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Case Report

Abdominal Leiomyoma in an Adult Transgender with Adrenogenital Syndrome: A Rare Case Report

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ABSTRACT

Leiomyomas of the abdomen are rare intra-abdominal tumors, particularly in male patients with adrenal hyperplasia. We report the case of a 50-year-old considers himself a with a man, who was born as a female, and has several surgeries related to transitioning from intersex to male gender. These procedures include bilateral oophorectomy with silicone testicular implantation, bilateral mastectomy with closure of the labial cleft, reconstruction of the urethral canal, and placement of testicular implants. He has a wife and two adopted children. The patient presented to the emergency department suffering from lower abdominal pain and vomiting. On examination, the abdomen appeared distended and soft. In addition to positive bowel sounds, there is a palpable large, firm, slightly tender mass occupying the lower abdomen and extending above the umbilicus, a small penis, and bilateral suspected testes in the proximal inguinal region. Abdominal radiographs, abdominal ultrasounds, and computer tomography (CT) of the abdomen and pelvis with intravenous contrast revealed a large pelvic-abdominal mass, bilaterally enlarged adrenal glands, and bilaterally implanted tests seen in the inguinal region. An ultrasound-guided tissue biopsy and an exploratory laparotomy were performed, and the tumor was completely resected. A rounded, firm mass arises from the pelvis, with histopathology revealing its origin from the remnants of the uterus and vaginal adhesions adherent to the mesentery. A tissue biopsy confirmed the leiomyoma

Introduction

Congenital adrenal hyperplasia (CAH) is a group of autosomal recessive diseases that impair the steroidogenesis of the adrenal glands. The 21-hydroxylase gene, located at chromosome 6p21, has mutations that lead to 21-hydroxylase deficiency, which is the most prevalent variant (1).

The carrier frequency for classical CAH is around 1 in 60 people (2,3). The reported incidence of classical 21-hydroxylase deficiency varies by ethnic/racial origin and ranges from 1 in 5000 to 1 in 15,000 (4,5). The presentations range from hirsutism and irregular menstruation in adults to newborn salt waste and unusual genitalia. In many nations, newborns with elevated 17-hydroxyprogesterone levels are screened

Table 1: Initial laboratory investigations.

Investigation	Result	References value
WBC	25.7 x10^3/uL	4.0-10
RBC	4.6 x10^6/uL	4.5-5.5
Hgb	13.8 gm/dL	13-17
Hct	41.6 %	40-50
MCV	90.0 fL	83-101
MCH	29.9 pg	27-32
MCHC	33.2 gm/dL	31.5-34.5
RDW-CV	12.5 %	11.6-14.5
Platelets	192 x10^3/uL	150-400
MPV	12.0 fL (H)	7.4-10.4
PDW	15.5 fL (H)	9.4-10.6
Absolute Neutrophil Count #	22.4 x10^3/uL (H)	2-7
Lymphocyte #	1.5 x10^3/uL	1-3
Monocyte #	1.7 x10^3/uL (H)	0.2-1
Eosinophil #	0.0 x10^3/uL	0-0.5
Basophil #	0.08 x10^3/uL	0.02-0.10
Neutrophil %	87.2 % (H)	54%-62%
Lymphocyte %	5.9 %	25%-33%
Monocyte %	6.5 %	3%-7%
Eosinophil %	0.1 %	1%-3%
Basophil %	0.3 %	0%-0.75%
Urea	9.2 mmol/L (H)	2.8-8.1
Creatinine	161 umol/L (H)	62-106
eGFR	>60 mL/min	
Sodium	133 mmol/L (L)	136-145
Potassium	4.7 mmol/L	3.5-5.1
Chloride	98 mmol/L	98-107
Bicarbonate	21 mmol/L (L)	22-29
Calcium	2.28 mmol/L	2.15-2.50
Adjusted calcium	2.44 mmol/L	2.15-2.50
Phosphorus	0.81 mmol/L	0.81-1.45
Magnesium	0.55 mmol/L (L)	0.66-1.07
Bilirubin T	20 umol/L	0-21
Total protein	65 gm/L (L)	66-87
Albumin	32 gm/L (L)	35-52
Uric acid	324 umol/L	200-430
Alkaline phosphatase	52 gm/L	40-129
ALT	14 U/L	0-41
AST	14 U/L	0-40
LDH	295 U/L	125-220
GGT	32 U/L	10-71
Troponin-T HS	8 ng/L	3-15
Iron	21 umol/L	6-35
TIBC	83 umol/L (H)	45-80
Trans ferritin	3.3 gm/L	2-3.6
Fe% saturation	25 %	15-45
B-Hydroxybut	0.30 mmol/L	0-0.60
Lipase	39 U/L	8-78
Glucose	812.4 mmol/L (H)	3.3-5.5
HbA1C %	6.3	4.8-6
CRP	326.4 mg/L (H)	0-5
Lactic acid	2.6 mmol/L	0.5-2.2
Procalcitonin	7.87 ng/mL	< 0.5 ng/mL
Prothrombin Time	13.8 seconds (H)	9.7-11.8
INR	1.3	
APTT	33.2 seconds (H)	24.6-31.2
CA 19-9	12.5 U/mL	0-27
CEA	1.5 ug/L	3.8- 5.0 ug/L
AFP	4	0-6

