Our Dermatology Online

Case Report

# Skin manifestations during Autoimmune Polyendocrinopathy-Candidiasis-Ectodermal Dystrophy syndrome

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## ABSTRACT

Autoimmune polyendocrinopathy candidiasis ectodermal dystrophy (APECED), Dermatological manifestations are at the forefront of clinical manifestations and may appear in early childhood before any endocrinopathy. Aa 7-year-old child was treated in pediatrics for adrenal insufficiency under hydrocortisone. Our examination revealed multiple achromic and hypochromic macules on the face, trunk and the roots of the legs. Diffuse alopecic patches, well limited with irregular contours with an erythematous-squamous background On the scalp. At the nails level, there was a diffuse onychodystrophy of the 4 nails with onycholysis of the fingers, xanthonychia, beauty marks. The examination of the mucous membranes showed a bilateral angular cheilitis with thrush. The dermatological manifestations during APECED syndrome are diverse and may appear at an early stage of the disease, hence the need to be aware of them in order to make an early diagnosis and prevent serious complications that may be fatal.

**Key words:** Polyendocrinopathy; Skin; manifestations

## INTRODUCTION

Autoimmune polyendocrinopathy candidiasis ectodermal dystrophy (APECED), also known as APS-1 (autoimmune polyendocrine syndrome type 1) is a monogenic autosomal recessive syndrome caused by mutations in the autoimmune regulator gene (AIRE) [1]. According to the classic criteria, clinical diagnosis requires the presence of at least two of three main components: chronic mucocutaneous candidiasis, hypoparathyroidism and primary adrenal insufficiency [2]. Patients often suffer from other endocrine or non-endocrine autoimmune conditions throughout life. Dermatological manifestations are at the forefront of clinical manifestations and may appear in early childhood before any endocrinopathy, In most populations investigated, Clinical manifestations varied between 17 and 100% of APECED patients [3].

## CASE REPORT

A 7-year-old child of second-degree consanguineous marriage, with the history of 3 brothers who died at the age of 1 month, 5 years and 11 years for undocumented chronic diseases. He was being treated in pediatrics for adrenal insufficiency with hydrocortisone, referred by his pediatrician for hypopigmented skin lesions with alopecia patches of the scalp. Our examination revealed manifest xerosis and multiple achromic and hypochromic patches on the face, trunk, and leg roots, well limited, Highlighted under Wood's Lamp, related to vitiligo (Figs. 1a and 1b). Dermoscopy of the scalp patches showed an erythematous background, comma hairs and corkscrew hairs (Figs. 2a and 2b). On the nails, there was a diffuse onychodystrophy with onycholysis of the fingers (Fig. 3). The examination of the mucous membranes showed a bilateral angular cheilitis with thrush (Fig. 4). In view of all these dermatological

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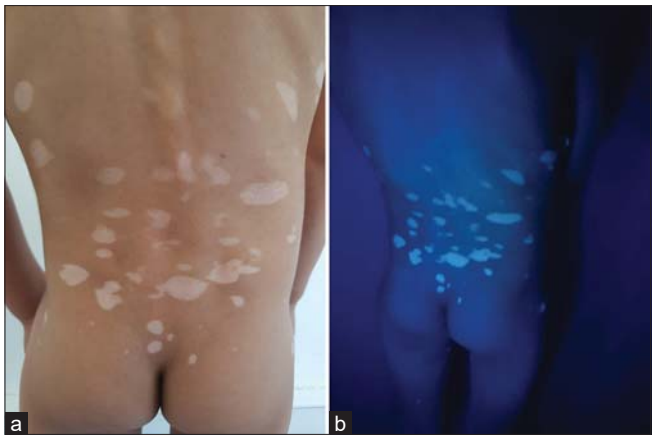


**Figure 1:** (a) Clinical picture showing achromic macules related to vitiligo (b) Wood's light contrast.

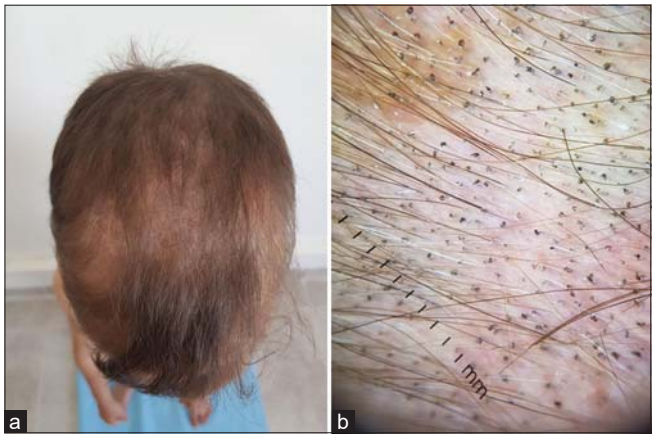


**Figure 2:** (a) Clinical picture showing alopecic plaques with a scaly erythematous background on the scalp (b) Dermoscopic image showing erythema, scales, corkscrew hairs.

