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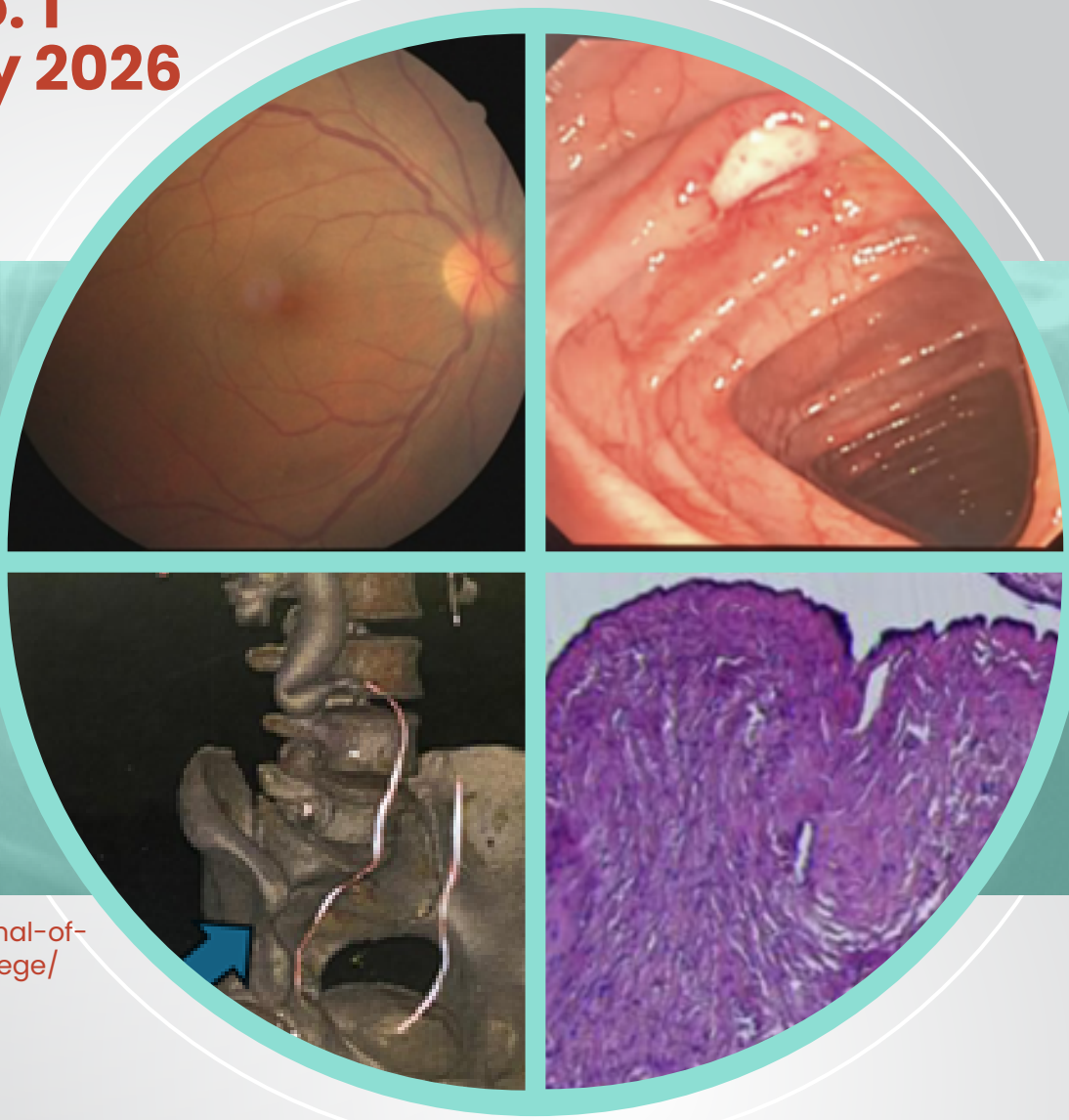

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JOURNAL of RAWALPINDI MEDICAL COLLEGE

*Clinical Case Reports*

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# The need for a Clinical Case Reports Journal from Pakistan

Muhammad Umar, Arsalan Manzoor Mughal

It is with a sense of quiet satisfaction and shared responsibility that we present the first issue of JRMC Case Reports, a special supplement of the Journal of Rawalpindi Medical College devoted exclusively to clinical case reporting. At Rawalpindi Medical University, our affiliated hospitals Holy Family Hospital, Benazir Bhutto Hospital, and Rawalpindi Teaching Hospital serve a very large and diverse patient population. Every day, clinicians working in these settings encounter patients who challenge routine thinking and remind us that medicine is learned most deeply at the bedside. Some encounters offer small but practical insights, while others fundamentally change how we understand a disease process. This journal has been created to capture those lessons, document them with care, and share them with colleagues who face similar realities in their clinical practice.

Clinical case reports hold a modest but powerful place in medical literature. They translate everyday clinical complexity into structured, meaningful narratives that can be easily understood and remembered.[1,2] For medical students and early career doctors, writing a case report encourages close observation, ethical sensitivity, disciplined engagement with literature, and clarity in clinical communication.[3] For practicing clinicians, particularly those working in resource constrained environments, a well written case report often provides timely guidance when standard protocols do not fully apply, such as in patients with unusual presentations, overlapping comorbidities, or limited diagnostic options.[4] Through JRMC Case Reports, we aim to link bedside learning with scholarly writing and connect local clinical experience with the wider medical community.

The cases presented in this inaugural issue reflect the breadth and relevance of focused clinical reporting. They include a young patient with primary AL amyloidosis presenting with macroglossia and spontaneous chest wall bruising, an early and severe ocular manifestation of Behçet's disease identified through occlusive retinal vasculitis, a child with Budd–Chiari syndrome secondary to Burkitt's lymphoma, a life threatening postpartum choriocarcinoma initially mistaken for retained products of conception, and an infant who developed iatrogenic Cushing syndrome following inappropriate use of topical steroids. Each report is concise, clinically grounded, and rooted in local practice realities. Collectively, they represent a growing body of practical knowledge that we believe will be of value to a wide range of readers.

Publishing case reports from RMU affiliated hospitals offers a particular strength. Our institutions receive patients from varied socioeconomic backgrounds, often presenting late and with complex clinical needs. This diversity naturally produces cases that are educational and clinically relevant. While rare and striking cases deserve attention, equal importance must be given to well documented presentations of known conditions, clear descriptions of clinical techniques, transparent reporting of complications, and honest reflections on decision making in difficult situations. Such reports strengthen everyday clinical judgment and support safe patient care.

Upholding ethical and scientific standards remains central to this initiative. JRMC Case Reports is firmly grounded in principles of responsible publishing, transparency, and integrity.[5] All submissions are expected to meet established requirements including informed consent where applicable, appropriate ethical oversight, clear disclosure of conflicts of interest, and accurate attribution of authorship. The editorial team is committed to guiding authors through a supportive and constructive review process, particularly for trainees and early career clinicians. Attention to clinical images, timelines, and evidence based conclusions will remain a priority, with patient dignity always at the centre of our work.

For postgraduate trainees and young clinicians who may be unfamiliar with academic writing, this journal provides an accessible and meaningful starting point. Preparing a case report allows authors to reflect on their clinical reasoning and communicate it in a structured manner that benefits others. Through mentorship and editorial guidance, we hope to build confidence and competence in clinical scholarship across our institutions.

To our readers, we invite you to approach this journal as you would a trusted colleague. Read with curiosity, reflect critically, and contribute when possible. In Pakistan, the medical knowledge we most urgently need often grows from our own clinical experiences. By sharing these experiences with honesty, humility, and academic rigour, we strengthen not only our literature but also our collective practice. We hope JRMC Case Reports becomes a dependable companion for trainees preparing for assessments, clinicians navigating complex decisions, and small research teams documenting local challenges.

With sincere appreciation for the authors who entrusted us with their work, and for the reviewers and editorial colleagues who shaped this issue, we welcome you to JRMC Case Reports. We look forward to its growth as a platform that remains relevant, ethical, and consistently present in the service of patients and the medical community.

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# Primary AL Amyloidosis Presenting with Macroglossia and Spontaneous Chest Wall Ecchymosis in a Young Male

Aamina Masud[1], Seemab Abid[2], Muhammad Afique[3]

## Abstract

**Summary:** We report the case of a 35-year-old male who presented with progressive macroglossia associated with dysarthria and worsening dyspnea with no accompanying stridor, dysphagia, or features suggestive of acromegaly; however, the patient specifically reported difficulty pronouncing words starting with the letter "R." Clinical examination revealed evidence of both cardiac and pulmonary involvement. Comprehensive investigations were conducted to rule out alternative causes of macroglossia. An abdominal fat pad biopsy demonstrated Congo red positivity with apple-green birefringence under polarized light, confirming the diagnosis of primary AL amyloidosis.[4] The patient was initiated on chemotherapy, stabilized, and subsequently referred to the oncology department for outpatient follow-up.

**Keywords:** Immunoglobulin Light-chain Amyloidosis, Macroglossia, Ecchymosis.

## Introduction

Primary (AL) amyloidosis is a plasma cell disorder resulting from excessive production of monoclonal light chains that misfold and deposit as insoluble amyloid fibrils in various organs, resulting in progressive organ dysfunction[1] The kidneys, heart, and lungs are most commonly affected organs[2] Macroglossia, although rare, is considered a pathognomonic clinical feature of AL amyloidosis[3]

## Case Presentation

We report the case of a 35-year-old man, Zahid, a chronic smoker, who presented to the Emergency Department with macroglossia of sudden onset, progressive in nature, accompanied by spontaneous ecchymotic patches over the anterior chest wall. It was associated with difficulty pronouncing words beginning with the letter "R," yet notably, there was no associated stridor, dysphagia, drooling of saliva, or airway compromise. The onset of chest wall ecchymosis occurred simultaneously with the tongue enlargement.

The patient also reported progressive exertional dyspnea corresponding to NYHA Class III, generalized weakness, extreme fatigue, and frothy urine. His past medical history was significant for hypertension and previously diagnosed and operated on bilateral carpal tunnel syndrome. There was no history of trauma, anticoagulant use, or liver disease to explain the ecchymotic lesions.

On examination, there was a diffusely enlarged, non-tender tongue occupying the oral cavity without ulceration or restriction of movement. Multiple ecchymotic patches were noted over the anterior chest wall. Cranial nerve examination was intact. Jugular venous pressure was elevated, and bilateral pitting pedal edema was present. Respiratory examination revealed signs of bilateral pleural effusions. Cardiovascular examination was remarkable for a pansystolic murmur and an audible S3 gallop. Abdominal examination revealed hepatomegaly with ascites. Neurological assessment was unremarkable.

Differential diagnoses considered included primary amyloidosis, acromegaly, congestive cardiac failure, nephrotic syndrome, and hypothyroidism. Baseline laboratory investigations showed 2+ proteinuria, elevated ESR and CRP, and hypoalbuminemia. Brain natriuretic peptide (BNP) levels were raised, indicating cardiac strain. Serum β2-microglobulin was elevated. Serum protein electrophoresis demonstrated a homogeneous monoclonal band in the gamma region, consistent with monoclonal gammopathy, which was further characterized as IgA lambda on immunofixation.

Chest radiography revealed cardiomegaly, while abdominal ultrasonography showed ascites and coarse hepatic echotexture. Echocardiographic findings were consistent with restrictive cardiomyopathy. An abdominal fat pad biopsy demonstrated apple-green birefringence under polarized light on Congo red staining, confirming amyloid deposition. A skin biopsy taken from the right medial scapular region also tested positive for Congo red staining with birefringence.

Bone marrow aspiration revealed increased plasma cells with binucleate forms, suggestive of plasma cell dyscrasia. Bone marrow trephine biopsy showed hypocellular marrow with predominance of plasma cells and megakaryocytes, concluding bone marrow plasmacytosis.

A final diagnosis of primary systemic (AL-type) amyloidosis with multi-organ involvement secondary to plasma cell dyscrasia was established. The patient was initiated on injection cyclophosphamide, bortezomib, and Decadron (dexamethasone) as part of the CyBorD chemotherapy regimen, along with supportive therapy including furosemide, losartan, omeprazole, and prophylactic anticoagulation. He showed initial clinical improvement and was discharged with oncology follow-up for continuation of therapy.

## Discussion

Amyloidosis is a systemic disorder characterized by extracellular deposition of insoluble misfolded protein fibrils composed of glycosaminoglycans, proteoglycans, and serum amyloid P component.[1]

**Contributions:**
AM SA MA - Conception, Design
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AM SA MA - Drafting
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It is classified according to the biochemical nature of the deposited protein. Primary light-chain (AL) amyloidosis results from monoclonal immunoglobulin light-chain deposition secondary to clonal plasma cell proliferation.[2] It predominantly affects individuals in the sixth decade of life, with fewer than 10% of cases reported below 50 years and demonstrates a slight male predominance.[3] The kidneys and heart are the most commonly involved organs, presenting with nephrotic-range proteinuria or restrictive cardiomyopathy, respectively.[4] Although macroglossia is considered a classical sign of AL amyloidosis, it is observed in only 10–20% of patients.[5] Spontaneous ecchymosis, particularly over the chest or periorbital region, is even rarer and is attributed to amyloid infiltration of vascular walls leading to increased capillary fragility.[6]

This case is unusual due to its presentation in a 35-year-old male with simultaneous macroglossia and anterior chest wall ecchymosis as initial manifestations. Differential diagnoses of macroglossia, including acromegaly, hypothyroidism, and neoplastic infiltration, were appropriately excluded through hormonal assays and clinical evaluation. Although the initial tongue biopsy was negative for Congo red staining, elevated beta-2 microglobulin on serum protein electrophoresis raised clinical suspicion. Subsequent abdominal fat pad and skin biopsies confirmed amyloid deposition, and bone marrow examination demonstrated clonal plasma cell infiltration, consistent with AL amyloidosis. The patient was initiated on a bortezomib-based chemotherapy regimen consisting of bortezomib, cyclophosphamide, and dexamethasone (VCD protocol), which is currently recommended as first-line therapy for transplant-ineligible patients.[7,] Early recognition of atypical mucocutaneous manifestations is essential, as delayed diagnosis significantly worsens prognosis. This case highlights the importance of maintaining a high index of suspicion for AL amyloidosis even in younger individuals presenting with isolated physical signs such as macroglossia and unexplained ecchymosis.

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# Eye as the First Clue: Behçet's Disease Unveiled by Retinal Vasculitis and Vein Occlusion

Ambreen Gull[1], Arsalan Sajjad[2], Wajeeha Rasool[3], Fuad Ahmad Khan Niazi[4]

## Abstract

**Summary:** Behçet's disease is a multisystem vasculitis of unknown etiology that is chronic and recurrent. One of the most severe and vision-threatening implications is ocular inflammation. We present a case of severe ocular Behçet's disease with occlusive retinal vasculitis in a young male. A 20-year-old boy had gradually deteriorating vision in both eyes for three months before experiencing a sudden loss of vision in the left eye for two weeks. Ocular examination showed bilateral anterior uveitis with vitritis and perivasculitis. Branch retinal vein occlusion (BRVO) with macular edema and macular pseudohole was seen in the left eye, which was confirmed by fundus fluorescein angiography and optical coherence tomography. A positive pathergy test, as well as repeated oral and vaginal ulcers, confirmed the diagnosis of Behçet's syndrome with panuveitis and occlusive vasculitis. Adalimumab, azathioprine, and systemic corticosteroids were used to treat the patient; this led to a partial recovery of vision and a noticeable reduction in inflammation. Early detection of Behçet's uveitis and timely beginning of immunomodulatory therapy are essential to prevent irreversible retinal damage and subsequent visual loss.

**Keywords:** Behçet's Disease, Retinal Vasculitis, Branch Retinal Vein Occlusion, Macular Edema, Adalimumab, Uveitis.

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## Introduction

Behçet's disease (BD) is a chronic, relapsing vasculitis affecting many organs with uncertain etiology. The disease involves a triad of recurrent oral aphthous and genital ulcers, skin lesions, and ocular inflammation.[1] It is most prevalent along the ancient Silk Road region, with greater rates in Turkey, the Middle East, and East Asia.[1] The condition primarily affects young adults, mainly boys, and can cause considerable morbidity due to vision loss when ocular involvement develops.[1,2]
Behçet's uveitis is usually bilateral, recurring, and non-granulomatous, with a relapsing-remitting history.[3] Ocular symptoms appear in up to 70-80% of patients and frequently affect the disease outcome.[4] The most frequent ocular manifestations are pan-uveitis with retinal vasculitis, which can cause vascular occlusions along with macular edema, which is cystoid in nature. Anterior uveitis in Behçet's disease is non-granulomatous with mobile hypopyon.[4,5] On fluorescein angiography, BD uveitis exhibits fern-pattern leakage, superficial hemorrhages, and occlusive periphlebitis, in contrast to sarcoid or tuberculosis-associated uveitis.[5,6]
The International Criteria for Behçet's Disease (ICBD) combines ocular signs with systemic features such as recurring oral mucosal and genital ulcers, erythema nodosum, papulopustular skin lesions, and a positive pathergy test, which serve as the basis for the predominantly clinical diagnosis.[1,7] For accurate diagnosis and treatment of non-granulomatous uveitis, other differentials need to be ruled out, such as sarcoidosis, reactive arthritis, systemic lupus erythematosus, inflammatory bowel disease, or herpes infections.[8]

## Case Presentation

A 20-year-old man presented at OPD with a gradually progressive, painless blurring of vision in both eyes for three months, followed by an abrupt onset of loss of vision in the left eye. He stated that for the past four years, he had suffered from excruciating, recurrent genital and oral ulcers, each of which healed on its own but left scars. He had intermittent erythematous papulopustular skin lesions but no history of fever, joint pain, backache, cough, weight loss, or prior ocular injury.
On examination, best-corrected visual acuity (BCVA) was 6/12 in the right eye and finger counting at 1 meter in the left eye. The anterior chamber of both eyes showed 2+ cells with mild flare along with bilateral vitritis (2+), with more pronounced haze in the left eye. Fundus examination revealed temporal mild disc pallor, perivasculitis, scattered hemorrhages in all quadrants, ghost vessels inferonasally, and a macular pseudohole in the left eye.
Systemic evaluation revealed a history of recurrent oral and genital ulcers, as well as erythematous papulopustular skin lesions.

**Investigations**

**Ocular Investigations**

Optical coherence tomography (OCT) confirmed macular edema in the left eye. Fundus Fluorescein Angiography (FFA) showed fern-pattern leakage and areas of capillary non-perfusion consistent with occlusive vasculitis and BRVO.
Dermatographia may be seen in some Behçet's patients as a sign of skin hyperreactivity, but it is nonspecific. The pathergy test remains the relevant diagnostic skin test for Behçet's disease.

**Systemic Investigations**

| Parameter | Findings |
|---|---|
| **ESR** | **62 mm/hr.** (elevated) |
| **CBC, LFTs, RFTs** | Within normal limits |
| **Serum ACE** | 13 U/L (normal 9–67 U/L) |
| **Serum Calcium** | 9.9 mg/dL |
| **Pathergy Test** | **Positive** (pustule formation at 48 hours) |
| **HLA-B51** | **Positive** |
| **Chest X-ray/HRCT** | No hilar lymphadenopathy or pulmonary lesions |
| **ANA, ANCA, Infectious panel (TB, syphilis, HSV)** | Negative |

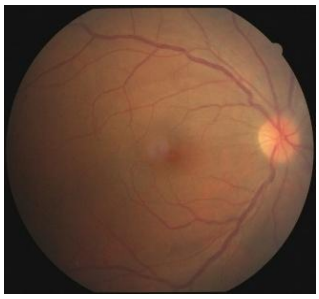

**Figure 1: Fundus Photograph showing hazy fundal view in the Right eye and temporal disc pallor with occlusive perivasculitis, and a macular pseudohole in the left eye**

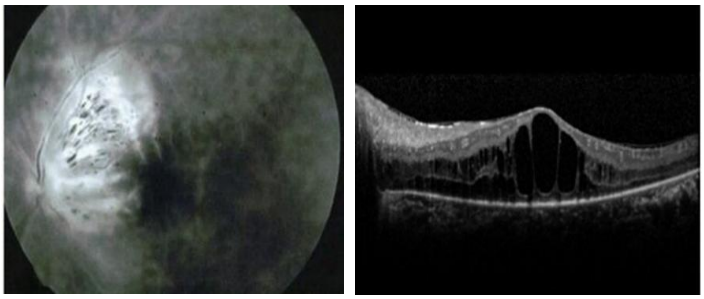

**Figure 2: FFA image of the Late phase showing fern pattern vasculitis in the Left eye and OCT of the Left eye showing macular edema**

**ICBD Classification Criteria Fulfillment**

Diagnosis was made clinically using the International Criteria for Behçet's Disease (ICBD).

According to the International Criteria for Behçet's Disease (ICBD), the patient met the diagnostic threshold:

- Recurrent oral ulcers history (2 points)
- Genital ulcers history (2 points)
- Ocular lesions (uveitis + retinal vasculitis) (2 points)
- Positive pathergy test (1 point)

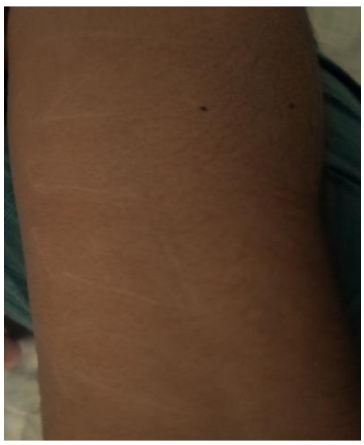

**Figure 3: Dermatographia**

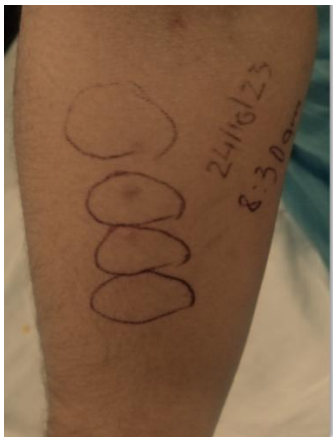

**Figure 4:** Positive Pathergy test

The total score of **7 points** exceeds the ≥4 requirement, confirming the diagnosis of Behçet's disease.

The uniqueness of this case lies in the unusually early and severe posterior segment involvement, including BRVO and macular pseudohole, as the first major presenting ocular feature.

These results excluded other granulomatous and autoimmune diseases, including sarcoidosis, tuberculosis, systemic lupus erythematosus (SLE), and inflammatory bowel disease (IBD).

**Differential Diagnosis**

**1. Inflammatory Bowel Disease (IBD)**

IBD can present with oral ulcers and uveitis; however, this patient had no gastrointestinal symptoms or colonoscopic evidence of IBD. The ocular pattern—occlusive retinal periphlebitis with BRVO and macular edema—is more typical of Behçet's, as IBD rarely causes occlusive posterior vasculitis.

