[Downloaded free from http://www.sjkdt.org on Thursday, January 24, 2013, IP: 137.215.193.100] || Click here to download free Android application for t

Saudi J Kidney Dis Transpl 2013;24(1):89-92
© 2013 Saudi Center for Organ Transplantation

Saudi Journal of Kidney Diseases and Transplantation

Case Report

Bladder Hamartoma: A Unique Cause of Urinary Retention in a Child with Goldenhar Syndrome

Ahmed Adam[1], Keshree Gayaparsad[2], Matthys J. Engelbrecht[1], Evelyn M. Moshokoa[1]

[1]Department of Urology, Steve Biko Academic Hospital, University of Pretoria; [2]Department of Anatomical Pathology, National Health Laboratory Service, Tshwane Academic Division, University of Pretoria, Tshwane, South Africa

ABSTRACT. The bladder hamartoma is an extremely rare entity. We report on its presence in a 5-year-old boy with Goldenhar syndrome. Most probably, this is the first report of a bladder hamartoma presenting with obstruction of the bladder outlet resulting in urinary retention. The obstructive lesion was resected endoscopically. This proved to be curative for the lesion, since the follow-up voiding cysto-urethrogram revealed only a negligible post-void residual volume. Although urogenital anomalies have a well-known correlation with the Goldenhar syndrome, the existence of the bladder hamartoma found in association with this syndrome, according to the best of our knowledge, has not been previously reported in the world literature. With this report being only the 11th described case of bladder hamartoma, we highlight on the management options for this exceptional histological finding. The incidence, screening, treatment decisions and important urogenital associations of the Goldenhar syndrome are also discussed.

Introduction

The Goldenhar syndrome (oculo-auriculo-vertebral syndrome) is a multifaceted entity composed of craniofacial anomalies that have been found to have an associated urogenital system anomaly in up to 70% of the cases.[1] According to the best of our knowledge, hamartomas of the urinary bladder have only been described in ten previous instances in the world literature, and have not been documented to present with urinary retention.[2,3] Here, we report a case of hamartoma of the urinary bladder presenting as a polypoid lesion causing mechanical obstruction to the bladder outlet in a child with Goldenhar syndrome.

Correspondence to:

Dr. Ahmed Adam,
Department of Urology,
Steve Biko Academic Hospital,
University of Pretoria,
Tshwane, South Africa
E-mail: aadam81@gmail.com

Case Report

A 5-year-old boy presented with a two-month duration of episodic urinary retention. He had a background diagnosis of Goldenhar syndrome with the following features: craniofacial asymmetry, ear microtia, perimembranous ventral septal defect (VSD), discrete subaortic membrane, kyphoscoliosis, lumbar hemivertebrae,

[Downloaded free from http://www.sjkdt.org on Thursday, January 24, 2013, IP: 137.215.193.100] || Click here to download free Android application for t

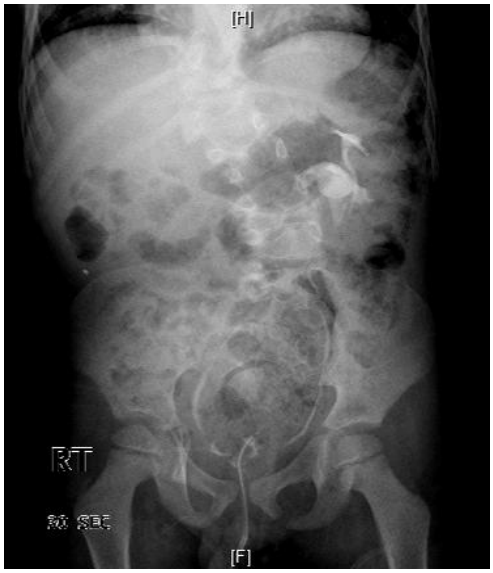

Figure 1. IVP demonstrating the fused right-to-left CRE. The lumbar scoliosis with a convex to the left is also seen.

anorectal malformation (ARM) and developmental delay. For this patient, a posterior sagittal ano-recto-plasty was performed for the ARM four years ago; however, the patient still has episodes of mild fecal incontinence.

