A case of hermaphroditism that presented as a pelvic cystic mass

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Abstract This report presents the case of a 14-year-old individual who was raised as a male child. The patient had a history of bilateral cryptorchidism combined with hypospadias, he visited our hospital with complaints of urinary retention and a cystic mass behind the bladder after the operation of urethroplasty for hypospadias. Pathological tests of the cystic mass showed that it was uterine tissue, but no ovarian tissue was found. The clinical diagnosis of mixed gonadal dysgenesis was confirmed in the case.
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Case report

The patient was a 14-year-old boy who had undergone urethroplasty and bladder fistulization for second-degree hypospadias at another hospital 2 months before being admitted to our hospital. He had a weak urine stream after the operation and experienced dysuria for 2 days before admission to our hospital. Moreover, 20 days after the surgery, leakage of urine was found near the ventral root of penis, and the urine was seen as a thin stream. No special treatment was provided; however, 7 days before he was admitted to our hospital, urethral dilatation was performed at another hospital. Two days earlier, dysuria had aggravated with dribbling and cloudy urine accompanied by frequent urination, urgent urination, odynuria, and tolerable intermittent lower abdominal pain. The results of the physical examination showed that his height was 154 cm, and he showed normal male sex characteristics such as a developed Adam's apple, normal beard, and pubic hair distribution. The lower abdominal area around the bladder was bulging. The penis and the scrotum were normally developed. The penile appearance was similar to that after urethroplasty and an urethral orifice was located at the glans. In the normal state, the length of the penis was 6 cm, and a fistula with a diameter of 1 mm was found near the ventral root of the penis. The patient had difficulty in urination; the proximal urethra at the urethral anastomotic site expanded significantly during urination, and the urine stream was thin. The size and texture of the left testis were normal. The left epididymis was about 1.5 cm × 1 cm × 1 cm, was hard on touch, and showed tenderness, while the right scrotum was empty. The testicle was not palpable in the right scrotum and inguinal region. A digital rectal examination showed the presence of an elongated, palpable cystic mass in front of the rectum, with a left-right diameter of 4.5 cm. The upper boundary could not be touched, and the surface was smooth without obvious tenderness. The patient's medical history included surgery for bilateral cryptorchidism at a local hospital when he was 3 years old. Orchidopexy of only the left testis was performed by surgery, and biopsy of the sex gland confirmed that it had testicular tissue, while the right testis was not checked. The patient had undergone urethroplasty 2 months before being admitted to our hospital for hypospadias.

Additional examinations

Retrograde urography (Figure 1): Retrograde urography with an indwelling catheter was performed through the urethral orifice. About 300 ml of slightly cloudy yellow urine was drained, and the contrast agent was injected through the catheter. The angiographs showed that the catheter was indwelled into a strip-like cyst cavity whose length was 14.5 cm and diameter was 5 cm. The cyst cavity was seen as an abnormality of the bladder. After the catheter removal and urination, the volume of the cyst was not significantly reduced.

Computed tomography (CT) (Figure 2): Although abnormalities in the bladder size and shape were not evident in the CT scans, a cystic mass with a diameter of about 5 cm was found behind the bladder. This mass had a clear border, and a smooth cystic wall and shared its boundary with the bladder and the rectum. The CT value of the cystic fluid was close to that of urine in the bladder.

Urinary system ultrasonography: The ultrasonography of the urinary system showed normal size and morphology for both the kidneys. The renal capsule was smooth, and solid echo was uniform. The upper poles of both the kidneys were normally placed, while the lower poles drew closer to the spine and assembled as low-echo regions in front of the spine. Bladder filling was good and the continuity of the bladder wall was complete. An irregular cystic echo of

115 mm × 52 mm × 31 mm with a clear boundary was found at the right rear side of the bladder. After urination, the pattern of the cystic echo did not change.

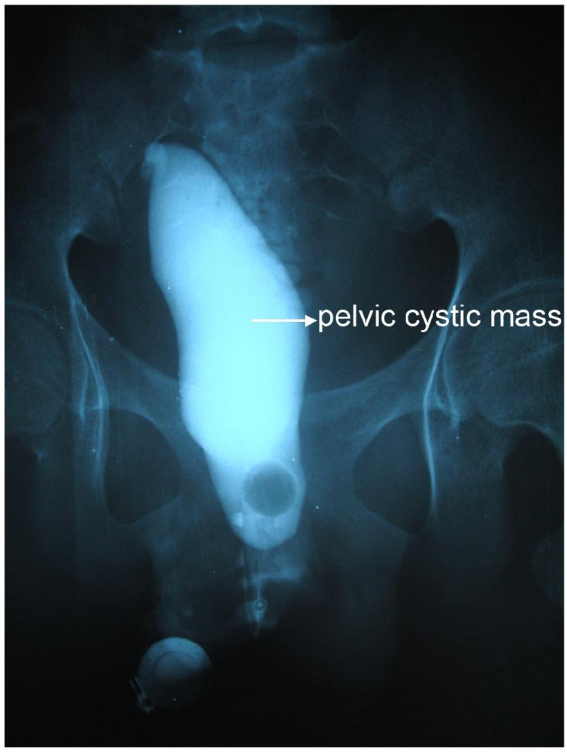

Figure 1. The angiographs showed that the catheter was indwelled into a strip-like cyst cavity but the urinary bladder did not develop.

