Case Reports

CONGENITAL ANOMALY PRESENTING AS LUMP ABDOMEN IN ADOLESCENT GIRL

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Introduction

The close embryological proximity of the mullerian, wolffian and metanephric systems increases the potential for a common ipsilateral embryological error around the fourth week of gestation. Genital anomalies are four times as common in females as in males with unilateral renal agenesis. Uterine anomalies associated with congenital renal agenesis and skeletal abnormalities represent an uncommon pathology that often presents important diagnostic and therapeutic problem [1]. The Mayer-Rokitansky-Kuster-Hauser syndrome is an eponym often applied to individuals with mullerian duct anomalies in association with a solitary kidney. We present a rare variant where left sided renal agenesis was associated with a large ipsilateral ovarian cyst and a bicornuate uterus. The left sided cornua was not communicating with the cervix hence there was ipsilateral hemihematometra. We could not find a report of this particular triad inspite of an extensive search of literature.

Case Report

An 18 year old girl presented with pain in the lower abdomen of one year duration. The pain was dull, aching, continuous and non-radiating. Additionally she used to have severe left lower abdominal pain with menstruation periods. She attained menarche at 16 years of age and periods were scanty although regular. She was married for 3 months and complained of dyspareunia.

Clinical examination revealed a large 15X12 cms cystic lump arising from the pelvis which the patient was unaware of. It was bimanually palpable on per-rectal and per-vaginal examination. A clinical diagnosis of left ovarian cyst was made.

Ultrasound revealed an 11x13x12.5 cms pelvic mass of mixed echogenecity with cystic and solid areas suggestive of left ovarian cyst/dermoid. Additionally the left renal fossa was empty and left kidney could not be visualized. Intravenous urogram and DTPA

renal perfusion seen confirmed an absent left kidney. The right kidney was normal.

At staging laparotomy, the left fallopian tube was stretched over and inseparable from the large ovarian cyst. The uterus was bicornuate with larger left cornua (Fig-1,2). On aspiration, old collected blood came out of the cornua suggesting that it was blind and the cause of the patients' symptomatology. Hence a left hemihysterectomy with a salpingo-oophorectomy was performed preserving the right hemiuterus, ovary and fallopian tube for fertility. Washings were taken from both subphrenic spaces and paracolic gutters for cytological examination. An omental biopsy was also taken. Post operative recovery was uneventful. Presently the patient is symptom free. Histo-pathological examination revealed a follicular cyst of the left ovary and a blind left hemiuterus. There was no evidence of malignancy.

Discussion

The association of renal agenesis with anomalies of mullerian ducts has long been recognized. However the precise relationships during development have re-

Fig. 1 : Bisected specimen of excised blind cornua of uterus with fallopian tube of same side and benign ovarian cyst

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Fig. 2 : Sketch diagram of pathology showing bicornuate unicervical uterus with no communication of cavity of the left cornua to cervix and filled with old dark, altered blood of menstruation. Ipsilateral ovarian cyst stretching the fallopian tube is also seen

mained elusive. Rokitsky anomalad refers to a group of associated anomalies that include unilateral renal agenesis or renal ectopia, ipsilateral mullerian defects, unicornuate or bicornuate uterus, vaginal atresia/agenesis, hematocolpos and genital duplication [2].

30% of women with uterine malformation have a demonstrable urinary tract anomaly. Conversely, women with urinary tract anomalies have a higher incidence of mullerian duct anomalies. In addition, bony skeleton anomalies can occur in 12% of cases especially those of the lumbar and cervical spine and limb abnormalities. Mullerian aplasia has been noted to occur in association with hypoplastic thumbs [3]. Hence renal agenesis should be suspected in any female with a uterine or major vaginal anomaly. A thorough work up including ultrasonography and intravenous urography is indicated and cannot be overemphasized. Intravenous urography and ultrasound in combination is probably comparable to magnetic resonance imaging in permitting an accurate preoperative diagnosis thus allowing early and appropriate surgical intervention [4].

Various classifications of uterine malformations are available covering all aspects, from embryological to functional. From a practical standpoint the most common anomalies are the didelphic uterus, bicornuate uterus (unicollis or bicollis) and septate uterus (partial or complete). Each has distinct anatomical characteristics and different clinical implications. Abnormalities of urinary tract can be found in conjunction with all three uterine malformations. Obstructive malformations almost always occur with renal agenesis on

the obstructed side. In our case, the problem was of bicornuate uterus with blind left hemiuterus resulting in ipsilateral hematometra along with renal agenesis on the same side. This complex has been designated as Type II unilateral renal agenesis representing faulty differentiation of the mesonephric duct, ureteral bud and a laterally placed mullerian duct that forms a blind pouch [5].

Congenital malformations of the vagina, cervix and uterus, although rare, may have profound implication for the young gynaecological patient. These anomalies are often detected in adolescent period. For proper management, the physician requires a thorough understanding of normal embryology and sexual differentiation. Each individual who presents with a defect must be thoroughly evaluated because genital tract aberrations don't necessarily follow any defined and consistent pattern. Genital malformations can be particularly disturbing to the patient and her family because they not only have reproductive implications but also significant psychological and sexual overtones that need to be addressed and dealt with in a sensitive and reassuring manner [6]. Although a congenital anomaly, diagnosis of this anomaly is usually not made until menarche. Patients may present with cyclical abdominal pain, inability to conceive, dyspareunia, frequent loss of pregnancy and premature labour etc. The confusing clinical sign is the associated menstruation from the other side and the diagnosis may be missed if a careful examination is not performed in teenagers. The course can be complicated by abscess formation and malignant transformation but most patients remain symptom free [7]. There may very rarely be familial occurrence of renal and mullerian duct hypoplasia, craniofacial anomalies, severe growth and developmental delay [8]. If agenesis of the kidney or malformation of the uterus is found in a patient, the physician should look for combined malformation in other members of the family [9].

These malformations have been linked to mutations in the WT1 gene. A recent study from Boston, Massachusetts USA shows that absence of WT1 gene mutations in patients with congenital absence of the uterus and vagina disproves this hypothesis [10]. Lindenman has hypothesized that activating mutations of either the antimullerian hormone gene or its receptor gene may cause mullerian duct regression in a genetic female during embryogenesis [11]. Early accurate diagnosis and a thorough workup are essential for complete relief of symptoms and preservation of reproductive potential. Candiani et al reviewed the experience of the Milan University First Dept of Obstetrics and

Gynaecology and studied 36 patients. They concluded that early accurate diagnosis after menarche followed by excision and marsupialization of the blind hemivagina offers complete relief of symptoms and preserves reproductive potential. Partial morphologic changes are evident but metabolic modifications comparable to those of the adjacent normal vagina have not yet been documented [12]. Appropriate counseling about the potential long term risks to the solitary kidney should be explained to the patient and family. A greater awareness of the syndrome should lead to accurate diagnosis [13].

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