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Effectiveness of Treatment in Patients with Mayer–Rokitansky–Küster–Hauser Syndrome Combined with Ovarian-Origin Hyperandrogenism

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Abstract Mayer–Rokitansky–Küster–Hauser (MRKH) syndrome is a rare congenital anomaly characterized by uterine and vaginal aplasia in women with a normal 46,XX karyotype. When combined with ovarian-origin hyperandrogenism, it presents additional clinical complexity. This study assessed the clinical and hormonal effectiveness of combined medical and surgical treatments in 66 patients with MRKH syndrome, 30 of whom had signs of hyperandrogenism. The primary treatment included weight reduction, biguanides, and combined oral contraceptives, with some undergoing sigmoid colpopoiesis and ovarian drilling. Results showed significant improvements in clinical symptoms (e.g., acne, hirsutism, unstable blood pressure) and endocrine profiles (e.g., testosterone and prolactin reduction). Ultrasound data revealed decreased ovarian volume and follicle count post-surgery. The Dermatology Life Quality Index (DLQI) improved from 30 to 8 (p<0.05), and overall treatment effectiveness reached 89%. The integration of ovarian drilling with sigmoid colpopoiesis is especially recommended in cases resistant to conservative therapy.

Keywords MRKH syndrome, Vaginal aplasia, Hyperandrogenism, Ovarian drilling, Sigmoid colpopoiesis, Quality of life

1. Introduction

Uterine and vaginal aplasia is a rare congenital anomaly of the reproductive organs characterized by the absence of the uterus and the upper two-thirds of the vagina in women with a normal karyotype (46, XX) [1,4]. This pathology accounts for 5–10% of all genital anomalies [1,4,8]. It is caused by a disruption in the embryonic development of the Müllerian ducts, resulting in the absence of these structures while ovarian function is preserved. The frequency of genitourinary anomalies, including uterine and vaginal aplasia, is increasing and ranks fourth among all congenital malformations, comprising 9.7%, of which 4.0% are related to female reproductive system malformations [1,2].

Mayer–Rokitansky–Küster–Hauser syndrome (MRKH) is one of the most well-known forms of uterine and vaginal aplasia. It is characterized by a normal female phenotype, absence of the uterus and vagina (or upper two-thirds of the vagina), and fully functional ovaries [6,7]. The syndrome is named after the scientists who described it: August Mayer, Carl von Rokitansky, Hermann Küster, and Georges Hauser [1,4,8,10].

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Patients with MRKH experience challenges such as primary amenorrhea, infertility, and significant psychological distress. Although surgical correction of anatomical defects is well-established, data on the combination of MRKH with endocrine and metabolic disorders, particularly ovarian-origin hyperandrogenism, remain limited [2,4,5,6,7,8,11,12].

Ovarian-origin hyperandrogenism manifests as excessive androgen production, leading to acne, hirsutism, and menstrual disorders. It is often associated with metabolic abnormalities, including metabolic syndrome, which affects 60–70% of hyperandrogenic patients and significantly worsens their overall health [2,4,5,8-12]. These factors necessitate an individualized approach to diagnosis and treatment.

Given the rarity of the combination of uterine and vaginal aplasia with ovarian-origin hyperandrogenism, further research is essential. Optimizing diagnostic and therapeutic strategies for these patients can improve their reproductive and general health, quality of life, and emotional well-being, which is of great clinical importance.

2. Materials and Methods

A total of 66 patients with vaginal and uterine aplasia were examined and divided into two groups:

- **Main group** (n = 30): vaginal and uterine aplasia combined with signs of ovarian-origin hyperandrogenism.
- **Comparison group** (n = 36): vaginal and uterine aplasia without hyperandrogenism.

Patients in the main group received medical therapy (weight loss, biguanides, and combined oral contraceptives). Some patients additionally underwent surgical treatment (sigmoid colpopoiesis + ovarian drilling). The effectiveness of therapy was assessed dynamically based on clinical and laboratory changes.

