Clinical Image

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Waarendburg syndrome

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Clinical Image

A 5-year-old girl with hearing impairment was brought to the ophthalmology clinic by her parents for a very marked photophobia. On examination, the bright blue color of the iris was striking (Fig.1), the child had a large nasal base, synophris, a cantal dystopia with diffuse choroidal hypopigmentation in both eyes. The auditory evoked potentials revealed bilateral sensorineural hearing loss at 65 dB The diagnosis of Waardenburg syndrome was retained given the association of more than 2 major criteria: sensorineural deafness, canthi dystopia and depigmentation anomalies. The child received a hearing aid and speech therapy.

In 1947, Waardenburg, a Dutch ophthalmologist, identified for the first time an unknown autosomal dominant syndrome, now called Waardenburg syndrome type 1, which associates a dystopia of the internal canthi, a frontal white streak, an enlargement of the base of the nose, an internal brow hypertrichosis, an iris heterochromia and a Sensorineural hearing loss. The ocular involvement of Waardenburg syndrome is often limited to iris heterochromia and choroidal depigmentation. Other ophthalmologic abnormalities have been described in association with this syndrome, including cataract, retinal vein occlusion and strabismus, thus compromising the visual prognosis.

Genetic diagnosis consists in searching for mutations in PAX3 gene; the first 3 types of WS are autosomal dominant, while type 4 is autosomal recessive. Treatment consists of a hearing aid with speech rehabilitation, photoprotection of the eyes and skin is highly recommended; the surgical treatment for aesthetic purposes can be proposed for the telecanthus. Genetic and familial counselling is also a good tool for these syndromed patients.

***Figure 1:** Photograph showing internal canthal dystopia, hypertelorism, synophris, a wide base of the nose and a blue iris in a 5-year-old child with Waardenburg syndrome type 1.*

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