Ind J Dermatol Venereol Leprol 1993; 59 : 261-3

WAARDENBURG'S SYNDROME

Sangeeta Amladi *, Saroj Sahdev **, Ashish Sukthankar *, H R Jerajani *

A 2 1/2 year old female child presented with heterochromia irides and a depigmented macule on the hand with central hyperpigmentation. There was presence of medial eyebrow hyperplasia, broad nasal root and dystopia canthorum. The fundus on the affected side was albinotic. There was no white forelock or deafness. Biopsy from the depigmented area showed an absence of melanocytes. A diagnosis of Waardenburg's syndrome type 1 was made.

Key Words : Waardenburg's syndrome, Heterochromia irides

Introduction

Waardenburg's syndrome[1] is a rare autosomal dominant disorder characterised by the following features : 1. depigmentation of hair, skin, or both; 2. congenital deafness; 3. partial or total heterochromia irides; 4. medial eyebrow hyperplasia; 5. broad nasal root; and 6. dystopia canthorum. However many observations demonstrate a heterogeneity in this syndrome. These clinical variants are described as type 1, type 2, Klein's syndrome (type 3), pseudo-Waardenburg's syndrome, and Waardenburg's syndrome-ocular albinism (table I). Other occasional associations reported are cleft lip and palate, EEG abnormalities, epilepsy, microphthalmia, anterior lenticonus and high refractive errors.[2,3] Our patient is of type 1 Waardenburg's syndrome.

Case Report

A two and a half year old girl presented with different coloured eyes (right iris with blue colour and left with brown) and a depigmented patch on the right middle finger since birth. The child was born of a nonconsanguineous marriage, was a full term normal delivery and has 3 siblings, 2 males and 1 female, all unaffceted. Her milestones were normal.

On examination, the most striking feature was heterochromia irides. This was associated with telecanthus, a slightly depressed nasal bridge and confluence of eyebrows in the midline (Fig. 1). Dystopia canthorum was present. The patient's vision was normal. Fundoscopy revealed an albinotic fundus on the right side. The corneal diameter and ocular tension were normal.

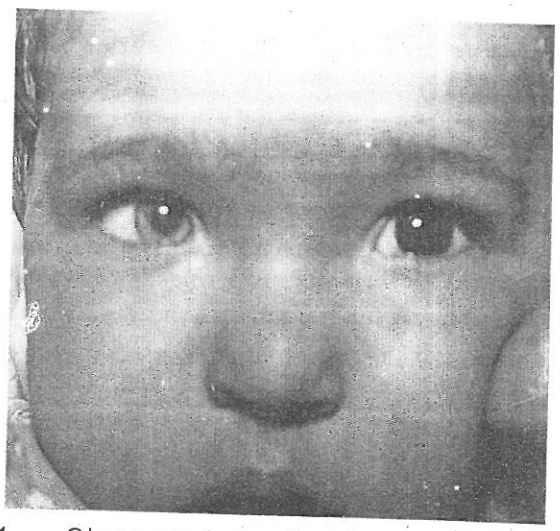

Fig.1. Close-up face showing heterochromia irides, confluence of eyebrows, hypertelorism and borad nasal root

In addition, she had an annular depigmented macule measuring 1 cm in diameter on the dorsum of the right middle finger with a central area of

From the Departments of Dermatology and Ophthalmology, L T M M College & L T M G Hospital, Bombay, India.

Address correspondence to: Dr Ashish Sukthandar B 58/554, M I G Conoly, Bandra East, Bombay-400 051.

moderate hyperpigmentation. Wood's lamp examination of the lesion indicated absence of pigment in the affected skin. Skin biopsy stained with heamatoxylin and eosin and Fontana-Masson stains revealed absence of melanocytes in the affected skin.

There was no clinical hearing loss as was confirmed on free field audiometry. Mental retardation, spinal, skeletal and gait defects were absent. No bowel incontinence was observed.

Comments

Waardenburg's syndrome type 1 is an autosomal dominantly inherited condition, with variable penetrance and expression. Isolated cases have also been reported.[1]

The white forelock is the most frequent cutaneous pigmentary abnormality with an incidence ranging from 17 to 58.4%.[4,5] However this feature was not present in our patient. White macules anatomically distinct from the white forelock are also a predominant feature with an estimated prevalence of 15%.[4,5] The patches of depigmentation resemble those of piebaldism, are present at birth, do not grow, and do not repigment. Hyperpigmented macules may be found within the amelanotic area. Depigmentation results from an absence of melanocytes in that area (melanocytopenic depigmentation) and this was confirmed in the present case with special stains.

Heterochromia irides may be partial or total and is found in 20% of reported cases.[4,5] In our patient albinotic fundus was seen on the affected side which is a known association.[4] Dystopia canthorum is characterized by an increase in the distance between the inner angles of the eyelids with normal distances between the pupils and the outer canthi.[1] The incidence of deafness ranges from 9 to 37.5%[5] which was absent in this patient.

Cases of piebaldism associated with heterchromia irides have been described.[6] As both abnormalities are features of Waardenburg's syndrome, the combination may represent a forme fruste of this syndrome. However the possibility of a

Table I. Clinical variants of Waardenburg's syndrome

	Type 1	Type 2	Klein's syndrome (Type 3)	Pseudo Waardenburg syndrome	WS-Ocular Albinism
White forelock	+	+	+		+
Depigmented macules	+	+	+		+
Heterochromia irides	+	+	+	Hetero / Isohypochromia	Hypoplastic Heterochromia
Deafness	+/-	+	Musculo skeletal abs	+	+
Confluent eybrows	+				
Dystopea canthorum	+	-		+	-
Broad nasal root	+			+	
Hirchsprung's	-	++			
Others				Congenital unilat. ptosis Light hair with scattered white hair on scalp	Prominent nasal root Premature graying Hyperopia-esotropia amblyopia

chance association or even a distinct genetic entity cannot be ruled out. Considering the presence of other characteristic facial features we propose this patient to be of Waardenburg's syndrome type 1.

References

1. Ortonne JP. Piebaldism, Waardnburg's syndrome, and related disorders: "Neural Crest Depigmentation Syndrome." Dermatologic Clinics. Vol 6 / Number 2, 1988: 205-16.
2. Waardenburg PJ. New syndrome combining developmental anomalies of eyelids, eyebrows and nose root with pigmentary defects of iris and head hair and with congential deafness. Am J Hum Genet 1951; 3: 195.
3. Pantke OA, Cohen MM Jr. The Waardenburg syndrome. Birth Defects 1971; 7: 147.
4. Goldberg MF. Waardenburg's syndrome with fundus and other anomalies. Arch Ophthalmol 1966; 76: 797-810.
5. Reed WB, Stone VM, Border E, Ziprkowski L: Pigmentary disorders in association with congenital deafness. Arch Dermatol 1967; 95: 176-86.
6. Comings DE, Odland GE: Partial albinism. JAMA 1966; 195: 519.