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Case report

Primary umbilical endometriosis in a nulliparous woman: A rare case report

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ABSTRACT

Introduction and importance: Endometriosis describes the presence of endometrial tissue outside the uterine cavity. These patients often experience cyclic pain, dysmenorrhea, dyspareunia and infertility. Extra-pelvic endometriosis, particularly at the umbilicus, is rare. The exact incidence of endometriosis is unknown; definitive diagnosis requires surgical exploration and histopathological confirmation. Conservative, medical and surgical approaches are used in treatment. A combined approach is most useful in cases where pain is a prominent symptom. There is limited discussion of umbilical endometriosis in the literature, most information is derived from case reports.

Case presentation: Herein we present the case of a 35-year-old nulliparous woman in Northern Tanzania with a 10-year history of infertility. On presentation this patient reported a three-year history of a cyclical aching pain associated with an umbilical mass. An abdominal MRI revealed an ill-defined, enhancing mass measuring 3 × 4 × 6 cm located along the right anterior abdominal wall. The mass was connected to a sinus tract extending inferiorly to the suprapubic region but showed no communication with the peritoneal cavity, suggestive of endometriosis. Furthermore, bilateral adnexal lesions demonstrated hyperintense signals with focal hypointense areas and variable restrictions, consistent with bilateral ovarian endometriomas. The patient underwent excision of the umbilical mass, and histopathological examination confirmed the diagnosis of primary umbilical endometriosis. Despite her stable condition following management, she did not conceive over the course of the following year.

Clinical discussion: This case describes a case of primary umbilical endometriosis and bilateral ovarian endometriomas in a 35-year-old nulliparous woman. Surgical excision and histopathological analysis confirmed endometriosis. However, following intervention the patient was still unable to conceive. Whilst this is perhaps not unsurprising given the patient's age it is important for us to consider all possible explanations for her infertility. Critically, this case emphasizes the need for early intervention and comprehensive management of endometriosis-related fertility challenges.

Conclusion: We aim to provide a detailed description of this case in order to support clinicians who may encounter similar cases in the future especially in resource poor settings. We are providing data to support the theory that primary umbilical endometriosis can occur spontaneously in patients without a surgical history. Enhancing clinician awareness of this condition and fostering interdisciplinary collaboration is fundamental in providing timely support in relation to fertility challenges and symptom relief.

1. Introduction

Endometriosis is a chronic and benign gynecological condition characterized by the presence of endometrial-like tissue outside the uterine cavity [1]. It predominantly affects women of reproductive age [1]. The condition most commonly involves pelvic structures such as the

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ovaries and fallopian tubes. Extra-pelvic manifestations have been reported, including the bladder, kidneys, bowel, omentum, lymph nodes, lungs, pleura, abdominal wall, umbilicus, and rarely the brain and heart [2]. These cellular changes are associated with oxidative stress, inflammation and angiogenesis [3]. Concurrently, the formation of fibrotic tissue can occur.

Primary umbilical endometriosis is an uncommon form of extrapelvic endometriosis and occurs in the absence of prior abdominal surgery [4]. It accounts for approximately 0.5–1.0 % of all cases of extragenital endometriosis, making it exceedingly rare [5]. 6–10 % of reproductive-aged women is an estimate for worldwide cases of endometriosis; but most probably underdiagnosed [6]. Endometrial cells can spread via lymphatic channels or venous circulation, seeding distant sites, including the umbilicus, lungs, and pleura [7]. This mechanism explains cases of umbilical endometriosis occurring in patients without pelvic disease and supports why umbilical involvement sometimes presents as an isolated lesion [8].

The clinical presentation of endometriosis is variable. Common symptoms include abdominopelvic pain, dysmenorrhea, menorrhagia and infertility [9]. Cyclical umbilical pain and bleeding synchronized with the menstrual cycle is highly suggestive of umbilical endometriosis. It is suggested that the pathogenesis of umbilical endometriosis may involve hematogenous or lymphatic spread in cases concurrent with pelvic endometriosis, or metaplasia of urachal remnants in isolated cases [10].

