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Mayer-Rokitansky-Küster-Hauser (MRKH) Syndrome Presenting with Condylomata Acuminata in a 28 Years Old Zambian Woman: A Case Report

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ABSTRACT

We present a rare form of Differences in sex development (DSD) that presented with Human papilloma virus (HPV) infection. The case is that of Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome presenting with condylomata acuminate in a 28 years old Zambian woman. The diagnosis was made clinically and in the operating theatre. The case underscores the importance of taking a thorough gynaecological history and physical examination in all female patients that present with sexually transmitted infections as rare DSD may be missed due to cultural barriers. Imaging techniques are key to confirming the diagnosis of DSD such as MRKH syndrome.

Keywords: Rare, Differences, Sex, Development, Diagnosis, Management.

INTRODUCTION

Differences in sex development (DSD), also known as Disorders of Sex development or Variations in Sex Characteristics (VSC) are rare congenital conditions involving genes, hormones, and reproductive organs, including genitals. In these disorders, development of chromosomal, genital, or anatomical sex is atypical [1]. One of these disorders is called Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome [2]. MRKH syndrome is named after four authors that originally described this condition over a period of 130 years. These were German anatomist Augus Franz Josef Karl Mayer in 1829, Austrian anatomist Carl von Rokitansky in 1838, German gynaecologist Hermann Küster in 1910, and Swiss gynaecologist Georges Andres Hauser in 1961. MRKH syndrome, also known as Mullerian aplasia or congenital absence of the uterus and vagina, is congenital disorder characterized by agenesis or aplasia of the uterus ad upper part of the vagina in females with a normal female karyotype (46, XX) [3]. In this syndrome the external genitalia appear normal and patients will have a normal reproductive andocrine function and will reach puberty showing normal signs of thelarche and pubarche [3]. Here the patients will typically present with primary amenorrhoea during adolescence. The condition is sometimes

associated with extragenital malformations involving mainly the kidneys and skeleton. The prevalence of MRKH syndrome is about one in five thousand live female births [4]. Condylomata acuminata (CA) are anogenital warts caused by Human papillomavirus (HPV) [5]. The strains of HPV that cause most of anogenital warts are 6 and 11. The virus is a double stranded DNA virus that is transmitted via penetrative sexual intercourse. Age, lifestyle, and sexual practices all play a role in one' susceptility to developing this sexually transmitted infection [6]. The condition is characterized by skin-coloured, fleshy papules in the anogenital region. HPV is the most common sexually transmitted infection globally with 9 to 13 percent of the world population being infected [7]. We present a case of MRKH syndrome presenting with CA. The case demonstrates the cultural obstacles in early diagnosis and management of MRKH syndrome.

THE CASE

A 28 years old female Zambian patient was seen in our Gynaecologic clinic at a University Teaching Hospital in Zambia years ago. Her presenting complaint was growths on her external genitalia. She gave no history of menstrual disorder of any type, dyspareunia, and dysuria. In addition, she gave no history of having had children and having ever been pregnant. On physical examination, the patient had all the features of a woman. She was a beautiful woman. Gynaecological examination by inspection revealed genital warts on the vulva. Digital examination was not done. A diagnosis of CA was made. The patient was booked for electrocauterisation under general anaesthesia. When the patient was under general anaesthesia and before the operation commenced, the lead doctor conducted a thorough vaginal examination digitally and by Sims speculum. The examination revealed a shallow vagina of about 3 centimetres deep. The diagnosis at this moment changed from mere CA to include a substantive DSD in the name of MRKH syndrome. A diagnosis of MRKH syndrome fitted well in the definition considering the depth of the vagina on digital examination. The CA lesions were successfully cauterized. We could not confirm the diagnosis of MRKH syndrome by radiological imaging such as Computer Tomography or Magnetic Resonance Imaging since the hospital did not have such specialized diagnostic techniques at the time. The patient was discharged and reviewed in the clinic for the CA. Nothing was done for the MRKH syndrome at that time.

