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CASE REPORT

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Esophageal Involvement and Bleeding in Bullous Pemphigoid Disease

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ABSTRACT

Bullous pemphigoid (BP) is the most common autoimmune bullous disorder and is characterized by autoantibodies against hemidesmosomal proteins in the skin and mucous membranes. BP typically presents as large, fluid-filled blisters on normal skin or red, enflamed areas of skin, mainly in the armpits, lower abdomen, inner thighs, and groin. Blisters on the lining of the oral and pharyngeal mucosa occur in up to 35% of cases, but blisters in the esophagus are rare, especially when there are no oral blisters. We report a case of newly diagnosed BP in a 48-year-old woman. She was admitted to our hospital with the sudden onset of hematemesis. This case highlights the importance of gastroenterologists who are cognizant of the potential association between skin illnesses and digestive disorders. It is important to exercise caution during endoscopic procedures in patients with pemphigoid disorders, especially in the absence of apparent symptoms.

Keywords: Bullous pemphigoid, hematemesis, endoscopy

INTRODUCTION

Bullous pemphigoid (BP) is the most common autoimmune bullous disorder and is characterized by autoantibodies against hemidesmosomal proteins in the skin and mucous membranes [1]. The incidence of BP, which is more common in older adults, is 2.5-42.8 cases per million per year, and its occurrence is gradually increasing due to longer life expectancy [2]. BP typically presents as large, fluid-filled blisters on normal skin or red, enflamed areas of skin, mainly in the armpits, lower abdomen, inner thighs, and groin. Blisters on the lining of the oral and pharyngeal mucosa occur in up to 35% of cases; however, blisters in the esophagus are rare, especially when there are no oral blisters. Patients with esophageal involvement may or may not have symptoms such as chest pain, difficulty swallowing, and pain during swallowing. Upper endoscopy may reveal blisters and necrosis in the esophagus [3].

CASE PRESENTATION

We report a case of newly diagnosed BP in a 48-year-old woman with a medical history of hypertension. Five months prior, she had been taking a penicillin group antibiotic for a dental enfection, after which bullous lesions appeared on her body. A skin biopsy confirmed the diagnosis of BP, and prednisolone at 1 mg/kg was started. During follow-up, the lesions regressed with oral prednisolone therapy. However, two months ago, she experienced an ischemic cerebrovascular event due to thrombosis in the truncus brachiocephalicus, resulting in left hemiplegia. Consequently, while continuing prednisolone treatment, additional acetylsalicylic acid therapy was initiated.

The patient was admitted to our hospital with the sudden onset of hematemesis. Urgent upper endoscopy revealed multiple blister lesions in the hypopharynx, a significant bleeding esophageal hematoma, and active esophageal bleeding (Figure 1). The procedure was completed by applying an Ankaferd blood stopper to the mucosal lesions where leakage-type bleeding was observed. In a follow-up endoscopy performed under sedation after two days of restricted oral intake and proton pump inhibitor infusion, it was noted that the bleeding in the mucosal lesions had ceased, and the edema had subsided (Figure 2). The patient, whose bleeding was controlled, was started on azathioprine treatment, as recommended by the dermatology department.

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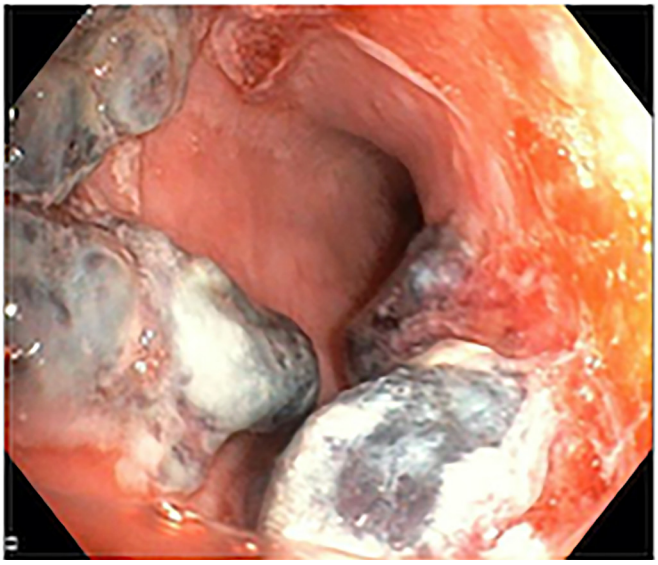

Figure 1. Esophageal hemotamas

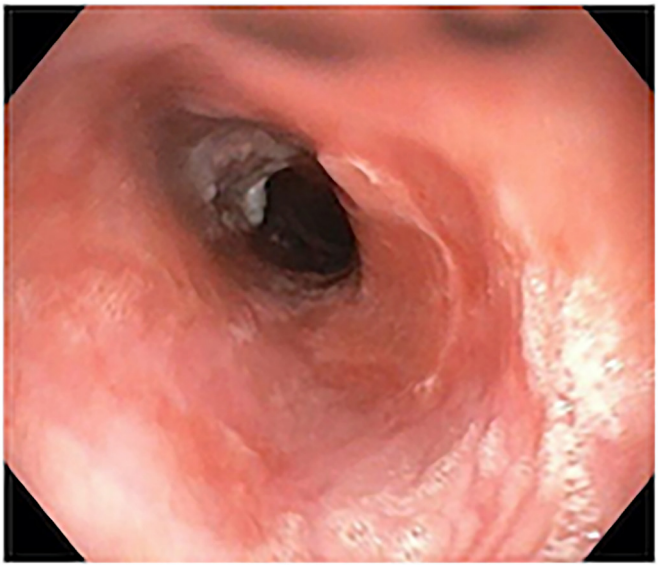

Figure 2. The esophagus after treatment

DISCUSSION

BP is the most common autoimmune disease causing blisters on the skin. It is becoming more common in older adults. BP typically presents as itchy, inflamed skin covered with blisters. However, the appearance of the blisters can vary greatly, and sometimes there are no blisters. Therefore, BP should be considered in any older adult with itchy, enflamed skin [4].

This is a rare case of a patient with a unique gastrointestinal symptom related to skin disease. Pemphigoid disorders, including BP and mucous membrane pemphigoid, are a very rare group of autoimmune skin diseases. These lesions are caused by antibodies that attack the basement membrane of the squamous epithelium, triggering enflammation.

It is noteworthy that the gastrointestinal symptoms exhibited by the patient were not consistent with the observations made during the endoscopic examination. The patient did not exhibit any gastrointestinal problems. Performing esophagogastroduodenoscopy can be challenging in such situations, as the esophagus has the potential to develop blisters and undergo sloughing even with minimal contact from the endoscope. It is advisable to employ a cautious and deliberate approach when manipulating the endoscope to minimize the risk of hemorrhage, lacerations, and perforation. The primary objective of treatment is to effectively manage the underlying immunological disorders. Systemic corticosteroids are commonly used as the initial therapeutic approach to induce remission. However, in cases of extensive gastrointestinal bleeding resulting in hemodynamic instability, therapeutic endoscopy may be used as an intervention to halt the bleeding.

This case underscores the importance of gastroenterologists who are aware of the possible association between skin diseases and gastrointestinal symptoms. Caution should be exercised during endoscopic procedures in patients with pemphigoid disorders, even in the absence of overt symptoms.

Ethics

Informed Consent: Written informed consent was obtained from the patient before procedure.

