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Beyond Anatomy: I Don't Have a Uterus, What Now? Case Report and Literature Review

Maria Clara Machado Briefs [α], Sheila R. Niskier [σ], Christiane de Morais Junqueira Camargo [ρ], Márcia Nunes Gaspar [Ω], Bárbara Soares da Silva [¥] & Maria Sylvia de Sousa Vitalle [§]

Summary- Rokitansky - Küster-Hauser syndrome (MRKH) is characterized by agenesis or aplasia of the uterus and upper part of the vagina and is considered the second most common cause of primary amenorrhea. The psychological repercussions are significant and are directly related to sexual and reproductive health. This work reports the case of a teenager with MRKH and reviews the syndrome and the main factors that impact mental health.

Case Description: Adolescent 14 years old, female, absence of menarche, sexual life not initiated, Tanner M5P5. Pelvic ultrasound with hypoplastic uterus, magnetic resonance imaging of the pelvis with non-individualized uterus, unidentified vagina and ovaries with normal topography and morphology. Diagnosed with MRKH and has no desire to perform vaginal dilation. He has difficulty talking about the syndrome and aspects of his sexual life. She appeared sad and felt inadequate, especially after her father told her she was "less of a woman."

Discussion: MRKH is the second most common cause of primary amenorrhea and its psychosocial impact is relevant. Historically, the uterus is described as primordial in female identity and menarche is an important milestone in puberty in girls. People with the syndrome tend to present more anxious and depressive symptoms, image disturbances, influencing self-esteem and identity construction.

Conclusion: The diagnosis of MRKH involves several factors that go beyond anatomical changes. Therefore, it is essential to include emotional aspects in the clinical approach, aiming to improve sexual, reproductive and mental health and, above all, the quality of life of these people.

Keywords: adolescent; amenorrhea; teenager behavior; infertility female.

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I. Introduction

Rokitansky - Küster-Hauser syndrome (MRKH) is a congenital syndrome characterized by malformation of the Müllerian system , resulting in agenesis or aplasia of the uterus and upper part of the vagina. The ovaries are functional and the development of secondary sexual characteristics occurs physiologically, so hormonal levels are generally normal. [(1)] The main symptom is primary amenorrhea and the diagnosis is normally made in adolescence, a period of complex physical and psychological transformations. [(2)] Therefore, the psychosocial repercussion is quite relevant and involves delicate aspects in a woman's life such as self-affirmation, sexual and reproductive health, impacting the formation of identity and interpersonal relationships. [(3)] Despite the well-established influence on mental health, especially among adolescents, much of the medical care is focused on physical changes and little directed towards emotional and psychological support. [(2)] Thus, this work describes the case of a teenager with MRKH, covering the etiopathogenesis , diagnosis and therapy, as well as the psychological aspects that involve the absence of the uterus and, consequently, menstruation for people with the syndrome. The work was approved by the Research Ethics Council number 3524696.

II. Case Report

A 14-year-old female adolescent, with no pathological history, reported the absence of menarche despite the complete development of secondary characteristics. The mother reported thelarche between the ages of eight and nine, pubarche at nine and growth spurt at eleven. The patient denies beginning sexual life. The physical evaluation presents Tanner staging M5P5 , external morphology of the genitalia without abnormalities, annular hymen and indirect vaginometry less than 2cm. Hormonal tests within the references for age and sex. Pelvic ultrasound (USG) with hypoplastic uterus of 1cm^3 and ovaries of compatible size for age. Magnetic Resonance Imaging (MRI) of the pelvis showed a non-individualized uterus, unidentified vagina, ovaries with normal topography and morphology. Karyotype 46,XX . To screen for extra-genital

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malformations , USG of the kidneys and urinary tract, echocardiogram transthoracic and cervical - thoraco - lumbar spine radiography without abnormalities.

In view of the clinical characteristics, combined with laboratory and imaging tests, the diagnosis of MRKH syndrome type 1 was made. The teenager had difficulty discussing the syndrome and its repercussions, so the mother took the lead in the consultation. She appeared sad and felt inadequate, especially after her father said that she was "less of a woman" due to her anatomical changes. There is no desire to perform vaginal dilation or any therapeutic intervention. Psychological support was offered, but she was unreceptive and preferred not to talk about topics involving her emotions and sexuality.

