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CASE REPORT

Ectodermal dysplasia: a case study of a toddler reported to a tertiary care hospital

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Abstract:
We reported a rare case of Ectodermal dysplasia in a young child born to a consanguineous married couple presented to a tertiary health care centre characterized by triad of defects of partial or complete absence of sweat glands, anomalous dentition and hypotrichosis. There are four types, of which X-linked recessive type is the most common. The basic defect is in non development or under development of certain ectodermal structures namely the skin and its appendages and the teeth. Diagnosis is usually clinical, but can be confirmed by skin biopsy which shows deficiency of sweat glands and other skin appendages. These patients have a normal life expectancy if they avoid exposure to hot environments.

Keywords: *Ectodermal dysplasia, Anhidrosis, Anodontia, Hypotrichosis, Toddler*
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Introduction

Ectodermal dysplasias are a rare heterogeneous group of disorders characterized by abnormal development of ectodermal structures. The etiology of ectodermal dysplasia appears to be genetic in nature, usually an X linked Recessive disorder affecting predominantly male and female are carriers. It is characterized by the triad of signs which comprises of sparse hair (atrichosis or hypotrichosis), abnormal or missing teeth (anodontia or hypodontia) and inability to sweat due to lack of sweat glands (anhidrosis or hypohidrosis).[1,2]

Case report

A 1year and 6 month old male child born to a second degree consanguineous married couple, presented with cough, cold, fever and breathlessness for 2 days. On examination, the patient had temperature of 104^0F, frontal bossing, malar hypoplasia, depressed nasal bridge, sparse scalp hair, no eye brows, thick everted lips, large ears, dry skin and complete absence of teeth. On auscultation he had scattered crepitations all over the lung fields, his chest radiograph was suggestive of bronchopneumonia. On direct questioning the parents, they revealed that he had multiple episodes of high grade fever particularly during summer and also did not sweat at all. His elder sibling (girl child) was normal. His mother had some features like patchy alopecia and pegged teeth (Figure I), along with similar features in her brother (maternal uncle) indicative of Autosomal recessive type of inheritance. X-ray of skull lateral view shows anodontia (Figure II). Skin biopsy was done; it was consistent with our diagnosis of anhydrotic ectodermal dysplasia.

Figure I: Mother with her child showing alopecia

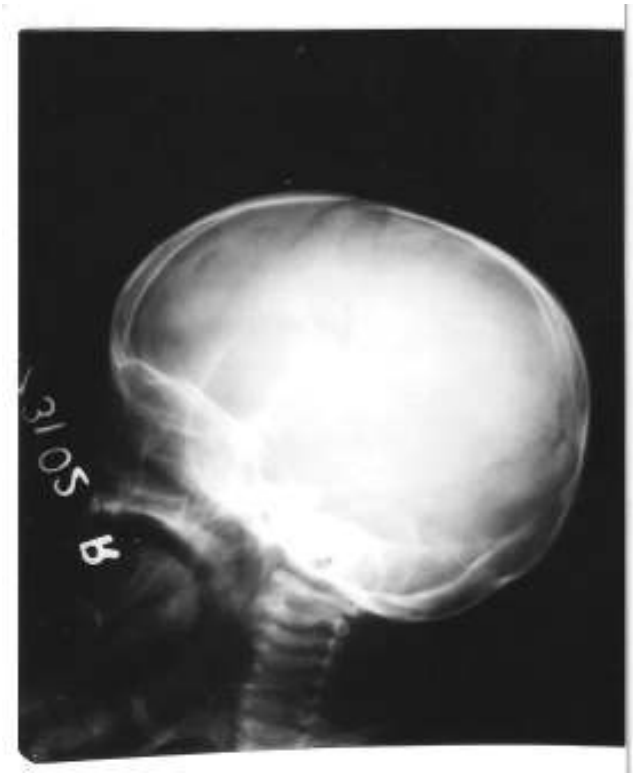

Figure II: Radiograph of skull lateral view showing Anodontia

Discussion

There are 4 recognised types of this disease; ED-1 (XLR), ED-anhidrotic (AR) EDAR, ED-3(AD), and ED-anhidrotic with immune deficiency. The X-linked Heriditary anhidrotic ectodermal dysplasia (also known as Christ - Siemens Touraine syndrome) is a rare disease characterized by non development or underdevelopment of certain ectodermal structures namely the skin and its appendages and the teeth. The incidence at birth is 1 in 100,000 males. The complete syndrome does not occur in females but female may show dental defects, sparse hair, reduced sweating and dermatoglyphic abnormalities.

