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DOI: http://dx.doi.org/10.1590/abd1806-4841.20153343

Abstract: Waardenburg syndrome is an inherited disease characterized by sensorineural hearing loss, pigmentation changes and minor facial malformations. It has four clinical variants. We report the case of a girl who, like her mother, was affected by this syndrome. The diagnosis was made after detection and treatment of deafness.
Keywords: Skin diseases, genetic; Genetic diseases, inborn; Deafness; Waardenburg syndrome.

CASE REPORT

A 12-year-old girl sought dermatological consultation regarding warts. She had a history of bilateral sensorineural deafness, first noticed at the age of 2 years, and received cochlear implant in right ear at the age of 5 years.

Physical examination revealed perioral warts. In addition, we noticed isohypochromia iridis, dystopia canthorum, hypertrichosis of the medial part of the eyebrows (synophrys), slight obliteration of the philtrum, and achromic patches on the back of the fingers of the right hand (Figure 1). We noted that the girl's mother had heterochromia of the left eye and achromic patches on the anterior aspect of the left forearm (Figure 2). Moreover, the mother reported early graying of her hair and that the patient's sister also had similar patches.

An ophthalmologic evaluation revealed diffuse thinning of the retinal pigment epithelium.

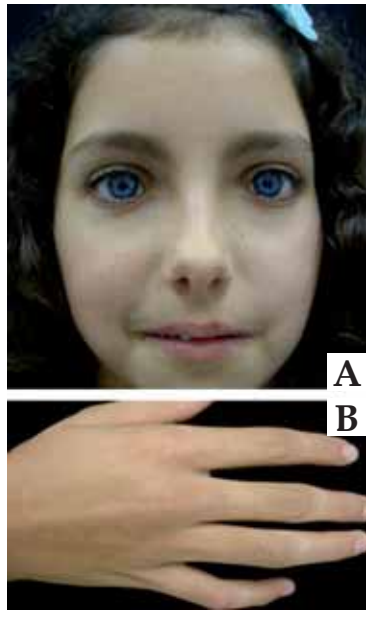

FIGURE 1: A. Isohypochromia iridum, canthorum dystopia, broad nasal bridge, hypertrichosis of the medial part of the eyebrows and obliteration of the philtrum. We can also see perioral keratotic papules compatible with viral warts. **B.** Achromic maculae and patches on the medial phalanges of the fingers of the right hand

FIGURE 2: Isohypochromia iridum in the patient (left) and heterochromia iridum in the patient's mother (right). We can see obliteration of the philtrum and a broad nasal root

Received on 25.12.2013.
Approved by the Advisory Board and accepted for publication on 10.02.2014.
* Study conducted at the Hospital de Clínicas de Porto Alegre - Federal University of Rio Grande do Sul (UFRGS) – Porto Alegre (RS), Brazil.
Financial Support: None.
Conflict of Interest: None.

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DISCUSSION

Waardenburg syndrome (WS) is an inherited disease with an estimated incidence of 1: 42,000 births. [1,2] It has 4 clinical variants, which share varying degrees of sensorineural hearing loss and changes in pigmentation.

In most cases, it is an autosomal dominant disorder caused by mutations in the PAX3 gene on chromosome 2q. These mutations result in changes in the differentiation, survival and migration of melanocytes derived from the neural crest in the embryonic period. [3-5] The great clinical variability of the syndrome is attributed to different penetrance and expression of the responsible genes. [6]

SW type 1 is the classical form. [2] It is characterized by varying degrees of hearing loss, defects in neural crest-derived structures and changes in pigmentation. [7]

Deafness, which is of sensorineural origin, may be unilateral or bilateral, total or partial. It occurs in approximately 20% of patients. [7] About 2% of all cases of congenital deafness are due to the SW. [1,4]

Dystopia canthorum (or telecanthus) is characterized by an increased distance between the inner corner of the eyelids, with normal distance between the pupils. It is a minor defect, the most common finding in SW type 1 (98% of cases) and the main feature that distinguishes it from other types of SW. [4,8,9] Other manifestations include broad nasal root, obliteration of the philtrum, synophrys (confluence of the eyebrows) and low anterior hairline.[3]

The main pigmentary changes of SW type 1 are heterochromia or isohypocromia iridum, poliosis and early whitening of the hair. In addition, there may be retinal pigmentary changes, areas of leukoderma and hyperchromic maculae.

Chart 1: Diagnostic criteria for Waardenburg syndrome type 1

MAJOR CRITERIA	MINOR CRITERIA
Hair hypopigmentation	Congenital leukoderma
Pigmentary change of the iris	Early graying of the hair
Dystopia canthorum	Synophrys
Sensorineural hearing loss	Broad nasal root
First-degree relative with WS	Hypoplasia of the nasal ala

Diagnosis is based on clinical findings (Chart 1). For diagnosis, 2 major criteria or 1 major criterium and 2 minor criteria must be present. [2,9]

There is no specific treatment for this syndrome. Early diagnosis enables the treatment of deafness. Dermatological findings are often unnoticed. However, they are essential for early diagnosis. Other than allowing the treatment of deafness, early diagnosis prevents permanent disabilities and is thus important for the development of the child.

The patient in our case had 4 major criteria and one minor criterium, which confirmed the diagnosis of SW type 1. Furthermore, the patient had received cochlear implant at an early age and showed improvement of hearing loss. ❑

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How to cite this article: Peruzzo J, Nazar FL, Tubone MQ, Escobar GF, Cestari TF. Waardenburg syndrome type 1. An Bras Dermatol. 2015;90(4):589-90.