III. Discussion

Congenital uterine anomalies manifest themselves in different ways, both in the development and shape of the uterus, commonly involving adjacent organs such as the cervix and vagina. Among these malformations, Müllerian agenesis (or MRKH) stands out. Next, the biological aspects of the syndrome, etiopathogenesis , clinical manifestations, diagnosis and therapeutic options will be presented. Subsequently, the psychological impact will be addressed, including the meaning of the absence of a uterus for these women, as well as the consequences on menstruation and fertility, important factors in the construction of identity and interpersonal relationships.

a) Epidemiology and Etiopathogenesis

MRKH syndrome, also known as Müllerian agenesis , is a rare disease, with an estimated prevalence of 1 in 5,000 [4] and occurs due to embryological changes in the differentiation and migration of the mesoderm, the tissue responsible for the formation of the reproductive, urinary, and skeletal and cardiac (systems most affected by the syndrome).[5] The etiology remains uncertain, possibly multifactorial, with genetic, chemical, mechanical and environmental components. Some chromosomal regions described and possible genes involved are 1q21.1 (RBM8A gene), 1p31-1p35 (WNT4 gene), 7p15.3 (HOXA gene), 16p11 (TBX6 gene), 17q12 (LHX1 and HNF1B genes), 22q11.21 and Xp 22. [6] Despite the increasing advancement of genetic research, there are still barriers to this investigation given the low availability of tests and their high cost, factors that prevented such an investigation in the case described.

b) Classification

The syndrome is classified as type 1 when only the reproductive system is affected and type 2 is accompanied by other extragenital malformations, generally renal, skeletal, cardiac, ocular, auditory and inguinal hernias. [1,8] The case described was classified as type 1 and no extragenital malformation was detected.

c) Clinical Presentation and Diagnosis

The karyotype is 46,XX and there are no abnormalities in hormonal levels or development in childhood. The syndrome is considered the second most common cause of primary amenorrhea and the diagnosis usually occurs in adolescence, during investigation of the absence of menarche. Other common symptoms are dyspareunia and cyclical abdominal pain. [3] On physical examination, the physiological development of secondary sexual characteristics is observed and the vagina is typically blind, generally measuring 0 to 3 cm. [1] In the case reported, the teenager had completed pubertal development for more than 2 years, without menarche and with a short vagina.transabdominal or transperineal USG shows absence of the upper part of the vagina and uterus. [10] MRI provides more image details, with the typical finding being uterine agenesis, although it is possible to find remaining Müllerian ducts. [10] The case presented showed a non-individualized uterus and topical ovaries, better visualized on MRI.

d) Therapeutic Options

Since 2002, the American College of Gynecology and Obstetrics has recommended the use of vaginal dilators as the first line of treatment, however the technique's success rates depend directly on the patient's disposition and motivation. Surgical treatment with vaginoplasty is reserved for patients who have not responded to conservative treatment. [1.11]

Although most adolescents do not have a reproductive desire, it is important to provide guidance and clarification on current techniques and monitoring with a specialized team is recommended. As the ovary is functional, "*in vitro*" fertilization and a surrogate uterus, popularly known as "surrogacy", can be performed, although the practice is prohibited in several countries. [1]In Brazil, according to the resolution of the Federal Council of Medicine (CFM) number 2294/21, the temporary transfer of the uterus can be carried out as long as the surrogate mother belongs to the family of one of the partners in a blood relationship up to the fourth degree and has at least one living child. [12]

Another promising option for treating infertility is uterine transplantation. There are already reports of successful cases, but it is still an experimental procedure. [1]

e) Psychosocial Impact

The psychological repercussions of the syndrome are relevant and directly impact intimate aspects of women, such as sexual activity and fertility. Furthermore, most diagnoses are made during adolescence, a period of great physical, psychological, social and affective transformations.

