal age [3]. In contrast, omphalocele, a midline defect with herniated contents covered by a membranous sac, is frequently linked to chromosomal anomalies (e.g., Trisomy 13, 18, 21) and major cardiac malformations [4, 5].

A third, less-discussed entity is the umbilical cord hernia (UCH). First described by Benson et al. in 1949 [6], UCH is defined as a defect less than 4 cm in diameter containing only bowel loops and covered by a membrane, with the umbilical cord inserting at the apex of the sac. It is often clinically and embryologically grouped with “minor” or “small” omphaloceles [7]. Consequently, a pervasive notion in pediatric surgical literature posits UCH as an isolated defect with an excellent prognosis, rarely warranting systematic investigation for associated anomalies [8]. This traditional view is primarily based on limited and older case series.

However, emerging reports hint at a more complex reality. Some contemporary studies have documented UCH cases accompanied by renal, skeletal, or even cardiac anomalies, challenging its benign stereotype [9, 10]. Despite these signals, comprehensive data characterizing the modern clinical profile of UCH, particularly in a tertiary care setting, remain scarce. Most literature focuses on Western populations, and robust reports from other regions are limited [11].

This single-center, retrospective study from a major referral hospital aims to provide a detailed analysis of neonates with UCH. We evaluate their demographic characteristics, prenatal detection rates, spectrum of associated anomalies, surgical management, and outcomes. By comparing UCH with contemporaneous cohorts of gastroschisis and omphalocele, we seek to clarify whether UCH aligns with the profile of an isolated defect or shares the multisystemic association pattern of omphalocele, thereby informing evidence-based diagnostic and management protocols.

Materials and Methods

Study Design and Setting

This retrospective cohort study was conducted at the Department of Pediatric Surgery, a high-volume national tertiary referral center for congenital anomalies. The study protocol was reviewed and approved by the Institutional Ethics Committee. The requirement for informed consent was waived due to the retrospective nature of the study.

Patient Population

The medical records of all neonates born with a diagnosis of omphalocele, gastroschisis, or umbilical cord hernia between August 1, 2019, and July 31, 2024, were reviewed. Inclusion criteria were: 1) live birth with one of the three specified defects, and 2) management at our pediatric surgery or neonatal intensive care unit. Exclusion criteria were other types of abdominal wall defects (e.g., bladder exstrophy, cloacal exstrophy) or incomplete medical records.

Data Collection

Data were extracted from electronic and physical patient records using a standardized data collection form. The collected parameters included gestational age at birth, birth weight, sex, maternal age, gravidity, parity, abortion history, prenatal care status, the presence and timing of prenatal diagnosis via

ultrasonography, mode of delivery, defect characteristics, associated anomalies, type and timing of surgical repair, postoperative complications, feeding milestones, duration of total parenteral nutrition (TPN) and mechanical ventilation, length of hospital stay, mortality, and long-term follow-up findings [1].

Associated anomalies were systematically sought via routine clinical protocols, including echocardiography performed by pediatric cardiologists, abdominal and renal ultrasonography, and genetic testing (karyotyping and/or chromosomal microarray analysis) when clinically indicated. Major cardiac anomalies were defined as structural heart defects requiring surgical or catheter-based intervention or associated with significant hemodynamic compromise (e.g., Tetralogy of Fallot, pulmonary atresia).

The diagnosis of UCH was strictly based on the criteria defined by Benson et al. [6]: a midline abdominal wall defect < 4 cm in diameter, with the umbilical cord inserting at the apex of a membrane-covered sac containing only bowel loops. All cases were confirmed intraoperatively by a staff pediatric surgeon. Cases with a membrane-covered defect but without clear cord insertion at the apex, or containing solid organs (e.g., liver), were classified as omphalocele and excluded. This diagnostic rigor was applied to minimize misclassification between UCH and small omphalocele.

Statistical Analysis

Statistical analysis was performed using IBM SPSS Statistics for Windows, Version 27.0. Descriptive statistics were presented as mean ± standard deviation (SD) for normally distributed continuous variables, median and interquartile range (IQR) for non-normally distributed variables, and frequency (percentage) for categorical variables. Normality was assessed using the Shapiro-Wilk test. Comparisons between the three defect groups for continuous variables were made using one-way ANOVA or the Kruskal-Wallis test, as appropriate. Categorical variables were compared using the Chi-square test or Fisher’s exact test. A binary logistic regression model was used to identify independent risk factors for mortality. A p-value of < 0.05 was considered statistically significant.