DISCUSSION

Our patient presented with a rare DSD in the form of MRKH coupled with HPV infection in the form of CA. Whereas CA diagnosis was made clinically in the clinic the diagnosis of MRKH was made under general anaesthesia. The diagnosis of MRKH would have been made earlier if a thorough gynaecological history had been taken and examination done in the clinic. The diagnosis of MRKH would have been confirmed with imaging techniques such as Computer Tomography and magnetic Resonance Imaging.

To understand how MRKH syndrome comes about, knowledge of the embryology of the female reproductive tract in humans is critical. The oviducts, uterus, cervix, and upper two thirds of the vagina originate from the paramesonephric (mullerian) duct, whereas the lower part of the vagina originates from the urogenital sinus [8]. The aetiology of MRKH syndrome is currently unclear. Possible causes include monogenic, oligogenic, polygenic, multifactorial, and environmental factors [3]. Studies supporting familial occurrence of MRKH syndrome and its associated anomalies give credence to a monogenic cause [9]. However, in most cases occurring sporadically the cause may be either polygenic or multifactorial or non-genetic [10]. Patients typically present with primary amenorrhoea during adolescence. Additional complaints include

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dyspareunia or apareunia and abdominal pains. Diagnoses that show similarities with MRKH syndrome include vaginal agenesis occasionally misinterpreted as imperforate hymen or transverse vaginal septum, rare form of mullerian agenesis associated with clinical virilization or hyperandrogemia caused by mutations in WNT4 and complete androgen insensitivity syndrome, which is an X-linked disorder affecting genetically males (46, XY). To confirm the diagnosis of MRKH syndrome, physical examination of external genitalia is conducted with imaging. Imaging includes transabdominal and vagina ultrasonography that reveals the absence of the uterus and presence of ovaries. The gold standard imaging method for the diagnosis of uterovaginal agenesis in MRKH syndrome is Magnetic Resonance Imaging (MRI). MRI should be performed always when available. Laparoscopy may be indicated for diagnosis where surgical removal of this tissue is needed [11]. Chromosomal analysis by G/Q-banding is often done to confirm normal female karyotype (46, XX) [3]. Creation of a functional neovagina is central in the treatment of vaginal agenesis in MRKH syndrome. Over the years various surgical and non-surgical methods have been prescribed for vaginal construction. Surgical procedures have included vaginoplasties using various autografts such as M^c Indoe vaginoplasty, Baldwin vaginoplasty, Davydow vaginoplasty, and Williams vulvovaginaplasty [12]. Self-dilation of vagina, also known as Frank's method, is the most commonly used non-invasive method [13]. Another non-invasive method is dilation by sexual intercourse, also known as D'Alberton's method [14]. Diagnosis of CA is typically made clinically. This follows a definitive history and thorough visual examination of the lesions. There is no cure for HPV. Available treatments are directed towards the removal of the lesions. However, these lesions may also involute on their own without any treatment. [15]. There is no evidence suggesting that removal of visible lesions reduces transmission of underlying HPV infection. Treatment for CA is classified as either physical ablative or topical agents. Physical ablative therapies are more effective at initial wart removal but have significant relapse rates [15]. Physical ablation methods include Liquid Nitrogen Cryosurgery, electrocauterization or Loop electrical excision procedure, and formal surgical procedures under general anaesthesia or Spinal anaesthesia [15]. Topical agents include 0.15-0.5% Podophyllotoxin, also called Podofilox [16], Imiquimod cream [17], and Sinecatechins, an ointment of Catechins [18].

CONCLUSION

This case has emphasized the importance of a through gynecological history taking and physical gynecological examination to diagnose DSD such as MRKH syndrome in patients that present with STI such as CA. Many patients experience difficulties talking about feelings and emotions related to DSD such as MRKH syndrome. It is therefore important to be aware of potential cultural aspects and their influence on reactions to the diagnosis in patients and their families and friends.

