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WAARDENBURG'S SYNDROME

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A case of Waardenburg's syndrome with typical clinical features and unusual developmental anomalies of thoracic cage and upper extrmitites is presented.

Key Word : Waardenburg' s syndrome

Introduction

Waardenburg's syndrome is determined by an autosomal dominant gene with variable panetrance. The clinicial features include lateral displacement of medial canthi with dystopia canthorum, hypertrophy of the nasal root, heterochromia of iris (total or partial), confluence and hyperplasia of medial portion of eyebrows (synophrys), perceptive deafness and white forelock. In this report we describe a case of Waardenburg's syndrome having additional developmental abnormalities.

Case Report

A 20-year-old male Libyan patient was admitted in April 1991 for evaluation. The patient was deaf-mute since birth. He also had a big white forelock involving the frontoparietal area of scalp and patches of depigmentation and hyperpigmentation over the central area of face, neck and trunk since birth. (Fig. 1). His nasal bridge was thick and the medial canthi of eyes were displaced laterally. The iris of his both eyes were lighter with different grades of colour and the pupil on the left side was irregular and dialated. The eyebrows were confluent. His thoracic cage, clavicles and upper extremities were poorly developed with syndactyly of both hands and fin-like appearance (Fig. 2). His mental development was normal. Non of his parents had any manifestation of the

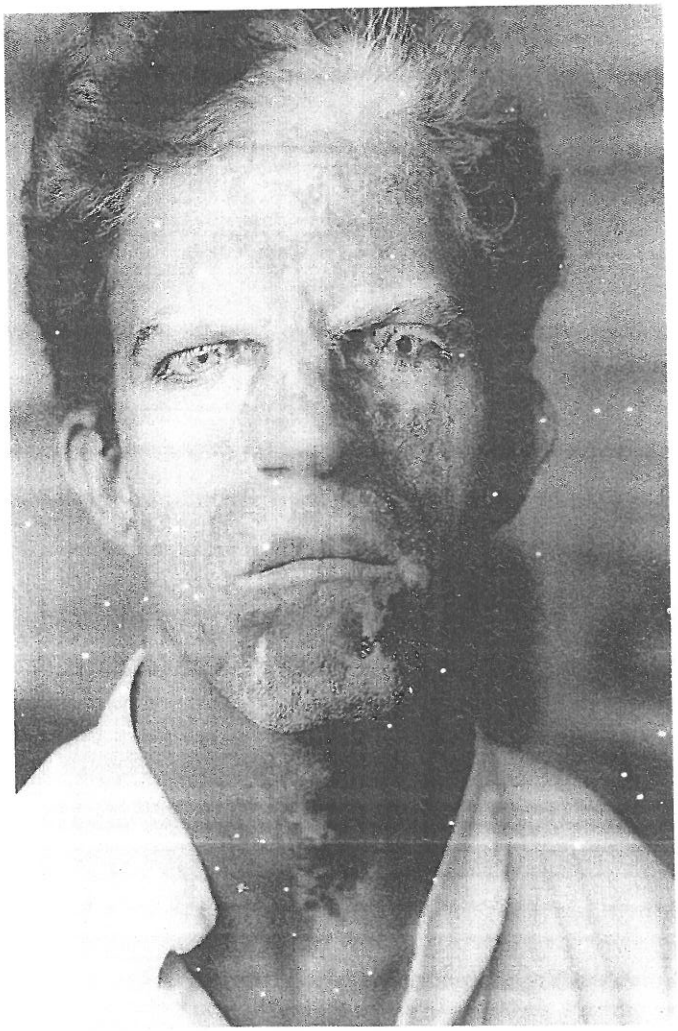

Fig. 1. Pigmentary changes on face

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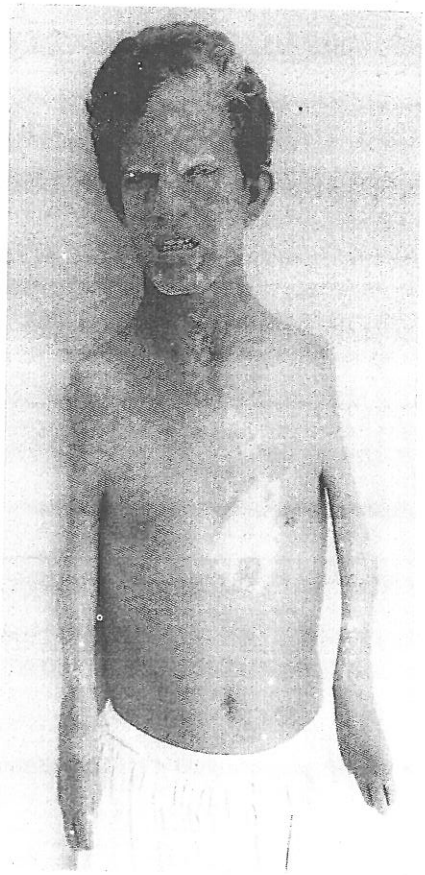

Fig. 2. Abnormalities of upper limbs

syndrome. One of his first cousins is supposed to have similar abnormalities but has not been seen by us.

Routine laboratory investigations on blood, urine and stools did not reveal any abnormality. Audiometry confirmed a severe sensineural deafness on both sides. X-rays of the chest and upper extremities revealed poor development of clavicles and thoracic cage with crowding of upper ribs, shortness and bowing of both ulna and radius, synostosis of carpal and metacarpal bones with reduced joint spaces and new bone formation.

Comments

The patient had all the characteristic constituents of the syndrome. He also had piebaldism. Such an association has been reported.[1,2] The developmental abnormalities of clavicles, thoracic cage, upper extremities and hands in our patient were additional prominent findings. Wiedemann et al[3] have mentioned diverse anomalies of upper extremities in this condition. Additionally, association of abnormalities like hare lip, high-arched palate, and massive jaw have been reported earlier.[2]

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