CHALLENGES IN THE MANAGEMENT OF LATE MANIFESTATION OF MAYER ROKITANSKY KUSTER-HAUSER SYNDROME: A CASE SERIES

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Abstract

The treatment of infertility caused by congenital abnormalities, especially of the mullerian dysfunction Like Mayer syndrome, remains a challenge in most developing nations. This is because of limited corrective or palliative surgical skills across the various disciplines involved in its management. The advent of Artificial Reproductive Techniques (ART) is yielding positive results for some of the challenges. Our intention is to highlight the diagnostic and management challenges associated with Mayer syndrome in our practice. Importantly, gynecologists should be on the alert due to the increasing occurrence of the condition, similarly, adequate training and re training should be the watch word.

Keywords: Challenges, laparoscopy, mullerian dysfunction, management

Introduction

Mayer Rokitansky-Kuster-Hauser Syndrome (MRKHS) is a congenital anomaly of the reproductive tracts affecting the normal development of the female internal organs which occur at about 4-12 weeks of intrauterine life with some sparing of the external genitalia. It is as a result of a halt in the development of the paramesonephric duct from which most of the female reproductive organs developed.¹ Its incidence has been widely acknowledged to be 1 in about half a million populations,² variant of this condition has been described.³ MRKHS is the second commonest cause of primary amenorrhea in developed countries, affecting 1 in 4 to 5000 live births.^{1,2}

Two types of MRKHS exist; Type I is an entirely pelvic abnormality affecting the development of proximal vagina and uterus, whereas the type II is external with associated heart, kidneys and spinal bone abnormalities,^{1,4} and sometimes coexisting with imperforate anus.⁵ Each can be corrected surgically to restore normal coitus but reproduction may be via artificial reproductive technology through egg donation and or adoption.^{1,2} These corrective interventions required training, skills, and team work among surgeons of many disciplines most of which are not available in one center. Thus, the need for

retraining and collaboration as the number of new cases increases. The commonest presentations of MRKHS are those of amenorrhea, coital difficulty and infertility.⁶ A hormonal survey is usually normal with slight changes in the testosterone levels. Radiological evaluation may reveal absent or rudimentary uterus with or without calyceal defects. Management is basically surgical requiring multiple disciplines. We present three cases of MRKHS, highlighting the general challenges associated with its management. This findings, we hope will alert the gynecologists on the rising incidence of this rare cause of primary infertility.

Case presentation 1

Mrs. SH was a 21 year old nulliparous woman who presented with inability to menstruate since maturity and a two-year history of infertility. She had been married for 2 years and had been experiencing coital difficulties. Thelarche was at 13 years and pubarche was normal. She experienced cyclical lower abdominal pain that lasted for two days. She had no history of surgery, chemotherapy, anosmia or features of Poly cystic ovarian syndrome (PCOs). She was a young woman with normal female physical features. No facial dysmorphic features. She had well-

developed breasts and female external genitalia. Pelvic examination showed blindly ended vagina with a depth of less than 6cm, no pelvic organs felt. A digital rectal examination also revealed no uterus. She had an ultrasound that showed atretic uterus, there was a focal hypoechoic area seen on the right adnexa (fig.1), other abdominal organs were normal. Hormonal profile indicated normal FSH and LH with slightly elevated prolactin.

She had laparoscopy with findings of a clean peritoneal cavity. She had normal tubes and ovaries bilaterally, but these ended in hypoplastic uterine tissue on each side. She did well post operatively and was counseled for serial vaginal dilatation (to enable coitus) and possible invitro fertilization (IVF) or adoption.

Case presentation 2

Mrs. HA was a 22-year old P0+0 with primary amenorrhoea. She presented with a 5 year history of recurrent cyclical lower abdominal pain and a year history of inability to conceive. She has been married for two years with initial regular intercourse but without satisfactory penetration associated with deep dyspareunia. She had no history of recurrent pimples, hoarseness of the voice or growth of beards, no history of anosmia, weight gain, heat or cold intolerance. Mrs. HA was married to a long distance driver in a monogamous setting, with no past history of surgery or mumps orchitis.

She was a young lady, with well-developed breast (fig.2), axillary and pubic hair Tanner Stage V. The abdomen was flat and moved with respiration, with no palpable organomegaly, there was female type external genitalia with normal female pubic hair pattern, the vagina had normal opening but measured 5cm in depth (fig.3), no vaginal fornixes' felt, no palpable cervix and uterus. Adnexae was free. She was evaluated for primary infertility with primary amenorrhea secondary to outflow obstruction query atretic uterus. Buccal and peripheral blood smears showed positive Barr body (XX karyotype). An abdominopelvic ultrasound revealed right ovary with no uterus seen. Other hematological investigations were normal.

She had laparoscopy with findings of a clean peritoneal cavity, bilateral well developed ovaries, both fallopian tubes were visualized but fimbria ends were not well formed on both sides and the uterus was absent. A diagnosis of Mayer Rokitansky-Kuster-

Hauser syndrome was made. She was counseled on her condition and the need for IVF/ adoption.

