

Congenital Ectodermal Dysplasia

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Arif-un-Nisa Naqvi (Department of Biological Sciences, Quaid-i-Azam University, Islamabad, Pakistan Agricultural Research Council, Islamabad.)

Abstract

Two cases of phenotypically similar ectodermal dysplasia characterised by reduction of hair, diminished sweat pores, missing teeth and somewhat cracked and abnormal skin were studied to understand the genetic basis of this disorder with the help of pedigrees. The mode of inheritance in one case was suggested to be x-linked where as in the other case it was autosomal recessive. (JPMA 35 225, 1985).

Introduction

Congenital ectodermal dysplasias (EDs) are a group of familial disorders that affect tissues and organs of ectodermal origin.¹ Ectodermal dysplasia in humans is a complex and heterogeneous group which has been classified into thirty two conditions².

An autosomal recessive inheritance was suggested for a form of hypohydrotic ectodermal dysplasia in which sweat pores were reduced in number but were not completely absent³

The, mode of inheritance in two siblings affected with ectodermal dysplasia with sensorineural hearing loss was suggested to be autosomal recessive.¹

Bixler et al.⁴ have reported two cases of ectrodactyly-ectodermal dysplasia clefting (EEC) syndrome and suggested an autosomal dominant inheritance with incomplete penetrance for the syndrome.

An association between ectodermal dysplasia, cleft lip, cleft palate and scrubbing brush hair was seen and this disorder was determined by an autosomal gene⁵.

The present study includes two cases of phenotypically similar ectodermal dysplasia characterised by reduction of hair, diminished sweat pores, missing teeth and somewhat cracked and abnormal skin. An attempt has been made to understand the genetics of this disorder with the help of pedigrees.

Pedigrees.

Figure 2 contains a pedigree of ectodermal dysplasia in which 1-4 and 1-5 are first cousins whereas all other couples of the pedigree are unrelated. The propositus is the resident of Islamabad and is forty years old.

Figure 3 shows the pedigree of a family of Rawalpindi district. A brother and sister are affected with ectodermal dysplasia and most of the marriages in this family are consanguineous.

Discussion

Symbols generally used in a pedigree are shown in figure 1.



Fig. 1 Symbols generally used in a pedigree.