for the most common type of congenital adrenal hyperplasia, classic (severe) 21-hydroxylase deficiency. However, cosyntropin stimulation testing may be required to confirm the diagnosis or identify non-classic (milder) subtypes (4). Patients with no medical compliance are more likely to suffer the late consequences associated with untreated CAH. High levels of androgen and estrogen will stimulate the growth of estrogen-dependent organs, like Leiomyomas. We present a patient with a large abdomen leiomyoma and bilateral adrenal hyperplasia due to longstanding treatment noncompliance. He had ambiguous genitalia and was raised as a male according to the medical record of the patient outside our institute.

Case Presentation

A 50-year-old patient with ambiguous genitalia presented to the emergency department after experiencing lower abdominal pain and vomiting for one week. The pain was scored at 9/10. The pain had no aggravating or relieving factors. No history of fever, nausea, significant weight loss, or anorexia. His vitals were as follows: body temperature: 36.8 °C, heart rate: 66 beats per minute, respiratory rate: 17 per minute, blood pressure: 122/72 mm Hg, a saturation of peripheral oxygen: 99%>. The Physical examination revealed lower abdominal tenderness with guarding, a soft distended abdomen, positive bowel sounds, a palpable large firm non-tender mass in the lower abdomen extended above the umbilicus, a small penis, and bilateral undescended testicle with ambiguous genitalia. The patient was given an intravenous paracetamol infusion, which did not relieve the abdominal pain. The laboratory tests revealed elevated white blood cells, absolute neutrophil count, monocyte, PT and PTT, urea and creatinine, c-reactive protein, and glucose. Low sodium, albumin, total protein, and bicarbonate were found. Testicular tumor markers were normal as shown in Table 1. His past surgical history includes the removal of both ovaries and silicone testicular implantation at the age of 14. At the age of 15, he underwent excision of both breasts with closure of the labial cleft using multiple Z-plasty techniques, as well as reconstruction of the urethral canal and placement of a testicular implant. According to the patient, he was diagnosed with congenital adrenal hyperplasia during childhood through laboratory tests but did not undergo any genetic testing.

An abdominal radiograph was done which showed a large soft tissue shadow occupying most of the abdomino-pelvic region, push the bowel loop peripherally (Figure 1).

Ultrasound of the abdomen and pelvis was taken, which demonstrated a large, hard, soft tissue mass of heterogeneous echo pattern at the mid-abdomen extending to the lower abdomen. The mass was causing slight compression of the posterior abdominal organs and vessels, along with deviation of the mid-abdominal organs laterally. It was difficult to determine organ of origin. Measuring 15 x 16.5 x 9.9 cm (1277 cc in volume) with feeding vessels seen at the periphery. No clear cystic changes were seen. (Figure 2).

The patient was treated conservatively and a Follow-up ultrasound of the abdomen after 1 week showed a change in the texture of the mass, which became more hypoechoic with vascularity on Doppler, suggestive of necrotic changes (Figure 3).

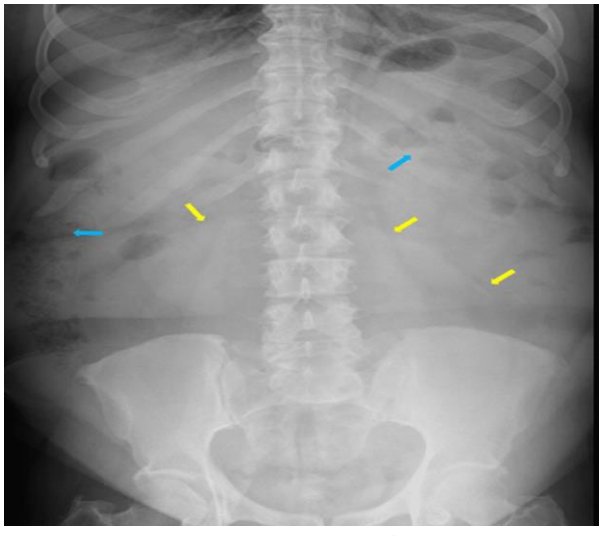

Figure 1: Abdominal radiograph showing a large soft tissue shadow occupying most of the pelvi-abdominal region (Yellow arrows) with displaced bowel loops peripherally (Blue arrows).

