

# HIV infection in a woman with Mayer-Rokitansky-Küster-Hauser syndrome – psychological and clinical implications: a case report and literature review

Gabrielle Saden, Aleksandra Jastrzębska, Brygida Knysz, Monika Stępień

Department of Infectious Diseases, Liver Diseases, and Acquired Immune Deficiencies, Wrocław Medical University, Wrocław, Poland

## Abstract

**Introduction:** Inflammation and injury within reproductive organs, including artificial vagina, can facilitate human immunodeficiency virus (HIV) transmission. However, HIV infection in a patient with Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome has been rarely described. Both MRKH and HIV diagnoses are linked to psychological distress in patients.

**Case description:** We report on a case of a 34-year-old HIV-infected woman with MRKH syndrome. The presented report is the second described case of HIV infection in a woman with MRKH syndrome, the previous one dating back to 1992. Entry of infection were squamous cells, covering the wall of artificial vagina. Acquired immunodeficiency syndrome (AIDS) diagnosis was delayed because symptoms, such as thrombocytopenia, subfebrile illness, cachexia, malaise, and persistent diarrhea were misdiagnosed by physicians.

**Conclusions:** HIV infection is rarely described in women with MRKH syndrome; however, the procedure of artificial vagina can favor sexually transmitted diseases (STD) transmission, including HIV. In any case, a patient with unexplained symptoms or illnesses that may have any connection with HIV infection should be tested for HIV, even in the absence of risk factors. Moreover, these two diagnoses combined can be considered as huge psychological burden for the patient. Receiving information about incurable infertility due to MRKH during young adolescence might cause severe psychological complications. Despite many sources of HIV infection, ways of transmission, and risk factors, HIV patient still can face stigma associated with the infection. Psychological circumstances of HIV infection can include depressive symptoms, feeling of guilt, fear of consequences, and escape from reality.

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**Key words:** HIV infection, Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, HIV transmission, vaginal agenesis, neovagina.

## Introduction

The routes of human immunodeficiency virus (HIV) transmission are well-known, primarily through both heterosexual and MSM (men who have sex with men) contacts

as well as the use of drugs intravenously [1]. Since the end of the eighties of the last century, the highest percentage of HIV infections in Poland was found in drug addicts [2]. But currently, the number of transmissions through sexual

**Address for correspondence:** Gabrielle Saden,  
Department of Infectious Diseases, Liver Diseases, and Acquired  
Immune Deficiencies, Wrocław Medical University, 5 Koszarowa St.,  
51-149 Wrocław, Poland, phone: +33-661344704,  
e-mail: [Gabrielle.saden@student.umw.edu.pl](mailto:Gabrielle.saden@student.umw.edu.pl)

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contacts has increased [3]. The number of infections mainly depends on risky behaviors among people having sex with multiple partners, but infection can also happen after sexual contacts with only one HIV-positive partner [4]. Inflammation and injury within the reproductive organs facilitate HIV transmission as well as artificial vagina in women with Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome. However, HIV infection in such patients has been rarely described [5]. In the course of HIV infection, many different symptoms are observed, with HIV diagnosis necessary but often omitted. These symptoms include thrombocytopenia, weight loss, protracted low-grade fever, seborrheic dermatitis, shingles, and even lymphadenopathy, etc. [6]. Both MRKH and HIV diagnoses are linked to psychological distress in patients. We report a case of HIV-infected women with MRKH syndrome associated with diagnostic problems of symptoms arising from the infection and immune deficit.

## Case description

A 34-year-old female, a farmer by profession, was referred to the Department of Infectious Disease due to fatigue, cachexia, chronic diarrhea persisting for 5 months, and HIV infection diagnosed 3 weeks earlier.

Her medical history revealed that she had never been an intravenous drug user. However, she could have been infected from her sexual partner, with whom she had vaginal sexual contacts for one year. She denied having anal and oral sex. At that time, the couple was not aware of HIV infection of the man. At 18 years of age, after the diagnosis of Mayer-Rokitansky-Küster-Hauser syndrome, she underwent a procedure of creating an artificial vagina.

