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ANHIDROTIC ECTODERMAL DYSPLASIA: A RARE CASE REPORT OF CHRIST–SIEMENS–TOURAINE SYNDROME IN AN ADULT MALE

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ABSTRACT

Anhidrotic ectodermal dysplasia (AED), also known as Christ–Siemens–Touraine syndrome, is a rare inherited disorder characterized by the triad of hypotrichosis, hypodontia, and anhidrosis. It results from abnormal development of ectodermal structures during embryogenesis. We report a case of a 28-year-old male with typical features of AED, including conical teeth, partial anodontia, sparse hair, dry skin, and absence of sweating. The diagnosis was clinical, supported by dermatologic and dental findings, and there were no similar cases in the family. This case highlights the importance of early recognition and multidisciplinary management of AED, a syndrome with significant aesthetic and functional consequences.

KEYWORDS

Anhidrotic ectodermal dysplasia, Christ–Siemens–Touraine syndrome, Rare genetic disorder

MAIN ARTICLE

Introduction

Anhidrotic ectodermal dysplasia (AED) is a rare genetic disorder affecting the development of ectoderm-derived tissues, including skin, hair, teeth, nails, and sweat glands. It is most frequently inherited in an X-linked recessive pattern, affecting primarily males, with female carriers showing variable phenotypic expression due to lyonization [1]. AED is characterized clinically by a classic triad of hypodontia or anodontia, hypotrichosis, and anhidrosis or hypohidrosis, which can lead to overheating episodes, recurrent infections, and major dental abnormalities [2]. The oral and facial features of the disease, such as conical teeth, midface hypoplasia, and periorbital hyperpigmentation, are highly distinctive and can help clinicians recognize the syndrome early [3]. Multidisciplinary care involving dermatologists, dentists, geneticists, and ophthalmologists is crucial to mitigate complications and improve quality of life in affected patients [4].

Case report

A 28-year-old male, with a history of asthma since childhood, presented with chronic anhidrosis and was referred to dermatology for evaluation of suspected ectodermal dysplasia. Physical examination revealed a dysmorphic facies with thick lips, enophthalmic appearance due to periorbital bone prominence, and marked periorbital hyperpigmentation (Figure1).

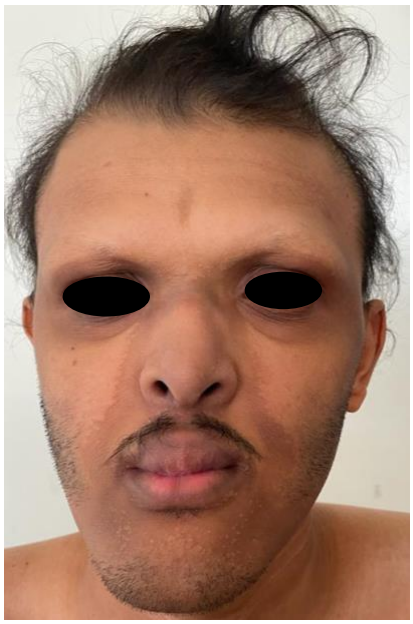

***Figure 1**: periorbital hyperpigmentation*

His scalp hair was fine, sparse, and lightly pigmented, while eyebrows and eyelashes were nearly absent. Axillary and pubic hair were also missing, although beard and mustache hair were preserved. The skin was dry, hypopigmented, and glabrous on the trunk and limbs. Multiple small white papules were noted on the chin, nasal wings, and glabellar region, corresponding to hyperplastic sebaceous glands. Oral examination showed hypodontia with retention of only five conical incisors and several dental caries (Figure 2).

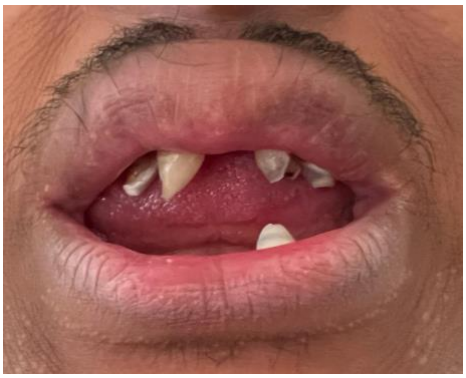

***Figure 2**: conical teeth*

No nail abnormalities were observed. Psychomotor and staturo-ponderal development were normal, and no family history of similar symptoms was reported.

A clinical diagnosis of Christ–Siemens–Touraine syndrome was made based on the presence of the full triad: hypodontia, hypotrichosis, and anhidrosis. Cutaneous biopsy and further genetic confirmation were recommended. No ocular, ENT, or neurological complications were present at the time of evaluation, but preventive workup was advised.

Discussion

Anhidrotic ectodermal dysplasia is a rare congenital condition caused by mutations in genes involved in ectodermal signaling pathways, particularly EDA, EDAR, and EDARADD, all of which influence the development of ectodermal derivatives [1]. The X-linked recessive form, caused by mutations in the EDA gene, is the most common, affecting males almost exclusively, while female carriers typically exhibit minor signs or remain asymptomatic [5]. Clinical diagnosis is based on the classic triad, though additional signs such as dry, hyperkeratotic skin, sebaceous gland hyperplasia, and periorbital darkening may provide supportive evidence [2].

Dental anomalies are often severe and include hypodontia or anodontia affecting both dentitions, conical teeth, and defective enamel, predisposing to early caries [3]. In this case,

the patient retained only five incisors of conical shape. Salivary hypofunction is common and may further compromise oral hygiene and mastication [3]. Hair abnormalities typically include fine, sparse, light-colored hair with early-onset alopecia. Eyelashes and eyebrows are usually sparse or absent, as in our patient. Although sweat glands may be hypoplastic or absent, partial anhidrosis is more common than complete anhidrosis, and this can lead to episodes of hyperthermia [2].

Management is primarily supportive and requires coordination between dermatologists, maxillofacial specialists, dentists, and sometimes ophthalmologists or pulmonologists depending on associated complications. Dental rehabilitation at an early age, including prosthetic or orthodontic care, is essential for functional and psychosocial improvement [4]. Genetic counseling should also be offered to the patient and family due to the hereditary nature of the disorder [5].

Conclusion

Anhidrotic ectodermal dysplasia is a rare genetic disorder with distinctive clinical features that enable diagnosis through careful physical examination. Recognition of the classical triad of hypotrichosis, hypodontia, and anhidrosis is critical for early diagnosis and comprehensive management. Our case illustrates the full phenotypic expression of the disease in a young adult male, reinforcing the importance of multidisciplinary care to address both functional and aesthetic consequences of the syndrome.

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