**2. Systemic Lupus Erythematosus (SLE)**

SLE may cause retinal vasculitis and oral ulcers, but this patient had no systemic features (photosensitivity, rash, serositis, renal disease) and negative ANA. The presence of genital ulcers and a positive pathergy test further favored Behçet's over lupus.

**3. Seronegative / Reactive Arthritis**
These can present with mucocutaneous lesions and uveitis, but posterior occlusive vasculitis and BRVO are rare. Absence of arthritis, prior infection, and the presence of recurrent oral/genital ulcers with positive pathergy ruled out reactive arthritis.
**4. Herpetic Infections (HSV/VZV)**
Herpetic retinitis is usually unilateral, necrotizing, and rapidly progressive. The bilateral, occlusive periphlebitis without necrosis, and absence of corneal or serologic evidence excluded viral etiology.
**5. Sarcoidosis**
Sarcoidosis causes granulomatous uveitis with hilar lymphadenopathy and elevated ACE. This patient had non-granulomatous uveitis, normal imaging, and normal ACE, making sarcoidosis unlikely.
**6. Other Systemic Vasculitides (e.g., Granulomatosis with Polyangiitis, Polyarteritis Nodosa)**
These typically cause systemic involvement (renal, ENT, or neurologic) with positive serology such as ANCA. The absence of multisystem disease and mucocutaneous ulceration pointed instead toward Behçet's.
**7. Relapsing Polychondritis**
Characterized by auricular and nasal cartilage inflammation with audio vestibular symptoms, which were not present in this case.
**8. Multiple Sclerosis (MS)**
MS can present with intermediate uveitis but lacks oral/genital ulcers, pathergy positivity, or occlusive retinal vasculitis. Normal neuroimaging further excluded MS.

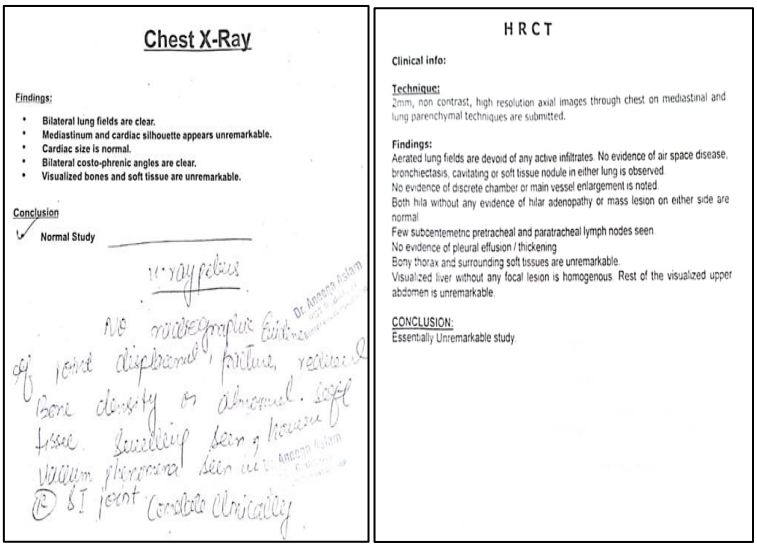

**Figure 5: Normal chest X-ray and HRCT**

**Diagnosis**
Based on clinical findings and systemic manifestations, a diagnosis of Behçet's disease with severe ocular involvement (Pan-uveitis with occlusive vasculitis) was made.
**Treatment**
The patient was started on oral prednisolone 1 mg/kg/day for 3 weeks, followed by gradual tapering. Azathioprine 100 mg twice daily was initiated as a steroid-sparing agent.[4,9] Owing to severe posterior segment inflammation, adalimumab was added (loading dose 80 mg subcutaneously, followed by 40 mg every other week).[1,10] Supportive therapy included omeprazole, folic acid, and calcium with vitamin D3 supplementation.
**Outcome And Follow-Up**
At one month, ocular inflammation had markedly subsided with clearing of vitreous haze. At three months, OCT demonstrated resolution of macular edema. The final BCVA was 6/6 in the right eye and 6/18 in the left eye. The patient remains under close follow-up on maintenance immunomodulatory therapy with Adalimumab 40 mg administered subcutaneously every two weeks, planned for 18–24 months to maintain remission and prevent ocular relapses.[9,10]

## Discussion

Behçet's disease is a systemic vasculitis that affects arteries and veins of all calibers, with ocular involvement being the most serious and vision-threatening component.[1,2] Ocular Behçet's typically presents as bilateral, recurrent, non-granulomatous uveitis, most often involving the posterior segment.[3,4] Retinal vasculitis, particularly occlusive periphlebitis, is the hallmark feature, observed in up to 85% of cases.[6] Recurrent inflammatory episodes can cause retinal ischemia, macular edema, and optic atrophy, and the disease has a relapsing-remitting course.[1]
Our patient demonstrated the characteristic ocular features of vitritis, occlusive retinal vasculitis with perivenular sheathing, along with recurrent oral and genital ulcers and a positive pathergy test. The presence of branch retinal vein occlusion (BRVO) and a macular pseudohole indicated severe vascular inflammation and ischemic damage. A diagnostic gap exists because early ocular Behçet's disease can mimic several inflammatory and infectious uveitic conditions. Retinal periphlebitis and BRVO may be misattributed to other vasculitides, delaying immunomodulatory treatment. This case reinforces the need to recognize ocular clues that point toward Behçet's before systemic manifestations become prominent.[3-6]

Differential diagnoses include infectious and non-infectious uveitic entities such as sarcoidosis, SLE, systemic lupus erythematosus, multiple sclerosis, and inflammatory bowel disease.[8] However, these were excluded based on clinical presentation and relevant investigations. Behçet's disease remains a clinical diagnosis supported by systemic manifestations and exclusion of mimicking conditions. [1,7]

Management of Behçet's uveitis depends on disease severity. Acute attacks require prompt high-dose corticosteroids, either oral or intravenous methylprednisolone (1 g/day for 3 days in severe inflammation), to control acute inflammation.[9,10] To prevent relapses and minimize corticosteroid dependency, long-term immunosuppressive agents such as azathioprine, cyclosporine, and methotrexate are recommended.[2,10] Biologic medicines, especially anti-tumor necrosis factor (TNF) treatments like infliximab and adalimumab, have shown notable effectiveness in managing severe or resistant posterior segment illness. These agents have demonstrated efficacy in reducing inflammation, clearing up macular edema, and lowering the likelihood of relapses.[4,9]

Up to 25% of patients with ocular Behçet's may suffer from serious sight loss despite vigorous therapy, particularly young boys with early-onset posterior illness.[3,5,6] Immunomodulatory treatment must be started as soon as possible to avoid irreparable vascular blockage and optic nerve injury. Long-term disease activity tracking and treatment response, along with side effects associated with therapy, are crucial. When prompt and appropriate therapy is started, about 60% of patients experience remission after the first several years.

Unlike typical Behçet's disease, which usually begins with recurrent oral/genital ulcers followed by anterior uveitis, this case is noteworthy because Behçet's disease rarely presents initially with severe occlusive vasculitis and branch retinal vein occlusion (BRVO) in a young adult. The presence of a macular pseudohole further reflects significant ischemic damage at first presentation, a combination infrequently reported in the literature.[3,10] Early detection of this aggressive ocular phenotype is clinically important because it necessitates rapid escalation to biologic therapy. Reporting this case is significant because it illustrates the importance of considering Behçet's disease in young patients presenting with sudden retinal vein occlusion and occlusive vasculitis. Early recognition allows timely initiation of immunosuppressive and biologic therapy, which is critical for preventing irreversible visual loss.

Young individuals who present with recurrent oral/genital ulcers and ocular inflammation should be suspected of having Behçet's illness. Occlusive retinal vasculitis, macular edema, and recurrent uveitis are the hallmarks of Ocular Behçet's, a unique and potentially blinding condition. To salvage vision, systemic corticosteroids and immunomodulatory medication must be started as soon as possible. In cases where these treatments are ineffective, biologics may be used. To track disease activity and side effects from treatment, routine ocular and systemic follow-up is required.

## Conclusion

Behçet's uveitis primarily affects the posterior segment of the eye and is bilateral, recurrent, and non-granulomatous. Retinal vasculitis and vein occlusion are two common disorders that can result in severe vision loss. Biologic treatment and early immunosuppression are essential for maintaining vision. Before establishing BD, always rule out mimics, including lupus, sarcoidosis, and tuberculosis. The best results are guaranteed by multidisciplinary management, which includes dermatology, rheumatology, and ophthalmology. Regular OCT and FFA monitoring helps identify recurrences early.

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# Budd–Chiari Syndrome Secondary to Burkitt's Lymphoma in a 5-Year-Old Child

Sadaf Ijaz[1], Amal Hasham[2], Aqeela Ayub[3], Masooma Ayesh[4], Maria Shamsher[5], Jaweria Zain[6]

## Abstract

**Summary:** We report a rare case of Budd–Chiari syndrome secondary to Burkitt's lymphoma in a 5-year-old male presenting with jaundice and abdominal pain. The case highlights the diagnostic challenges in pediatric patients presenting with hepatomegaly and cholestatic jaundice, and emphasizes the importance of considering underlying malignancy in atypical cases of hepatic venous outflow obstruction.

**Keywords:** Burkitt lymphoma, Budd Chiari syndrome, venous thromboembolism/ etiology.

## Introduction

Burkitt's lymphoma (BL) is a highly aggressive non-Hodgkin lymphoma (NHL) of B-cell origin, characterized by the translocation and deregulation of the MYC gene.[1,2] It accounts for 6–8% of childhood malignancies, with a median age of presentation around 10 years. Pediatric NHLs are predominantly high-grade, and BL is known for its rapid progression and frequent extranodal involvement, particularly of the abdomen, bone marrow, and central nervous system.[1]

Budd–Chiari syndrome (BCS) is a rare disorder caused by obstruction of hepatic venous outflow, which may be primary (due to venous abnormalities) or secondary (due to external compression or invasion by tumors).[3] Association of BL with BCS is extremely rare and can lead to diagnostic confusion, as both conditions may initially present with hepatomegaly and jaundice.

## Case Presentation

A 5-year-old male, resident of Bahria Town, Rawalpindi, presented to the emergency department with complaints of abdominal pain, anorexia, and yellow discoloration of the skin and sclera for 20 days. The pain was gradual in onset, intermittent, and generalized. There was no history of itching, clay-colored stools initially, fever, or bleeding tendencies.

**Past and Family History**

He had no history of previous transfusions, injections, or chronic illnesses. He was a product of normal vaginal delivery with normal developmental milestones. His immunization status was up to date. No family history of similar illnesses was reported.

**Examination**

The child appeared unwell and deeply icteric. Vitals were stable. Anthropometric measurements were between the 10th and 25th percentiles. There was no pallor, lymphadenopathy, or edema.

Abdominal examination revealed hepatomegaly (4 cm below the costal margin, firm, 13 cm span) and splenomegaly (1 cm below the costal margin). Ascites was clinically present, with shifting dullness positive.

**Investigations**

CBC: Hb 10.3 g/dL, WBC 9500/µL, Platelets 531,000/µL

LFTs: Total bilirubin 9.9 mg/dL (Direct 8.2 mg/dL), ALT 182 U/L, ALP 120.9 U/L

Serology: Hepatitis A, B, C, and E negative

Ultrasound: Mild hepatosplenomegaly with mild ascites

During hospitalization, jaundice worsened, and the child developed abdominal distension and tender hepatomegaly. Clay-colored stools and low-grade fever appeared.

**Advanced Imaging**

CT abdomen and pelvis revealed multifocal thrombosis of the inferior vena cava with hepatomegaly, nutmeg appearance of liver, and flip-flop pattern suggestive of Budd–Chiari syndrome, along with acute pancreatitis and right renal vein thrombosis.

A triphasic CT scan later showed periportal hypodensities, hepatic and renal hypodense lesions, and mesenteric lymphoid masses, suggesting a lymphoproliferative disorder.

Ascitic Fluid Analysis:

Protein 49 g/L, LDH 2296 U/L, ADA 34.3 U/L, with 80% lymphocytes and a few atypical cells.

Histopathology: Liver biopsy revealed sheets of monotonous intermediate-sized lymphoid cells with a "starry-sky" appearance, confirming Burkitt's lymphoma.

**Final Diagnosis:**

Budd–Chiari syndrome secondary to hepatic infiltration by Burkitt's lymphoma.

## Discussion

This case demonstrates a rare presentation of BL manifesting as Budd–Chiari syndrome. Burkitt's lymphoma can involve the liver either by direct infiltration or secondary thrombosis due to

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hypercoagulability and venous obstruction.[4] Pediatric lymphomas are known to induce a prothrombotic state, and studies report venous thromboembolism in up to 10–15% of cases within the first year of diagnosis.

In this patient, hepatic venous thrombosis and IVC obstruction likely resulted from lymphomatous invasion. The diagnosis was exigent due to overlapping symptoms of hepatic illness and the scarcity of such a presentation in children. Imaging and biopsy were crucial investigations that led to the diagnosis.

The gold standard for the diagnosis of lymphoma is Histopathology, which shows hyperchromatic monomorphic lymphoid cells with phagocytic histiocytes—the classic "starry sky" pattern.[5]

**Management and Outcome**

Burkitt's lymphoma management includes chemotherapy or chemoimmunotherapy, depending upon the disease stage. Prophylaxis for tumor lysis syndrome, febrile neutropenia, and monitoring for thrombotic complications remains the main supportive treatment.

The child was then referred to the pediatric oncology unit for further management and initiation of chemotherapy after the diagnosis was confirmed

## Conclusion

This case highlights the importance of considering unusuall diagnosis like malignancy in pediatric patients presenting with unexplained hepatic venous obstruction. Imaging and histological evaluation early in the course of disease are essential for making a timely diagnosis and management of rare diseases like Budd–Chiari syndrome secondary to Burkitt's lymphoma.

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# Postpartum Secondary PPH Leading to Diagnosis of Choriocarcinoma: A Case Report

Zainab Maqsood[1], Tallat Farkhanda[2], Sara Ejaz[3], Tayyaba Kousar[4], Sobia Nawaz Malik[5], Humaira Bilqis[6]

## Abstract

**Summary:** One uncommon but extremely aggressive kind of gestational trophoblastic neoplasia (GTN) is postpartum choriocarcinoma. Its clinical manifestation frequently resembles typical subsequent postpartum hemorrhage sources, which could impede identification and raise maternal morbidity.
Case Presentation:
A woman with a history of three prior cesarean sections appeared with recurring severe vaginal bleeding. Twelve days after an emergency cesarean delivery, Conservative management was started after an initial examination revealed retained products of conception. However, hypovolemic shock caused the patient's condition to worsen. Suspicion of GTN was raised by significantly high levels of β-human chorionic gonadotropin (β- hCG) in the serum and pelvic magnetic resonance imaging. An urgent surgical procedure was carried out since the bleeding was potentially fatal.

**Keywords:** Postpartum Hemorrhage, beta-Human Chorionic Gonadotropin, Cesarean Section, Choriocarcinoma, Gestational Trophoblastic Neoplasia

## Introduction

Choriocarcinoma is one of the most aggressive types of gestational trophoblastic neoplasia (GTN), which is a spectrum of malignant trophoblastic diseases.[1] Due to its rarity, postpartum choriocarcinoma may initially resemble more frequent causes of subsequent postpartum hemorrhage (PPH), delaying identification.[2] This case emphasizes how crucial it is to raise suspicions as soon as possible when postpartum bleeding is unusual or prolonged.

## Case Presentation

Twelve days following an emergency caesarean section, a 30-year-old lady, P3+0, who had three prior cesarean sections, began experiencing significant vaginal bleeding. Before experiencing two to three episodes of severe bleeding with clot passage during the next five days, she was well for the first fifteen postoperative days. She saw her treating physician on the twentieth postoperative day because she was still bleeding. She was actively bleeding but hemodynamically stable at the time of the initial assessment. She was given transaminic acid both orally and intravenously. She was prescribed misoprostol 200 µg TDS for seven days after a trans-abdominal ultrasound revealed the possibility of retained products of conception (RPOCs). She started taking misoprostol, but instead of getting better, her bleeding got much worse.
After that, she went to her obstetrician in hypovolemic shock, and prompt resuscitation was performed. Once more, a transvaginal ultrasound revealed a large postpartum uterus that raised the possibility of RPOCs. Doppler ultrasonography, conducted by a skilled radiologist, revealed no aberrant vascularity but did not reveal a distinct endomyometrial junction.
A significantly higher β-hCG level of 115,897 mIU/mL was found after additional testing. An MRI of the pelvis revealed a fluid-filled, enlarged endocervical canal with an ill-defined lobulated lesion affecting the anterior uterine wall. The findings were suggestive of an invasive mole or GTN and included subtle enhancement, invasion of the myometrium, loss of the junctional zone, and adjacent neovascularization. The patient experienced dizziness, sweating, pallor, hypotension (BP 90/60 mmHg), a weak pulse (110 bpm), chilly, clammy extremities, and sudden, profuse vaginal bleeding with clots on the third day of stay. For an urgent surgical procedure, she was taken to the operating room.
Hemoperitoneum was found to be between 100 and 200 mL. Both tubes and ovaries looked normal, and the uterus was the size of a 12-week gestation. Peritoneal washings were performed together with a complete abdominal hysterectomy, bilateral salpingo-oophorectomy (TAH+BSO), and infracolic omentectomy.
**Histopathology:**
A highly mitotic trophoblastic tumor made up of cytotrophoblasts, syncytiotrophoblasts, and intermediate trophoblasts with myometrial invasion, lymphovascular invasion, necrosis, and bleeding was visible under a microscope. There were no tumors in the omentum, ovaries, tubes, or cervix. The final diagnosis was uterine-confined gestational trophoblastic neoplasia, sometimes known as choriocarcinoma.
Follow-up: Beta HCG on 18th July 2024 came out to be 830miu/ml. Chronic carcinoma of Figo stage I with a scoring of 4 (low risk) was diagnosed, and the patient was referred to the oncology clinic for chemotherapy (methotrexate and folinic acid).

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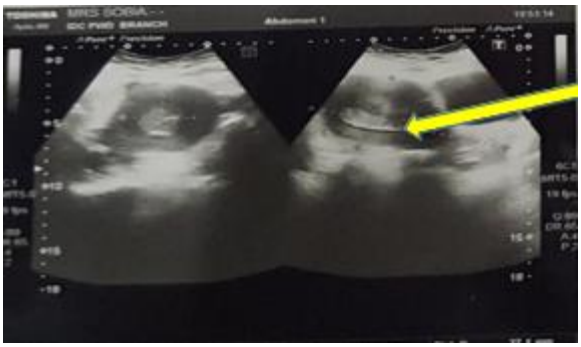

**Figure 1: Heterogeneous lesion predominantly hyperechoic measuring 37x 31 mm**

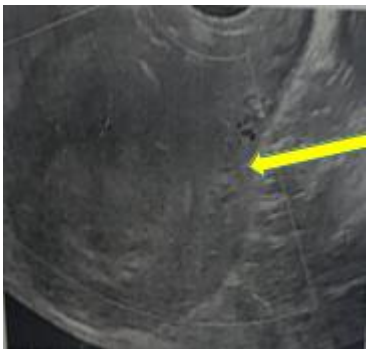

**Figure 2: Cystic solid mass in endolumen**

## Discussion

Rare as it is, postpartum choriocarcinoma sometimes mimics common causes of secondary PPH, delaying diagnosis.[2,3] Since GTN can happen after any pregnancy event, including term birth or caesarean section, early β-hCG testing should be prompted if bleeding persists or gets worse despite treatment for suspected RPOCs. Although results may be nonspecific, imaging modalities such as Doppler ultrasound and MRI are essential for distinguishing RPOCs from GTN.[4] Since choriocarcinoma is extremely chemosensitive and has excellent results when treated promptly, early detection is essential. As demonstrated in this instance, where surgical intervention proved to be life-saving, failing to detect GTN may result in catastrophic bleeding.[5]

## Conclusion

This case shows that GTN should be suspected in cases of chronic postpartum bleeding that do not respond to conventional treatment. It is essential to use sophisticated imaging and β-hCG for early assessment. In this instance, prompt diagnosis and surgical intervention saved lives. Even though it is uncommon, postpartum choriocarcinoma needs to be considered when making a differential diagnosis for subsequent PPH.

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# Exploring the Intricacies and Clinical Perspectives of Chronic Diarrhea: A Case Report

Tanveer Hussain[1], Anum Abbas[2], Sadia Ahmed[3], Muhammad Umar[4]

## Abstract

**Summary:** Crohn's disease (CD) is a chronic inflammatory disorder that may affect any segment of the gastrointestinal tract, with a predilection for the terminal ileum and colon. It commonly presents chronic diarrhea, abdominal pain, and per rectal (PR) bleeding. We report the case of a gentleman who experienced recurrent diarrhea and PR bleeding, initially managed as infective colitis but subsequently diagnosed with Crohn's disease following colonoscopy and histopathological confirmation. He was treated successfully with corticosteroids for induction and azathioprine for maintenance therapy. This case emphasizes the need to consider Crohn's disease as a differential diagnosis in patients with chronic diarrhea and PR bleeding, particularly in regions where infectious causes are more prevalent, and may delay diagnosis.

**Keywords:** diarrhea, disease, bleeding.

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## Introduction

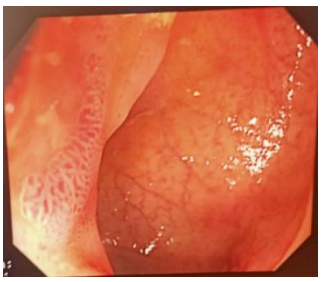
Crohn's disease (CD) is one of the two main forms of inflammatory bowel disease (IBD). It may involve any part of the gastrointestinal tract, most often the terminal ileum and proximal colon, and is characterized by transmural inflammation, skip lesions, strictures, and fistulae.[1]
The global incidence of CD continues to rise, particularly in newly industrialized regions of Asia, the Middle East, and South America.[2] In Western countries, incidence ranges from 0.1 to 16 per 100,000 person-years, but recent studies indicate a growing burden in South Asia.[3]
The pathogenesis is multifactorial, involving genetic susceptibility, mucosal immune dysregulation, environmental triggers, and gut microbiota.[4] Genome-wide association studies have identified key genes including NOD2, ATG16L1, and IL23R, highlighting the role of innate immunity and microbial recognition.[5]
Diagnosis remains challenging in infection-endemic settings where CD may mimic intestinal tuberculosis, infectious colitis, or ulcerative colitis. Colonoscopy typically demonstrates longitudinal ulcers, cobblestoning, skip lesions, and strictures, supported by cross-sectional imaging and histology.[6] Most patients develop progressive disease requiring immunosuppressants or biologics, while only 20–30% follow an indolent course.[8]

## Case Presentation

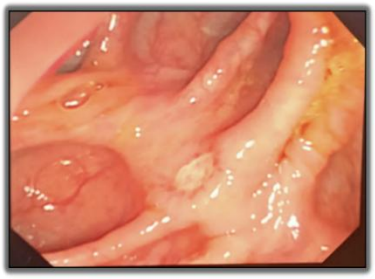
A 44-year-old gentleman from Azad Kashmir presented to the gastroenterology outpatient clinic with a 2-year history of chronic diarrhea. The diarrhea was of small volume, loose in consistency (Bristol stool scale type 6–7), occurring intermittently, and occasionally mixed with blood. Over the past 3 months, he also developed abdominal pain. The pain was dull, continuous throughout the day, and significantly worsened after meals, reaching an intensity of 7–8/10 on the pain scale. The diarrheal episodes were frequently associated with urgency, tenesmus, and abdominal bloating. He denied any history of weight loss, extraintestinal manifestations such as arthralgia, aphthous ulcers, or skin lesions. He had sought medical care at several centers in Saudi Arabia and local hospitals, where he was managed empirically with antibiotics, antispasmodics, and probiotics, but his symptoms persisted without improvement. On examination, he appeared thin and lean, with a BMI of 24 kg/m². There was mild tenderness in the right iliac fossa, but no guarding or signs of peritonism. Perianal inspection revealed a small skin tag, with no evidence of abscess or fistula.

