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ISOLATED MAYER-ROKITANSKY-KUSTER-HAUSER (MRKH) SYNDROME: A CASE REPORT AND REVIEW OF THE LITERATURE.

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Abstract:

Introduction: *Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is a congenital disorder of unknown etiology, characterized by mullerian agenesis and can be associated with renal, skeletal and cardiac malformation. Most cases are sporadic.*

Case Report: *We report a typical case of MRKH syndrome of a 16 year old female with normal renal system.*

Discussion: *The diagnostic workup of patient presenting with primary amenorrhea should include history taking, physical examination, hormonal evaluation, imaging and chromosomal analysis. Normal levels of LH (Leutinizing Hormone) and FSH (follicle stimulating hormone) indicate appropriate ovarian function. On the other hand, imaging techniques such as USG and MRI highlight the exact anatomical defects.*

Conclusion: *Absence of normal menstruation in young normal looking adolescent female should raise suspicion towards Mayer-Rokitansky-Kuster-Hauser Syndrome. Proper counseling of the patient regarding her future fertility and available therapies (surgical and non-surgical) should be explained. While MRKH syndrome is not a life-threatening disease, the diagnosis in adolescence has a significant influence on a patient's life. Hence, counseling and proper supportive care are the mainstay for the management.*

Key Words: *Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome, Mullerian agenesis, Neovagina, aplasia of the uterus, Primary amenorrhea.*

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INTRODUCTION:

TheMayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is characterized by congenital hypoplasia of the uterus and the upper part of the vagina. The incidence of MRKH syndrome has been estimated as 1 in 4500 newborn girls [1]. Its features include partial or complete absence (agenesis) of the uterus with an absent or hypoplastic vagina, normal fallopian tubes, ovaries, normal external genitalia and the typical 46, XX, female chromosome pattern. Breast development and growth of pubic hair are also normal. Associated renal and/or skeletal abnormalities are common [2].

Often, the first noticeable sign of MRKH syndrome is that menstruation does not begin by age 16 (primary amenorrhea) [3].

HISTORICAL BACKGROUND:

This syndrome was first described by Mayer in 1829 and Rokitansky in 1838. Uterine and vaginal agenesis were reported by Rokitansky and Mayer described some vaginal duplication [4].
In 1910, Kuster recognized urologic association and in 1961 Hauser differentiated MRKH from androgen insensitivity syndrome (AIS) [5].

CLASSIFICATION OF MRKH SYNDROME:

MRKH syndrome is classified into three groups[6]:

1. Type I/Typical MRKH**:** It is characterized by isolated uterovaginal agenesis.
2. Type II/ Atypical MRKH: It is associated with renal and ovarian malformations.
3. MURCS associated MRKH: Third type is Mullerian duct aplasia, renal aplasia, and cervico-thoracic somite dysplasia association with renal, skeletal, and cardiac malformations, and hearing impairment.

CASE REPORT:

A 16 year old girl presented with a history of primary amenorrhea. She did not complain of any gastrointestinal or urinary symptoms. Her Primary amenorrhea diagnostic workup was performed, as follows;

1. History: There was no history of delayed menses or primary amenorrhea or any other endocrine diseases in her family. Her age during the onset of breast development (thelarche) was 13 years. She never had any complaint of cyclical abdominal pain. There was no history of smell perception disorders. There was no history of any abdominal or inguinal surgery/ chemotherapy/ radiotherapy.
2. Physical Examination: She had normal growth parameter (normal height). Her BMI was recorded to be 18.75 Kg/m^2. The vitals were normal, blood pressure being 110/68mmHg. On physical examination, she had normal secondary sexual characteristics. Breast development was tanner stage IV, axillary hair were present and pubic hair growth was tanner staging IV. She had normal looking external genitalia and vaginal opening could be seen. There was no palpable abdominal mass, inguinal hernia or gonads in the inguinal region.
3. Imaging: On Ultrasonography, the pelvis showed normal right and left ovaries measuring 2.8 X 2.3cm and 2.5 X 1.4cm respectively with non-visualization of uterus suggestive of aplastic uterus.On MRI, the kidney, ureter and urinary bladder were visualized without any abnormality. The uterus was not visualized and suspected as a bud on the postero-inferior aspect of urinary bladder was seen, suggestive of aplastic or hypoplastic uterus. Ovaries were normal in size, shape and intensity with multiple small follicles.
4. Hormonal Evaluation: Her hormonal profile; FSH, LH, Prolactin and TSH was within normal range.
5. Karyotyping: On chromosomal analysis she had a karyotype of 46, XX.