Diagnosis is primarily clinical but histological confirmation is required for definitive diagnosis. Imaging modalities such as ultrasonography ± doppler, computed tomography (CT), and magnetic resonance imaging (MRI) may assist in preoperative evaluation [11,12]. Surgical excision remains the definitive treatment for umbilical endometriosis, while medical management, including analgesics and hormonal therapy, may offer temporary symptom relief but is non-curative [13]. Here, we report a rare case of primary umbilical endometriosis in a nulliparous woman with a 10-year history of infertility. The patient also presented with bilateral ovarian endometriomas, underscoring the complexity of this condition.

Currently, there are no case reports of primary umbilical endometriosis from limited settings in Sub-Saharan Africa such as Tanzania. This work has been reported in line with the SCARE criteria [14].

2. Case report

A 35-year-old nulliparous woman with a history of infertility presented to the outpatient clinic. She reported a three year history of cyclical dull pain at her umbilicus. The pain was associated with an umbilical nodule that bled during menstruation. Of note, the pain worsened during menstruation and was relieved by analgesics. The patient also reported dysmenorrhea, menorrhagia and dyspareunia. The patient had a copper IUD inserted 3 years prior and had been using Zinnia P (levonorgestrel and ethinylestradiol) for the last 3–4 months in order to try relieve the pain. Her past gynecological history was otherwise unremarkable and she had a regular menstrual cycle. There was no medical, surgical or social history of note.

On examination, her vital signs were stable. A hyperpigmented swelling (3 × 2 cm) at the umbilicus was noted on abdominal examination. The swelling was firm, immobile, non-tender, not reducible and was not tethered to underlying structures (Fig. 1A–B). An abdominal MRI revealed an ill-defined enhancing mass measuring 3 × 4 × 6 cm located along the right anterior abdominal wall and was connected to a sinus tract extending inferiorly to the suprapubic region but did not communicate with the peritoneal cavity, findings suggestive of endometriosis (Fig. 2A). The differential diagnosis was broad, including an umbilical hernia, granuloma, keloid scar, nodular malignant neoplasms and embryological abnormalities. Additionally, bilateral adnexal lesions were noted, they were hyperintense with focal hypointense areas and variable restrictions, the right side lesion measured 1.6 × 1.7 cm and left side lesion measured 2.0 × 0.8 cm, findings consistent with bilateral ovarian endometriomas (Fig. 2B).

The patient was scheduled for surgical local excision of the umbilical nodule. The specimen was sent for histopathological examination. The results revealed a normal epidermis with scattered endometrial glands and stroma within the dermis (Fig. 3A). Photomicroscopy highlighted the presence of endometrial stroma and subcutaneous fat tissue. This morphology is consistent with endometriosis (Fig. 3B). The patient's postoperative recovery was uneventful, and she was deemed fit for discharge on postoperative day 2. She received counseling regarding her future fertility and possibility of IVF, risk of recurrence and was scheduled for a follow-up visit in two weeks. She was commenced on Dienogest, Visanne and a combined oral contraceptive for the