Case presentation 3

A 22 year old nulliparous woman presented with inability to menstruate since puberty associated with lower abdominal pain. There was no history of anosmia, persistent headaches, excess hair growth or thyroid symptoms, she was yet to get married and have never had sexual contact.

Examination showed a young girl with well-developed secondary sexual characteristics. The abdomen was flat and moved with respiration with no organ palpable. Pelvic examination revealed normal female external genitalia with blind end vagina admitting only the index finger and a depth of about 3cm, there was no uterus palpable. Buccal smear showed 46xx. Other investigations were normal. She had diagnostic laparoscopy which confirmed a bulb connecting the fallopian tube (likely the atretic uterus) (fig.4), normal tubes and ovaries with stigmata of ovulation. She did well subsequently and was counseled on adoption.

Fig. 1. Sonographic image showing absent uterus, only right ovary

Fig.2. Showing a well-developed breast Tanner Stage V

Fig.3. A blind-ending vagina, about 5cm in length

Fig.4: laparoscopic image of right tube and ovary, with rudimentary uterus

Discussion

MRKHS is a rare abnormality of the mullerian system; the condition is usually not detectable in children and adolescents who may not be sexually active and also because of the development of normal secondary sexual characteristics. In our environment where parents are shy discussing sexuality with their female children, it makes the condition difficult to diagnose or even unnoticeable until after marriage where they manifest. Even at that level, due to the cultural and religious belief, couples find it difficult discussing such issues and seeking for medical intervention. Similarly, the diagnosis is also challenging since the condition is rare and the majority of gynecologists have not managed the condition in their practice.⁶ The best diagnostic tool is the 3D ultrasound and MRI which are not readily available in most centers in the developing nations.^{7,8,9} Additionally, Buccal smear for karyotyping also serves a great deal in the diagnosis, interestingly, two of the three clients had karyotyping done to aid evaluation. This is an easy tool for the diagnosis but may not be highly specific. A side from the diagnostic challenges, there is a psychologic effect of the disease that creates difficulty in assigning gender to the

client,¹ especially for some clients with other related abnormalities but the three patients presented had normal female external genitalia suggesting that most likely, they had the type I MRKH syndrome.

Both conservative and surgical management options have been proposed for MRKHS.^{8,10,11} but these two only offers palliative therapy. As it is currently, no definitive treatment option has been confirmed to be superior to the other.⁶ In a center like ours, however, due to late presentations, only a little can be done. Serial vaginal dilatation is one option of therapy but often associated with loss of self-esteem as well as the psychological effect of discomfort because of the number of hours the dilator is to be kept in the vagina; a range of 30 min to 2 hours per day.^{1,12,13} Where the expertise is available, surgical correction of the vaginal stenosis can be done using flaps/grafts from the skin on the buttocks {Mc indo- Reed procedure} and thighs,¹⁴ as well as the intestine or sigmoid colon. There have been reported cases of MRKHS that had sigmoid colovaginoplasty in Sokoto, Nigeria in 2010.¹¹

The management of MRKHS requires a multidisciplinary approach with the expertise of a general, plastic and urologic surgeons.^{1,8} This surgical expertise is not widespread in the country, thus, a great barrier to the management of MRKHS in most centres' in Nigeria. Neovaginoplasty can be done in childhood if condition is noticed early, however, the risk of stenosis and subsequent surgical intervention has been reported¹¹, that is to say, the late presentation as seen in our environment could have a positive advantage of improving success rates since tissues would have been more developed and serial dilation strictly performed. Laparoscopic intervention has also been done through the application of acrylic olive between the neovagina and abdominal wall for continuous pressure¹⁵, this was also a challenge in our facility and most centers across the country, largely due to lack of equipments and training. Even where the facility and expertise are available to conduct these procedures, there had been documented complications of vaginal stenosis and scarring. Thus, due to the above challenges, the majority of our clients only benefitted from serial vaginal dilatation as an option to improve their sexual satisfactions and ease of difficulties.

Another major intervention that could be done for MRKH syndrome is uterine transplant⁸, a procedure that could salvage the situation especially in our setting where a premium is given to child bearing.

This intervention is also a big obstacle in the management of MRKHS in the country largely because of the lack of expertise and the cost of post-op support for these clients. Additionally, invitro fertilization and surrogate mother can be an option to the childlessness associated with MRKS,^{8,16} but this option requires training on ART and also has great financial implications which might not be affordable to the client and even where the finances are available, the stigma associated with the procedure might not be tolerated by the couple. The last resort for most clients in our practice area will be adoption largely due to the costs associated with all the interventions mentioned and because of the common practice of extended family system.

Conclusion

MRKHS is a rare cause of primary infertility in young couples which is difficult to manage, routine examination of all girl child will help in its early detection. The role of training and retraining of both gynaecologists, urologist, and plastic surgeons is a key to effective management.

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