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Case Report

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Rare Causes of Abdominal Mass in Infants: Hydrocolposis and Hydronephrosis Due to Imperforate Hymen

Bebeklerde Karın Kitlelerinin Nadir Nedeni: İmperfore Hymen'e Bağlı Hidrokolpos ve Hidronefroz

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ABSTRACT

The epidemiology of pediatric abdominal masses varies according to age. Although malignant masses are more common in older children, benign masses are more common during the neonatal period and infancy. Here, we present a case of a baby with hidrocolpos and hydronephrosis due to imperforated hymen (IH). A 6-week-old female infant presented with abdominal distension after 2 weeks. The patient was admitted to our clinic after an abdominal mass was detected on ultrasonography (USG) performed at another center. Physical examination revealed abdominal distention and dullness on percussion, suggesting the presence of fluid or a mass, and a firm, non-palpable abdomen on palpation. A mass was visible on the direct abdominal X-ray. Subsequent USG performed at our hospital revealed a 10-cm cystic mass extending to the right upper quadrant of the abdomen, accompanied by hydronephrosis. Pediatric surgery was requested from the patient. IH was detected in the infant. Abdominal computed tomography revealed vaginal atresia. The patient was surgically treated with hymenotomy and pus drainage. The appearance of the mass improved on direct abdominal X-ray taken after the surgery. In conclusion, IH is a rare disease that is often missed in diagnosis. Therefore, clinicians should adopt a multisystemic approach that emphasizes the importance of thorough physical examination, including a complete genital examination.

Keywords: Hydrocolpos, hydronephrosis, imperforate hymen

ÖZ

Pediatrik karın kitlelerinin epidemiyolojisi yaş gruplarına göre değişiklik gösterir. Malign kitleler yaşlı çocuklarda daha yaygınken, benign kitleler neonatal dönem ve bebeklik döneminde daha yaygındır. Burada, imperforate hymen (IH) nedeniyle hidrokolpos ve hidronefroz olan bir bebek vakasını sunuyoruz. 6 haftalık bir kız bebek, 2 haftadır karın şişliği şikayetiyle başvurdu. Hasta, başka bir merkezde yapılan ultrasonografi (USG) sonucunda karın kitlesi tespit edilmesinin ardından kliniğimize kabul edildi. Fizik muayenede karın distansiyonu, sıvı veya kitle varlığını düşündüren perküsyonla matlık ve palpasyonda sert, non-palpable bir karın gözlemlendi. Direkt karın röntgeninde bir kitle görüldü. Hastanemizde yapılan USG'de, karın sağ üst kadranına uzanan 10 cm'lik kistik bir kitle ve hidronefroz görüldü. Pediatrik cerrahi konsültasyonu istendi. Bebekte IH tespit edildi. Hastanın abdominal bilgisayarlı tomografisi vajinal atrezisi ile uyumluydu. Hasta, hymenotomi ve apsenin drenajı ile cerrahi olarak tedavi edildi. Cerrahiden sonra çekilen direkt karın röntgeninde kitlenin görünümü iyileşti. Sonuç olarak, IH nadir bir hastalıktır ve teşhis sıklıkla gözden kaçabilir. Bu nedenle, klinik olarak çok sistemli bir yaklaşım benimsenmeli ve kapsamlı fizik muayenelerin, özellikle tam bir genital muayenenin önemi vurgulanmalıdır.

Anahtar Kelimeler: Hidrokolpos, hidronefroz, imperfore hymen

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INTRODUCTION

Abdominal masses are common in pediatric patients. Abdominal masses have a wide range of differential diagnoses, including benign and malignant tumors (1). When examining pediatric "abdominal masses", malignancies are a possibility to be considered (2). The epidemiology of pediatric abdominal masses varies according to age. Although malignant masses are more common in older children, benign masses are more common during the neonatal period and infancy (3-8). Here, we present an infant with hydrocolpos and hydronephrosis due to imperforate hymen (IH). Consent was obtained from the patient and her family.

CASE REPORT

Anamnesis and Physical Examination

A 6-week-old female infant was born at 38 weeks of gestation via normal vaginal delivery, weighing 4.030 g. The patient had a 2-week history of abdominal swelling. On physical examination, abdominal distension was noted, and a mass was observed on the direct abdominal X-ray. However, inadequate evaluation during primary care led to delayed diagnosis and unnecessary referral to a higher-level specialist. The increasing patient load and limited time available for thorough assessments in primary care often result in early referrals, which can cause unnecessary workload in the health system and undue anxiety for parents.

Diagnostic Evaluation

a. Laboratory Tests

• White blood cell (WBC) count, hemoglobin (Hb), mean corpuscular volume (MCV), and platelet count: these tests were requested to assess the presence of an underlying inflammatory process or hematological disorder that might be contributing to abdominal distension.

