Pregnancy in a case of Mayer-Rokitansky-Küster-Hauser Syndrome
Gravidez num caso de Síndrome Mayer-Rokitansky-Küster-Hauser

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Abstract

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome refers to the congenital absence of the upper part (2/3) of the vagina with variable uterine development. In this disorder, infertility may be the most difficult aspect for the patient to accept. This review will describe a rare case of pregnancy in a woman with MRKH syndrome through assisted reproductive technology.

Keywords: Mayer-Rokitansky-Küster-Hauser Syndrome; Pregnancy; Assisted reproductive technologies.

INTRODUCTION

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome refers to the congenital absence of the upper part (2/3) of the vagina with variable uterine development and it is due to interrupted embryonic development of the müllerian ducts in the fifth week of gestation[1-3]. In the majority of cases it manifests through primary amenorrhea due to uterine hypoplasia or agenesis[3]. In this disorder, infertility may be the most difficult aspect for the patient to accept.

We will describe a rare case of pregnancy in a woman with MRKH syndrome achieved through assisted reproductive technology. The literature has been describing offspring in women with this syndrome but by using a gestational carrier[4,7,8].

CASE REPORT

A 33-year-old woman was referred to our centre for assisted reproductive technologies.

This patient had telarche and adrenarche when she was 12-year-old. At 13 years of age, still without menarche, she started complaining of cyclic pelvic pain.

By that time the patient was admitted to the hospital due to crampy abdominal pain associated with a history of amenorrhea. External examination revealed completed puberty with normal secondary female sexual characteristics and normal external genitalia. Patient underwent gynecological examination that revealed a rudimentary vagina or a transverse vaginal septum. Pelvic ultrasound showed a normal uterus distended by fluid suggestive of blood, normal ovaries, and a linear image not seen in all of it's extension, suggestive of vagina. The suspicion of MRKH syndrome was raised.

An abdominopelvic MRI was then carried out and showed and uterus distended by blood, normal ovaries and a short blindly ended vagina, with 2,5 cm of length, in it's inferior part.

An echocardiogram and a karyotype were performed and both were normal. At this moment it was concluded that this patient had the type I MRKH syndrome. She was submitted to vaginal permeabilization to resolve the cyclic pain due to arrested menstrual blood allowing her to have regular, although very painful menses.

At the age of 16, the patient started vaginal dilatation and some years later she was submitted again to vaginal permeabilization, and after referred unpainful regular menses.

When the patient became 20-years-old, with a vaginal length of 5cm, she started to have sexual activity but with intense dispareunia and blood loss.

Seven years later, to improve her sexual life, she was submitted to sigmoidal vaginoplasty, and since then refers normal sexual activity.

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At 31 years of age the patient started trying to conceive. After one and a half year of unsuccessful pregnancy she was submitted to assisted reproductive technologies treatments, that included two laparoscopic GIFTs, but none achieved pregnancy.

With 33 years of age the patient arrived at our sector and after the protocolar evaluation of the couple, the diagnosis of primary infertility due to congenital anomaly of the female reproductive tract was confirmed.

She was submitted twice to controlled ovarian hyperstimulation for IVF, by means of a flexible antagonist protocol.

In the first attempt, 6 oocytes were obtained on oocyte retrieval performed by vaginal route and guided by ultrasound, under general anesthesia. The thickness of the endometrium was 9mm. Two embryos were transferred transvaginally, at blastocyst stage, but this was technically very difficult due to severe stenosis of the upper third of the vagina and cervical canal. None of the other embryos had quality to be cryopreserved. Fifteen days after the β-hCG was negative.

In the second IVF attempt, 14 oocytes were aspirated. The endometrium had a thickness of 9 mm. Two embryos were transferred, again at blastocyst stage, but this time, by a transabdominal, transmiometrial route. It was technically easier but 2 weeks later the result was the same: no pregnancy was achieved. However, at this time, six embryos were cryopreserved, through slow freezing method.

Knowing that the vaginal route for embryo transfer was the best option to get pregnant, she was advised to improve the vaginal and cervical canals, so she was submitted to another surgery, a vaginoplasty with split-thickness skin graft from the abdominal wall.

Since six embryos were cryopreserved, she was submitted to a frozen embryo transfer. A blastocyst was transferred by vaginal route, using a hard catheter. Technically the transfer was easy. After two weeks, β-hCG was positive.

During the pregnancy she had two episodes of first trimester metrorraghia.

At 36 weeks of pregnancy she went into labour and was submitted to a cesarean section giving birth to a healthy baby.