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A 2021 systematic review, focusing on the psychological impact, showed that the majority of patients had negative emotions upon diagnosis, including suicidal thoughts. They also had a higher incidence of anxious and depressive symptoms, low self-esteem and image disorders, as well as feelings of fear, guilt, denial and inadequacy. [2,13,14] It is estimated that two thirds of patients experience anxiety and, in approximately one quarter of cases, the manifestations are moderate to severe. [15]

Most affected women report feeling "incomplete", "different", "sexually inadequate" or "defective ". [16,17] These feelings show the power of the uterus and menstruation as symbols of the social role of women and how, today, they still influence society, although there may be variations according to the historical moment, religion and culture. [18]

I didn't menstruate . Is there something wrong with me? – Meaning of menarche throughout history

Menarche is considered an important milestone in a woman's life and has received different connotations throughout history and different cultures. It is probably the most important physical change in girls' puberty and involves not only biological transformation, but all the emotional and social adjustment linked to it. Etymologically, it originates from the Greek – "*men"* referring to "moon" and "month" and – "*arkhe"* meaning beginning, representing a rite of initiation into female adult life [19] , also described as the first "mystery feminine". [20]

In the era of matriarchy, birth, menstruation, sexuality, menopause and death were considered the milestones of a woman's life. Menstrual blood was linked to the sacred and was used in mystical rituals, to fertilize the earth and as a representation of feminine strength, associated with the idea of life, death and cycles of nature. More contemporary theories, such as the one proposed by Langer [21] , reinforced the idea of menstrual bleeding as an indicator of female normality, acceptance of one's own sex and sexuality, maintaining a relationship with fertility, capacity for regeneration, strength and power. [19]

However, from the 19th century onwards, the association of menstruation with negative ideas grew, largely influenced by the spread and domination of patriarchy in current cultures. As a result, ideas of fear of blood and female inferiority were disseminated because they were unable to "control their bodies" by bleeding every month, in addition to emphasizing the harm involved in the menstrual period and before it. [19]

Over time, the symbology and rites linked to menarche lost importance and women were taught to deal with the menstrual period as a physiological and mechanical event. Regardless of the positive or negative connotation in relation to the menstruation process, it is known that it currently maintains great psycho-emotional and socio-cultural significance, influenced by several factors, such as the way of preparing for the period, expectations, age, emotional support and personality traits. [19,22]

Given this, the inability to menstruate also gains considerable significance for women at an individual and social level, especially in adolescence, triggering ambivalent feelings regarding this process. [19]

An English qualitative study brings reports from patients aged 18-22 years diagnosed with MRKH and assesses the social and emotional impact. One of the participants draws attention to the fact that she feels different and is unable to participate in dialogues with her peers on simple subjects, such as menstruation. Furthermore, the absence of menstruation was shown to be related to the loss of part of the identity with the female gender, portrayed in the speech of one of the girls that part of her feminine side "was gone" when she learned that she would never have a menstrual period "like all women". woman ". [14]

The fact that they feel "different" makes many choose not to share their diagnosis with other people and even lie about their menstruation during medical appointments because they feel embarrassed and afraid of being judged. [17] In addition to self-judgment, many of these people also suffer from social pressure from their environment, including friends, family and partnerships. [18] In the case described, in addition to the difficulty in organizing her own feelings regarding the repercussions of the syndrome, the patient appeared extremely fragile after her father's comment that she would be "less of a woman" due to her anatomical changes.

f) Uterus and Reproductive Function

The relationship between the uterus and femininity dates back to antiquity and has also received different analytical, religious and cultural connotations over the centuries. [19] According to Colling [23], Plato introduced the idea of the uterus as a "matrix", which would correspond to a "rabid animal" that would live inside women with the desire to procreate. This theory was incorporated by several thinkers, assigning the organ a fundamental role in women's identity, as their main function would be procreation. Psychoanalysis was also influenced by this thought, as portrayed in one of Freud's theories in 1923 [24], in which motherhood would be the "normal destiny of femininity".

