CASE REPORT

APPEARANCE SAYS IT ALL; A RARE CASE OF HYPOHIDROTIC ECTODERMAL DYSPLASIA

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Ectodermal Dysplasia (ED) is a rare genetic condition characterized by the involvement of ectoderm derivatives such as hair, nail, sweat glands, and teeth. It has many variants, but the two most common ones are hypohidrotic/anhidrotic ectodermal dysplasia and hidrotic ectodermal dysplasia. Herein, we present a case of a 20-year-old female with hypohidrotic ectodermal dysplasia who had anodontia, hypohidrosis, and hypotrichosis, and her condition went unrecognized until she was seen for gastroenteritis at a tertiary care center. This case report will help spread education and awareness regarding such a rare and under-recognized condition. Early diagnosis and intervention help improve the quality of life.

Keywords: Hypotrichosis; Anodontia; Hypohidrosis; Ectodermal dysplasia

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INTRODUCTION

Ectodermal Dysplasia (ED) is a rare hereditary disorder that involves the defect of two or more tissues of the embryonic ectoderm. The frequency of ectodermal dysplasia is approximately 1/100,000 live births.[1] It was first described by Thurnam in 1848.[2] There are many known variants or types of ED but hypohidrotic/anhidrotic ED (Christ-Siemens-Touraine Syndrome) and hidrotic ED (Clouston Syndrome) is the most common variants, which are differentiated from each other based on the absence or presence of sweat glands.[3] In Pakistan, very few cases of ectodermal dysplasia have been reported so far. It is considered one of the underdiagnosed conditions due to limited knowledge and unawareness among parents, leading to delayed diagnosis and appropriate treatment. Therefore, by reporting this case of a 20-year-old female with no family history of ED, we hope to bring this topic to realization.

CASE

A 20-year-old female presented to the emergency department with nausea, vomiting, and diarrhoea for 3-day. Vital signs were within the normal range, but the blood pressure was on the lower side of normal (98/74 mmHg). She was given an intravenous (IV) fluid bolus right after sending a blood sample for the initial workup, and also received antibiotics and anti-pyretic. Her condition improved rapidly.

On examination, she had a depressed nasal bridge, hypotrichosis, everted lips, short height, and did not have teeth. Figure-1, 2, 3, and 4. Further information was sought given her clinical features and she reported decreased sweating, increased body temperature, and speech problems such as slurred speech and could not pronounce words properly. Her intellectual ability appeared normal and was studying in 12th grade. Interestingly, no family history of ED was reported. Her condition was only diagnosed incidentally and remained untreated until she visited a tertiary care hospital for gastroenteritis. Therefore, the patient and her parents were counselled and educated regarding the condition.

All her baseline investigations were normal. A genetic test was planned but not performed due to unavailability and non-affordability. Whenever possible, she will undergo genetic testing as an outpatient for confirmatory diagnosis as well as to establish the subtype at an outside send-out laboratory given the scarcity of such facilities in our healthcare setup. A clear-cut diagnosis was made given the prominent clinical features of hypohidrotic ectodermal dysplasia. A multi-disciplinary approach was planned to address ectodermal dysplasia, and outpatient follow-ups with a primary care physician, psychologist, odontologist, and speech therapist were arranged.