Clinical presentation included primary amenorrhea and difficulties in sexual life. Secondary sexual characteristics developed adequately to sex and age. Gynecological examination revealed muscle bridges in the place of vagina extending from the urethra to posterior commissure, lack of vagina, lack of uterus, and ovaries of normal structure. There were no other organ defects. The patient underwent procedure of vaginal reconstruction; the surgeon made an incision through the abdomen to access the peritoneal cavity, reached the area of the pelvis, and a neovagina was created through tunneling in the vesico-rectal space. During this procedure, a space between urethra, bladder, and rectum was extended, and to such prepared neovagina, a glass dilatator was inserted in post-operative period. For the first ten days, dilatators were exchanged every day, followed by only nightly insertion. She was advised to start sexual intercourses after three months after the procedure. In the interval between intercourses, she was advised to wear a prosthesis until termination of epithelialization of neovagina (for about 2-3 years). Such a procedure should have prevented obliteration of the vagina tube. After the epithelialization of neovagina surface, epithelial cells were found in smears.

Fourteen years later, at the age of 32, thrombocytopenia was observed and diagnosed as idiopathic. Platelet count was mainly between 9,000 and 51,000/ $\mu\text{l}$ . Due to purpura,

she was treated with corticosteroids without any effect, and eventually, she underwent splenectomy. After initial improvement and temporary increase of platelet count to 640,000/ $\mu\text{l}$ , in the further course of the disease, thrombocytopenia occurred again, with platelet count between 26,000 and 117,000/ $\mu\text{l}$ . She received corticosteroids again, and because of the poor response for that treatment, azathioprine was added.

About one year after the splenectomy, the patient was admitted to a hospital for hormonal tests. During that stay, suppression of pituitary-adrenal axis and corticosteroids side effects, such as hypokalemia and low bone mass were observed as well as multinodular goitre with autonomy of the left lobe. Due to uncertain result of the first histopathological examination and second one that revealed follicular thyroid cancer, near-total thyroidectomy was performed.

During the next 4 months, the patient was hospitalized twice in different internal departments, due to low-grade fever, chronic diarrhea, and abdominal pain. At first, colitis, biliary reflux, hiatal hernia, and stage 2 hemorrhoids were diagnosed. During subsequent hospitalization, the attention was focused on cachexia that could not be justified by the above-mentioned symptoms and lactose intolerance. Eventually, HIV test was performed with positive result.

Next, on admission to the department of infectious diseases, cachexia (body mass index, 15.4), fatigue, watery diarrhea (up to 7 stools per day) as well as advanced immune deficiency with a very low number of CD4 T-cells (9 cells/ $\mu\text{l}$ ) and HIV RNA with 175,130 copies/ml, were present. It was found that the cause of chronic diarrhea was an invasion of *Cryptosporidium* spp. According to guidelines, a combination of antiretroviral therapy (ART) was initiated. The patient received tenofovir disoproxil, lamivudine, darunavir, and ritonavir, and azithromycin due to cryptosporidiosis. Three weeks after the initiation of antiretroviral treatment, there was an improvement of the patient's condition, with better well-being, increase of body mass index (BMI) to 18.4, decrease of the number of stools from 7 to 1 per day, increase of number of platelet count up to 193,000/ $\mu\text{l}$ , and a significant increase in CD4 T-cell count up to 80 cells/ $\mu\text{l}$ . During the next several years of observation and ART, there was a subsequent improvement of the clinical state in terms of platelet count (435,000/ $\mu\text{l}$ ), CD4 T-cell count (607/ $\mu\text{l}$ ), and HIV RNA below detection limit (20 copies/ml). No additional comorbidities were observed.

## Discussion and conclusions

According to our knowledge, the presented report is the second described case of HIV infection in a woman with MRKH syndrome, the previous one dating back to 1992 [5]. MRKH syndrome is a congenital malformation that leads to the absence of uterus, cervix, and 2/3 upper part of vagina, with a properly developed sexual secondary characteristics and a normal karyotype 46XX. The frequency of MRKH syndrome is 1 case in 4,000 to 5,000 alive female births [7]. Malformations of the kidney, spine, ears, and heart can also

occur. The diagnosis is usually made while seeking the causes of amenorrhea after puberty. Treatment is based on surgical creation of an artificial vagina, enabling beginning of sexual life [8-10].

