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RESEARCH ARTICLE

AUTOIMMUNE POLYGLANDULAR SYNDROME TYPE 2 PRESENTING WITH PERICARDIAL EFFUSION: A DIAGNOSTIC CHALLENGE

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ABSTRACT

Background: Autoimmune Polyglandular Syndrome Type 2 (APS-2), or Schmidt syndrome, is a rare autoimmune disorder marked by the coexistence of Addison's disease with autoimmune thyroid disease and/or type 1 diabetes mellitus. Its nonspecific symptoms often lead to delayed diagnosis. While pericardial effusion is an uncommon manifestation of APS-2, it can pose diagnostic challenges. ***Case Presentation:*** A 44-year-old female presented with progressive pedal edema, facial puffiness, and dyspnea for three months. Clinical examination revealed non-pitting pedal edema, hyperpigmentation, dry skin, and muffled heart sounds. Investigations showed pericardial effusion confirmed by echocardiography, with transudative fluid secondary to hypothyroidism, as evidenced by elevated TSH and positive anti-TPO antibodies. The patient was treated with thyroxine and diuretics. During hospitalization, she developed worsening dyspnea, hypotension, and hypoglycemia, raising suspicion of adrenal insufficiency. Morning cortisol and ACTH levels confirmed primary adrenal insufficiency, leading to the diagnosis of APS-2. Treatment with intravenous hydrocortisone stabilized her condition, with significant clinical improvement. ***Discussion:*** This case highlights the diagnostic complexity of APS-2, particularly with atypical initial presentations like pericardial effusion. The pericardial effusion was attributed to hypothyroidism, a rare but recognized cause. Delayed recognition of adrenal insufficiency often complicates APS-2 diagnosis, necessitating high clinical suspicion for timely intervention. ***Conclusion:*** This case emphasizes the importance of recognizing multisystemic symptoms suggestive of APS-2. Prompt diagnosis and treatment with hormone replacement therapy can prevent life-threatening complications, underscoring the need for a comprehensive endocrine evaluation in patients with atypical presentations.

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INTRODUCTION

Autoimmune Polyglandular Syndrome Type 2 (APS-2), also known as Schmidt syndrome, is a rare disorder characterized by the coexistence of Addison's disease with autoimmune thyroid disease and/or type 1 diabetes mellitus.[1,2] APS-2 primarily manifests in adulthood with a female predominance, often with subtle and nonspecific symptoms, leading to diagnostic delays.[2,3] It results from autoimmune-mediated loss of immune tolerance, causing progressive endocrine gland destruction. Addison's disease, commonly the earliest sign of APS-2, is frequently underdiagnosed because symptoms like fatigue, hyperpigmentation, and orthostatic hypotension overlap with more common conditions.[4] Recognizing the multisystemic nature of APS-2 is essential, as delayed diagnosis can lead to severe complications such as adrenal crises or myxedema coma.

Pericardial effusion, although an uncommon presentation of APS-2, can occur due to autoimmune thyroid disease.[6] This case report discusses a 44-year-old female who presented with pericardial effusion, pedal edema, and progressive dyspnea, followed by an Addisonian crisis during hospitalization. The report highlights the diagnostic challenges posed by rare manifestations of APS-2, emphasizing the importance of a high clinical suspicion.

CASE PRESENTATION

A 44 year old female presented to medicine emergency with complaints of Pedal edema and facial puffiness for 3 months and Shortness of breath for 1 month The pedal edema was insidious in onset, progressively increasing, and associated with facial puffiness. The shortness of breath was also insidious in onset and progressive in nature. Initially, it was limited to exertional activities, such as climbing stairs, but later, the patient experienced breathlessness even after walking

