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Original Research Article

A Rare Case of Autoimmune Polyglandular Syndrome (Type 1 Diabetes Mellitus, Hypothyroidism, Hypogonadism), Systemic Lupus Erythematosus Associated with Dyslipidemia

Kumar Kamal[1], Mehta Neha[2], Goel RK[3], Raihan MD[4], Kumar Sushil[5]

[1]DM Student, Department of Cardiology, Sardar Patel Medical College, Bikaner, Rajasthan
[2]PG Student, Department of Dermatology, Rama Medical College, Hapur, Uttar Pradesh
[3]Professor and Head, Department of Medicine, Maharaja Agrasen Medical College, Agroha, Haryana
[4]Professor and Head, Department of Dermatology, Rama Medical College, Hapur, Uttar Pradesh
[5]Assistant Professor, Department of Medicine, Maharaja Agrasen Medical College, Agroha, Haryana

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Corresponding author: Dr. Kamal Kumar
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Abstract:

Introduction: Autoimmune polyglandular syndromes (APS) are rare disorders characterized by the presence of multiple autoimmune diseases. This abstract highlights a unique case of a 26-year-old female with coexisting Type 1 Diabetes Mellitus (T1DM), hypothyroidism, hypogonadism, systemic lupus erythematosus (SLE), and dyslipidemia, emphasizing the complexity of APS and the importance of early recognition.

Case Presentation: A 26-year-old female presented with T1DM at age 15, hypothyroidism at age 18, hypogonadism at age 24, and SLE symptoms at age 25. Lab tests confirmed SLE and revealed dyslipidemia. No familial autoimmune history was evident.

Conclusion: This case underscores the diverse clinical manifestations within APS. Co-occurrence of T1DM, hypothyroidism, hypogonadism, SLE, and dyslipidemia in one patient is rare. Prompt identification and management of such complex autoimmune conditions are vital for patient well-being and outcomes, requiring a multidisciplinary approach to care.

Keywords: Autoimmune polyglandular syndromes (APS), Coexistence of autoimmune disease, Complex clinical manifestations.

Introduction

Autoimmune polyglandular syndromes (APS) are rare conditions where two or more autoimmune diseases occur simultaneously within a person. APS has two subtypes: APS-1 caused by genetic mutations in the autoimmune regulator (AIRE) gene, characterized by a unique triad of chronic mucocutaneous candidiasis, hypoparathyroidism, and Addison's disease [1] and APS-2, more common involves autoimmune disorders like thyroid diseases, celiac disease, Addison's disease, and type 1 diabetes. [2] Nevertheless, APS-2 is not limited to these conditions and can involve other autoimmune diseases such as pernicious anemia and vitiligo. [3]

Case Description: A 26-year-old female presented with a complex medical history, including a known diagnosis of Type 1 Diabetes Mellitus (T1DM) and hypothyroidism. She complained of multiple reddish-yellow lesions on both hands and had experienced the following symptoms over the past 3-4 years; facial rash with photosensitivity, hair loss, recurrent oral and nasal ulcers, and absence of secondary sexual characteristics.

Notably, the patient had never reached menarche. There was no family history of similar complaints. She reported decreased appetite but had a normal sleep pattern and no urinary or gastrointestinal symptoms.

Physical examination revealed normal vital signs, diffuse non-scarring alopecia, oral and nasal ulcers, multiple reddish-yellow firm papules on the ventral and dorsal aspects of both hands, and a malar rash. Laboratory investigations revealed a positive antinuclear antibody (ANA) profile with a speckled pattern (end point titre 1:160), negative dsDNA, negative anti-phospholipid antibody (APLA) profile, and low serum calcium levels.

Ultrasonography of the abdomen and pelvis showed bilateral streak ovaries and a small-sized uterus (3.5x1.2 cm). Hormonal assessments, including ACTH and PTH, were not performed. Histological examination of the lesions on the hands revealed foamy, xanthomatous histiocytes

scattered throughout multiple levels of the dermis, with the highest concentration in the superficial dermis.

The diagnosis of SLE was based on clinical symptoms (malar rash, ulcers, alopecia, photosensitivity) and a positive ANA test (speckled pattern, titer 1:160), despite a negative anti-dsDNA, consistent with autoimmune features.