Footnotes

Authorship Contributions

Surgical and Medical Practices: D.A., Concept: D.A., Design: M.C.Y., Data Collection or Processing: K.K., Analysis or Interpretation: D.A., Literature Search: K.K., M.C.Y., Writing: K.K., D.A.

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CASE REPORT

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Machado-Joseph Disease, A Case Report of Treatment Based on Phototherapy

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ABSTRACT

Machado-Joseph disease (MJD) is a rare autosomal dominant disease caused by a mutation in exon 10 of the *ATXN3* gene resulting from a cytosine-adenine-guanine trinucleotide repeat. A case of a 48-year-old man with MJD is reported. His father, two paternal aunts, and his older sister all died because of this disease, and his younger brother had the same disease. Due to the absence of therapeutic options, phototherapy sessions were offered as an alternative between 425 and 650 nm 1.33 Joules/cm^2, 30 cm above the chest. After three months of phototherapy sessions, the following items showed a decrease in their scores: physical functioning, role limitations due to physical health, role limitations due to emotional problems, energy/fatigue, emotional well-being, social functioning, and general health. The only variable that remained unchanged was pain. He discontinued treatment, attributed the deterioration to phototherapy. A year later, as the disease progressed, he decided to resume the same scheme, and the following trends were observed: a) Improvement in role limitations due to physical health, emotional well-being, social functioning, and general health; b) No changes in role limitations due to emotional problems and pain and c) deterioration in energy/fatigue.

Keywords: Palliative care, phototherapy, spinocerebellar ataxia type 3

INTRODUCTION

Machado-Joseph disease (MJD), also known as spinocerebellar ataxia type 3 (SCA3), is a hereditary and multisystem neurodegenerative disease belonging to the group of spinocerebellar ataxias, which are diseases characterized by degeneration of the pyramidal, extrapyramidal, oculomotor, and cerebellar systems, in addition to motoneurons [1]. Approximately 40 subtypes of ataxias have been classified, of which SCA3 is the most common subtype globally [2]. Clinically, it is found to be heterogeneous even among family members (peripheral neuropathy, dystonia, ophthalmoplegia, parkinsonism, cerebellar ataxia, non-motor manifestations, cognitive impairment, sleep disorders, olfactory dysfunction, and psychiatric symptoms), but progressive in all [3]. The first neurological involvement occurs at the level of coordination of extremities, control of voluntary movements, speech, walking, and swallowing. Moreover, with the course of the disease, clinical findings in the pyramidal, extrapyramidal, peripheral, and cranial nerves will be noted, reducing the life expectancy to approximately 21 years from the beginning of the clinical picture. Although this disease has a variable onset and can begin in adolescence to old age, its onset is commonly observed in adulthood [4].

MJD was first classified by neurologist Coutinho and Sequeiros [5] among Machado-Joseph families who were inhabitants of the Azores islands. This autosomal dominant disease is a rare genetic pathology that is caused by a mutation in exon 10 of the *ATXN3* gene due to the repeat of a cytosine-adenine-guanine (CAG) trinucleotide [6] located on chromosome 14q32.1, which is a gene that, in the absence of its correct functioning, leads to the translation of an abnormal expansion of polyglutamine (polyQ) in the gene product (ataxin-3) and incorrect protein folding, resulting in alterations in different cellular processes and neuronal dysfunction and death. After the toxic aggregation of the protein, ataxin-3 with the abnormal expansion of the polyQ sequence neuronal inclusions is aggregated, promoting neuronal toxicity and degeneration [7].

The accumulation of toxic ataxin 3 comprises a C-terminal region and domain where an abnormal expansion of the polyglutamine tract occurs. This protein, in turn, binds to the specific intracellular calcium channel InsP3R1, thereby increasing its release. Excess intracellular calcium can kill cells by inducing

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cytotoxic processes, such as oxidative stress, mitochondrial permeability, and calpain activation [8]. Furthermore, loss of neurons occurs in the neostriatum and cerebellar cortex, whereas gliosis occurs in the spinal and cranial motor nuclei, substantia nigra, and cerebellar dentate nucleus. The presence of these aggregates in axons can negatively affect axonal transport mechanisms and result in neuronal degeneration [9]. Pathological ataxin-3 tends to aggregate as part of its natural function [10].

Unfortunately, to date, no specific cure for this pathology has been established, and this has urged doctors to seek alternative treatments, such as phototherapy, which has shown favorable treatment outcomes for other diseases of the central nervous system [11].

CASE PRESENTATION

The study investigated a 48-year-old man with MJD. His father, two paternal aunts, and older sister died from this disease. His younger brother also has the same affection. In this case, the disease manifested when the patient was 37 years old and compromised the mobility of his left ankle. Subsequently, several symptoms and signs of the disease progressively manifest in the patient (Table 1). Due to the absence of therapeutic options, phototherapy sessions were offered.

With the patient in the supine position, the phototherapy lamp (Federal Ministry of Health registration number: 1694E95) was turned on within a range between 425 and 650 nm, 11.33 Joules/cm^2, and 30 cm above the chest, following the next scheme: (a) 30-min daily sessions from Monday to Friday. The reason for the lamp's position above the thorax is that blood is a highly conductive material, and the lamp near the chest may increase the likelihood of energy reaching the aorta where the greatest systemic effect would occur by interacting with a high blood volume. Furthermore, the patient refused to undergo any laboratory tests.

Table 1. Clinical characteristics of the patient

Affected organ	Clinical characteristics
Ocular	Diplopia
Psychological	Depression
Sleep disorders	Obstructive sleep apnea
	Staggering or ataxic gait Progressive gait imbalance Uncoordinated body movements Increase in the support base Unsafe gait Clumsiness in movements
Facial	Difficulty swallowing and controlling saliva
Speaks	Impairment of normal speech with difficulty speaking
Muscular	Dystonia

The QR clarifies the actual status of the patient

The short form 36 health survey (SF-36) is a popular instrument for evaluating health-related quality of life in patients with several diseases and conditions [12]. The SF-36 measures eight dimensions: physical functioning, role limitations due to physical health, role limitations due to emotional problems, energy/fatigue, emotional well-being, social functioning, pain, and general health. For each dimension, the items were coded, aggregated, and transformed into a scale ranging from 0 (the worst health status for that dimension) to 100 (the best health status). Moreover, scores higher or lower than 50 indicate better or worse health status, respectively, than the average of the reference population. The component analyses revealed two distinct concepts measured by the SF-36: a physical dimension represented by the physical component summary and a mental dimension represented by the mental component summary. Moreover, this scale was registered in the patient before treatment and after 3 months, and it was repeated after 1 year.

After three months of phototherapy sessions, the following items were scored: physical functioning, role limitations due to physical health, role limitations due to emotional problems, energy/fatigue, emotional well-being, social functioning, and general health. The only variable that remained unchanged was pain. However, he discontinued treatment, attributed the deterioration to phototherapy.

A year later, as the disease progressed, he decided to resume the same scheme, and the following trends were observed: (a) improvement in role limitations due to physical health, emotional well-being, social functioning, and general health; (b) no changes in role limitations due to emotional problems and pain; and (c) deterioration in energy/fatigue and health changes (Table 2).

DISCUSSION

Although the age at onset of MJD is significantly varied (with extremes ranging from 4 to 70 years of age), the average age of onset is 32-40 years. The clinical presentation will begin to become progressive and disabling, with an average life expectancy of 20-25 years. In the present case, symptoms began at the average of the highest incidence in the world.

