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Probable Ovarian Agenesis with Hypoplastic Prepubertal Uterus: A Case Report

O. Tounsi[1], Y. Bouktib[1], B. Boutakioute[1], M. Oualiel Idrissi[1], N. Guanouni Idrissi Cherif[1]

[1]Department of Radiology Arrazi Hospital, Mohammed VI University Hospital Center, Marrakech Morocco

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***Corresponding author:** O. Tounsi
Department of Radiology Arrazi Hospital, Mohammed VI University Hospital Center, Marrakech Morocco

Abstract | **Case Report**

We report the case of a 43-year-old woman, being treated for hypothyroidism, nulligravid, presenting in the perimenopausal period with secondary amenorrhea and chronic pelvic pain. Pelvic MRI revealed a right-lateralized hypoplastic uterus with a prepubertal morphology and no identifiable ovaries. Associated genitourinary anomalies were noted, including an ectopic left kidney. This presentation suggests a previously undiagnosed disorder of sexual development (DSD). This case highlights the importance of a comprehensive etiological workup for pelvic anomalies in adult patients.

Keywords: Ovarian Agenesis, Prepubertal Uterus, MRI Findings, Amenorrhea.

INTRODUCTION

Disorders of sexual development (DSD) in females comprise a heterogeneous group of congenital anomalies affecting structures derived from the Müllerian ducts and/or gonads. These conditions are usually diagnosed during adolescence when patients present with primary amenorrhea or pubertal disorders.

Ovarian agenesis is a rare condition, often confused with gonadal dysgenesis. When it occurs in association with a hypoplastic or prepubertal uterus, it suggests a significant developmental abnormality requiring multidisciplinary evaluation [1, 2].

