to show flow void areas within the lesion, suggesting aneurysm formation.

Discussion

In this case the diagnostic problem was that the patient’s medical records of the year 2000, after total resection of the primary cardiac tumour, revealed no evidence of residual myxoma tissue in the heart, albeit her cranial CT demonstrated multiple metastatic deposits widely scattered in both cerebral hemispheres. Two possible postulates can be offered to explain this phenomenon. Firstly, the embolisation of this myxoma would have taken place at the time of original resection of the tumour, and the deposits in the cerebral tissue would have grown ever since. Secondly, the myxoma that embolised at that time must have lodged in the cerebral vasculature, forming a mycotic aneurysm, which ruptured some years later releasing the contents, and the local invasive property of the myxoma led to the florid lesions. Although the magnetic resonance angiography failed to show aneurysm formation, the possibility cannot be ruled out without performing a digital subtraction angiography.

Emboli occur at a frequency of 30–50% with myxoma. Left atrial myxoma gives rise to systemic embolisation. In the majority of cases cerebral arteries are involved. Transient or permanent visual loss may result from involvement of retinal arteries. The behaviour of embolised tumour fragments, within the central nervous system is controversial. In rare instances tumour fragments continue to grow and thus create a symptom complex, compatible with an expanding intracranial mass [5,3]. A rarer delayed complication of embolised myxoma is arteriolar aneurysm at the site of embolisation [1,2]. These aneurysms enlarge progressively and can cause neurological symptoms, years after the removal of the primary tumour [4]. Rupture of such aneurysms has been reported [1,2,5]. Angiographic studies have shown aneurysmal change in cerebral vasculature [2]. It has been postulated that a tumour embolus weakens the arterial wall and permits viable myxoma cells to penetrate the damaged endotheium, causing weakening of the media, and aneurysmal dilation. A cerebral arteriogram may demonstrate this as a filling defect.

As this patient had evidence of multiple lesions, a surgical removal was not feasible. Our patient was managed with intravenous steroids to reduce the cerebral oedema and antiepileptic medication to control the fits.

References


An unusual presentation of female urethral leiomyoma

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(Index words: Diagnostic approach, histology, urethral and bladder reconstruction)

Abstract

Leiomyoma of the urethra is a rare clinical entity. We report an unusual presentation of this tumour, which led to a diagnostic and surgical dilemma. The patient was a 16-year old female who presented with a labial mass which was palpable abdominally. Imaging methods and laparoscopy demonstrated a well defined soft tissue mass arising from the pelvis, without any obvious involvement of the urinary tract. The tumour (8 × 10 cm) was completely excised with reconstruction of the bladder and urethra. Histology confirmed a cellular leiomyoma.

Introduction

Urethral leiomyoma is rare and only about 50 cases have been reported in literature. This benign smooth muscle tumour often presents as a midline extramucosal vaginal mass, which is often mistaken for a urethral diverticulum, fibroid or prolapse. We report an unusual presentation of a large lateral tumour, which led to a dilemma in diagnosis and surgical approach.

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Case report

A 16-year old unmarried girl presented with a history of progressively enlarging painless lump at the vulva for 6 months. Other than the discomfort it caused during walking, there were no symptoms. Examination of the external genitalia revealed a visible and palpable lump under the right labium majus, which was firm, spherical and non-tender. The urethral meatus and vaginal opening were distorted and deviated to the opposite side. Bimanual palpation per rectum revealed the tumour to be extending as a mobile pelvic mass.

Abdominal ultrasound and CT scan demonstrated a well defined pelvic soft tissue mass lying between the bladder and the rectum, displacing the uterus and vagina towards the left side. It extended superiority up to the lumbosacral articulation and inferiorly to the right labial fold. There was no sign of infiltration into the surrounding structures (Figure 1).