The abnormal repeat of the CAG trinucleotide tends to alter the *ATXN3* gene located on chromosome 14, in its fragment 32 of the short arm, and the protein ataxin-3, a soluble cysteine, weighing 42 kD, which is part of the cysteine-protease group with activity in the ubiquitin-proteasome system and participates in regulating protein degradation. Intracellular accumulation occurs in some parts of the brain. An excess of polyQ is also

Table 2. The 36 health survey questionnaire changes				
Parameter	**Date**			
	January 2023	**March 2023**	**January 2024**	**March 2024**
Physical functioning	70	50	35	30
Role limitations due to physical health	100	25	0	50
Role limitations due to emotional problems	100	66.7	66.7	66.7
Energy/fatigue	65	45	45	40
Emotional well-being	68	44	32	40
Social functioning	75	62.5	37.5	50
Pain	77.5	77.5	77.5	77.5
General health	60	40	15	35
Health change	75	25	25	25
This scale provides a profile of health status and is applicable to both patients and the general population. The scale goes from zero (the worst) to 100 (the best) in each dimension				

produced, creating a toxic environment for neurons and forming inclusions with the mutated protein ataxin-3, thereby resulting in MJD [7]. In the case of our patient's family, although several family members died from the same disease, no autopsy has been performed to corroborate intracellular accumulations.

The severity of abnormalities observed in the images depends on the length of the CAG repeats. Other studies, such as spect, are useful for observing certain abnormalities, such as the reduction in the density of the dopamine transporter in the striatum. In molecular diagnosis, different techniques are used to identify the main genes underlying MJD. It is important to differentiate MJD from other diseases because they share several similarities, such as autosomal dominant striatonigral degeneration, hereditary dentatorubral-pallidoluisian atrophy, and syphilis. In relation to this, it is crucial to insist with the patient to accept nuclear magnetic resonance imaging and other imaging studies of the central nervous system.

MJD is a progressive pathological condition with no specific treatment and varies symptoms. It responds in a limited way to symptomatic treatment. Unfortunately, since MJD is a rare and clinically heterogeneous pathology, it has not been possible to conduct double-linked clinical trials with controls and placebo. Currently, no proven treatment that can prevent SCA3, as well as other ataxias, has been established. Some experimental treatments include astragaloside IV, MJD1 deactivation, and interference with ribonucleic acid (RNA).

The average survival time of these patients was 21 years, with a range of onset of 7-29 years. In clinically confirmed cases of MJD, three groups of causes of death have been noted: neurological, respiratory, and enfectious diseases. Most patients die of pulmonary complications, usually within 6-29 years of onset, considering that no disease-modifying treatments have been established.

The case we present of application of phototherapy was in the absence of effective treatments that stop or cure cases of MJD and at the patient's request and applying the principle of "first do no harm" given the safety of phototherapy. However, it has not yet been determined whether the slowdown in disease deterioration was due to phototherapy, which requires a molecular explanation. Although the engineer who developed the lamp used with the patient based his work on the concept of isomerization of sugars such as those contained in RNA and deoxyribonucleic acid [13] and considering the evidence of the photobiomodulation effects on Messenger RNA [14], the role of phototherapy cannot be clearly attributed to the enflammatory cascade reduction (Figure 1) [15].

One of the limitations of this study was the lack of application of the following scales were not applied: the International

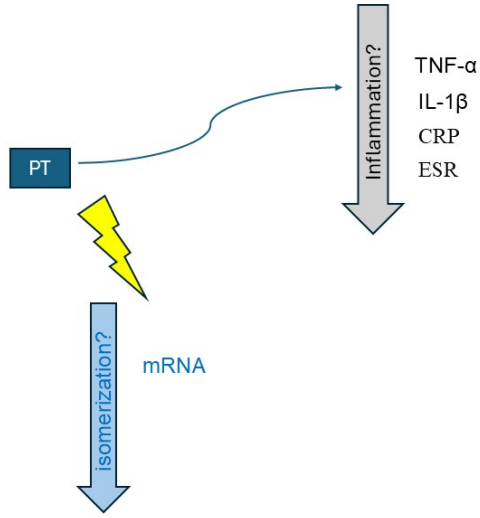

Figure 1. Possible actions of phototherapy on Machado-Joseph disease
CRP: C-reactive protein, ESR: Erythrocyte sedimentation rate, IL: Interleukin, PT: Phototherapy, TNF-α: Tumor necrosis factor-α

Cooperative Ataxia Rating Scale and the Scale for the Assessment and Rating of Ataxia. However, the SF-36 documented clinical evolution stability, stopping the deterioration.

Ethics

Informed Consent: The patient written informed consent was obtained.

Footnotes

Authorship Contributions

Concept: H.M.Z., Design: H.M.Z., Data Collection or Processing: D.E.G.M., P.L.R., M.D.R.P., H.M.Z., Analysis or Interpretation: D.E.G.M., H.M.Z., Literature Search: D.E.G.M., P.L.R., M.D.R.P., H.M.Z., Writing: D.E.G.M., P.L.R., M.D.R.P., H.M.Z.

Conflict of Interest: No conflict of interest was declared by the authors.

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CASE REPORT

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Successful Management of Hemolytic Anemia Due to Rh-immunization, Diagnosed Antenatally

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ABSTRACT

Hemolytic disease of the fetus and newborn (HDFN) remains a significant concern in maternal-fetal medicine, despite advancements in prevention and management. HDFN occurs when maternal alloimmunization leads to the production of immunoglobulin G (IgG) antibodies that cross the placenta and trigger fetal red blood cell hemolysis. Severe HDFN can cause hydrops fetalis and fetal death if left untreated, whereas survivors often face complications like neonatal anemia and hyperbilirubinemia, potentially causing kernicterus. A 42-year-old Rhesus (Rh)-negative woman with Rh-positive fetal anemia underwent intrauterine transfusions at 23 and 29 week of gestation. The fetus received Rh-negative donor blood during cordocentesis, which led to improvement in hemoglobin levels and normalization of Doppler parameters. Delivery at 34 weeks was via cesarean section because of premature membrane rupture. Postnatal management included exchange transfusion, phototherapy, and Ig therapy, resulting in stabilized hemoglobin and bilirubin levels. This case highlights the critical role of antenatal and postnatal interventions in managing severe HDFN. Regular monitoring and timely intervention, including Doppler ultrasound for anemia assessment and transfusion strategies, can significantly improve outcomes for high-risk neonates.

Keywords: Rhesus, newborn, anemia

INTRODUCTION

Hemolytic disease of the fetus and newborn (HDFN) still poses a significant risk to pregnant women, despite advancements in managing and treating affected pregnancies and stopping red blood cell alloimmunization during pregnancy [1]. HDFN occurs when a mother is alloimmunize, either by being exposed to red blood cell antigens that are not compatible with the fetus or by receiving blood that is not compatible. The placenta actively transports the resulting immunoglobulin G (IgG) antibodies, leading to fetal hemolysis and anemia [2,3]. If left untreated, progressive fetal anemia results in hydropsing of the fetal body and, eventually, fetal death. If the fetus survives, ongoing hemolysis causes neonatal anemia and hyperbilirubinemia, which, if untreated, ultimately leads to serious cerebral injury ("kernicterus") [1].

