Congenital genitourinary malformation in a woman with three primary gynaecological tumours: synchronous findings never before described

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DESCRIPTION

The authors describe the case of a 69-year-old nulliparous White woman without any relevant personal or family history who, in the course of the study of a progressive increase in abdominal circumference, underwent an abdominal-pelvic ultrasound, which revealed a large pelvic mass, lateralised to the left, suggestive of fibroid, and a single left kidney. The pelvic CT confirmed the ultrasonographic findings and showed left ureterohydronephrosis with signs of nephropathy in relation to extrinsic compression (figures 1 and 2). Renal function was normal. The patient underwent exploratory laparotomy, and a bulky lobulated pelvic mass occupying the entire pelvis, extending cranially to the umbilical plan, apparently suggestive of a large uterine or ovarian fibroid, was visualised. Washed peritoneal fluid (ascites did not exist) and debulking surgery with total hysterectomy and bilateral adnexectomy were performed (figure 3). The cytology was negative for malignant cells and histological results revealed a fibroid of the left ovary of about 2500 g, right unicornuate uterus

Figure 3 Right unicornuate uterus with a rudimentary communicating type left uterine cavity. Large left adnexal tumour, histological examination of which revealed an ovarian fibroid (A—posterior image of the uterus and uterine attachments/pelvic tumour, B—side image of the bulky left ovarian fibroid).

with rudimentary left horn, both with complex endometrial hyperplasia with atypia, and bilateral fallopian tube in situ serous carcinoma (serous tubal intraepithelial carcinoma with the *p53* signature sparing the fimbriated end of the fallopian tube). As per advice from the multidisciplinary group, no complementary treatment was provided. The patient was asymptomatic through 2-year follow-up.

Figure 1 CT showing a large pelvic tumour of about 218.1×131.0×209.8 mm (A—axial section, B—sagittal section).

Figure 2 CT with contrast highlighting a single left kidney with ureteropelvic dilation of about 32.0 mm (A—axial section) and left ureterohydronephrosis with maximum anteroposterior diameter of 17.1 mm (B—sagittal section).

Learning points

- ▶ The authors report the case of a never before described association of a right unicornuate uterus with a rudimentary left horn, both with complex endometrial hyperplasia with atypia, a bulky fibroid of the left ovary and bilateral fallopian tube in situ serous carcinoma in a woman with a single left kidney.
- ▶ Congenital abnormalities of the Müller ducts can lead to a number of conditions—with many small variations from the norm—that are asymptomatic and do not require treatment. The association of uterine malformations with congenital abnormalities of the urinary tract is common, owing to their relationship in embryonic development (urinary malformations are present in 20–30% of women with abnormalities of Müllerian ducts). The unicornuate uterus corresponds to about 5% of Müllerian duct anomalies (type II, according to the American Fertility Society classification system), occurring in the general population, with an estimated prevalence of 1:4020. This congenital anomaly occurs due to failure of the

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development of the Müllerian ducts (often partially) and is frequently associated with a rudimentary cavity that can communicate with the main cavity of the uterus and has functional endometrium. In 2012, the European Society of Human Reproduction and Embryology, and the European Society of Gynaecological Endoscopy, proposed a new clinical classification system for congenital uterine malformations, based on anatomical changes from the normal uterus, taking into account their embryological origin. According to this classification, the uterine malformation reported in this case forms part of the U4a, C0, V0 class (hemi-Uterus (U4) with rudimentary uterine cavity (a), normal Cervix (C0) and normal Vagina (V0)).¹

- Synchronous gynaecological tumours in two organs, usually ovary and endometrium (50–70% of cases), are rare, occurring at a prevalence of 1–2% of women with gynaecological malignancy. In turn, triple gynaecological cancer is extremely rare. There are three published clinical cases of primary synchronous tumours of the ovary, endometrium and fallopian tube. In these cases, the ovarian tumours belonged to the epithelial type. The aetiology of synchronous gynaecological tumours is uncertain, but Lauchlan (1968) suggested a possible simultaneous interaction between the Müllerian system, hormones and/or carcinogenic agents at the origin of tumour tissues with the same embryogenesis. Risk factors for synchronous gynaecological malignancies include young age (premenopausal women), obesity, pelvic endometriosis, nulliparity, family history, immunological and genetic defects, prolonged exposure to carcinogens, radiation and chemotherapy for the primary cancer, and field cancerisation (when the body is exposed to carcinogens, other organs besides the organ with cancer are also exposed to the carcinogen and carry a high risk of cancer). The use of combined hormonal contraception can be a protective factor.^{2 3} In this case, only nulliparity is present. Ovarian fibroid is a benign or low malignant potential ovarian stroma tumour, usually without hormonal activity, and therefore not associated with endometrial hyperplasia. Despite this, malignant tumours of the fallopian tube are very rare, accounting for less than 1% of cancers of the female genital tract; they have been described to have an increased risk in women with mutations of the breast

cancer susceptibility 1 and 2 genes (*BRCA 1* and 2 genes), and 40–50% of cases are bilateral. The existence of these three synchronous tumours of the female reproductive tract can be linked to Peutz-Jeghers syndrome, but the lack of skin pigmentation disorders (skin and mucosa), and the absence of typical personal and family history of gastrointestinal polyps excluded this diagnosis in our patient. The Lynch syndrome can also be associated with the development of synchronous gynaecological tumours.^{2 3} Given the age of the patient and the absence of family history, particularly of breast and ovarian cancers, this association may have originated at some point from a genetic defect that occurred *de novo* in this patient, so no genetic study was performed. The aetiology of the clinical association presented is unknown and it may represent an incidental finding during the study of a pelvic mass. However, the occurrence of fallopian tube carcinoma is very rare especially when related to genetic predisposition, so it is possible that this case is an account of a never before described syndrome.

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Contributors EV-F, CM, LC and PS evaluated the patient in inpatient clinic. PS followed the patient in outpatient clinic. PS and EV-F performed the surgery. EV-F collected the data and wrote the manuscript. All the authors were involved in the conception of the work and revised it critically for important intellectual content. All the authors approved the final version to be submitted/published.

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