Table 1: Showing patient investigation results with reference value

S.N.	Lab Test	Value	Reference Value
1	Blood Sugar	260 mg/dl	60-160 mg/dl
2	HbA1C	14.4%	5.7—6.4%
3	Total Cholesterol	1734 mg/dl	125-200 mg/dl
4	S. Triglycerides	692 mg/dl	60-170 mg/dl
5	Anti TTG	Negative	
6	ACTH Leve	>15 ug/dl	15-25 ug/dl
7	TSH	44 mlU/L	0.5-5 mlU/L

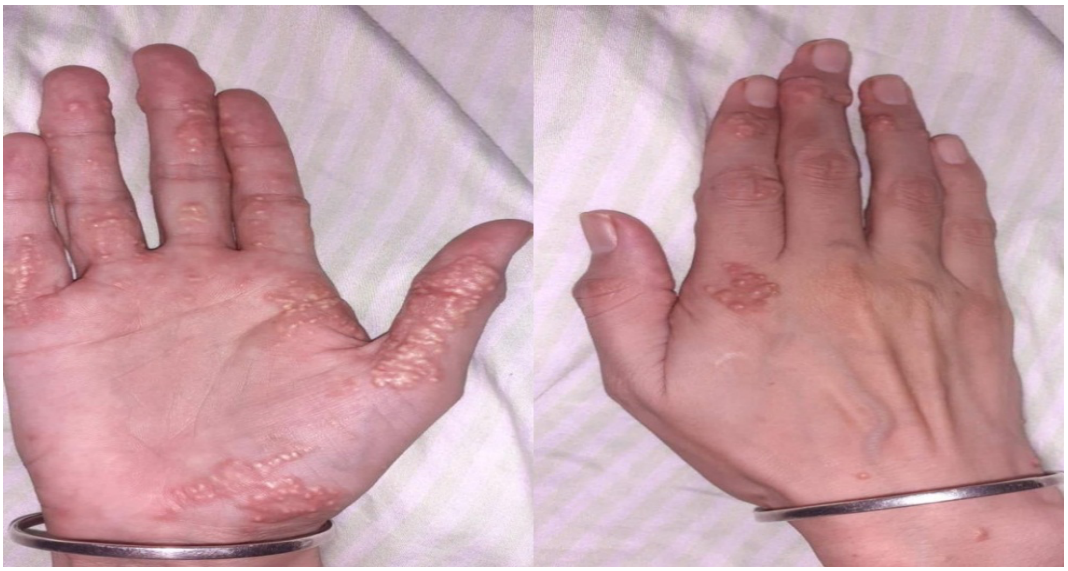

Figure 1: Reddish yellow firm papules on ventral and dorsal aspects

Discussion

Autoimmune polyglandular syndrome, a rare condition with an incidence of 1 in 200,000, is characterized by simultaneous presence of multiple autoimmune diseases. [4] In this case report the diagnosis of APS in the 26-year-old female was established through a comprehensive approach. Clinical evaluation and medical history assessment revealed a rare manifestation of APS, characterized by co-occurrence of T1DM, hypothyroidism, hypogonadism, non-scarring alopecia, SLE, and dyslipidemia, indicative of multiple autoimmune diseases. To confirm APS, laboratory tests and genetic testing were performed. Type 1 Diabetes Mellitus (T1DM) is an autoimmune condition characterized by the destruction of insulin-producing beta cells in the pancreas. While it's common in adolescence, simultaneous occurrence with other autoimmune disorders, as seen here, is rare. [5]

The presence of hypothyroidism, diagnosed at a young age, is an additional diagnostic complexity. Hypogonadism at age 24 and its coexistence with other diseases suggests shared pathogenic mechanisms or genetic factors. The early onset of SLE, a chronic autoimmune connective tissue disease highlights the unpredictable nature of autoimmune diseases. The identification of dyslipidemia, risk factor for cardiovascular disease further complicates the clinical picture. [6] Notably, the absence of familial autoimmune history suggests the potential involvement of sporadic genetic mutations or environmental factors in disease development. Our case involves multiple critical factors. Firstly, diagnosing APS is challenging due to its diverse and nonspecific clinical presentations. Secondly, the management of APS is a multidisciplinary and holistic approach, involving endocrinologists, rheumatologists, dermatologists, and other specialists. Lastly, Patients must receive comprehensive education about chronic nature of disease, emphasizing strict adherence to treatment regimens and regular follow-up to optimize long-term health outcomes. [7]

The diagnosis of APS was made through a

combination of clinical evaluation, medical history, laboratory tests, and genetic testing.

Conclusion

This case report highlights the complexity of autoimmune polyglandular syndromes and emphasizes the need for early recognition and a multidisciplinary approach for optimal care and management.

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