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OPIS PRZYPADKU
CASE REPORT

Hidden umbilical anomalies: case reports of newborns with a persistent urachus and a persistent vitellointestinal duct

Ukryte anomalie pępkowe: opis przypadków noworodków z przetrwałym moczownikiem i przetrwałym przewodem żółtkowo-jelitowym

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ABSTRACT

INTRODUCTION: Anomalies in the umbilical region of newborns encompass a range of congenital malformations that require careful diagnosis. These conditions may lead to delayed healing of the umbilical stump, discharge from the navel, and pose challenges for pediatricians. Among these, vitellointestinal duct (VID) anomalies are the most common, occurring in approximately 2–3% of the population. The VID, an embryonic structure connecting the yolk sac to the primitive midgut, normally regresses during development. Failure of this process can result in various anomalies, the most common being Meckel's diverticulum. Similarly, urachal anomalies arise when embryonic urachus persists, potentially presenting as abnormalities at the umbilicus.

CASE REPORTS: A 6-day-old male newborn with a draining fistula at the umbilicus was transferred to our institution with a suspected diagnosis of persistent urachus. After admission, the visible fistula was catheterized under ultrasound guidance, revealing a connection to the intestinal loops. This finding indicated a diagnosis of a persistent VID, which was subsequently excised. Another patient was a 15-day-old female newborn with a bladder defect detected during a prenatal ultrasound examination. An everted patent urachus was diagnosed after birth, which was subsequently treated by means of surgical excision.

CONCLUSIONS: Abnormalities in the umbilicus may raise the suspicion of persistent fetal structures. A thorough physical examination, supplemented by ultrasound and catheterization, can effectively make the correct diagnosis. Surgical treatment is recommended, involving resection of the persistent urachus or resection of the persistent VID with the bowel fragment.

KEYWORDS

umbilical fistula, omphalomesenteric duct remnant, patent vitellointestinal duct, congenital malformations, patent urachus

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STRESZCZENIE

WPROWADZENIE: Nieprawidłowości w okolicy pępka u noworodków obejmują szereg wrodzonych wad rozwojowych, które wymagają dokładnej diagnostyki. Stany te mogą prowadzić do opóźnionego oddzielenia kikuta pępowiny, wycieku z pępka, a także stanowić wyzwanie diagnostyczne dla pediatrów. Spośród nich anomalie przewodu żółtkowo-jelitowego (*vitellointestinal duct* – VID) należą do najczęstszych – występują u około 2–3% populacji. Przewód żółtkowo-jelitowy, struktura embrionalna łącząca pęcherzyk żółtkowy z pierwotnym jelitem środkowym, zwykle ulega regresji w trakcie rozwoju. Brak tego procesu może prowadzić do różnych nieprawidłowości, z których najczęstszą jest uchyłek Meckela. Podobnie anomalie moczownika powstają w wyniku przetrwania tej struktury zarodkowej, co może skutkować zmianami w obrębie pępka.

OPIS PRZYPADKÓW: 6-dniowy noworodek płci męskiej z przetoką drenującą w pępku został przetransportowany do naszego ośrodka z podejrzeniem przetrwałego moczownika. Przy przyjęciu widoczną przetokę cewnikowano pod kontrolą ultrasonografii, ujawniając połączenie z pętlami jelitowymi, co pozwoliło na rozpoznanie przetrwałego VID, który został chirurgicznie usunięty. U drugiego pacjenta, 15-dniowego noworodka płci żeńskiej z podejrzeniem wady pęcherza moczowego w badaniach prenatalnych, zdiagnozowano przetrwały moczownik, który poddano resekcji.

WNIOSKI: Nieprawidłowości pępkowe mogą budzić podejrzenie przetrwałych struktur płodowych. Dokładne badanie fizykalne, wspomagane ultrasonografią i cewnikowaniem, umożliwia przeprowadzenie precyzyjnej diagnostyki. Zalecane jest leczenie chirurgiczne, obejmujące resekcję przetrwałego moczownika lub resekcję przetrwałego VID z fragmentem jelita.

SŁOWA KLUCZOWE

przetoka pępkowa, pozostałość przewodu żółtkowo-jelitowego, drożny przewód żółtkowo-jelitowy, wady wrodzone, przetrwały moczownik

INTRODUCTION

The umbilicus, though a small structure, holds significant medical importance not only as a symbolic marker of birth, but also as a site where several fetal structures are present during development. These structures typically tend to involute, however, they may persist in some cases. The navel forms during the fourth week of fetal life with the occurrence of the embryonic plate. Initially, it appears as a primitive umbilical ring that consists of a connecting stalk, umbilical vessels, the vitelline duct, allantois, and loops of the intestine. Subsequently, it develops into the definitive cord, including one vein and two arteries suspended in Wharton's jelly. After birth, these structures obliterate and develop into ligaments [1]. Congenital malformations in the umbilical region are most commonly vitellointestinal duct (VID) anomalies affecting ca. 2–3% of the population, followed by urachal remnants (ca. 1%) [2,3]. While the prevalence of VID varies between 1 in 5000–8000, the prevalence of patent urachus is extremely rare, occurring in only 1–2 cases per 100 000 [4].