The pedigree shown in

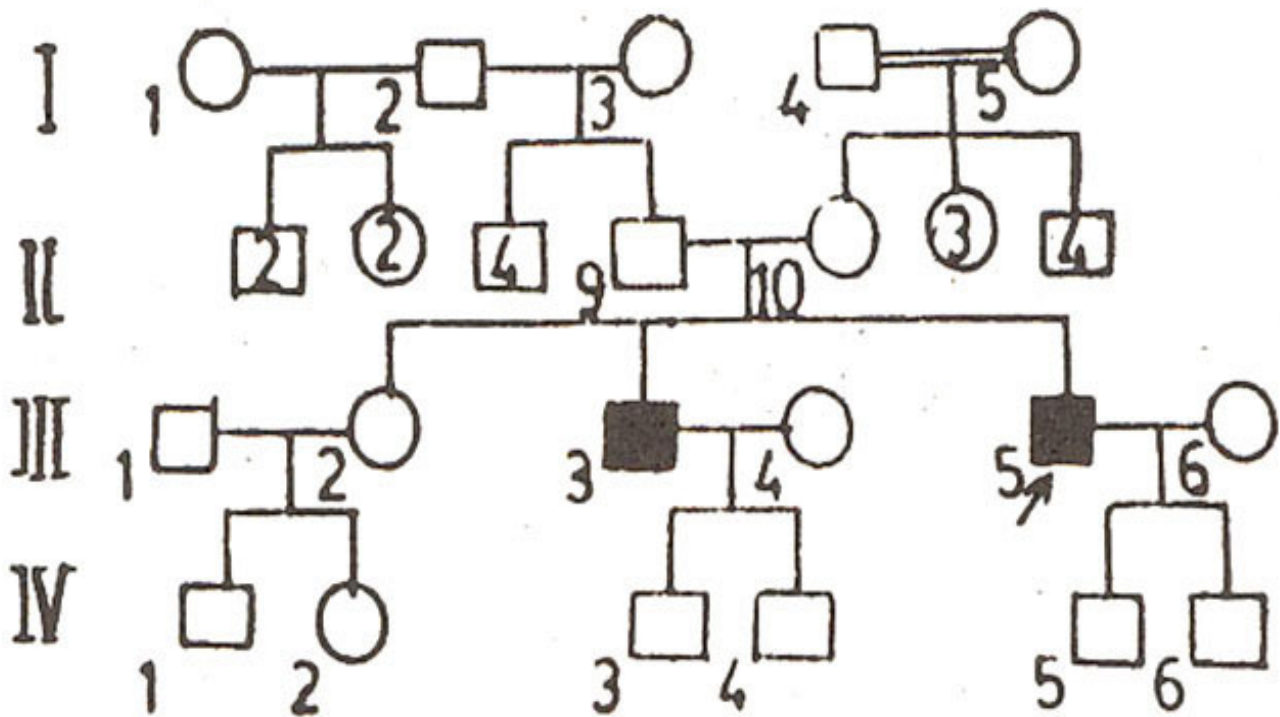


Fig. 2. Pedigree of Ectodermal dysplasia. 1-4 and 1-5 are first cousins whereas all other couples of the pedigree are unrelated.

figure 2 can be best explained by postulating an x-linked recessive inheritance for the trait. According to this hypothesis 11-10 is a carrier and the trait is expected to affect her male offspring only. The female offspring 111-2 can be normal as well as a carrier. For the following reasons it is less likely that the trait is determined by an autosomal recessive gene.

(a) As the couple 11-9 and 11-10 are not blood relatives the probability that both of the are heterozygous carriers is relatively low considering the low incidence of ectodermal dysplasia in the general population.

(b) On the basis of this hypothesis, irrespective of sex, a quarter of the offspring are expected to be affected. While the pedigree shows two out of the three offspring affected and both the affected persons happen to be males, though the possibility that it is merely a coincidence cannot be ruled out.

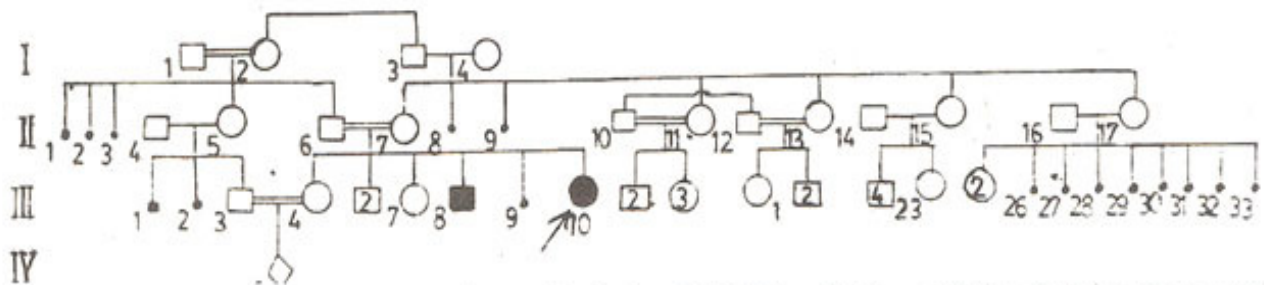


Fig 3. Pedigree of Ectodermal dysplasia. The couples I - 1 and 1-2; II-6 and II-7; and III-3 and III-4 are first cousins whereas the couples II-10 and III-1; and II-12 and II-13 are second cousins. III-8 and III-9 died at the age of 6 months and seven days, respectively, both were affected. The symbols II-1-2-3-8-9, III-1, 2, 6-27-28-29-30-31-32-33 signify deaths in early infancy.

The pedigree shown in Figure 3 can be explained by postulating an autosomal recessive nature of the trait.

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The cases reported by Crump³ and Mikaelian¹ also have similar genetic basis. The couple 11-6 and 11-7 are first cousins and probably both are heterozygous carriers because two out of seven of their offspring, which is roughly a quarter, are affected. Some of the early deaths referred to in the pedigree might have been caused by ectodermal dysplasia.

Reference

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