HDFN has no cure. Therefore, interventions have focused on prevention and minimizing the adverse effects of associated complications. By providing Kell-negative donor blood and using Rhesus (Rh) Ig as a preventative measure, women of childbearing age can lower the risk of red blood cell alloimmunization and the number of Rh(D)- and K-mediated HDFN. However, the gap between supply and demand for anti-Rh(D) drugs remains large in low-income countries and is below optimal thresholds in high-income countries. Moreover, although the disease still poses a significant risk of mortality and morbidity in developing countries, it is considered treatable with favorable outcomes in developed countries [4].

CASE PRESENTATION

A 42-year-old repeat pregnant woman presented to the department of obstetrics and gynecology with the diagnosis: fetal anemia due to Rhesus immunization. The mother's blood group was A (II) Rh (-), and fathers was B (III) Rh (+). We determined the fetal blood group to be AB (IV) RH (+). The woman underwent cordocentesis and fetal hemotransfusion at 23 and 29 weeks. According to the examination results at week 23, the fetal weight was 527 g, and the preoperative tests showed a hemoglobin level of 3 g/dL and an middle cerebral artery peak systolic velocity (MCA-PSV) of 79.165 m/s (2.604 MOM). During the surgery, the fetus received 87 mL of donor blood [0 (I) Rh (-)]. There were no complications in the postoperative period, laboratory parameters of the hemogram

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improved, and the MCA-PSV level normalized (37.14; 1.222 MOM). We repeated the hemotransfusion at 29 gestational weeks. The preoperative examination results showed that the fetal weight was 1.213 g, the hemoglobin level was 8.2 g/dL, and the MCA-PSV was 67.77; 1.76 MOM. The fetus received a transfusion of 100 mL of donor blood [0 (I) Rh (-)] during repeat cordocentesis. The postoperative period was uneventful. The laboratory results showed positive dynamics: the hemoglobin level increased to 14.3 g/dL, and the MCA-PSV normalization was 41.17; 1.042 MOM.

At 34 weeks of gestation, due to premature rupture of the membranes and the presence of a uterine scar, emergency cesarean section was performed. The neonate was born with a body weight of 2170 kg and an Apgar score of 6/7 points. The infant's condition stabilized, leading to his transfer to the intensive care unit. Due to signs of respiratory failure, he received respiratory support in the form of nasal continuous positive airways pressure during the 24-hour period. The umbilical vein and artery. Complete blood count revealed a hemoglobin level of 8 g/dL. It is decided to perform an isovolumetric double-volume exchange blood transfusion, considering the neonate's condition, RH-immunization, hemoglobin, and bilirubin levels. We transfused 350 mL of donor blood to the infant within 30 minutes and extracted 320 mL of the recipient blood. Hemodynamic parameters were stable during hemotransfusion, and posttransfusion complications were not observed. During hospitalization, the neonates underwent continuous phototherapy and received two injections of Ig at a rate of 1 g/kg at 24-hour intervals. We monitored the patient's blood tests and noted positive dynamics in the form of an increase in hemoglobin and a decrease in total bilirubin. The newborn were discharged in a stable condition, and the relevant recommendations were made.

DISCUSSION

Rh hemolytic disease can be mild (mild anemia and jaundice) or severe (severe anemia, hydrops fetalis, and death). Therefore, antenatal and postnatal follow-up of high-risk babies is important [1]. Currently, the peak systolic velocity of the midbrain artery with Doppler ultrasound, a non-invasive method, is measured to determine the severity of fetal anemia and to perform intrauterine transfusions (IUT) if necessary. This procedure is performed to prevent severe fetal anemia and other complications [5].

Two IUT and postnatal exchanges were performed in this case. Following the exchange, blood tests revealed a hemoglobin level of 16.7 g/dL. The serum bilirubin level decreased more effectively because the exchange took longer to allow for extravascular and intravascular bilirubin equilibration. Serum bilirubin level was 6.9 mg/dL after exchange transfusion. The newborn underwent phototherapy after the exchange transfusion, and the intravenous immune globulin treatment was successful. In this case, we did not observe any vascular complications, infection, coagulopathic, electrolyte abnormalities, acidosis, alkalosis, necrotizing enterocolitis, nutritional intolerance, anemia, polycythemia, hypothermia, hyperthermia, graft-versus-host disease, apnea and bradycardia, hypotension, or hypertension [6].

We conclude that severe hemolytic diseases requiring IUT may also occur during the postnatal period. Although easy and postnatal exchange transfusions may be necessary, the duration of postnatal phototherapy and transfusions may vary.

Ethics

Informed Consent: Written informed consent was obtained from the parents for the publication of this report.

Footnotes

Authorship Contributions

Surgical and Medical Practices: T.J., S.E., Concept: S.E., Design: N.H., Data Collection or Processing: G.E., Analysis or Interpretation: S.M., Literature Search: S.M., G.E., Writing: S.M.

Conflict of Interest: No conflict of interest was declared by the authors.

Financial Disclosure: The authors declared that this study received no financial support.

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CASE REPORT

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Lipoma of the Ischiorectal Fossa: A Rare Case Report

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ABSTRACT

Ischiorectal fossa (IRF) masses are uncommon and pose significant diagnostic challenges because of their varied etiologies. This case report presents a rare case of an IRF lipoma in a 55-year-old female patient characterized by a large, benign gluteal mass. Clinical evaluation included imaging with contrast-enhanced magnetic resonance imaging (MRI), which confirmed the presence of a 147x67 mm transverse lipomatous mass extending into the right gluteal fold without malignant features. Surgical resection was performed using the posterior midline approach, which allowed complete tumor removal with optimal cosmetic outcomes. Postoperatively, the patient remained symptom-free, with no recurrence noted after 8 months. This report emphasizes the importance of MRI in the diagnostic process and surgical planning of IRF masses and highlights the rarity and unique characteristics of lipomas in this anatomical location.

Keywords: Ischiorectal fossa, lipoma, benign tumor, tumor recurrence

INTRODUCTION

The ischiorectal fossa (IRF), also known as the ischioanal fossa, is a fat-filled anatomical space. Its boundaries include the levator ani muscle superiorly, the obturator internus muscle and fascia laterally, the external anal sphincter muscle medially, the perineal muscles anteriorly, and the skin inferiorly [1]. Tumors in this region are typically benign, although they may manifest as perianal or gluteal swellings, sometimes compressing adjacent structures such as the rectum, anus, or bladder, causing symptoms like obstruction. The diagnostic tools of choice include computed tomography and magnetic resonance imaging (MRI), with biopsies being recommended primarily when malignancy is suspected [2,3].

CASE PRESENTATION

A 55-year-old woman presented with a growing mass in the right gluteal region, which had been developing for 3 years. She experienced symptoms such as discomfort, dysmenorrhea, and constipation. A clinical examination revealed a large protruding mass from the right gluteal area. MRI demonstrated a 147x67 mm mass in the right ischiorectal region, extending into the gluteal fold, exerting pressure on the anal sphincter and pelvic floor muscles. The differential diagnosis included lipoma, low-grade liposarcoma, and epidermoid cyst [4]. Pathological examination confirmed the diagnosis of a benign lipomatous mass (Figure 1). An open biopsy was performed, and histopathological examination of the specimen revealed fibrolipoma with no signs of atypia. As the surgical method we used “Perineal approach” technique. This technique improves the visibility and removal of lipomas.