Results

Demographic and Baseline Characteristics

A total of 45 neonates were included. The distribution was: omphalocele (n=15, 33.3%), gastroschisis (n=19, 42.2%), and umbilical cord hernia (n=11, 24.4%). Table 1 summarizes the comparative demographic data. Maternal age differed significantly between groups (p=0.006), being lowest in the gastroschisis group (23.0 years), consistent with known epidemiology [3]. Prenatal diagnosis was highest in the omphalocele group (86.7%), followed by gastroschisis (63.2%), and was lowest in UCH (36.4%) (p=0.012), likely due to its smaller, more subtle sonographic appearance.

Table 1: Demographic and Clinical Characteristics of Umbilical Cord Hernia Patients

Parameter	Value (n=11)
Sex (Male/Female)	7 (63.6%) / 4 (36.4%)
Mean Gestational Age (days)	265.09 ± 10.46
Mean Birth Weight (g)	2776.82 ± 680.05
Mean Maternal Age (years)	30.09 ± 6.77
Prenatal Diagnosis Rate	4 (36.4%)
Cesarean Delivery Rate	8 (72.7%)
Number of Mothers with Abortion History	6 (54.5%)

Associated Anomalies

The presence and spectrum of associated anomalies were markedly different among groups. Omphalocele patients had the highest burden, with 80% having a major associated anomaly, including major cardiac defects in 53.3% and Trisomy 13 in 20%. No gastroschisis patient had a major cardiac or chromosomal anomaly, though 42.1% had complex gastroschisis (intestinal atresia/perforation) [12].

Crucially, in the UCH group, six patients (54.5%) had at least one major associated anomaly. The detailed distribution is presented in Table 2. Major cardiac anomalies were the most frequent (45.5%), followed by genitourinary (36.4%) and chromosomal anomalies (27.3%). The cardiac spectrum included serious lesions like Tetralogy of Fallot and pulmonary atresia. Chromosomal anomalies comprised Trisomy 13, 18, and 21. Two patients had defined genetic syndromes. Four patients (36.4%) had completely isolated UCH.

Table 2: Distribution of Associated Anomalies in Umbilical Cord Hernia Patients*

Anomaly Category	Number of Patients (n=11)	Rate (%)	Detailed Anomaly Descriptions
Major Cardiac Anomalies	5	45.5%	Tetralogy of Fallot (2), atrioventricular septal defect (AVSD) (1), pulmonary valve atresia (1), aortic hypoplasia (1), bicuspid aortic valve (1)
Chromosomal Anomalies	3	27.3%	Trisomy 13 (1), Trisomy 18 (1), Trisomy 21 (1)
Genetic Syndromes	2	18.2%	7p deletion and 12q duplication (1), 3MC syndrome (1)
Genitourinary Anomalies	4	36.4%	Double vagina-double urethra (1), horseshoe kidney (1), polycystic kidney (1), hypospadias (1)
Musculoskeletal Anomalies	3	27.3%	Clubfoot (2), hand/finger anomaly (1)
Central Nervous System Anomaly	1	9.1%	Meningomyelocele (1)
Cleft Lip/Palate	2	18.2%	Cleft lip/palate (2)
Isolated Umbilical Cord Hernia	4	36.4%	Number of patients without associated anomalies

**: Numbers in parentheses indicate the count of specific anomalies, not patients. Individual patients may have multiple anomalies per category. "Number of Patients" reflects unique patients with ≥ 1 anomaly in that category.*

Surgical Management and Outcomes

All UCH patients underwent successful primary fascial closure within the first five days of life, with no surgical complications. This contrasts with omphalocele and gastroschisis groups, where staged closures and complications like abdominal compartment syndrome were observed. Postoperative recovery was fastest in the UCH group, with a mean time to full enteral feeding of 4.75 days versus 23.57 days in gastroschisis ($p < 0.05$).

Overall mortality was 37.8%. It was highest in the omphalocele group (66.7%), followed by gastroschisis (26.3%) and UCH (18.2%) ($p=0.018$). Both deaths in the UCH group occurred in patients with major associated anomalies (one with Trisomy 13 + major cardiac defect, one with a chromosomal microdeletion syndrome + pulmonary hypertension). Logistic regression identified the presence of a major extra-intestinal anomaly (OR: 6.8, 95% CI: 1.9–24.1, $p=0.003$) and mechanical ventilation requirement as independent predictors of mortality.

