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A RARE CASE OF HYPOHIDROTIC ECTODERMAL DYSPLASIA

Genetics

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ABSTRACT

Ectodermal dysplasias constitute a rare group of genetic disorders primarily affecting tissues of ectodermal origin, including teeth, sweat glands, hair, and nails. These congenital conditions often present challenges in diagnosis and management. Hypohydrotic ectodermal dysplasia, also known as Christ–Siemens–Touraine syndrome, is the most common variant and follows an X-linked recessive inheritance pattern. This syndrome is characterized by a classic triad of reduced sweating, defective dentition, and hypotrichosis. We present a rare case of hypohydrotic ectodermal dysplasia involving an 18-year-old female patient. The patient exhibited the hallmark features of the disorder, including reduced sweating, defective dentition, and sparse hair. The diagnosis was established based on these distinctive clinical findings. A multidisciplinary management plan has been developed to address the patient's unique needs and improve her quality of life. This case underscores the importance of early recognition and comprehensive management of hypohydrotic ectodermal dysplasia to optimize the long-term well-being of affected individuals. Further research is warranted to explore therapeutic interventions and genetic counselling for families at risk of this X-linked recessive disorder.

KEYWORDS

INTRODUCTION

Ectodermal dysplasias are rare group of genetic disorders which primarily manifest through defects in the ectodermal structures like skin, nails, teeth and sweat glands. Other ectodermal derivatives like eyes, ears, adrenal and neural structure may also be involved(1,2). There are more than 150 variants described in the literature. The most common variant being hypohydrotic form. They are primarily classified based on the extent of involvement of sweat glands. Hypohydrotic form and hydrotic forms. Hypohydrotic form is also called as Christ□□Siemens□□Touraine syndrome. It is by a characterized by the triad of reduced sweating, defective dentition and hypotrichosis. Typical facial features include frontal bossing, sunken cheeks, saddle nose and everted thickened lips, sparse eyebrows.(3). Dental manifestations of the disorder include conical teeth, hypodontia, delayed eruption of permanent teeth. Sweat glands may be absent or rudimentary. In few cases mucous glands maybe absent in oesophagus, duodenum and in respiratory tract. Other reported signs being short stature, photophobia and ocular abnormalities.(4) The hydrotic variant of ectodermal dysplasia is called as Cloustone syndrome in which sweat glands are normal and is inherited in an autosomal dominant pattern.

Case Report

An 18-year-old girl presented with a raised body temperature and heat intolerance. She had abnormal facial features, including absent eyebrows, frontal bossing, and thick, everted lips (Figure 1). Her scalp hair was thin and brittle, and she reported a history of hair loss and premature greying. An examination of her oral cavity revealed cone-shaped teeth and hypodontia (Figure 1 and 2). She also complained of difficulty in chewing food. She had experienced heat intolerance since childhood and rarely sweated. Dystrophic changes were noted over her fingernails. Her intellectual status was normal, and her thyroid function tests, liver function tests, and renal function tests were normal. Based on her characteristic clinical features, she was diagnosed with hypohydrotic ectodermal dysplasia and was referred for dental consultation, where she was advised on prosthesis for her teeth. Environmental modifications, such as cold water baths, were suggested to alleviate heat intolerance.

DISCUSSION

Ectodermal dysplasias are hereditary genetic disorders primarily affecting two or more ectodermal derivatives(5). Ectodermal dysplasias are categorized into two categories based on the presence or absence of sweat glands: hydrotic and hypohydrotic, respectively. The hypohydrotic form, also called Christ-Touraine syndrome, is inherited in an X-linked recessive manner and is characterized by the classic triad of hypodontia, hypohydrosis, and hypotrichosis. The hydrotic form of ectodermal dysplasia is characterized by abnormalities in teeth, hair, and sweat glands. Due to absent sweat glands, individuals with this form present with unexplained pyrexia and heat intolerance. Fine, sparse hair is seen over the scalp, which lacks luster. Other common facial features include frontal bossing, sunken cheeks, a depressed nasal bridge, thick everted lips, low-set ears, wrinkled, hyperpigmented periorbital skin, and normal intelligence(6,7,8). The most common oral manifestation is hypodontia and conical-shaped teeth, affecting both deciduous and permanent teeth(9).

The patient reported hyperthermia and heat intolerance secondary to absent sweating, along with hypodontia featuring conical teeth. Additionally, she had absent eyebrows, loss of scalp hair, frontal bossing, thick and everted lips, and dystrophic nails. Based on these characteristic clinical findings, the diagnosis of hypohydrotic ectodermal dysplasia was made.

Management involves addressing the various manifestations, including the use of wigs or special hair care formulas for managing sparse and dry hair, environmental modifications to combat hyperthermia (e.g., ensuring accessibility to an adequate water supply and using air conditioning to maintain a cool temperature), and the use of skin care products to manage dry skin. Dental implants and prostheses are required for hypodontia, and lubricating eye drops are recommended. Management of recurrent respiratory infections and asthma is often necessary(10).

CONCLUSION

In summary, this case report highlights the presentation and diagnosis of hypohydrotic ectodermal dysplasia in an 18-year-old female. Her clinical features, including absent eyebrows, frontal bossing, thick and everted lips, thin and brittle scalp hair, cone-shaped teeth, hypodontia, heat intolerance, and dystrophic fingernails, align with the classic characteristics of this rare genetic disorder. Timely diagnosis and a multidisciplinary approach to management, including dental consultations and environmental modifications, are essential for enhancing the patient's quality of life. Further research and awareness are necessary to improve the understanding and care of individuals affected by ectodermal dysplasias and their associated challenges.

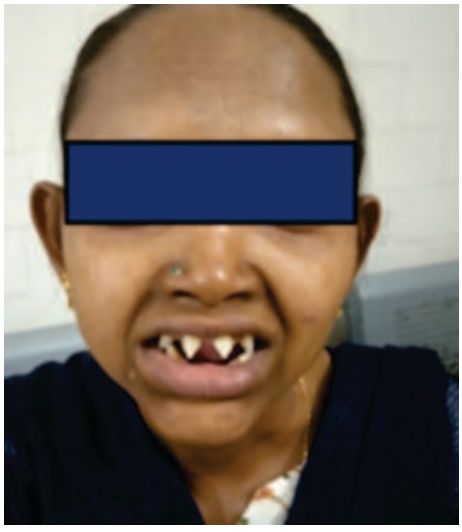

Figure 1: Clinical Photograph Showing Conical Teeth, Frontal Bossing, Sparse Scalp Hair, Thick And Everted Lips.

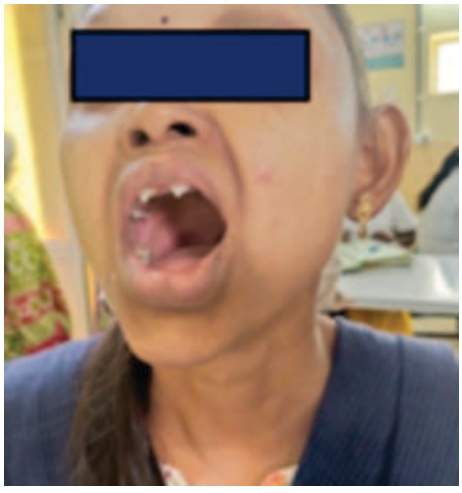

Figure 2: Clinical Photograph Showing Hypodontia With Conical Teeth

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