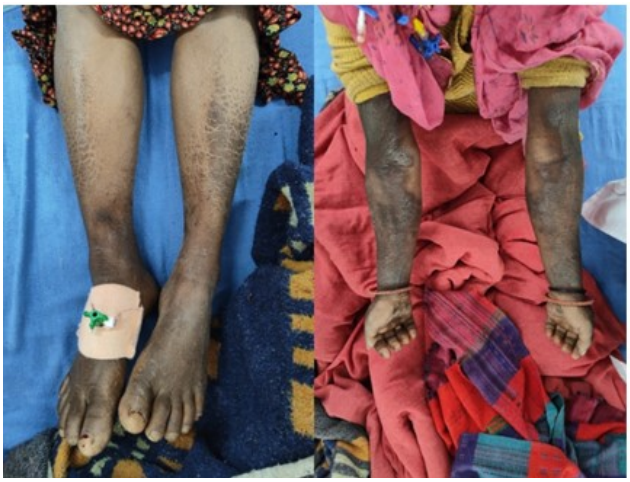

Figure 1. Hyperpigmentation of hands and legs

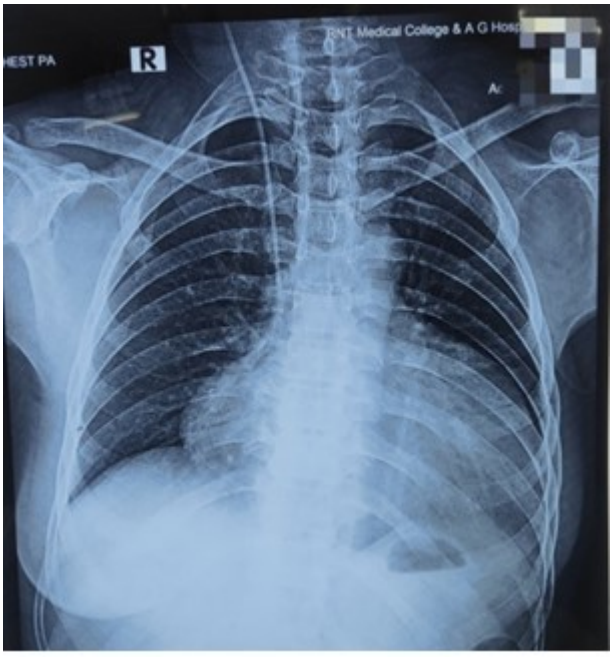

Figure 2. X ray showing enlarged cardiac silhouette

a few steps The symptoms were aggravated in the supine position and relieved by sitting upright. There was no history of fever, cough with expectoration, chest pain, palpitations, or decreased urine output. The patient also reported amenorrhea for the past seven years but denied any previous diagnosis of chronic illness. On clinical examination, she had non-pitting pedal edema, facial puffiness, and dry skin associated with hyperpigmentation of both upper and lower limbs. Respiratory examination revealed fine crepitations bilaterally in the infra-scapular and infra-axillary areas. Cardiac examination showed muffled heart sounds, but no murmurs were appreciated. Vital signs were recorded as: blood pressure: 100/70 mm Hg, pulse rate: 70/min, respiratory rate: 24/min, SpO2:93% on room air, and temperature: 98.6°F.

Initial investigations included a chest X-ray, which showed an enlarged cardiac silhouette, and an ECG, which revealed low voltage QRS complexes. A provisional diagnosis of pericardial effusion was made, and the patient was admitted for further evaluation. Routine blood investigations were done along with thyroid function tests. Her complete blood count showed hemoglobin 9.6g/dL, WBCs 6900/mm³, and platelets 203000/mm³.