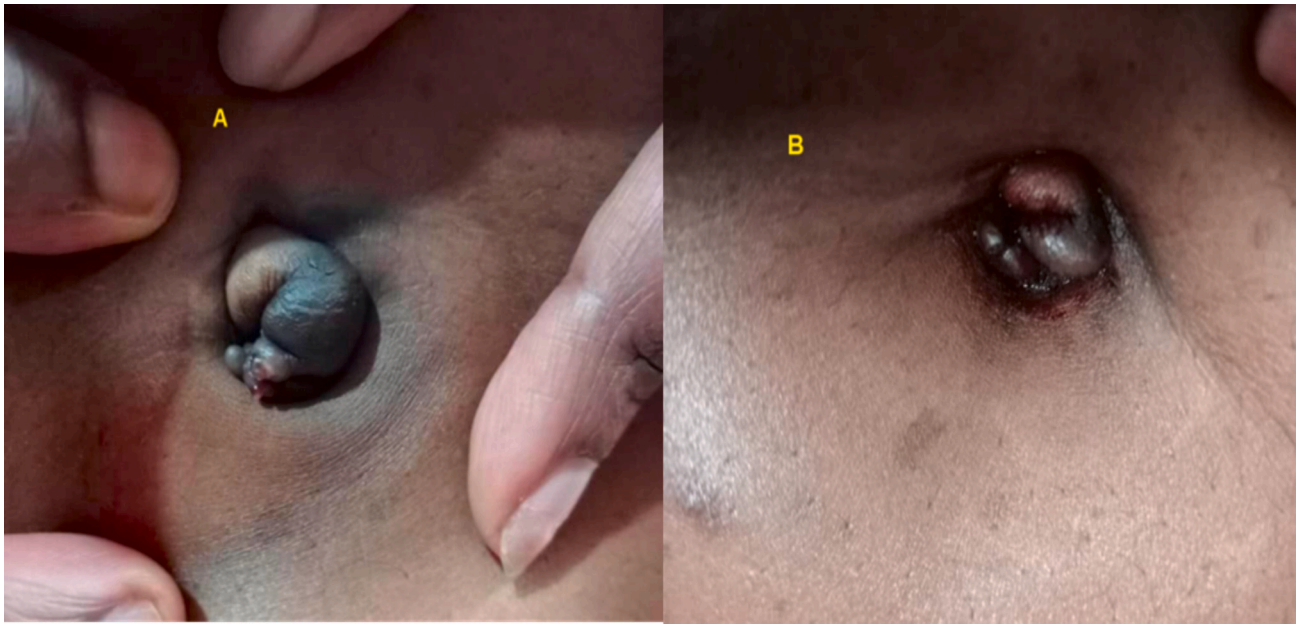

Fig. 1. (A) Hyperpigmented swelling at the umbilicus, measuring 3 × 2 cm firm, non-tender not reducible or mobile, and was not attached to the underlying structures or the overlying skin. (B) The appearance of umbilicus during menstruation.