Diagnostic laparoscopy showed a solid mass protruding postero-laterally between the bladder and the uterus. Trans-abdominal exposure showed a large (8 x 10 cm) retroperitoneal tumour attached to the posterior urethral wall. Complete excision of the mass was done, which resulted in a partial urethrectomy. The bladder and the urethra were reconstructed with tabularised anterior bladder wall. Presence of the J stent was a useful guide to ureteric orifices. The recovery was uneventful with good urinary continence.

Macroscopically the tumour tissue was soft, tan, with a uniform cut surface. Microscopy revealed a cellular tumour with interlacing bundles of spindle cells with abundant eosinophilic cytoplasm and elongated blunt-ended nuclei with minimal cellular atypia. This appearance was consistent with the cellular urethral leiomyoma (Figure 2).

A differential diagnosis of an ovarian neoplasm or retroperitoneal teratoma was contemplated. Tumour markers were negative. Radiologically there was no involvement of the urinary tract. Pre-operative ureteric stenting was attempted to prevent operative trauma, and was successful only on the right side. Urethral mucosa was intact and appeared normal during cystourethroscopy.

Discussion

Leiomyoma of urethra is rare, with a strong female preponderance. The exact aetiology and pathogenesis remain unclear, though an endocrine dependency is suspected as several cases of urethral leiomyoma were reported to enlarge during pregnancy and regress after delivery [1–3].

The morphological features of urethral leiomyoma are similar to its counterparts elsewhere. They tend to grow as isolated, encapsulated, oval or spherical masses of varying diameter, ranging from few millimeters to several centimetres, with the largest reported up to now measuring 8 cm [3]. The most common site is the posterior wall of the urethra. The common clinical presentations include periurethral or vaginal mass, dysuria, dyspareunia, haematuria, and rarely obstructive urinary symptoms [1,2,4–6]. In the presence of a urethral mass it may be confused with urethral diverticulum, caruncle, papilloma and carcinoma.

The recommended treatment is complete local excision through the vagina. Large tumours as in the present case, need a trans-abdominal approach with urethral and...
bladder neck reconstruction. Prognosis is excellent as recurrence is very rare and malignant transformation is not known to occur [7].

A labial mass with intra-abdominal extension, as in the present case, is an extremely rare presentation which can lead to a diagnostic dilemma. This case illustrates the importance of surgical expertise for urethral reconstruction.

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Case reports

Distal aphalangia, microcephaly and mental retardation

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(Index words: Distal aphalangia, microcephaly, seizures, consanguinity, mental retardation)

Case history

A 3-year old boy from Hatton presented with generalised convulsions. He was the third child born to consanguineous parents. The antenatal and perinatal period of this child had been normal. At 3 months of age he developed generalised convulsions and was treated with phenobarbitone at the Nawalapitiya Base Hospital. However, subtle seizures persisted at a frequency of about 1 or 2 seizures a month. At about 15 months of age the seizure frequency increased. He also had global development delay.

On examination he had dysmorphic features. The occipito-frontal circumference was 46 cm (<3rd centile, [1], length of child 89 cm (< 3rd centile) and weight 13.5 kg (between 10th and 25th centile).

The dysmorphic features were mainly confined to hands and feet. The distal phalanges of all the four fingers of the right hand and fourth and fifth of left hand were hypoplastic. All affected fingers showed some degree of camptodactyly and nails were absent. Both thumbs and, left index and middle fingers appeared normal (Figure 1). Xray of the hand revealed that the distal phalanges of the affected fingers were absent. The distal phalanx of the left index finger was hypoplastic. Other bones of the both hands were normal.

All toes appeared short. There was overriding of the left fourth toe. Toenails appeared normal (Figure 2). Xray of both feet showed that there were only two phalanges in each toe. Phalanges of both big toes were normal in size and shape. The distal phalanges of all other toes were hypoplastic (acro-osteolysis). There was duplication of the proximal phalanx of fourth toe (Figure 3). Neither hands nor fingers showed polydactyly or syndactyly.

He had generalised hypotonia and global development delay. He could sit without support but could not stand. He reached out for objects but the pincer grasp was poor.

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