**Investigations**

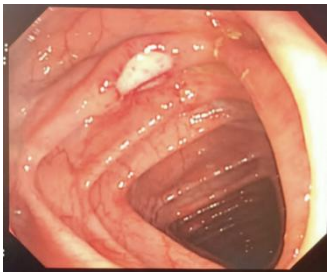
**Blood tests:** Hemoglobin 13.3 g/dL, MCV 74 fL, CRP 36 mg/L, ESR 52 mm/hr. LFTs and renal functions were normal. His TSH, Anti-Ttg (IgA), ANA c, and p- ANCA were also normal, but ASCA was positive, with Fecal calprotectin 250ug/g and Vitamin B12 150pg/dL and Folic acid 2.1ng/mL.
**Stool culture:** It was negative for Clostridium difficile toxin.
**Colonoscopy:** There were multiple deep ulcers with surrounding erythema in the terminal ileum, ascending colon, and hepatic flexure with intervening normal-looking mucosa, but the transverse, descending, sigmoid colon, and rectum were normal.



Terminal Ileum

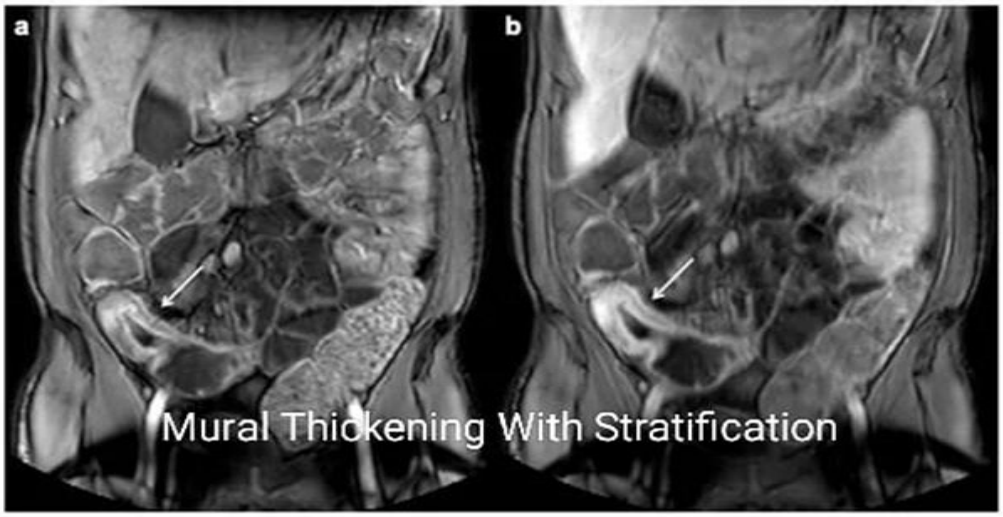


Cecum



Ascending Colon

**Histopathology:** There was focal chronic inflammation with mild cryptitis and crypt abscesses, ulceration, and no granulomas.
**Imaging:** MR enterography revealed jejunal wall segmental thickening with delayed mural enhancement, suggestive of IBD.



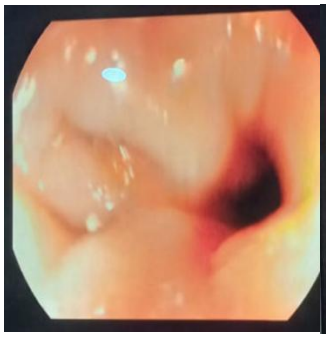

**Crohn's disease (Montreal classification A2 L3 B1).**

**Treatment**

The patient was put on budesonide 9mg once daily orally for 8 weeks; the patient's symptoms were completely resolved within two months. He started azathioprine 2 mg/kg for maintenance therapy. Cobalamin, folic acid, and vitamin D deficiency were also replaced.

**Outcome And Follow-Up**

After 8 weeks, the patient had complete resolution of diarrhea, bleeding, and abdominal pain, normalization of CRP ( 6mg/L) and fecal calprotectin (50 µg/g), and repeat colonoscopy at 12 weeks showed mucosal healing. He remained in clinical remission at 12 months on azathioprine monotherapy.



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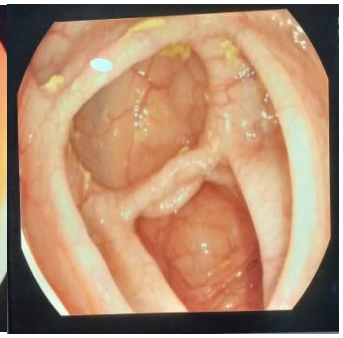

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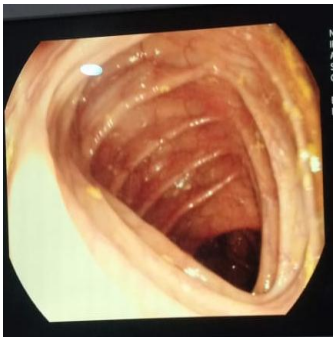

Ascending Colon

## Discussion

This case illustrates the diagnostic challenges in regions endemic for infectious diseases. The absence of granulomas on histology initially raised suspicion for an infective etiology. However, the chronic nature of symptoms, persistently elevated fecal calprotectin, positive ASCA serology, and colonoscopic evidence of skip lesions were more typical of Crohn's disease (CD). Granulomas are identified in only 15–65% of CD biopsies and are not essential for diagnosis.[5]

Endoscopic findings such as aphthous ulcers, longitudinal fissures, skip lesions, and ileocecal involvement are highly characteristic of CD.[6,7] Radiological features, including segmental mural thickening and delayed mural enhancement on MR enterography, provide further diagnostic support.[9,10]

Management of CD has shifted considerably in recent decades. Corticosteroids were once the cornerstone of induction therapy, but the introduction of biologics has transformed treatment strategies.[12] Current guidelines advocate an individualized approach: budesonide is recommended for mild-to-moderate ileocecal disease, while systemic corticosteroids, immunomodulators, and biologics (anti-TNF, anti-integrin, and anti-IL-12/23 agents) are reserved for more extensive or refractory cases.[11]

Therapeutic objectives now follow a treat-to-target model, moving beyond symptom relief. This includes short-term clinical remission, intermediate biomarker reduction, and long-term mucosal healing demonstrated by colonoscopy.[13,14] Mucosal healing is associated with reduced hospitalization, need for surgery, and colorectal cancer risk. In our patient with mild ileocolonic disease, budesonide monotherapy with dietary support achieved both clinical and endoscopic remission. Early recognition and timely therapy in such cases are vital to prevent disease progression and long-term complications.

**Learning Points:**

- Crohn's disease should be suspected in chronic diarrhoea even in infection-endemic regions.
- Endoscopic and radiological features are more reliable than granulomas for diagnosis.
- Budesonide is effective in mild ileocolonic disease.
- Treat-to-target strategies improve long-term outcomes.

## Author Information

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# Topical Steroid Misuse: An Unrecognized Cause Of Iatrogenic Cushing Syndrome In Infancy

Verda Imtiaz[1], Naghmana Iftikhar[2], Hina Sattar[3], Noshina Riaz[4]

## Abstract

**Summary:** Cushing's syndrome represents a condition of excess cortisol activity, arising either from internal overproduction or external administration of glucocorticoids, leading to a wide range of clinical features.[1] Cushing's syndrome in infancy is rare and most often iatrogenic. We report an infant who presented with rapid-onset weight gain and hypertrichosis due to chronic application of an over-the-counter steroid-containing ointment for diaper rash. Recognition of exogenous steroid exposure allowed timely withdrawal and clinical recovery. This case highlights the importance of medication history and the regulation of topical corticosteroid sales in resource-limited developing countries.

**Keywords:** Cushing Syndrome; Glucocorticoids; Administration, Topical; Infant; Iatrogenic Disease; Adrenal Insufficiency.

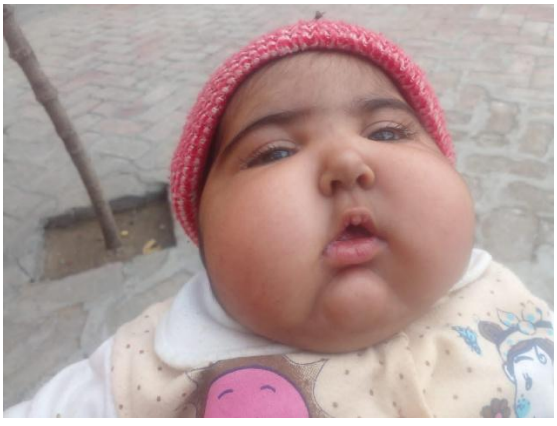
## Introduction

Infants are particularly vulnerable to developing topical corticosteroid–induced iatrogenic Cushing's syndrome because of their thin, highly permeable skin, a relatively greater surface area–to–body weight ratio, and the frequent occurrence of dermatologic conditions requiring topical therapy. Most reported iatrogenic cases have involved the use of potent preparations such as clobetasol or betamethasone for diaper dermatitis. The clinical manifestations in iatrogenic cases are similar to those of endogenous Cushing's syndrome. Characteristic findings include moon-shaped facies, abnormal weight gain, truncal adiposity with supraclavicular and subscapular fat accumulation, excessive hair growth, facial plethora, and skin fragility with telangiectatic changes. Complications related to mineralocorticoid or androgenic activity—such as hypertension, acne, and hirsutism—are less frequently encountered than in endogenous disease. Abrupt cessation of exogenous steroid exposure may precipitate acute adrenal insufficiency, which can be life-threatening if unrecognized.

## Case Presentation

A previously healthy 9-month-old female infant presented to the Paediatric Medicine outpatient clinic with progressive weight gain and facial puffiness for 2 months. The parents initially perceived the chubby cheeks as healthy growth, but became concerned as the weight continued to rise rapidly. They also noticed excessive hair growth over the forehead, nape of the neck, periauricular area, and trunk. There was no history of irritability, vomiting, seizures, polyuria, polydipsia, abdominal pain, cough, dyspnoea, oedema, or jaundice. There was no prior systemic steroid use. Birth History was unremarkable, with birth weight 3 kg, uneventful perinatal course. Past History also not significant. Developmental milestones were appropriate for age. She was fully immunised. She was breast-fed until 6 months, and from 6 months onwards was on complementary feeds also. On further enquiry, parents revealed daily use of a commercially available nappy-rash ointment for 4 months. The cream was later identified to contain a potent fluorinated corticosteroid, Clobetasol.

On examination, the child was active, playful, cushingoid with rounded plethoric facies, central obesity, and hypertrichosis. She was vitally stable with a heart rate of 120/min, respiratory rate 34/min, and BP 80/40 mmHg (normal for age). There were no striae, acne, skin thinning, oedema, hypertension, hepatosplenomegaly, or features of virilization.



**Contributions:**
VI NI - Conception, Design
NI HS NR - Acquisition, Analysis, Interpretation
VI NI - Drafting
HS NR - Critical Review

All authors approved the final version to be published & agreed to be accountable for all aspects of the work.

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**Differential Diagnosis**
- Iatrogenic Cushing's syndrome from exogenous steroids
- Adrenocortical tumour (adenoma/carcinoma)
- Pituitary-dependent Cushing's disease
- Ectopic ACTH production
- Obesity/metabolic syndrome

**Investigations**
Morning serum ACTH was quite suppressed, i.e., 1.5 pg/mL (reference 7.2–63.3 pg/mL), while random blood sugar, serum electrolytes, thyroid profile, HbA1c, and lipid profile were within normal limits. No imaging was performed as adrenal pathology was unlikely given suppressed ACTH and a clear drug history.

**Treatment**
The offending topical steroid ointment was discontinued immediately. Oral Hydrocortisone was started in physiologic doses, i.e., 10mg/m2/day in 3 divided doses, as such patients can go into adrenal crises if steroids are withdrawn immediately. Parents were counseled on avoiding unsupervised steroid use. Supportive skin care for diaper dermatitis was advised.

**Outcome And Follow-Up**
At the 4-week follow-up, the patient showed a gradual regression of cushingoid facies, reduction in hypertrichosis, and reduction in weight. Vital signs and developmental progress remained normal. Long-term follow-up was planned to monitor for hypothalamic–pituitary–adrenal axis recovery and growth parameters.

## Discussion

In developing regions, unsupervised or inappropriate use of pharmacological agents remains prevalent because of inadequate regulation of over-the-counter drug sales and the use of potent corticosteroid-containing ointments for diaper rash. Management centres on the withdrawal of the exogenous steroid slowly, treatment of the primary dermatologic condition, and monitoring for adrenal insufficiency during recovery. Public health measures, including the regulation of over-the-counter steroid sales and parent education, are crucial to prevent such cases. Pediatricians should maintain a high index of suspicion for steroid-induced Cushing's syndrome in infants presenting with cushingoid appearance, particularly if topical preparations have been used in the diaper area. Caregivers should always be questioned about the use of any creams or ointments applied to the skin.[3]

**Learning Points:**
- Always obtain a detailed drug history, including topical agents, in any child with cushingoid features.
- Topical steroids under occlusion in infants can lead to systemic Cushing's syndrome.
- Early recognition and gradual withdrawal of the offending agent usually result in reversal of clinical features without long-term sequelae.
- There is a pressing need for stricter regulation of potent topical corticosteroids in resource-limited settings.

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# Eagle's Syndrome: A Diagnostic Dilemma in Chronic Throat and Neck Pain

Ashar Alamgir[1], Tabassum Aziz[2], Aimen Sohail[3]

## Abstract

**Summary:** Eagle's syndrome is caused by either elongation of the styloid process or calcification of the stylohyoid ligament. It is a rare and important cause of chronic neck, pharyngeal, and cervicofacial pain. Its vague symptoms often simulate neurologic pain and temporomandibular joint disorders, delaying diagnosis. We present a case report of a 21-year-old male with a one-year history of neck and facial pain that was not responding to medication. It was eventually diagnosed on imaging and successfully managed by styloidectomy via trans-oral approach. This case was selected because such cases often remain undiagnosed for many months due to symptoms that cannot be attributed to any routine disorders. This case report highlights the importance of delayed diagnosis and reinforces the need for a detailed history, comprehensive intra-oral examination, and investigations in cases of persistent neck pain.

**Keywords:** eagle's syndrome, elongated styloid process, stylohyoid.

## Introduction

Eagle's syndrome is characterized by an elongated styloid process (>30 mm) or calcification of the stylohyoid ligament.[1] Its classic symptoms comprise recurrent throat pain, dysphagia, foreign-body sensation, and earache.[2] Its presentation mimics glossopharyngeal neuralgia, trigeminal neuralgia, and temporomandibular joint (TMJ) disorders, which frequently result in diagnostic confusion and extending patient's ailment.[3]
CT scan is the diagnostic gold standard modality with orthopantomogram and 3D reconstructions as supplemental tools.[4] Surgical options like styloidectomy are the definitive treatment for symptomatic patients, performed using either trans-oral or extraoral approaches.[5] This report presents a young male with chronic throat pain, initially misdiagnosed as neuralgia, who ultimately was diagnosed as Eagle's syndrome and achieved an excellent outcome following surgical management.[6]

## Case Presentation

A 26-year-old male student from Attock presented to the ENT OPD of Holy Family Hospital on October 25, 2024, with a one-year history of pain in the throat radiating to the right side of his face. The pain was intermittent, moderate to severe, and exacerbated by swallowing, opening his mouth, and moving his neck. He also reported a persistent foreign body sensation in the throat. His symptoms were temporarily relieved by over-the-counter analgesics.
Over the previous year, he had consulted multiple physicians, a dentist, and an oral maxillofacial surgeon, receiving treatments for trigeminal neuralgia, glossopharyngeal neuralgia, and dental issues without success. His past medical, surgical, and personal history was unremarkable.
On examination, the patient was vitally stable. Extra-oral examination revealed that the face was symmetrical. No palpable mass or tenderness on the face, neck, or in the muscles of mastication during palpation. No tenderness was elicited in the temporomandibular joints during mandibular movements. Intra-oral examination revealed marked tenderness and a palpable bony lesion in the right tonsillar fossa.

**Investigations**

Baseline investigations, like CBC, LFTs, RFTs, ESR/CRP, were within normal limits. An orthopantomogram (OPG) was advised, in which the styloid process was elongated. A CT scan of the base of the skull to the clavicle was done to see the length of the styloid process. It was significantly enlarged (35 mm).

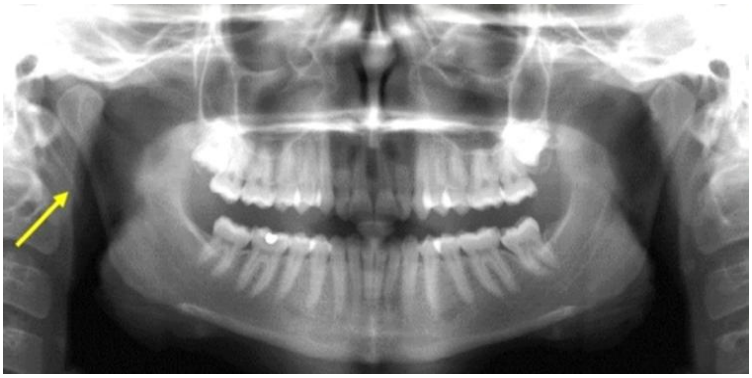

**Figure 1: An orthopantomogram (OPG) is done to see the elongated styloid process. An elongated styloid process can be seen in this OPG**

**Contributions:**
AA- Conception, Design
TA AS - Acquisition, Analysis, Interpretation
TA AS - Drafting
AA - Critical Review

All authors approved the final version to be published & agreed to be accountable for all aspects of the work.

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**Differential Diagnosis**
glossopharyngeal neuralgia, trigeminal neuralgia, temporomandibular joint (TMJ) disorders
**Management**
After counseling the patient, informed consent was acquired for surgical intervention. The patient underwent styloidectomy via trans-oral approach of the right styloid process under general anesthesia. The elongated portion was carefully amputated and removed with the preservation of the neurovascular bundle. Hemostasis was achieved, and the wound was closed primarily. The patient had an uneventful recovery and was discharged on the third postoperative day with a prescription for oral antibiotics, analgesics, and antiseptic mouthwash.
**Outcome And Follow-Up**
The patient was followed up at one-week and one-month with complete resolution of pain and foreign-body sensation. The surgical site healed well without any complications.

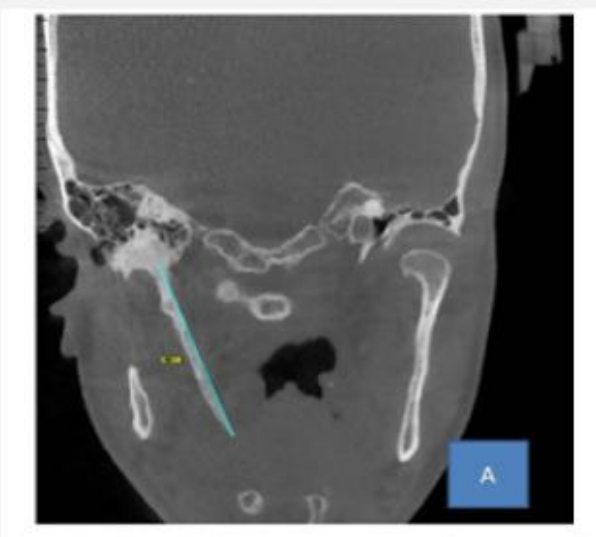


**Figure 2: A CT scan of the neck is done to measure the length of the styloid process. This image shows elongation of the right styloid process**

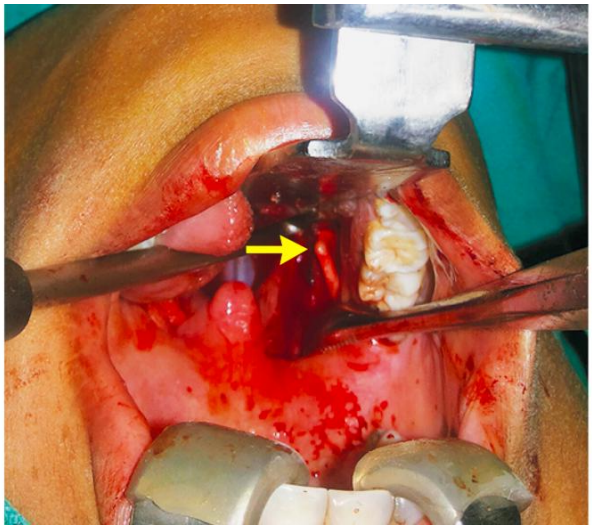

**Figure 3: Styloidectomy being done under general anesthesia via transoral approach**

## Discussion

The diagnosis of Eagle's syndrome has always been a great challenge, as its symptoms correspond with neurological pain and dental pathologies.[7] In our case, the patient underwent multiple assessments and treatments before the definitive diagnosis was discovered. The mainstay of diagnosis is hidden in clinical suspicion and careful palpation of the tonsillar fossa, which may potentially elicit tenderness over a bony prominence.[8] Although an OPG can raise suspicion, a CT scan is the gold standard diagnostic tool, clearly demonstrating the styloid length and its relation to nearby structures.[9] Management options comprise conservative therapy with analgesics and steroids; however, surgical excision, i.e., styloidectomy, constitutes the definitive management for symptomatic patients.[10]
The trans-oral approach, utilized in our case, has the advantages of no external scarring and reduced tissue dissection.[11] While it demands a meticulous surgical approach to avoid neurovascular injury. The excellent outcome in our patient affirms its safety and efficacy.
Eagle's syndrome should be considered in patients with unexplained and refractory throat pain or facial pain.
Careful intra-oral examination of the tonsillar fossa is the cornerstone for early diagnosis. A CT scan is the gold standard investigation, confirming elongation of the styloid process. Trans-oral styloidectomy offers a cosmetically favorable and effective treatment option

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# When Identity Feels Divided: A Case Report On Gender Dysphoria

Asad Tamizuddin Nizami[1], Ifrah Javed Malik[2], Kainat Kareem Mirza[3], Zarnain Umar[4]

## Abstract

**Summary:** A psychological condition in which an individual's feeling of gender is not in congruence with the assigned biological sex is known as gender dysphoria.
We shall present the case of a 17-year-old girl who presented with primary amenorrhea to the endocrinology department and was referred to psychiatry. This case report highlights the importance of a multidisciplinary approach.

