A syndrome of hypohidrotic ectodermal dysplasia with normal teeth, peculiar facies, pigmented disturbances, psychomotor and growth retardation, bilateral nuclear cataract, and other signs*

Summary. This paper describes a 7-year-old girl with trichodysplasia, normal teeth, onychogryposis, hypohidrosis, psychomotor and growth retardation, dry and warm skin with follicular hyperkeratosis, pigmentary disturbances (hyper- and hypochromic spots), bilateral nuclear cataract, dermatoglyphic anomalies, and other signs. This condition is considered a new form of ectodermal dysplasia.

The concept of ectodermal dysplasia varies widely among different authors. A recent review included 32 syndromes (Freire-Maia, 1971). A further unpublished review (Freire-Maia, 1975) lists 45 apparently different syndromes with signs involving at least two of the following groups of disturbances: trichodysplasia, abnormal dentition, onychodysplasia, and dyshidrosis.

The patient to be described here combines an unusually diverse variety of manifestations. As far as we know, this syndrome has not been previously described by other authors. A preliminary note on this case was published in 1973 (Freire-Maia et al., 1973).

Case report

The patient, who is a very dark mulatto, was born on 9 May 1967, and was initially examined on 22 February 1973, at 5 years 9 months; subsequent examinations were made up to June 1973.

On physical examinations, she presented trichodysplasia, ie, partial alopecia, especially in the fronto-temporal regions with straight, dry, and brittle hair showing a slightly reddish colour at the tips; almost complete absence of eyebrows, scarce eyelashes; apparently complete absence of body hair, and a greater than normal tendency to lose hair at the parieto-occipital region where she rests her head on the pillow (Fig. 1); frontal bossing and depressed bridge of nose; normal teeth; severe onychogryposis (Fig. 2); dry and warm skin, with slight follicular hyperkeratosis which is most evident at the scalp; hypochromic spots at the feet, knees, arms, neck, and forehead; large hypochromic spot at the antero-external region of the right forearm. In April 1973 weight (12350 g) and height (90 cm) were those of an approximate age of about 2.5 years; head circumference was at the 10th centile for age.

Language development is very limited, and she is able to enunciate only very few words clearly and correctly. Language development is at a 15–18 month level. Mental age lies between 1.5 and 2 years. IQ is probably not greater than 33 (Mehl, 1974). Neuromotor development corresponds to the approximate age of 2 years. The patient began to walk at 5.5 years. Coordination and fine motor skills are at a level of 2.5 years. Deep tendon reflexes are symmetrically hyperactive; the superficial reflexes are immature. EEG and brain scan are normal (Abdala, 1974).

Bone age falls between 3–4 years. Radiographs of the skeleton were normal. Ophthalmological examination showed a bilateral nuclear cataract, probably congenital. Fundoscopy was normal.

* This study was supported by Grants from the National Research Council (Brazil), the World Health Organization, The Bamberindus Foundation and the Teaching and Research Council of the Federal University of Parana to the Department of Genetics of the same University. Dr Opitz was supported by PHS/NIH Grants GM 1542 and 5 KO4 18982. Contributed in part as Paper No. 1722 from the University of Wisconsin Genetics Laboratory.
Case reports

Comedingly rare. On the basis of Freire-Maia's classification (1971), the patient's disorder belongs to the tricho-onycho-dys(hypo)hidrotic (or 1–3–4) subgroup of the ectodermal dysplasias.

No other primary anomalies of differentiation are present; this child, therefore, does not have a multiple congenital anomaly/mental retardation syndrome, but a 'pure' dysplasia, in this case an ectodermal dysplasia, with apparent involvement of the CNS (ie, a neuro-ectodermal dysplasia).

The low TRC (56) reflects not only traumatic absence of the ridges of the left fifth finger but a true reduction below the mean for Brazilian females (127.2; SD = 49.9; Toledo et al, 1969) and of the average finger count (12.7 in that normal sample versus 6.2/9 fingers in our patient). The significance of this finding is unknown since her first-degree relatives cannot be studied; however, it is possible that reduction of TRC may represent another manifestation of ectodermal dysplasia in this child. A correlation between reduced head size and IQ and decreased TRC has been demonstrated in the Brachmann-De Lange syndrome (Barr et al, 1971). The fact that all the fingers of our patient have ulnar loops may be coincidental since in the normal population mentioned above ulnar loops represent 68% of the patterns. The absence of digital triradius c of the left palm may also be a normal variation. However, Walker's ratios (12.8% and 13.7%) are somewhat lower than the normal values for the above mentioned Brazilian series (20.7%, SD = 9.6 for the right hand, and 20.2%, SD = 9.5 for the left). The excess of secondary creases marking the hands and fingers of our patient is clearly abnormal.