Liver function tests revealed SGOT 29 U/L, SGPT 36 U/L, total bilirubin 0.56 mg/dL, direct bilirubin 0.23 mg/dL, ALP 60 U/L. Renal function tests indicated urea 26.8 mg/dL and creatinine 1.0 mg/dL. Electrolytes were Na:116 mmol/dl and K:4.1 mmol/L. Thyroid function tests indicated primary hypothyroidism with fT3 of 0.3 nmol/L, fT4 of 5.4 nmol/L, and TSH of 100 IU/ml. Echocardiography showed gross pericardial effusion without signs of cardiac tamponade with Grade 2 Diastolic dysfunction. Diagnostic pericardiocentesis was done which revealed transudative type ruling out infective and inflammatory etiologies. A diagnosis of pericardial effusion secondary to hypothyroidism was made. Further testing revealed anti-TPO positivity (202 IU/ml), confirming autoimmune hypothyroidism. The patient was started on Thyroxine 100 mcg once daily and Furosemide 40 mg twice daily. On the third day of admission, the patient developed worsening shortness of breath and uneasiness. Her systolic blood pressure dropped to 40 mm Hg, with a pulse rate of 110/min and respiratory rate of 30/min. She was shifted to the ICU, started on vasopressors, and provided oxygen support. An urgent echocardiogram was performed, which again revealed no signs of cardiac tamponade and >50% IVC collapse, ruling out hypovolemia. Repeat total leukocyte counts were within normal limits, ruling out sepsis. Later in the day, the patient experienced an episode of hypoglycemia, raising the suspicion of an Addisonian crisis. Morning cortisol levels were 7.6 mcg/dL, and ACTH levels were 156 pg/ml, confirming primary adrenal insufficiency. Given the coexistence of autoimmune hypothyroidism and primary adrenal insufficiency, a diagnosis of Autoimmune Polyglandular Syndrome Type 2 (APS-2) was established. She was started on intravenous hydrocortisone 100 mg TDS, which led to stabilization of her vitals within 24 hours. Her sodium levels improved progressively, and she was eventually shifted back to the general ward.

DISCUSSION

This case highlights the diagnostic challenges of autoimmune polyglandular syndrome type 2 (APS-2), especially with rare initial presentations like pericardial effusion. APS-2, though primarily defined by adrenal and thyroid dysfunction, can present with atypical features that complicate diagnosis and delay treatment.[6] Pericardial effusion in APS-2 is exceedingly rare, with only a few reports documenting this as an initial manifestation.[7] The pericardial effusion in this case was likely due to hypothyroidism, a known cause of pericardial fluid accumulation.[8] Hypothyroidism-induced pericardial effusions are generally slow to develop and often asymptomatic; however, they can progress to tamponade if untreated.[8] Adrenal insufficiency, the cornerstone of APS-2, is frequently underdiagnosed because of nonspecific symptoms like fatigue, hypotension, and pigmentation changes.[4] These nonspecific symptoms, coupled with the rarity of APS-2, often contribute to significant delays in diagnosis.[9] High clinical suspicion and a thorough endocrine evaluation are essential, especially in patients with multisystemic involvement or atypical presentations, such as cardiovascular manifestations.[6,10] Management involves addressing the underlying endocrinopathies. In this case, glucocorticoid and thyroid hormone replacement were initiated promptly after diagnosis, resulting in clinical improvement. Timely recognition and treatment are critical to prevent life-threatening complications, including adrenal crises and myxedema coma.[4,11]

This case emphasizes the importance of maintaining a broad differential diagnosis when encountering unexplained multisystemic symptoms. It highlights the importance of maintaining a high index of suspicion for APS-2 in patients presenting with atypical features like pericardial effusion. Prompt recognition and comprehensive endocrine evaluation are essential to prevent life-threatening complications and improve patient prognosis.[7,12]

CONCLUSION

This case highlights the complex nature of Autoimmune Polyglandular Syndrome Type 2 (APS-2), especially when it manifests through atypical symptoms like pericardial effusion. The initial clinical features, including pedal edema, facial puffiness, shortness of breath, and enlarged cardiac silhouette on Chest X ray and low voltage ECG led to a provisional diagnosis of pericardial effusion, confirmed by echocardiography, which was later linked to hypothyroidism. The development of adrenal insufficiency, highlighted by the episode of hypoglycemia and confirmed by low cortisol levels, prompted the diagnosis of APS-2. Early recognition of the multisystem involvement and prompt treatment with glucocorticoid and thyroid hormone replacement resulted in stabilization and recovery. This case emphasizes the importance of maintaining a high clinical suspicion for rare endocrine syndromes in patients with unexplained multisystemic symptoms, as timely intervention can significantly improve patient outcomes and prevent potentially life-threatening complications.

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