## Introduction

A psychological condition in which an individual's own feeling of gender is not congruent with the assigned biological sex is known as gender dysphoria. About two-year-old children have gender dysphoria, and it persists into adolescent years, which accounts for between 12% and 27% of the cases.[1]
Gender dysphoria is related to 5 alpha reductase deficiency, a rare cause of disorder of sexual development in males. It is presented as a 46XY karyotype. This case of a 17-year-old girl highlights the importance of a multidisciplinary approach to mitigate the distress and suffering of the individual.

## Case Presentation

A 17-year-old female patient, educated till 6th grade, resident of Rawalpindi, was referred to the Institute of Psychiatry, Benazir Bhutto Hospital, Rawalpindi, in January 2025 by the Endocrinology department of Holy Family Hospital, where she was admitted for evaluation of primary amenorrhea.
The patient presented with 3 years of distress during early adolescence (13-14 years of age). The patient had a strong desire to be treated as male. The informant reported that the patient had no genital ambiguity at the time of birth, but the secondary sexual characteristics were absent, and the patient had primary amenorrhea. The patient was legally and socially raised as female.
As the patient was distressed by being perceived as female, it resulted in social withdrawal and reduced interest in activities. At 15 years of age, the patient noted phallic enlargement concurrent with the first testicular descent.
On examination, the breast was not developed. The pubic hair was absent. The external genitalia were ambiguous. There was a clitoromegaly/phallic-like structure and vaginal slit.
On mental state examination, a young girl of stated age, wearing an abaya and veil, had feminine features. Her thought content as she stated.

مجھے سمجھ نہیں آتا میں کیا ہوں ...میں کون ہوں ...نہ مجھے میرا جسم ٹھیک لگتا ہے، نہ مجھے اپنا آپ باقی لڑکیوں جیسا لگتا ہے۔ میں کبھی نارمل زندگی گزار سکوں گی؟

**Investigations:**
The results of imaging karyotyping and hormonal profile are mentioned in Tables 1 and 2, respectively.

**Table 1: Results of Abdominal Ultrasound, Pelvic Ultrasound, and Cytogenetic Study**

| Investigation | Findings |
|---|---|
| **Abdominal Ultrasound** | Unremarkable |
| **Pelvic Ultrasound** | A soft tissue was visualized posterior to the urinary bladder and anterior to the rectum, likely representing the prostate with a volume of 8ml.<br>Normal testis visualized bilaterally in a **malformed scrotal sac.** |
| **Cytogenetic study** | **46XY Karyotype** |

Using the Revised Child Anxiety and Depression Scale, scores indicated borderline to clinical levels of generalized anxiety and depression.
The IQ was assessed using Raven's Standard Progressive Matrices- average range (55th percentile). No evidence of cognitive impairment.
Based on the detailed evaluation, the patient was diagnosed with Disorders of Sex Development (DSD)- 5 Alpha Reductase Deficiency with Gender Incongruence (ICD-11) or Gender Dysphoria (DSM-5-TR)

**Contributions:**
ATN IJM KKM ZU - Conception, Design
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**Treatment:**
The patient was recommended gender affirming surgery to align the patient's physical characteristics with male gender identity, as advised by the Endocrinology department. A multidisciplinary team was involved, including an endocrinologist, plastic surgeon, gynecologist, psychiatrist, and radiologist.

**Table 2: Hormonal Profile**

| Name Of Test | Level |
|---|---|
| **Serum Estradiol** | 21.50 pg/ml |
| **Ratio of Testosterone/DHT** | 21 |
| **LH** | 5.1 mIU/mL |
| **Testosterone** | 315 ng/mL |
| **Serum 17 OH Progesterone** | 0.17 ng/mL |
| **Dihydrotestosterone** | 149.39pg/ml |

## Discussion

According to DSM-5, Gender dysphoria is a psychological condition in which a person feels incongruent with one's biological gender. This shows that the definition for gender dysphoria in children and adults is the same. It is like our case in which a young girl had primary amenorrhea and felt incongruent with her gender.

A rapid rise has been seen in the number of children and adolescents presenting with gender dysphoria in their dysphoria/incongruence or identifying as transgender in several countries in the last 10 years. The prevalence of gender dysphoria has shown a rapid rise in birth-registered females. The most prevalent mood disorder was gender dysphoria was depression, which was more common in females.[3] The patient who presented to us was being raised as a female. She was expected to fulfill the social responsibility as a female, which resulted in her mood symptoms.

The prevalence of gender dysphoria is increasing, particularly in children. The factors that cause this increase are the surge in role models like gender non-conforming celebrities and media exposure. As the children are not legally allowed to give informed consent, they rely on caregivers and health professionals to make decisions on their behalf. The dysphoria is accompanied by psychosocial stressors and psychiatric illnesses. Although it is important to diagnose gender dysphoria, the psychiatric comorbidities persist over a long time and affect the future of the child.[1]

The deficiency of 5-alpha reductase is a rare cause of sex development-related disorders.

This enzyme is responsible for converting testosterone to 5α-dihydrotestosterone (DHT). The absence leads to impaired virilization and phenotypic ambiguity. The deficiency of the second type of 5α-reductase enzyme affects urogenital development. About 60% of the patients with ambiguous genitalia were raised as females who transitioned to male during puberty. Studies have shown that early diagnosis can limit medical and psychosexual complications due to gender dysphoria.[2]

The psychiatric comorbidities include depression, anxiety, personality-related disorders, and suicidality. It is recommended to have delayed surgery until the gender identity is confirmed. Family counselling is very important for irreversible genital surgery. It is a difficult and delicate task for a multidisciplinary team.[4]

Our case highlights that the presence of 5-alpha-reductase deficiency and gender dysphoria necessitates a holistic and multidisciplinary approach. This report aims to contribute to the growing understanding of gender dysphoria, emphasizing the crucial role of empathy, patient autonomy, and collaborative management in clinical practice. Continued awareness, education, and research in this domain are essential to ensure compassionate and evidence-based care for all individuals navigating gender identity.

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# Unmasking The Culprit: Recurrent Nosebleeds From Masson's Tumor

Javeria Awan[1], Sundas Masood[2], Haitham Akaash[3], Sadia Chaudhary[4]

## Abstract

**Summary:** Masson's tumor, also called Intravascular papillary endothelial hyperplasia (IPEH), is a benign condition affecting various parts of the body; nonetheless, sinonasal cavity involvement is uncommon.
We present the case of a 32-year-old male who experienced recurrent epistaxis and frontal headaches for 13 years. Despite multiple nasal surgeries, his symptoms persisted without a confirmed diagnosis. Physical examination revealed numerous adhesions near the middle turbinate, which were more pronounced on the left side. A contrast-enhanced CT scan revealed a heterogeneously enhancing soft-tissue lesion in the left maxillary sinus that extended into the nasal cavity, with erosion of the sinus walls. Histopathology confirmed Masson's tumor. The patient underwent several surgeries, including lateral rhinotomy and excision of a fleshy mass in the maxillary cavity. One-year post-surgery, there was no recurrence or re-bleeding.
IPEH of the sinonasal region should be part of the differential diagnosis of recurrent epistaxis; otherwise, it can present an ENT surgeon with a diagnostic conundrum.

**Keywords:** Nasal cavity, Epistaxis, Maxillary sinus, Nasal surgical procedures, Vascular malformations.

## Introduction

Masson's tumor, also known as intravascular papillary endothelial hyperplasia, is a rare entity that originates in various sites of the body, including the Head and Neck, digits, and trunk. Until now, only a few cases have been reported in the Sinonasal cavity, particularly the maxillary sinus. It is a benign vascular malformation that results in thrombus formation, inflammation, and vascular stasis within a vessel, leading to endothelial cell proliferation.[1] This paper describes a case of recurrent epistaxis diagnosed as Intravascular papillary endothelial hyperplasia of the Maxillary sinus.

## Case Presentation

A 32-year-old male, known hypertensive, presented with recurrent epistaxis for the past 13 years. It was associated with a Frontal Headache. There was no history of trauma or Visual Disturbances.
The patient underwent multiple Nasal surgeries over the course of 13 years, which led to no relief of his symptoms nor a confirmed diagnosis of his pathology.
The physical examination revealed no definite mass in the nasal cavities; however, multiple adhesions were noted near the middle turbinate, which were more pronounced on the left side.

### Investigations

A computed tomography scan with contrast showed a heterogeneously enhanced soft tissue lesion measuring about 2.7 x 2.3 x 1.8 cm in the left maxillary sinus.It is shown to expand into the left nasal cavity, accompanied by erosion of the medial and anterior walls of the maxillary sinus.
The histological analysis indicated intravascular proliferation of reactive endothelial cells, resulting in many papillary forms lined by a single layer of endothelial cells, thus validating the diagnosis of Masson's tumor.

### Treatment

The patient underwent multiple surgeries, including a previous lateral rhinotomy. In addition, his anterior and medial maxillary walls were deficient, providing complete access to the maxillary cavity. A visible fleshy mass was present along the anterior half of the cavity, which was then excised. A nasal endoscopy was done, and a point of origin in the apex of the maxillary cavity was identified and removed.
There has been no recurrence or rebleeding at a one-year follow-up.

## Discussion

Intravascular papillary endothelial hyperplasia (IPEH), also known as Masson's tumor, is an uncommon benign vascular lesion characterized by reactive endothelial proliferation and thrombus formation. Since its initial description in 1923, it has been documented in numerous anatomical sites; however, involvement of the sinonasal cavity remains exceedingly rare. Recent reviews consistently highlight the infrequency of maxillary sinus involvement, with fewer than 20 cases documented in the English literature to date.[3,11,12]
IPEH typically arises within the lumen of dilated vessels, pre-existing vascular malformations, or organizing hematomas, supporting its classification as a reactive rather than neoplastic process.[2] Contemporary studies reaffirm that endothelial proliferation is secondary to vascular stasis and thrombosis, rather than uncontrolled cellular atypia.[3,11]

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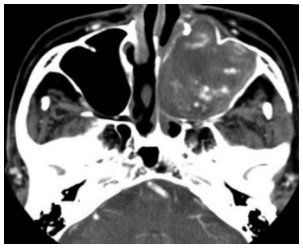

**Figure 1 Axial CT showing a soft tissue lesion in the left maxillary sinus extending into the left nasal cavity**

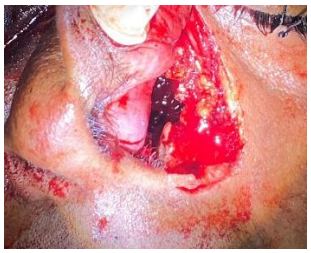

**Figure 2 (A) shows an Open lateral rhinotomy owing to past surgeries**

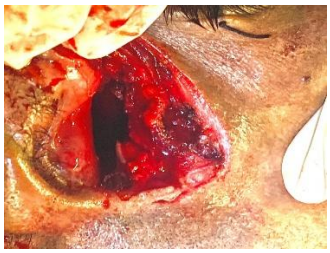

**Figure 2 (B) shows an anteriorly deficient Maxillary wall on an Open Lateral rhinotomy.**

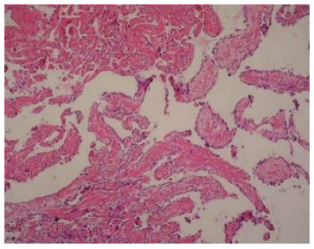

**Figure 3: Histopathological image showing papillary structures in a vessel (H&E x 40)**

Clinically, sinonasal IPEH presents with nonspecific symptoms depending on the site and extent of involvement. Recurrent epistaxis remains the most frequently reported presenting complaint, followed by nasal obstruction, rhinorrhea, facial pain, headache, epiphora, and anosmia.[11] Radiologically, IPEH poses a diagnostic challenge due to its aggressive-appearing features. Recent reports consistently describe heterogeneously enhancing sinonasal masses on contrast-enhanced CT, often accompanied by bone remodeling or erosion, findings that may raise suspicion for malignancy.MRI has been shown to provide superior soft-tissue characterization, typically demonstrating intermediate T1 and heterogeneous T2 signal intensities with contrast enhancement, aiding in surgical planning and assessment of local extension.[12]

**Table 1: Reports on maxillary IPEH in literature**

| Author | Year of publication | Age/sex | Location | Symptoms | Imaging | Surgical techniques |
|---|---|---|---|---|---|---|
| **Stern et al.[6]** | 1991 | 17/M | Right maxillary sinus, ethmoid, and nasal cavity. | Frontal headache, pain in the right cheek, proptosis. | CT | Caldwell Luc excision. |
| **Lancaster et al.[7]** | 1998 | 67/F | Left maxillary sinus and ethmoid. | Left nasal blockage, rhinorrhea, postnasal discharge, cheek pain. | CT | Endoscopic excision. |
| **Wang et al.[8]** | 2009 | 42/M | Left maxillary sinus, ethmoid, frontal, and nasal cavity. | Left nasal blockage, rhinorrhea, frontal headache, epistaxis. | CT/MRI | Endoscopic excision. |
| **D'Aguanno et al.[9]** | 2019 | 67/F | Right maxillary sinus, nasal cavity. | Right cheek pain, rhinorrhea, and postnasal drip. | CT/MRI | Caldwell Luc, and endoscopic excise. |
| **Cooke et al.[10]** | 2020 | 28/M | Right maxillary sinus, B/L nasal cavity, and B/L ethmoid sinus | B/L nasal blockage, epistaxis, headache, Right cheek pain, and itchy eyes. | CT | Endoscopic excision and skull base repair. |
| **Voruz F et al.[3]** | 2020 | 46/M | Left maxillary sinus | Left nasal blockage and bloody serous rhinorrhea. | CT/MRI | Endoscopic excision. |
| **Voruz F et al.[3]** | 2020 | 76/M | Right maxillary sinus, nasal cavity. | Epistaxis and rhinorrhea. | CT/MRI | Endoscopic excision. |
| **Voruz F et al.[3]** | 2020 | 33/F | Right maxillary sinus. | Rhinorrhea, orbital pressure, and headache. | CT/MRI | Endoscopic excision. |
| **Nakamura et al.[11]** | 2023 | 58/M | Left maxillary sinus, B/L nasal cavity, and B/L ethmoid sinus. | B/L nasal blockage, epiphora, epistaxis. | CT/MRI | Endoscopic excision. |
| **Maiti et al.[12]** | 2025 | 29Y/M | Right nasal cavity, nasopharynx, maxillary sinus | Right-sided nasal obstruction, blood-tinged mucopurulence | CT/MRI | Endoscopic excision |
| **Present case** | 2025 | 32/M | Left maxillary sinus and left nasal cavity. | Epistaxis. | CT | A combined approach of open lateral Rhinotomy and endoscopic excision. |

The differential diagnosis of sinonasal IPEH is broad and includes angiosarcoma, inverted papilloma, inflammatory polyps, lymphoma, squamous cell carcinoma, and metastatic disease.[4] Among these, angiosarcoma represents the most critical diagnostic pitfall due to its close histologic resemblance. Recent literature highlights the importance of correlating radiologic, histopathologic, and immunohistochemical findings to avoid overtreatment.[9,11]

Definitive diagnosis relies on histopathological examination, which typically reveals papillary fronds composed of a single layer of bland endothelial cells lining fibrinous cores, with minimal atypia and absence of necrosis. Immunohistochemistry consistently demonstrates positivity for endothelial markers such as CD31, CD34, and factor VIII–related antigen, findings that have been reconfirmed in recent series.[3,5,12]
Surgical excision remains the treatment of choice, with complete resection being curative in nearly all reported cases. Recent studies support endoscopic excision as the preferred approach when feasible, offering excellent visualization with minimal morbidity. However, open or combined approaches may be necessary in cases with extensive disease, previous surgeries, or compromised anatomy, as demonstrated in our patient.[11,12] Recurrence is exceedingly rare and is typically associated with incomplete excision.
This case underscores the importance of considering IPEH in the differential diagnosis of long-standing recurrent epistaxis, particularly when imaging reveals a vascular sinonasal mass with bone erosion. Awareness of this rare entity among otolaryngologists, radiologists, and pathologists is essential to ensure accurate diagnosis, appropriate surgical management, and avoidance of unnecessary aggressive treatment.

Learning Outcomes
• Despite its rarity in the nasal cavity, IPEH should be considered in cases of recurrent epistaxis and included in the differential diagnosis of hemorrhagic nasal masses.
• While radiologic imaging is crucial, a definitive diagnosis requires histopathological examination.
• The treatment involves complete surgical excision.

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# Otosclerosis: Presentation, Diagnosis And Management

Khadija Mazhar[1], Sadia Chaudhary[2], Sundas Masood[3], Abdur Rehman[4]

## Abstract

**Summary:** Hearing loss profoundly affects quality of life, limiting communication, social interaction, and occupational performance. It is also associated with emotional distress, social isolation, and an increased risk of cognitive decline when left untreated. This case report describes a young male presenting to the ENT OPD with progressive hearing loss in bilateral ear. After exclusion of all associated symptoms and previous history, etiology was sought via otoscopy, revealing a perfectly mobile and normal-looking tympanic membrane. PTA and tympanometry revealed bilateral carhart notch and A-type curve on tympanometry, respectively, making the diagnosis of clinical otosclerosis. Patient underwent exploratory tympanotomy and Stapedotomy with a prosthesis. Follow-up PTA showed closure of air bone gap.

Otosclerosis, though benign, is twice as common in females than male. This case illustrates the classical presentation of otosclerosis in a male patient and highlights the role of keen history taking and high clinical suspicion in a male patient where otosclerosis is often missed as a differential diagnosis of progressive hearing loss, resulting in diagnostic delay, making it a clinical rarity, and hence it is selected as a case report.

**Keywords:** Otosclerosis, Deafness, Quality Of Life, Tympanic Membrane.

## Introduction

Otosclerosis is a localized disorder of bone metabolism affecting the otic capsule, characterized by abnormal resorption and deposition of endochondral bone, most commonly around the stapes footplate. This pathological process leads to stapes fixation and progressive conductive hearing loss, which may develop a mixed or sensorineural component in advanced cases. The condition typically presents in early to middle adulthood and accounts for 5–9% of adult hearing loss in Caucasian populations, with a lower but variable prevalence in Asian cohorts.[1,2] Clinically, otosclerosis manifests as gradually progressive bilateral hearing loss, while tinnitus and vertigo are less common. The two principal forms are fenestral otosclerosis, limited to the stapes footplate and oval window, and cochlear otosclerosis, which involves the otic capsule and may contribute to sensorineural deficits.[3,4]

The present case is reported to highlight the classical presentation of bilateral fenestral otosclerosis in a young adult male, emphasizing the diagnostic role of audiometric and tympanometric assessments, and to demonstrate the effectiveness of stapedotomy with piston prosthesis in restoring hearing thresholds. Such case documentation is of clinical relevance in regions where otosclerosis may be underdiagnosed due to limited access to specialized otological services and radiological investigations, and serves to reinforce surgical intervention as a definitive treatment modality with favorable outcomes.

## Case Presentation

A 32-year-old unmarried male shopkeeper presented to the ENT outpatient clinic. He belonged to a lower-middle socioeconomic stratum and was the sole provider for his family. His past medical history was noncontributory, except for an appendectomy performed in 2016. He reported a history of pollen allergy and a smoking history of seven pack-years. There was no history of major systemic illness or trauma. Family history was significant only for maternal hypertension.

**History of Presenting Complaint**

The patient reported progressive bilateral hearing impairment for the past 10 years. The hearing loss was insidious in onset, gradually progressive, and non-episodic. There were no associated otological or vestibular symptoms, including tinnitus, vertigo, otalgia, otorrhea, or aural fullness. He denied any preceding upper respiratory tract infections, head trauma, or prior ear surgery.

**Examination**

General physical examination revealed a healthy adult male with stable vital parameters and no stigmata of chronic systemic illness. Local otological examination demonstrated normal auricles and external auditory canals. The tympanic membranes were intact, pearly in appearance (figure 1), and demonstrated a well-visualized cone of light bilaterally. Tuning fork tests revealed a negative Rinne bilaterally, with Weber lateralized centrally, and absolute bone conduction comparable to the examiner, consistent with a conductive hearing deficit. The fistula test was negative, and there was no evidence of facial nerve dysfunction. Comprehensive examination of the nasal cavity, oral cavity, oropharynx, and larynx was unremarkable. Systemic examination, including cardiovascular, respiratory, abdominal, and neurological assessments, was within normal limits.

**Investigations**

Routine hematological investigations were within reference ranges. Audiometric evaluation demonstrated bilateral conductive hearing loss on pure tone audiometry(Figure 2) as well as a carhart notch seen at 2kHz. Tympanometry revealed findings compatible with stapes fixation(figure 3), while acoustic reflex testing confirmed absent reflexes(figure 4).

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**Differential diagnosis:**

Based on the clinical and audiological profile, the differential diagnoses considered included serous otitis media, adhesive otitis media, tympanosclerosis, and congenital stapes fixation.

A working diagnosis of **otosclerosis** was established on the basis of clinical suspicion corroborated by audiometric and tympanometric findings

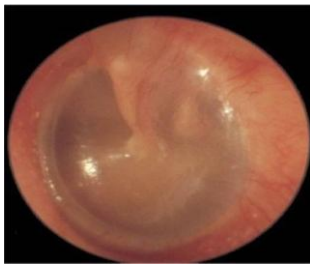

**Figure 1: Showing shiny and intact left tympanic membrane with no retraction and cone of light present at 7'o clock position as it appeared on otoscopy**


**Figure 2: PTA of patient showing bilateral air bone gap of 30dB and presence of carhart notch at 2kHz suggestive of bilateral otosclerosis**

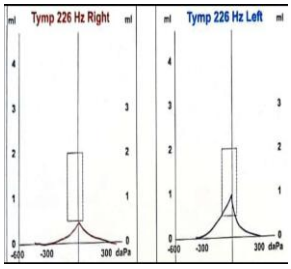


**Figure 3: showing low compliance and normal pressure & volume on tympanometry of right ear plotted as As type curve. Left ear tympanometry appears to be normal**

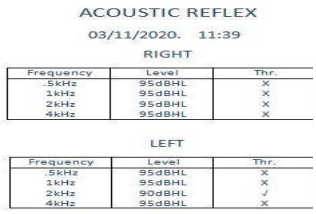


**Figure 4: shows absent acoustic reflex on both sides**

**Management**

The patient underwent a right stapedotomy with insertion of a Teflon piston of size 4.5mm prosthesis under general anesthesia. A tympanomeatal flap was elevated, and an exploratory tympanotomy was done to assess middle ear structures. Stapes food plate was found fixed to the oval window while the rest of the ossicles were intact and mobile( ruling out malleolus fracture, congenital stapes fixation). A fenestration was created in the stapes footplate. A piston prosthesis was then placed across the oval window, followed by closure of the operative site(figure 5 A-E). The intraoperative and immediate postoperative courses were uneventful.

