Case report

Anhydrotic Ectodermal Dysplasia With Pregnancy- A Case Study

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Abstract

Hereditary Ectodermal Dysplasia is an inherited disorder involving skin, teeth, hair and nails. Hypohidrotic ectodermal dysplasia or Anhydrotic ectodermal dysplasia is the most common syndrome among this large group of heriditory disorders. Hypohydrosis/anhydrosis, hypotrichosis and hypodontia constitute the main symptoms of the syndrome. The case of a 28 year old lady with Anhidrotic ectodermal dysplasia and pregnancy with positive family history is described.

Introduction

Ectodermal dysplasia (ED) is not a single disorder, it is a large and complex group of disorders defined by the abnormal development of two or more structures derived from the embryonic ectoderm layer. These are congenital, diffuse and non progressive disorders. More than 192 distinct disorders have been described till date. Most common of them are X-linked recessive anhydrotic (Christ-Siemens-Touraine syndrome) and hydrotic ectodermal dysplasias (Clouston syndrome) . It is also rare and non progressive and presents a triad of partial or total absence of sweat glands, hypotrichosis, and hypodontia . In addition, there are other signs and symptoms that can be found depending on the involvement of the ectodermal tissue . The ectoderm, one of three germ layers present in the developing embryo, gives rise to the central nervous system, peripheral nervous system, sweat glands, hair, nails, and tooth enamel . As a result, patients of ED exhibit the following clinical sign: hypotrichosis, hypohidrosis, and cranial abnormalities. The patients often exhibit a smaller than normal face because of frontal bossing, a depressed nasal bridge, the absence of sweat glands results in very smooth, dry skin and/or hyperkeratosis of hands and feet. Oral traits may express themselves as anodontia, hypodontia, and conical teeth. Anodontia also manifests itself by lack of alveolar ridge development . The earliest recorded cases of ED were described in 1792. Since then, nearly 200 different pathologic clinical conditions have been recognized and defined as ED. These disorders are considered relatively rare, 1 in 10,000 to 1 in 100,000 births[1].

Current classification of ectodermal dysplasias is based on clinical features. Freire-Maia and Pinheiro proposed the first classification system of the ectodermal dysplasias in 1982 , with additional updates in 1994 and 2001. Their original classification system stratified the ectodermal dysplasias into different subgroups according to the presence or absence of (1) hair anomalies or trichodysplasias, (2) dental abnormalities and (3) nail.

From the clinical point of view two main forms have been distinguished-

Hypohidrotic/anhydrotic form /Christ-Seimens-Tourian Syndrome.

2. Hydrotic form /Clouston syndrome[2].

ED is characterized by the 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 (anhidrosis or hypohidrosis). The lack of teeth and the special appearance were reported to be major concerns. Most patients with ED have a normal life expectancy and normal intelligence. However, the lack of sweat glands may lead to hyperthermia, followed by brain damage and death in early infancy, if unrecognized.Thus an early diagnosis is important.

The Hypohidrotic/anhydrotic form exhibits the classic triad- hypohidrosis/anhydrosis, hypotrichosis and hypodontia. Usually X-linked recessive inheritance is seen. Males are affected severely, while females show only minor defects[3].

Case Study

A Bangladeshi woman who was 28 years old, primigravida, was admitted in ISMCH with 39

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weeks of pregnancy and slight abdominal pain. Her pregnancy was uneventful. She was normotensive and non diabetic. She had highest degree from University and she is a service holder.

She is a known case of Anhydrotic ectodermal dysplasia and has characteristic appearance having under developed nasal bridge, absent eye brows, poorly developed breast with rudimentary nipple and areola, less number of teeth, fragmented nails. Her hair is fine in texture and short, hair line drawn backward.

She gave history of admission in hospital in her childhood due to feeling of excessive heat without any sweating. She was diagnosed as anhydrotic ectodermal dysplasia by skin biopsy. Her father was suffering from mild form of the disease.Her menstrual history was normal, she was married for 1and half years and conceived spontaneously without any drugs.