On abdominal Ultrasonography, a fused right-to-left crossed renal ectopia (CRE) was observed without any hydroureter or hydronephrosis in both systems. The bladder wall appeared smooth and regular. The blood urea and electrolyte level along with the urine dipstick analysis was unremarkable. Intravenous pyelogram (IVP) (Figure 1) demonstrated the CRE with normal implant positions of both ureters. Cystoscopy revealed a mobile pedunculated lesion obstructing the bladder outlet, with a stalk attached distal to the trigone. This lesion was resected at its base and sent for pathological assessment.

Macroscopy of the resected specimen revealed a polypoid tissue mass measuring 0.5 cm × 0.3 cm × 0.2 cm. The attached stalk was 1.1 cm in length.

Histology revealed a polyp (Figure 2), covered by mildly hyperplastic stratified squamous epithelium. In the sub-epithelial stroma, round to oval-shaped von Brunn's nests were visible (Figure 3). Occasionally, some of the nests had

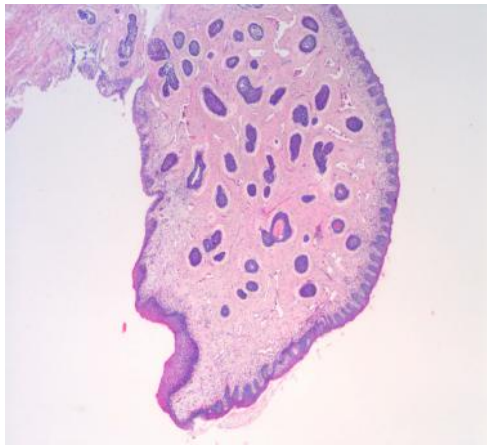

Figure 2. Histological view of the lesion showing a polyp comprised mostly of epithelial aggregates of von Brunn's nests. (polyp full view): (H&E) Magnification ×2.

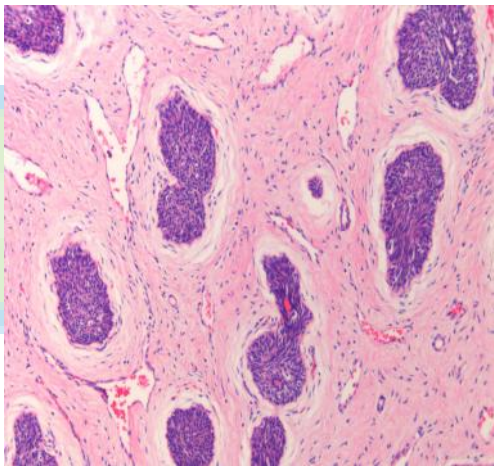

Figure 3. The von Brunn's nests with a surrounding fibromyxoid–vascular stroma (close-up): (H&E) Magnification ×10.

the appearance of cystitis cystica. The stroma surrounding these nests had a fibro-myxoid quality and appeared vascular. The above features were in keeping with that of a bladder hamartoma.

Voiding cysto-urethrogram (VCUG) performed six weeks post-resection revealed only minimal post-void residual volume. The above patient is planned for corrective surgery for the VSD and scoliosis and is to follow-up at urology for close surveillance of the corrected bladder outlet obstruction.

[Downloaded free from http://www.sjkdt.org on Thursday, January 24, 2013, IP: 137.215.193.100] || Click here to download free Android application for t

Discussion

A hamartoma is defined as an excessive focal overgrowth of normal mature tissue, with cellular elements similar to that of the organ of origin. It is only differentiated from the organ of origin due to hyperplastic change.[2,3] This lesion may occur within various organs in the body, particularly the spleen, liver, kidney and lung. It is however an extremely rare occurrence to be isolated in the urinary bladder.[2,3] The first report of a bladder hamartoma was described by Davis in 1949.[2,3] Since then, ten other cases (including our index patient) have been reported in the world literature and, to date, none have been described to have malignant potential.[2,3] As described here and in four previous reports, the location of this lesion has been found in approximation to the trigone.[2,3] Hematuria, pyuria, suprapubic discomfort and, now, urinary retention may be the presenting symptoms.[2]

Although treatment recommendations in the management of the bladder hamartoma have not been stipulated, partial cystectomy or transurethral resection have both been performed. Long-term follow-up with cystoscopy has also been advised.[2] Hamartomas of the urinary bladder have been described in association with the Peutz–Jeghers and Beck-with-Wiederman syndromes.[2,3] However, this is probably the first report of a bladder hamartoma found in association with the Goldenhar syndrome.