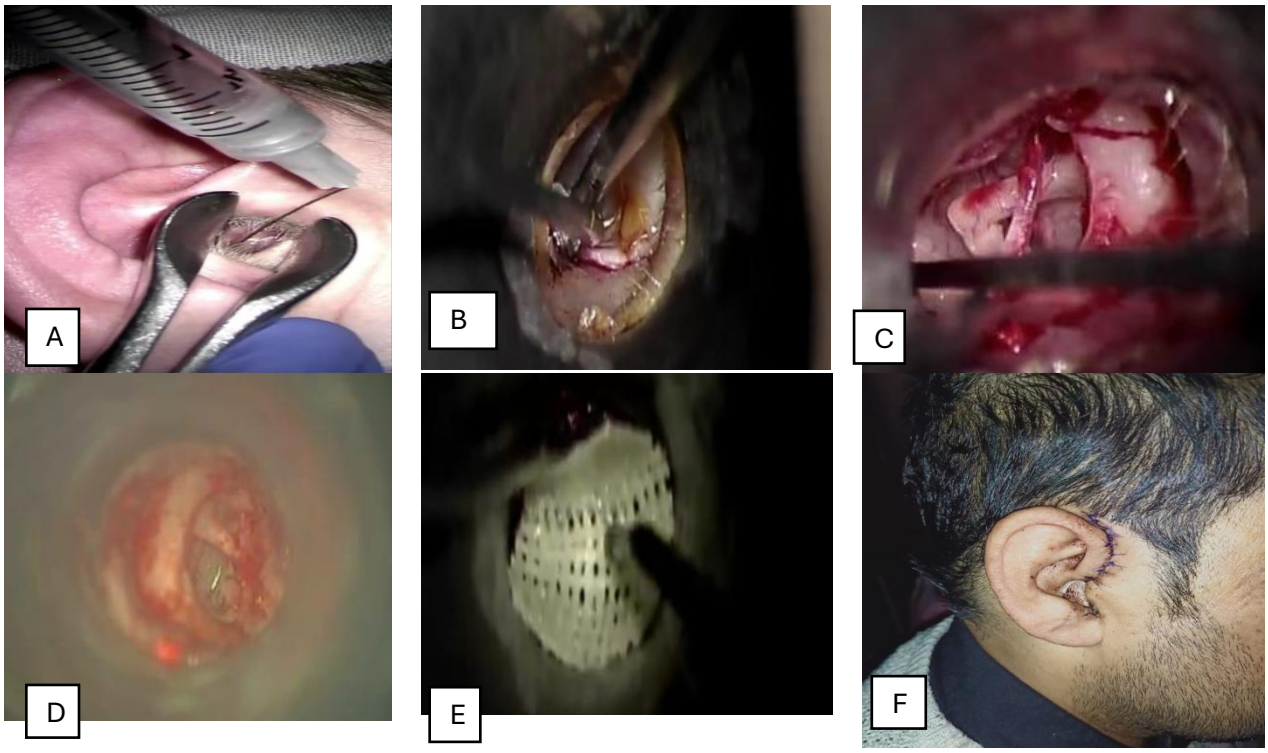


**Figure 5: Steps of surgery are as follows:(A) Local anaesthesia given. (B) tympanomeatal flap elevated. (C) shows middle ear structures visualized and per-op diagnosis of otosclerosis confirmed. (D) fenestration made in stapes foot plate piston inserted. (E) shows wound closed. (F) post op scar**

**Outcome**

The patient demonstrated satisfactory recovery with no early postoperative complications. Follow-up pure tone audiometry after 3 months revealed a significant improvement in air conduction thresholds, closure of ABG, and diminished carhart notch, which was consistent with a successful functional outcome (**Figure 6**). He reported subjective improvement in hearing acuity, confirming the effectiveness of stapes surgery in this case.

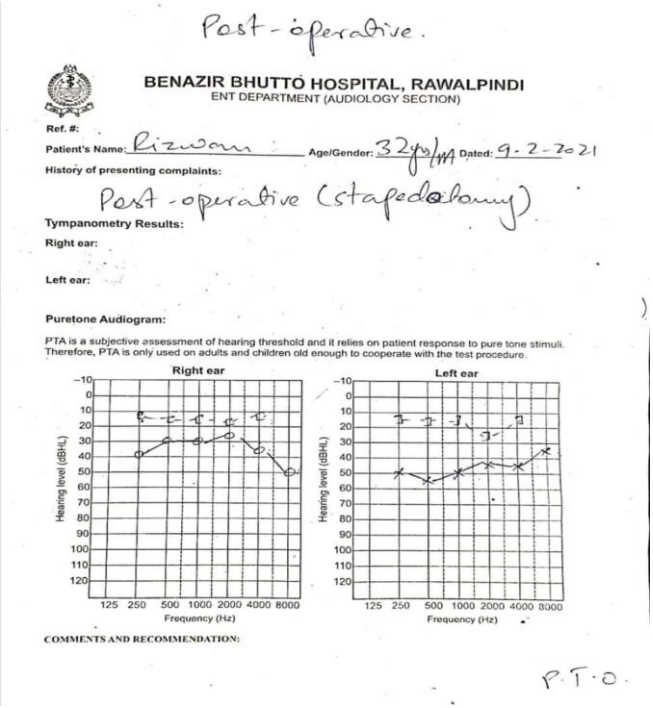


**Figure 6: Post-op audiogram shows closure of air-bone gap on operated(right) ear as well as no carhart notch formed**

## Discussion

Otosclerosis is a progressive osteodystrophic disorder marked by abnormal bone remodeling within the otic capsule and is a leading cause of acquired hearing loss, with histological evidence in approximately 10% of Caucasians.[5] The condition primarily induces conductive hearing loss due to stapes fixation, but sensorineural hearing loss may also occur, potentially resulting from disrupted inner ear fluid homeostasis or dysfunction of the cochlear lateral wall fibrocyte network.[6] Recent synchrotron radiation phase-contrast imaging has revealed that otosclerotic plaques can form shunts and abnormal vascular connections to the labyrinth, leading to hypervascularization, venous overload, and subsequent sensorineural hearing loss.[7] Histologically, otosclerosis is present in 12% of white individuals, but only 0.3–0.4% develop clinical symptoms.[8] Prevalence is lower among Black, Asian, and Native American populations. The average age of onset is around 30 years, and the female-to-male ratio is approximately 1.5–2:1.[8]

To date, only two genome-wide association studies have examined otosclerosis, with the latest identifying 18 significant loci. Targeted resequencing has found 13 variants in five genes—MARK3, LINC01482, AHSG, SUPT3H, and RELN—implicated in bone metabolism, chromatin remodeling, and extracellular matrix regulation, suggesting both protective and pathogenic roles. Variants in TGFβ1 and EYA2 were not significantly associated after correction, though TGFβ1 pathways remain relevant.[9]

Environmental factors, notably viral infections such as measles, are implicated in otosclerosis pathogenesis, possibly by triggering autoimmune responses that disrupt bone remodeling.[10] Hormonal influences, particularly estrogen, have been proposed, given the higher prevalence in females and during periods of hormonal fluctuation such as pregnancy. However, a large case-control study of 1,196 women found no significant association between pregnancy and otosclerosis.[11,12]

Normal bone remodeling occurs at a rate of 10% per year in the skeleton, but the otic capsule remodels at only 0.13% per year.[13] In otosclerosis, this rate increases, resulting in bone deposition that impairs auditory structures and sound transmission. Lesions most commonly occur anterior to the oval window and stapes footplate (80%), but can also affect the round window (30%), pericochlear region (21%), and anterior internal auditory canal (19%).[14]

Clinically, patients experience gradual hearing loss, especially at low frequencies, making it difficult to perceive male voices or vowel sounds. Nearly half report tinnitus, while vertigo is rare (10%) and typically occurs only with vestibular involvement.[8,15] Bilateral disease develops in 80% of cases, though initial presentation is often unilateral.[16]

Otoscopic examination is usually normal, except for the Schwartz sign—reddening of the promontory—seen in about 20% of cases, indicating active osteosclerotic activity, but it is not required for diagnosis.[17] Diagnosis relies on clinical history, examination, and audiometry. Audiometry assesses air and bone conduction, with thresholds above 25 dB considered abnormal. Conductive loss at low frequencies and the Carhart notch at 2,000 Hz are classic findings, though the latter is not a reliable diagnostic marker. Audiometry is also valuable for monitoring disease progression. Early disease causes ossicular stiffening and a small air–bone gap, which widens with stapes fixation. Cochlear involvement in about 10% of patients leads to high-frequency sensorineural loss and a mixed hearing pattern. Tympanometry is typically normal, except in advanced cases with marked ossicular fixation.

High-resolution CT is the first-line investigation for identifying otosclerotic foci, with diagnostic accuracy exceeding 90% in some studies, and can help differentiate other middle ear pathologies. The degree of hearing loss may correlate with the size of otosclerotic lesions, though some

studies report no significant correlation between CT and audiometric findings, suggesting audiometry alone may suffice when CT is unavailable.[18-20]

Medical management aims to slow abnormal bone remodeling. Bisphosphonates and sodium fluoride have shown potential for stabilizing hearing and slowing progression in observational studies and small trials, but large randomized trials are lacking. Systematic reviews indicate that at least six months of sodium fluoride can stabilize hearing thresholds, improve vestibular symptoms, and delay tinnitus progression. Bisphosphonates administered for at least six months have demonstrated improvements in hearing, dizziness, and tinnitus remission, with double-blind studies showing greater stabilization of hearing thresholds compared to placebo.[21]

Surgical advances have refined both traditional and novel techniques. Stapedotomy remains the gold standard, providing significant improvement in air conduction and speech discrimination with low complication rates in experienced hands.[22] Lasers and microdrills have enhanced precision in footplate fenestration, though superiority over conventional methods is still under investigation.[23] Preoperative high-resolution CT is now routinely used for surgical planning. For advanced cases with profound sensorineural loss, cochlear implantation is effective, even with cochlear ossification, and outcomes are improved by technical modifications and electrode selection.[24] Endoscopic approaches offer better visualization and may reduce morbidity, while 3D-printed models are being developed to improve surgical training.[25] Ultimately, the choice between stapes surgery and cochlear implantation is individualized, based on disease extent, hearing profile, and patient factors, with both approaches demonstrating high success rates in recent studies.

## Conclusion

This case report emphasizes detailed, careful history taking, examination, and keeping otosclerosis as part of the differentials when there is progressive conductive hearing loss. Audiometry can help diagnose as well as monitor disease progression. Despite the availability of radiological and audiometry facilities, a trained mind with a caring heart is the foundation of patient care.

## Author Information

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# Endocrine Dysregulation and Functional Ovarian Cyst: A Case from Adolescent Gynecology

Tabinda Khalid[1], Rubaba Abid Naqvi[2], Aqsa Ikram Ul Haq[3], Shama Bashir[4]

## Abstract

**Summary:** Functional ovarian cysts are common in reproductive-age women, but their occurrence secondary to hypothyroidism is rare. We report a case of a 21-year-old female with poorly controlled hypothyroidism who presented with bilateral multiloculated ovarian cysts mimicking neoplastic pathology. Emergency laparotomy was performed due to worsening symptoms, and bilateral cystectomy with ovarian conservation was achieved. Histopathology confirmed corpus luteal cysts. Postoperative thyroid hormone replacement led to normalization of menstrual cycles and resolution of cysts. This case highlights the importance of thyroid evaluation in young women with ovarian masses to avoid unnecessary surgery and preserve fertility.

**Keywords:** Ovarian Cysts, Corpus Luteum, Hypothyroidism, Thyroid Hormones/therapeutic use, Adolescent, Young Adult, Gynecology, Fertility Preservation.

## Introduction

Ovarian cysts are fluid-filled sacs that may arise from normal ovulatory processes or pathological conditions. Ovarian cysts are a common gynecologic finding in adolescents, yet their evaluation and management require careful consideration due to age-specific risks and outcomes.[1] Updated guidelines emphasize structured approaches for diagnosis and treatment in children and adolescents, ensuring both safety and fertility preservation.[2] Recent retrospective studies further highlight the clinical spectrum of adolescent ovarian cysts and the importance of individualized management strategies.[3] Endocrine disorders like hypothyroidism can disrupt the hypothalamic–pituitary–ovarian axis.[4]

It may affect ovarian function through complex endocrine and immune pathways.[5] Recognition of this link is crucial, as thyroid hormone replacement can reverse changes and restore reproductive function.[6]

## Case Presentation

A 21-year-old unmarried female, a postgraduate student from Rawalpindi, presented to the Gynecology outpatient department of Rawalpindi Teaching Hospital on 11 April 2025 with complaints of lower abdominal pain ongoing for two years, with acute worsening over the preceding week, along with menstrual irregularities. She had a three-year history of hypothyroidism but was non-compliant with levothyroxine therapy.

Her menstrual cycles were previously regular (5/28); however, for the past two years, she had become oligomenorrheic, with scanty flow every 40–60 days. She reported progressive weight gain but denied cold intolerance, constipation, acne, hirsutism, galactorrhea, or urinary and bowel disturbances.

On examination, she was well-oriented with a BMI of 29.8 kg/m², pulse rate 106/min, and blood pressure 100/60 mmHg. The abdomen was protuberant with a palpable, firm, non-tender, irregular pelvic mass measuring approximately 20 cm, with restricted mobility.

**Investigations:**

- Hemoglobin: 10.1 g/dL
- TSH: 41 mIU/L (↑), Free T4: low
- CA-125: 18.4 IU/mL
- Ultrasound: Bilateral multilocular adnexal cysts- right cyst (16x11x4) and left cyst (10 × 11 × 5 cm)—with normal uterus.
- CT scan: Confirmed complex cysts with mild ascites
- Tumor markers: CA-125 18.4 IU/ml, β-hCG < 1 IU/ml, AFP 5 IU/ml.

**Treatment**

We planned a laparoscopy; however, due to worsening pain, an emergency laparotomy was performed. Bilateral multilocular cysts with hemorrhagic areas were found. Bilateral cystectomy with ovarian conservation was completed. Histopathology confirmed corpus luteal cysts. Levothyroxine 100 µg/day was initiated postoperatively.

**Outcome and Follow-Up**

At 3-month follow-up:

- Menstrual cycles normalized
- Ultrasound showed normal ovaries
- TSH decreased to 4 mIU/L

**Contributions:**
TK - Conception, Design
TK - Acquisition, Analysis, Interpretation
TK RAN AIUH SB - Drafting
TK RAN AIUH SB - Critical Review

All authors approved the final version to be published & agreed to be accountable for all aspects of the work.

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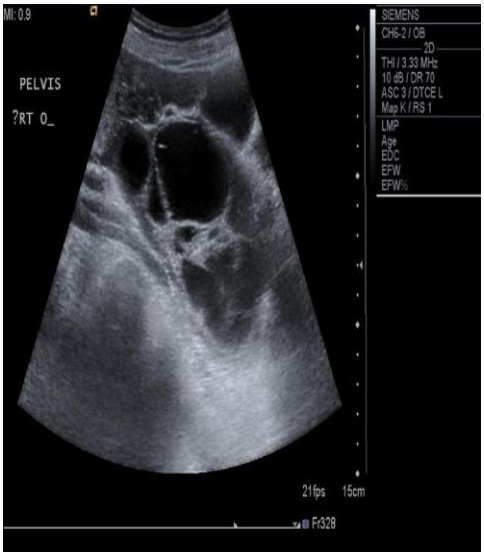


**Figure 1A: (ultrasound image showing multiloculated ovarian cysts)**

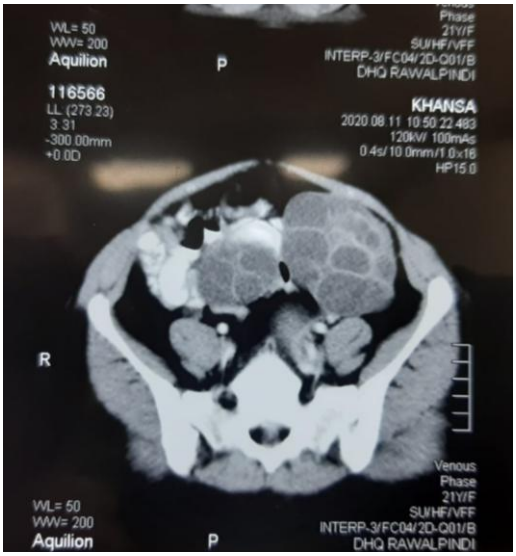


**Figure 1B: (C-T Scan showing bilateral complex cysts with ascites)**

## Discussion

Functional ovarian cysts arise from disturbances in normal follicular development and are most common in reproductive-age women.
The corpus luteal cysts observed in this patient are typically transient structures that may enlarge due to hormonal dysregulation. Primary hypothyroidism has been implicated in their pathogenesis through complex interactions between the hypothalamic–pituitary–ovarian axis. In hypothyroidism, several mechanisms can lead to the development of large ovarian cysts.

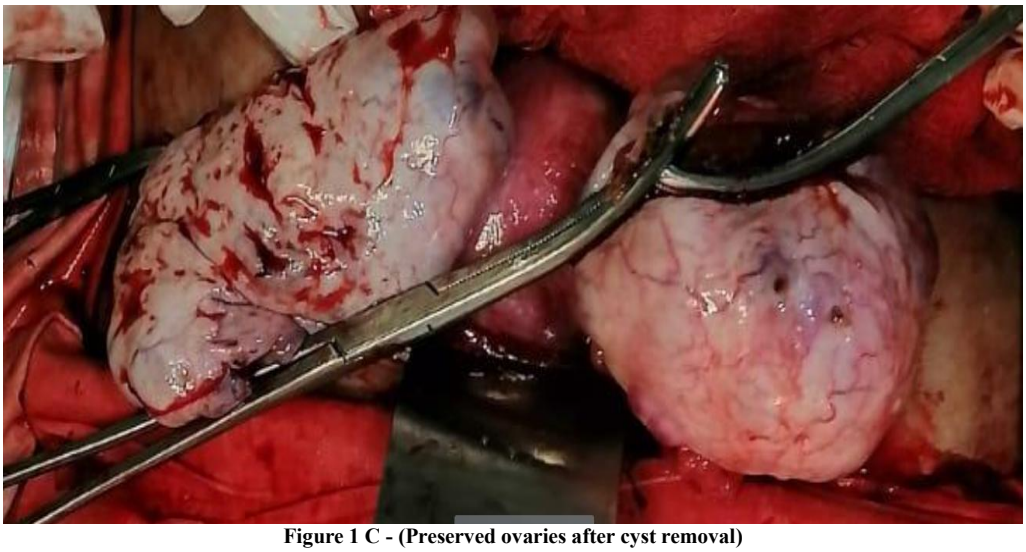

**Figure 1 C - (Preserved ovaries after cyst removal)**

Elevated thyroid-stimulating hormone (TSH) can cross-react with follicle-stimulating hormone (FSH) receptors on ovarian tissue, promoting excessive follicular growth. Increased thyrotropin-releasing hormone (TRH) causes hyperprolactinemia, which disrupts gonadotropin-releasing hormone (GnRH) pulsatility, leading to anovulation and follicular persistence. Additionally, altered gonadotropin ratios with relatively low luteinizing hormone (LH) and higher FSH levels prevent normal luteinization, resulting in cyst formation. Chronic hypothyroidism may also increase ovarian sensitivity to gonadotropins and insulin-like growth factors, further stimulating cyst development. Lastly, thyroid hormone deficiency directly impairs ovarian follicular maturation and angiogenesis, contributing to the persistence of large functional cysts.
Hypothyroidism presenting with ovarian cysts is an uncommon but clinically important condition. Large cysts may mimic neoplastic disease, creating diagnostic uncertainty and the risk of unnecessary surgical intervention. Similar to our patient, huge bilateral ovarian cysts associated with hypothyroidism have been reported, highlighting the potential for misinterpretation as ovarian malignancy.[7]
Other reports describe bilateral ovarian masses due to primary hypothyroidism, further emphasizing the spectrum of presentations and the importance of considering endocrine causes in the differential diagnosis.[8] Rare complications such as pituitary adenoma and chronic adnexal torsion have also been documented in hypothyroid patients, underscoring the need for timely recognition and multidisciplinary management.[9]

Additionally, giant ovarian cysts have been described following thyroidectomy, reinforcing the link between thyroid dysfunction and ovarian pathology.[10] Collectively, these cases demonstrate that ovarian cysts secondary to hypothyroidism can regress with appropriate thyroid hormone replacement, thereby avoiding unnecessary surgical procedures. Our case adds to this growing body of evidence, supporting routine thyroid evaluation in adolescents and young women presenting with ovarian cysts.

Collectively, these cases demonstrate that ovarian cysts secondary to hypothyroidism can regress with appropriate thyroid hormone replacement, thereby avoiding unnecessary surgical procedures. Our case adds to this growing body of evidence, supporting routine thyroid evaluation in adolescents and young women presenting with ovarian cysts. Thus, a multidisciplinary team approach involving gynecologists, endocrinologists, and radiologists is essential for accurate diagnosis, timely intervention, and fertility preservation, as functional cysts secondary to hypothyroidism are reversible with appropriate hormonal supplementation.

## Conclusion

Functional ovarian cysts secondary to hypothyroidism, though uncommon, should be considered in reproductive-age females presenting with bilateral ovarian masses. Early recognition and thyroid hormone replacement can lead to complete regression of cysts and normalization of reproductive function, thereby avoiding unnecessary surgical procedures and preserving fertility.

**Learning points**

- Thyroid function testing should be routine in young women presenting with bilateral ovarian cysts.
- Functional cysts secondary to hypothyroidism may mimic malignancy but regress with thyroid hormone therapy.
- Multidisciplinary management involving gynecologists, endocrinologists, and radiologists is essential to prevent unnecessary surgery and preserve fertility

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# Laparoscopic Management of a Giant Paratubal Cyst: A Case Report

Sadia Khan[1], Ismat Batool[2], Nighat Naheed[3], Hina Saleem[4], Arshia Kanwal[5], Ayesha Javed[6]

## Abstract

**Summary:** Large paratubal cysts are uncommon benign adnexal lesions that can closely resemble ovarian cysts on both clinical and radiologic assessment. Their laparoscopic excision can be technically demanding, particularly when fertility preservation is desired. We describe a 24-year-old woman who presented with gradually increasing abdominal distension and right lower abdominal pain. Imaging revealed a 25 × 15 × 10 cm right-sided adnexal cyst with the ovary draped over its surface, suggesting a paratubal origin. All tumor markers were within normal limits. Laparoscopic cystectomy with controlled decompression was carried out, and all pelvic organs were preserved. Histopathology confirmed a benign paratubal cyst. The postoperative course was smooth, and the patient was discharged the next day. This case demonstrates that, with careful preoperative evaluation and meticulous surgical technique, laparoscopy can be safely and effectively used even for very large paratubal cysts.

