Exploring Congenital Anomalies Associated with Omphalocele: A Comprehensive Review of Pediatric Cases at Mulago National Referral Hospital, Uganda

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ABSTRACT

Introduction

Omphalocele is a congenital midline abdominal wall defect resulting from failure of normal embryological development, characterized by herniation of abdominal viscera into a membranous sac [1,2]. The condition is frequently associated with other congenital anomalies and syndromes, which significantly influence neonatal outcomes, particularly in low- and middle-income countries (LMICs) where diagnostic and treatment resources are limited [3].

Objective

This study aimed to determine the prevalence and characterize congenital anomalies associated with omphalocele among neonates managed at Mulago National Referral Hospital (MNRH), Uganda.

Methods

A cross-sectional descriptive study was conducted over eight months among neonates aged ≤28 days diagnosed with omphalocele at MNRH. Thirty-three neonates were consecutively enrolled after obtaining parental informed consent. All participants underwent detailed clinical evaluation, abdominal ultrasonography, babygram radiography, echocardiography, and random blood sugar testing. Data were collected using a structured questionnaire and analysed using SPSS version 20.

Results

The prevalence of omphalocele among neonates at MNRH was 0.1%. The male-to-female ratio was 1.1:1, and the median age at diagnosis was 4 days. Seventy-six percent of neonates were delivered at term, and approximately two-thirds had a birth weight above 2.7 kg. Beckwith–Wiedemann syndrome was the most common associated condition (33%), followed by pentalogy of Cantrell (3%). Cardiac anomalies were identified in 9% of cases, while musculoskeletal anomalies were present in 12%.

Conclusion

Omphalocele, though rare, is commonly associated with additional congenital anomalies, with Beckwith–Wiedemann syndrome being the most frequent. Comprehensive neonatal evaluation is essential for early identification of associated defects and optimal management.

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BACKGROUND

Omphalocele is a congenital defect of the anterior abdominal wall occurring at the umbilical ring, resulting in herniation of abdominal organs into a peritoneal sac. This anomaly arises from failure of the midgut to return to the abdominal cavity during embryogenesis [1,2]. The condition has a well-documented association with chromosomal abnormalities, syndromic conditions, and other structural congenital anomalies, which significantly affect prognosis and survival [3].

Management of omphalocele presents substantial challenges in LMICs due to limited access to prenatal screening, specialized neonatal care, and advanced diagnostic modalities [4]. Delayed diagnosis and incomplete evaluation often result in missed associated anomalies, contributing to increased morbidity and mortality [5]. Early identification and comprehensive assessment of neonates with omphalocele are therefore critical to improving outcomes [6].

MATERIALS AND METHODS

Study Design

This cross-sectional descriptive study was conducted at the Pediatric Surgery Unit of Mulago National Referral Hospital and focused on neonates aged 28 days or younger with a confirmed diagnosis of omphalocele.

Study Setting

Mulago National Referral Hospital (MNRH) is Uganda's largest tertiary healthcare facility and the primary teaching hospital for Makerere University College of Health Sciences. The hospital has a bed capacity of approximately 1,500 and serves as the only center in Uganda where neonatal surgery is routinely performed.

Study Participants

Neonates admitted through the Emergency Department, Newborn Special Care Unit (SCU), or Pediatric Surgical Outpatient Clinic (PSOP) with a confirmed diagnosis of omphalocele were eligible for inclusion. Diagnosis was confirmed by the Principal Investigator through physical examination. Neonates who died before completion of investigations or before informed consent could be obtained were excluded.

Sample Selection and Size

All neonates with omphalocele admitted between a period of 1 year who met the eligibility criteria were recruited using consecutive non-probability convenience sampling. Although a minimum sample size of 37 was estimated using Daniel's formula for prevalence studies, the rarity of the condition necessitated inclusion of all available cases during the study period, resulting in a final sample of 33 neonates.

Data Collection

Data were collected using a structured, pre-tested questionnaire administered by the Principal Investigator and a trained Research Assistant. Collected data included demographic information, clinical and physical examination findings, sonographic results, radiological findings, echocardiographic findings, and random blood sugar measurements.

Radiological images were interpreted independently by two radiologists, ultrasonography was performed by a radiologist and senior sonographer, and echocardiography by a paediatric cardiologist.

Data Management and Analysis

Data were cross-checked, double-entered into Microsoft Excel, cleaned, and exported to SPSS version 20 for

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analysis. Continuous variables were summarized using means and standard deviations or medians, while categorical variables were presented as frequencies and percentages.

Ethical Considerations

Ethical approval was obtained from the Makerere University College of Health Sciences Research and Ethics Committee, Institutional Review Board, Uganda National Council for Science and Technology, and Mulago Hospital administration. Written informed consent was obtained from parents or guardians, and confidentiality was strictly maintained.

RESULTS

A total of 34 neonates with omphalocele were identified during the study period. One neonate died before recruitment, leaving 33 neonates for final analysis. All participants underwent complete clinical assessment and all planned investigations.

The prevalence of omphalocele at MNRH during the study period was 0.1%. The male-to-female ratio was 1.1:1, and the median age at diagnosis was 4 days. Most neonates (76%) were delivered at term, and approximately two-thirds had a birth weight above 2.7 kg.

Beckwith–Wiedemann syndrome was identified in 33% of cases, making it the most common associated condition. Pentalogy of Cantrell was present in 3% of neonates. Cardiac anomalies were detected in 9% of cases, while musculoskeletal anomalies were observed in 12%.

DISCUSSION

The findings of this study demonstrate that omphalocele at MNRH is frequently associated with additional congenital anomalies, particularly Beckwith–Wiedemann syndrome. This observation is consistent with published literature indicating a strong association between omphalocele and syndromic conditions [3]. The identification of cardiac and musculoskeletal anomalies further emphasizes the need for routine comprehensive evaluation of affected neonates [6].

The observed prevalence aligns with reports from other Sub-Saharan African settings, though underreporting is likely due to missed cases from home deliveries and limited prenatal detection [5]. Prenatal ultrasound remains the primary screening tool for detecting omphalocele; however, its sensitivity in low-resource settings is variable due to equipment limitations and inadequate training [4].

The absence of genetic and chromosomal testing in this study highlights an important gap in neonatal care within LMICs, potentially leading to underdiagnosis of associated anomalies that influence long-term outcomes [5].

Limitations

The study was limited by its small sample size due to the rarity of omphalocele. Stillbirths, abortions, and home deliveries were not captured. Genetic and chromosomal analyses were not performed, and cranial ultrasonography was excluded due to financial constraints.

CONCLUSION

Omphalocele, though uncommon, is frequently associated with other congenital anomalies at Mulago National Referral Hospital, particularly Beckwith–Wiedemann syndrome. Early diagnosis and comprehensive neonatal evaluation are essential for optimal management and improved outcomes in resource-limited settings.

RECOMMENDATIONS

Strengthening prenatal ultrasound screening, establishing multidisciplinary neonatal care teams, providing

structured parental counseling, and promoting further research—including genetic studies—are essential steps toward improving the care and outcomes of neonates with omphalocele.

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