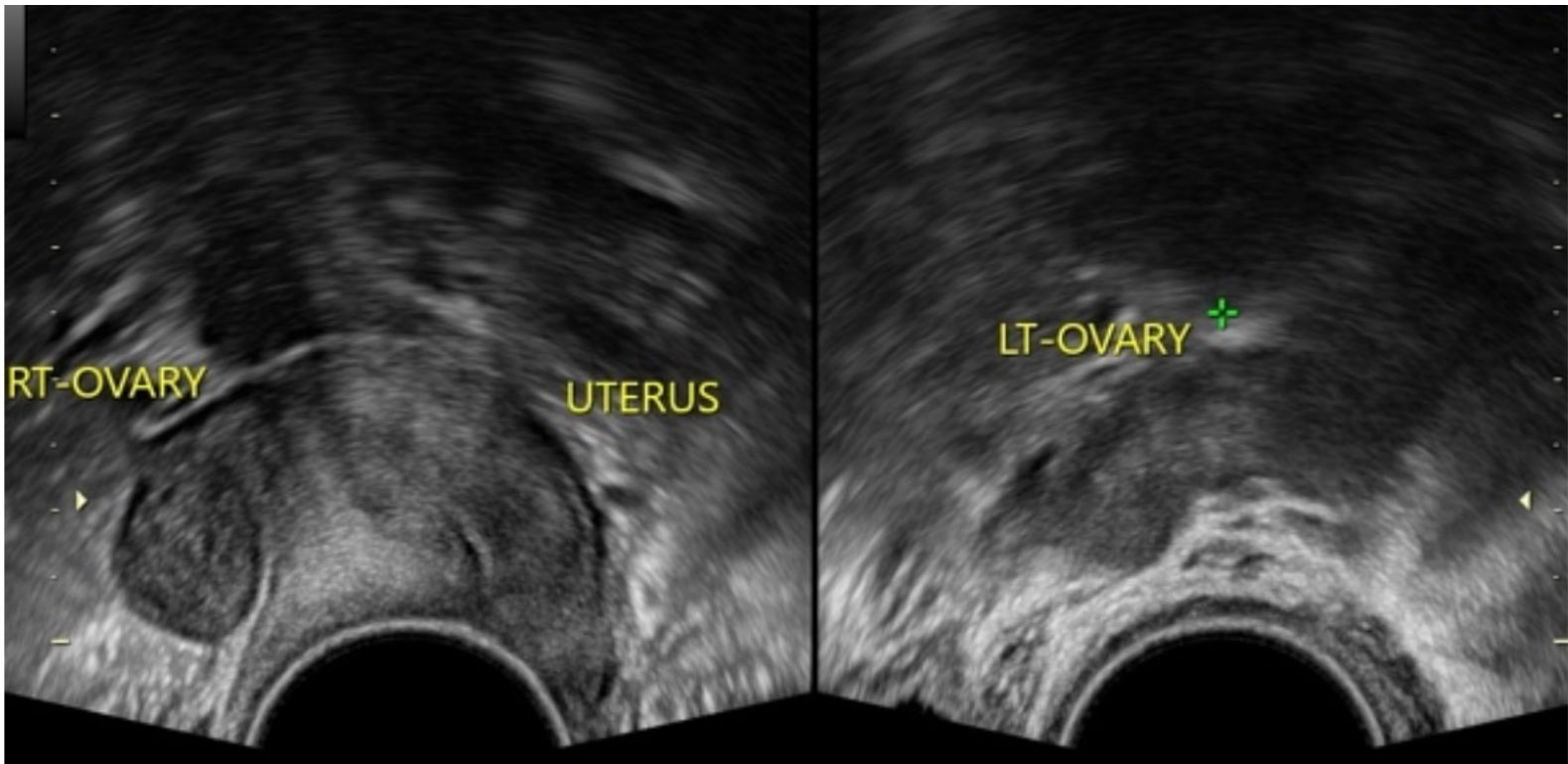




INTRODUCTION

- Gonadal dysgenesis is a group of rare heterogeneous disorders.
- The spectrum of disease includes both primary and secondary amenorrhoea.
- Mayer-Rokitansky-Küster-Hauser syndrome (MRKH) is a syndrome whereby typically Mullerian structures are absent or rudimentary, associated with genitourinary and/or skeletal anomalies [1].
- Ovarian anomalies have a reported prevalence in MRKH of 5–10% including hypoplasia, streak ovaries, malposition and tumours [3].
- The literature describes limited case reports where gonadal dysgenesis and MRKH co-exist.
- We describe an atypical presentation of uterine hypoplasia and ovarian dysgenesis with secondary amenorrhoea

CASE PRESENTATION

- We present a 26 year old Nullip presenting to gynaecology outpatients with a 18-month history of secondary amenorrhoea & primary infertility
- Previously her periods were regular but ‘light’ since menarche at age 15.

CASE PRESENTATION

- She had full development of secondary sexual characteristics. Normal sexual function with no dyspareunia. No PMH of note.
- OE:** A normal BMI of 20. Abdomen soft, non-tender, no organomegaly. Normal Vulva/vagina and cervix.

Ultrasound Pelvis

- retroverted hypoplastic uterus, measuring 38mm x14mm x20mm.
- No increased vascularity.
- Both ovaries were hypoplastic with small immature follicles.
- The right ovary volume was 1.8mls, left ovary volume 0.6mls.
- No adnexal masses

- She had responded to a progesterone challenge however, bleeding remained light.
- In the interim she was commenced on combined oral contraceptive pill with high oestrogen component and referred for further investigation to a specialist centre for genetic testing & endocrinology review

Test	Result
Haemoglobin	126 g/l
Renal function	Normal
Thyroid function	Normal TSH 0.95
FSH	<1 unit/l
LH	<1 unit/l
Oestradiol	46pmol/l
Progesterone	<1
SHBG	Low 16nmol/; (32-128)
Testosterone	0.5 nmol/l (0.3-1.7 nmol/L)

DISCUSSION

- Typically MRKH is not included in the differential diagnosis of delayed menses which can lead to misdiagnosis [2]
- Preeti et al. (2020) highlight the importance of seeing peripheral oestrogenisation, a hormone profile & karyotype to facilitate diagnosis in such cases. Cases of mis-diagnosis of gonadal dysgenesis coexisting with Mullerian agenesis have been reported and later found to be pure gonadal dysgenesis only [1].
- El-Feky (2021) described a similar case with uterine hypoplasia & ovarian dysgenesis however, the patient had primary amenorrhoea.
- Differential diagnoses of hypogonadotrophic hypogonadism should be evaluated in the diagnostic work up.
- Sensitive Counselling regarding longer term effects of a lack of oestrogen impact on quality of life and fertility potential are important.
- Including counselling on treatments e.g. HRT.
- Longer term surveillance maybe required and beneficial in such cases due to the paucity of evidence regarding the natural progression of their condition.
- This case highlights the need for further research and increased awareness of gonadal dysgenesis and atypical MRKH presentations

CONCLUSION

- The association of MRKH and gonadal dysgenesis is rare but important to consider in patients with a hypoplastic uterus and gonadal dysgenesis presenting with secondary amenorrhoea to avoid delayed diagnosis and treatment.
- Incorporating peripheral oestrogenisation, a hormone profile and karyotyping can facilitate diagnosis.