Giant cystic degeneration of a uterine leiomyoma in a patient with autosomal dominant polycystic kidney disease

Katrin Arnoldsa,⁎, Emily Sendereya, Michael L. Spraguea, Rodolfo Blandonb, Diane L. Carlsson, Stephen Zimberga

aCleveland Clinic Florida, Department of Gynecology, Weston, FL, USA
bCleveland Clinic Florida, Department of Radiology, Weston, FL, USA
cCleveland Clinic Florida, Department of Pathology, Weston, FL, USA

Abstract

Objective: To report the management of a large uterine leiomyoma with diffuse cystic degeneration in a patient with autosomal dominant polycystic kidney disease (ADPKD).

Design: Case Report.

Setting: Cleveland Clinic Florida, Department of Gynecology, Section of Minimally Invasive Gynecologic surgery, Weston Florida.

Patient(s): A 52-year old woman with ADPKD with a large abdominal mass, abnormal uterine bleeding and symptomatic anemia. Imaging revealed a giant intramural cystic lesion of the uterus compressing the inferior vena cava.

Intervention(s): Uterine artery embolization and blood transfusion followed by a computed tomography guided cyst aspiration were performed on admission to alleviate anemia and abdominal pain and distension. Total laparoscopic hysterectomy with bilateral salpingectomy was performed in an outpatient setting.

Main Outcome Measure(s): Management of large cystic degeneration of leiomyoma.

Results: Normal recovery from definitive surgery. Surgical pathology confirmed a benign, cystically dilated leiomyoma.

Conclusion: This case demonstrates the management of giant intramural cyst lesion of the uterus using a minimally invasive surgical approach, as opposed to emergency surgery via laparotomy.

Capsule: Large uterine leiomyoma with diffuse cystic degeneration in a patient with autosomal dominant polycystic kidney disease, in which step-wise treatments allows successful minimally invasive hysterectomy.

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1. Introduction

Autosomal dominant polycystic kidney disease (ADPKD) is frequently associated with extrarenal manifestations such as hepatic or pancreatic cysts as well as the formation of cerebral aneurysms. The presence of uterine cysts as an extrarenal manifestation of ADPKD is extremely rare and has only been reported in two cases in the English literature [1,2]. We report a case of a patient with ADPKD and giant cystic degeneration of a uterine leiomyoma, the largest reported thus far.

2. Case Report

A 52-year-old Caucasian woman, diagnosed with ADPKD at age 28, presented to the emergency room with a large abdominal mass, approximately 30 weeks pregnancy size, abnormal uterine bleeding with menorrhagia, as well as symptomatic anemia. The patient reported a history of increasingly heavy menses over the past 2 months filling 9 pads a day. She had a history of one full term pregnancy, which resulted in vaginal delivery. On review of her old records, a 4 cm intramural cyst was described 12 years previously; but due to lack of insurance, the patient declined to have this further evaluated.

On physical examination, a tense, poorly mobile, mass was palpable on her abdomen extending 6 cm above the umbilicus, causing severe abdominal distention and pain. The patient was also experiencing active vaginal bleeding and reported weakness and fatigue.

Ultrasound evaluation of the pelvis revealed a large cystic mass with inability to visualize the uterus or the adnexa and computed tomography revealed a 20.3 cm intramural cystic lesion of the uterus consistent with cystic degeneration of a large intramural leiomyoma (Figs 1, 2).

Magnetic resonance imaging (MRI) confirmed the suspected central cystic degeneration of a leiomyoma measuring 19.6 cm leading to mass

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effect and compression of the inferior vena cava and proximal common iliac veins without thrombosis (Fig 3).

Multiple cysts were visualized in the kidneys and the liver consistent with the patient’s diagnosis of ADPKD.

The patient’s hemoglobin was noted to be 4.6 g/dl and she required blood transfusions to treat symptomatic anemia. Her heavy vaginal bleeding persisted and the patient underwent successful bilateral uterine artery embolization (Fig 4), which significantly slowed down her vaginal bleeding to 2 pads a day. Tumor markers were drawn due to the size of the mass and CA-125 was elevated at 173 U/ml.

