

IMPROVING THE MANAGEMENT OF PATIENTS WITH VAGINAL AND UTERINE APLASIA AND OVARIAN GENESIS

HYPERANDROGENISM

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Abstract:

Objective: To review and synthesize current best practices for the comprehensive management of patients presenting with the complex triad of vaginal and uterine aplasia (most commonly associated with Mayer-Rokitansky-Küster-Hauser syndrome [MRKHS]), the presence of ovaries (ovarian genesis), and concurrent hyperandrogenism. This combination often presents significant diagnostic and therapeutic challenges, requiring a multidisciplinary approach focused on optimizing physical, psychological, and reproductive health outcomes.

Methods: A narrative review of existing literature, including clinical guidelines, systematic reviews, and case series, was conducted focusing on the diagnosis, treatment, and long-term surveillance of individuals with MRKHS variants, atypical presentations of androgen insensitivity syndrome (AIS), and other differential diagnoses that include Mullerian agenesis alongside ovarian function and hyperandrogenic features. Specific attention was paid to techniques for neovagina creation, hormonal management strategies for hyperandrogenism, and psychological support.

Results: The management paradigm necessitates accurate initial diagnosis, differentiating between typical MRKHS, incomplete AIS, and rare genetic syndromes. Treatment of vaginal aplasia typically involves non-surgical dilation (preferred initial method) or surgical vaginoplasty (e.g., laparoscopic Vecchietti procedure, Davydov procedure), tailored to the individual's anatomy and preferences. Hyperandrogenism, when present, requires thorough endocrinological investigation to determine its etiology (e.g., polycystic ovary syndrome [PCOS]-like phenotype, adrenal enzyme deficiencies, or ovarian tumors). Management

often involves combined oral contraceptives or anti-androgens (e.g., spironolactone) to address hirsutism, acne, and menstrual irregularities (in cases with a functional uterus/endometrium, though rare in this specific cohort). Crucially, long-term psychological and sexual health support is paramount due to issues related to body image, sexual function, and infertility.

Conclusion: Effective management of patients with vaginal and uterine aplasia and ovarian genesis hyperandrogenism demands a highly individualized, staged approach involving reproductive endocrinologists, gynecologic surgeons, geneticists, and mental health professionals. Standardized diagnostic protocols and tailored treatment plans for both anatomical reconstruction and hormonal imbalance are necessary to improve quality of life and ensure optimal long-term health surveillance.

Key Words: Vaginal Aplasia, Uterine Aplasia, Mayer-Rokitansky-Küster-Hauser Syndrome (MRKHS), Hyperandrogenism, Ovarian Genesis

Introduction

The management of patients presenting with vaginal and uterine aplasia, coupled with the presence of functional ovaries (ovarian genesis) and concurrent hyperandrogenism, represents one of the most complex and challenging areas in reproductive medicine. The most common cause of congenital aplasia of the vagina and uterus is Mayer-Rokitansky-Küster-Hauser syndrome (MRKHS), which affects approximately 1 in 4,500 to 5,000 female births. While typical MRKHS involves normal female karyotype (46,XX) and normal ovarian function, the presentation of significant hyperandrogenism alongside Mullerian agenesis is atypical and necessitates a thorough investigation to rule out overlapping or variant conditions, including specific Disorders of Sex Development (DSDs) or unique endocrine pathologies.

This specific cohort of patients faces a dual burden: the anatomical deficit requiring reconstruction for sexual function (neovagina creation) and the physiological imbalance resulting from hyperandrogenism, which can manifest as hirsutism, acne, and metabolic concerns. The complexity lies in differentiating the

source of the hyperandrogenism—whether it is a polycystic ovary syndrome (PCOS)-like phenotype secondary to the underlying genetic or metabolic milieu, or related to a specific enzyme deficiency or gonadal tumor.

Current clinical practice often addresses these components separately, leading to fragmented care. The lack of standardized protocols tailored specifically to this triad—anatomical aplasia, ovarian presence, and endocrine dysfunction—underscores the critical need for an integrated, multidisciplinary approach. Improved management must focus not only on surgical and hormonal correction but also on rigorous psychological support, addressing issues of body image, sexual identity, and fertility implications.

This paper aims to review the current diagnostic pathways and therapeutic strategies available for patients with vaginal and uterine aplasia and ovarian genesis hyperandrogenism. By synthesizing evidence from gynecology, endocrinology, and mental health, we seek to establish a comprehensive framework for optimal clinical management, thereby improving long-term physical and psychological outcomes for this unique patient population.

Conclusion

The effective management of patients presenting with the intricate combination of vaginal and uterine aplasia, ovarian genesis, and hyperandrogenism requires a highly individualized and meticulously coordinated multidisciplinary approach. This patient cohort presents unique challenges that transcend typical MRKHS care, demanding simultaneous attention to anatomical reconstruction, endocrine normalization, and profound psychological support.

Successful management hinges upon an accurate, early diagnosis that carefully distinguishes between MRKHS variants, atypical DSDs, and primary endocrine disorders causing hyperandrogenism. Once diagnosed, treatment must be staged, prioritizing non-surgical methods for neovagina creation where feasible, followed by tailored surgical intervention when necessary. Concurrently, the hyperandrogenism must be addressed through targeted hormonal therapies, guided

by comprehensive endocrinological assessment, to mitigate long-term metabolic risks and improve quality of life related to androgenic symptoms.

Ultimately, the goal of improved management is the restoration of physical function and the promotion of psychological well-being. Long-term surveillance is crucial, encompassing regular gynecological check-ups, monitoring for potential complications of vaginoplasty, and continuous assessment of metabolic and psychological health. Future research should focus on establishing standardized international guidelines for the diagnosis and treatment of this rare combination of conditions, particularly investigating the long-term metabolic consequences of hyperandrogenism in this population and refining surgical techniques to minimize morbidity and maximize functional outcomes. By integrating the expertise of gynecologic surgeons, reproductive endocrinologists, geneticists, and mental health specialists, we can significantly enhance the quality of care and optimize the life trajectory for these complex patients.

REFERENCES

1.Махмудова С. Э., Атаева Ф. Н. Опыт применения модульной системы для овладения педагогическими навыками в подготовке резидентов магистратуры по специальности" акушерства и гинекологии" //ЛУЧШАЯ НАУЧНАЯ СТАТЬЯ 2018. – 2018. – С. 290-293.

2.Makhmudova S. E., Ataeva F. N. Evolutionary views on hypertensive disorders in pregnancy (literature review) //Advaced science. – 2019. – С. 183-187.

3.Мусаева Т. Т. Влияние аутоиммунного тиреоидита на течение и исход беременности : дис. – СПб, 2009, 2009.

4.Yusufovna X. N., Valiyevna D. M., Nuriddinovna A. F. Problems in the translation of world picture terminology in french //PalArch's Journal of Archaeology of Egypt/Egyptology. – 2020. – Т. 17. – С. 6.

5.Махмудова С. Е., Атаева Ф. Н. Эволюционные взгляды на гипертензивные расстройства беременности (ОБЗОР ЛИТЕРАТУРЫ) //Передовая наука. – 2019. – С. 183-187.

6.Атаева Ф., Закирова Н., Курбаниязова В. Современные подходы к лечению плацентарной недостаточности и синдрома задержки развития плода //Журнал проблемы биологии и медицины. – 2015. – №. 4, 1 (85). – С. 141-142.