These ideas of biological determinism, which emphasize women's reproductive function as a fundamental role, began to be questioned at the end of the 19th century with feminist demonstrations, when women began to occupy spaces outside the domestic nucleus and claim sexual freedom. From this, associated with the development of contraceptive methods, women began to have the power to decide regarding pregnancy and the restructuring of family

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configurations began with women expanding their roles in society. [25,26] Furthermore, together with feminist movements, discussions are beginning to emerge about gender and the meaning of the feminine beyond biological aspects, as Simone de Beauvoir brings in her theories on the social construction of women, emphasizing the fact of "becoming herself as a woman", the central subject of his work "The second sex" published in 1949. [27]

Since then, women have been gaining more space in the job market and expanding their roles in society. Thus, there are those who dream of motherhood while others do not express this desire or postpone pregnancy due to various factors. Despite this, the symbolism of the uterus in femininity continues to be very relevant and the absence of this organ associated with the impossibility of gestation has an important impact on women in the most diverse cultures, gaining even greater relevance in more conservative societies. [25]

A review study on the psychological impact shows that the inability to conceive is one of the main stressors in the syndrome, having greater influence as age increases, while in adolescence ambivalent perceptions are observed. [16]

A 22-year-old participant in an English qualitative study, when she learned that she could not carry a pregnancy, reported feeling childish and not being "completely feminine". In the statement she says that one of the basic points of being a woman is being able to produce children and increase the population. So, the fact that she doesn't achieve this makes her feel like less of a woman. She also reports that she began to force herself to "be more feminine" by wearing more makeup and skirts to ensure that people knew she was a woman. Another patient in the same study, also 22 years old, reports that the absence of a uterus does not affect her because until now she has not needed this organ. A third patient, aged 18, reported not being as affected as she prefers to focus on other areas of her life and avoids thinking about her diagnosis. [14]

g) Repercussions on Interpersonal Relationships

The feeling of being "different" and "sexually inadequate" leads many patients to a place of isolation, accompanied by insecurity about themselves and questioning their own identity. [18] It is important to highlight that they often compare themselves negatively with other women, feeling "inferior". [28]

Several participants reported feeling less confident after the diagnosis and less worthy of intimate relationships, as their partner would be at a "disadvantage" with them in relation to other women. [14] Still others feel "less attractive" and accept violent or abusive relationships because they believe they are not worthy because they cannot conceive. [28]

To deal with such situations, some prefer not to get involved in intimate relationships because they feel embarrassed [14] or because of the constant fear of rejection. [17]

In countries like Malaysia, where a more conservative culture prevails, patients reported that they did not have any access to information about sexual intimacy from the medical team because it was considered "taboo" to talk about sexuality or that it should only be mentioned after marriage. As a result of this misinformation, many of them had traumatic experiences during sexual intercourse that could have been prevented if there was greater dialogue and guidance on these issues. [17]

Furthermore, a 2022 review analyzed 14 articles that evaluated the impact of the syndrome on sexual function, indicating a relationship with greater difficulty in maintaining lubrication, experiencing orgasm, in addition to experiencing pain during sexual intercourse. Furthermore, one of the studies highlighted that girls with MRKH tend to start relationships later and have lower sexual frequency. [29]

As the syndrome impacts women's intimate aspects, many teenagers and even adults find it difficult to speak openly about the topic in the face of prejudice, in addition to religious and social issues. Furthermore, a large part of medical care is focused on physical and objective aspects of the syndrome and patients complain that they have not been able to adequately understand the complexity of the syndrome's implications for sexual and reproductive life. [2,18]

In adolescent care, mothers or guardians often assume the central role in the approach and the patient herself does not actively participate in decisions. [2] This fact was mentioned by the teenager in a study when she complained that her autonomy was not recognized and that decisions were mediated between the health professional and her mother. [14] In the above case, the mother also assumed a central role in therapeutic decisions, while the teenager had difficulty talking about the syndrome and its repercussions, perhaps due to cognitive and emotional immaturity inherent to her age, not having assimilated the information adequately or preferring focus on other aspects of your life.

h) An Adolescence Perspective

Adolescence, according to the World Health Organization [30] , covers the period from 10 to 19 years and involves major biopsychosocial transformations. The set of signs and symptoms typical of this phase are called by some authors as "normal adolescence syndrome" and the individual goes through the loss of the child's body and the child's social roles to enter the search for themselves and adult identity. [31]