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References

1. Lee PA, Houk CP, Ahmed SF, and Hughes IA (2006). Consensus statement on management of intersex disorders. International consensus conference on intersex. Pediatrics. 118(2): e488-500.

2. Hauser GA and Schreiner WE (1961). Mayer-Rokitansky- Küster -Hauser Syndrome. Rudimentary solid bipartite uterus with solid vagina. Schweiz Med Wochenschr. 91: 381-384.

3. Morten Krogh Herlin, Michael Bjørn Petersen, and Mats Brännström (2020). Mayer-Rokitansky- Küster-Hauser syndrome: a comprehensive update. Orphanet J Rare Diseases. 15: 214.

4. Herlin M, Bjørn A-MB, Rasmussen M, Trolle B, and Petersen MB (2016). Prevalence and patient characteristics of Mayer-Rokitansky-Küster -Hauser Syndrome: a nationwide registry-base. Hum reprod. 31: 2384-2390.

5. James William D, Elston Dirk, Treat James R, Rosenbach Misha A, and Neuhaus Isaac. 19 viral diseases; genital warts. Andrews' Diseases of the skin: Clinical Dermatology. (13th Ed). Edinburgh: Elsevier (2020). PP. 406-410.

6. O'Mahony C, Gomberg M, Skerlev M, Alraddadi A, de Las heras-Alonzo ME, Majewski S, Nicolaidou E, Serdaroglu S, Kutlubay Z, Tawara M, Stary A, Al Hammadi A, and Cusini M (2019). Position statement for the diagnosis and management of anogenital warts. J Eur Acad Dermatol Venereol. 33(6): 1006-1019.

7. Kaderli R, Schnuriger B, and Brugger LE (2014). The impact of smoking on HPV infection and the development of anogenital warts. Int J Colorectal Dis. 29(8): 899-908.

8. Kobayashi A and Behringer RR (2003). Developmental genesis of the female reproductive tract in mammals. Nat Rev Genet. 4: 969-980.

9. Williams LS, Demir Eksi D, Shen Y, Lossie AC, Chorich LP, Sullivan ME, et al. (2017). Genetic analysis of Mayer-Rokitansky- Küster-Hauser syndrome in a large cohort of families. Fertil Steril. 108: 145-151.

10. Friedler S, Grin L, Liberti G, Saar-Ryss B, Rabinson Y, and Meltzer S (2016). The reproductive potential of patients with Mayer-Rokitansky- Küster-Hauser syndrome using gestational surrogacy: a systematic review. Reprod Biomed Online. 32:54-61.

11. ACOG committee Opinion. No. 728 (2018): Mujllerian Agenesis: Diagnosis, management, and Treatment. Obstet Gynecol. 131: e35-42.

12. Davydov S (1969). Colpopoiesis from the peritoneum of the ureterorectal space. Obstet Gynecol. 12: 252-257.

13. Frank R (1938). The formation of an artificial vagina without operation. Am J Obs Gynecol. 35: 1053-1055

14. D'Alberton A and Santi F (1972). Formation of a neovagina by coitus. Obstet Gynecol. 40: 763-764.

15. Lacy CJ, Woodhall SC, Wikstrom A, and Ross J (2012). European guideline for the management of anogenital warts

16. Scheinfield N, and Lehman DS (2006). An evidence-based review of medical and surgical treatment of genital warts. Dematol. Online J. 12(3): 5.

17. Meltzer SM, Monk BJ, and Tewari KS (2009). Green tea catechins for treatment of external genital warts. Am J Obstet. Gynecol. 3: 233. E1-7.

18. Mayeaux EJ and Dunton C. Modern management of external genital warts. J Low Genit Tract Dis (2008). 12(3): 185-

URL: http://dx.doi.org/10.14738/bjhr.1202.18386.