Odontomicronychial ectodermal dysplasia

Marta Pinheiro, Andrea Luiza Snel, Newton Freire-Maia

Abstract

This paper describes odontomicronychial dysplasia, a pure ectodermal dysplasia of the 2-3 subgroup of group A. It is characterised by precocious eruption and shedding of deciduous dentition, precocious eruption of secondary dentition with short, rhomboid roots, and short, thin, slow growing nails. This condition probably results from an autosomal recessive gene.

(*J Med Genet* 1996;33:230-232)

Key words: ectodermal dysplasia; dentition; autosomal recessive inheritance.

The ectodermal dysplasias form a nosological group of over 150 conditions, classified into 11 clinical subgroups, and encompasses both pure dysplasias and syndromes; 101 (66%) of them have a genetic cause (41 autosomal dominant, 52 autosomal recessive, and eight X linked) and the remaining 53 (34%) conditions have an unknown cause.¹ Here we report on an apparently hitherto undescribed pure ectodermal dysplasia of the 2-3 (odonto-onychia) subgroup of group A, probably resulting from an autosomal recessive gene, and characterised by the dental and nail alterations that warrant its classification as a 2-3 condition. We propose the name odontomicronychial dysplasia.

Family history

The condition was found in two sibships of a large Brazilian family with remote Portuguese ancestry (fig 1). The unaffected parents of the proband (VI.2) are second cousins ($F=1/64$) and third cousins once removed ($F=1/512$): total $F=9/512=0.01758$. The unaffected parents of IV.101, IV.102, IV.103, and IV.110 are first cousins ($F=1/16$). Apart from patients IV.101 and IV.102, who have already died, all the others are healthy. In the first inbred sibship there are an affected male and a normal female. In the second inbred sibship there are three affected males, one normal male, one affected female, and nine normal females. The total of the two sibships is 10 normal females, one affected female, one normal male, and four affected males. There are 11 normal sibs and five affected sibs, a ratio close to 3:1 ($\chi^2=0.33$, $p>0.50$) which becomes closer to it (11:3) by the elimination of one affected in each sibship ($\chi^2=0.09$, $p>0.80$). The total sex ratio (5:11) does not depart significantly from 1:1 ($\chi^2=2.25$, $p>0.10$).

Case reports

CASE 1

The proband VI.2 (figs 1 and 2) was the second child of a 23 year old mother and a con-

sanguineous 27 year old father. He was born at term after a normal pregnancy and delivery. At birth, the proband weighed 4800 g (97th centile) and measured 50.0 cm (25th centile). During infancy he had several dental anomalies that led the parents to seek dental assistance. Nail alterations (short, thin, with slow growth) were noted later.

At 7 years 9 months he weighed 27 kg (75th centile) and measured 135 cm (97th centile). Apart from the dental and nail disturbances he was healthy.

ODONTOLOGICAL HISTORY

Deciduous dentition (denoted by Roman numbers) $\overline{\text{II}}$, $\overline{\text{I}}$, $\overline{\text{I}}$, and $\overline{\text{II}}$ erupted when the proband was 1½ months of age and exfoliated about one month later; teeth $\overline{\text{III}}$, $\overline{\text{III}}$, $\overline{\text{IV}}$, $\overline{\text{III}}$, $\overline{\text{II}}$, $\overline{\text{I}}$, $\overline{\text{I}}$, $\overline{\text{II}}$, and $\overline{\text{III}}$ exfoliated at 4½ years and $\overline{\text{IV}}$, $\overline{\text{V}}$, and $\overline{\text{V}}$ were extracted because of crown destruction caused by caries. At this time, teeth $\overline{\text{V}}$, $\overline{\text{IV}}$, $\overline{\text{IV}}$, and $\overline{\text{V}}$ were also affected by caries and were filled with silver diamine fluoride to restrain progression of caries. Permanent dentition (denoted by Arabic numbers) 6, 6, 6, and 6 erupted at 2½ years of age and had short and rhomboid roots; 1 and 1 erupted at 3 years of age and exfoliated about two years later owing to their increased mobility secondary to short and rhomboid roots; 2 and 2 exfoliated at 6 years of age; 2, 1, 1, and 2 erupted at 7 years of age. Teeth 1 and 1 were extracted (owing to the absence of osseous implantation) some months later. Canines, premolars, and second molars (both maxillary and mandibular) have not erupted yet. In summary, both the upper and lower dentition had precociously erupted teeth; all deciduous teeth exfoliated precociously; and deciduous and permanent teeth had short crowns and roots. Besides being short, the roots of the permanent teeth are also rhomboid, the process of development of the permanent teeth is advanced for the patient's chronological age, and neither enamel hypoplasia nor primary hypodontia were noted in either dentition.

