A rare case of Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome: relief of symptoms with fractional CO_2 laser therapy

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ABSTRACT

We report the case of a 27-year-old woman with Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome suffering from vaginal dryness, dyspareunia and recurrent vaginal infections, unresponsive to non-hormonal local treatment (lubricants and moisturizers) and to antibiotics drugs, who underwent CO_2 laser therapy. She underwent a series of three laser treatments (one every four weeks), and a further treatment 18 months later. Objective (Vaginal Health Index, VHI score) and subjective (Visual Analogue Scale, VAS, scores for dyspareunia and vaginal dryness) evaluations were employed. After the three laser treatments, there was an improvement in all the scores employed until the eighteen-month follow-up, when the patient again complained of vaginal dryness (VAS score 4). She underwent another laser application and felt well again. Vaginal CO_2 laser therapy was effective in treating vaginal dryness in this patient. This treatment can be used not only in post-menopausal patients, but also in other women who suffer from vaginal dryness and dyspareunia due to particular pathological conditions.

KEYWORDS

Mayer-Rokitansky-Kuster-Hauser syndrome; vaginal reconstruction; vaginal CO_2 laser therapy; dyspareunia.

Introduction

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is characterized by congenital aplasia of the uterus and the upper part of the vagina associated with physiological development of the secondary sexual characteristics and with a normal female karyotype (46 XX) [1]. Its incidence has been reported to be 1/4000–1/5000 in newborn girls [1] and it is characterized by amenorrhea and infertility [1]. Etiologically, MRKH syndrome is due to a failed fusion on the median line of the Mullerian ducts [1]. The smooth dorsal bundle of muscles of the bladder and of the rudimentary vagina is regularly shaped, because these structures arise, respectively, from Wolff's duct and from Gartner's duct. The ovaries arise from mesenchyme and from the epithelium of the genital crest of the intermediate mesoderm, so, generally, there are no anomalies of ovarian development [1]. This syndrome seems to be transmitted as an autosomal dominant characteristic [1]. There are two different types of MRKH syndrome: the first is characterized by complete aplasia of the uterus and the presence of two rudimentary horns connected by a peritoneal fold and normal Fallopian tubes; the second type is characterized by uterine hypoplasia and hypoplasia or aplasia of one or both tubes and it may be associated with urinary, skeletal, auditory and cardiac anomalies [1,2]. Generally, the first symptom of the syndrome is primary amenorrhea, with normal ovarian anatomy, no signs of androgen excess, and a shorter vagina that causes dyspareunia [1,3].

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The diagnosis is usually completed with abdominal and pelvic ultrasound, magnetic resonance imaging and laparoscopy, which is the gold standard for evaluation of MRKH syndrome [1]. In the last few years, laser therapy has been introduced in the field of gynecology as a new non-hormonal option for the treatment of vulvo-vaginal atrophy (VVA) in post-menopausal women [4-7]. Here we report the case of a patient in whom vaginal CO_2 laser therapy was used to treat symptoms of MRKH syndrome.

Case presentation

A 27-year-old Italian woman with MRKH syndrome, suffering from vaginal dryness, dyspareunia, and recurrent vaginal infections, unresponsive to topical non-hormonal treatments (lubricants and moisturizers) and to antibiotics, was referred to our center for vaginal laser treatment. The patient had been

diagnosed with MRKH syndrome at 14 years of age, when she was admitted to a regional hospital with recurrent pelvic pain and primary amenorrhea. She had no history of medical illness, there was no parental consanguinity, and she had no siblings with similar complaints. Detailed gynecological examination revealed normal secondary sexual characteristics with well-developed external genitalia and a blind vaginal pouch measuring about 3 cm in length. She had no urinary or bowel symptoms.

To establish the cause of the recurrent pelvic pain and amenorrhea, ultrasonography of the abdomen and pelvis was performed, which showed absence of the uterus and the presence of normal shaped ovaries. Hormone levels (follicle-stimulating hormone, luteinizing hormone, estradiol, testosterone and thyroid function test) were within normal limits. Karyotyping was normal (46 XX). Given the ultrasound findings, the patient underwent exploratory laparoscopy that confirmed the absence of the uterus and the presence of two rudimentary uterine hemihorns, one on each side. The ovaries were normal, both for morphology and position, as were the tubes. No other organ abnormalities were revealed. In conclusion, the girl was diagnosed with MRKH syndrome, type one. The gynecologists suggested that the patient undergo surgical therapy (construction of a neovagina) to restore a normal vaginal anatomy. Because of the patient's young age, she decided to wait and to do exercises with an intruder for vagina elongation for about twenty minutes a day for two years.

After only three months of this physical therapy, the vaginal length had increased from 3 cm to 6 cm, and after two years it had reached 11 cm. However, she persistently suffered from vaginal dryness and dyspareunia. During the screening visit at our center, we used objective and subjective parameters to evaluate her symptoms. We used the Vaginal Health Index (VHI) as an objective evaluation of vaginal dryness and the appearance of the vagina. This test consists of five measures: elasticity, fluid volume, pH, epithelial integrity and moisture. Each parameter is graded from 1 to 5: a higher score reveals normal vaginal health, whereas a low score indicates a dry vagina [8].

