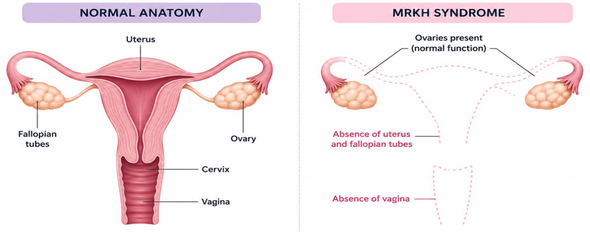
PRIMARY OVARIAN INSUFFICIENCY MASQUERADING AS MÜLLERIAN AGENESIS: A CASE REPORT

Heather Wang, DO[1], Nicolas Johnson, MD[2], Michael Thomas, MD[2]
Department of Obstetrics and Gynecology, Mercy St. Vincent Medical Center, Toledo, Ohio
Division of Reproductive Endocrinology and Infertility, University of Cincinnati, Cincinnati, Ohio

BACKGROUND

- Primary ovarian insufficiency (POI) is characterized by ovarian dysfunction before age 40 and may present with infertility, hypoestrogenism, and amenorrhea.[1]
- Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is classically defined by the congenital absence of the uterus and upper vagina with normal ovarian function and secondary sexual characteristics.[2]



- Distinguishing these diagnoses is critical due to differing management, fertility implications, and long-term health outcomes.

OBJECTIVE

- To describe a case of POI initially misdiagnosed as Müllerian agenesis.
- To emphasize the importance of diagnostic reassessment in patients with inconsistent clinical findings.

CASE PRESENTATION

Clinical History

- 36 y.o. GoP0 with primary amenorrhea and osteopenia
- She was previously diagnosed with MRKH syndrome after imaging suggested uterine agenesis
- Other notable findings included normal pubic hair development and minimal breast development
- Prior CT confirmed remnant ovaries and uterine agenesis with otherwise normal anatomy
- She was initiated on transdermal estradiol at age 28 for presumed diagnosis of MRKH syndrome
- She presented to REI clinic after episodes of vaginal bleeding

Diagnostic Reassessment

EVALUATION	FINDINGS
Repeat pelvic US	Uterus identified (5.7 x 2.2 x 3.6 cm) Endometrial thickness: 2 mm Ovaries unremarkable
Hormonal evaluation	Estradiol <5 pg/mL FSH 10.9 mIU/mL AMH 0.075 ng/mL
Karyotype	46, XX
Autoimmune antibody screen	Negative

Table 1: Repeat pelvic US demonstrated a uterus, which led to reconsideration of MRKH syndrome. Subsequent laboratory evaluation was consistent with POI.

Management

- Increased transdermal estradiol dosage
- Added oral progesterone for endometrial protection
- Planned longitudinal follow-up

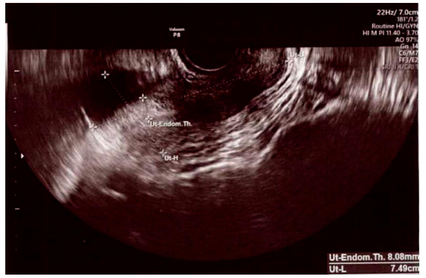


Figure 1: Repeat pelvic ultrasound identified a uterus with a measurable endometrial stripe, findings inconsistent with the prior diagnosis of Müllerian agenesis.

CONCLUSION

- POI may be misdiagnosed as Müllerian agenesis when initial evaluation is limited.
- Incomplete pubertal development should prompt consideration of endocrine dysfunction.
- Repeat evaluation should be considered in patients with atypical or evolving presentations.
- Prompt recognition of POI is essential for appropriate initiation of hormone replacement, bone health preservation measures, and individualized reproductive counseling.

REFERENCES

1. Nelson LM. Primary ovarian insufficiency. *N Engl J Med.* 2009;360(6):606-614.
2. Morcel K, Camborieux L, Guerrier D. Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome. *Orphanet J Rare Dis.*2007;2:13.