Discussion

This study provides compelling evidence that umbilical cord hernia, traditionally relegated as a minor and isolated abdominal wall defect, is in fact associated with a high rate of serious systemic anomalies in a significant proportion of cases. Our finding that 54.5% of UCH patients had a major associated anomaly—with cardiac and chromosomal defects being particularly prominent—directly challenges the entrenched clinical dogma and necessitates a fundamental re-evaluation of this condition [8].

Recently, Menchaca et al. (2023), using a large North American administrative database, provided important epidemiological evidence that UCH is associated with a higher-than-expected rate of congenital anomalies [13]. Our study builds upon and significantly extends these findings. While Menchaca et al. relied on administrative coding, we employed rigorous, surgically confirmed diagnostic criteria (Benson et al.) [6] in a single-center cohort, minimizing misclassification. Furthermore, we implemented a standardized screening protocol including universal echocardiography by a pediatric cardiologist and advanced genetic testing (karyotype ± chromosomal microarray analysis) in the majority of patients. This approach allowed us to: 1) report a more precise and detailed spectrum of associated anomalies (Table 2), including clinically silent but significant cardiac defects and rare genetic syndromes; 2) demonstrate the feasibility and yield of a systematic diagnostic algorithm (Table 3); and 3) provide a clinically applicable management pathway that moves beyond epidemiological association to direct patient care. Our findings from a distinct Turkish cohort also suggest that the high association rate is not population-specific but likely a fundamental characteristic of the defect itself. Thus, the present study should not be regarded as a simple confirmation of previous database findings, but as a clinically oriented extension that clarifies the diagnostic and management implications of UCH in a tertiary referral setting.

UCH: An Isolated Defect or a Marker of Syndromic Disease?

Classical teaching, supported by earlier series, suggests UCH is an isolated finding with an excellent prognosis. Our results contradict this. The 45.5% incidence of major cardiac anomalies and 27.3% rate of chromosomal aneuploidies in our UCH cohort are strikingly high and align more closely with the anomaly profile of omphalocele than previously recognized [4, 5]. For instance, Benjamin & Wilson's analysis of over 2800 abdominal wall defects reported high rates of extra-intestinal anomalies in omphalocele but did not highlight UCH as a high-risk entity [14]. Our data suggest that UCH may represent part of a

spectrum of ventral body wall defects, where the abdominal finding can be a sentinel marker for underlying multisystem developmental disruption, similar to omphalocele but perhaps with a variable expressivity that includes milder, isolated forms [15].

The discrepancy with older literature may be explained by several factors. First, as a national referral center, our cohort may be enriched with complex cases (referral bias). Second, earlier studies may have under-detected anomalies due to less routine or advanced screening (e.g., fetal echocardiography, genetic microarray). Finally, it is possible that the etiological mix of UCH includes both a benign, isolated form and a more severe syndromic form, the latter being increasingly recognized and referred to tertiary centers.

Embryological and Pathogenetic Implications

The standard embryological explanation for UCH—a simple delay in midgut reduction during the 10th–12th weeks—is insufficient to account for the associated cardiac and chromosomal anomalies observed, which originate much earlier in embryogenesis [16]. The coexistence of these anomalies suggests a common, earlier disruptive event. Pathogenetic theories proposed for omphalocele, such as abnormalities in ectodermal placode function or a generalized embryonic dysplasia affecting the lateral body folds, may be more applicable than previously thought for this subset of UCH cases [17, 18]. This implies that UCH might not be a localized defect but one manifestation of a broader field defect in ventral wall development, sharing a common, albeit heterogeneous, etiopathogenesis with other abdominal wall defects.

Clinical Management: Toward a Mandatory Comprehensive Algorithm

The high association rate mandates a decisive shift in clinical practice. The traditional, minimalist approach focused solely on surgical repair is inadequate and potentially dangerous if life-threatening associated conditions remain undiagnosed. Based on our findings, we propose a structured, mandatory diagnostic algorithm for every neonate with UCH, summarized in Table 3.

