Case Report

Solitary kidney and bicornuate uterus in mother and child

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Introduction

Congenital solitary kidney occurs in 1–3/1000 births and is occasionally associated with hypertension renal failure in later life [1]. Although usually sporadic, this malformation can be inherited either in isolation or as part of a multiorgan disorder such as the branchio-oto-renal and Kallmann syndromes [1]. We now report a pedigree with unilateral renal agenesis associated with a malformation of the female genital tract, the bicornuate uterus.

Case

The index case (Figure 1) presented at the age of 14 years with a 5-day history of abdominal pain following her second period. On ultrasound scan (USS) a pelvic mass was noted and the right kidney was not visualized. At operation the mass comprised the right-sided entity of a bicornuate uterus and double cervix which was distended with endometrial blood: the cervical opening to the vagina was absent. An opening was fashioned in the right cervix and a vaginal septum was excised. Subsequent investigation revealed a normal blood pressure (120/70 mmHg) and plasma creatinine (65 μmol/l) with a solitary, hypertrophied left kidney as assessed by intravenous urogram (Figure 2a). 99mTc-Technetium-dimercaptosuccinic acid (DMSA) scan (Figure 2b) demonstrated no functional kidney tissue on the right, thus excluding an ectopic organ.

The mother of the index case was known to have the same uterine malformation and she also had an absent right kidney with a normal organ on the left as assessed by USS and intravenous pyelogram: she was normotensive. The non-identical twin sister of the index case, the second sister and the father of the index

Fig. 1. Family pedigree with affected females shown by solid circles and index case indicated by an arrow.

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Inherited urogenital anomalies case were subsequently assessed by USS and each had two, normal kidneys (Figure 1).

Discussion

The bicornuate uterus and cervix represent a degree of failure of Mullerian duct fusion. Lesser degrees of this phenomenon generate a bicornuate uterus connected to a single cervix, while a more severe phenotype is the double vagina with uterine and cervical anomalies (uterus didelphys). Heinonen [2] analysed 26 cases of uterus didelphys and found that dysmenorrhoea, dyspareunia, and leukorrhoea were the commonest symptoms. The presentation of unilateral vaginal/cervical obstruction with abdominal mass usually occurs in the teenage years (haematocolpos) but has also been recorded in neonates (hydrocolpos). In one study, 17 affected subjects had intravenous pyelograms, and four had a solitary functioning kidney [2]. In all cases the absent kidney is on the same side as the obstructed Mullerian derivatives. A literature review reveals that diverse mesonephric duct derivative and metanephric malformations, including duplex [3] and pelvic kidney as well as Gartner’s duct cyst, are less commonly associated with the double uterus.

Although the association of Mullerian duct fusion anomalies with renal agenesis has become relatively well recognized by the obstetric community, the syndrome is perhaps less familiar to nephrologists. Moreover, published reports of these anomalies in more than one generation are rare. Our kindred is compatible a dominant gene on an autosome or the X chromosome. In fact it is notable that the syndrome was absent in the non-identical twin of our index case, consistent with a genetic vs a teratogenic aetiology. Biedl et al. [4] described two families compatible with autosomal dominant transmission in which affected members had unilateral or bilateral renal agenesis and a spectrum of uterine canal anomalies including fusion malformations or vaginal atresia, sometimes accompanied by absent uterus (Rokitansky–Kuster–Hauser syndrome). Interestingly, some males in this report [4] were affected by renal agenesis and absence of the vas deferens or seminal vesicles, indicating that the disease is not sex limited. Recently, Battin et al. [5] reported a another family with renal and Mullerian congenital anomalies.

Regarding candidate genes for this syndrome, we suggest that mutations of PAX2 should be sought based on the facts that this transcription factor gene is expressed in the human Wolffian (mesonephric) duct and Mullerian (paramesonephric) ducts (see figure 2F in reference [6]) and null mutations in mice inhibit both metanephric and Mullerian differentiation [7].

In conclusion, based on our current report and that of other kindreds [3–5], we suggest that it would be reasonable clinical practice to screen first-degree relatives of patients with Mullerian and renal malformations to identify individuals with congenital renal anomalies.

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References


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