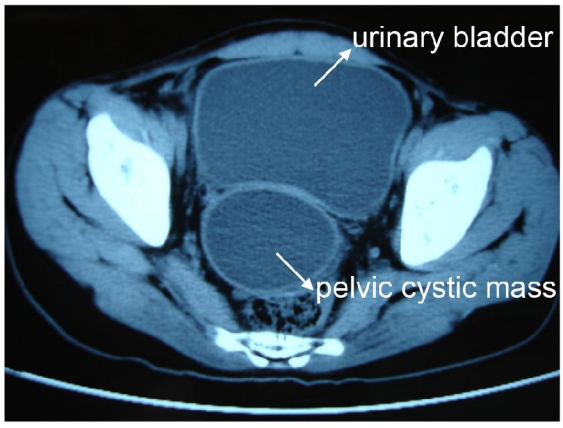

Figure 2. A cystic mass behind the urinary bladder was showed in CT.

Treatment course: After anti-infection treatment, surgical treatment was performed. A long fusiform cystic mass similar to the uterus was found behind the bladder during surgery. A clear, pale yellow liquid was extracted from the puncture. Running downward along the rectovesical pouch and ending below the neck of the bladder, the uterine broad ligament-like tissue had blood vessels running within it and was found to connect with the lateral peritoneum on the right side. A gonadal tissue (1.5 cm × 1.0 cm) similar to that of the ovary was found in the right iliac fossa. Both the ureters and the bladder were normal. A cystic mass was isolated from the place where the lower part of neck of the bladder meets the posterior urethral, and then the mass was removed. The gland similar to the ovary was removed by surgery, and the urinary fistula was repaired.

Pathological findings: The tested tissues were uterine and oviduct tissue, and no ovarian tissue was found.

Chromosomal examination: The karyotype was 45X/46XY, sex-determining region of Y chromosome (SRY) (+).

Reviewed CT scans: No obvious abnormalities in the bladder size and shape were seen. The cystic mass behind the bladder disappeared.

Discussion

The patient in this case was raised as a male child and had normal scrotal development but showed hypospadias at birth. When he was first admitted to the hospital, he underwent surgical treatment for bilateral cryptorchidism for the left testis, which was confirmed by histological examination. However, the cryptorchidism in the right testis was not identified. On the second admission, he was diagnosed with cryptorchidism combined with hypospadias, while hermaphroditism was not considered. The chromosome karyotype was not examined, and simple urethroplasty was performed without checking the right sex gland. For the 2 times that the patient was admitted to the hospital, the diagnosis of hermaphroditism was missed. This shows that some medical units still lack an understanding for diagnosing hermaphroditism. Therefore, we should be vigilant in cases of children with bilateral cryptorchidism, hypospadias, and chromosome karyotype analysis, SRY inspection, sex hormone examination, and imaging tests should be performed in early childhood to identify the chromosome sex.

Postoperative pathology confirmed the presence of effusion from the uterus that was present behind the long strip-like cystic mass situated behind the bladder. The effusion may be caused by the following reasons: urethral stenosis occurred after urethroplasty; therefore, the patient had dysuria and urinary retention. Since the uterus was connected to the posterior urethra by remnants of the degraded Mullerian tube [1, 2], effusion was easily caused during uroschesis. Since the detrusor muscle rather than the uterine smooth muscle contracted during urination, the volume of the

cystic mass behind the bladder did not reduce after urination. Conditions such as mixed gonadal dysgenesis, incomplete degradation of the Mullerian system, and the presence of an additional uterus, vagina, or 1 fallopian tube can present as masculine insufficiency [3]. For this patient, the nature of the cystic mass behind the bladder was unknown at the time of previous operations. Uterine effusion was considered according to the medical history and chromosome karyotype. This case is clinically significant because it suggests that in the case of urethral stricture and urinary retention, the cystic mass behind the bladder may be the uterus with effusion in patients with hermaphroditism

A case of hermaphroditism with a chromosome karyotype of 45X/46XY can present as mixed gonadal dysgenesis or true hermaphroditism. In this case, pathological testing of the strip-shaped right gonadal tissue did not show presence of ovarian tissue, suggesting mixed gonadal dysgenesis. The strip-shaped gland was removed by using surgery for its malignant tendency [4, 5].

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