**Keywords:** Paratubal cyst, Broad ligament cyst, Laparoscopic cystectomy, Giant adnexal mass, Fertility preservation.

## Introduction

Most adnexal masses originate from the ovary, but a small fraction arise from adjacent structures such as the fallopian tube or broad ligament [1]. Paratubal cysts account for roughly 10 % of all adnexal lesions [2]. They are believed to arise from mesothelial inclusions or remnants of the Wolffian or Müllerian ducts [3]. Typically, these cysts are small and asymptomatic, but in rare cases they can enlarge considerably, leading to abdominal distension and being mistaken for ovarian tumors [2,3]. Managing such large cysts laparoscopically can be challenging because of limited working space and the risk of rupture [4]. We present a rare case of a giant paratubal cyst managed successfully through laparoscopy at a tertiary care center

## Case Presentation

A 24-year-old unmarried, nulliparous woman presented with a one-year history of right-sided lower abdominal pain and gradually increasing abdominal distension over six months. Initially, she was treated for a presumed urinary tract infection after being misdiagnosed with bladder distension by a general practitioner. Her symptoms persisted, prompting referral to the Department of Obstetrics and Gynecology, Benazir Bhutto Hospital (BBH), Rawalpindi.

Her past medical and surgical history was unremarkable, and her menstrual cycles were regular. On examination, she was hemodynamically stable, with a BMI of 23 kg/m². Abdominal inspection revealed a distended abdomen with an everted umbilicus. Palpation showed a large, soft, cystic, mobile, and non-tender mass extending up to the xiphisternum. (Figure 1)

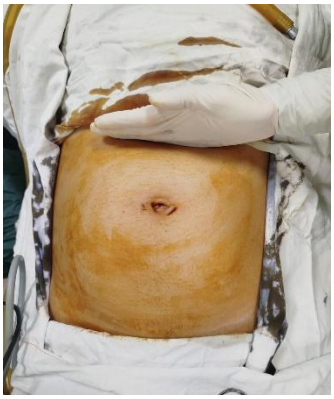

**Figure 1: Distended abdomen with an everted umbilicus**

Routine laboratory investigations, including complete blood count and renal and liver function tests, were normal. Tumor markers were within reference ranges: CA-125 = 10 U/mL, CEA = 0.9 ng/mL, AFP = 3.6 ng/mL, and β-hCG < 0.2 mIU/mL.

**Contributions:**
SK IB NN HS AK AJ - Conception, Design
SK IB NN HS AK AJ - Acquisition, Analysis, Interpretation
SK IB NN HS AK AJ - Drafting
SK IB NN HS AK AJ - Critical Review

All authors approved the final version to be published & agreed to be accountable for all aspects of the work.

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**Financial Support:** None to report
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Ultrasonography demonstrated a unilocular cystic mass in the right adnexa measuring approximately 29 × 17.6 × 8.7 cm. MRI revealed a large, thin-walled, well-defined right adnexal cyst measuring 25 × 15 × 10 cm, displacing the uterus to the left. The right ovary appeared draped around the cyst, suggesting a paratubal origin. (Figure 2)

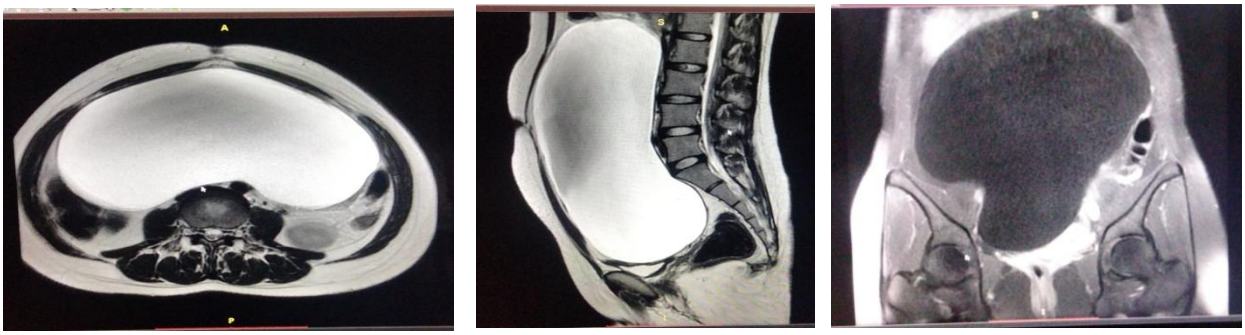

**Figure 2: Axial & sagittal T2WI precontrast coronal post contrast T1WI**

After appropriate preoperative counseling, a laparoscopic cystectomy was performed using the open umbilical entry technique. Two 5 mm accessory ports were inserted under direct vision. Intraoperatively, the pelvic organs were poorly visualized initially due to the large size of the cyst, which made identification of the contralateral adnexa, uterus, bowel, and major pelvic vessels difficult.

Controlled decompression was achieved by aspirating approximately 4 liters of clear fluid, which significantly improved visualization. Following decompression, the uterus, left fallopian tube, and ovary appeared normal and healthy. The right ovary was visualized separately, and the right fallopian tube was stretched over the cystic mass. The cyst was identified as arising from the right broad ligament, consistent with a paratubal cyst.

The ureter and internal iliac artery were clearly identified and carefully preserved throughout the procedure (Figure 3). The cyst wall was gradually freed from surrounding structures by meticulous dissection and counter-traction. One of the 5 mm accessory ports was converted to a 10 mm port to facilitate safe retrieval of the specimen, with residual cyst fluid aspirated to allow controlled decompression. The cyst was completely excised while preserving the right ovary, fallopian tube, and all vital structures. (Figure 4,5). The patient remained hemodynamically stable throughout the procedure and postoperative period and was discharged the following day in good condition.

**Histopathology:**

Grossly, the specimen consisted of a thin-walled, unilocular cyst containing clear straw-colored fluid. Microscopically, the cyst wall was lined by a single layer of cuboidal to flattened epithelial cells supported by fibrous stroma. There was no atypia, and no ovarian tissue was identified. (Figure 6) These findings were consistent with a benign paratubal cyst of the broad ligament.

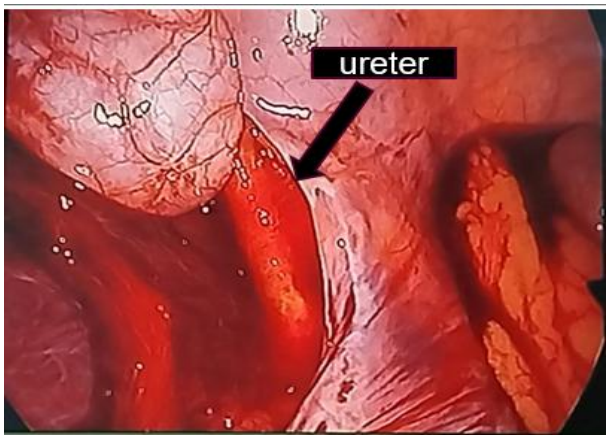


**Figure 3: Identification of the Ureter**

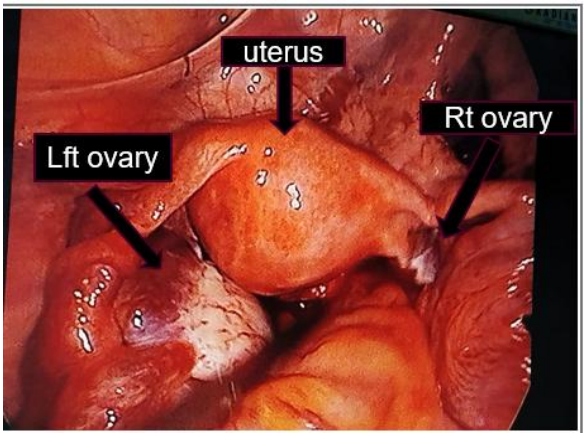


**Figure 4: Identification of Uterus and Ovaries**

## Discussion

Paratubal cysts are typically small, but rarely they can reach massive dimensions, mimicking ovarian cysts on clinical and imaging evaluation [5]. The nonspecific presentation—abdominal distension, discomfort, or urinary symptoms—usually results from the compressive effect of the expanding cyst [6]. It is notable that only about one in fifteen paratubal cysts are correctly diagnosed before surgery [7]. MRI can aid in diagnosis, as the ovary appearing draped over the cyst is a helpful distinguishing feature [6,7].

Laparoscopic removal of such large cysts requires technical adjustments and careful planning. The open entry method reduces the risk of visceral or vascular injury, while decompression under direct visualization minimizes the chance of rupture or spillage. Identification of the ureter and major vessels during dissection is essential for safety [4,7].

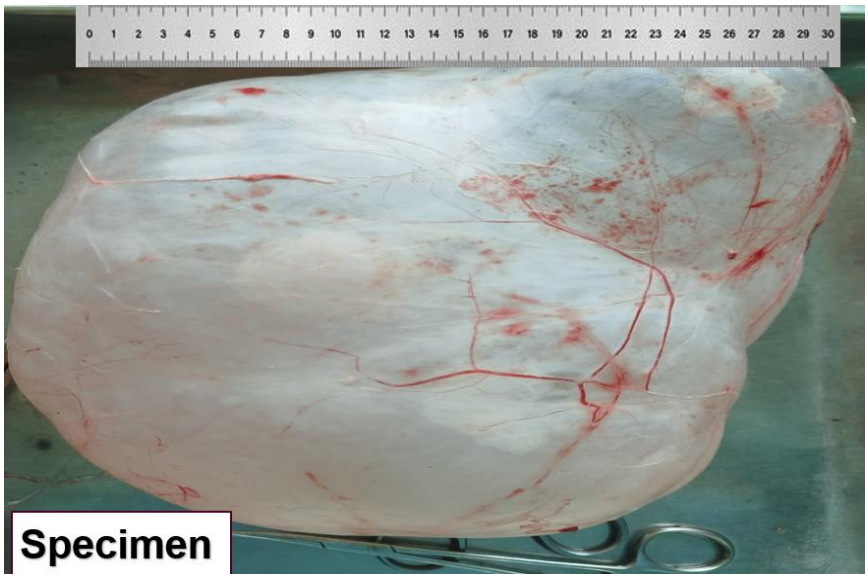


**Figure 5: Paratubal Cyst**

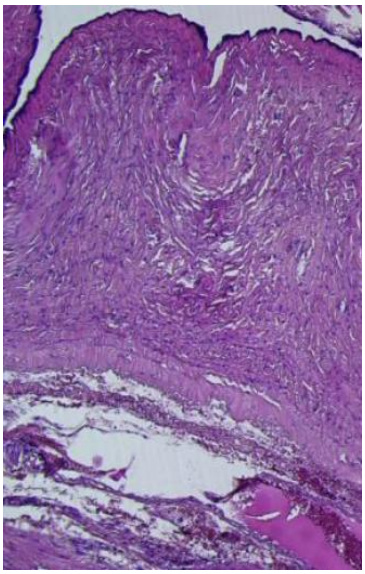

**Figure 6: Histopathology of Cyst**

A systematic review by Stefanopol and colleagues identified only 17 published cases of giant paratubal cysts, with few managed laparoscopically [8]. The largest paratubal cyst previously reported from Pakistan measured 10 × 8 cm and was excised via laparotomy [9]. To the best of our knowledge, the present case with a cyst measuring 25 × 15 × 10 cm managed entirely through laparoscopy ranks among the largest successfully treated minimally invasively in the country. This reinforces the view that cyst size alone should not preclude laparoscopic management when expertise and appropriate facilities are available.

## Conclusion

Although giant paratubal cysts are uncommon, they should be included in the differential diagnosis of large adnexal masses. When tumor markers are normal and imaging suggests a benign nature, laparoscopy offers a safe, fertility-preserving, and minimally invasive option even for very large cysts. Successful outcomes depend on adequate preoperative assessment and surgical precision.

## Author Information

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# Pleural Adenocarcinoma Presenting with Deep Venous Thrombosis: An Unusual Incidental Finding

Zaid Umar[1], Samar Saleem[2], Faran Maqbool[3], Sumayyah Maryam[4]

## Abstract

**Summary:** Deep venous thrombosis (DVT) is a common vascular condition with recognized risk factors such as immobility, trauma, and hypercoagulable states. However, in patients without identifiable causes, the possibility of an underlying malignancy should be strongly considered. We report the case of a 50-year-old non-smoker male who presented with lower limb swelling secondary to extensive DVT and was incidentally diagnosed with pleural adenocarcinoma following evaluation of a right-sided pleural effusion. This case highlights the importance of maintaining a high index of suspicion for occult malignancy in patients with unprovoked DVT to enable early diagnosis and timely management.

**Keywords:** Adenocarcinoma, Pleural Neoplasms, Venous Thrombosis.

## Introduction

Venous thromboembolism (VTE) is strongly associated with cancer, with a relative risk increase of 5–20 times.[1-3] Approximately 7–12% of patients with idiopathic DVT are later diagnosed with occult malignancy.[3] Pleural adenocarcinoma, a rare malignancy (<5% of pleural cancers), can manifest with pleural effusion, chest pain, or respiratory symptoms.[10,11]DVT may complicate the disease course in up to 10% of cases.[12] This case highlights pleural adenocarcinoma initially presenting with unprovoked DVT, which is an unusual clinical scenario.

## Case Presentation

A 50-year-old non-smoker male, a security guard by occupation, presented with a history of left leg pain for 18 days, which was gradual in onset, sharp, more around the ankle, described as 7/10 on the Visual Analog scale (VAS), persistent, and aggravated by walking, relieved partially by oral or IM analgesics. It was associated with swelling that had progressed to the mid-thigh at the time of presentation. Other risk factors, such as prolonged immobilization, trauma, insect bite, prolonged travel, or drugs contributing to such symptoms, weren't reported. On systemic inquiry, he reported having a dry cough and dull right-sided chest pain on inspiration with MRC grade II dyspnea. Past medical and surgical history was unremarkable. The patient was a *Naswar* addict for 30 years, but had no history of smoking, alcoholism, or IV drug abuse. Upon admission, the patient was calm and comfortable, vitally stable, maintaining saturation on room air with a notable left leg swelling extending from ankle to mid-thigh with 2cm of girth difference on comparison with the right leg, with overlying skin being shiny and erythematous, warm, and tender to touch with grade II pitting edema and positive Homan's sign. Peripheral pulses were palpable with no lymphadenopathy.

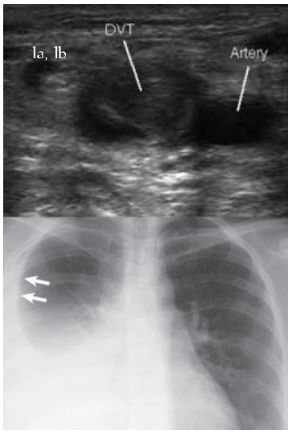


**Figure 1: Leg Doppler and Chest X-Ray**

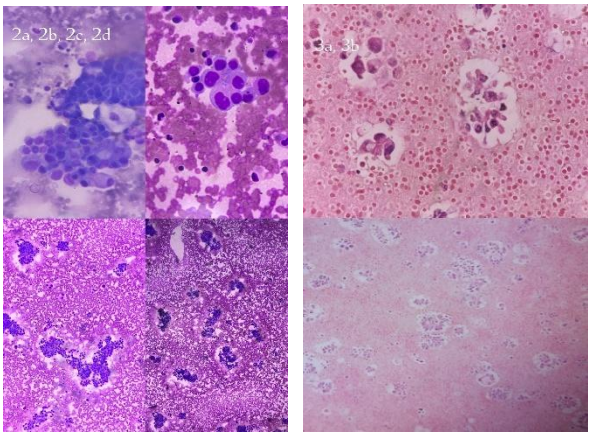


**Figure 2 & 3: Pleural fluid cytology**

Systemic examination, unmasked presence, reduced chest movements, reduced focal fremitus, stony dull percussion note, and reduced breath sounds in the right middle and lower chest.

**Contributions:**
ZU SS FM SM - Conception, Design
ZU SS FM SM - Acquisition, Analysis, Interpretation
ZU SS FM SM - Drafting
ZU SS FM SM - Critical Review

All authors approved the final version to be published & agreed to be accountable for all aspects of the work.

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**Investigations**

Preliminary laboratory workup revealed normocytic normochromic anemia with normal liver and kidney functions. Inflammatory markers were raised along with D-dimer levels (2000ng/mL). The coagulation profile was normal. Doppler Ultrasound (Leg) revealed acute DVT extending to the left common, internal, and external iliac, superficial femoral, and great saphenous veins (Figure 1a). Chest X-ray (Figure 1b) showed right-sided costo-phrenic angle blunting and opacifications consistent with right-sided pleural effusion, thus prompting the need to get USG chest for quantification as well as to perform radiologically guided diagnostic pleurocentesis. Pleural fluid was exudative lymphocytic on microscopy with negative Gram and ZN staining, thus ruling out tuberculous effusion, which is quite common in our setup. Pleural fluid cytology revealed atypical cell clusters with pleomorphic, eccentrically placed nuclei, a high nuclear to cytoplasmic ratio, and abundant cytoplasmic mucin, consistent with metastatic adenocarcinoma. (Figure 2a, 2b, 2c, 2d) (Figure 3a, 3b). Immunohistochemistry was positive for TTF-1, CK7, AE1/AE3, and Napsin-A, which are markers associated with adenocarcinoma.[7-9] Molecular genetics studies were negative for EGFR mutation.

CECT chest (pre- and post-contrast) showed massive right pleural effusion with compressive collapse, pleural thickening, and an enlarged pre-tracheal lymph node (Figure 4a, 4b, 4c). Coronal images revealed diaphragmatic and mediastinal pleural thickening with right lung collapse and mediastinal shift (Figure 5). CT abdomen and pelvis showed a central filling defect extending from the left common iliac to the femoral vein, consistent with extensive DVT (Figure 6a, 6b, 6c). The bone scan showed no evidence of metastasis. Bronchoscopy was normal.

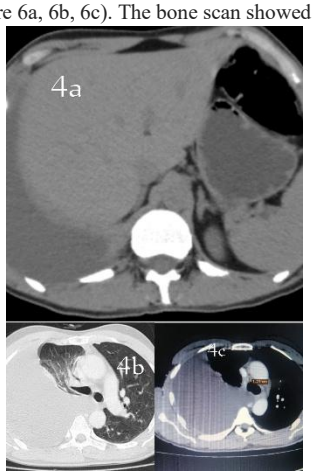


**Figure 4: CECT chest**

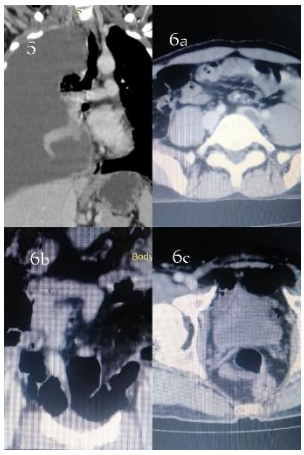


**Figure 5 & 6: CT Abdomen & Pelvis**

**Treatment**

As per TNM staging,[13-15] our patient had stage IV adenocarcinoma with the VTE phenomenon. With good functional status, he underwent chest tube insertion and talc pleurodesis.[10,11] In a poorer status, an indwelling pleural catheter is preferred. DVT was managed with LMWH 80 mg twice daily. The family was counseled about prognosis and palliative options. The patient received four cycles of platinum-based chemotherapy (cisplatin + pemetrexed). EGFR mutation was negative, precluding targeted therapy.[4-6]

**Outcome And Follow-Up**

The patient tolerated chemotherapy and pleurodesis. Symptomatic improvement was noted in breathlessness and limb swelling. Despite the challenges, coordinated care between both facilities (pulmonology and oncology) ensured optimal management, highlighting the value of specialized centers and collaborative medical practice.[10,11]

## Discussion

This case demonstrates the importance of a thorough workup in patients presenting with apparently idiopathic DVT [1-3]. Malignancy is a major risk factor of DVT with a relative risk of 5-20 times. Occult malignancy is present in 7– 12% of idiopathic DVT. In diagnosed malignancies, the 12-month incidence is 4.2% to 4.7%. Approx. 10% case of Pleural Adenocarcinoma develop DVT during the whole disease course. Pleural adenocarcinoma is rare,[7-9,12] and most often presents with pleural effusion, dyspnea, or chest pain. Diagnosis is usually delayed as symptoms are nonspecific and mimic other benign pleural diseases. In our case, the patient presented with left leg DVT and right-sided malignant pleural effusion, an unusual initial combination. While thromboembolic events are well-documented in association with malignancy, reports describing pleural adenocarcinoma presenting with DVT are scarce.

Venous thromboembolism (VTE) in the context of cancer is associated with poor prognosis and worse long-term survival.[5,6,12] Therefore, timely diagnosis and treatment are crucial. Current evidence favors LMWH over warfarin and DOACs for both treatment and prophylaxis of malignancy-related VTE, given its superior efficacy and safety profile.[4-6]

The diagnosis of pleural adenocarcinoma is established primarily through pleural fluid cytology and confirmed with immunohistochemistry. Markers such as TTF-1, CK7, and Napsin A are typically positive and help differentiate pleural adenocarcinoma from malignant mesothelioma and pleural metastases of non-pulmonary origin (e.g., breast or gastrointestinal cancers). Molecular profiling, including EGFR mutation testing, can further refine diagnosis and guide targeted therapy.[7-9]

Treatment of pleural adenocarcinoma is stage-dependent. Early stages (I–II), though rare, may be managed surgically with adjuvant platinum-based chemotherapy or radiotherapy. Stage III, with mediastinal nodal involvement, is usually unresectable and treated with systemic

chemotherapy such as cisplatin/carboplatin plus pemetrexed, often combined with immunotherapy (e.g., pembrolizumab). Stage IV, defined by malignant effusion or distant metastasis, is treated palliatively with chemotherapy, targeted therapy (EGFR/ALK/ROS1 inhibitors if mutations present), and immunotherapy (PD-1/PD-L1 inhibitors). Malignant effusions are managed with pleurodesis (e.g., talc) or indwelling pleural catheters in poor functional status for symptom relief.

The prognosis of pleural adenocarcinoma remains poor, with median survival ranging between 6 and 12 months depending on stage and treatment response. However, early recognition and multidisciplinary care may improve outcomes.

**Learning Points**

This case highlights left leg DVT as a rare initial sign of pleural adenocarcinoma with malignant effusion in a patient without typical risk factors. It underscores the need to investigate unexplained DVT for occult malignancy. Early recognition of the cancer–thrombosis link and multidisciplinary management, including pleural drainage and systemic therapy, are key to improving outcomes.