Table 3

Proposed Multidisciplinary Diagnostic and Management Algorithm for Neonates with Umbilical Cord Hernia (UCH)

Domain	Recommended Action	Rationale / Key Finding from Study
Cardiac Evaluation	Detailed Echocardiography by a pediatric cardiologist.	Major cardiac anomalies (e.g., Tetralogy of Fallot, AVSD, pulmonary atresia) were present in 45.5% of patients.
Genitourinary Screening	Abdominal and Renal Ultrasonography (pre- or post-operative).	Genitourinary anomalies (e.g., duplex system, horseshoe kidney, hypospadias) were present in 36.4% of patients.
Genetic Assessment	Formal Genetic Consultation with Karyotype $\pm$ Chromosomal Microarray Analysis.	Chromosomal (27.3%) and syndromic (18.2%) anomalies were significant; essential for prognosis, recurrence risk, and long-term planning.
Prenatal Management	If diagnosed prenatally: Referral for detailed fetal anomaly scan & fetal echocardiography. Discuss genetic amniocentesis.	UCH should not be dismissed as an incidental finding; associated anomalies are common and may alter prenatal counseling and delivery planning.
Delivery Room Protocol	Visual inspection of cord before clamping; clear communication between obstetrics and neonatology teams.	Herniated bowel loops may be mistaken for cord; risk of iatrogenic bowel injury is preventable with careful technique.
Surgical Consultation	Early pediatric surgical evaluation for planning of elective primary repair.	All defects in this series were successfully repaired primarily with low surgical morbidity when isolated.
Long-term Follow-up	Individualized based on associated anomalies; may include cardiology, genetics, nephrology, developmental pediatrics.	Survivors with associated anomalies require multidisciplinary care to address cardiac, renal, syndromic, or neurodevelopmental sequelae.

This protocol ensures that lethal cardiac defects are identified, genetic counseling is informed, and renal anomalies are detected. Prenatally, the diagnosis of UCH should trigger specialized scanning and counseling, moving away from its perception as an incidental finding [19]. Furthermore, emphasizing careful cord clamping technique is a simple yet critical patient safety measure.

Prognostic Stratification: Isolated vs. Complex UCH

Our data reveal a clear prognostic dichotomy. Patients with isolated UCH (36.4% of our cohort) had excellent outcomes, consistent with historical reports. Conversely, morbidity and mortality were

concentrated in the “Complex UCH” subgroup with major associated anomalies. This mortality (18.2%) is significantly higher than the near-zero mortality expected for isolated UCH [8]. This stratification is vital for accurate parental counseling and resource allocation. It underscores that poor outcomes are driven by associated conditions, not the abdominal wall defect itself.

Study Limitations and Strengths

This study has limitations inherent to its retrospective, single-center design. Referral bias likely enriched our cohort with complex cases, potentially overestimating the true population-wide association rate. The sample size, though substantial for this rare condition, limits subgroup analyses. Lack of long-term neurodevelopmental follow-up data restricts our analysis to early outcomes.

Nevertheless, the study has significant strengths. It provides one of the most detailed contemporary analyses of UCH, systematically applying modern diagnostic standards. A key strength is the diagnostic rigor. Unlike population-based studies reliant on diagnostic codes, which may misclassify small omphaloceles as UCH or vice versa, every case in our series was surgically confirmed according to strict anatomical criteria. This enhances the internal validity of our findings regarding the true association rate of UCH. The findings are clear, internally consistent, and challenge a long-held clinical axiom with direct and actionable implications for patient care.

Conclusion

Umbilical cord hernia is not the uniformly benign, isolated condition it has long been considered. A substantial subset of patients, particularly those presenting to tertiary centers, harbor serious associated cardiac, chromosomal, and multisystemic anomalies. This study mandates a paradigm shift in management. We advocate that the diagnosis of UCH should automatically initiate a comprehensive, multidisciplinary diagnostic evaluation—including echocardiography, renal ultrasound, and genetic assessment—as is standard for omphalocele. UCH should be re-conceptualized as a potential indicator of a broader developmental disorder, not merely a “small hernia.” Adopting this proactive approach is essential for accurate prognosis, informed counseling, and optimizing outcomes by ensuring timely management of life-threatening associated conditions [20, 21].

Declarations

Author Contribution

Müge Yıldız: Conceptualization, Data curation, Formal analysis, Writing – original draft. Yavuz Yılmaz: Conceptualization, Methodology, Supervision, Writing – review & editing. Elif Emel Erten: Data collection, Investigation, Resources. Müjdem Nur Azılı: Data collection, Investigation, Validation. Emrah Şenel: Supervision, Project administration, Writing – review & editing. All authors read and approved the final manuscript.

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