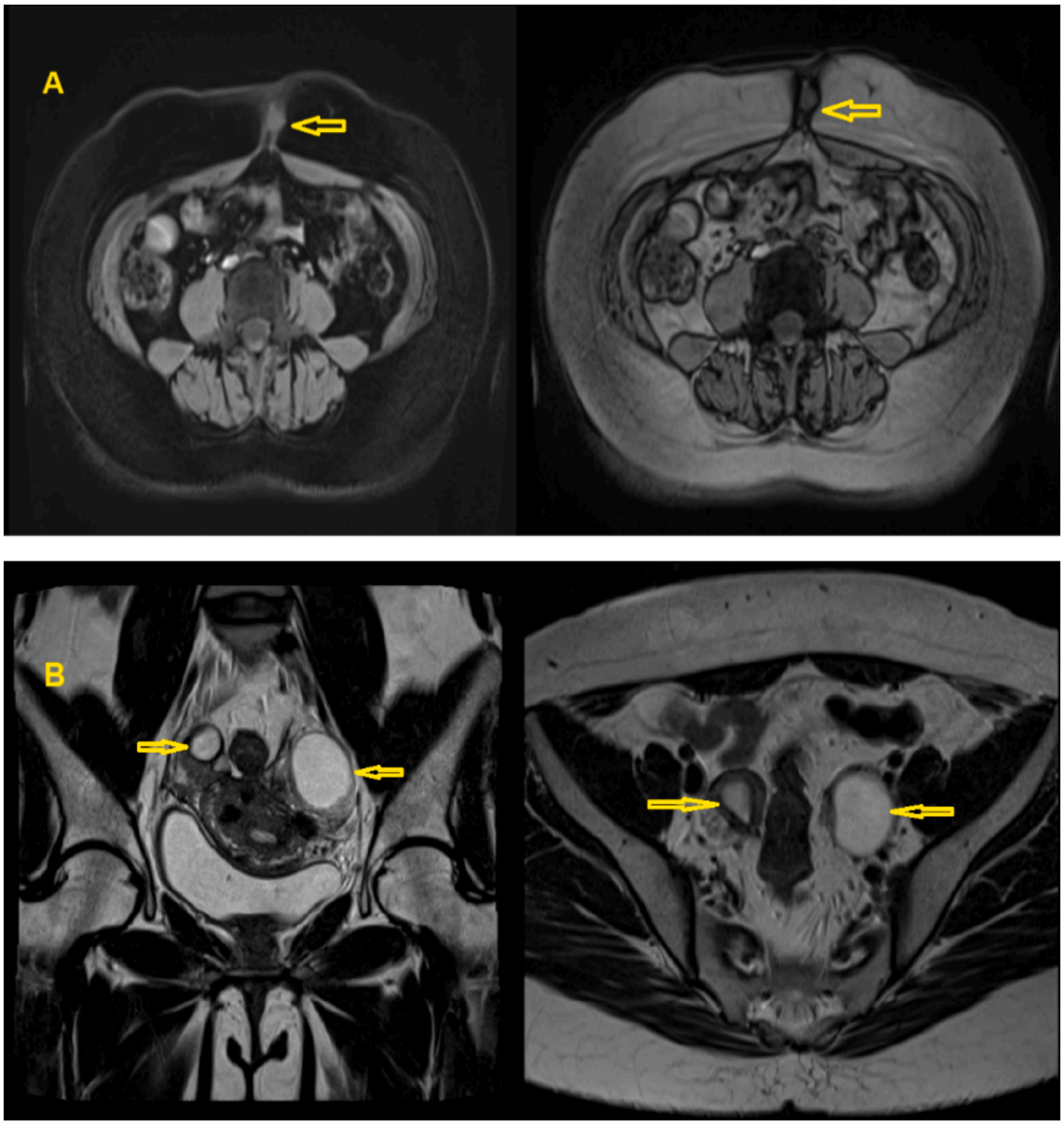

Fig. 2. A: An abdominal MRI (axial view) revealed an ill-defined enhancing mass (indicated by yellow arrow) measuring 3 × 4 × 6 cm located along the right anterior abdominal wall and was connected to a sinus tract extending inferiorly to the suprapubic region but did not communicate with the peritoneal cavity, findings suggestive of endometriosis.
B: An abdominal MRI (coronal and axial view) respectively, revealed bilateral adnexal lesions appeared hyperintense with focal hypointense areas and variable restrictions, right measures 1.62 × 1.70cm and left measures 2.0 × 0.8 cm, findings consistent with bilateral ovarian endometrioma.

management of endometriosis. She has been monitored clinically every four weeks, and no symptoms or signs of local recurrence have been observed during the 8-month follow-up period. Of note, despite her condition remaining stable following management, she did not conceive over the course of the following year.

3. Discussion

Here we present a rare case of primary umbilical endometriosis in a nulliparous woman This case report explores the rarity of this condition, particularly in Tanzania where no data on this specific condition currently exists, to our knowledge. There are two cases in the literature describing umbilical endometriosis, both cases refer to secondary umbilical endometriosis [15,16]. Our report also reviews the available literature on similar cases and discusses the challenges associated with managing umbilical endometriosis.

Umbilical endometriosis (UE), also known as Villar's nodule, was first described by Villar in 1886. The nodules are defined by the presence of endometrial glands and/or stroma within the umbilicus [1]. In the literature, primary umbilical endometriosis remains a rare entity, with few cases of spontaneous development and even fewer in nulliparous women. Primary umbilical endometriosis arises spontaneously without prior surgical intervention, whereas secondary umbilical endometriosis is associated with a history of surgical procedures, such as laparoscopy or open abdominal surgery [4]. Our patient had no history of surgery. The pathogenesis of umbilical endometriosis has been attributed to several mechanisms, including the translocation of endometrial cells through the peritoneal cavity, dissemination via lymphatic channels, or the involvement of embryonic remnants within the umbilical fold, such as the urachus or umbilical vessels. Additionally, genetic susceptibility and immunological abnormalities are considered contributing factors [4].