## Author Information

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# Retrocaval Ureter: Diagnostic Dilemma - A Case Report

Rameez Ahmed Mughal[1], Zeeshan Qadeer[2], Umer Javed Chughtai[3], Faraz Basharat Khan[4]

## Abstract

**Summary:** Retrocaval ureter is one of the unique and rarest congenital anomalies resulting from abnormal development of the inferior vena cava. The characteristic J-shaped or fishhook appearance is pathognomonic for this on CT urogram.[1]
Case presentation: A middle-aged man with a prolonged history of right lumbar pain spanning over three years presented to us. CT urogram depicted right moderate hydronephrosis with a fish-hook ureter. Subsequently, open surgical repair was performed successfully. At 6 months follow-up, he was pain-free with improvement in RFTs. In patients with progressive hydronephrosis or declining renal function, surgical repositioning of the ureter, either by open or laparoscopic method, is the treatment of choice.

**Keywords:** Retrocaval ureter, congenital anomaly, inferior vena cava, hydronephrosis.

## Introduction

Retrocaval ureter results from the abnormal embryological development of the inferior vena cava. This rare congenital anomaly is signified by the posterior course of the ureter with respect to the IVC, resulting in its compression against the lumbar vertebrae.[3] Pathogenesis is the persistence of the right subcardinal vein, which fails to regress and causes entrapment of the ureter behind the IVC.
The classical presentation is flank pain with a history of recurrent infections or obstructive uropathy, usually in the third or fourth decade of life.[2] The condition is more frequent in males, with a male-to-female ratio of approximately 3:1, and is predominantly right-sided; left-sided cases are extremely rare and usually associated with situs inversus or double IVC.[1]
Diagnosis is challenging due to nonspecific symptoms. CT urography is considered the most reliable modality, demonstrating the pathognomonic "fishhook" deformity of the proximal ureter as it loops behind the IVC.[5]

## Case Presentation

A 46-year-old man presented with dull, intermittent right flank pain persisting for three years, associated with nausea and occasional vomiting. He had a history of multiple hospital admissions in the periphery, where supportive treatment was given, which temporarily relieved his symptoms. He denied fever, hematuria, or lower urinary tract complaints.
On examination, he was stable with mild right flank tenderness.

**Investigations**

Laboratory tests were normal, including urea and creatinine. Ultrasound revealed Moderate Hydronephrosis with no evidence of stone, proximal ureteric dilatation, and distal ureter not visualized. (figure-1).
X-ray KUB was unremarkable. Contrast-enhanced CT urography demonstrated a dilated right renal pelvis and proximal ureter with a fishhook configuration looping behind the IVC at L3, consistent with a type I retrocaval ureter. (Figure-2 and Figure-3)
After obtaining informed consent, the patient underwent ureteroureterostomy through a right flank incision using an extraperitoneal approach.
Intraoperatively, we noted a dilated renal pelvis and a retrocaval ureter looping behind the inferior vena cava at the level of the L3 vertebra, with a narrowed retrocaval segment. (Figure-4)

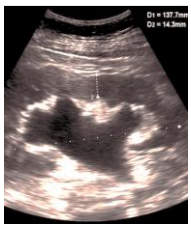

**Figure 1: Ultrasound depicting moderate hydronephrosis**

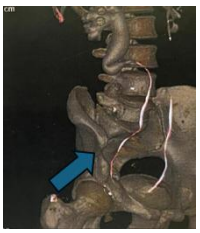

**Figure-2: CT urogram showing classic fish-hook appearance of the ureter with hydronephrosis**

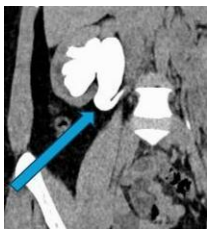

**Figure-3: Coronal section of excretory urogram confirming the findings. No stone was seen anywhere in the urinary tract**

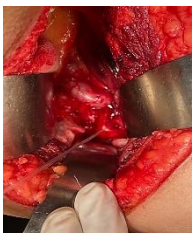

**Figure 4: dilated renal pelvis and narrowed ureter slinged by a loop**

The right renal pelvis and ureter were carefully isolated, and the ureter was looped with a vessel marker, then mobilized cranially and caudally around the stenotic segment. Gentle, atraumatic handling was ensured throughout to preserve vascularity and reduce the risk of anastomotic leak or ischemic atrophy.

**Contributions:**
RAM ZQ - Conception, Design
UJC FBK - Acquisition, Analysis, Interpretation
RAM FBK - Drafting
ZQ UJC - Critical Review

All authors approved the final version to be published & agreed to be accountable for all aspects of the work.

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The adventitia at both ends of the ureter was freed from scar tissue, maintaining adequate blood supply, and the segment was brought together without tension. The stenotic retrocaval portion was excised, and the proximal and distal ends were spatulated about 7–10 mm, positioned 180° apart. (Figure 5)
A ureteric stent was placed, and corner sutures with 5-0 Vicryl were applied. The anastomosis was then completed with interrupted sutures to ensure a watertight seal. (Figure 6)

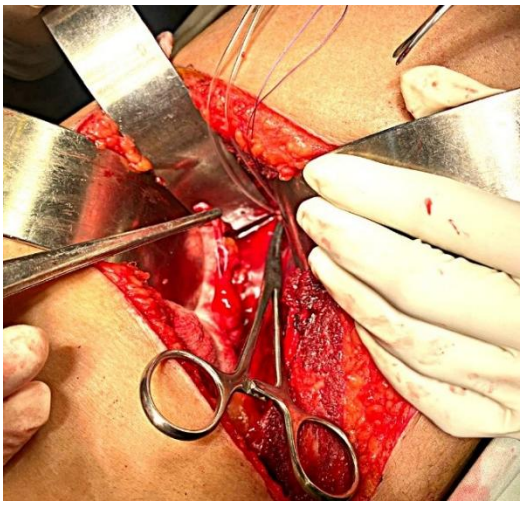

**Figure 5: Proximal and distal ureteric ends freed and excised**

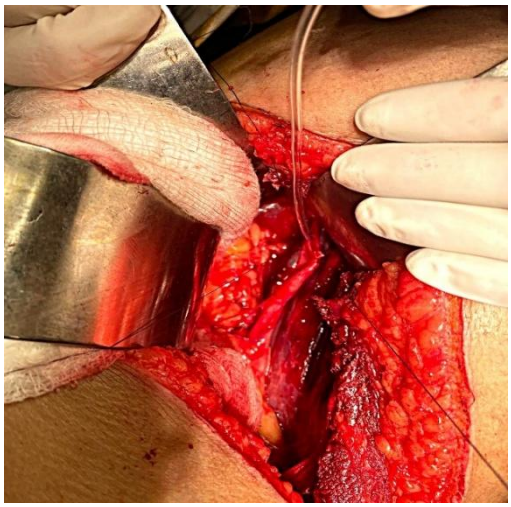

**Figure 6: Anastomosis being done of both proximal and distal ends**

A surgical drain was inserted and removed on the fourth postoperative day once drainage became minimal. A Foley catheter was kept indwelling for 72 hours.
**Follow up**
Ultrasound at six weeks showed resolving hydronephrosis, and the DJ stent was removed. At six months, he remained asymptomatic with good renal function and no recurrence of obstruction.

## Discussion

Retrocaval ureter is a vascular anomaly rather than a true urinary tract malformation. Bateson and Atkinson originally classified it into two subtypes:

- **Type I (low loop):** Most common, with a sharp medial deviation of the ureter crossing posterior to the IVC at L3, causing marked hydronephrosis.
- **Type II (high loop):** Less common and less obstructive, where the ureter crosses higher near the renal pelvis, usually producing only mild dilatation.[1]

Patients typically present with flank pain, recurrent urinary tract infections, or nephrolithiasis. Rarely, delayed presentations have been reported even in the fifth decade.[7]
Imaging is key to diagnosis. While ultrasound may show hydronephrosis, it cannot delineate the course of the ureter. CT urography provides three-dimensional visualization of the ureter's anomalous course behind the IVC.[4]
Surgery is indicated in symptomatic patients, progressive hydronephrosis, or deteriorating renal function. The principle is repositioning the ureter anterior to the IVC and restoring continuity. Both open and minimally invasive approaches (laparoscopic and robotic) have been described, but the latter approach is superior in terms of patient satisfaction, enhanced recovery, and less morbidity.[6,] Unfortunately, due to resource limitations, open surgery was performed in our hospital.

## Conclusion

In young or middle-aged adults with a prolonged history of pain or hydronephrosis, thorough clinical examination and investigations should be performed to rule out rare causes like retrocaval ureter. In this respect, CT urography remains the gold standard. Surgery is the treatment of choice with excellent results and symptom resolution with renal function improvement. Postoperatively, follow-up imaging is required to confirm resolution.

## Author Information

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# Post-Abortion Septic Pelvic Thrombophlebitis With Right Ovarian Vein Thrombosis Complicated By Septic Pulmonary Emboli

Muhammad Khurram[1], Arslan Ahmad[2], Sara Mustafa[3], Nida Anjum[4]

## Abstract

**Summary:** Septic pelvic thrombophlebitis (SPT) is an uncommon but important cause of persistent fever after delivery or gynaecological procedures[1]. Anticoagulation and parenteral antibiotics are the main treatment goals for ovarian vein thrombosis.[2] Ovarian vein thrombosis (OVT) may complicate and lead to septic pulmonary embolism (SPE).[3]A woman in her early 20s presented with high-grade fever and right iliac fossa pain seven days after an induced abortion. Her fever persisted despite 72 hours of broad-spectrum intravenous antibiotics. Contrast-enhanced abdominopelvic computed tomography (CT) revealed a right OVT. Subsequent CT pulmonary angiography confirmed multiple peripheral, partly cavitating nodules consistent with SPE. She was successfully treated with optimized antimicrobial therapy and therapeutic anticoagulation. Fever abated, and interval imaging confirmed thrombus regression. She completed a three-month course of anticoagulation and remained well at follow-up with radiological resolution of pulmonary lesions. SPT should be suspected when postpartum or post-abortion fever fails to respond to adequate antibiotics. Prompt diagnosis with cross-sectional imaging and combined antimicrobial-anticoagulant therapy are essential to prevent complications and ensure a good outcome.

**Keywords:** Ovarian Vein; Thrombophlebitis; Ovarian Venous Thrombosis; Septic Pulmonary Embolism; Puerperal Disorders.

**Contributions:**
MK SM- Conception, Design
AA NA - Acquisition, Analysis, Interpretation
MK SM - Drafting
AA NA - Critical Review

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## Introduction

SPT encompasses infection-associated thrombosis of the ovarian veins or deep pelvic venous plexus. It arises from the convergence of endothelial injury, hypercoagulability, and pelvic sepsis. Although rare, a missed diagnosis can lead to embolic complications. Ovarian vein thrombosis (OVT) has a predilection for the right side, a phenomenon attributed to uterine dextro-rotation, the greater length of the right ovarian vein, and its acute angle of insertion into the inferior vena cava. POVT mainly occurs within the first ten days postpartum, especially after cesarean delivery.[5] Diagnostic imaging, like contrast-enhanced abdominopelvic CT scan are gold standard for diagnosis. In case of contraindication, MRI can be used.[4]

## Case Presentation

A previously healthy woman, early 20s, presented with seven days of fever (spiking to 39°C) and right lower quadrant pain one week after surgical termination at 9 weeks' gestation. There were no urinary or gastrointestinal symptoms and no thrombotic history.
On admission: temperature 38.9°C, pulse 106/min, blood pressure 108/64 mmHg. Abdominal examination revealed suprapubic and right iliac fossa tenderness without peritonism. Vaginal examination demonstrated uterine and right adnexal tenderness. There was no limb swelling, and the respiratory examination was unremarkable.

**Investigations**
Laboratory results showed leukocytosis with neutrophilia and raised C-reactive protein; renal and liver functions were within reference limits. Blood and urine cultures were negative. Serum β-hCG declined appropriately.
Pelvic ultrasound revealed a bulky postpartum uterus with trace free fluid. Because fever persisted beyond 72 hours of broad-spectrum antibiotics, a contrast-enhanced CT abdomen/pelvis was requested, demonstrating an enlarged right ovarian vein with a central low-attenuation thrombus, enhancing wall, and perivenous fat stranding—typical of OVT (Figure 1). New pleuritic pain prompted CT pulmonary angiography (Figure 2, 3), which showed multiple bilateral, peripheral nodules—some cavitary—compatible with SPE.

**Differential diagnosis**
- Endometritis without thrombosis
- Pelvic abscess or infected haematoma
- Acute appendicitis or right ureteric colic
- Urinary tract infection/pyelonephritis
- Catheter-related bloodstream infection

**Treatment**

The patient's empiric antibiotic regimen was optimized to provide broad coverage for polymicrobial pelvic pathogens. Therapeutic anticoagulation was initiated with low-molecular-weight heparin, followed by transition to an oral agent for three months due to embolic involvement beyond the pelvis.

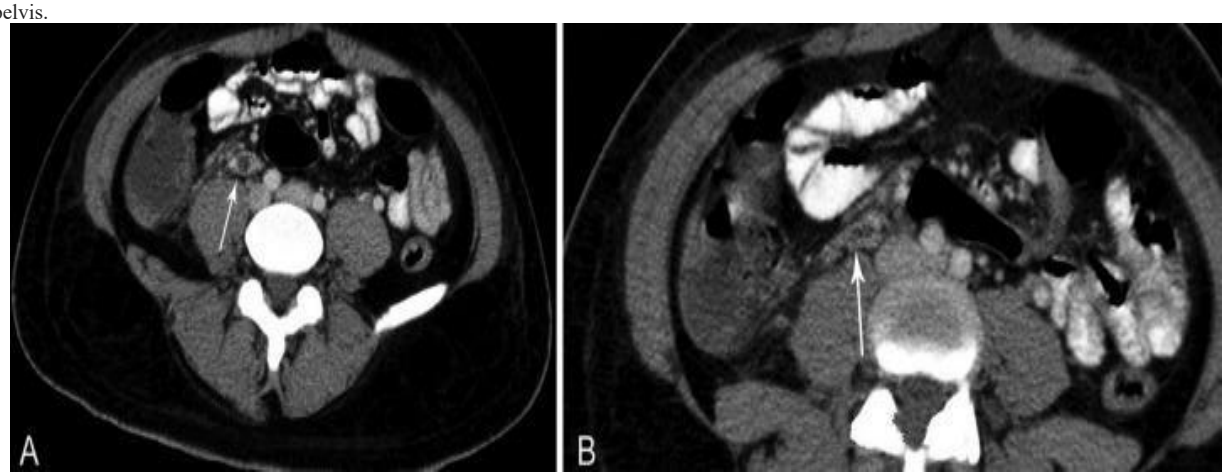


**Figure 1: Axial contrast-enhanced CT of the abdomen and pelvis. The arrow indicates an enlarged right ovarian vein with a central low-attenuation thrombus and surrounding perivenous fat stranding**

Analgesia and supportive care were provided. No invasive source control was required.

**Outcome and follow-up**

Defervescence occurred within 72 hours of initiating anticoagulation plus optimised antibiotics. At six weeks, the patient was asymptomatic; repeat CT abdomen/pelvis showed a reduction in thrombus calibre with re-established venous flow. By three months, chest CT demonstrated complete resolution of nodules, and anticoagulation was discontinued.

**Timeline**

| Date/Day | Event |
|---|---|
| **Day 0 (Post-abortion day 7)** | Fever and right iliac fossa pain; admitted; broad-spectrum antibiotics started |
| **Day 3** | Persistent fever; CT abdomen/pelvis: right ovarian vein thrombosis (OVT) with perivenous stranding |
| **Day 4** | CT pulmonary angiography: peripheral nodules consistent with septic pulmonary emboli (SPE); anticoagulation commenced |
| **Day 6–7** | Defervescence; pain improved; markers declining |
| **Week 6** | Asymptomatic; interval imaging: thrombus regression |
| **Month 3** | Pulmonary nodules resolved; anticoagulation stopped. |

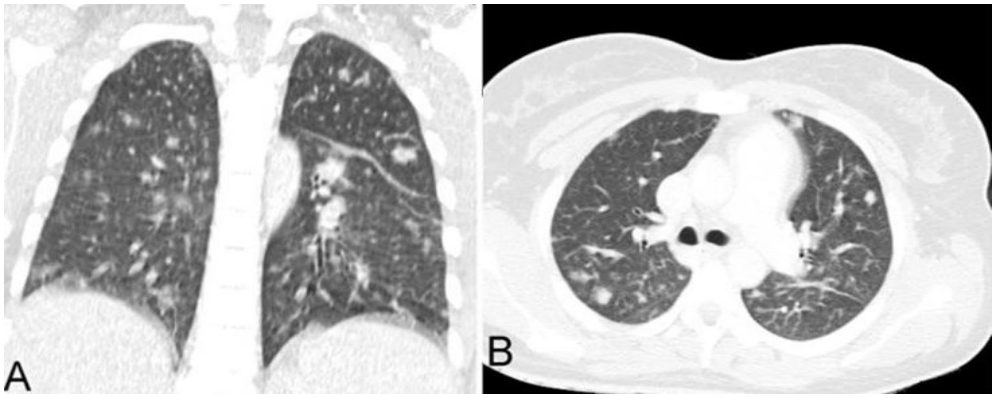


**Figure 2: Axial CT pulmonary angiography showing multiple bilateral, peripheral nodules compatible with septic pulmonary emboli/ Coronal CT pulmonary angiography reconstruction demonstrating peripheral, partly cavitating nodules**

**Patient's perspective**

"I kept spiking fevers despite strong antibiotics. Scans showed a clot in a pelvic vein and spots in my lungs. After starting blood thinners and continuing antibiotics, the fevers stopped, and I steadily recovered."

**Informed consent**

Written informed consent for publication of the case details and images was obtained using the BMJ consent form; a copy is held by the authors and is available to the journal on request.

**Learning points**

- Suspect septic pelvic thrombophlebitis when postpartum/post-abortion fever persists despite adequate antibiotics.
- Right ovarian vein thrombosis is common; CT or MR venography confirms the diagnosis.
- In the presence of septic pulmonary emboli, treat with appropriate antibiotics plus therapeutic anticoagulation and extend duration (typically ~3 months).

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# Sheehan's Syndrome with Dilated Cardiomyopathy: An Overlooked Sequela of Postpartum Hemorrhage

Jabar Imran[1], Shahroze Nayyar[2], Jahanzaib Hamid[3], Iqra Ashraf[4]

## Abstract

**Summary:** We present the case of a 43-year-old female who developed panhypopituitarism as a sequela of postpartum hemorrhage. She manifested with recurrent vomiting, diarrhea, hypotension, and hypoglycemia, and was subsequently diagnosed with Sheehan's syndrome complicated by dilated cardiomyopathy.Prompt initiation of hormonal replacement therapy resulted in remarkable clinical improvement. This case emphasizes the critical need for early recognition of Sheehan's syndrome in women with a history of obstetric hemorrhage, particularly when presenting with chronic, nonspecific complaints, to prevent life-threatening complications.

**Keywords:** Sheehan syndrome; postpartum hemorrhage; hypopituitarism; dilated cardiomyopathy; heart failure.

## Introduction

Sheehan's syndrome is a form of hypopituitarism that occurs as a result of ischaemic necrosis of the anterior pituitary gland following severe postpartum haemorrhage (PPH).[2] The pituitary gland undergoes hyperplasia during pregnancy, making it particularly vulnerable to vascular compromise in the setting of massive blood loss and hypovolaemic shock.[1,2] This condition, first described by Harold Sheehan in 1937, remains a major cause of hypopituitarism in developing countries where maternal health services and emergency obstetric care are often limited.[2,6,5]

The clinical manifestations of Sheehan's syndrome are variable and may appear immediately or many years after the inciting obstetric event. Acute presentations can include failure of lactation, severe fatigue, adrenal crisis with hypotension, and hypoglycaemia.[13,14] More chronic or delayed features include secondary amenorrhoea, loss of axillary and pubic hair, generalized weakness, cold intolerance, alopecia, coarse dry skin, and progressive features of hypothyroidism and adrenal insufficiency. Because these symptoms are nonspecific and evolve gradually, diagnosis is often delayed, sometimes until life-threatening metabolic or systemic complications develop.[2,6,15]

Although endocrine and metabolic manifestations are well documented, cardiac involvement in Sheehan's syndrome is exceedingly rare. Only isolated case reports have described dilated cardiomyopathy (DCM) as a complication.[4,7] The pathophysiology is thought to be multifactorial, involving prolonged untreated hypothyroidism, glucocorticoid deficiency impairing myocardial function, and possible autoimmune mechanisms. Importantly, DCM in this context has been shown to improve with appropriate hormone replacement, suggesting that early recognition and treatment are crucial.[8]

We present a case history of a middle-aged woman of Punjab, Pakistan, who had developed Sheehan syndrome following massive PPH and later on presented with panhypopituitarism, which was complicated by dilated cardiomyopathy.[3,8] The case contributes to the scarcity of the literature on cardiac manifestations of Sheehan syndrome and emphasizes the role of early diagnosis in women who have experienced obstetric haemorrhage and who present.

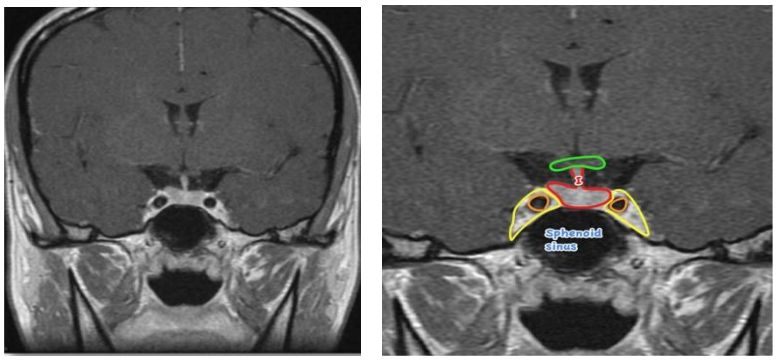


**Figure 1: A.B Normal Pituitary Gland(Coronal Images)**

## Case Presentation

A 43-year-old homemaker, married for 18 years, with no prior medical comorbidities, presented with recurrent vomiting, diarrhea, and abdominal pain of three days' duration. These episodes were associated with dizziness, drowsiness, and symptomatic hypoglycaemia. She also reported cold intolerance, progressive alopecia, reduced libido, generalized weakness, and increasing exertional dyspnoea over the past several years.

**Contributions:**
JI SN JH IA - Conception, Design
- Acquisition, Analysis, Interpretation
JI SN JH - Drafting
IA - Critical Review

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Her obstetric history was significant. She was Gravida 4, Para 4. Her last childbirth in 2013 was complicated by massive postpartum haemorrhage due to delayed placental expulsion. She required intensive care unit admission, surgical evacuation, and transfusion of eight units of red cell concentrated. Although she survived the acute event, she developed secondary amenorrhoea and infertility within two months of delivery, accompanied by progressive fatigue and loss of secondary sexual characteristics.

On examination, her blood pressure was 100/70 mmHg supine and dropped to 80/60 mmHg on standing, with a pulse of 78/minute, low volume. Random capillary blood glucose was 68 mg/dL. Physical features included dry coarse skin, thinning of scalp hair, loss of the lateral third of eyebrows, and bilateral pedal oedema.