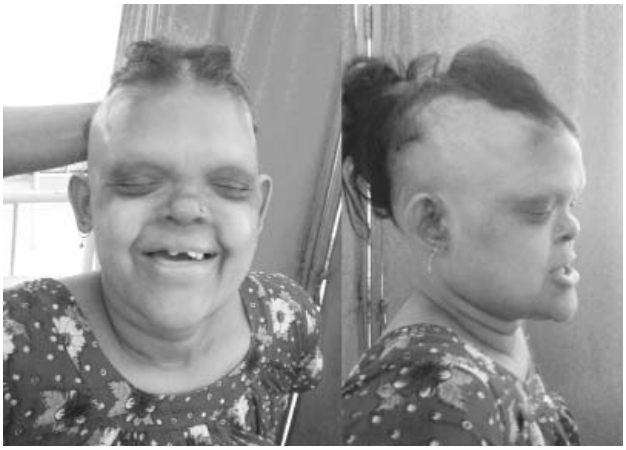

Fig I & II- Sparse hair, drawn backward hairline, conical teeth and saddle nose

Her height was 5'3"ft, weight was 68kg. She was mildly anaemic, oedema was absent.
Examination of cardiovascular, respiratory and other systems revealed no abnormality.
On obstetrical examination, her fundal height corresponded to period of amenorrhoea, presentation was found to be breech.
Her complete blood count, renal function tests were fairly normal, there was no proteinuria, blood group AB+ve, blood sugar was normal. ECG showed anterior ischemia but ECHO was normal.

Caesarean section was done and a healthy female of 2.7 kg was delivered. No congenital abnormality was found. As, the mother has very poor breast and nipple development, the baby was advised artificial feeding. The patient was discharged after 5 days of C/S without any complication.

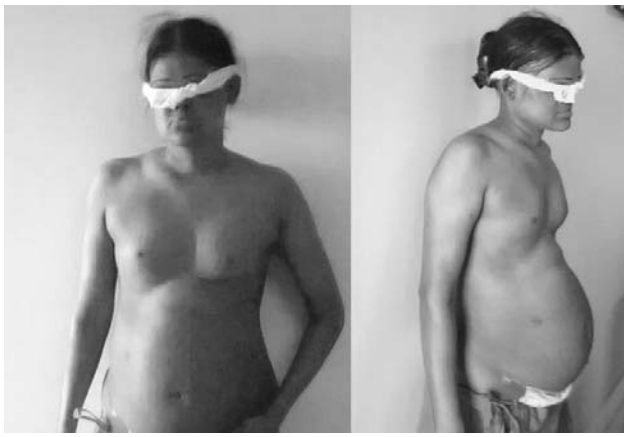

Fig III & IV- Poor breast and nipple development associated with AED

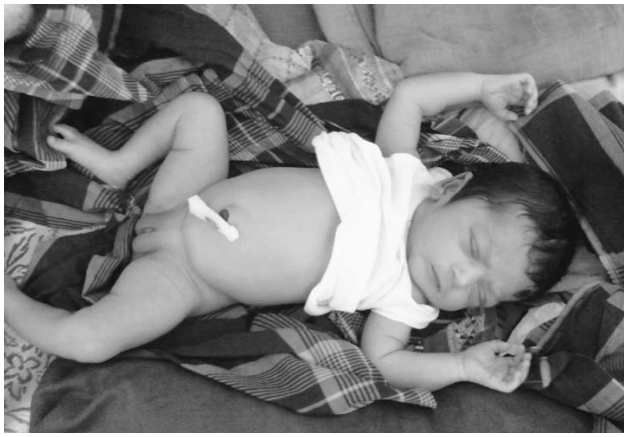

Fig V- The baby

Discussion

Anhydrotic ectodermal dysplasia in this case was diagnosed earlier due to the complaints of heat intolerance produced by absence of sweat glands. Ectodermal dysplasia of the mother did not pose any threat to the fetus.
The breasts of the case were not developed. Breastfeeding by the mother will not be possible.The complete syndrome does not occur in females[3]. Conception and childbirth is not an abnormal event in such cases. As in most cases of ED her intellectual and mental domains were normal.

Conclusion

Laboratory identification of genes and mode of inheritance of mutant genes associated with X

chromosome or autosomes can be done if facilities are available and it requires further probing through genetic analysis. Conception and childbearing is a normal event in female cases with ectodermal dysplasia with otherwise normal features. As these patients lead almost near normal lives prenatal diagnosis for early detection and abortion is not necessary.

Ethical consideration

Consent was taken from the patient for publishing her case history and photographs.

References

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