Thus, the above workup determined the diagnosis of Mayer-Rokitansky-Kuster-Hauser syndrome. The patient was counseled for surgical correction of vagina (neovaginoplasty).

DISCUSSION:

The typical clinical presentation of MRKH syndrome is primary amenorrhea in association or not with cyclic colic pain, in an adolescent with secondary sexual characteristics compatible with age with no sign of virilization. As discussed in the introduction there are two subtypes of MRKH syndrome, the typical (type I or isolated) and the atypical form (type II). The frequency of type II being much greater than type I [7-9]. The typical form is characterized by sonography findings of symmetric muscular buds (the Mullerian remnants) and normal Fallopian tubes; this is referred to as Rokitansky sequence, where only the caudal part of the Mullerian duct (upper vagina and uterus) is affected. On the other hand, atypical form shows, in addition to what was mentioned above, asymmetric hypoplasia of one or two buds, with or without dysplasia of the Fallopian tubes. It may be associated with renal alterations (40%): renal agenesis, ectopic kidney, horseshoe kidney [10]. Patient usually undergoes puberty with normal thelarche and adrenarche, however, menses do not begin (i.e., primary amenorrhea). Because ovarian

function is normal, patients experience all bodily changes associated with menstruation. There is usually infertility, difficulty with intercourse, voiding difficulties, urinary incontinence, or recurrent UTIs and/ or vertebral anomalies (most commonly scoliosis) [11].

The etiology of this syndrome is unclear; however, genetic involvement has been confirmed by various researchers, showing autosomal dominant inheritance or limited chromosomal imbalance undetectable in standard karyotypes [12].

For the diagnosis of MRKH syndrome, transabdominal sonography is a useful and non-invasive technique. It is the first investigation in evaluating patients suspected of having the syndrome. Magnetic resonance imaging provides a more accurate diagnosis.

The karyotype of the patient in the current case report is 46, XX and the endocrine balance was normal, with transabdominal sonography showing aplastic uterus, suggesting mullerian agenesis/ MRKH syndrome. Once the MRKH syndrome is suspected a complete checkup must be undertaken to evaluate the associated malformations such as renal and vertebral abnormalities. The differential diagnosis of MRKH syndrome includes congenital absence of vagina, a low transverse vaginal septum, an imperforate hymen, as well as 46, XY disorders of sex development, including androgen insensitivity syndrome (AIS) and 17α-hydroxylase deficiency [12].

Treatment is usually delayed until the patient is ready to start sexual activity. The management depends upon the type and underlying abnormality. It may be either surgical or non-surgical but the chosen method has to be individualized. The non-surgical options commonly used are the Franks'technique or perineal dilatation. The surgical techniques include various options like McIndoe technique, William vaginoplasty, Rotational flap procedure, Intestinal neovagina and Vacchietti technique. In addition, it is important to manage psychological symptoms in women with mullerian agenesis. This is because a young woman who discovers that she has a congenital malformation involving her reproductivity may develop extreme distress about her fertility, feminity and physical appearance [13].

CONCLUSION:

The prevalence of MRKH syndrome is often underestimated due to lack of indepth evaluation, late diagnosis and undefined aetiology. Physicians should be well equipped to counsel a patient with MRKH syndrome regarding her option for future fertility, diagnosis and available therapies. Especially in the event of surgery of the neovagina, the pivotal point is taking account of patients' maturity, carefully considering dealing with any precluded feelings of inferiority or solitude, both on the part of the patients as well as of their families. While MRKH syndrome is not a life-threatening disease, the diagnosis in adolescence has a significant influence on a patient's life. For such patients, with adequate treatment, sexual relations are possible, and fertility options are available.

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