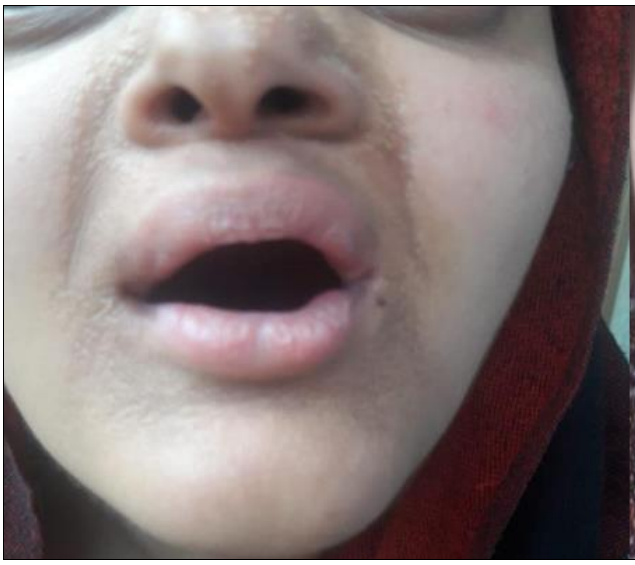

Figure-1: Depressed nasal bridge and hyperpigmentation around eyes

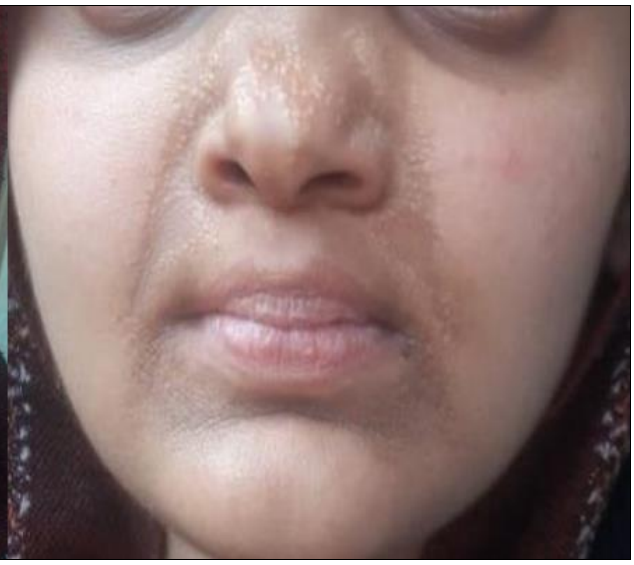

Figure-2: Anodontia and everted lips

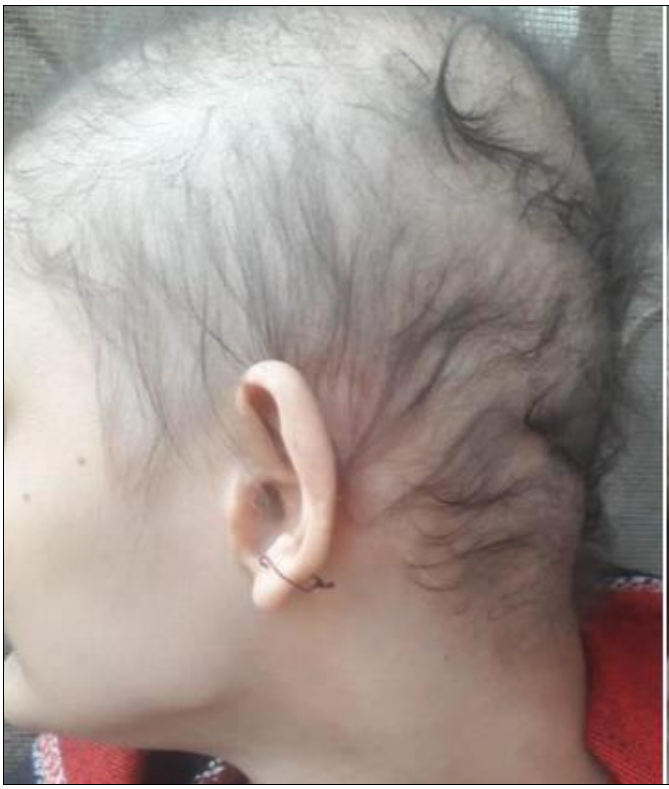

Figure-3: Hypotrichosis

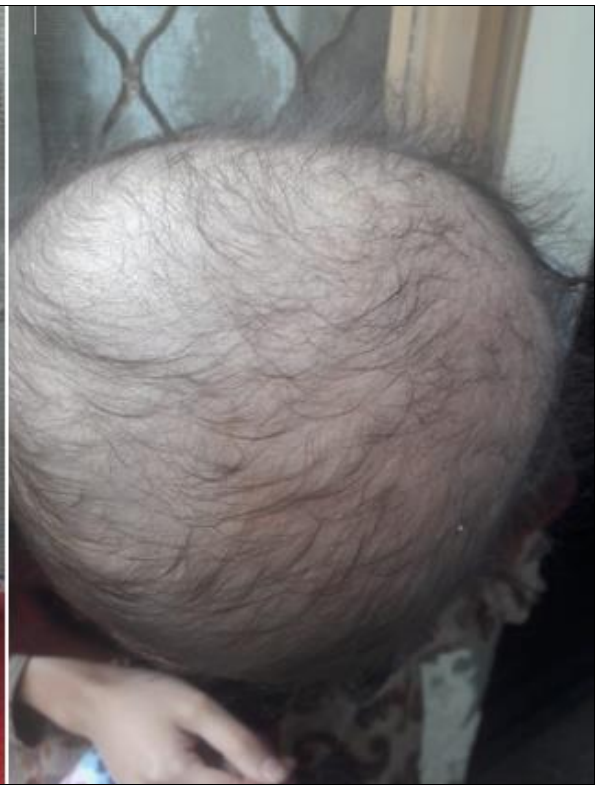

Figure-4: Hypotrichosis

DISCUSSION

More than 200 subtypes of ED have been formulated based on genetic mutations, clinical features, and molecular pathway involvement.[4] However, Hypohidrotic ectodermal dysplasia is a common variant among this large group of hereditary disorders with a prevalence of 1/20,000 newborns worldwide. The characteristic features of hypohidrotic ectodermal dysplasia include hypohidrosis (decreased sweating) or anhidrosis (absent sweating), hypodontia (decreased teeth), or anodontia (no teeth), and hypotrichosis (decreased hair). The facial features are also very specific such as depressed nasal bridge (saddle nose), frontal bossing, everted and

thick lips, prominent supraorbital ridges, hyperpigmentation around the eyes, and sunken cheeks.[5] In some cases, photophobia, xeropthalmia, xerostomia, and short stature have been noted as well.[6]

The most common inheritance pattern of hypohidrotic ectodermal dysplasia is X-linked recessive followed by autosomal recessive and autosomal dominant. However, in rare cases, hypohidrotic ectodermal dysplasia might occur because of a de novo mutation which is thought to be the reason in cases with no family history of ED.[7] Our patient had features of hypohidrotic ectodermal dysplasia with no family history of ED including siblings (two female and three male) or any immediate family members. X-linked recessive HED is because of mutation in ectodysplasin A (EDA). While autosomal recessive and autosomal dominant HED is due to a mutation in ectodysplasin A receptor (EDAR) and EDAR-associated death domain (EDARADD) genes respectively.[8] Each of these genes activates signalling pathways such as tumour necrosis factor α-pathway, WNT-signalling pathway, and nuclear factor KB-pathway, which involves the interaction of ectoderm-mesoderm, and helps in the differentiation and organogenesis of the developing embryo.[9]

