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Case Report of a 20-Year Diagnostic Delay; Difficulty in Diagnosing a Uterine Anomaly

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ABSTRACT

This is the case of a 46-year-old woman who first presented to gynaecology clinic aged 26 with severe dysmenorrhoea. She has medical history of left renal agenesis and a previous left oophorectomy. Over the course of 20 years she was extensively imaged with ultrasound (US), magnetic resonance imaging (MRI) and computerised tomography (CT) with the diagnosis of a right unicornuate uterus with an obstructed left rudimentary horn only made recently following 11 scans. There are many factors in this case that confused the picture and made diagnosis more challenging including an incomplete history, language barrier, possibility of a pelvic kidney and changes in imaging of an obstructed horn.

Keywords

Anomaly, Diagnosis, Horn, Unicornuate, Uterine.

Case Report

This 46-year-old woman had a long journey towards diagnosis and treatment of a uterine anomaly. She is gravida 0 and never been sexually active with no smear tests. Past medical history includes surgery aged 16 in Turkey for an ovarian cyst where she believed her left ovary had been removed, left renal agenesis and raised prolactin (arachnoid cyst visualised on MRI but no pituitary lesion).

She first presented to gynaecology clinic aged 26 in 2001 with a 10-year history of left sided pelvic pain and an ultrasound suggestive of a uterine anomaly (which was not mentioned in any subsequent imaging). She had a total of 16 follow up appointments, only coming to a full diagnosis and understanding of her symptoms in 2020. Her main symptom was dysmenorrhoea, further complicated by irregular periods and menorrhagia. Over the years she tried various treatments with varying success including mefenamic acid, combined oral contraceptive and progesterone only contraceptive of different regimes. She did not proceed to laparoscopy.

During this diagnostic process she underwent many scans which are summarised in table 1, with MRI images shown in figure 1. She was also referred to general surgery following imaging in 2015 which suggested a pelvic kidney. She had a hysteroscopy in 2019 with benign histology from a cervical polyp and the endometrium (anomaly not noted). The left sided mass had been slowly increasing in size across all her imaging and was now approaching 10cm. It was finally at this point in 2020 after review of all her imaging that the possibility of a uterine anomaly was discussed with the patient. Her final diagnosis is right unicorn ate uterus with a left sided rudimentary obstructed horn with associated endometrioma and adhesions. She was started on continuous progesterone suppression to prevent further obstruction of this horn and her symptoms are finally improving. She would now like to explore her fertility.

Discussion

Congenital uterine anomalies result from embryonic maldevelopment of the Müllerian ducts. Prevalence is estimated at 5.5% in the general population and is increased to 8% in infertile women [1]. This case demonstrates the difficulty of diagnosing uterine anomalies, and its need to be considered as a diagnosis. This patient has left renal

Table 1: Summary of imaging performed.

Year	Imaging	Report
2001	US pelvis	Rudimentary left uterine horn, normal ovaries.
2003	US pelvis	Poor views so no report given.
2006	US pelvis	Normal
2007	CT AP	Highly abnormal appearance of the left sided ovary, showing a solid well-defined mass localised under the abdominal wall, measuring 7.5x 4.6 x 4.1 cm. No left sided kidney was seen.
2011	US pelvis	Elongated anteverted uterus measuring 10.5 x 3.19 x 3.95 cm with normal myometrium. There was a very thick endometrial plate measuring approximately 1.5cm. The left ovary was not well visualised. The right ovary appeared normal with a small functional cyst. No other adnexal masses seen.
2015	US pelvis	Anteverted normal appearing uterus with endometrial thickness 8mm. Right ovary was normal. On the left side there was a solid inhomogenous mass in the adnexa measuring 82 x 71 x 57mm. This was suspected to be ovarian in origin.
2015	MRI pelvis	Endometriosis with endometrial masses in the left peritoneal and retroperitoneal pelvis. MDT review 2015 - ? pelvic kidney
2019	US pelvis	Anteverted uterus which was normal in size shape and echotecture. The ovaries were obscured by bowel gas and a 9.6 x 6.0 x 5.5cm left iliac fossa solid highly vascularised mass was seen. The left kidney was not seen.
2019	CT AP	The lesions were thought to represent haemorrhage or abscess, and could also be in keeping with endometriosis. A previous report on this scan did mention the possibility of a uterine anomaly however this report was apparently authorised in error and there is documentation saying to ignore it.
2019	US pelvis	Normal uterus and midline echo, with a normal right ovary. There was a heterogenous longitudinal mass with internal vascularity measuring 9.2 x 5.4 x 7.1 cm. This was thought to be an ectopic left kidney.
2020	CT urogram and DMSA	No evidence of any functioning renal tissue seen in the left iliac fossa masses, and they do not resemble normal renal architecture.