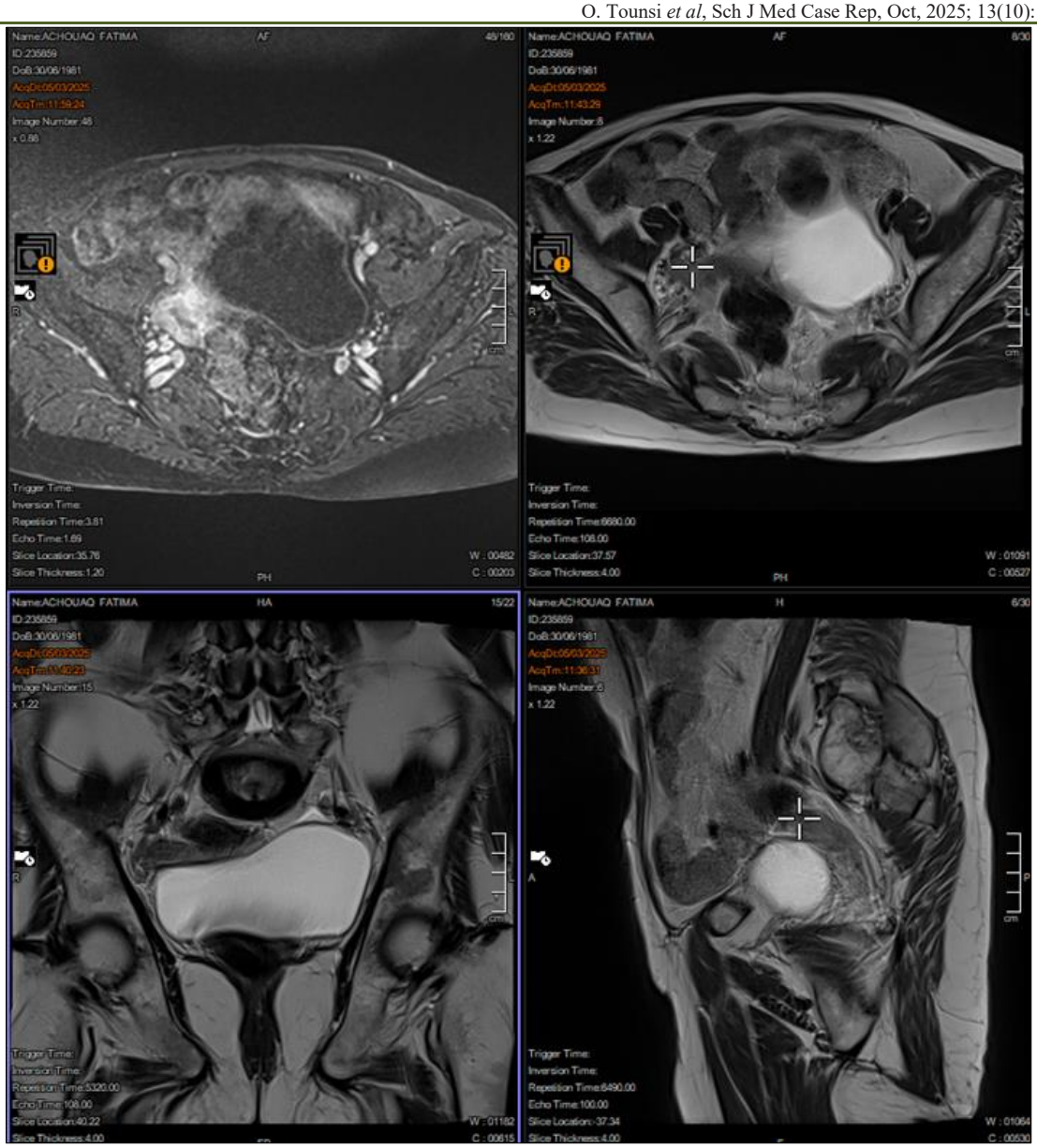
CASE REPORT

A 43-year-old woman, followed for well-controlled hypothyroidism on replacement therapy, nulligravid, presented with secondary amenorrhea and chronic pelvic pain.

Clinical examination revealed a normal female body habitus without signs of virilization. Breast development was Tanner stage V, and secondary sexual characteristics were unremarkable.

Pelvic MRI Findings:

- **Uterus**: Hypoplastic, right-lateralized, measuring approximately 50x30 mm, with a prepubertal morphology (cervix-to-body ratio >1).
- **Endometrium**: A fine endometrial stripe was visible.
- **Vagina**: A small hemivagina was present.
- **Ovaries**: Not visualized despite extended exploration.
- **Kidneys**: Ectopic left kidney; right kidney in normal position. No dilation of urinary tracts.
- **Bladder**: Normal morphology.
- **Other**: Small amount of free fluid in the pouch of Douglas.



Images 1-4: axial T1 and T2 weighted sequences with and without Fat saturation

DISCUSSION

1. Diagnosis Hypothesis

Several conditions must be considered in this atypical presentation:

a. Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome

Characterized by uterovaginal agenesis or hypoplasia with normal functioning ovaries and a 46, XX karyotype. This diagnosis is less likely here due to the presence of a small but visible uterus and partial vagina [1-4].

b. Gonadal Dysgenesis (e.g., Turner Syndrome or Mosaic Forms)

Typically associated with 45, X or mosaic karyotypes (45, X/46, XX). The uterus is usually hypoplastic and the ovaries are streak gonads, leading to absent or delayed puberty. A karyotype is essential here to rule out mosaic Turner syndrome, especially since the ovaries were not visualized [2].

c. Complete Androgen Insensitivity Syndrome (CAIS)

Individuals have a 46, XY karyotype with a complete female phenotype. There is absence of uterus and fallopian tubes (due to Müllerian duct inhibition), and undescended testes. This is unlikely here due to the presence of a uterus and absence of testicular structures on imaging [3].

d. Isolated Ovarian Agenesis or Hypoplasia

Rare, sometimes associated with uterine anomalies. May be of genetic origin (e.g., FSHR, LHX8, NR5A1 mutations) or secondary to perinatal ischemic events [1, 2].

2. Hypothyroidism and Sexual Development

Although hypothyroidism can lead to menstrual disorders and amenorrhea, it is not directly responsible for congenital uterovaginal or gonadal anomalies. In this case, the disease is well-managed, making an endocrine etiology unlikely [3].

3. Associated Anomalies

The ectopic left kidney is noteworthy, as Müllerian anomalies are frequently associated with renal malformations in 30–40% of cases [1]. This supports the diagnosis of a congenital developmental syndrome.

Additional Investigations Required

- **Karyotyping**: To identify chromosomal DSD such as Turner syndrome or mosaic variants.
- **Comprehensive hormonal panel**:
  - FSH, LH, estradiol: to evaluate gonadotropic axis.
  - Testosterone, AMH: to assess ovarian function or presence of testicular tissue.
- **Extended pelvic ultrasound or MRI**: To search for ectopic ovaries or dysgenetic gonads.
- **Renal function assessment**: Via urological MRI or renal scintigraphy if needed.

CONCLUSION

This case highlights a rare and likely previously undiagnosed disorder of sexual development discovered in adulthood. The association of a hypoplastic prepubertal uterus, probable ovarian agenesis, and a renal anomaly suggests an incomplete Müllerian differentiation syndrome or partial gonadal dysgenesis.

Cytogenetic and hormonal evaluations are crucial for establishing a diagnosis and planning appropriate multidisciplinary care. This includes long-term surveillance for gonadal remnants (due to tumor risk), hormonal therapy if needed, and psychosocial support.

REFERENCES

1. Morcel, K., Camborieux, L., & Guerrier, D. (2007). Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome. *Orphanet Journal of Rare Diseases*, 2(1), 13.
2. Grynberg, M., et al. (2010). Gonadal dysgenesis: diagnosis and management. *Gynécologie Obstétrique Fertilité*, 38(6), 387–392.
3. Sultan, C., et al. (2009). Disorders of sexual development: clinical, biological and genetic aspects. *EMC - Pédiatrie*.
4. Guerrier, D., et al. (2006). The Mayer-Rokitansky-Küster-Hauser syndrome—phenotypic manifestations and genetic approaches. *Journal of Negative Results in BioMedicine*, 5, 1.