Mullerian Duct Cyst Causing Bladder Outlet Obstruction in a Patient with HNF-1 β Gene Deletion

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Key Words

Mullerian duct cyst • Urinary outlet obstruction •
Hepatocyte nuclear factor-1 β (HNF-1 β) • Prostate cyst

Abstract

A 24-year-old male was referred to a tertiary hospital for a possible prostatic abscess. The patient went into acute urinary retention. Transurethral drainage was performed. MRI pelvis three days post-operatively identified the prostatic cystic structure as a müllerian duct cyst.

Several other phenotypical features were noted on examination as well as findings on investigations. From these diagnosis of hepatocyte nuclear factor-1 β (HNF-1 β) gene deletion was made.

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Case Presentation

A 24-year-old male was referred to a tertiary hospital for a possible prostatic abscess. His initial presentation was vomiting and lethargy with a diagnosis of diabetic ketoacidosis being made. He subsequently developed fevers and acute urinary retention. Pelvic magnetic resonance imaging was performed showing a thick walled mid-line cystic structure associated with the prostate (fig. 1).

The patient was commenced on broad-spectrum antibiotics. Due to the size and location of the abscess it was deemed too difficult for percutaneous drainage. The patient underwent a transurethral drainage. Preoperative digital rectal examination demonstrated a large soft fluctuant mass. Intra-operatively there was no evident urethral connection. The prostatic urethra was incised using a Colins knife 10 mm lateral to the verumontanum and a capacious cystic structure opened. Internally there was clear fluid with the walls of the structure covered in an adherent white fibrotic sloughy tissue (fig. 2). A 26 Fr resection loop was used to bluntly enucleate the cyst contents, which included several small gelatinous prostatic stones. Histopathology showed fibrous tissue infiltrated by neutrophils and histiocytes. Nil spermatozoa were identified. Culture of the cystic fluid grew *Staphylococcus aureus*.

Further review of the medical history and examination revealed that the patient was diagnosed with insulin dependent diabetes 5 years ago. It was noted that he had bilateral cataracts from childhood. He had also experienced orgasmic anejaculation for the past 2 years. There was no significant family history with both parents alive and well.

Radiological investigation revealed multiple cortical and medullary renal cysts bilaterally up to 15 mm in diameter. Mild bilateral hydronephrosis and hydroureter was evident. Splenomegaly was also identified. Magnetic resonance cholangiopancreatography showed a normal pancreas with dilatation of the pancreatic duct. Repeat

Fig. 1. Axial and sagittal view of magnetic resonance images (T2 weighted). White arrows identifying Müllerian duct cyst.

magnetic resonance imaging pelvis three days post-operatively identified the prostatic cystic structure as a Müllerian duct cyst. Notable blood tests demonstrated normal renal function, elevated liver function tests, hypomagnesemia and hypoalbuminemia.

A diagnosis of hepatocyte nuclear factor-1 β (HNF-1 β) gene deletion was made.

Discussion

HNF-1 β Mutation

HNF-1 β is involved in developmental regulation of the kidneys, pancreas, urogenital tract and biliary system. This mutation is commonly termed renal cysts and

Fig. 2. An endoscopic photo looking into cyst. White arrows identifying slough lining cyst (left) and verumontanum (right) lateral to which small incision was made.

diabetes syndrome, as these are the predominant features [1]. As more cases are analysed it has become apparent that there are a wide range of phenotypical features. Spontaneous HNF-1 β deletions are seen to be as high as 50% [1]. This high frequency of de novo mutations explains why parents are often unaffected.

Renal Disease

The most common renal finding in HNF-1 β anomalies is multiple bilateral renal cysts, often misdiagnosed as autosomal dominant polycystic kidney disease. Other findings include unilateral renal agenesis and renal hypoplasia [1]. Renal function can be variable between patients. Some patients display normal renal function with clinically asymptomatic proteinuria while others present with rapid deteriorations in function and chronic kidney disease [1].

There has been a novel observation of a chromophobe renal cell carcinoma develop in a patient with known HNF-1 β anomaly [2].

Diabetes

Maturity onset diabetes of the young 5 is the most frequent extra-renal phenotype associated with HNF-1 β anomalies. Age of diagnosis can range from the neonatal period to late middle age with a mean age of 24. An analysis of a cohort with known HNF-1 β mutation showed that 48% had diabetes [1].

Urogenital Malformation

Most HNF-1 β associated urogenital malformations have been described in females. They are usually related to müllerian duct developmental abnormality and include bicornuate uterus, uterus didelphys and single ovaries [1]. The occurrence of male urogenital malformation is much rarer and very few cases have been reported. Cryptorchidism, hypospadias and epididymal cyst have been noted but due to normal population prevalence their presence is of uncertain significance [1].

Hematology

Elevated liver function tests are common among patients with HNF-1 β anomalies. They are usually seen as rises in alanine aminotransferase and γ -glutamyl transpeptidase. These increases are usually not associated with jaundice or liver insufficiency [1]. In contrast several studies have presented patients with severe neonatal cholestasis requiring surgical intervention as well as late onset cholestasis in middle age adults [3].

Hypomagnesaemia seen in patients with HNF-1 β gene anomalies is due to loss of the γ subunit of the Na⁺/K⁺ ATPase. Malfunction leads to decreased magnesium reabsorption in the distal convoluted tubule [1].

Other Clinical Features

A recent study has shown some previously unrecognised HNF-1 β anomaly phenotypical associations in one

family group. These are bilateral childhood cataracts and splenomegaly [1]. Pectus excavatum has also been noted to be part of the clinical spectrum [1].

Case Relevance

The identification of a müllerian duct cyst in a patient with a HNF-1 β gene anomaly appears to be a previously unreported occurrence. Other Müllerian duct abnormalities have been seen in female cases. There are several types of cystic structures associated with the lower male genital tract and consideration needs to be given to these as a differential diagnosis in this case. These can be broadly divided into intraprostatic and extraprostatic cysts. Intraprostatic cysts include prostatic utricle cysts, Müllerian duct cysts, ejaculatory duct cysts, and prostatic abscesses. Extra prostatic cysts include seminal vesicle cysts and cysts of the vas deferens [6].

In this case the patient had a midline intraprostatic cystic structure not communicating with the prostatic urethra and extending above the prostate. On opening it contained clear fluid and two gelatinous prostate stones. Nil spermatozoa were identified. All of these features are in keeping with a müllerian duct cyst. There have been case reports of Müllerian duct cysts causing ejaculatory impairment as well as becoming infected and masquerading as a prostatic abscess [1]. This however is the first documented case of a patient with HNF-1 β gene deletion presenting with an obstructing Müllerian duct cyst.

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