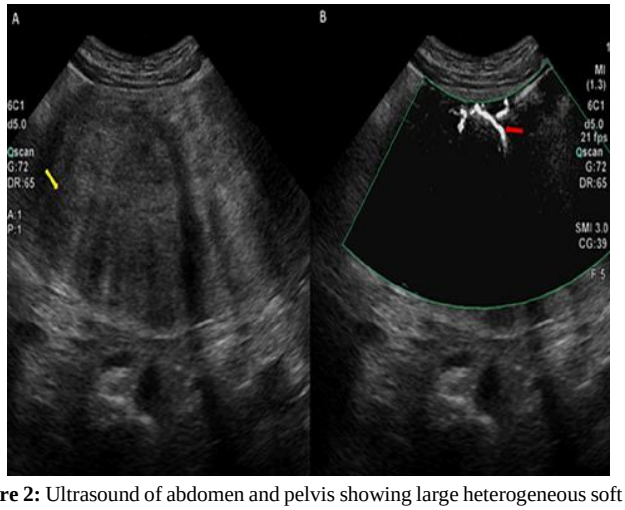

Figure 2: Ultrasound of abdomen and pelvis showing large heterogeneous soft tissue mass occupying the superior epigastric and mid abdomen extending to the lower abdomen (Yellow arrow) with feeding vessel seen in the periphery in super microvascular imaging technique (Red arrow).

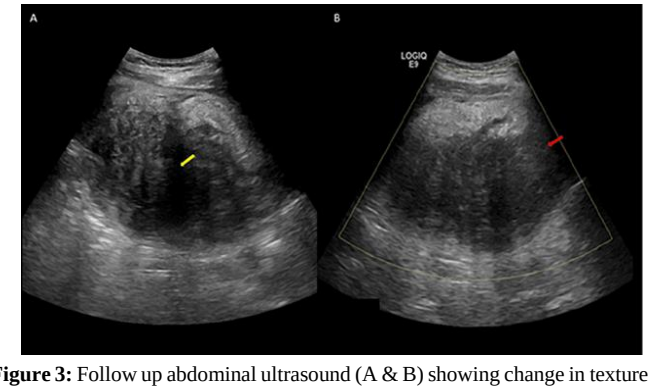

Figure 3: Follow up abdominal ultrasound (A & B) showing change in texture of the mass, which became more hypoechoic (Yellow arrow) with a vascularity by Doppler study (Red arrow), indicating necrotic changes.

CT abdomen and pelvis with contrast was done for further characterization of the lesion, which showed a large heterogeneous well-defined solid soft tissue, intrabdominal mass extending from the right lower quadrant to the mid abdomen, measuring 15.1 x 11.7 x 16.5 cm, which had no significant contrast enhancement, causing mass effect displaying the adjacent bowel loops. Small and large bowls were unremarkable. Both adrenal glands were enlarged and nodular, the largest nodule was seen in the left adrenal gland measuring 2cm. Both implanted testes with peripheral calcification and were seen at the lower part of the both inguinal region (Figure 4 and 5).

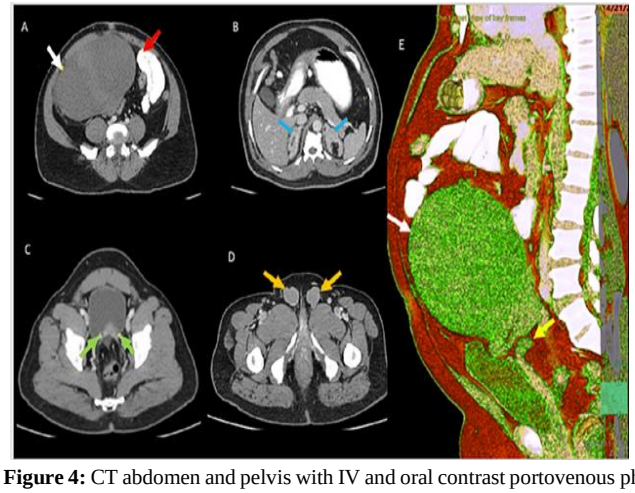

Figure 4: CT abdomen and pelvis with IV and oral contrast portovenous phase A and E. Axial and E Sagittal cuts color-coded 3D rendering demonstrating a large heterogeneous enhanced right paramedian abdominal soft tissue mass (white arrows), displacing the bowel loops peripherally (red arrow). The mass is connected to the retro-vesical plate-like soft tissue density by a twisted pedicle (yellow arrow). Soft tissue enhancing structure seen posterior to the urinary bladder (Green arrows) and 7 bilateral implanted testes (Orange arrows).

