FAMILY CASE OF GRANULAR DYSTROPHY AND OCULOCUTANEOUS ALBINISM

ASOCIACIÓN DE ALBINISMO OCULOCUTÁNEO Y DISTROFIA GRANULAR EN UNA FAMILIA

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ABSTRACT

Clinical case: A 35-year-old female patient with blurred vision since childhood, for which no treatment had been given, presented with poor visual acuity. She had white skin and fair yellow hair. There were several well circumscribed deposits in the central and anterior corneal stroma, and iris transillumination and foveal hypoplasia were evident. The clinical diagnosis was oculo-cutaneous albinism and granular corneal dystrophy. We found oculo-cutaneous albinism in two brothers and granular dystrophy in three brothers, the mother and a son.

Discussion: Corneal dystrophy is an autosomal dominant disorder inherited independently of oculo-cutaneous albinism, which is inherited as an autosomal recessive condition. This is the first case report of granular dystrophy concurrent with oculo-cutaneous albinism (Arch Soc Esp Oftalmol 2006; 81: 289-292).

Key words: Oculocutaneous albinism, granular dystrophy.

RESUMEN

Caso clínico: Paciente femenino de 35 años de edad con mala agudeza visual desde la infancia. A la exploración se encontró baja agudeza visual, nistagmo, hipopigmentación de piel y cabello amarillo, córnea con depósitos blanquecinos en estroma central y anterior, transiluminación de iris e hipoplasia foveal. Se diagnosticaron albinismo oculocutáneo y distrofia corneal granular. Se encontró albinismo oculocutáneo en dos hermanos y distrofia granular en tres hermanos, la madre y el hijo.

Discusión: La distrofia corneal granular se transmite genéticamente siguiendo un patrón autosómico dominante e independiente del albinismo oculocutáneo. Este es el primer caso publicado de presentación concomitante de ambas entidades.

Palabras clave: Albinismo oculocutáneo, distrofia granular.

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INTRODUCTION

Granular dystrophy (GD) or Groenouw type I is a bilateral corneal pathology characterized by the presence of discrete and variable sized whitish opacities in the anterior and central stroma. It appears in the first or second decade of life. As the disease evolves, lesions increase in number and size and begin to coalesce; the stroma between opacities remains transparent but subsequently it assumes the appearance of sand polished glass. Visual alterations are infrequent before the fifth decade of life. However, some patients can exhibit epithelial erosions and increased corneal opacity with important reduction of visual acuity.

Albinism is a heterogeneous group of hereditary pathologies characterized by abnormalities in the synthesis of melamine associated to changes in the visual system development. Oculocutaneous albinism (OCA) involves eyes, skin and hair. The ophthalmological signs are variable and include nystagmus, hypo-pigmentation of the uveal tract and the pigmented epithelium of the retina, transillumination of the iris, foveal hypoplasia and abnormal decussation of the optic nerve fibers at the level of the chiasm (1).

This communication presents a case of simultaneous expression of GD and OCA.

CASE REPORT

A 35 year-old woman came to the practice complaining of poor visual acuity (VA) since childhood, which remained untreated. Upon exploration, yellowish hair was found on the scalp as well as on the brows and eyelashes, in addition reddish skin without nevus (fig. 1); visual acuity with 20/400 correction (−9.50 −5.50 x 135º) in right eye (RE) and 20/200 (−6.50 −5.50 x 20º) in left eye (LE); exotropy of 50 diopters, horizontal nystagmus; both eyes had IOP of 12 mmHg, normal conjunctiva, cornea with some well-defined central and anterior whitish stromal deposits (fig. 2), anterior chamber without cells or fla-re, clear iris with transillumination, transparent lens and foveal hypoplasia. With the ocular and systemic clinical data the OCA and GD diagnostic was establishes and the family was asked to undergo a clinical assessment.
The family comprises 9 individuals, with parents aged 74 and 64. No kinship was found and they come from different towns. GD is present in the mother, three brothers and the patient’s son, whereas OCA appears in two brothers but without GD data (fig. 3).

**DISCUSSION**

Granular dystrophy is the most common stromal dystrophy and is transmitted through dominant autosomic inheritance. It is produced due to a mutation in the big-h3 gene of the 5q31 chromosome, which encodes the keratoepitheline molecule.

In most cases, treatment only endeavors to correct the refractive defect. Some patients can exhibit recurring skin erosions which require the use of therapeutic contact lenses and artificial tears. When the visual acuity is highly affected surgery can be considered depending on the depth and extension of the lesions.

Penetrating keratoplasty treatment is performed in 4.1-37.7% of cases with a recurrence rate of up to 87% (2) which requires a second operation (3).

Phototherapeutic keratotomy has also been utilized for treating said dystrophy (4).

OCA is a group of diseases characterized by reduced melanine synthesis. If this synthesis is deficient in the skin, hair and eyes, the OCA term is utilized, whereas if it only occurs in the eyes it is defined as ocular albinism. There are at least 10 variants of OCA which have been classified in two categories according to the presence or absence of tyrosinase activity in melanocytes: tyrosinase-negative (OCA 1) and tyrosinase positive (OCA 2). The inheritance pattern is autosomic recessive. OCA 1 is the severest form of albinism which occurs due to alterations in the tyrosinase gene found in chromosome 11q14-21. In OCA 2 there is a production of feo-melamine and the mutation is localized in chromosome 15q11.2-q12 (5).

The data observed in the patient such as poor visual acuity, nystagmus, absence of nevus, low pigmentation of skin, hair and eyes point to a diagnostic of OCA 1. In turn, the clinical characteristics of the patient suggest that the poor visual acuity is due mainly to oculo-cutaneous albinism and therefore the treatment focuses at this time on correcting the refractive defect. As regards corneal dystrophy, the family data point to a dominant autosomic pattern independent of the albinism pathology. It is interesting to note the association of both pathologies in the same family because they are independent events and to date no relationship has been found between the genes involved and the known loci for these pathologies are in different chromosomes. As far as our knowledge goes, this is the first publication of concomitant presentation of both entities.

**REFERENCES**