The construction of identity is determined by several factors, including family, cultural and social. The direct influence of the syndrome on sexual and

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reproductive life can lead to questioning one's identity and confusion regarding one's body, gender, social and sexual roles. [28] This variability in emotions was described by a study participant when she said that she felt sad one day, happy the next day, then depressed again and that this diagnosis took a lot away from her. [14]

In a didactic way, adolescence can be classified as early, middle and mature, based more on psychic and behavioral characteristics than chronological ones, although age also, admittedly, has its value, especially in defining periods. In the initial period, around 10 to 14 years old, biological changes and emotional fluctuations begin, attention turns to oneself, and self-reference is common. They begin to question their parents and authority figures, reducing interest in the family cycle and increasing the desire for independence. [32.33]

Most diagnoses occur in middle adolescence, between 14 and 17 years old, with greater influence from social groups, tendency to experiment, test limits, improve abstract thinking skills, reasoning and creativity. [33,34] When they discover the syndrome at this stage, the feeling of "not belonging" has a strong influence on self-esteem and identity construction, after all, not menstruating at the right time like other girls makes them feel more different at the moment. who seek precisely to resemble their peers. [28]

Age-related immaturity can manifest itself in difficulty in processing and dealing with so much new information in a short space of time and great variability in emotional responses. [28] One of the participants in a qualitative study mentioned that she was unable to understand the diagnosis and its implications, given the many physical and psychological changes she was already experiencing . He adds that, some time later, he began to have a different view and that if he had been more mature, he would have asked different questions and requested other information at the time of the diagnosis. [18]

Late or mature adolescence, commonly occurs from the age of 17-18, when there is progress in the consolidation of identity, the ability to rationalize for making decisions independently, establishing limits, planning and beginning to assume roles and responsibilities typical of adults. [32,33] With the development of hypothetical-deductive thinking, they begin to worry about long-term projects and women affected by MRKH may be more bothered by their inability to become pregnant. Furthermore, with emotional and cognitive maturity, sexual intercourse becomes more frequent and it is possible that they begin to show greater interest (or aversion) to gynecological interventions such as the use of vaginal dilators and the sexual act itself. [28]

The Medical Consultation. How to improve the approach?

Although the psychological repercussions of MRKH syndrome are well established, most services do not have the structure to provide adequate care for these patients, especially mental health ones. The moment of diagnosis is often marked by pain and confusion, with some authors even establishing a relationship with symptoms of post- traumatic stress. [28,35,36] socio-emotional aspects in the consultation when explaining anatomical anomalies and their repercussions, going beyond the objective and biological point of view. Furthermore, the importance of ensuring that the patient assumes her autonomy and is the protagonist of care is highlighted, especially during adolescence. [2] The consultation during this period of life takes on some particularities, such as moments together with the family member and with the adolescent alone, in order to ensure privacy, confidentiality and medical secrecy. [34] Individualization of care is a key factor in the approach and the professional must seek to understand how the individual feels and what resources they use to deal with the situation, as these are intimate matters that the person being assisted may not want to share with others, even if they are family or close people. [18.28]

Furthermore, a multidisciplinary approach is necessary, including individual psychology and/or support groups and mutual strengthening among these women, so that they feel more belonging and the long-awaited improvement in self-esteem and mental health in general occurs. [3.14]

IV. Conclusion

MRKH syndrome is the second cause of primary amenorrhea and directly impacts women's health, specifically their sexuality and reproductive health. Most diagnoses occur during adolescence, a period already marked by major biopsychosocial transformations in addition to emotional and cognitive immaturity, demanding greater attention to these factors. Therefore, it is essential that the clinical approach contemplates not only the physical aspects related to the malformation of the reproductive tract, but also encompasses emotional factors, individualizing care and making the adolescent occupy a protagonist role in her process. The establishment of a bond that allows dialogue and clarification about sexual health is an exceptional element in consultations, in addition to adequate guidance regarding therapeutic options and reproductive future, aiming to improve quality of life and mental health.

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