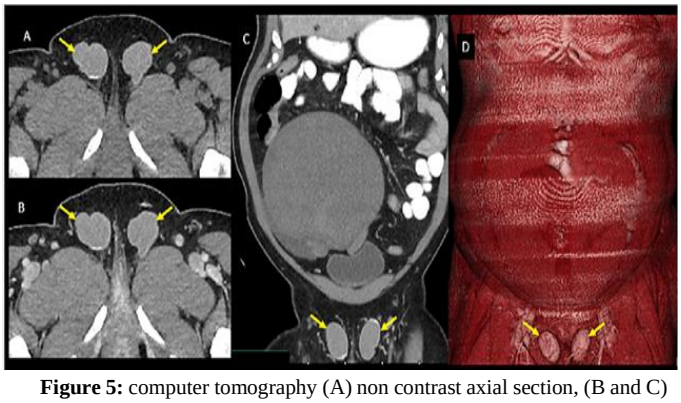

Figure 5: computer tomography (A) non contrast axial section, (B and C) post contrast, porto-venous, axial and coronal (D) 3D color coded coronal shows Bilateral implanted testis with homogenous CT density about 70 Hounsfield in pre and post contrast (yellow arrows).

Ultrasound-guided intra-abdominal lesion biopsy was done, and the core biopsy report was smooth muscle neoplasm. It was difficult to categorize the mass as benign versus malignant based on the material received, therefore excision was recommended. He underwent an exploratory laparotomy. A large, rounded firm mass arising from the pelvis and the rudimentary uterus and vagina were seen. There were adhesions with the mesentery and the bowel also. The mass along with the appendix and the residue of the uterus and vagina were removed.

Grossly the mass was a well-circumscribed sphere-like tumor measuring 18 cm in maximum dimension characterized by a smooth tan outer surface and red-maroon hemorrhagic soft cut surface with a focal whorled appearance (Figure 6).

Figure 6: Gross specimen of the tumor showing tan and glistening on the surface. B) Cut surface of the mass demonstrating hemorrhagic red-maroon areas.

Light microscopic examination revealed smooth muscle tumor consisting of intersecting bundles of necrotic smooth muscle cells with bland nuclear features. the tube- like structure measuring 2×2×1 cm which was adherent to the tumor on light microscopic examination appeared to be a primitive genital tract that has a lumen surrounded by a muscular coat. The lumen consisted of a branched epithelial lining that resemble an endocervix characterized by tall columnar cells with basal nuclei and basophilic abundant cytoplasm. The lumen was surrounded by intersecting bundles of smooth muscle fibers forming a muscle coat (Figure 7).

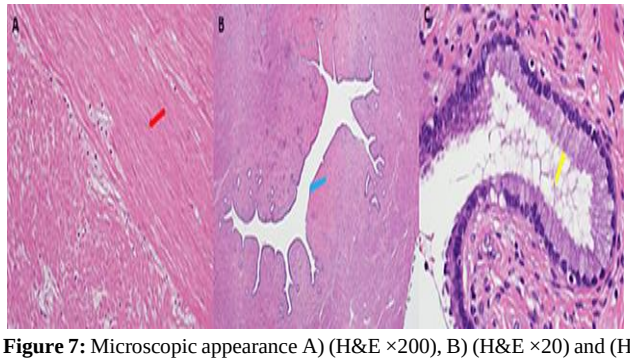

Figure 7: Microscopic appearance A) (H&E ×200), B) (H&E ×20) and (H&E ×400), showing leiomyoma with intersecting bundles of necrotic smooth muscle cells (Red arrow), tubelike structure compatible with primitive genital tract (Blue arrow) and lumen showing endocervical like epithelium (Yellow arrow).

Post-operative days were uneventful, and patient became symptomatically better and was discharged home with surgery outpatient follow up.

Discussion

Congenital adrenal hyperplasia (CAH) is a rare group of inherited autosomal recessive disorders characterized by a deficiency in mineralocorticoids and glucocorticoids (1). The most common type is caused by a 21-hydroxylase deficiency in 95% of cases. CAH can be further divided into salt wasting, simple virilizing, and non-classical forms. The most severe form is salt-wasting CAH, which presents early hypovolemia, adrenal insufficiency, and hyperandrogenism. Patients who have sufficient mineralocorticoids may present with the less severe form, simple virilizing CAH, but they can still have severe hyperandrogenism. Non-classical CAH is the mildest form with varying presentations from asymptomatic to mild hyperandrogenism. (2). The majority of classic cases are recognized and treated in infancy since CAH is now a part of newborn screenings (3).

