ANIRIDIA, CONGENITAL GLAUCOMA AND WHITE CORNEAS IN A NEWBORN BABY

ANIRIDIA, GLAUCOMA CONGÉNITO, CÓRNEAS OPACAS EN RECIÉN NACIDO

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ABSTRACT

Case report: We present the case of a newborn baby in whom we observed aniridia, congenital glaucoma and edematous corneas, without clearance of the cornea after glaucoma surgery, and in whom a keratoplasty was considered. The patient had no evidence of systemic diseases and no deletion of chromosome 11.

Discussion: It is important to perform a thorough ophthalmological and systemic exploration in newborn patients with aniridia due to other diseases that can be associated with it. We present the unusual case in which aniridia, congenital glaucoma and white corneas coexisted in a newborn baby. To our knowledge, such cases have only been reported twice before in the literature (Arch Soc Esp Oftalmol 2009; 84: 573-576).

Key words: Aniridia, glaucoma, cornea, newborn.

INTRODUCTION

Aniridia is an infrequent disease, usually bilateral, characterized by iris hypoplasia (1,2). The prevalence is of 1 case every 50,000-100,000 inhabitants (3).

Two-thirds of aniridia cases are due to familial inheritance, with dominant autosomic transmission and complete penetrance. The affected gene in familial aniridia is PAX6 in the short branch of chromosome 11 (1).

When performing a gonioscopy, a rudimentary iris is seen, even in the most extreme cases. In these patients it is usually frequent to have associated a foveal and optic nerve hypoplasia.
Progressively, it is also common in aniridia to see the appearance of cataracts, glaucoma and corneal opacity. Glaucoma usually develops with time due to the progressive closure of the angle as it is occluded by the rudimentary iris. The cornea can also progressively be affected and become opacified. It is rare to find glaucoma and corneal opacification at birth in aniridia patients (1,2).

**CLINICAL CASE**

**Personal antecedents**

A male patient, Caesarian birth (controlled pregnancy, no prenatal or peri-natal infection) at 36 weeks of gestation. The patient was referred to our centre due to suspected bilateral congenital glaucoma three days after birth.

**Ophthalmological exploration**

The ophthalmological exploration under general anesthesia revealed an intra-ocular pressure of 27 mmHg with Perkins tonometry in both eyes, a corneal diameter of 11.5 mm, a large corneal opacity and bilateral aniridia.

**Treatment**

A bilateral trabeculectomy was performed at 12, with triangular scleral flap and conjunctival flap based in the fornix. No anti-mitotics were utilized.

**Evolution**

At 15 days an ophthalmological exploration was carried out under sedation, which showed persistence of the corneal opacity (figs. 1 and 2) and an IOP of 17 mmHg in right eye and 14 mmHg in left eye.

It was considered to carry out a keratoplasty due to the persistence of corneal edema in both eyes.

**Genetic study**

The possible presence of genetic deletions or duplications was assessed with the MLPA technique (Multiples Ligation-dependent Probe Amplification), utilizing the SALSA P245 sound. This method allows the assessment of the doses of gene PAX 6 in chromosome region 11p13, the deletion of which causes aniridia and other ocular alterations.

All the sounds that were utilized exhibited normal relative intensity in the sample studied. Accordingly, the possibility of the patient exhibiting the deletion predisposing to the WAGR syndrome was discarded.

**DISCUSSION**

Aniridia is an infrequent disease, characterized by a hypoplastic iris which can be associated to other ocular anomalies such as optic nerve or foveal hypoplasia, cataracts or glaucoma. It can also be associated to systemic alterations such as anomalies in the genital-urinary apparatus, Wilms tumor and mental retard, giving rise to the so-called WAGR
Aniridia in a newborn baby

syndrome (Wilms tumor, aniridia, genital urinary apparatus alterations and mental retard).

Aniridia is a congenital disease which can express sporadically or due to familial heritage. The defective gene is PAX6, in charge of ocular development in chromosome 11p13 (4).

A bibliographic search in MEDLINE yields only 2 references which exhibit all the ophthalmological characteristics of our patient: aniridia, edematous corneas and newborn congenital glaucoma (3,5). Lee et al present a case having said characteristics in a patient with Brachmann-de Lange syndrome (Cornelia de Lange syndrome), which belongs to the group of syndromes with multiple congenital anomalies and mental retard. The patient was submitted to multiple operations: Ahmed valve and penetrating keratoplasties. At the age of five and a half, this patient achieved a transparent cornea in one eye and amblyopia in the contralateral eye due to a graft failure due to recurring infections (5).

Lise-Schneider B et al presented the case of two sisters, one of whom exhibited congenital glaucoma with corneal edema and bilateral aniridia without the PAX6 being affected, and the other exhibited congenital glaucoma with corneal edema but without aniridia (3).

In our case, the ocular tension remained controlled but this had no effect on the clearing of the cornea. Therefore, we believe the corneal edema is due to a corneal anomaly associated to aniridia.

In patients affected by aniridia, keratopathies usually appear not at birth but in late childhood. This situation is due to an insufficiency of the limbar stem cells, giving rise to the impossibility of corneal epithelial regeneration, causing corneal vascularization and it being covered by conjunctival epithelium (2).

The possibility of performing a keratoplasty is considered due to the central involvement of both corneas.

REFERENCE