WAARDENBURG'S SYNDROME WITH LEPROSY

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Summary

A rare association of Waardenburg's syndrome and tuberculoid leprosy in a 13 year old male patient is described. This is an unrecorded feature in the literature. These two disorders are quite unrelated entities and their occurrence in the same patient is a casual one. All the classical features of Waardenburg's Syndrome except deafness were present and the disease manifested as an isolated case in the family. The pertinent literature is briefly reviewed.

Waardenburg's syndrome is a rare hereditary anomaly characterized by laterally displaced medial canthi, confluence of medial eyebrows with frequent synophrys, total or partial heterochromia iridum, unilateral or bilateral neural type of deafness and a white forelock with occasional depigmented patches on the body. Although initial description of this entity was by Van Der Hoeve in 1916 in two deaf-mute twins, Waardenburg[1], a dutch ophthalmologist was the first to correlate the ocular, auditory and dermal features of this disorder. In 1951, he surveyed the five institutes for the deaf in Holland and proposed a new syndrome. Hence this anomaly is referred to as Waardenburg's syndrome or interoculo-irido-dermato auditive syndrome.

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Received for publication on 17—9—1976

Waardenburg[1] reported that this syndrome is transmitted as an autosomal dominant trait comprising of the following components with varying degrees of penetrance :— dystopia canthi medialis lateroversa (99%); prominant nose root with absence of nasofrontal angle (78%); hyperplasia of the medial part of the eyebrows with frequent synophrys (45%); total or partial heterochromia iridum (25%); unilateral deafness or deaf-mutism (20%) and a white forelock, occasionally premature greying and very slight leucistic spots (17%). The distance between the inner angles of the eyelids is markedly increased with lateral displacement of the lacrimal puncti which lie in front of the cornea. The interpupilary and outer canthal distances are within the normal range but there is shortening of the palpebral fissure in the horizontal direction, resulting in blepharophomosis and decreased visibility of the medial part of the sclera. Epiphora and chronic dacryocystitis may occur due to the elongation of the lacrimal passage.

Following the original description of Waardenburg, further cases have been

published by various workers from different parts of the world with additional characteristic features. DiGeorge et al[2], reported this syndrome in American negroes for the first time and pointed out the occurence of double-sided blue eyes (ischypochromia iridum) in some affected persons. Fish[3] in 1959 described the first histological report of Waardenburg's syndrome and observed the absence of the organ of Corti and the atrophy of spiral gangeion and auditory nerve. Thorkilgard[4] pointed out the typical configuration of the skull; the occurrence of metopic suture and massive jaw and the presence of depigmented or brown patches on the legs, face and body of the family members. Goldenberg[5] reported 14 cases of this disorder in different races and observed that the hypochromia iridum (light blue) is characteristically associated with hypochromic or albinoid fundus. Two of his patients had associated hare lip; one had cleft palate and one had high arched palate. Amir and El-Shazly[6] in 1974 reported the association of familial hyperbetalipoproteinemia with Waardenburg's syndrome and attributed both these disorders to mutation of a single dominant gene.

Very few case reports of Waardenburg's syndrome have been published from India[7-10]. The purpose of this communication is to report a hitherto unreported association of Waardenburg's syndrome and tuberculoid leprosy in a patient.

Case Report

Male patient, aged 13 years reported to the Skin out-patient clinic of Sir Sunderlal Hospital, Varanasi on 20—6—1975 with the complaint of anaesthetic patches on the arm and thighs for 9 months.

Examination revealed three discrete, nonscaly plaques situated on the lower one third of the extensor aspect of uperarm (2x2 cms), lower third of the front of right thigh (3 cms x 4 cms) and the upper third of the front of left thigh (8 cms x 4 cms). The skin over the lesions was atrophic, hypopigmented and anaesthetic (75% loss of thermal and pain sensations) and the borders were raised. Sweating and hair were absent over the lesions. Peripheral nerves were not thickened. Histological examination of the lesions revealed features of tuberculoid leprosy.

During examination some interesting findings were noted on this patient. He had a white forelock; lateral displacement of the inner canthi and the inferior lacrimal puncti; high broad nasal root, confluence of the eyebrows on the medial part; ischypochromia irides (pale blue eyes) leukoderma on the lower lip and high arched palate (Fig. 1 Page No. 47). As revealed by the parents these lesions were present since birth. The patient was of dark complexion and average intelligence. There was no history of consanguinity in the family and none of the family members were known to have similar lesions.

Ocular examination revealed normal visual acuity. On both sides the sclera was visible on the medial aspect, the cornea placed in front of the inferior lacrimal puncti, and the iris pale blue. Ophtholmoscopy revealed hypochromia of the fundus in both eyes. The following eye measurements were obtained :—

Inner canthal distance (A) = 51 m.m.
Near Pupillary distance (B) = 66 m.m.
Outer canthal distance = 98 m.m.
Palpebral fissure = 21 m.m.

$$\frac{\text{Inner canthal distance}}{\text{Near Pupillary distance}} = \frac{A}{B} = \frac{51}{66} = 0.77$$

(If A/B index is more than 0.6 it indicates dystopia canthorum[11]).

Audiometry :— was normal.

No abnormality could be detected on examination of other systems.

Discussion

Waardenburg's syndrome is transmitted as an autosomal dominant disorder with variable penetrance of the individual components; but cases may occur sporadically also. 5 out of 16 patients in Waardenburg's series exhibited this syndrome as sporadic cases and the assumption that they were new mutations led to an estimated mutation rate of 1 per 2,70,000 gametes. In our patient also the disease manifested as a sporadic occurrence.

This syndrome manifesting individual features by themselves are not uncommon but the incidence with complete spectrum of the disorder in a single individual is very rare[13]. Our patient exhibited all the classical features of this syndrome except deafness (deafness was reported to occur approximately in 20% of the cases[1]).

In addition to the classical Waardenburg's syndrome, our patient manifested tuberculoid leprosy. This association is a casual one. Another interesting feature in our case is the occurrence of bilateral isohypochromia irides (Pale blue eyes). Di George et al[2] observed blue eyes in some affected persons of American negro families and pointed out that it is an important feature because blue eyes are not found in that race. Similarly the occurrence of blue irides is not common in our country and hence this feature may be important especially if there is a family history of this disorder. Thorkilgard[4] stated that the affected area of the iris presents as a startling white-blue colour caused by scattered areas of white hypoplastic mesenchyme of the anterior iris leaf showing against a dark blue background.

The association of pigment abnormalities and deafness in Waardenburg's syndrome is a curious feature and may be explained on the commonneural crest origin of melanocytes and acoustic nerve cells. Fish[3] reported the absence of the organ of Corti and the atrophy of spiral ganglion and auditory nerve in Waardenburg's syndrome. Further he postulated that the developmental fault in the neural crest may be responsible for the anomaly of the auditory vesicle, pigmentation of the iris and skin and of sympathetic elements.

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