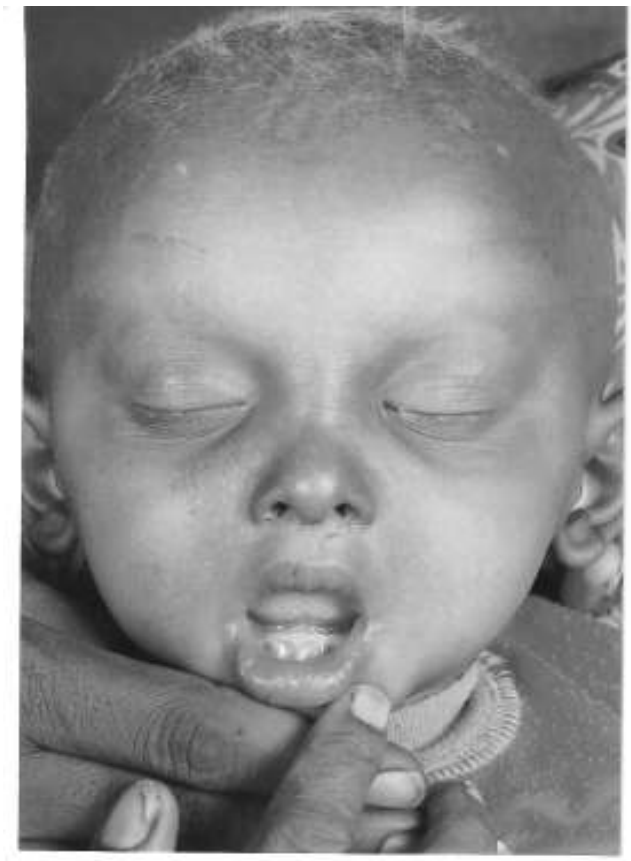

Figure III: Showing characteristic facial features.

Clinically there is anhidrosis, anodontia or hypodontia and hypotrichosis [3]. They have a characteristic facies with protruding thick lips, large deformed ears, broad depressed nose and frontal bossing (Figure III) resembling syphilitic facies [4]. Scalp hair is sparse, fine and blond. Alopecia is often the first feature to attract but it is seldom total. Absent or reduced sweating causes heat intolerance and affected patients can present with unexplained fever. Mental retardation is reported in 30-50% cases and is believed to be due to prolonged fever and febrile seizures [6]. The nails are usually normal or may be brittle.

Sexual development is usually normal. The otolaryngological manifestations include thick nasal secretions and impaction sinusitis. The absence of mucus glands in the respiratory tract predisposes these children to recurrent respiratory tract infections. The findings of equally affected male and female in single sibships, as well as the presence of consanguinity, supports the AR mode of inheritance which is otherwise almost identical to X-linked recessive ED.

Diagnosis is usually clinical, but in case of ambiguity, it can be confirmed by skin biopsy which will show deficiency of sweat glands and other skin appendages. Survival of these patients depends on avoidance of precipitating factors like exposure to hot climate. Life expectancy is usually not affected provided sufficient precautions are taken[5,6].

Conclusion

In this toddler, ectodermal dysplasia was diagnosed clinically by classical triad of hypotrichosis, anodontia and hypohidrosis and the diagnosis was confirmed by skin biopsy. This case study signifies the importance of evaluating hyperpyrexia when associated with physical abnormalities. Survival of these patients is largely dependent on avoidance of exposure to hot environment, provided such precaution is taken; their life expectancy is good to survive middle and old age.

Acknowledgment: Written consent was taken from the mother for publishing the case and including her child's photographs.

REFERENCES

1. Jayantilal PR, Rajesh S, Naveen YG, Nandini P. Treatment Considerations for a Patient with Ectodermal Dysplasia: A Case Report. JIOH 2010; 2(4): 73-78.
2. Babu SG, Castelino RL, Shetty SR, Rao KA. Hereditary Ectodermal Dysplasia - Case Report. WebmedCentral DENTISTRY 2011; 2(3): WMC001711.
3. Joseph G. Ectoderma dysplasia. Morelli-Nelson textbook of paediatrics; Kleigman, Behrman, Jenson, Stanton Saunders International edition, Ed 18th 2008; p2666.
4. Anoop TM, Simi S, Mini PN, Ramachandran M, Jabbar PK, Rajkumari PK, Sujathan P. Hypohydrotic Ectodermal dysplasia. JAPI 2008 April; 56: 268-70.
5. Shigli A, Reddy RPV, Hugar S, Deshpande D. Hypohydrotic ectodermal dysplasia: A unique approach to esthetic and prosthetic management: A case report. J Indian Soc Pedo Prev Dent 2005; 31-34.
6. Munoz F. Lestrignant G, Sybert V, Frydman M, Alswaini A. Definitive evidence for an autosomal recessive form of hypohidrotic ectodermal dysplasia clinically indistinguishable from the more common X-linked disorder. Am J Hum Genet. 1997; 6: 94-100.