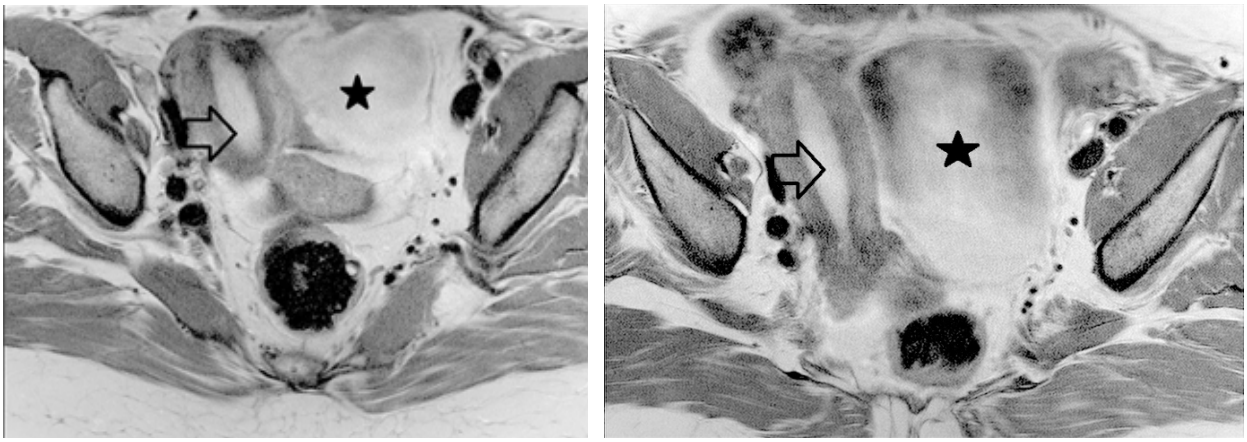

Figure 1: MRI of patients' uterine anomaly. Unicornuate uterus ⇨. Obstructed horn ★

agenesis; 70 – 89% of patients with unilateral renal agenesis also have a genital anomaly [2] therefore a uterine anomaly should have been considered. We often find these anomalies incidentally or when investigating subfertility (where most research is focussed), but it should be remembered that they can present in a variety of ways and more unusual diagnoses need to be thought of if a diagnosis is not found, imaging is confusing, or treatments are not working.

Many imaging techniques have been used for diagnosis including US, hysterosalpingography (HSG), sonohysterography and MRI. 2D US is the most common imaging performed on women and is often the initial imaging to flag anomalies. Unfortunately it is operator dependent and gives variable results unlike 3D US, which is more reliable but does require specialist training. 3D US is the gold standard for diagnosis of Mullerian anomalies and is cheaper, less invasive and more accessible than MRI with evidence to suggest reproducibility and good concordance between sonographers [3]. MRI is comparable to 3D US but requires specialist interpretation and is reserved for complex or inconclusive cases [4-6]. Invasive techniques such as laparoscopy and hysteroscopy are not often needed due to the advances of imaging [7]. HSG also has good concordance with 3D US, however this is more invasive with exposure to radiation, and it cannot differentiate between endometrium and myometrium, therefore is less helpful [8].

This patient did not have any 3D US, but she did have many 2D US (6) and MRI scans. MRI is comparable to the gold standard, therefore why was her anomaly not detected? There may have been some confusion in the appearance of the rudimentary horn, it would have appeared as a blood-filled structure in imaging due to its obstruction, therefore may look more like an endometrioma. Uterine anomalies are not common, therefore the reporting radiologist may not have seen one before and may not have considered it as a diagnosis, especially if it was not specified on the imaging request. The question over her absent kidney being a pelvic kidney also confused the picture and acted as a red herring

in the diagnosis of her condition. She did not have any records from Turkey with her previous surgical history, so could not be confirmed whether or not her ovary had been removed in the previous surgery or whether it was just the cyst.

This patient also did not attend many of her appointments and initially had a language barrier making confirmation of history difficult, and assessment of treatments less useful as some appointments were years apart.

This patient is only just thinking about becoming a mother. At 46 years old this is much more difficult and it could be wondered whether an earlier diagnosis may have made a difference for her in this regard. Uterine anomalies are known to lead to adverse reproductive outcomes including miscarriage, preterm labour, small for gestational age, malpresentation, and increase in perinatal mortality [9] (significance depends on the specific anomaly), and the fertility and obstetric complications have been well researched. Older women with features such as dysmenorrhoea or abnormal bleeding should be investigated as prompt treatment may improve reproductive outcomes [10].

In conclusion, common things are common, however the less common do need to be considered, especially when there are clues to its presence.

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