DISCUSSION

Tumors originating in the IRF are rare. Due to the anatomical complexity and deep location of this region, they pose significant diagnostic and therapeutic challenges. The IRF is a fat-filled space and is prone to various types of masses, but most are benign and non-aggressive. However, due to their proximity to critical pelvic structures, including the rectum, anus, and pelvic floor, these tumors can cause different symptoms depending on their size and location. Common presenting symptoms include swelling, discomfort, pain, and, in some cases, obstructive symptoms like constipation or urinary retention. This case is notable for its presentation with a large lipomatous mass causing pressure-like symptoms but without malignant features, which is consistent with previously reported cases of benign lipomatous tumors in the IRF [5].

Lipomas which are common benign tumors, are infrequently found in the IRF, with only a handful of cases documented in the literature. These tumors arise from mature adipose

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tissue and are typically slow-growing, as seen in this case, in which the patient had a history of gradually enlarging masses over 3 years. The use of MRI has played a crucial role in both diagnosis and surgical planning. MRI is considered the gold standard for imaging soft tissue tumors, providing excellent differentiation between fatty and non-fatty tissue, as well as detailed information on the size, shape, and relationship of the tumor to adjacent structures [1,6]. MRI findings of a homogenous, well-defined mass with signal characteristics similar to subcutaneous fat strongly suggest a diagnosis of lipoma. However, it is important to distinguish between benign lipomas and liposarcomas, which can have overlapping imaging features. In cases in which MRI findings are ambiguous or in which there is a suspicion of malignancy, biopsy may be warranted to confirm the diagnosis [4].

Surgical excision remains the treatment of choice for symptomatic IRF lipomas, particularly when they cause significant discomfort or functional impairment. The posterior midline approach in this case allowed for adequate exposure and complete tumor resection. This approach is often favored for IRF tumors due to the deep-seated location and proximity to vital structures. Complete excision is important to minimize the risk of recurrence, which has been documented in other IRF tumors, particularly those with malignant potential, such as aggressive angiomyxoma [7,8]. In contrast, lipomas have a much lower risk of recurrence following complete resection. In this case, at the eight-month follow-up, the patient did not present with symptoms, and no recurrence was observed, highlighting the efficacy of the surgical intervention.

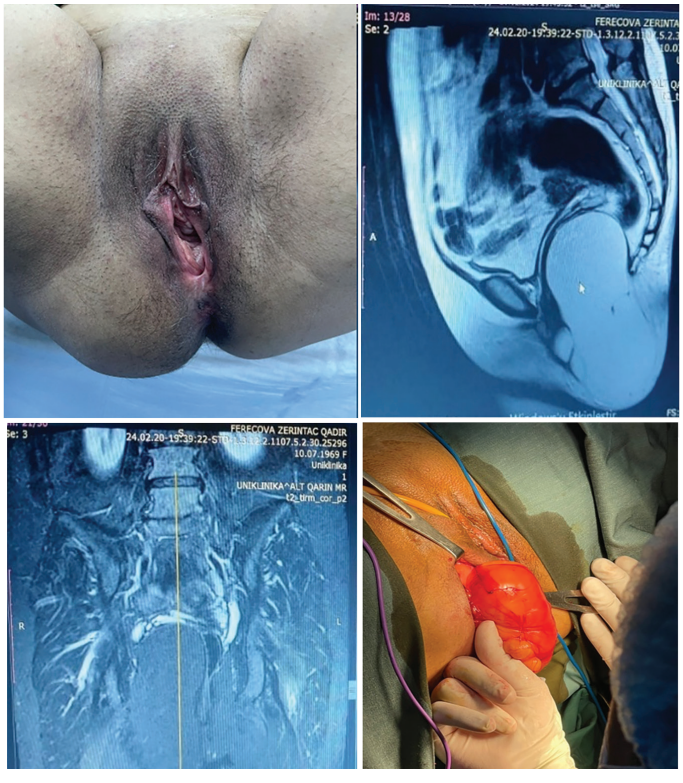

Figure 1. Preoperative examination and operative imaging

One of the challenges in managing IRF tumors is ensuring that no residual tumor tissue remains postoperatively, as incomplete excision is associated with a high risk of recurrence. Although benign tumors like lipomas have a low recurrence rate, careful follow-up is still warranted to monitor for signs of recurrence, particularly within the first year after surgery. In the case of aggressive tumors, such as liposarcomas and angiomyxoma, early recurrence is a well-documented phenomenon, and close postoperative surveillance is critical [2].

Furthermore, it is important to note that although most IRF tumors are benign, a thorough differential diagnosis is necessary. Other potential tumors include cystic lesions, such as epidermoid cysts, which can also present as well-circumscribed masses in the IRF, or more aggressive neoplasms like sarcomas. Therefore, imaging, combined with clinical judgment, plays a pivotal role in the management of these patients.

In this case, the benign nature of the lipomatous tumor led to a favorable outcome with minimal morbidity. The cosmetic outcomes were also satisfactory owing to the use of the posterior approach, which minimizes visible scarring and preserves the function of surrounding structures. This case underscores the importance of a multidisciplinary approach involving radiologists, surgeons, and pathologists to ensure accurate diagnosis and optimal treatment of IRF tumors (Figure 2) [5].

This case highlights the rarity of IRF lipomas and the importance of MRI for both diagnosis and surgical planning. Surgical resection remains the mainstay of treatment for symptomatic tumors, with a low risk of recurrence if complete excision is achieved. Long-term follow-up is necessary to ensure the absence of recurrence, especially in tumors with malignant potential. Although IRF tumors are rare, this report adds to the growing body of literature on the presentation, diagnosis, and management of lipomatous tumors in this anatomically complex region.

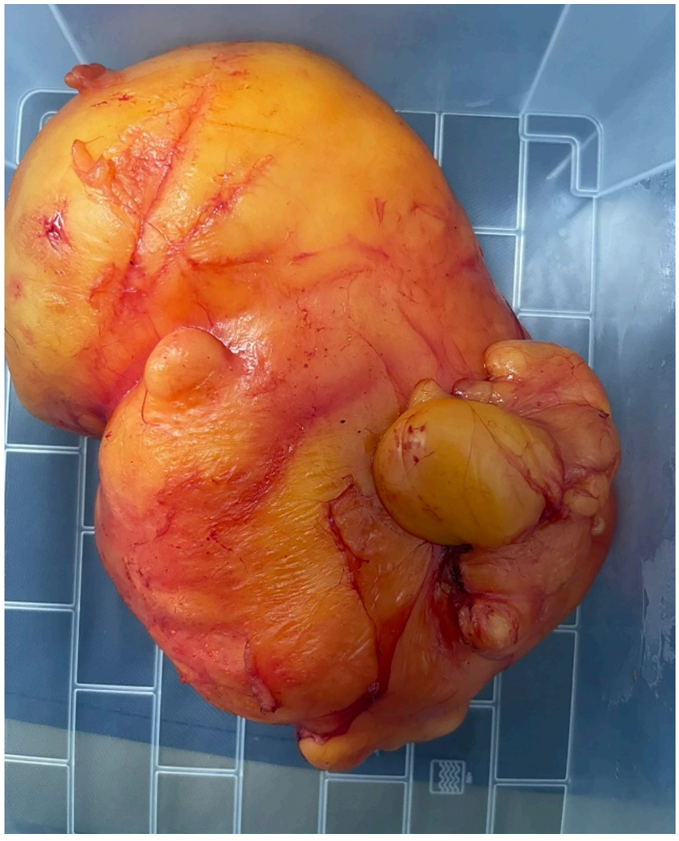

Figure 2. Surgical material: Lipoma

Ethics

Informed Consent: Informed consent was obtained from the patient.

