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WAARDENBURG'S SYNDROME

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Three children in a family of five presented with heterochromia iridis, lateral displacement of inner canthi and varying degrees of sensorineural deafness. All the 3 showed iris atrophy. The father of the children had only heterochromia iridis. A diagnosis of Waardenburg's syndrome Type I was made in the children with the father probably representing a forme fruste of the condition.

Key Words : Heterochromia iridis, Deafness, Waardenburg's syndrome

Introduction

Waardenburg's syndrome is an autosomal dominant disorder with variable penetrance characterised by lateral displacement of inner canthi and of lacrimal puncta, heterochromia iridis, white forelock, prominence of nasal root and of the medial eyebrows, congenital deafness, and hypomelanotic macules.

However, there are considerable differences between individual cases due to variable expressivity of the defective genes. Thus, several types of Waardenburg's syndrome occur.[1] Several associated features have also been reported and include EEG abnormalities, cleft lip and palate, high refractive errors and micro-ophthalmia.[2] We report the occurrence of Waardenburg's syndrome in 3 children in a family of 5.

Case Reports

The patients were aged 10, 5 and 3 years and were born of a non-consanguineous marriage. They presented with depigmented patches of skin on the upper and lower limbs since birth, lateral displacement of the inner canthi and varying degrees of sensorineural deafness.

Table I shows the clinical details. In the family, the eldest daughter aged 16 and the youngest aged 1½ years were unaffected.

Discussion

The above clinical features suggest two characteristics of Waardenburg's syndrome - the autosomal dominant type of transmission and the variable expressivity of the disease. In this family, one child had all the characteristics of Waardenburg's syndrome while the other two had only a few features. The father manifested only a forme fruste of the disease in the form of

Table I. Clinical details of 3 patients and the father

	Eldest patient	Second child	Third child	Father
Heterochromia iridis	+	+	+	+
Outward displacement of inner canthi	+	+	+	-
Hypopigmented patches	+	+	+	-
White forelock	+	-	-	-
Sensorineural deafness	bilateral	only right side	-	-
Iris atrophy	+	+	+	-

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heterochromia iridis. The fact that there were also 2 unaffected children suggested the variable penetrance.

The most constant feature (99%) of the syndrome is the lateral displacement of the medial canthi. This was seen in all the children. White forelock is found in 17 to 58.4% of patients.[3-5] In our cases, only the oldest boy showed this finding. Depigmentation is seen in 5 to 8.3% of cases.[3,5] It was observed in all our cases. The patches of depigmentation were present since birth. Heterochromia iridis may be total or partial and is found in about 2% of the reported cases.[5] This was seen in all our cases.

Deafness is seen in 2% of the cases.[2] In our report, the oldest affected child had bilateral complete sensorineural deafness. The next sibling had deafness in only 1 ear while the youngest was normal on audiological examination.

A unique feature in our cases was the presence of iris atrophy which has not been reported in earlier literature, though iris hypoplasia has been recorded earlier.[5] Even though iris atrophy and hypoplasia are both manifested as small iridi, in atrophy there is depigmentation with lacunae in the substance of the iris thus differentiating it from hypoplasia.

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