CASES 2-5

Patients IV.101, IV.102, IV.103, and IV.110 are double first cousins twice removed ($r=1/16$), double second cousins twice removed ($r=1/64$), first cousins three times removed ($r=1/64$), and second cousins three times removed ($r=1/256$) of the proband (total $r=25/256=0.09766$). They are described by other family members as having similar dental and nail alterations as those seen in the proband. Two of them (IV.101 and IV.102), as mentioned

Department of
Genetics, Federal
University of Paraná,
Caixa Postal 19071,
81531-990 Curitiba,
PR, Brazil
M Pinheiro
A L Snel
N Freire-Maia

Correspondence to:
Dr Pinheiro.

Received 13 December 1994
Revised version accepted
for publication
7 November 1995

Figure 1 Pedigree of the family.

before, have already died; the other two have unknown addresses and could not be located.

Discussion

Several facts, such as the corrected ratio of 11:3, the occurrence of the condition in both sexes

(1F, 4M), the presence of affected subjects in (two) related sibships who are the offspring of normal consanguineous couples, and the presence of only normal children (five males and three females) in the offspring of affected subjects, suggest that the cause of the condition is an autosomal recessive gene.

Subgroup 2-3 (odonto-onychial) of the ectodermal dysplasias at present includes eight conditions,¹ excluding the present one: deafness-onycho-osteodystrophy-mental retardation (DOOR) (MIM 220500, BDE 0262, POS 3733, FMP 1), deafness and onychodystrophy (MIM 124480, BDE 2034, POS 3581, FMP 3), ectodermal defect with skeletal abnormalities (FMP 2), odonto-onychodysplasia (POS 4102, FMP 5), Kirghizian dermatosteolysis (MIM 221810, BDE 3044, POS 3604, FMP 6), corneodermato-osseous (CDO) (MIM 122440, BDE 2760, POS 3753), lacrimo-auriculo-dento-digital (LADD) syndrome (MIM 149730, BDE 2180, POS 3546), and hearing loss, sensorineural, with enamel hypoplasia and nail defects (MIM 234580). (MIM refers to McKusick number,² POS to POSSUM number, BDE to the numbers referred to in the *Birth Defects Encyclopedia*,³ and FMP to those referred to in our book.⁴) The condition described here, odontomichronychial dysplasia, is different from all the above ectodermal dysplasias and increases the number to nine in the subgroup 2-3 of EDs.

We are most grateful to all family members, particularly to Mrs Maria Regina Pereira Espindula (mother of the proband), whose valuable cooperation made this investigation possible. The authors also acknowledge the assistance of Dr Eleidi A Chautard-Freire-Maia for critically reading this paper, Dr Chandal Maria Meirelles Nasser for being an efficient contact between us and the family described, Mrs Néria Amorim Maia Ehmke for dermatoglyphic analysis, and Miss Irene Sedoski for typing this manuscript. This work was supported by grants from the Conselho Nacional de Desenvolvimento Científico e Tecnológico.

Figure 2 Left hand and orthopantomogram of the proband (VI:2 in fig 1) at the age of 4 years.

lógico (CNPq, Brazil). The first and third authors are research fellows of CNPq.

- 1 Pinheiro M, Freire-Maia N. Ectodermal dysplasias: a clinical classification and a causal review. *Am J Med Genet* 1994; 53:153–62.
- 2 McKusick VA. *Mendelian inheritance in man: a catalog of human genes and genetic disorders*. 11th ed. Vols 1 and 2. Baltimore: Johns Hopkins University Press, 1994.
- 3 Buyse ML, ed. *Birth defects encyclopedia*. Cambridge, MA: Blackwell, 1990.
- 4 Freire-Maia N, Pinheiro M. *Ectodermal dysplasias: a clinical and genetic study*. New York: Alan R Liss, 1984.