CONGENITAL BILATERAL SENSORY NEURAL DEAFNESS, POSTAXIAL POLYDACTYLY, SCROTAL TONGUE AND RECURRENT PYOGENIC INFECTIONS

Puneet Bhargava, C M Kuldeep, Kailash Kumar, N K Mathur

A case report of a 40-year-old male with congenital bilateral sensory neural deafness, postaxial polydactyly involving hands and feet, scrotal tongue and recurrent pyogenic infections is described.

Key Words : Deafness, Polydactyly, Scrotal tongue

Introduction

Oro-facio-digital syndrome type IV is an autosomal recessively inherited syndrome complex with clinical characteristics including lobulated tongue, pseudocleft of lip, pre and postaxial polydactyly of hands and feet, severe talipes equinovarus, mesomelic limb shortness associated with tibial hypoplasia and severe bilateral deafness.[1] A case report of a patient who presented with some of these features is described.

Case Report

A 40-year-male patient with normal intelligence presented to us with recurrent pyogenic infections for last 20 years. Past history showed that he was born with bilateral deafness, duplication of little fingers in both hands and feet and an abnormal tongue. No other family member had similar illness. Clinical examination revealed multiple lesions of folliculitis and impetigo scattered on the face, back, chest and arms. (Figs. 1,2). There was duplication of little fingers in both hands and little toes in feet (Fig.3). Tongue had a deep logitudinal groove with more or less deep radiating grooves dividing the tongue in a transverse cerebriform configuration consistent with diagnosis of scrotal tongue. There was no other systemic abnormality. Clinical and laboratory investigations including routine haemogram, X-ray chest, ECG, EEG, CT scan skull, USG abdomen, and liver function tests were normal. Audiometry revealed severe bilateral sensory neural deafness.

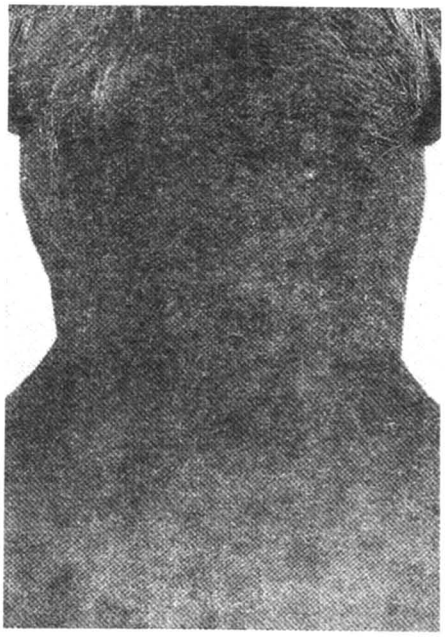

Fig. 1. Pyoderma lesions on the back neck.

Discussion

Teratology studies have shown that intake of drugs such as acetazolamide,

From the Department of Dermatology, SMS Medical College, Jaipur, India.

Address correspondence to : Dr N K Mathur C-24, Peeyush Path, Bapu Nagar, Jaipur-302015.

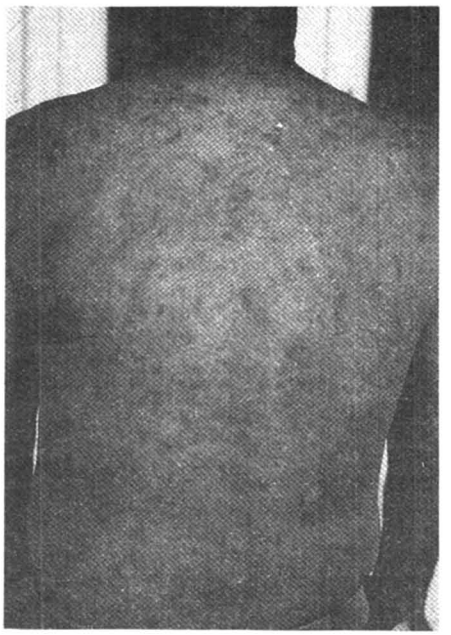

Fig. 2. Pyoderma lesions on the back.

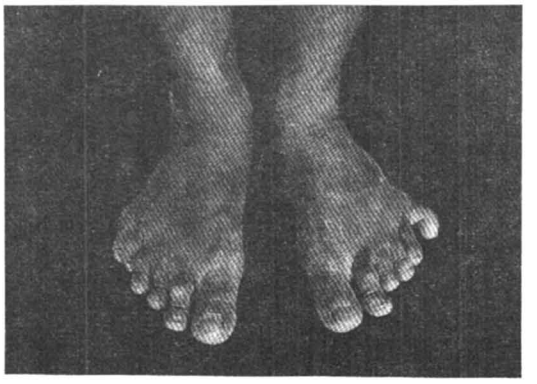

Fig. 3. Polydactyly of feet.

adenine, 1,7-dimethylxanthine, aminophylline, retinoic acid, aspirin, valproic acid and acetoxy-methyl-methylnitrosamine during pregnancy can cause limb defects including polydactyly in some children.[2,3] However, drug history could not be extracted in our patient due to his ignorance and death of his mother due to old age.

Polydactyly can be a feature of neurofibromatosis type-I[4] but lack of cafe-au-lait spots, cutaneous neurofibromas and other features excluded this diagnosis. Polydactyly with lobulated tongue and congenital deafness can be present in oro-facio-digital syndrome type-IV. This syndrome has autosomal recessive inheritance, but complete features of this syndrome were also not present in this patient. It appears that the patient had a new constellation of abnormalities which arose due to a sporadic mutation. Lack of similar abnormalities in 3 generations of his family supports this concept.

References

1. Nevin NC, Thomas PS. Oro-facio-digital syndrome type-IV : report of a patient. Am J Med Genetic 1989; 32: 151-4.
2. Sanders DD, Stephens TD. Review of drug-induced limb defects in mammals. Teratology 1991; 44: 335-54.
3. Buntinx IM. Preaxial polydactyly in the fetal valproate syndrome. Eur J Pediatr 1992; 151: 919-20.
4. Merlob P, Levy Y, Shuper A. Postaxial polydactyly in association with neurofibromatosis. Clin Genet 1987; 32: 202-5