• Tumor markers (CA-15-3, AFP, B-HCG): these markers were measured to rule out malignancy and to determine whether the abdominal mass was benign or malignant.

b. Imaging Methods

• Direct abdominal X-ray: this was performed to identify the cause of the abdominal distension and confirm the presence of an abdominal mass (Figure 1).

• Ultrasonography (USG): the initial USG performed at another center identified well-circumscribed bilateral masses. Follow-up USG at our hospital was conducted to further evaluate the size, location, and associated conditions of hydronephrosis. Direct abdominal X-ray findings were also considered.

• Abdominal computed tomography (CT): this technique was requested to provide detailed imaging of the mass (Figure 2), assess anatomical relationships, and identify other structural anomalies, such as ureteral and vaginal atresia.

Laboratory results showed a WBC count of 13.1 x 10^3/µL, Hb of 11 g/dL, and MCV of 91 fL. The tumor markers were CA-15-3 at 13.3 U/mL, AFP at 82.1 µg/L, and B-HCG <0.5. USG performed at another center revealed bilateral mass lesions measuring 12x8x11 cm, which were presumed to originate from the kidneys. Direct abdominal X-ray identified a mass. USG at our hospital revealed a 10-cm cystic mass extending to the right upper quadrant, accompanied by hydronephrosis, dilated pelvicalyceal systems, and a tortuous left ureter.

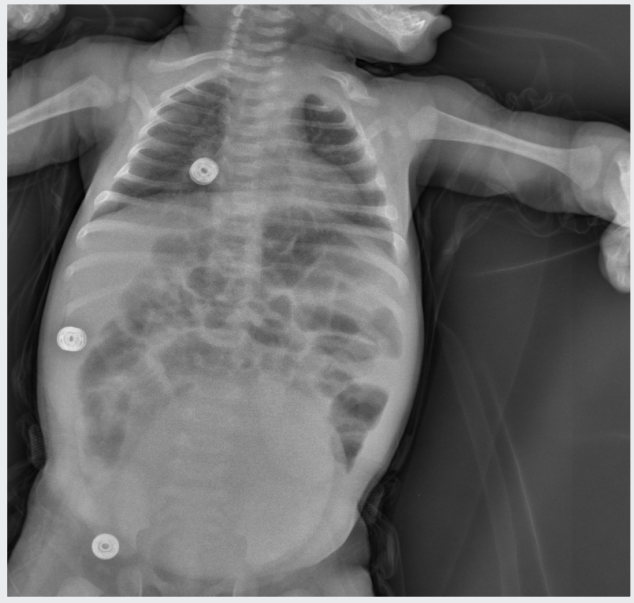

Figure 1. Patient's direct abdominal X-ray, demonstrating abdominal mass

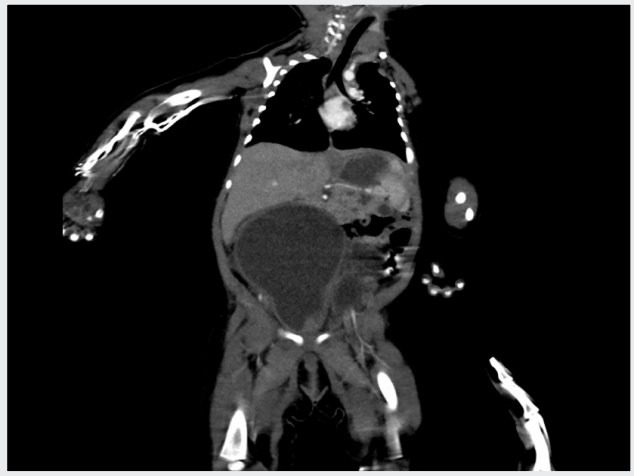

Figure 2. Abdominal CT showing cystic structure with fluid in the abdomen

CT: Computed tomography

Abdominal CT imaging revealed two distended cystic structures thought to be the uterus, cervix, and bladder, and a fluid-filled cystic structure in the abdomen, suggestive of ureteral and vaginal atresia. Grade 2 ectasia was detected in the pelvicalyceal systems of both kidneys.

Diagnosis and Treatment

A pediatric surgery consultation was performed to further evaluate the findings. The diagnosis of IH with hydrocolpos was based on imaging and clinical presentation. The treatment plan included surgical intervention, which involved incision of the imperforate hymen and drainage of accumulated pus. Postoperative evaluation revealed improvement in the appearance of the abdominal mass, as seen on direct abdominal X-ray taken after the operation (Figure 3).

DISCUSSION

Neonatal hydrocolpos is a rare condition (9). In infants with IH, hydrocolpos caused by maternal estrogen is often discovered incidentally (10-12). An imperforate hymen may exert pressure on the urinary system, leading to hydronephrosis (13,14). The incidence of IH is approximately 0.1% (15). Diagnosis is frequently delayed because the condition is asymptomatic and rare (16). IH is commonly diagnosed after menarche, and its symptoms include amenorrhea, abdominal pain, and urinary retention. Typically, IH patients are asymptomatic. In rare cases, hydrocolpos can cause hydrocolpos, presenting as an abdominal mass in approximately 0.006% of infants (17-19). Our patient presented with an abdominal mass Ramareddy et al. (20) reported eight consecutive cases of IH from 2010 to 2015, of which two were identified in infancy. In seven of these cases, genitourinary obstruction was noted.

