

EXTENSIVE UTERINE MYOMATOSIS IN A WOMAN WITH SPINAL MUSCULAR ATROPHY: CASE REPORT

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ABSTRACT

Uterine fibroids, the most prevalent benign tumors in the female pelvis and common in women of reproductive age, have variable characteristics and are influenced by hormonal factors, such as estrogen and progesterone. This report describes the case of a 43-year-old female patient with a large uterine fibroid associated with spinal muscular atrophy (SMA) type II. The patient was at high surgical risk due to severe ventilatory disorders and restricted mobility. After diagnosis by pelvic ultrasound, she underwent total abdominal hysterectomy and bilateral salpingectomy. Surgery revealed a myomatous uterus comparable to a 7-month gestation, weighing 1340 g. The anatomopathological examination identified a myometrium with leiomyoma, secretory endometrium and chronic cervicitis, with no signs of malignancy. The study reinforces the relevance of early diagnosis and individualized management of uterine fibroids, especially in cases associated with challenging health conditions such as SMA.

Keywords: Leiomyoma, Hysterectomy, Case reports, Spinal muscular atrophy.

INTRODUCTION

Uterine fibroid, also known as leiomyoma or uterine fibroma, is the most common benign tumor in the female pelvis¹, primarily affecting women of reproductive age. It is a monoclonal mesenchymal neoplasm originating from smooth muscle cells of the myometrium and exhibits heterogeneous characteristics in terms of size, number, and location. The high prevalence of this type of tumor underscores its importance in women's health, requiring careful attention in both diagnosis and clinical management.

From a physiological perspective, fibroids are influenced by hormonal factors, primarily estrogen and progesterone, whose receptors are often overexpressed in these lesions. These hormones play a crucial role in tumor growth, explaining the relationship between fibroids and hormonal changes typical of reproductive age and the menopausal transition^{2,3}.

The clinical presentation of fibroids can be highly variable. Some women remain asymptomatic, while

others may experience significant symptoms such as abnormal uterine bleeding, pelvic pain, a sensation of pressure in the pelvic region, and anemia. Additionally, fibroids can cause compression symptoms in adjacent organs, including increased urinary frequency and bowel disturbances. The location and size of the fibroids are key factors in determining the severity of these symptoms, as well as their impact on fertility and reproductive outcomes^{2,3}.

The treatment of fibroids is individualized and considers the severity of symptoms, the size and location of the fibroids, as well as the patient's reproductive desires. For those seeking to preserve fertility, myomectomy—performed through techniques such as laparoscopy or hysteroscopy—is often recommended, as it allows selective removal of fibroids with faster recovery and symptom improvement. On the other hand, hysterectomy is considered a definitive solution for women who do not wish to conceive in the future^{2,4}.

Alternatively, there are conservative management options, particularly for patients who prefer to avoid surgery. These include selective progesterone receptor modulators, gonadotropin-releasing hormone (GnRH) agonists, and levonorgestrel-releasing intrauterine devices. These therapies aim to reduce the size of the fibroids and control symptoms, providing clinical relief without surgical intervention^{2,3}.

CASE REPORT

Female patient, 43 years old, virgin, with no active sexual life, under the care of Dr. Waldemar Naves do Amaral at Hospital Amparo and Clínica Fértil. During consultation and clinical anamnesis, uterine myomatosis was hypothesized. To confirm the diagnosis, a transabdominal pelvic ultrasound was requested, revealing a large uterine fibroid measuring 9.9 cm x 8.3 cm in its largest area and 5.6 cm x 4 cm and 4.8 cm x 4.7 cm in smaller areas. The patient had Spinal Muscular Atrophy (SMA) type II, was on Risdiplam, presented significant musculoskeletal alterations, had limited abdominal access, a high pneumological risk, severe ventilatory disorder, and relied on a wheelchair for mobility.

The patient underwent a total abdominal hysterectomy and bilateral salpingectomy. The surgical procedure was performed by Dr. Waldemar and his gynecological surgery team. Extensive uterine myomatosis was identified, with the uterine size comparable to a 7-month pregnancy, weighing 1340 grams, and with apparently normal fallopian tubes and ovaries.

The sample was sent for examination. The histopathological examination of the uterus revealed a myometrium containing leiomyoma, a secretory endometrium, a cervix with chronic cervicitis, and no signs of malignancy. Upon evaluation of the fallopian tubes, the smaller tube showed vascular congestion, a focus of chronic perivascular inflammatory process, and no signs of malignancy. The larger fallopian tube displayed Morgagni's hydatids and no malignancy.

The surgery progressed successfully, and the patient had a good recovery, with no complications. During her hospital stay, she remained afebrile and showed no signs of alarm.



Figure 1 - Uterine Myomatosis and Adnexa



Figure 2 - Patient with SMA Type II

DISCUSSION

This study presents a case report aimed at describing the surgical treatment of a large uterine fibroid. Uterine fibroids affect about 40% of women of reproductive age. The main risk factors include advanced age, early menarche, family history, obesity, and nulliparity. Fibroids originate from the uterine smooth muscle tissue and follow a benign course, with their origin linked to the hormones estrogen and progesterone.⁵ Large fibroids are difficult to treat clinically, often leading to heavy menstrual bleeding, which can progress to severe anemia and a higher chance of requiring additional surgeries. In patients who do not wish to become pregnant, the traditional treatment is hysterectomy. For women who wish to preserve fertility, abdominal myomectomy is the treatment of choice. However, complication rates in subsequent conceptions are high, including uterine perforation, excessive bleeding, and fluid overload. Additionally, treatment is more complex in cases with concomitant comorbidities⁶.

Spinal muscular atrophy (SMA), for example, is an autosomal recessive genetic condition that affects the lower motor neurons and can be divided into 5 different subtypes. Type II, also known as Dubowitz disease, is the intermediate phenotype, with the following characteristics: trunk and limb weakness, inability to stand or walk, progressive scoliosis, contractures in the knees and hips, and joint laxity in the hands and fingers. Despite advances in new medications for treatment and the discovery of the responsible gene, SMA remains characterized as an incurable disease.⁷

The diagnosis of fibroids is made through ultrasound, which provides reliable data, especially when combined with transvaginal ultrasound, which is accurate in more than 90% of cases.^{5,8} To determine the treatment, whether surgical or medical, it is necessary to identify the size of the fibroid. In the presented case, the complexity lies in the size of the fibroid associated with the existing comorbidities. Surgery for large fibroids is challenging, and the standard approach consists of total hysterectomy along with bilateral salpingo-oophorectomy⁹, which was performed in the reported case. For the best surgical outcome, a multidisciplinary approach, involving specialties such as general surgery, plastic surgery, and urology, may be recommended to assist the gynecological surgeon.

CONCLUSION

The study reports a successful case of total hysterectomy combined with bilateral salpingectomy for a large fibroid. Fibroids are benign tumors with a high prevalence, primarily affecting women of reproductive age. The diagnosis is made through ultrasound, as the clinical presentation can vary. For treatment, it is essential to analyze the factors reported by the patient and the size of the fibroid, with options for both medical and surgical approaches.

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