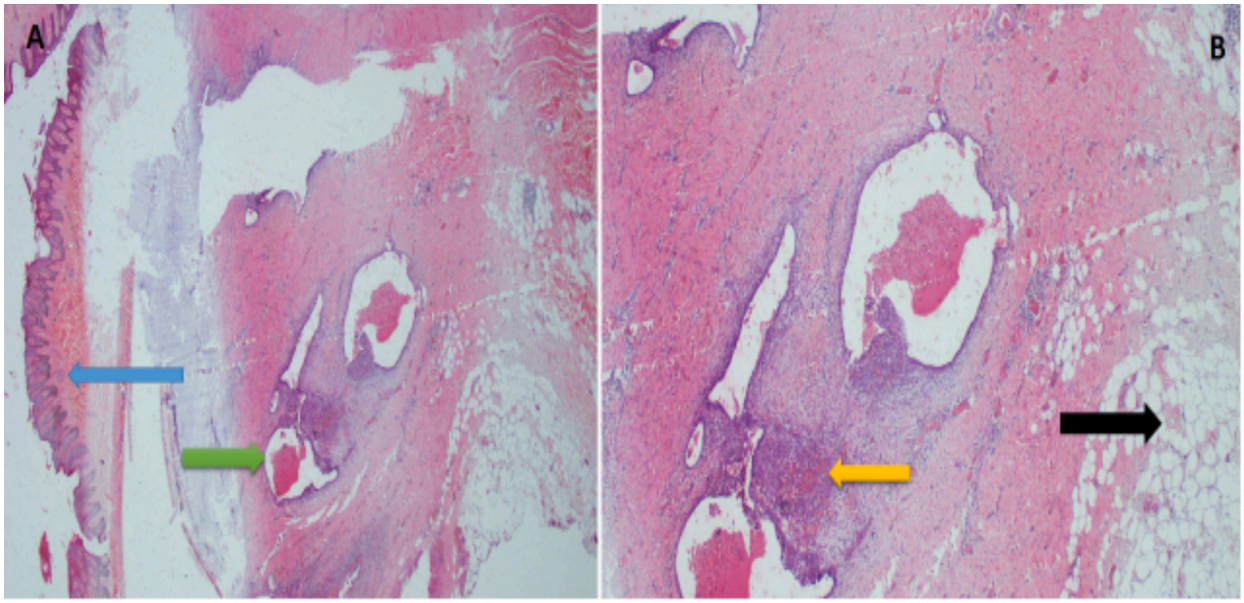

Fig. 3. A & B: Histopathology of cutaneous endometriosis demonstrating a normal epidermis (blue arrow) and scattered endometrial glands and stroma in the dermis, H&E staining at 20× original magnification (A); photomicroscopy highlighting endometrial stroma (yellow arrow), and subcutaneous fat tissue (black arrow), H&E staining at 40× original magnification (B).

The clinical presentation typically includes an umbilical swelling, observed in approximately 90 % of cases, often accompanied by cyclical pain in 82 % of cases and bleeding or discharge in 49 %. Our patient presented with an umbilical swelling, severe dysmenorrhea, dyspareunia, and heavy menstrual bleeding which correlates with endometriosis. However, some patients may remain asymptomatic [10,17]. Subfertility is a frequent complication of endometriosis, affecting up to 50 % of women with the condition [18]. In our case, the patient had been unable to conceive for 10 years, which may further support the theory that umbilical endometriosis could contribute to infertility. However, more research is required. Endometriosis is linked to infertility, though the exact mechanism underlying this association in the early stages of the disease remains unclear [19]. Umbilical endometriosis is sometimes associated with deep infiltrating endometriosis (DIE), which can involve the ovaries, fallopian tubes, or uterosacral ligaments, further worsening reproductive outcomes [20].

The differential diagnosis of umbilical endometriosis includes conditions such as an umbilical granuloma, umbilical polyp, hemangioma, melanocytic nevus, umbilical hernia, lipoma, pyogenic granuloma, hernia, urachal residual, pemphigus vegetans, and keloid, hypertrophic scars, and cutaneous metastases of malignancies [21,22].

Histopathological confirmation remains the gold standard for diagnosis. Various imaging modalities provide valuable information in confirming the diagnosis and ruling out the involvement of other organs [11,12,23]. In our case, imaging with MRI suggested our patient had primary umbilical endometriosis as well as bilateral ovarian endometriomas. An ovarian endometrioma is commonly detected in individuals affected by endometriosis as demonstrated in our patient by MRI.

In our case, the diagnosis of umbilical endometriosis was initially suspected based on clinical presentation and imaging findings, and subsequently confirmed through histopathological analysis. The histopathological analysis of the excised tissue confirmed the presence of endometrial-type glands scattered within the endometrial stroma beneath the skin, consistent with endometriosis.

