TRICHORHINOPHALANGEAL SYNDROME TYPE 1

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A case of trichorhinophalangeal syndrome type I (TRPS- I) in a 10 year old Muslim girl is reported. She has typical features of hypotrichosis, pear-shaped nose, long philtrum, receding chin, and deformed angulated fingers. Radiology showed characteristic cone-shaped epiphyses of middle phalanges. An autosomal recessive mode of inheritance is surmised.

Key Words: Sparse hair, Long philtrum, Bulbous nose, Deformed fingers, Cone shaped phalangeal epiphyses

Introduction

This disorder, which is more common in women, is usually determined by an autosomal dominant gene although there is evidence of a recessive pattern of transmission,[1] and is caused by deletion of chromosome band 8q 24.12.[2] It is characterised by fine, sparse, brittle hair with variable degrees of alopecia, a long pear-shaped nose, high philtrum, tubercle of normal skin below the lower lip, and brachyphalangeal dysostoses.[3,4] In addition, eyebrows are dense medially and sparse laterally but nail changes are not consistent. Radiologically, the epiphyses of middle phalanges are typically cone-shaped which results in their shortening together with deformity of the proximal interphalangeal joints. Craniofacial abnormalities include mandibular hypoplasia and maxillary prognathism.

Case Report

A 10 year-old Muslim girl presented with fronto-temporal alopecia with fine, sparse hair present since early childhood. Eyebrows were sparse on lateral compared to the medial sides. Nose was bulbous. Philtrum was high and the lips were large compared to the lower part of the face with receding chin (Fig. 1). A tubercle of normal skin could be felt below the lower lip. Ears were everted. There was swelling of the proximal interphalangeal joints with angulation of the fingers at the same joints (Fig. 2). Intelligence was below average, Speech development was normal, There were no other skin or nail changes. The girl's height was 125 cm and weight was 21 kg. There was no history of inability to sweat. Straight X-ray of the limbs revealed increased density of the epiphyses of distal phalanges of 2nd, 3rd and 5th fingers and toes. Premature epiphyseal fusion was noted in some. Elder brother and 3 younger brothers and a sister

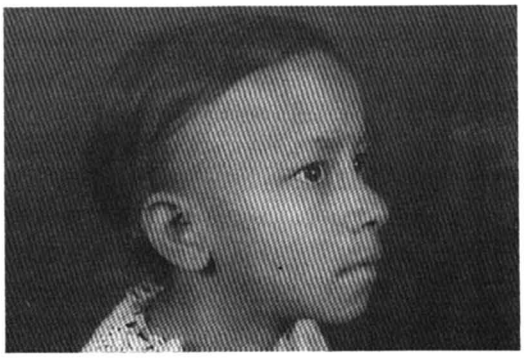

Fig. 1. Profile view of the patient.

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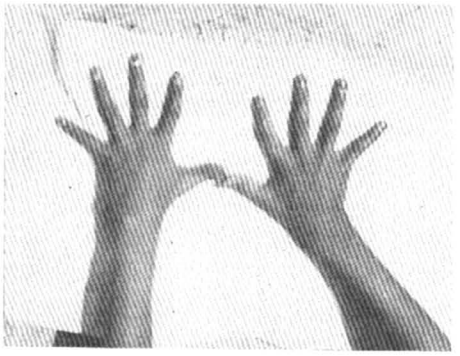

Fig. 2. Deformed angulated fingers.

were all normal, except that one of the brothers aged 8 years showed increased density in some of the epiphyses of distal phalanges of hands. Parents are not affected and there was no history of consanguinity. Systemic examination and routine haematological and urinalysis did not reveal any abnormality. There was no ECG abnormality.

Discussion

Though in most of the cases this genetic disorder is transmitted by autosomal dominant gene, in our case an autosomal recessive transmission is most likely as one of the brothers out of a total of 5 siblings had increased density in some of the epiphyses of the distal phalanges of the hands and might even develop other features in future as a late onset of the disorder is known. Even though there is no history of consanguinity the patient is Muslim and inter marriages are known within that community leading to increased prevalence of autosomal recessive genes. It can be differentiated from TRPS Type II by the absence of features like mental retardation, multiple exostoses on X-ray, microcephaly, loose redundant skin, delayed speech, winged scapulae, joint hyperlaxity, pes planus, osteoporosis, scoliosis, avascular necrosis of femoral head.[2,3] However in our case, intelligence of the patient was not in accordance with the chronological age. Another variant, TRPS-Type III has been described which includes severe generalized shortness of all phalanges, metacarpals and metatarsals in addition to the usual features of type I.[2]

References

1. Gorlin RJ, Cohen MM, Wolfson J. Tricho-rhinophalangeal syndrome. Am J Dis Child 1969; 118 : 595-9.
2. Corrington PR, Chen H, Altick JA. Tricho-rhinophalangeal syndrome, type 1. J Am Acad Dermatol 1994; 1 : 331-6.
3. Harper J. Genetics and genodermatoses. In: Textbook of Dermatology (Champion RH, Burton JL, Ebling FJG, eds). 5th edn. London: Blackwell Scientific publications, 1992 : 340-1.
4. Bertolino AP, Freedbug IM. Hair. In : Dermatology in General Medicine (Fitzpatrick TB, Eisen AZ, Wolff K, et al, eds), 4th edn. New York: McGraw-Hill 1993: 681.