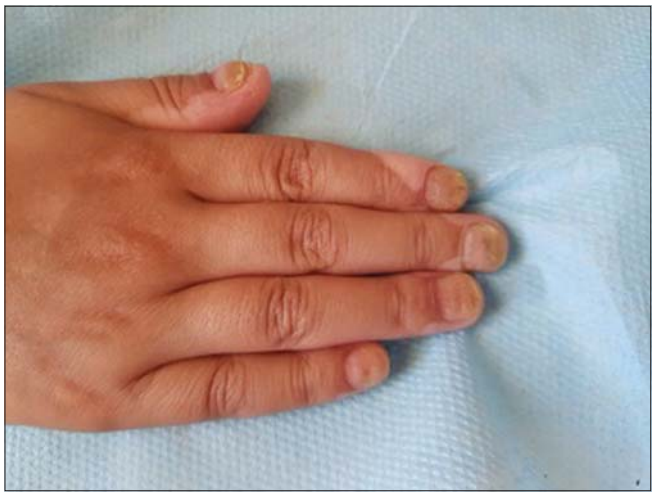

**Figure 3 :** Clinical picture showing diffuse onychodystrophy.

manifestations with adrenal insufficiency, the diagnosis of APECED syndrome was obvious. The result of mycological sampling of the nails and scalp was in favor of candidiasis. Our patient received oral fluconazole

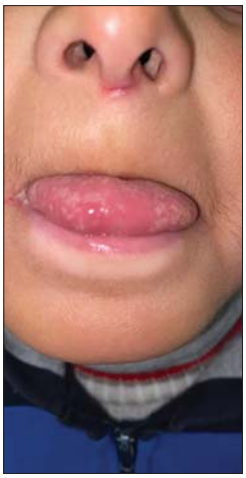

**Figure 4:** Clinical picture showing angular chelitis with oral thrush.

with improvement of oral, nail and scalp lesions. For the vitiligo lesions, the patient was treated with tacrolimus cream.

## DISCUSSION

Autoimmune polyendocrinopathy type 1 (AEP1) is a rare disease, apart from certain populations in which there is a very high prevalence, such as the Finnish population, the Sardinians. It is an autosomal recessive disease, linked to mutations in the AIRE gene [4]. The clinical diagnosis is based on the presence of at least two criteria among the characteristic triad of the syndrome: chronic mucocutaneous candidiasis, autoimmune hypoparathyroidism and autoimmune adrenal insufficiency [5]. Dermatological manifestations are at the forefront of clinical manifestations and may appear in early childhood before any endocrinopathy [6]. Mucocutaneous candidiasis is the most frequent manifestation in patients with AEP1, it is often diagnosed in early childhood, usually before the age of 5 years, it is chronic and intermittent. It particularly affects the mucous membranes, especially the mouth, esophagus and genitals, but also the nails and more rarely the skin. Oral candidiasis may present as angular cheilitis, thrush or diffuse oral candidiasis with possible esophageal involvement responsible for dysphagia and retrosternal pain [7]. Nail involvement is also common, such as candidal paronychia and/or onychomycosis with dystrophy. The nails of the hands are more frequently affected than those of the feet. This nail involvement usually requires long-term oral treatment with fluconazole or itraconazole. Nail dystrophy is also common without identification of a fungal infection [4]. The association of alopecia areata

and PEA-1 has been described since 1946, and it may be single or multiple episodes of varying severity. The pathogenesis of alopecia areata in APECED syndrome is multifactorial, favored by the autoimmune context and the severe deficiency syndrome related to intestinal malabsorption, which is frequent in these patients. Its prognosis is identical to that of alopecia areata outside the context [6]. Vitiligo occurs in 8-25% of patients. It usually begins in childhood, as well as halo nevi and poliosis. Dental abnormalities such as tooth enamel hypoplasia are often associated, and may be secondary to recurrent oral infections, malnutrition or prolonged hypocalcemia [5]. The dermatologic manifestations during APECED syndrome are diverse. Therefore, any patient presenting with recurrent cutaneous-mucosal candidiasis, isolated or associated with autoimmune pathologies, should be considered as having APECED syndrome and should have a family investigation with a clinical, biological, endocrine and immune workup [7].

## CONCLUSION

The dermatological manifestations during APECED syndrome are diverse and can appear at an early stage of the disease, which makes it necessary to be aware of them in order to make an early diagnosis and prevent serious complications that can be lethal.

### Consent

The examination of the patient was conducted according to the principles of the Declaration of Helsinki.

The authors certify that they have obtained all appropriate patient consent forms, in which the patients gave their consent for images and other clinical information to be included in the journal. The patients understand that their names and initials will not be published and due effort will be made to conceal their identity, but that anonymity cannot be guaranteed.

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