The patient went to a gynecologist at the age of 18 because of amenorrhea. MRKH syndrome was diagnosed and she underwent the procedure of creation of an artificial vagina. A long-term follow-up study [11] of patients, who underwent this procedure reported good results, with 80% of physiological epithelialization of neovagina, 96% of persisting sufficient vaginal depth, and 91% of lack of contractions. Some disadvantages of this method include the risk of secondary stenosis that requires life-long dilation as well as the risk of rectoceles and cystoceles [12]. Our patient did not suffer from any adverse event, and to this date, she is sexually active without pain during or after intercourse. However, this procedure could be a factor facilitating HIV transmission via vaginal sexual contacts [1]. HIV was transmitted unknowingly, probably during vaginal sexual contacts with her seropositive partner. The entry of infection were squamous cells, covering the wall of artificial vagina. Also, the infection could be favored by vaginal epithelium injuries from sexual contacts due to lack of wetting, as in elderly women with vaginal mucosal atrophy. Kell *et al.* [5] presented a case of a 31-year-old patient with a short vagina and repeated sexual intercourses. The extension of the vagina during sexual contacts and micro-traumas were most probably the cause of HIV transmission.

A very small part of the literature addresses the gynecological care of patients with MRKH syndrome, and it was described as a barrier to regular gynecological attention [13]. Regular check-ups and sexual education are important in these patients to prevent infections and implement appropriate screening. A study on human papillomavirus (HPV) vaccination in this population revealed that most respondents (73%) had never received any education about HPV vaccine [14], and their knowledge about the virus and sexually transmitted diseases was very low. A third of the patients was unsure if routine Pap smear should be performed in such individuals. This study confirms a lack of proper gynecological care for patients with MRKH. Another study reported 29% of recurrent urinary tract infections in patients with MRKH [15]. The risk of underlying malignancy of residual Müllerian tissues should not be excluded [16]. On the other hand, and given the advances in surgical techniques for the management of this condition, it is important to note that the tissue of neovagina is susceptible to infection not only by HIV but also by human papillomavirus (HPV) [17]. Extragenital skin tissue used for neovagina reconstruction could reduce its keratinization capacity after being grafted, which can increase predisposition to HPV infection [18]. It has also been described that the stress suffered by these implanted tissues from mechanical dilation, micro-abrasion, trauma from sexual activity, and exposure to substances, such as semen, urine, or feces, are also the risk factors of tissue dysplasia [17]. A study among patients, who previously underwent neova-

gina reconstruction due to MRKH and who were referred for evaluation and treatment of HPV neovaginal vulval-related lesions [19], found that the neovaginal pH ranged between normal values, and microbiological cultures provided results consistent with those of normal vaginal flora. The authors concluded that sexually active patients, who underwent neovagina reconstruction are HPV-exposed and they should be evaluated for infections after surgery. So far, there is no research describing the mechanism of HIV infection in patients with MRKH syndrome. Similarly, the impact of neovagina on HIV acquisition needs to be studied. There is an ongoing study conducted by the Thai Red Cross AIDS Research Center with primary aim to describe the neovagina in transgender women, evaluating the willingness and obstacles for transgender women with neovaginal and rectal HIV, and sexually transmitted infections (STI). Data collection started in 2013, and is supposed to end in 2024.

In the current case, AIDS diagnosis was established after 6 years of different therapies because of undiagnosed symptoms, such as thrombocytopenia and subfebrile illness, progressive cachexia, malaise, and persistent diarrhea lasting for several months. The symptoms occurred in the patient due to HIV infection, but were misdiagnosed, and therefore, it delayed the correct diagnosis.