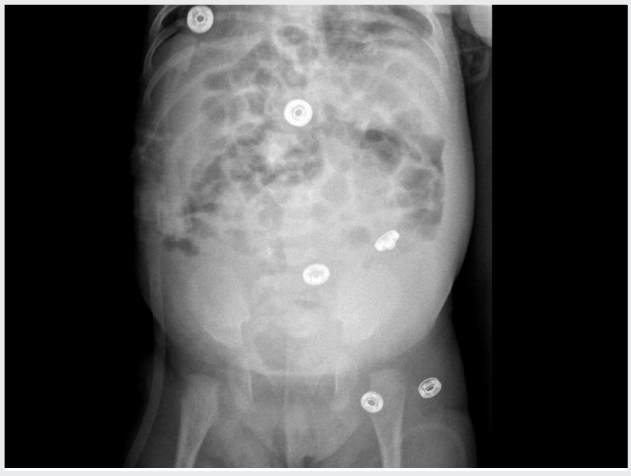

Figure 3. Patient's direct abdominal X-ray, taken after surgery demonstrating disappearance of abdominal mass

Eksioglu et al. (21) described a rare case of IH associated with a bicornuate uterus and bilateral hydroureteronephrosis while investigating a pelvic mass in an 8-month-old infant. In our case, both hydronephrosis and IH were present.

Neonatal hydrocolpos is an exceptionally rare condition that is often discovered incidentally in infants with an IH (9). This condition is typically linked to maternal estrogen exposure, which can lead to the accumulation of fluid in the vagina, resulting in hydrocolpos (10-12). The pressure exerted by hydrocolpos can affect the urinary system, potentially causing hydronephrosis. Although IH has an incidence rate of approximately 0.1%, its diagnosis is frequently delayed because of its asymptomatic nature in the neonatal period (13-15). Symptoms and diagnoses are often made later in menarche, when typical symptoms such as amenorrhea, abdominal pain, and urinary retention become apparent.

In rare cases, IH can present with hydrocolpos as an abdominal mass, with an incidence of approximately 0.006% in infants (17-19). Our case is consistent with this rare presentation. The infant presented with an abdominal mass, which is consistent with the findings in the literature. For instance, Ramareddy et al. (20) reported eight cases of IH, two of which were diagnosed in infancy. Notably, seven out of these eight cases exhibited genitourinary obstruction, highlighting the significant effect of IH on urinary tract function.

Similarly, Eksioglu et al. (21) reported an unusual case of IH associated with a bicornuate uterus and bilateral hydroureteronephrosis in an 8-month-old infant. This case further illustrates the diversity of clinical presentations of IH and the importance of thorough diagnostic evaluation when encountering atypical findings.

In our case, the presence of both hydronephrosis and IH underscored the complexity of the condition. The abdominal mass was detected via imaging studies and managed surgically by incision of the hymen and drainage of hydrocolpos. Postoperative imaging showed significant improvement, supporting the effectiveness of timely surgical intervention in managing such rare conditions.

Clinical Implications

Early Detection: Given the rarity and potential for delayed diagnosis of IH, clinicians should maintain a high index of suspicion when confronted with an abdominal mass in an infant, especially if other symptoms are present.

Imaging and Diagnosis: Comprehensive imaging, including ultrasound and computed tomography, plays a crucial role in the diagnosis and management of IH. Accurate imaging is essential for identifying associated conditions like hydronephrosis and understanding the extent of obstruction.

Management: Surgical intervention, including incision of the hymen and hydrocolpos drainage, is often necessary for symptomatic relief and to prevent further complications. Early surgical intervention can improve outcomes and prevent long-term sequelae.

Future Research: Further studies are needed to better understand the pathophysiology of IH and its associated complications. Additionally, studies on the long-term outcomes of infants with IH who are treated surgically could provide valuable insights into the management and prognosis of this rare condition.

CONCLUSION

In conclusion, IH is a rare disorder that is often missed in diagnosis. Therefore, clinicians should adopt a multisystemic approach that emphasizes the importance of thorough physical examination, including a complete genital examination.

Ethics

Informed Consent: Informed consent was obtained from all participants.

Author Contributions

Concept: M.Ç., B.Y.K., Design: Ş.Ç.K., M.Ç., B.Y.K., Data Collection or Processing: Ş.Ç.K., M.Ç., Analysis or Interpretation: B.Y.K., Literature Search: Ş.Ç.K., M.Ç., B.Y.K., Writing: Ş.Ç.K.

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