**Table 1: Baseline Laboratory Investigations**

| Investigation | Result / Findings |
|---|---|
| **Peripheral film** | Microcytic hypochromic anemia |
| **Serum Iron** | **48 µg/dL** (Reference: 50–170 µg/dL) |
| **Serum TIBC (Total Iron Binding Capacity)** | **184 µg/dL** (Reference: 250–400.9 µg/dL) |
| **Serum Folate Levels** | **2.4 ng/mL** (Reference: >3.3 ng/mL) |
| **Serum Vitamin D** | **14.6 ng/mL** (Normal: ≥20 ng/mL) |
| **Serum Vitamin B12** | 270 pg/mL (Reference: 208–964 pg/mL) |
| **Urine routine examination** | Normal |
| **Abdominal Ultrasound** | Unremarkable |
| **Chest X-Ray** | Normal |
| **Serum Cholesterol** | **211 mg/dL** (Reference: 125–200 mg/dL) |
| **Serum Triglyceride** | 127 mg/dL (Reference: <150 mg/dL) |
| **Anti-nuclear antibody (ANA)** | Negative |
| **Anti-TTG Ab (IgA)** | 1 (Reference: <10) |
| **TSH** | 1.34 µl U/mL (0.3-4.5) |
| **T3** | 1.15 ng/mL (0.69-2.15) |
| **T4** | 7.97 ng/mL (52-127) |
| **Prolactin** | 14.1 µl U/mL (54-490) |
| **FSH** | 6.18 IU/L (3.03-8.08 IU/L) |
| **LH** | 1.60 IU/L (1.80-11.78 IU/L) |
| **Serum IGF-1 levels** | 14.7 nmole/L (18.4-37.12) |
| **Estradiol** | 55.6 pg/mL (692-1286 pg/mL) |
| **Progesterone** | 0.394 ng/mL (143 ng/mL) |
| **C-peptide levels (Fasting)** | 1.9 ng/mL (1.1-5.0) |
| **Serum Insulin (Fasting)** | 7.5 µl U/mL (5-25) |
| **Serum ACTH (morning)** | 13.5 pg/mL (7-69) |

**Table 2: Follow-up Laboratory tests**

| Laboratory Test | 16/11/2022 | 13/01/2023 |
|---|---|---|
| **WBC** | 8.5 | 8.0 |
| **Hemoglobin (Hgb)** | **11.1** | **11.0** |
| **MCV** | 76.3 | 85.3 |
| **MCH** | 29.4 | 29.9 |
| **Platelets** | 179 | 364 |
| **PT** | 13 | — |
| **APTT** | 36 | — |
| **Urea** | **62** | 26 |
| **Creatinine** | **1.3** | 0.7 |
| **Total Bilirubin** | 0.6 | 0.3 |
| **ALT** | **66** | 19 |
| **ALP** | 134 | 65 |
| **Sodium** | 136 | 137 |

**Radiological Investigations:**

Electrocardiography showed nonspecific T-wave inversions(Fig 2). Echocardiography revealed globally reduced left ventricular contractility with an ejection fraction of 35–40%, while cardiac MRI confirmed dilated cardiomyopathy with biventricular dysfunction(Fig 3). Pituitary MRI demonstrated an empty sella(Fig 4B). Panthytopituitarism was being diagnosed by hormonal tests; the levels of cortisol, thyroid hormones, gonadotropins, oestradiol, and prolactin were reduced. Other laboratory tests, such as renal and liver, were normal.

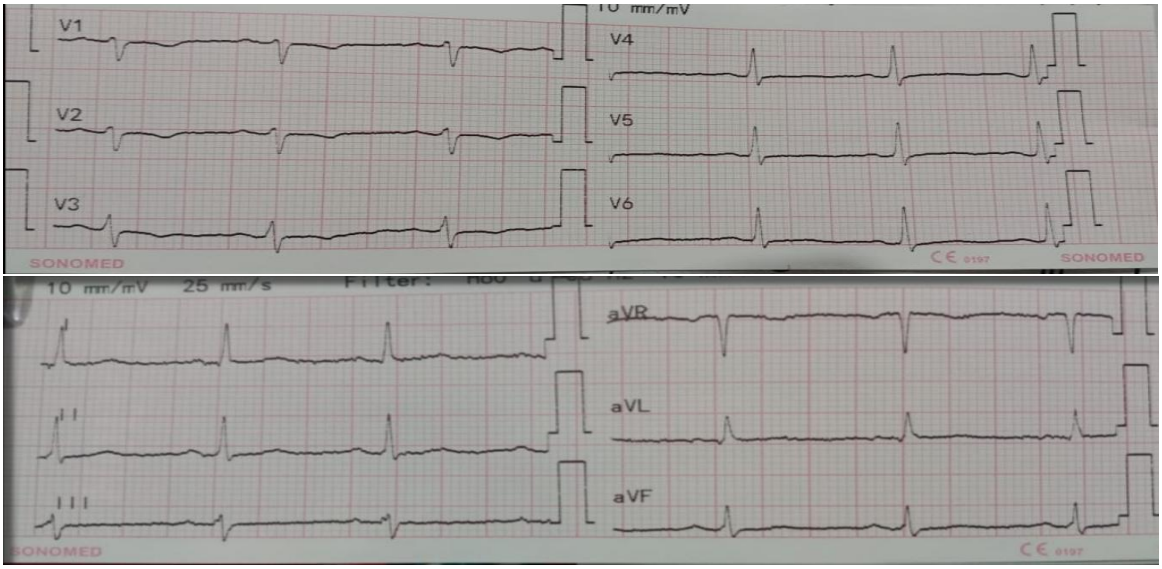


**Figure 2: ECG shows normal sinus rhythm with normal axis, intervals, and R-wave progression. Non-specific T-wave inversions in V1–V3 with T-wave flattening in V4–V6 and limb leads**

**Outcome and Follow-up**: Sheehan syndrome, complicated with dilated cardiomyopathy, was diagnosed. Her acute presentation involved stabilization using intravenous fluids and antiemetics. Prednisolone at 7.5 mg/day, levothyroxine at 50 0g/day, and oestrogen-progesterone therapy were started. Social therapy was also administered to support cardiomyopathy. She had recovered her symptoms of fatigue, postural dizziness, and hypoglycaemia on follow-up twice in two weeks. Blood pressure stabilized at 100/70 mmHg, and her thyroid profile normalized. Repeat echocardiography showed improvement in systolic blood pressure. The patient reported a significant improvement in energy, appetite, and mood, with resumption of daily household activities.

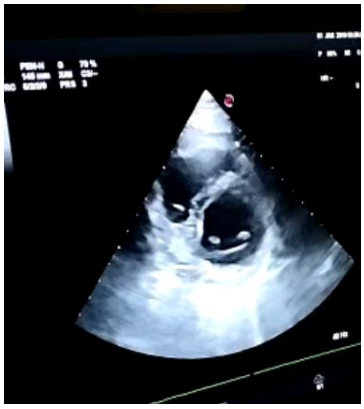

**Figure 3: EF 35% -40% Moderate LV systolic dysfunction. Global hypokinesia is present**

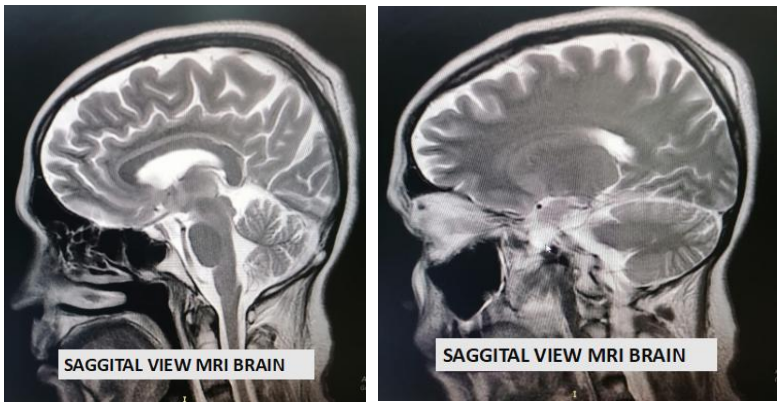


**Figure 4: (A) Early — enlarged pituitary with low T1, high T2 homogeneous signal, ring enhancement; (B) Late — empty sella of normal size**

## Discussion

Sheehan's syndrome remains a frequently underdiagnosed condition because of its insidious onset and non-specific manifestations. Women may present years after the inciting obstetric insult, as in our patient who developed clinical features eight years following a severe postpartum hemorrhage. This delay reflects both the subtlety of symptoms and the lack of awareness among healthcare providers.[2,3]

The endocrine deficiencies in Sheehan's syndrome often present gradually, with amenorrhea, fatigue, and generalized weakness being the earliest indicators.[3,11] Because these symptoms overlap with common post-pregnancy complaints, the diagnosis is easily overlooked. Recognition requires a high index of suspicion, especially in women with a history of complicated childbirth and subsequent menstrual irregularities.

In the present case, the most remarkable feature was the presence of dilated cardiomyopathy. While it is established that hypothyroidism and adrenal insufficiency can impair myocardial contractility, progression to overt dilated cardiomyopathy is unusual. This highlights the potential severity of long-standing untreated hypopituitarism.[7,8]

In Sheehan syndrome, the pathogenesis of cardiomyopathy is most likely multifactorial. Besides direct impacts of thyroid and adrenal hormone deficiency, the autoimmune process has been suggested to be a contributory factor.[12] This intersection indicates that the syndrome Sheehan has could not only be an endocrine disorder, but this syndrome could also have an immunological overlap.

Notably, timely and appropriate hormone replacement has been shown to improve the cardiac performance of such patients, and in some cases, the cardiomyopathy has been reversed.[9,10] This demonstrates the importance of early diagnosis and management of hypopituitarism.

On a larger population health scale, the case highlights the persistence of postpartum hemorrhage-associated morbidity in Pakistan. To avoid the development of long-term complications like the case of Sheehan, it is important to strengthen maternal healthcare services, improve obstetric care, and provide proper follow-up for high-risk women.

## Conclusion

The results of the work showed that two weeks of proper hormone replacement therapy resulted in a significant clinical improvement in the patient. Her fatigue, frequent instances of hypotension, and episodes of hypoglycemia were fully cured. The blood pressure leveled at 100 / 70 mmHg without the necessity to use support measures, her thyroid picture was restored to normal levels, which is a clear indication of sufficient endocrine treatment. There was a significant increase in the left ventricular systolic function on follow-up echocardiography, which indicates the recovery of the previously reported dilated cardiomyopathy. The patient also mentioned a major improvement in the overall well-being, such as an increase in the level of energy, the regained appetite, and a more stable mood. All these changes indicate the reversibility of cardiac dysfunction caused by endocrine with early hormone replacement.

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# A Hidden Pigment, A Damaged Joint: The Ochronotic Knee - A Case Report

Saad Riaz[1], Osama Ijaz[2], Talha Farooq[3]

## Abstract

**Summary:** Ochronotic arthropathy (alkaptonuria) is a rare hereditary disorder that poses a diagnostic dilemma. It is usually diagnosed perioperatively or by conducting investigations based on perioperative findings in the postoperative period, followed by retrospective analysis. This report discusses a case of a 37-year-old male who presented with bilateral knee pain. Radiological studies were indicative of grade III osteoarthritis but perioperatively black pigmentation was noted. Backtracking revealed it to be a case of alkaptonuria. This typically presents as early-onset osteoarthritis involving major joints like the knee, hip, shoulder, and spine. Orthopedic surgeons should have an inquisitive mind and keep this as their differential diagnosis while dealing with young patients with similar symptoms to avoid surprises during surgery. Our study aims to increase understanding of this condition for effective management and improving functional outcomes.

**Keywords:** Alkaptonuria, Ochronosis, Osteoarthritis, Rare Disease.

## Introduction

Ochronotic arthropathy is a rare condition that occurs in patients of alkaptonuria with a reported incidence of 1:100 000–1:250 000 live births worldwide. It follows an autosomal recessive mode of inheritance.[1] This disorder has a higher prevalence in regions of Slovakia and the Dominican Republic.[2,3] Pathophysiology of this rare metabolic disorder lies in the accumulation of homogentisic acid (HA) due to deficiency of the enzyme homogentisic oxidase[1,2]. HA oxidizes, converts to melanin-like pigments, and then deposits in connective tissue rich in collagen, like ligaments, tendons, and joints, inducing early degenerative changes.[4]
The knee joint is the most commonly affected large joint, whereas the hip and sacroiliac joints are the next most affected joints. Usually, stiffness and pain of the knees and hip become evident in the 3rd–4th decade of life. Other manifestations occur due to HA accumulation in the sclera, skin, heart valves, nose and ears cartilage, renal tubule epithelial cells, pancreas, central nervous system, endocrine organs, respiratory organs, and arteries.[6] Currently, nitisinone is the only approved medical treatment available to reduce HA levels, and symptomatic treatment can be done with local heat, physiotherapy, and analgesics. Some possible surgical options for the affected joint are synovectomy, arthroscopic debridement, fusion, or arthroplasty.[7-9] This report describes a case of ochronotic knee that presented with OA at a young age.

## Case Presentation

A 37-year-old male patient who was a driver by profession presented to our outpatient department. He had bilateral knee pain, which started 1.5 years back and gradually progressed over time. Pain was more on the left side and was recurrent in nature, as it only temporarily responded to oral analgesics and anti-inflammatory drugs, which he took from various small clinics he visited previously. He had multiple joint fluid routine examinations performed as well, which were normal. His uric acid levels used to fluctuate between 6.8-7.5 mg/dl, and he was put on anti-hyperuricemia agents (febuxostat, allopurinol) by some practitioners, which used to decrease his uric acid levels, but pain did not improve. There was no positive family history. On examining the patient, there were no cutaneous signs of ochronosis (**Error! Reference source not found.**). Although both knees were neutral aligned, mild effusion on the left side was noted (for which R/E was performed by some clinicians). Upon deep palpation, he experienced tenderness along the medial joint line of the left side. The range of motion on the right side was 0-100°, and on the left was 0-90°. Standing radiographs were taken with orthogonal views. These showed advanced degenerative disease, especially on the left. We see joint space narrowing, loose bodies, and marginal osteophytes. (**Error! Reference source not found.**). Since the patient was non-affording, there was a delay in intervention as multiple previous consultations advised him to undergo arthroscopic evaluation, but he refused due to financial limitations. We decided to proceed with total knee replacement after pre-anesthesia assessment, but as he could not afford it considering the needs of the patient, we planned to do open arthrotomy with synovial biopsy, thorough joint lavage, and removal of loose bodies, and proceed according to intraoperative findings. We proceeded by anterior midline incision with a medial para-patellar approach. We noticed multiple black patches over the articular surface of the joint along with arthritic changes. There were 3-4 loose bodies in the joint as well (Figure 3). Peri-operatively, it was discussed as well to proceed with knee arthroplasty, but due to financial concerns of the patient, we were limited to taking a synovial biopsy, removal of loose bodies, and joint lavage. Keeping in mind the operative findings patient was investigated again, starting from history. His mother revealed that his diaper used to get dark stains, which were very difficult to remove. Similarly, one of his sisters had chronic back pain. Careful clinical examination showed one of his ears to have a slight blackish tinge. Urine stored for 6-8 hours changed its color to black (Figure 4). Urinary HA levels were done, biopsy sample sent came negative for malignancy and synovial chondromatosis. Retrospective diagnosis of ochronotic arthropathy was established. The biopsy sample sent came back negative for malignancy and synovial chondromatosis. Post-operatively, knee physiotherapy was started immediately on the second postoperative day. His pain improved

**Contributions:**
SR OI - Conception, Design
OI - Acquisition, Analysis, Interpretation
OI TF - Drafting
SR - Critical Review

All authors approved the final version to be published & agreed to be accountable for all aspects of the work.

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postoperatively. On his first visit, 2 weeks after surgery, he was satisfied with the results as his pain improved, and he was able to carry out his daily activities with ease. He was advised regarding the probable need for arthroplasty in the future. At his last follow-up, six months post-surgery, the patient remained pain-free and had resumed his occupation comfortably; however, he was subsequently lost to follow-up.

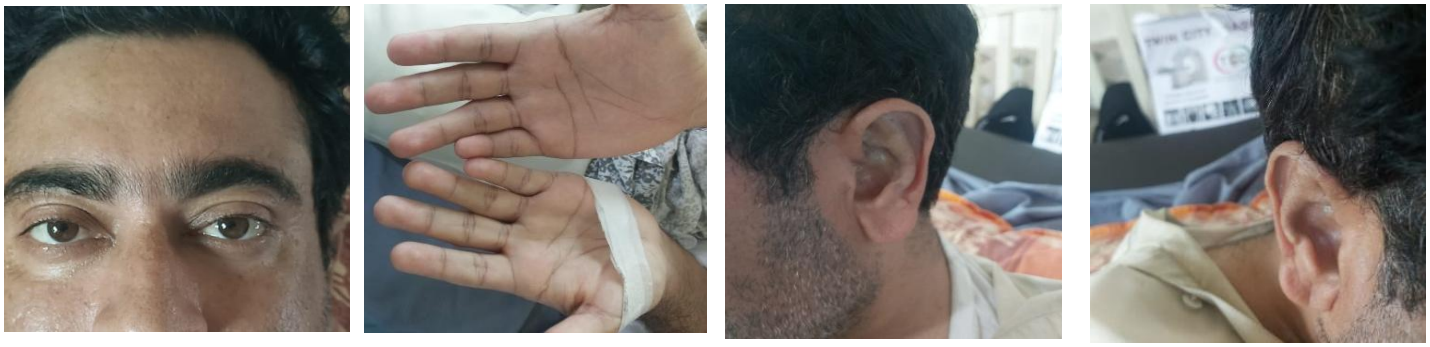

**Figure 1**: **Cutaneous Findings**

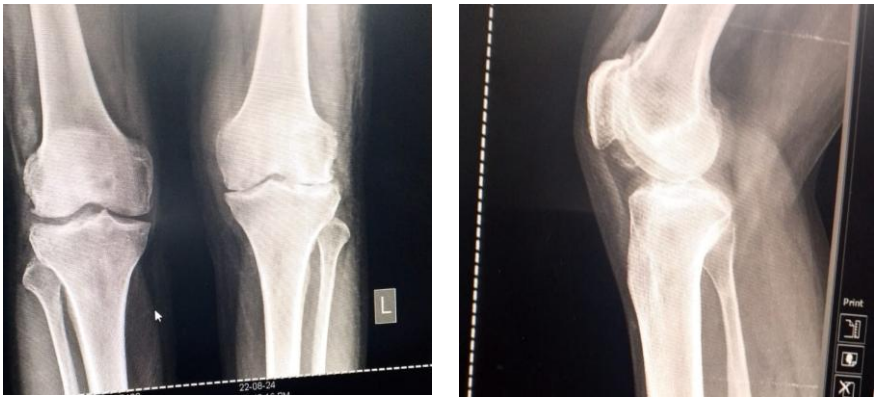

**Figure 2**: **Knee Radiographs showing advanced osteoarthritis**

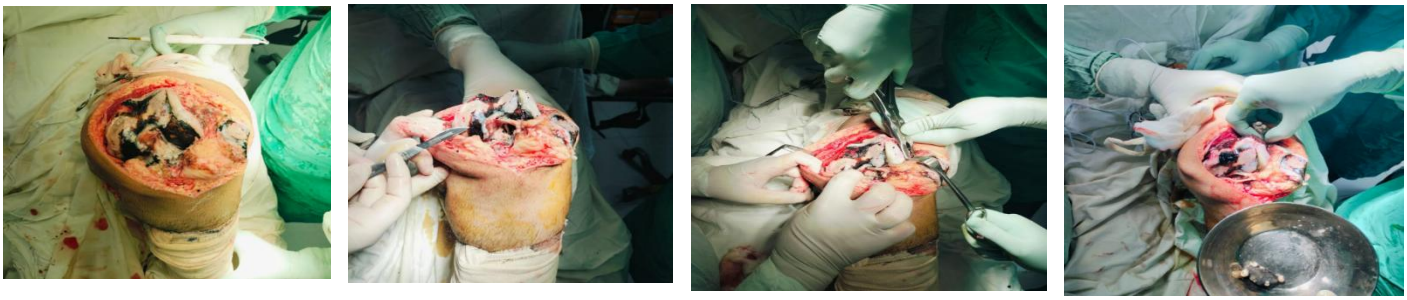

**Figure 3**: **Perioperative findings showing multiple black pigment patches over the articular surface and loose bodies in the joint**

## Discussion

Alkaptonuria is a rare disease that occurs due to the accumulation of homogentisic acid in the cartilages and joints. In the fourth to fifth decade of life, joint pain was reported mostly in the hips and knees.[4] In about half the cases, the involvement of intervertebral discs at the thoracolumbar region is early on. Noticing dark spots in the diapers of babies during the first months of life can be a sign of alkaptonuria.[10,11] Clinical manifestations occur mostly in males. Exposure to the air of the HA causes its oxidation to benzoquinone acetate (BQA), which darkens the urine. Additional clinical symptoms are found, including discoloration of the sclera and cornea[5]. You can easily see skin pigmentation on the ulnar or radial surface of your hand. The ulnar side of your fingers also show additonal skin pigmentation. Nitisinone is the first FDA-approved treatment for alkaptonuria. It works by blocking the enzyme 4-hydroxyphenylpyruvate dioxygenase and can lower urinary homogentisic acid levels by up to 97%.[12,13] Recent studies have shown that patients treated with nitisinone experience significant improvements in pain, energy levels, and physical functioning.[14,15] In the early stages of osteoarthritis (OA), conservative therapy is effective. Another study reveals that health workers find it effective if started early with the use of individual follow-up to prevent side effects.[3] However, in severe cases affecting the hip or knee joints, arthroplasty is the only treatment that can improve quality of life.[16] Multiple studies have indicated that arthroplasty is a suitable option for this condition. It reduces pain and enhances patient mobility. A literature review by Singh, Liu, Awad, and Couto confirms that this treatment is both safe and effective for cases of ochronotic arthropathy.[17-19] However, a study by Narvekar et al.,[20] suggests that arthroscopic intervention can be useful for diagnosis and to avoid unexpected findings during surgery.

So, orthopaedic surgeons should be vigilant for unusual signs in early-onset osteoarthritis to avoid surprises during surgery related to darkened cartilage.

**Figure 4**: **Change in urine color**

## Conclusion

Ochronotic arthropathy, although it is a rare condition, should always be considered in patients with early-onset osteoarthritis, particularly in younger patients. Awareness about this disorder helps to prevent perioperative surprises and helps in early diagnosis and proper management. Arthroscopy could be a useful diagnostic tool in doubtful cases, while arthroplasty remains an effective treatment option for this condition. Combining a high index of suspicion with thorough clinical history and careful observation of cutaneous findings could aid in early diagnosis. Early recognition of this uncommon entity and vigilance could improve functional outcomes.

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