Short communication

Hereditary glaucoma associated with oculodentodigital dysplasia

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ARTICLE INFO

Article history:
Received 16 September 2010
Accepted 24 April 2011
Available online 15 February 2012

Keywords:
Oculodentodigital syndrome
Glaucoma
Inheritance

ABSTRACT

Case report: A newborn evaluated at 20 days old due to occasional nystagmus. Her mother had presented with oculodentodigital dysplasia (ODDD) and glaucoma. The physical examination revealed opaque micro-corneas, and horizontal nystagmus. The tonometry showed 35 mm Hg in OD and 40 mm Hg in OS and the fundus examination was normal. She had a narrow nasal bridge with narrow nostrils, and fourth and fifth finger syndactylyl in both hands. A bilateral trabeculectomy was performed with a good response.

Discussion: ODDD is a rare autosomal dominant disease with heterogeneous phenotype manifestations. The most frequent cause of loss of visual acuity is the glaucoma, requiring long-term follow up with periodical control of the intraocular pressure (IOP).

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Glaucoma hereditario asociado a displasia oculodentodigital

RESUMEN

Caso clínico: Recién nacida de 20 días de vida con nistagmo ocasional, de madre con displasia oculodentodigital. En el examen físico se hallaban microcórneas veladas, nistagmo horizontal, tonometría de 35 en ojo derecho y 40 mm Hg en el izquierdo, fondo de ojo normal; pirámide nasal y narinas estrechas y sindactilia de los dedos cuarto y quinto de ambas manos. Buena respuesta a trabeculectomía bilateral.

Discusión: La displasia oculodentodigital es una enfermedad hereditaria con marcada heterogeneidad fenotípica. La causa más frecuente de pérdida de visión es el glaucoma, siendo necesario su diagnóstico temprano con un seguimiento continuo y controles periódicos de la presión intraocular.

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Introduction

Oculodentodigital dysplasia is an autosomal dominant hereditary disease that affects the normal development of the face, eyes, teeth and limbs, with a marked intra- and interfamilial phenotypical heterogeneity.

The most frequent ophthalmological expressions are microphthalmos and microcornea, and more rarely iridial anomalies, cataracts and secondary glaucoma; dental alterations include hypoplasia and yellowish color of teeth, with complete syndactyly of the fourth and fifth finger being the most characteristic malformation of limbs.

Clinic case

Newborn within term referred for ophthalmological assessment on day 20 of life due to the appearance of occasional nystagmus. Her mother, without relevant history during pregnancy, had been intervened for glaucoma at age 20 with bilateral trabeculectomy reconstructive hand surgery due to syndactyly (Figs. 1 and 2). The ophthalmological assessment evidenced microcorneas with discretely veiled appearance, occasional horizontal nystagmus, digital ocular tone bilaterally increased and ocular fundus (FO) with papilla having preserved characteristics. The general physical assessment revealed narrow nasal pyramid with narrowed nasal orifices and thin nasal wings, slight micrognatia and syndactyly in the 4th and 5th fingers of both hands. Under sedation, tonometry evidenced an intraocular pressure (IOP) of 35 and 40 mm Hg in the right and left eyes respectively. In addition, epithelial edema and superficial corneal clouding were observed.

A bilateral trabeculectomy was performed with good postop IOP control without the need of additional topical drugs. In the latest ophthalmological exploration at 14 months of life, the patient was in orthotropia, with good fixation and following light, refractive defect of −4 dioptres in both eyes, diffuse blebs and without inflammatory signs (Figs. 3 and 4), and adequate IOP control was confirmed. OF did not produce relevant alterations.

Discussion

Oculodentodigital dysplasia is an extremely rare hereditary disease that is mainly transmitted through a dominant autosomal pattern and exceptionally through recessive autosomal inheritance. It is caused by heterozygotic mutations in gene GJA1 located in chromosome 6q22-q23 and which codes the connexine 43 protein (Cx43), which seems to be a potent role in ocular development. Typically, it is characterized by the alteration of the normal development of face, eyes, teeth and limbs in addition to auditory and neurological
alterations although clinical expressions are highly variable, including among members of the same family.

The most frequent cause of vision loss is glaucoma, although the type, severity and age of presentation are variable. A number of mechanisms play a role in its presentation, including alterations in the development of the iridocorneal angle, gonioscopic changes similar to infantile glaucoma and chronic closed or open angle glaucoma associated to microcornea. The association of glaucoma with microcorneas during childhood is important as we do not always find the typical buftalamos. In addition, genetic alterations have been described that modify the normal anterior segment development, giving rise to trabeculodysgenesis which can express in the form of congenital glaucoma as in this case or during childhood.

Early diagnostic of this rare syndrome will allow us to identify the alterations that may require medical or surgical therapy such as congenital glaucoma which may give rise to poor vision and even blindness. In addition, it will enable multidisciplinary management of the other associated diseases in order to improve the quality of life of the patient.

In conclusion, it is necessary to bear in mind the possibility of glaucoma in this syndrome and the need of maintaining long-term follow-up with regular IOP controls due to the high variability in the presentation age.

**Conflict of interest**

The authors have no conflict of interests to declare.

**References**