Footnotes

Authorship Contributions

Surgical and Medical Practices: D.K., K.G., Concept: D.K., K.G., Design: D.K., K.G., Data Collection or Processing: K.G., Analysis or Interpretation: D.K., K.G., Literature Search: K.G., Writing: D.K.

Conflict of Interest: No conflict of interest was declared by the authors.

Financial Disclosure: The authors declared that this study received no financial support.

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CASE REPORT

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Sellar Chondroma Misdiagnosed as Craniopharyngioma: A Case Report

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ABSTRACT

Tumors in the saddle area comprise a wide range of benign and malignant entities, with chondroma accounting for only a small fraction. There are only few reported cases in the literature. Sellar chondroma needs to be distinguished from pituitary adenoma, meningioma, craniopharyngioma, and chordoma, which is sometimes difficult to diagnose due to its rarity and overlap with other imaging findings. Moreover, it is easy to be misdiagnosed on imaging alone. Definitive diagnosis should depend on pathological findings. Despite being a benign tumor, saddle area chondroma poses a significant surgical challenge and is prone to complications due to its unique location, warranting attention from clinicians.

Keywords: Headache, sellar chondroma, misdiagnosed, craniopharyngioma

INTRODUCTION

Chondroma is a benign bone tumor, most commonly observed in the short bones of the hands and feet, and rarely occurs intracranially. The majority of intracranial chondromas originate from the skull base cartilage, with a minority occurring in the convexity dura mater, cerebral falx, and other locations [1], leading to potential misdiagnosis in imaging studies. We present a case of a sellar chondroma initially misdiagnosed as a craniopharyngioma.

CASE PRESENTATION

A 61-year-old female patient with a 6-month history of headaches that occurred intermittently and lasted 20 minutes to 2 hours each time; symptoms do not relieve during rest, and there were no aggravating or alleviating factors. She has no symptoms of nausea, vomiting, limb weakness, visual decline, blurred vision, visual field defects, or polyuria; and has never undergone relevant treatment. The patient had a 5-year history of hypertension, with a fluctuating systolic blood pressure ranging from approximately 105 to 155 mmHg, and she did not adhere to regular medication. Furthermore, she denied having diabetes. Head computed tomography (CT) revealed a circular mass in the sellar region, approximately 3.0x2.4 cm in size, with uneven density and arcuate calcifications at the margin (Figure 1A). The physical examination of the patient revealed no abnormalities. Hematological examination revealed normal levels of plasma cortisol (16.15 ug/dL), testosterone (16.60 ng/dL), progesterone (0.42 ng/mL), estradiol (18.90 pg/mL), triiodothyronine (1.2 ng/mL), thyroxine (6.41 ug/dL), thyroid-stimulating hormone (0.32 uIU/mL), adrenocorticotropic hormone (25.50 pg/mL), follicle-stimulating hormone (37.78 mIU/mL), luteinizing hormone (9.41 mIU/mL), and prolactin (13.16 ng/mL). The patient underwent further cranial magnetic resonance imaging (MRI) plain scan + enhancement, revealing a mixed-signal mass in the sellar region with clear boundaries; T1-weighted imaging (T1WI) showed low signal (Figure 1B), T2-weighted imaging (T2WI) showed high signal (Figure 1C), and the enhancement scan showed uneven enhancement (Figure 1D), with compression of the pituitary causing flattening. Radiological diagnosis suggests the possibility of craniopharyngioma. Head computed tomography angiography (CTA) revealed a mass close to the bilateral internal carotid arteries, terminal branches of the basilar artery, and posterior cerebral arteries on both sides (Figure 2A).

The patient underwent surgical treatment via a left frontal subtemporal approach; intraoperatively, the mass had abundant blood supply, was closely adherent to surrounding vessels, and most of the mass was excised, achieving complete hemostasis. Postoperative pathological examination confirmed

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the diagnosis of a chondroma (Figure 2B). On the second day after surgery, the patient experienced severe headaches, bilateral pupils lost light reflex, and bilateral Babinski signs were positive. Head CT revealed a new-onset left frontal lobe intracranial hemorrhage, prompting emergency evacuation of the hematoma and decompression surgery with bone flap removal on the left frontal lobe. Postoperatively, the patient received fluid resuscitation, dehydration for intracranial pressure reduction, nutritional support for brain function, and electrolyte supplementation as symptomatic supportive treatment. Postoperative re-evaluation of pituitary hormones showed normal results, and the patient improved and was discharged on the 22nd day after admission.

DISCUSSION

Although chondromas are common benign bone tumors, they are rarely found in the sellar region, accounting for less than 1% of all intracranial tumors [2]; their clinical manifestations are highly diverse, including headaches, visual problems, and hormonal imbalances, among others [3].

Sellar chondromas have clear margins and may exhibit varying degrees of calcification or ossification, which is one of their characteristic features; on MRI, they typically present low signal intensity on T1WI and high signal intensity on T2WI, consistent with the nature of cartilaginous tissue [4]; MRI can also clarify the relationship between the tumor and the pituitary. CT bone window imaging provides a clear view of the bone structure of the sella turcica, aiding in the assessment of the tumor's impact on surrounding bone quality. Multiplanar reconstruction of CTA is crucial for elucidating the relationship between the tumor and surrounding structures, especially the pituitary gland, optic nerves, and major blood vessels.

Sellar chondromas should be differentiated from many other sellar tumors. Although chondromas do not have specific radiological manifestations, the literature reports that 60%-90% of chondromas have irregular and patchy calcification, which is an important

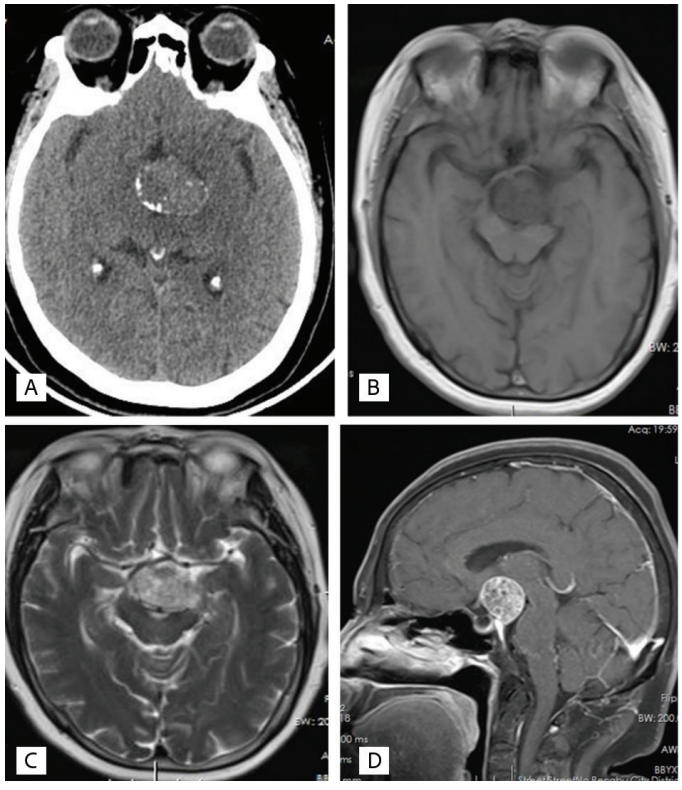

Figure 1. CT and MRI findings. (A) An axial CT image of the head shows a sellar mass with marginal calcification. (B) Axial T1-weighted image showing hypointensity. (C) Axial T2-weighted image showing hyperintensity. (D) Sagittal postcontrast T1 fat-saturated image showing uneven enhancement

