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Case Report / Olgu Sunumu

A case of autoimmune polyglandular syndrome type 1 with ectodermal dystrophy in her nail*

*Tırnağında ektodermal distrofi olan bir otozomal poliglandular sendrom tip 1 vakası**

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ABSTRACT

Autoimmune polyglandular syndrome type 1 (APS-1), also called autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy, has three major components including mucocutaneous candidiasis, hypoparatiroidism and Addison's disease. It is an autosomal recessive disorder. Mutations in the AIRE gene found on chromosome 21 are the cause of this disease. A 21-year-old female patient with Addison's disease was admitted to our outpatient clinic for routine control. The patient was diagnosed with Addison's disease at the age of six when she applied for a complaint of fatigue and hyperpigmentation. She was diagnosed with primary hypoparathyroidism after 6 months. Physical examination revealed that mucocutaneous candidiasis at her mouth and ectodermal dystrophy at her right hand second finger's nail. The patient had no hair on the pubic or axillary region. Asplenism was detected in the abdominal ultrasound examination. In conclusion, patients with APS-1 can be presented with ectodermal dystrophy and it must be paid attention to ectodermal dystrophy during diagnostic evaluation.

Keywords: APS-1, ectodermal dystrophy

ÖZ

Otoimmün poliendokrinopati-candidiazis-ektodermal distrofi olarak da adlandırılan otoimmün poliglandüler sendrom tip 1(OPS-1); mukokütanöz kandidiyazis, hipoparatiroidi ve Addison hastalığı olmak üzere üç tane major komponente sahiptir ve 21. kromozomda localize olan AIRE genindeki mutasyonlar bu hastalığın nedenidir. Yirmi bir yaşında Addison hastalığı tanısı olan kadın hasta polikliniğimize rutin kontrol için başvurdu. Hastaya 6 yaşında iken yorgunluk ve hiperpigmentasyon şikayetiyle başvurduğu hastanede Addison hastalığı tanısı konulmuş. Altı ay sonra primer hipoparatiroidi tanısı almış. Fizik muayenede hastanın ağzında mukokütanöz kandidiazis ve sağ el ikinci parmak tırnağında ektodermal distrofi tespit edildi. Hastanın pubik ve aksiller bölgede kıllanması yoktu. Abdominal ultrason incelemesinde asplenizm saptandı. Sonuçta, OPS-1' de ektodermal distrofi görülebilir. Bu nedenle ektodermal distrofili hastalarda OPS-1 açısından dikkatli olmak gerekir.

Anahtar Kelimeler: OPS-1, ektodermal distrofi

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INTRODUCTION

Autoimmune polyglandular syndrome type 1 (APS-1), also called autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED), has three major components including mucocutaneous candidiasis, hypoparatiroidism and Addison's disease. APS-1 was first described in 1946. It is an autosomal recessive disorder. APS-1 develops due to monogenic mutation in the AIRE (autoimmune regulator gene) on chromosome 21 which codes for a putative transcription factor featuring 2 zinc motifs (1). It is not related with HLA allele genes. Three major components of APS-1 usually appear in a chronological order. Candidiasis is the first clinical finding that begins before the age of 5 years. Later, hypoparathyroidism is usually added to this syndrome before the age of 10. Primary adrenal failure occurs until the age of 15 (2). These three components are present in roughly 40% of cases. However, it is not mandatory for patients to follow this order. We previously reported a female patient who was firstly presented with hypoparathyroidism (3). On the other hand, ectodermal dystrophy as a component of APS-1 may not accompany all of cases.

In this case report, we aim to present a young woman with APS-1 who had ectodermal dystrophy in her nail.

CASE PRESENTATION

A 21 years-old woman with Addison's disease approved for our outpatient clinic for routine control. She had first applied to a hospital at the age of 6 with complaints of fatigue and hyperpigmentation, and Addison's disease was diagnosed. Six months later, primary hypoparathyroidism has been found as an additional diagnosis. Since two out of three main parts of APS-1 were present, the patient was diagnosed with APS-1.

Our physical examination findings were mucacutaneous candidiasis at her mouth (Figure1), absence of axillary and pubic hair and ectodermal dystrophy in her right second finger's nail (Figure2).