VID is defined as the embryonic connection between the yolk sac and the primitive midgut of the developing fetus. The anomaly occurs when these structures fail to resorb, the most frequent one being Meckel's diverticulum [5]. VID or its adjacent tissue can also contain ectopic mucosa, which may be found in pancreatic, gastric, duodenal or colonic tissue. Therefore, a thorough excision of the surrounding heterotopic tissue should be performed, along with histopathology examination [6]. The prolapse of the small bowel through the umbilical opening is considered to be a rare presentation of persistent VID [7].

Urachal anomalies occur when there is a persistence of embryonic urachal remnant without involution into the median umbilical ligament [2]. A study conducted in Canada reports a prevalence rate of approximately 1% in the general population. The preferred approach for managing patent urachus is surgical removal, even for asymptomatic remnants, due to the risk of future infection and potential malignancy [2,3].

Both VID and urachal anomalies should be considered when diagnosing congenital lesions in the umbilical region. Our report presents two neonatal cases that illustrate these malformations and emphasizes the crucial role of thorough physical examination, together with ultrasound imaging in distinguishing between them. Considering their distinct embryological origins and anatomical features, accurate diagnosis is essential.

CASE REPORTS

Case 1

A 6-day-old male neonate born by caesarean section at 38 weeks of an uncomplicated pregnancy was referred to our institution due to suspected persistent urachus. His general condition upon admission was good. Apart from secreting fistula at the umbilical cord stump (Figure 1), the newborn exhibited no significant abnormalities on physical examination and laboratory testing. Abdominal ultrasound revealed a urine-filled bladder that did not empty following catheter insertion into the fistula. Furthermore, the intestinal contents were visualized in the catheter. These findings suggested a diagnosis of VID. The child underwent

VID excision with wedge resection of the small bowel via an open laparotomy (Figure 2). The patient was discharged home on postoperative day 8 following an uncomplicated recovery. The histology examination of the resected VID confirmed the presence of small bowel mucosa with signs of inflammation. No complications were observed during the 6-month follow-up.

Fig.1. Patent vitellointestinal duct.

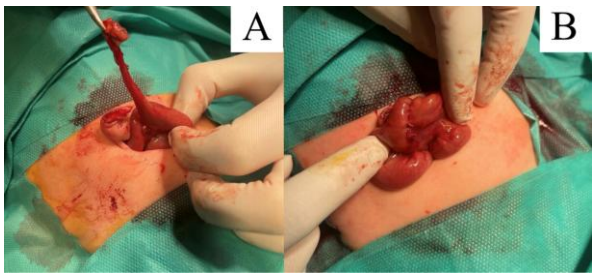

Fig. 2. Vitellointestinal duct before excision (A). Small bowel after wedge resection (B).

Case 2

A 15-day-old female neonate was referred to our institution with a prolapsed bladder through a patent urachus. The diagnosis was initially suggested on prenatal ultrasound and later confirmed during physical examination by inserting a catheter through the urethra, which was subsequently visualized within the fistula (Figure 3). The child underwent excision of the patent urachus with wedge resection of the adjacent bladder fragment (Figure 4). A catheter was kept in the bladder for 6 days. The child was discharged home on postoperative day 14 following an uncomplicated recovery. Histological examination of the excised specimen revealed urachal epithelium, which confirmed the initial diagnosis. No further complications were observed during the 7-month follow-up.

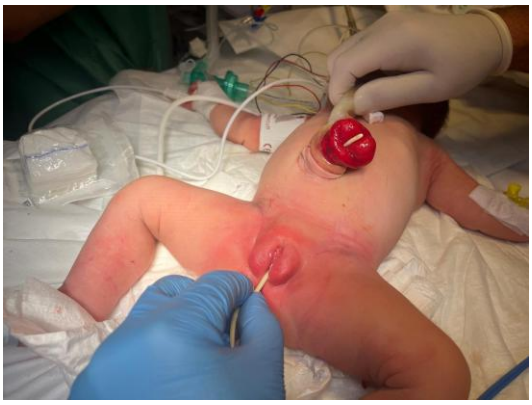

Fig. 3. Catheter inserted through urethra and visualised in patent urachus.