We also employed a subjective parameter, the Visual Analogue Scale (VAS), to evaluate both dyspareunia and vaginal dryness [9]. Because the patient gave higher VHI and VAS scores, we decided, with her agreement, to try CO_2 laser therapy. The patient signed a written informed consent document.

The patient was treated with a factional microablative CO_2 laser system (SmartXide2 V2LR, Monnalisa Touch, DEKA, Florence, Italy). A vaginal probe was inserted as far as the top of the vaginal canal; it was then rotated as it was being withdrawn in order to provide a complete treatment of the vaginal walls. The laser parameters employed for the treatment are summarized in table 1.

We employed the same protocol that is used in post-menopausal women: a series of three laser treatments, one every four weeks. The procedure was performed in the outpatient clinic. Nine key time points were considered for evaluation of the treatment results: before the first laser application (T1), before the second laser application (T2), before the third laser application (T3), three months after the third laser application (T4), six months after the third laser application (T5), nine months after the third laser application (T6), twelve months after the third laser application (T7), eighteen months after the third laser application (T8), three months after the fourth laser application (T9). At each evaluation, VHI and VAS scores were evaluated.

Before the first laser application the VAS score for dyspareunia was 7, falling to 5 before the second laser application, and 3 before the third laser therapy (Table 2). At three months of follow-up (FU) the patient did not have dyspareunia and the benefits were long lasting, until the 18-month FU. The VAS score for vaginal dryness showed the same pattern of improvement. Before the first laser application, the VAS score was 8, falling to 6 before the second laser application, and 4 before the third laser therapy. At three months of FU the patient no longer suffered from vaginal dryness and this result was long lasting, until the 18-month FU. Eighteen months after the last laser application the patient again complained of vaginal dryness (VAS score: 4). The VHI score improved from baseline to the 12-month FU. At the 18-month FU visit we observed decreased vaginal fluid volume and moisture with a reduced VAS score (Table 2). We decided, in agreement with the patient, to do another laser application to restore vaginal health, with benefit. Throughout the FU period, the patient never experienced irritation or inflammation of vaginal mucosa, and she never used topical cream containing hyaluronic acid, antibiotics, lubricants and moisturizers.

Table 1 Vaginal CO_2 laser parameters employed in the treatment.

	First laser application		Second laser application		Third laser application		Fourth laser application	
	Watt	Smart stack	Watt	Smart stack	Watt	Smart stack	Watt	Smart stack
Endovaginal	40	3	40	3	40	3	40	3
Introitus	20	1	25	1	30	1	30	1

Table 2 Vulvo-vaginal symptoms (dyspareunia and vaginal dryness) and Vaginal Health Index (VHI) during the observation period.

	T1	T2	T3	T4	T5	T6	T7	T8	T9
VAS for dyspareunia	7	5	3	0	0	0	0	0	0
VAS for vaginal dryness	8	6	4	0	0	0	0	4	0
VHI score	13	17	20	22	23	25	25	23	25

Discussion

The present case of MRKH syndrome merits discussion for the following reasons: it is notable that use of an intruder alone restored a normal vaginal length, and the use of CO_2 laser therapy in a young girl suffering from vaginal dryness, irritation and inflammation of vaginal mucosa and affected by MRKH syndrome constitutes an innovation.

It seems increasingly apparent that MRKH syndrome is a pathology with a complex and multifactorial etiology; new studies in the fields of genetics and embryology have been carried out in order to better clarify its etiology and open up possible new therapeutic horizons [1,10,11].

The aim of the treatment of MRKH syndrome is to create an anatomical and functional vagina that ensures psychological wellness and a satisfactory sexual life. A neovagina should have a correct axis canal of adequate size and a secretory capacity to allow intercourse [12]. Treatment must be offered to patients only when they are ready to start sexual activity, at or after adolescence, when the woman has reached physical and psychological maturity. There are two approaches: surgical treatment and non-surgical passive self-dilatation of the rudimentary vagina.

The first approach consists of different procedures including the Vecchietti procedure (increasing the vaginal size by gradually applying traction to the vaginal wall) or reconstruction of a neo-vagina using different tissues such as skin (McIndo-Reed), peritoneum (Davydov), or bowel [13].

The second approach is the first line of treatment and consists of regular use of dilatators of progressively increasing length and diameter [13]. The secretory capacity after passive dilatation depends on the single case, and some patients experience vaginal dryness even though they are not in menopause.

Salvatore et al. demonstrated that use of the microablative fractional CO_2 laser on the vaginal mucosa was immediately associated with initial tissue remodeling (i.e., activation of fibroblasts and neocollagenesis) [4]. To date, the largest studies [5-7; 14] looking at the use of CO_2 laser therapy have included only post-menopausal women. Moreover, in the field of gynecology, the other type of laser, the Erbium laser [15], is also currently used only in post-menopausal women.

No cases of MRKH syndrome treated with CO_2 laser have been reported in the literature, therefore there was no standardized protocol to follow. The results obtained in this case suggest that a treatment cycle of three laser applications can significantly improve the two most bothersome symptoms of VVA and vaginal health scores at 12-week FU in women not responding to or dissatisfied with previous local estrogen therapies.

This case report shows that the fractional CO_2 laser can be used not only in post-menopausal women, but in other women who suffer from vaginal dryness and dyspareunia due to particular pathological conditions.

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