In an effort to alleviate the patient’s painful abdominal distention, she underwent CT guided aspiration of 2.5 l fluid from the cyst (Fig 5), which significantly decreased the uterine size and symptoms. The aspirated fluid was clear and serous, negative for malignancy on cytology, and showed no evidence of infection. She was discharged in stable condition and was followed up in our clinic the following week, where endometrial biopsy and a PAP smear were collected and found to be negative for malignancy.

The patient desired definitive surgical management and underwent an uncomplicated total laparoscopic hysterectomy with bilateral salpingectomy as an outpatient with an uneventful recovery post.

Fig. 1. Computerized tomography scan shows coronal view of 20 × 13 × 17 cm cystic intramural mass and numerous hepatic cysts.

Fig. 2. Computed tomography and transverse view of intrauterine cystic degeneration of leiomyoma and numerous renal cysts.

Fig. 3. MRI sagittal view of complex cystic intramural mass suggesting cystic degeneration of a leiomyoma. Mass effect results in compression of the inferior vena cava and proximal common iliac veins without thrombosis.

Fig. 4. Angiogram prior to uterine artery embolization shows right uterine artery to be enlarged and tortuous. A large uterine mass is present.

Fig. 5. Computed tomography guided aspiration of cystic mass.

Image 1. 100× magnification demonstrating cystically dilated leiomyoma.
operatively. Surgical pathology revealed a 1478-g uterus with cystically
dilated leiomyoma (Images 1, 2) and myometrium with changes sec-
ondary to embolization (Image 3) as well as benign fallopian tubes
with paratubal serous cysts.

3. Discussion

ADPKD is a systemic disorder that primarily affects the kidneys, but
often presents with extra-renal manifestations, such as liver and pan-
creatic involvement. Cystogenesis is understood to be caused by a mu-
tation in one of two genes, which encode the membrane-spanning
proteins, polycystin-1 and polycystin-2 (PKD1 and PKD2) [3]. These
proteins work together to increase calcium entry uterine cysts as an
extra-renal manifestation [1,2].

Upon review of the patient’s records, she had been diagnosed with a
4 × 1 mm intrauterine fluid collection 12 years prior to her presentation.
She had been lost to follow-up and it is likely, that this uterine cyst was
present for years and gradually grew in size.

In this case, we were able to treat the patient’s symptomatic anemia
and stabilize the active vaginal bleeding with the help of uterine artery
embolization. Though not a treatment for the active disease process, the
percutaneous cyst aspiration by interventional radiology of 2.5 l allevi-
ated the patient’s abdominal bulk symptoms tremendously by shrink-
ing uterine size and relieved the pressure onto her great vessels,
which allowed for postponing of surgical intervention until her hemo-
globin was stable and planning intervention electively and in a mini-
mally invasive fashion, as opposed to emergency surgery at time of
admission in the face of heavy bleeding with multiple transfusions
and an operation requiring a large midline laparotomy.

Ultimately, we were able to achieve a minimally invasive approach
to hysterectomy by performing a laparoscopic hysterectomy after CT
guided cyst aspiration, whereas without this intervention she would
have required a large, midline laparotomy with incision from the
pubis to subxiphoid area. Complications related to giant leiomyomas in-
clude infection, necrosis, rupture, hemorrhage, deep vein thrombosis
and hydronephrosis due to mass effect [5]. The most common misdiag-
nosis could be cystic adenomyosis, hematometra, uterine sarcoma and
adnexal masses.

Although a rare entity, we would like to raise the awareness of uter-
ine cysts being a possible extrarenal manifestation in ADPKD patients.
Larger trials need to be conducted to prove a definitive connection be-
tween cystically dilated uterine cysts and PKD1 and PKD2 mutations
in ADPKD patients. We describe interventions, which avoided a large,
midline laparotomy and allowed an outpatient minimally invasive sur-
gical solution.

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