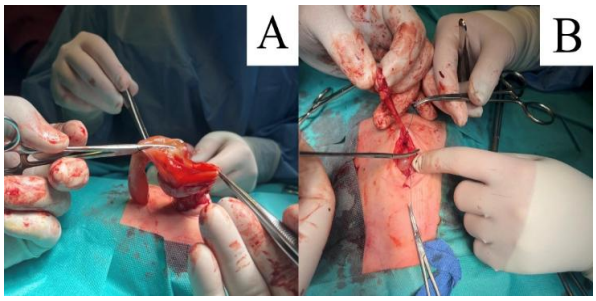

Fig. 4. Patent urachus before excision (A). Excision of patent urachus with wedge resection of adjacent bladder fragment (B).

DISCUSSION

Persistent embryonic structures in newborns may initially present with similar clinical manifestations, but the associated complications can vary significantly. Approximately 15% of VID anomalies are related to a patent VID, while other manifestations include Meckel's diverticulum, cysts, sinuses, and fibrous cords [7]. VID remnants can lead to complications such as an acute abdomen, painless rectal bleeding, intestinal obstruction, or umbilical anomalies, with these issues typically occurring in infancy [8]. Similarly, a persistent urachus may result in complications, including abscess formation in the surrounding area, further underscoring the need for timely surgical intervention [9,10]. Although rare, malignant transformation of urachal mucosa has been reported. Urachal carcinoma is a rare and aggressive tumor, with a poor prognosis once diagnosed [11].

Rare persistent embryonic structures in the umbilical region often pose a diagnostic challenge due to their overlapping clinical presentations. Without clear signs such as umbilical discharge or a fistula, these anomalies can be easily misdiagnosed or overlooked. For instance, cases have been reported where a suspected persistent VID was later diagnosed as an inflamed

appendix adjacent to the abdominal wall [12], or an umbilical cyst observed on prenatal ultrasound was ultimately identified as a persistent urachus postnatally [13]. Similarly, in our case, what initially resembled an urachal remnant was later confirmed as a VID anomaly following postnatal evaluation with physical examination and ultrasound.

Prenatal diagnosis of urachal anomalies remains uncommon as these conditions are primarily considered when umbilical drainage occurs during the neonatal period [14]. However, advancements in ultrasound technology and increasing expertise among ultrasonographers have improved the potential for prenatal detection. For example, a case report describing two cases of bladder prolapse through a patent urachus included one case diagnosed prenatally, enabling surgical intervention at just 9 hours of life [15]. In another report, Japanese authors presented a situation where the symptoms initially suggested a VID anomaly, but abdominal CT identified an anomalous congenital band instead [16]. This band, connecting the mesentery to the umbilicus, lacked the mucosal layer characteristic of VID histology.

Accurate diagnosis of congenital malformations relies heavily on imaging techniques. Our case highlights the utility of ultrasound in evaluating the umbilical region for suspected VID or urachal anomalies. Ultrasound is particularly beneficial for ruling out other conditions, such as polyps, umbilical remnants, or granulomas, and for preoperative planning or assessing connections to the bowel or urinary bladder [17]. While VID and urachal remnants are more likely in neonates and infants, conditions like umbilical granuloma, umbilical polyp, omphalolith, neoplasms, or benign soft tissue masses should be considered in older children [7,17,18].

When planning surgical intervention, the choice of operative approach depends on the type of anomaly. For VID or Meckel's diverticulum, wedge resection or segmental resection is preferred. If the base of the diverticulum is narrow, a wedge resection is typically performed; if the base is wide, segmental resection with anastomosis is required [19]. Histological findings often reveal ectopic gastric, duodenal, colonic, or pancreatic tissue within the VID or adjacent intestinal tissue, which may cause bleeding or ulceration. Consequently, excising a small portion of the intestinal wall near the duct opening is recommended to prevent complications. In a study analyzing symptomatic VID remnants in children, wedge resection was the predominant surgical approach, particularly in cases presenting with abdominal pain (63%) [20].

For persistent urachus, surgical excision involves removing the structure along with a section of the adjacent bladder tissue to mitigate the risk of malignancy. Given the potential for malignant transformation, excision of the surrounding tissue is considered to be a preventive measure [20].

This discussion underscores the importance of accurate prenatal and postnatal diagnosis, appropriate imaging modalities, and timely surgical intervention to address persistent embryonic structures in the umbilical region effectively.

CONCLUSIONS

Abnormalities in the umbilicus: a fistula, signs of inflammation, or fluid leakage may raise the suspicion of persistent fetal structures. A thorough physical examination, supplemented by ultrasound and catheterization, can effectively make the correct diagnosis. Surgical treatment is recommended, involving resection of the persistent urachus with the adjacent bladder fragment to prevent potential malignant transformation of the bladder or resection of the persistent VID with the bowel fragment to prevent bowel pathology associated with the presence of ectopic tissue.

Authors' contribution

Study design – G. Kudela, A. Wiernik
Manuscript preparation – A. Kątnik, K. Szala, G. Kudela, A. Wiernik
Literature research – A. Kątnik, K. Szala, G. Kudela, A. Wiernik
Final approval of the version to be published – G. Kudela, A. Wiernik

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