Diagnosis is based on clinical assessment, genotyping, hormonal sex determination, genetic sex, and an evaluation of the patient's potential for future sexual engagement and fertility in an infant born with ambiguous genitalia. The use of 17 hydroxyprogesterone levels in newborn screening for CAH is a helpful method for identifying the condition early before the affected infant has an adrenal crisis. As levels are influenced by preterm and birth weight, screening is connected to a high likelihood of false positive findings, nevertheless. In the newborn screening program, molecular genetics, particularly the genotyping of the CYP21A2 gene, should be regarded as a secondary screening test (4).

Adrenal insufficiency, infertility-related problems, and adrenal tumors can all result from inadequate therapy (5). High amounts of adrenal androgens can cause a variety of unfavorable gynecologic issues in patients with uncontrolled CAH. Untreated CAH patients have dramatically raised levels of both hormones because androgens are converted to estrogens. High amounts of androgen and estrogen promote the growth of organs that depend on estrogen, which results in the emergence and development of uterine leiomyoma (1).

In our case, the 50-year-old patient was born with ambiguous genitalia, and the decision was to raise as a male. And was diagnosed with CAH shortly after birth. The specific enzyme deficiency is unknown, and no records were available for review. The patient does not recall any hormonal treatment received in our hospital's record. And underwent bilateral ovarian removal at the age of 14 and a bilateral mastectomy at the age of 15. The patient refused to undergo any hormonal workup and was only treated as an acute case at the emergency correlation with hormonal assessment was not provided.

In our case, the old medical record when he was diagnosed as CAH reflects that our patient was raised as male and most of the 46, XX CAH cases raised male were diagnosed after two years of age. In these cases, hysterectomy and bilateral salpingo-oopherectomy, genital corrective surgeries, and testicular prosthesis operations were performed in a very wide age range (6).

Leiomyomas have been associated with high estrogen levels, although there is inconsistent and limited information on the effects of both estrogen and testosterone on the uterus. According to earlier studies, testosterone may even decrease uterine growth in vitro rather than stimulating it (7). The formation of uterine Leiomyomas was shown to be more likely in women with greater testosterone levels in research by Wong et al (8). That examined the effects of both hormones on the development of Leiomyomas. In addition, CAH cases with raised levels of both testosterone and estrogen were at a greater risk of developing leiomyomas than those with elevated testosterone levels alone.

In some cases, abdominal leiomyomas often present a diagnostic challenge due to the unusual locations they arise from. These fibroids have been associated with previous morcellated hysterectomies or myomectomies in the literature (9). Deep soft tissue leiomyoma is a very rare smooth muscle tumor that is now generally thought to be divided into two categories based on the occurrence site; both clinical and pathological features, as well as aspects of the histological diagnosis criteria, were significantly different (10).

Abdomen/retro peritoneum leiomyoma almost always occurs in women, and we report a case of abdomen/retro peritoneum leiomyoma. Large leiomyomas occur in men and are reviewed in the literature. The cornerstones of CAH treatment are glucocorticoids and mineralocorticoids, intending to provide enough replacement while lowering ACTH and adrenal androgens (1). Treatment in adults with CAH is a bit tricky as some patients do not want treatment due to social stigma. Our patient denied hormone studies and hormone replacement therapy and no previous treatment records were available. A persistently elevated ACTH in undertreated or untreated CAH can present with persistently high ACTH, which not only causes adrenal hyperplasia but also other adrenal masses (11). In the literature review, there are only 4 reported cases of Abdominal leiomyomas in patients with CAH (11,12). Surgery should be considered in patients with fibroids having constant abdominal pain, unrelieved by conservative management. Our patient underwent exploratory laparotomy and found a mass arising from the pelvis, rudimentary uterus, and vagina; and histopathology confirmed the leiomyoma.

Conclusion

Untreated CAH can present with gynecological complications like fibroids and rarely extra uterine fibroids. Patients may not reveal their medical history if there are no previous medical records available, as was the situation in our case, where all previous clinical follow-up was performed outside of our country and non-compliant with hormone therapy. Knowing the clinical and radiological findings can help steer the clinician toward timely, appropriate management and to avoid potentially harmful treatment.

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Conflict of Interest

Authors declare no conflict of interest.

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