Usher Deafblindness

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Abstract — Pigmentary retinopathy refers to a group of inherited degenerative diseases of the retina, which primarily affects the photoreceptor cells in the retina. The association with congenital hearing loss defines Usher syndrome.

Usher syndrome is a rare pathology of autosomal recessive transmission with a double sensory impairment (auditory and visual). We report the observation of a 12-year-old patient from a consanguineous marriage with congenital deafness, normal vestibular function and pigmentary retinopathy composing type 2 of Usher syndrome.

Index Terms — Usher Syndrome, Pigmentary retinopathy, Congenital hearing loss.

I. INTRODUCTION

Pigmentary retinopathy refers to a group of inherited degenerative diseases of the retina, which primarily affects the photoreceptor cells in the retina. The association with congenital hearing loss defines Usher syndrome.

Usher syndrome is a rare pathology of autosomal recessive transmission with a double sensory impairment (auditory and visual).

II. CASE REPORT

We report the case of a 12-year-old female patient followed for congenital bilateral hearing loss with hearing aids from the age of 8 months. Only daughter of a consanguineous marriage. The patient presented for a consultation for a decrease in bilateral visual acuity with a night blindness.

The ophthalmological examination found visual acuity corrected to 20/20 in both eyes. The examination of the anterior segment was normal, the intraocular pressure was 12mmHg on the right and 11mmHg on the left.

The examination of the fundus found at the level of the two eyes: a papilla of normal aspect, retinal lesions giving a pepper and salt aspect more marked on the periphery testifying to a depigmentation of the pigment epithelium as well as a narrowing arteriolar (Fig. 1).

The auto fluorescence reveal hypofluorescent lesions at the periphery, associated with heterogeneous macular hypofluorescence (Fig. 2).

The examination was completed by a macular OCT showing a loss of the pigment epithelium as well as the ellipsoid zone outside the perifoveolar region at the level of the two eyes (Fig. 3).

The electroretinogram (ERG) objectified a significant impairment of the responses of the cones and rods with a few functional retinal cells in the two eyes confirming the retinopathy of the pigments (Fig. 4).

The neurological examination was normal, showing in particular no cognitive impairment, vestibular, cerebellar syndrome or proprioceptive ataxia.
Fig. 2. Autofluorescence photography objectifying a peripheral hypofluorescence associated with irregular central hypofluorescence: a) right eye; b) left eye.

Fig. 3. Image of OCT revealing a disappearance of the epithelium pigmentary and the ellipsoid zone outside the region perifoveolar: a) right eye; b) left eye.

Fig. 4 ERG traces revealing a malfunction of cones and sticks at both eyes.

III. DISCUSSION

The association of pigmentary retinopathy and congenital deafness of perception in a context of inbreeding suggests in our patient the diagnosis of Usher syndrome [1].

The presence of moderate congenital deafness with language acquisition, normal vestibular responses are in favor of type 2 of this syndrome. Namely, type 1 is characterized by the depth of hearing loss associated with vestibular disorders, thus preventing the acquisition of speech and walking [2].

The gene USH2A located on chromosome 1q41, and codes for Usherin, a transmembrane protein present in the basement membrane of photoreceptor cells and hair cells of the cochlea, and which has an important role in the development and homeostasis of the inner ear and retina [3].

The transmission of this rare syndrome is of an autosomal recessive type which can lead to the appearance of a sporadic case within a healthy family, hence the importance of advising the family on a possible recurrence of the syndrome in the siblings [4].

Pigmentary retinopathies constitute a group of conditions leading to an often severe visual handicap, with a progressive deterioration of visual acuity from the age of 10 years for type 2 [5], visual acuity can sometimes be
worsened by installing a macular hole. [6].

A recent study has shown the effectiveness and efficiency of a new treatment based on autologous stem cells of the bone marrow, by retrobulbar, subtenonian, intravitreal or intravenous injections, with a statistically significant increase in acuity visual and visual field in 80% of patients [7].

In addition to early care, especially in specific low vision equipment, psychological care has an important place in the therapeutic arsenal of this pathology that is both blinding and deafening [8].

An alternative treatment of USH2 based on a retinal implant type Argus II, has shown encouraging results and which can be a real hope for patients with Usher syndrome [9].

IV. CONCLUSION

Pigmentary retinopathies are a group of conditions often leading to a visual impairment. The association with a Congenital deafness defines Usher syndrome. The complexity of pathophysiological mechanisms and the genetic heterogeneity constitute a difficulty for a global approach therapy.

REFERENCES