CT: Computed tomography, MRI: Magnetic resonance imaging

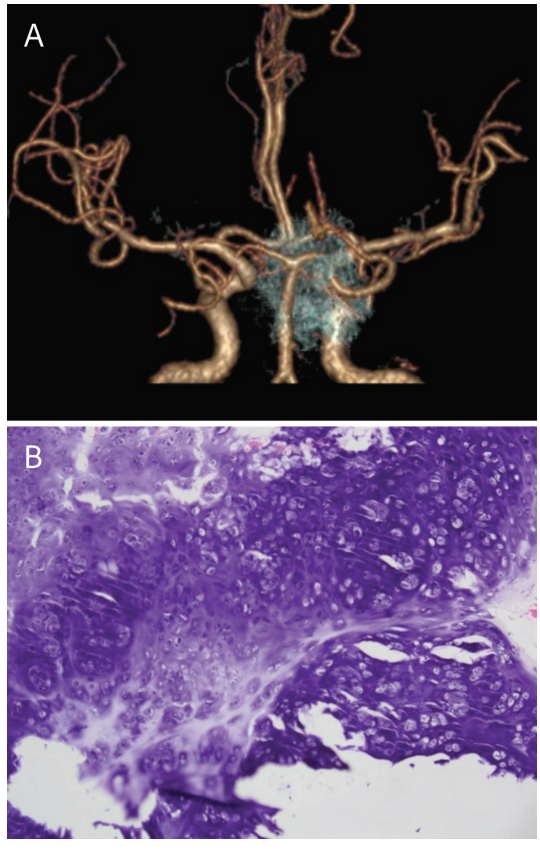

Figure 2. CTA and pathological findings. (A) A cerebral vascular virtual reality reconstruction image showing a close relationship between the mass and adjacent blood vessels. (B) Hematoxylin and eosin-stained image of the mass showing diffuse cartilaginous cells

CTA: Computed tomography angiography

basis for diagnosing chondromas [5-7]. Pituitary adenomas are common in the sellar region, but generally have less calcification and gonadotropin-secreting effects [6]. It is not difficult to differentiate the tumor from the chondroma by observing the relationship between the tumor and pituitary gland on imaging. Calcification is the manifestation of adamantinomatous craniopharyngiomas, which usually present as "eggshell"-like calcification at the edges [8]. The calcification of chondromas is mostly internal, and craniopharyngiomas are cystic and solid, and the signal is more complex than that of chondromas. Similarly, meningiomas in the sellar region may contain sand bodies but generally present with fewer calcifications. Most meningiomas are adjacent to the skull, with a wide base. The enhanced scan shows obvious and uniform enhancement, with an enhanced "dural tail", which is different from the manifestations of chondromas [7]. In addition, chordoma is an important differential diagnosis. Chordoma commonly occurs in the central occipital clivus, with unclear margins and extensive osteolytic bone destruction. In contrast, chondromas are usually located on one side of the midline. Chondromas cause less damage to bone and usually absorb adjacent bone under pressure; at the same time, some chondromas turn into chondrosarcomas [7]. It is difficult to distinguish a chordoma when the tumor is rapidly growing or when bone destruction is aggravated. In such cases, a definitive diagnosis relies on pathological and immunohistochemical analysis, and chordoma usually present with positive staining for creatine kinase, epithelial membrane antigen, and S-100 proteins [9].

Surgical excision is typically the preferred treatment for sellar chondromas, particularly for symptomatic tumors [2,3]. Surgical treatment of sellar chondromas poses some specific challenges because of the proximity of the sella turcica to the optic chiasm and pituitary gland; postoperative complications may include visual impairment or hormonal dysfunction. In addition, the sellar region contains many important vascular structures, and thus, surgery may pose a significant risk of bleeding. In our case, acute intracranial hemorrhage occurred on the second postoperative day, and timely surgery saved her life.

Sellar chondromas are generally benign, and the treatment outcome may depend on the size, location of the tumor, and the patient's underlying condition. For patients undergoing surgery, postoperative follow-up is crucial to monitor any potential recurrence or complications.

Ethics

Informed Consent: The patient's verbal consent was obtained and waived the written informed consent.

Footnotes

Authorship Contributions

Concept: C.H., W.Y., Data Collection or Processing: W.Y., Literature Search: C.H., W.Y., Writing: C.H., W.Y.

Conflict of Interest: No conflict of interest was declared by the authors.

Financial Disclosure: The authors declared that this study received no financial support.

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CASE REPORT

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J Cau Med Sci 2024;2(3):48-50

Ovarian Fibroma with Substantial Calcification: An Uncommon Case Presentation

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ABSTRACT

Ovarian fibroma is a rare benign tumor of the ovary that is composed of fusiform fibrous cell components. Ovarian fibroma can occur at any age, and it is more common in postmenopausal or postmenopausal women. The clinical manifestations of ovarian fibroma are not typical; they are usually characterized by no obvious symptoms and mostly accidental imaging findings. The density and signal of ovarian fibroma are generally uniform, and necrosis, cystic degeneration, and calcification are rare. We report a 25-year-old young woman who accidentally discovered a large calcified mass on the right side of the pelvic cavity during a computed tomography examination of the lumbar spine. The mass was pathologically confirmed as calcified ovarian fibroma.

Keywords: Ovarian fibroma, calcification, sex cord-stromal tumor

INTRODUCTION

Ovarian fibroma with large calcification is a rare presentation except in Gorlin syndrome [1,2]. Most ovarian fibromas exhibit minimal or no calcification, and when present, calcifications are usually sparse. The presence of significant calcification in ovarian fibroma is rare and may pose a diagnostic challenge, as it can mimic other calcified pelvic masses, such as teratomas and malignancies. In this report, we emphasize the importance of recognizing this rare presentation and considering ovarian fibroma in the differential diagnosis of calcified pelvic masses.

CASE PRESENTATION

A 25-year-old female patient presented to our hospital with lower back pain. The patient underwent lumbar spine computed tomography examination, and unexpectedly, an irregular high-density mass was detected in her pelvic region, approximately 6.2×7 cm in size (Figure 1a). Ultrasonography confirmed the presence of a right adnexal mass with posterior acoustic shadowing (Figure 1b). Pelvic magnetic resonance imaging was used to further characterize the pelvic mass. The axial T1-weighted images (T1WI) (Figure 2a) and sagittal T2-weighted images (T2WI) (Figure 2b) sequences both exhibited prominent low signal intensities, with the enhanced scan revealing mild enhancement (Figure 2c). The patient complained of significant dysmenorrhea and occasional irregular vaginal bleeding but denied any abdominal pain or bloating. In addition, she denied any family genetic disease or other medical history. Laboratory examination upon admission revealed normal results for complete blood count, liver function, and kidney function. Urinalysis revealed elevated white blood cell counts (+++), increased vitamin C levels (+), and 113/L squamous epithelial cells. Tumor markers, including alpha-fetoprotein at 2.0 ng/mL, carcinoembryonic antigen at 1.17 ng/mL, ferritin at 38.00 ng/mL, carbohydrate antigen 125 (CA-125) at 19.60 U/mL, CA-153 at 4.30 U/mL, CA-199 at 3.68 U/mL, and human epididymis protein 4 at 20 pmol/L, all fell within the normal range. Subsequently, the right ovarian mass was removed, and oophoroplasty was performed. The pathological diagnosis of ovarian fibroma with marked calcification.