Although information concerning HIV infection are widely available, there are still many late presenters, in whom HIV infection is identified when CD4 T-cell count is  $< 350/\mu\text{L}$  [20]. The reason is that the test is done when the symptoms occur, and despite the symptoms indicating HIV infection, the test is not ordered by medical staff [21]. The presented patient does not belong to the group of risky behaviors; she has never used illicit drugs, had few sexual partners, and worked as a farmer. The diagnosis of idiopathic thrombocytopenic purpura was the reason of corticosteroids and azathioprine treatment as well as splenectomy, which in fact excavated immunosuppression, leading to the thyroid cancer, cachexia, and cryptosporidiosis. Thrombocytopenia very often diagnosed as idiopathic, can be associated with HIV infection [22]. Frequently, the efficient treatment is highly active antiretroviral therapy [22]. There is no long-lasting effect after treatment with corticosteroids and azathioprine as well as after splenectomy. The use of ART resulted in an improvement, expressed as an increase in platelet count to  $193,000/\mu\text{L}$  after a few weeks from the beginning of the treatment. Also, the treatment of HIV wasting syndrome and cryptosporidiosis is based on ART. In fact, within three weeks after ART initiation, body weight increased, resolution of diarrhea was obtained, and a 9-fold increase in the number of CD4 T-cells was observed.

We would like to emphasize that both MRKH syndrome and HIV infection diagnosis can be considered as psychological burden for the patient. Receiving information about incurable infertility during young adolescence might cause severe psychological complications. Song *et al.* [23] observed that women with MRKH syndrome more often experience anxiety symptoms in comparison with healthy women.

Other psychological disruptions that can be found as comorbidities with anxiety are negative self-evaluation of femininity, neurotic personality traits, and depressive symptoms. Weijenborg *et al.* [24] reported that women with MRKH diagnosis can struggle pain during intercourses, sexuality-related distress, and sexual dysfunction. Interestingly, longer period since the creation of neovagina is correlated with less sexuality-related distress [24]. Women with MRKH diagnosis require interdisciplinary treatment, involving gynecologist, sexologist, and psychologist to help understand their condition, prepare for vaginal reconstruction, and adapt to sexual activity [7]. Additionally, a therapy should be considered, such as cognitive-behavioral group treatment [25]. Another aspect that should not be omitted is regular sexual activity in women with MRKH syndrome, which prevent from obliteration of the vagina tube. However, fear of pain or injuries during intercourses may cause discouragement or anxiety in patient's partner, or lead to erectile dysfunction. Therefore, the sexologist care is also crucial to maintain satisfying sex life of MRKH syndrome patients [7].

Another important aspect of this case is HIV infection. From the very beginning of the infection, patients with HIV require adequate care, also during giving the diagnosis. According to Senyuk *et al.*, patients can perceive that healthcare professionals inform about the diagnosis in an inappropriate way [26], which does not facilitate a good relationship with a physician and trust atmosphere from the very beginning of diagnosis. Despite many sources of HIV infection, ways of transmission, and risk factors, HIV-positive patients still might face stigma associated with the infection [27]. Psychological circumstances of HIV infection can result in depressive symptoms [28], such as feeling of guilt, the fear of consequences, and escape from the reality [29]. Surprisingly, healthcare providers are not excluded from discriminative behaviors towards HIV-infected individuals. According to Tavakoli *et al.*, the lowest stigma can be found in voluntary counseling and testing centers personnel, and the highest in paramedics, nurses' aides, and housekeeping staff [30]. Moreover, the patients might even face reluctance to being operated or undergo any medical interventions [26]. Taking into consideration all the above-mentioned psychological disturbances, the support for HIV-positive patients is crucial, and should be easily accessible [29].

HIV infection is rarely described in women with MRKH syndrome; however, the procedure of artificial vagina can facilitate STD transmission, including HIV.

In any case, a patient with unexplained symptoms or illnesses that may have any connection with HIV infection should be tested for HIV, even in the absence of risk factors. Early diagnosis of infection offers the possibility of a monitoring and immediate implementation of effective antiretroviral therapy, and can diminish psychological burdens associated with the lack of self-acceptance of their clinical state.

## Conflicts of interest

The authors declare no conflict of interest.

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