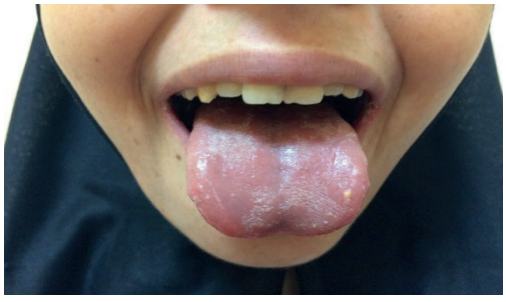

Figure 1. Mucocutaneous candidiasis at her tounge

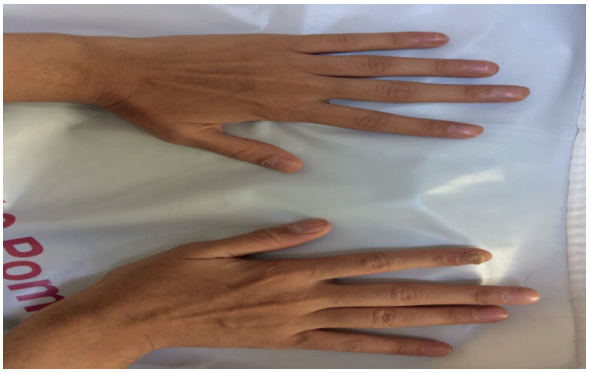

Figure 2. Ectodermal dystophy at her right second finger's nail

At the laboratory examination; PTH:7.86 pg/ml (15-65), Ca:7.01 mg/dl (8.6-10) P:4.73 mg/dl (2.5-4.5), albumin:4.43 g/dl, cortisol:1.09 µg/dl (6.2-19.4), ACTH:262 pg/ml (7.2-63), plasma aldosterone:<20 pg/ml (29.4-161), plasma renin activity: 4.18 ng/ml /h (0.5-1.90), DHEA-S:0.1 µg/dl, E_2:133 pg/ml, FSH:6.95 mIU/ml, LH:8.19 mIU/ml, progesterone:0.03 ng/ml Na:141 mmol/L, K:4.05 mmol/L, urea:43 mg/dl, creatinine:1.02 mg/dl, FT3:2.75 pg/mL, FT4:1.31 ng/ml TSH:0.831 µU/mL. There was asplenism at her abdominal ultrasonographic examination.

DISCUSSION

Ectodermal dystrophy is one of the clinical manifestations of APS-1 which is defined by certain abnormalities such as enamel hypoplasia of teeth, particular abnormalities of the nails, keratopathy, vitiligo, alopecia. They may be related to AIRE gene but not necessarily exists at every patient with APS-1 (4). AIRE modulates some transcription factors at medullar thymic epithelial cells (5). This affects both negative selection of effector T cells and positive selection of regulatory T cells (6). The presence of chronic inflammatory infiltrates composed mainly of lymphocytes in the affected organs and the presence of autoantibodies reacting to target tissue specific antigens are important factors that play a role in the pathogenesis of APS-1. Neutralizing antibodies to T helper are present in peripheral blood of the patients which causing defective antifungal response. This situation contribute to the development of mucocutaneous candidiasis. So chronic mucocutaneous candidiasis is usually the first clinical manifestation and is nearly always present at APS-1 (2).

Nail deformities are caused by chronic candidiasis. Alopecia and vitiligo are other disorders related to

autoimmunity (2). Nonendocrine manifestations as alopecia, vitiligo, intestinal malabsorbtion, pernicious anemia, chronic active hepatitis are also distinguished in progress of time. Nail dystrophy is one of the rare manifestations at APS-1(1). In our patients with APS-1, we detected ectodermal dystrophy in her hand finger's nail, but this finding has not attracted anyone until now. There are not so many cases reported within APS-1 patients who are presented by nail dystrophy. One of the cases was a 3 years-old boy who was hospitalized because of splenomegaly and general condition disorder. At his medical history and physical examination, nail changes was detected 2 years old. His hormone levels were normal. APS-1 was suspected and AIRE gene mutation is found at genetic tests (7). Another case was an eight years-old girl who was presented to the hospital with mucocutaneous candidiasis, idiopathic generalized epileptiform seizures, nail dystrophy, twitching left face and progressively increasing generalized skin hyperpigmentation and hypopigmented patches over both shins (8).

In the conclusion, patients with APS-1 can be presented with ectodermal dystrophy and it must be paid attention to ectodermal dystrophy during diagnostic evaluation.

DECLARATION OF INTEREST STATEMENT

The authors report no conflicts of interest. The authors alone are responsible for the content and writing of the paper

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