Brachydactyly and Ectodermal Dysplasia - Is It a New Association?

Sir,

Brachydactyly means shortness of fingers and toes. The shortness is relative to length of the other long bones and other parts of the body. Brachydactyly most often occur as an isolated dysmilia but can occur with other anomalies as part of many other congenital syndromes. The majority of the types of isolated brachydactyly are inherited as autosomal dominant conditions. Some of the medical causes of brachydactyly are pseudohypoparathyroidism, Down’s syndrome, gonadal dysgenesis, familial brachydactylism and Weil-Marchesani’s syndrome.

A 47 year old lady came to our outpatient department complaining of decreased sweating during heavy exertion. On examination, she had a dry skin and brachydactyly with cleft between big toe and second toe (Fig. 1). Her hair, teeth and nail were normal. X-ray of foot revealed short third and fourth metatarsals (Fig. 2). Her two children were found to have characteristic features suggestive of hypohydrotic ectodermal dysplasia including sparse hair, hypodontia and dry skin with confirmatory skin biopsy. This lady had only minimal expression of the disease like dry skin and brachydactyly.

Hypohydrotic ectodermal dysplasia is characterized by partial or complete absence of sweat glands, hypotrichosis and hypodontia. This condition is usually an X linked recessive disorder affecting predominantly males. Females are usually carriers. This disorder is transmitted through healthy female carrier, occasionally the heterozygous female may show some features of this condition. The complete syndrome does not occur in females.

There are about 63 medical conditions which have been associated with brachydactyly. Among the causes of brachydactyly, hypohydrotic ectodermal dysplasia has not been reported so far in the literature. Probably this may be the first case to report ectodermal dysplasia as a cause for the brachydactyly. Ectrodactyly has been associated with ectodermal dysplasia, cleft lip and/or cleft palate which is called EEC syndrome.

REFERENCES


Fig. 1: Photograph of foot showing dry skin and brachydactyly with cleft between big toe and second toe.

Fig. 2: X-ray foot showing short third and fourth metatarsals.