DISCUSSION

The development of the female genital tract is a complex process. Around twelve weeks, the caudal part of the müllerian ducts fuse to form the uterovaginal canal and then the uterus and the upper vagina. The cranial unfused portion develops into the fallopian tubes. The lumen of the lower vagina has a different origin as it is formed from the urogenital sinus. This process is dependent upon a series of events and the failure of any one of these results in a congenital anomaly, such as the MRKH syndrome[1].

MRKH syndrome is subdivided in two types: type I or Rokitansky sequence and type II or *MURCS* association (*MU*llerian duct aplasia, *R*enal dysplasia and *C*ervical *S*omite anomaly). Type I syndrome is characterized by an isolated absence of the upper 2/3 of the vagina whereas type II is marked by other malformations which include renal, vertebral, otologic anomalies and, more rarely, cardiac and digital defects[1,3,4,5].

The incidence of MRKH syndrome has been estimated to be 1 in 5000 live births[3].

Although the vast majority of cases of MRKH seem to be sporadic, familial aggregates have been reported (>20%)[4]. The mode of inheritance seems to be autosomal dominant with an incomplete degree of penetrance and variable expressivity[3,4]. The question of whether or not the MRKH is manifested in the male has been raised. In fact combinations of Wolffian duct agenesis or severe hypoplasia, with or without, renal and/or skeletal anomalies have been described and have been found in families with females with MURCS association[3,4].

The first clinical signal is primary amenorrhea in patients presenting a normal female phenotype, normal 46XX karyotype, normal and functioning ovaries, normal secondary sexual characteristics and normal external genitalia. Most women have a rudimentary non-functioning uterus but 2 to 7% have a uterus with functioning endometrium and may present cyclic pelvic pain due to hematometra[3].

Ultrasonography should be the first exam to be performed and if inconclusive or incomplete a MRI should be done, since it is more sensitive and specific[3,5].

Laparoscopy is performed in cases of doubtful diagnosis after ultrasonography and/or MRI and should be reserved for women in whom interventional therapy is likely to be undertaken[3].

Differential diagnosis includes androgen insensitivity syndrome, Turner syndrome, low-lying transverse vaginal septum and imperforated hymen[3,4,5].

Treatment consists in creating a neovagina, by surgical or nonsurgical way, allowing patients to have normal sexual function, but must be offered only when they are emotionally mature and ready to start sexual

activity[1,3,6].

Nonsurgical treatment is recommended as first line and the most commonly used is Franck's dilator method that involves the application of vaginal dilators (Hegar candles), progressively increasing in length and diameter. This option is highly successful (78-92%) and complications rare[3,6].

Surgery may be appropriate if nonsurgical therapy fails or if the patient chooses surgery as the first line approach. A number of techniques are appropriate but the approach is often dependent on the surgeon's experience.

The Abbe-McIndoe operation involves the dissection of a space between the rectum and the bladder, placement of a mold covered with a skin graft into the space, and diligent postoperative vaginal dilatation[1,3].

The peritoneal vaginoplasty, referred to as the Davydov procedure, adopted the use of peritoneum to cover a newly created vaginal space[1].

William's vaginoplasty uses a vulvar flap to form a vaginal tube[1].

The original Vecchietti`s operation involves the creation of a neovagina via dilatation with a traction device attached to the abdomen, sutures placed subperitoneally by laparotomy and a plastic olive placed in the vaginal dimple. Nowadays it is performed by laparoscopy[1,3].

Sigmoidal colpoplasty involves vaginal replacement or creation of a neovagina by grafting a 12-18 cm long segment of sigmoid[3].

All surgical techniques are associated with complications, which included vesicovaginal and rectovaginal fistulae, bladder perforation, graft contracture, keloid scar formation and skin graft site disfigurement. There have also been cases of malignancy reported in surgical neovagina.

Clinical follow-up and regular intercourse are important in the mid and long term successful process[3].

Ultimately, infertility will be the most difficult aspect of the disorder for the patient to accept. Nowadays, reproduction may be possible, in some cases, if assisted techniques are performed. The risk for transmission of the disorder cannot be accurately evaluated since very little is currently known about the genetics of MRKH syndrome.

This patient has type I MRKH syndrome, also called Rokitansky sequence, once the only anomaly present is the agenesis of the upper part of the vagina. She was submitted to treatment at the wrong age, since at the age of 16 she was far from starting sexual activity and, as we said before, that moment is the best to start treatment. She achieved a reasonable sexual life, but she wanted more: she wanted to get pregnant.

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