Case Report

Ectodermal dysplasia: Case report

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Abstract

Ectodermal dysplasia represents a group of inherited conditions characterized by aplasia or dysplasia of two or more ectodermally derived anatomical structures such as hair, nails, teeth and sweat glands .Two main clinical forms have been described-hypohidrotic and hidrotic type depending upon functioning of sweat gland. Main clinical manifestations include heat intolerance, anodontia or hypodontia, atrichosis or hypotrichosis and dystrophic nails. This is medical illness with psychosocial impact on child and parents. We present two cases one of hypohidrotic type and other of hidrotic type of ectodermal dysplasia. Importance of early prosthetic management has been discussed.
Keywords: Ectodermal dysplasia, Hypohidrotic, Prosthetic rehabilitation.

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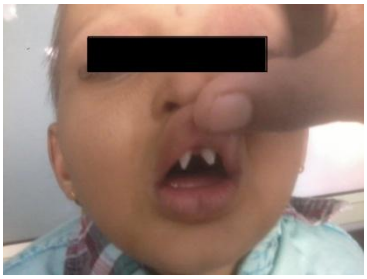
INTRODUCTION

Ectodermal dysplasia is a rare heterogeneous group of disorder characterized by a constellation of findings involving defect in two or more tissues developed from ectoderm. These tissues are skin, hair, nail, teeth and eccrine glands.[1] More than 150 distinctive syndromes have been described with all possible modes of inheritance. Depending upon number and functionality of sweat glands they are broadly classified as hypohidrotic (anhidrotic) and hidrotic ectodermal dysplasia.[2] Hypohidrotic ectodermal dysplasia is most common form of ectodermal dysplasia, most commonly inherited as X-linked recessive pattern. Because of such inheritance pattern males have full expression of clinical manifestations while females are asymptmatic carriers or mildly affected.[3] Hypohidrotic ectodermal dysplasia is characterized by triad of signs comprising sparse hair, (atrichosis or hypotrichosis) abnormal or missing teeth, (anodontia or hypodontia)and inability to sweat due to lack of sweat glands. Frequently they present as episodes of high fever in warm environments which may be mistakenly considered as a fever of unknown origin.[1] The typical facies is characterized by frontal bossing, malar hypoplasia, flattened nasal bridge, wrinkled periorbital skin.[4] Children with decreased sweating may have mortality rate of up to 30% in infancy or early childhood because of intermittent hyperpyrexia.[5] Dental manifestations include peg shaped teeth, hypodontia or anodontia. The scalp hair, eyelashes and eyebrows are sparse, fine and lightly pigmented. Hidrotic ectodermal dysplasia is mostly inherited as autosomal dominant disorder .Here sweat glands are normal but nails are dystrophic, hair are sparse and there is hyperkeratosis of palms and soles.[6] Treatment includes protection from high ambient temperature. Early dental evaluation is necessary so that prosthesis can be provided early for cosmetic reason and adequate nutrition[1] Here we present one case each of hypohidrotic and hidrotic ectodermal dysplasia.

CASE REPORT 1

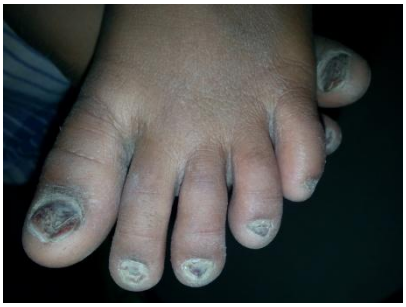
Seven year old boy, 1st in order, born of nonconsanguinous marriage presented with recurrent episodes of hyperpyrexia particularly in summer months and inability to tolerate heat. Parents also complained about poor scalp hair growth. Also there were problems while eating as only 2teeth were erupted. There was

history of peeling of skin during early infancy. On history, his maternal uncle also had similar problems. On examination, his vitals were stable .His face had typical old man look. His skin was dry, wrinkled with hyperkeratosis. The scalp hair, eyebrows were sparse, fine and lightly pigmented. He had only two peg shaped teeth. However his mental development was normal and was good in studies. He was clinically diagnosed as hypohidrotic ectodermal dysplasia.



CASE REPORT 2

Six year old female child, second in order born of consanguineous marriage brought with sparse scalp hair with its poor growth and abnormal nails. There was no family history of similar complaints. On examination, her vitals were stable. She had sparse scalp hair, eyebrows. Her nails were dark colored and dysplastic. There was no history of heat intolerance .she used to get sweating on exertion or in summer months. Her mental and other developmental milestones were normal. Her school performance was average. She was clinically diagnosed as hidrotic ectodermal dysplasia.



DISCUSSION

Ectodermal dysplasias are a group of inherited disorders that share common developmental defect involving at least two of the major structures classically hold to derive from embryogenic ectoderm-hair, teeth, nail and sweat glands. Depending upon number and functionality of sweat glands they are classified as anhidrotic (sweat glands absent) orhypohidrotic (sweat glands reduced) and hidrotic type. (sweat gland normal) Hypohidrotic ectodermal dysplasia is most frequently reported manifestation of ectodermal dysplasia, inherited as X-linked recessive patter affecting males. In developing countries like ours diagnosis is mainly clinical. In developed countries diagnosis pertains to laboratory identification of genes and mode of inheritance of mutations. It is to be considered that an absence of positive family history should not be factor in causing diagnostic dilemma as this condition shows multiple modes of inheritance.[9] In our first case of hypohidrotic ectodermal dysplasia maternal uncle was suffering from similar complaints suggesting X-linked recessive inheritance. In 2nd case of hidrotic ectodermal dysplasia there was no family history of similar complaints. In our first case that boy used to get recurrent febrile episodes at times needed hospitalization. Both children were getting difficulty in carrying on regular schools as they used to get bad comments from peers about their appearance. Clinical manifestatiosns of ectodermal dysplasia cause considerable social problems to individuals affected.[10] First case was advised light clothing, a cool water spray bottle, avoiding going in high ambient temperatures, application petrolatum jelly to nasal mucosa and was referred to dental specialties for dental rehabilitation. Treatment requires multidisciplinary team approach involving Pediatrician, Pediatric dentist, orthodontics and prosthodontics. Early prosthetic treatment in such children is important. This results in significant improvement in esthetics, masticatory and phonetic functions. Inaddition, positive psychological on child and his or her parents should be taken in to account.[10] Hypohidrotic ectodermal dysplasia associated with immunodeficiency is newly recognized primary immunodeficiency which presents with recurrent bacterial infections and high mortality.[11] Such history was not noted in present case. In present both cases intelligence and other development was normal. With proper care and prosthodontics treatment and regular follow up with multidisciplinary team affected children can enjoy relatively normal life.

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