DISCUSSION

Ovarian fibromas are the most common type of ovarian sex cord-stromal tumor, accounting for approximately 4% of all ovarian tumors [3]. In approximately 84% of cases, ovarian fibromas appear as solid masses [4]. Larger ovarian fibromas may show heterogeneity because of necrosis, cystic degeneration, or hemorrhage. Heterogeneous masses comprise

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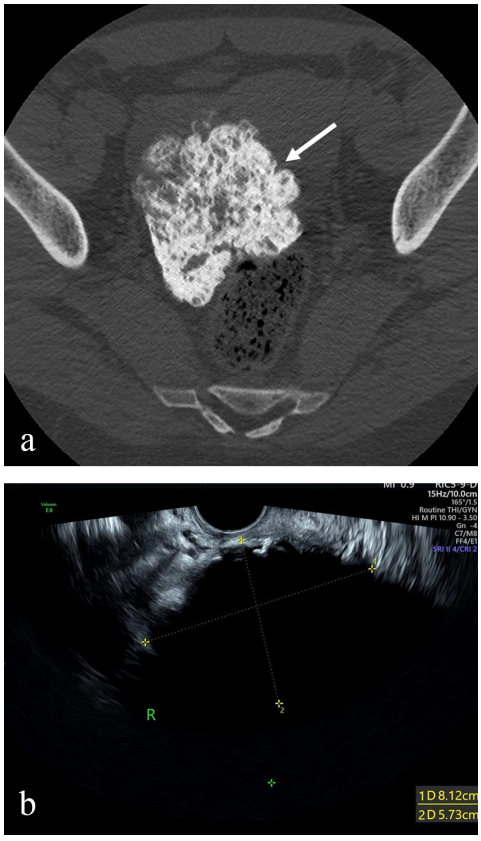

Figure 1. CT and Ultrasonography findings. (a) CT examination showed an irregular high-density mass in the pelvic region (white arrow); (b) ultrasonography of the right ovary

CT: Computed tomography

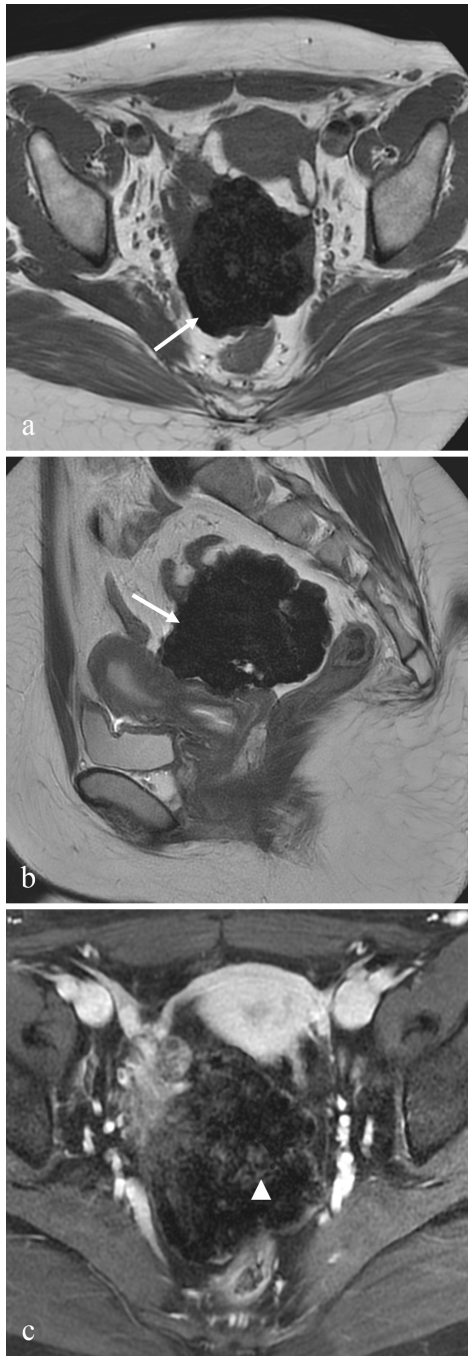

Figure 2. Pelvic MRI findings. (a) Axial T1-weighted image showing low-signal mass (white arrow); (b) Sagittal T2-weighted image showing low-signal mass (white arrow); (c) post-contrast T1 fat-saturated image showing mild enhancement (white triangle)

MRI: Magnetic resonance imaging

approximately 11% of all cases, whereas predominantly cystic lesions account for approximately 5% of cases. Calcified ovarian fibromas are rare, with a higher prevalence observed in Gorlin syndrome [5-8]. Gorlin syndrome is an autosomal dominant disorder characterized by specific diagnostic criteria, including multiple basal cell nevi, jaw cysts, and skeletal abnormalities [7]. Approximately 75% of affected individuals have a familial history of the syndrome [9]. In such cases, 75% of ovarian fibromas are bilateral and multifocal. In the current case, the patient had unilateral ovarian calcifying fibroma, no other skin or skeletal abnormalities, and no family history of Gorlin syndrome, effectively ruling out the possibility of Gorlin syndrome.

Subserosal uterine leiomyomas and ovarian Brenner tumors also demonstrate low signal intensity on both T1WI and T2WI, potentially leading to misidentification as ovarian fibromas. Subserosal uterine leiomyomas have pedicles extending to the uterus and blood vessels between the uterus and the tumor mass. They can be distinguished by assessing the relationship between the ipsilateral ovary and the tumor. For menopausal women, it is difficult to distinguish it from uterine leiomyoma calcification, but this case was a 25-year-old young woman, so the possibility of such a large amount of calcified leiomyoma is very small. Additionally, Brenner tumors predominantly consist of solid components, occasionally featuring amorphous

calcifications within the solid component, and widespread calcification is uncommon. Although ovarian teratoma may contain ossifying components, its fat content can serve as a distinguishing point from fibroma. Compared with fibroma, fibrothecoma may have estrogenic activity, less fibrosis and collagen components, higher T2WI signals, and more frequent necrosis and degeneration [10]. Ovarian fibroma is one of the most common ovarian sex cord-stromal tumors, and it is generally associated with no endocrine function. The tumor indicators in our case were all negative, although there are literature reports that some cases with ovarian fibroma have elevated serum CA-125 levels, tumor size, and ascites are associated with elevated CA-125 levels, and elevated serum CA-125 levels do not originate from tumor cells themselves [11]. Therefore, the tumor marker CA-125 can still be used as one of the distinguishing points between it and ovarian epithelial tumors.

In conclusion, widespread and diffuse calcification is an uncommon pathological feature of ovarian fibromas. Generally, small, asymptomatic tumors do not necessitate treatment, whereas larger tumors causing symptoms are best managed through surgical resection.

Ethics

Informed Consent: We obtained the patient's verbal assent and waived written informed consent.

Footnotes

Authorship Contributions

Concept: C.H., W.Y., Data Collection or Processing: W.Y., Literature Search: C.H., W.Y., Writing: C.H., W.Y.

Conflict of Interest: No conflict of interest was declared by the authors.

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