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Waardenburg syndrome: rare cause of iretic heterochromia About 2 family cases

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ABSTRACT

Waardenburg syndrome is a rare congenital condition first described in 1951 by Waardenburg. It is characterized in its most typical form by the association of pigmentation disorders, internal canthal dystopia and enlargement of the base of the nose. We report two familial cases of waardenburg syndrome, diagnosed in a 6-year-old girl and her 6-month-old younger brother. Both had complete irreversible heterochromia, internal canthal dystopia and albinoide fundus. Waardenburg syndrome is a rare, hereditary entity whose ophthalmologic signs are internal canthal dystopia and pigmentation disorders such as iris heterochromia and albinoide fundus. Neurosensory deafness, musculoskeletal abnormalities and Hirschsprung's disease should be systematically sought

INTRODUCTION

Waardenburg syndrome (SW) is a rare genetic disorder [1], characterized by the association of facial morphological abnormalities, pigmentation abnormalities and deafness. Other abnormalities have been described allowing to distinguish several subtypes (1-4) of SW according to the manifestations encountered and their clinical expressivity [2].

There are five major criteria and five minor criteria for diagnosing SW. Major criteria include neuro-sensory deafness, canthus dystopia, irish heterochromia, frontal white lock, and evocative family history. The minor criteria include disorders of skin pigmentation, synophrosis, a prominent nose root, hypoplasia of the nose and canities.

The purpose of our work is to recall this rare entity of congenital irish heterochromia through two family cases in a 5-year-old girl and her 6-month-old brother.

MATERIAL AND METHODS

Observation 1

It is a girl of 6 years, resulting from a not consanguineous marriage, consulted for a diminution of the visual acuity with a heterochromie irienne noticed by its teacher.

The anamnesis has objectified a similar case in his little brother.

Ophthalmologic examination found a decrease in visual acuity improved after correction, a dystopiecanthal (Figure 1), an iretic heterochromia (Figure 2) and an albinoid fundus (Figure 3).

The general examination did not find a clinically obvious auditory disorder, no musculoskeletal or dermatological disorders.

The diagnosis of Waardenburg syndrome was based on the presence of at least 2 major signs: pigmentation abnormalities (iris heterochromia and albinoide fundus) and dystopiecanthal (intercanthal distance = 42 mm).

In order to look for subclinical hearing problems we asked for an audiogram which objectified a unilateral perception deafness.

Genetic confirmation based on the demonstration of the EDNRB gene on chromosomes 20 and 22 for type IV and MITF leads on chromosomes 3, 1 and 8 for type II, thus the search for mutation of the PAX 3 gene on chromosome 2 for types I and III; was not done for technical and economic reasons.

Figure 1: Dystopia of canthus

Figure 2: Iretic heterochromia

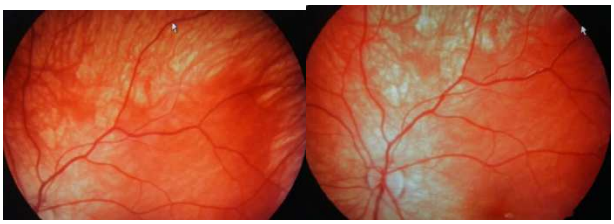

Figure 3: albinoide fundus

Observation 2

It is a boy of 6 months, having presented the same symptomatology as his sister with a heterochromia irian, a dystopiecanthale (figure 4) and a fundus albinoide, without auditory disturbance clinically detectable, nor dermatological.

The general examination was without particularity.

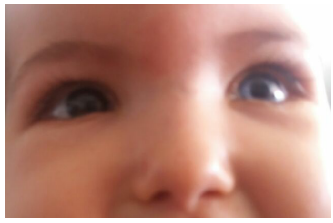

Figure 5: dystopiecanthal; ireric heterochromia

RESULT AND DISCUSSION

Waardenburg's syndrome is an autosomal dominant genetic disorder, characterized by oculo-dermato-auditory malformations with variable expressivity, associating in the most typical form an internal dystopiecanthal, an enlargement of the base of the nose, abnormalities of the pigmentation and sometimes neurosensory deafness [4]. This syndrome is reported for the first time in 1951 by Waardenburg [1]. There are 4 subtypes of SW depending on the clinical expressiveness.

Genetically, the first 3 subtypes of SW are of autosomal dominant inheritance whereas type IV is of autosomal recessive inheritance. Recent advances in genetic analysis and molecular biology have shown that types I and III are allelic variants while types II and IV are two distinct entities [2]. Several genes have been implicated in this syndrome [5]:

• The PAX 3 gene has been found in majorities of patients with SW types I and III.

• MITF gene damage for SW II

• Eradication of the EDN3 gene in the EDNRB receptor or the SOX 10 gene for SW IV.

Clinically, the clinical diagnostic SWest is based on the presence of major and minor criteria (already described). The Idu SW type is retained in the presence of at least two major signs or a single major sign associated with minus two minor signs. For SW type II, the diagnosis is made if the patient is carrying at least two major signs, without dystopiecanthal, premature discoloration of the hair becomes a major criterion of diagnosis. Type III is identical to type I with musculoskeletal involvement, the association with Hirschsprung's disease is suggestive of type IV [4,6].

In the 2 children, We have retained the diagnosis of SWsus-type I, given the presence of a dystopiecanthale and the absence of musculoskeletal disorders. skeletal, and Hirschsprung's disease. The presence of family history confirms the autosomal dominant inheritance of this type.

Other eye signs have been described in association with SW. For François, these signs are a coincidence. They are represented by microphthalmia, cataract, and strabismus. In their series, Delleman and Hageman estimate the frequency of strabismus converging at 19% [7]. However, our 2 patients did not show any other signs apart from a decrease in visual acuity in the 1st case related to a refractive disorder.

CONCLUSION

Waardenburg syndrome is an inheritance of the internal radicular hallucopia, and pigmentation abnormalities such as iris heterochromia and albinoid fundus represent the signs of theophyllmological. Neurosensory deafness, musculoskeletal disorders, as well as a Hirschsprung disease, must be systematically sought.

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