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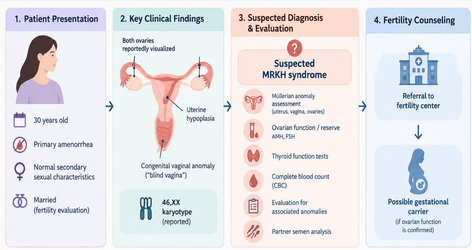
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Primary Amenorrhea Associated with Congenital Vaginal Anomaly and Uterine Hypoplasia: A Case Report of Suspected Mayer–Rokitansky–Küster–Hauser Syndrome

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Backgrounds

Mayer–Rokitansky–Küster–Hauser (MRKH) syndrome is a congenital disorder of the female reproductive tract resulting from abnormal development of the Müllerian ducts. It is characterized by congenital agenesis or hypoplasia of the uterus and upper vagina in individuals with a normal 46,XX karyotype and generally preserved ovarian function. The estimated prevalence of MRKH syndrome is approximately 1 in 4,000–5,000 live female births, making it one of the most common congenital causes of primary amenorrhea in individuals with a normal karyotype. This case report describes the clinical features, diagnostic evaluation, and fertility counseling of a woman with suspected MRKH syndrome.

Methods

A 30-year-old married woman with primary amenorrhea presented for fertility evaluation and counseling. She reported normal secondary sexual characteristics. Previous evaluations reportedly identified a congenital vaginal anomaly described as a “blind vagina,” uterine hypoplasia, visualization of both ovaries, and a normal 46,XX karyotype.

Findings

The patient had previously undergone evaluation for failure to initiate menstruation, as mentioned.

Given the presence of primary amenorrhea, normal secondary sexual characteristics, a reportedly normal 46,XX karyotype, uterine hypoplasia, and a congenital vaginal anomaly, MRKH syndrome was clinically suspected.

To complete the diagnostic evaluation and assess reproductive potential, tests of ovarian function and reserve, including anti-Müllerian hormone (AMH) and follicle-stimulating hormone (FSH), as well as thyroid function tests and a complete blood count (CBC), were requested. Given the patient's desire to pursue pregnancy, evaluation of the male factor through semen analysis of her husband was also recommended.

Summary of the patient's clinical findings and diagnostic evaluations

Variable	Finding
Patient age	30 years
Marital status	Married
Reason for presentation	Primary amenorrhea and fertility counseling
Menstrual history	No menstrual bleeding since puberty
Secondary sexual characteristics	Normal
Breast development	Normal according to the patient's history
Pubic hair growth	Normal according to the patient's history
Vaginal status	Congenital vaginal anomaly described as a “blind vagina,” according to the patient
Uterine status	Uterine hypoplasia, according to the patient's report of previous evaluations
Ovarian status	Both ovaries reportedly visualized on previous imaging
Karyotype	Normal (46,XX), according to the patient
Laboratory evaluation	AMH, FSH, thyroid function tests, and CBC were requested
Male-factor evaluation	Semen analysis was recommended
Previous imaging	Written reports of previous imaging studies were unavailable
Fertility counseling	Counseling regarding the potential use of a gestational carrier, if appropriate
Follow-up	Referred to a fertility center for further evaluation and treatment
Follow-up results	Not available

Conclusion

Comprehensive evaluation of women presenting with primary amenorrhea and normal secondary sexual characteristics is essential for identifying congenital reproductive tract anomalies. In patients with suspected MRKH syndrome, accurate assessment of reproductive anatomy, ovarian function, and fertility potential is important for appropriate counseling and individualized management. When previous diagnostic records are unavailable, further imaging and complementary investigations may be required to confirm the diagnosis.

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