The diagnosis of hypohidrotic ectodermal dysplasia is usually made based on clinical features. However, genetic testing for the genes related to this condition such as EDA, EDAR, and EDARADD can be done for confirmation. Our diagnosis of hypohidrotic ectodermal dysplasia was solely based on the clinical features because of the unavailability and unaffordability of genetic testing. Most of the cases of hypohidrotic ectodermal dysplasia are diagnosed during infancy or early childhood because obvious clinical features are not expressed in new-borns.[9]

The management of hypohidrotic ectodermal dysplasia requires coordinated efforts by a team of specialists which includes a paediatrician, internist, dental specialist, dermatologist, speech therapist, otolaryngologist, and psychologist. It is recommended to start dental treatment and oral rehabilitation as early as possible to prevent alveolar ridge atrophy and to improve both sagittal and vertical skeletal relationships during craniofacial growth and development.[10] Otolaryngologist has to deal with multiple problems in hypohidrotic ectodermal dysplasia such as hearing pathologies, saddle nose, nasal obstruction, and rhinosinusitis.[11] Moreover, cosmetics (wig placement), prosthodontics (dentures, implants), and psychological counselling, the early institution help gain confidence and overcome social challenges.[7]

Genetic counselling is also very important. The mode of inheritance is identified through family history alone or with molecular genetic testing. Once a pathogenic variant is recognized in a family, then prenatal testing for a high-risk pregnancy and preimplantation genetic testing can be done for hypohidrotic ectodermal dysplasia.[12]

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