Psychomotor retardation and cataracts are also interpreted as manifestations of ectodermal involvement. The slight difference between skin and rectal temperatures shows that the patient has a disturbance of skin heat regulation. The low percentage of buccal cell X-chromatin bodies is without explanation, however, it may again reflect a secondary manifestation of (subclinical involvement of the buccal mucosa in) the ectodermal dysplasia process.

Data on this patient were evaluated by Professor Carl Witkop of the University of Minnesota. He thinks: '... that the fingernail material does fall in the hyperkeratotic onycholysis type of defect. This is very similar to the type of defect we see in our syndrome of hypoplastic enamel, onycholysis and hypohidrosis, and ... is also similar to the ectodermal dysplasia of the hidrotic type reported by Jacobsen and Clouston and Wilkey and Stevenson among Canadian families.' Dr Witkop concludes that our

Discussion

Pilocarpine-iodine-starch method (also applied, at the same time, to a normal control girl) revealed severe hypohidrosis, suggestive of a reduced number of sweat glands; no fever or apparent heat discomfort (even on hot days) was verified. The difference between her axillary and rectal temperature is generally between 0.2–0.4°C.

The karyotype is normal (40 cells examined). Sex chromatin in squash preparations of oral mucosa were present in 12% of the 200 nuclei analysed.

Dermatoglyphic analysis showed absence of triradius c of the left palm; TRC = 56 in nine fingers (no pattern was present in the left finger due to trauma). There was an ulnar loop on the other nine fingers. Walker's ratio is 12.8% on the right hand and 13.7% on the left. Both palms and all the fingers show a marked increase in secondary creases ('white lines'). She has a tibial loop in both hallux areas. Her medical history is unremarkable, except that she had chronic bronchitis for some 10 months around the age of 2 years.

The family of the patient could not be located; she is in the care of the Lar das Meninas, a State institution in Curitiba.

Fig. 2. Onychogryposis.

The patient clearly has an ectodermal dysplasia, with extensive involvement of many tissues of ectodermal origin; however, some of them (for example, the teeth) remain uninvolved. Her condition combines a variety of signs of other ectodermal dysplasias. This again illustrates the well known fact that the existence of pathognomonic signs among malformation syndromes and dysplasias is ex-
patient has 'a new form of ectodermal dysplasia' and that he has 'not seen a combination like this before' (personal communication to J.M.O., 28 February 1974).

We are impressed that this patient has a genetic condition, however, the exact aetiology remains unknown. The senior author would be grateful for information about similar patients.

We are very grateful to Dr Paulo Zelter Grupenhacher (for the ophthalmological examination), to Professor Dr Octavio Augusto da Silveira (for the EEG), to Professor Rui F. Pilotto (for the photographs), to Professor Dr Haydée Abdala (for the neurological data), to Professor Nelly Mattos Mehl (for the psychological investigation), to Dr Roaldo A. Koehler (for the radiological analysis), and to Professor Eleidi A. Chautard-Ferreira-Maia and Néri Amorim Maia (for help in obtaining the dermatoglyphics).

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A syndrome associated with trisomy of the short arms of chromosome No. 9 was first identified by Rethoré et al (1970). This report describes a further example of the syndrome in an adult female, born to a mother who was a balanced translocation carrier 46,XX,t(1p+9q—).

Case report

The subject is a young Caucasian woman born on 30 August 1951 to non-consanguineous mother and father aged 24 and 25 years, respectively, at her birth. Birth weight was 2 900 g. There was a history of threatened abortion at 3 and 6 months. She had jaundice at 3 weeks of age and appeared to be mentally retarded by the age of 1 year. She was sitting up at 3½ years and standing at 5 years. She could say single words at 4 years. She had an operation for ptosis at the age of 6 and attended a special school until her admission to a hospital for the mentally handicapped on 13 March 1964. Since her admission to hospital there has been little change in her physical and mental state.

Physical state (Figs. 1 and 2). She is of small stature; height 134.0 cm and weight 3 600 g. She has fair hair, blue eyes, and rather small ear lobes. Cranial circum-

REFERENCES


A case of partial (9p) trisomy in a family with a balanced translocation 46,XX,t(1p+9q—)

Summary. A case of partial trisomy 9 is described, confirming that this will produce a recognizable syndrome of a characteristic facies with deep-set eyes and an unusual shape of the nose. Failure of secondary sexual characteristics to develop appears to be a feature in adults. In this case the mother had a balanced translocation between chromosomes 1 and 9 and must have, in addition, had a non-disjunction of her normal and her deleted No. 9 in order to produce the unbalanced state in her daughter.

Received 4 November 1974.