A standardized approach to the management of primary umbilical endometriosis has not been established, largely due to the rarity of the condition [24]. Medical therapy, including combined oral contraceptives, progestins, or GnRH agonists, may relieve pain but does not improve fertility and is typically reserved for symptomatic relief [25]. However, these therapies do not halt disease progression, making symptom recurrence and lesion growth significant limitations of this approach [13,26]. In our case, the patient was placed on hormonal therapy following surgery. This approach may have positively contributed to a notable reduction in symptoms following surgery.

Surgical management remains the primary treatment of choice for umbilical endometriosis. This typically involves resection of the umbilical mass, often performed with the assistance of laparoscopy. Complete excision minimizes the risk of recurrence and enables the identification and treatment of any concurrent pelvic endometriosis [4,27]. Furthermore, it is advisable to conduct a pelvic laparoscopic exploration, when possible, to identify any additional sites of endometriosis, thereby complementing the surgical intervention. The risk of recurrence of umbilical endometriosis after surgical resection varies from 5.4 % to 27 % [28]. The patient should be advised on the possible local recurrence. Surgical intervention, particularly laparoscopic excision of endometriotic lesions and adhesiolysis, has been shown to enhance spontaneous conception rates, particularly in cases of stage I-II disease [29]. For moderate-to-severe cases (stage III-IV), assisted reproductive technologies (ART), such as in vitro fertilization (IVF), are often recommended due to improved success rates compared to expectant management [30].

With reference to ovarian endometriomas, clinicians are responsible for supporting patients to make an informed decision as to whether pursue conservative, medical or surgical management. Growing evidence suggests that excisional surgery may negatively affect ovarian reserve. As a result, recent trends have shifted towards more conservative management of ovarian endometriomas, likely leading to a decrease in the number of surgical referrals. [31,32]. In our case, we opted for a conservative approach for managing bilateral ovarian endometriomas in a symptomatic women with history of subfertility. However, hormonal treatments such as oral contraceptives and progestins are not recommended for women actively trying to conceive, as they suppress ovulation and alter endometrial function. These therapies are primarily used for symptom management and are only considered in specific cases, such as when symptomatic patients require temporary relief while awaiting IVF or surgical intervention [33].

4. Conclusion

We aim to provide valuable insights for clinicians who may encounter similar cases in the future especially in resource limited settings such as Tanzania and other areas of Sub-Saharan Africa. Fundamentally, we aim to provide data to confirm that primary umbilical endometriosis can occur spontaneously in patients with no prior surgical history. With this knowledge we hope clinicians will consider primary umbilical endometriosis as a differential diagnosis when they may not have done previously. We hope that this report will enhance awareness of this rare condition and offer a better understanding of its clinical presentation and management. The paucity of literature on umbilical endometriosis with bilateral ovarian endometrioma with a long standing history of infertility in a premenopausal woman demonstrates the importance of sharing similar cases in order to guide clinicians to achieve the best outcomes for patients.

CRediT authorship contribution statement

John Lugata: Data collection, patient management conceptualization, study design, initial manuscript version preparation and final manuscript draft approval.
Tecla Lyamuya: Data collection, patient management conceptualization, study design, initial manuscript version preparation and final manuscript draft approval.
Laetitia Makower: Data collection, patient management conceptualization, study design, initial manuscript version preparation and final manuscript draft approval.
Ibrahim Salum: Data collection, patient management conceptualization, study design, initial manuscript version preparation and final manuscript draft approval.
Rafiki Mjema: A lead obstetrician and gynaecologist, provided expertise throughout the entire process and revised and approved the final draft.
Alex Mremi: A lead pathologist, provided expertise throughout the entire process, performed histopathological analysis and approved the final manuscript draft.
Tom Kakumbi: A lead gynaecologist, fertility specialist and a researcher in endometriosis associated infertility provided expertise throughout the entire process and revised, approved the final draft.

Consent

Written informed consent for the publication of clinical details and images was obtained from the patient. A copy of the consent is available for review by the chief editor of this journal.

Ethical approval

This case report was approved by the authors' institution review board committee. The patient provided written informed consent to allow for her de identified medical information to be used in this publication. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Guarantor

Dr. John Lugata.

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Declaration of competing interest

All authors have declared that no competing interests exist.

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