Autosomal recessive anhidrotic ectodermal dysplasia in a large Moroccan family

K Kabbaj, L Baala, H Chhoul, A Sefiani

Abstract
We studied a large Moroccan family in which anhidrotic ectodermal dysplasia is transmitted as an autosomal recessive trait. Fourteen family members, both males and females, were affected and they all had a common ancestor. Linkage analysis by homozygosity mapping in this family will permit the gene localisation of this rare form of anhidrotic ectodermal dysplasia.

Keywords: anhidrotic ectodermal dysplasia; autosomal recessive transmission; consanguinity

Anhidrotic or hypohidrotic ectodermal dysplasia (AED) is a disease characterised by total or partial anodontia, absent or reduced sweating (anhidrosis or hypohidrosis), and sparse hair (atrichosis or hypotrichosis). X linked recessive AED (MIM 305100), the most frequent form of this disorder, was first described by Thurnam in 18481 and then by Charles Darwin in 1875. A clinically indistinguishable autosomal recessive variant of AED (MIM 224900) has been described in a few cases.2

In this paper, we present a very large inbred family, which includes five generations with eight women and six men all affected with AED. An autosomal recessive mode of inheritance is suggested by the consanguinity of the healthy parents who had affected sons and daughters.

Case report
The proband was an 8 year old girl at the time of initial examination (fig 1). She was born in 1985 to a consanguineous couple. She had an uncomplicated prenatal period and a normal birth. The neonatal period was complicated by episodes of hyperthermia and recurrent chest infections and she failed to thrive. In addition, she had few conical shaped teeth with two maxillary canines, four molars, and two mandibular canines (fig 2). It was also noted that she had thin and sparse hair and she was unable to sweat because of the lack of sweat glands. Furthermore, she had a distinctive face with a depressed nasal bridge, prominent lips, periorbital wrinkles, and pigmentation. An X;autosomal translocation associated with the AED phenotype was excluded by cytogenetic analysis.

Family study
The family survey showed that 13 other patients were affected with AED. They all had consanguineous parents and lived in Shoul, about 50 km from Rabat. Seven patients were still living and we examined them. The other six had died from severe episodes of hyperthermia. They all had hypotrichosis, hypodontia, and anhidrosis as the most striking features. This triad is sufficient for the diagnosis of anhidrotic ectodermal dysplasia. The pedigree (fig 3) and the family history suggest autosomal recessive transmission of this disease.

Discussion
Anhidrotic (or hypohidrotic) ectodermal dysplasia is a heterogeneous condition which includes different clinically indistinguishable genetic variants.3 Thurnam1 reported the first description of the clinical features of ectoder-
Pedigree of the family. The arrows indicates the proband.

An autosomal recessive trait supports these findings. The localisation and identification of the gene involved in this family by homozygosity mapping and positional cloning should open the field for the molecular study of many other ectodermal dysplasias.

We would like to thank Professor M Hassar for his assistance during the preparation of this paper. We also thank the patients and their families for their cooperation.

2 Thurnam J. Two cases in which the skin and teeth were very imperfectly developed. Proc R Med Chin Soc (Lond) 1846;31:71-82.