The Goldenhar syndrome is a morphogenetic anomaly that involves the first two branchial arches, and was first described by Von Arlt (1845).[4] This syndrome has an estimated incidence of 1:3500 to 1:5600 and, almost two centuries later, the exact etiology still remains unclear.[4,5] Associated facial anomalies include preauricular skin tags, epibulbar dermoids, ear microtia and facial assymetry.[5] The malformed ear along with conductive hearing loss may be a harbinger of an associated underlying urogenital system anomaly. This is the case with the Goldenhar syndrome along with the Brachio-oto-renal, Townes–Brock and Mayer–Rokitansky–Küster–Hauser syndromes.[5,6] The urogenital associations found in the Goldenhar syndrome include crossed fused ectopia (as presented in our index case), pelvic kidney, renal agenesis, left retrocaval ureter, ureteral duplication, multi-cystic kidneys and ureteropelvic junction obstruction.[1,5,7] Because the above associated incidence is significant, it has been recommended that all patients with the Goldenhar syndrome should have ultrasound screening at an early age and cystoscopic evaluation if needed. Patients should be adequately counseled with a view to close follow-up with regular visits.[5] It is through the early detection of renal anomalies that obstructed hydronephrosis and vesicoureteric reflux can be optimally managed. Children with unilateral renal agnesis are also at an increased risk of hyperfiltration-induced renal injury; thus, early recognition may always prove beneficial.[5]

Although an exceptionally rare finding, this case has added the bladder hamartoma as another point to the ever expanding list of possible causes of urinary retention. The above report also adequately emphasizes the point that the screening, diagnosis, associations and management of the Goldenhar syndrome should be familiar to any physician who claims guardianship of the urogenital tract.

Acknowledgments

The authors would like to thank Dr. R.H. Kusche (Department of Urology) for assisting with the initial patient management and Dr. M. Louw (Department of Anatomical Pathology), University of Pretoria, for the above photomicrographs.

References

1. Marulaiah M, Chattopadhyay AV, Rao P. Goldenhar syndrome and urogenital abnormalities. Indian J Urol 2003;19:167-9.
2. Ota T, Kawai K, Hattori K, Uchida K, Akaza H, Harada M. Hamartoma of the urinary bladder. Int J Urol 1999;6:211-4.
3. Brancatelli G, Midiri M, Sparacia G, Martino R, Rizzo G, Lagalla R. Hamartoma of the urinary bladder: Case report and review of the literature. Eur Radiol 1999;9:42-4.
4. Kulkarni V, Shah MD, Parikh A. Goldenhar

[Downloaded free from http://www.sjkdt.org on Thursday, January 24, 2013, IP: 137.215.193.100] || Click here to download free Android application for t

syndrome: A case report. J Postgrad Med 1985; 31:177-9.
5. Ritchey ML, Norbeck J, Huang C, Keating MA, Bloom DA. Urological manifestations of Goldenhar syndrome. Urology 1994;43:88-91.
6. Adam A, Ebrahim ZI, Engelbrecht MJ, Van Heerden IJ, Moshokoa EM. Mayer-Rokitansky-Küster-Hauser syndrome associated with a urogenital sinus anomaly in a 4-year-old child: Report of a case. Curr Urol 2009;3:103-5.
7. Ishitoya S, Arai Y, Waki K, Okubo K, Suzuki Y. Left retrocaval ureter associated with Goldenhar syndrome (branchial arch syndrome). J Urol 1997;158:572-3..