Table 1. Main complaints of examined patients during treatment

Clinical symptoms	Before treatment	After treatment	p
Amenorrhea	30 (100%)	30 (100%)	>0.05
Irritability	8 (26.7%)	2 (6.7%)	>0.05
Sleep disturbances	11 (36.7%)	2 (6.7%)	>0.05
Fatigue	9 (30%)	2 (6.7%)	>0.05
Decreased libido	2 (6.7%)	2 (5.6%)	>0.05
Unstable blood pressure	11 (36.7%)	3 (10%)	<0.05
Edema	10 (33.3%)	—	<0.05
Sweating	9 (30%)	2 (6.7%)	>0.05
Mastodynia	11 (36.7%)	1 (3.3%)	>0.05
Hirsutism	26 (86.7%)	6 (20%)	<0.001
Acne	27 (90%)	3 (10%)	0.05
Inability to lose weight	10 (33.3%)	2 (6.7%)	<0.01
Headache	17 (56.7%)	1 (3.3%)	<0.05

3. Results and Discussion

Hormonal Profile.

Table 2. Hormonal profile of patients 6 months after treatment

Indicator	Reference range	Before treatment	After treatment	p
LH, IU/L	2–14	16.4±1.3	11.3±1.4	>0.05
FSH, IU/L	3.5–13.0	4.7±0.6	5.3±0.8	>0.05
Insulin, μIU/mL	3–20	9.7±1.4	9.2±1.4	>0.05
Prolactin, mIU/L	40–530	479.6±22.1	382.5±19.4	<0.001
DHEA-S, μmol/L	2.45–14.55	15.22±1.7	13.81±1.5	>0.05
Testosterone, nmol/L	0.45–3.75	4.5±0.4	2.81±0.6	<0.05
AMH, ng/mL	0.5–7.0	3.7±0.9	3.8±0.4	>0.05

Following treatment, LH levels normalized in several patients. The LH/FSH ratio averaged 2.1±0.02 in the main group and 2.3±0.02 in the comparison group ($p > 0.05$). Testosterone decreased from 4.2±0.5 to 3.2±0.7 nmol/L; DHEA-S remained within the age norm. In 7 patients (23.3 ±4.7%), free testosterone remained slightly above normal. Differences in other hormones, except prolactin, were not significant. A higher LH/FSH ratio (3.4±0.2 vs 2.7±0.1; $p<0.01$) suggests deeper hypothalamic-pituitary disturbances in metabolic syndrome.

Ultrasound Evaluation.

Table 3. Ultrasound parameters in treatment dynamics

Parameter (M±SD)	Before surgery	After surgery	p (Student's t-test)
Ovarian volume (cm^3)	17.7 ± 6.2	11.2 ± 2.2	0.95
Number of follicles (n)	16 ± 4	9 ± 4.2	0.07
Capsule thickness (mm)	1.0 ± 0.4	0.6 ± 0.4	0.70
Stroma thickness (mm)	11.1 ± 2.7	0.81 ± 2.2	0.84
Stroma volume (cm^3)	6.6 ± 0.7	1.8 ± 0.8	—

A month after surgery (sigmoid colpopoiesis + ovarian drilling), ovarian volume and follicle number decreased ($p< 0.05$). Capsule and stroma thickness showed no significant changes, though Doppler studies indicated improved blood flow.

The Dermatology Life Quality Index (DLQI) was used to assess treatment effectiveness for hyperandrogenism and ovarian function. DLQI scores decreased from 30 to 8 ($p<0.05$), and the effectiveness of comprehensive therapy was 89%.

4. Conclusions

In patients with vaginal and uterine aplasia combined with ovarian-origin hyperandrogenism, comprehensive therapy (weight reduction, biguanides, COCs, etc.) combined with surgery (sigmoid colpopoiesis and ovarian drilling) demonstrated positive clinical and laboratory outcomes and improved quality of life.

Ovarian drilling during sigmoid colpopoiesis is advisable in severe hyperandrogenism resistant to medical therapy and with a high software score (≥12).

Drilling promotes ovulation, corrects hyperandrogenism, and may serve as a preparatory stage for assisted reproductive technologies, considering individual characteristics and risks (adhesions, diminished ovarian reserve).

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