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Juvenile Open-Angle Glaucoma: A Report of a Pedigree

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Key Words

Juvenile open-angle glaucoma
Inheritable glaucoma

Abstract

Six patients with juvenile open-angle glaucoma have been studied clinically and genetically in a family pedigree consisting of 17 members. This study revealed that juvenile open-angle glaucoma has an autosomal dominant mode of inheritance and the detected patients showed incipient to severe disturbances of visual function.

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Introduction

Juvenile open-angle glaucoma is a rare type of primary open-angle glaucoma different from congenital 'open-angle' glaucoma in the following important respects: juvenile open-angle glaucoma appears after infancy but before the age of 30 years and lacks the congenital abnormalities of the cornea and the anterior chamber angle [1-5].

In most of the cases, juvenile open-angle glaucoma has an autosomal dominant mode of inheritance [1-5].

Material and Methods

A pedigree with juvenile open-angle glaucoma has been studied clinically and genetically by the authors in Epirus, Greece.

The purpose of this epidemiological study was the detection of the patients, the observation of the disease course and the determination of the mode of transmission in this family pedigree.

A patient with a juvenile open-angle glaucoma happened to be referred to one of the authors. The family history of this patient disclosed the existence of more glaucomatic patients in the same family, who were his mother (1st generation) and his two brothers (2nd generation). We considered our patient as the 'propositus' and thus built, the genea-logic tree (fig. 1).

This study started 6 years ago, and since then all the genealogic tree members have been examined and followed closely. The first examination was done in their homeland, and subsequently all the patients were evaluated in our clinic. In the entire genealogic tree, there are 17 members (male: 10, female: 7), who belong to 4 generations; all come from Epirus, Greece. Twelve members out of 17 are related by blood (male: 8, female: 4). One member, who belonged to the 1st generation, died during this study, but he had been examined before his death. The only member who has not been examined yet is a 3-year-old boy of the 4th generation. All the members were asked to give a detailed medical history, and the

patients' examination included: assessment of the best corrected visual acuity, slitlamp evaluation, tonometry, gonioscopy, fundus examination and automatic perimetry (Humphrey).

Results

Six members out of 17 of the genealogic tree, being related by blood, have been found to suffer from bilateral juvenile open-angle glaucoma (male: 3, female: 3). According to the generation they belong to, the patients were classified as follows.

In the 1st generation 1 female patient was found. Glaucoma was first diagnosed at the age of 42 years and since then she has been under topical treatment with β -blockers and pilocarpine.

In the 2nd generation, 3 patients (brothers) were found. Glaucoma was first detected in the propositus at the age of 29 years and subsequently in his other two brothers at the age of 25 and 23 years respectively. The propositus underwent surgical treatment (filtering operation) in both eyes at the age of 30 years, and since then he has been under topical treatment with β -blockers. His brothers have not received

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any surgical treatment yet, following topical installations of β -blockers and pilocarpine.

In the 3rd generation, 2 female patients, daughters of the propositus, were suspected of having glaucoma. At first, they were found to have intraocular hypertension (mean intraocular pressure: 25 mm Hg) at the age of 20 and 17 years, respectively. Later, during the 6 years of this study, the older patient presented all the features of incipient glaucoma, and she is now under topical treatment, refusing surgical treatment. The younger of the sisters presents only intraocular hypertension without any other glaucoma findings and she has been without any treatment.

During the 6 years of follow-up, these 6 glaucomatous patients of the genealogic tree showed the following characteristics.

The female patient of the 1st and the propositus of the 2nd generation presented severe glaucomatous abnormalities in visual fields, and the optic disk-cupping ratio was 0.7 and 0.6, respectively. The other 2 patients of the 2nd generation showed moderate changes in visual fields and an optic disk-cupping ratio of 0.4-0.5. The older patient of the 3rd generation who presented the full expression of the disease showed incipient glaucomatous abnormalities in visual fields and an optic disk-cupping ratio of 0.4. The younger sister, till now, presents only intraocular hypertension without any other glaucoma findings. Intraocular pressures of the patients who are under topical instillation treatment vary within normal rates. Slitlamp examination and gonioscopy revealed no pathognomonic characteristics of congenital or secondary glaucoma.

Discussion

In the members of our genealogic tree from Epirus, Greece, the establishment of a primary open-angle glaucoma or the onset of intraocular hypertension at an age under 30 years, confirms that our patients present the juvenile

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Fig. 1. Juvenile open-angle glaucoma in

a family pedigree. □, O = Male, female, healthy; ■, · = male, female, patients; JZÍ = male, dead; .
 ■ = male, patient, propositus; I, II, III, IV = 1st, 2nd, 3rd, 4th generation.

open-angle glaucoma. The inheritable mode of transmission has been apparent in our cases since 6 out of the 12 members related by blood (50%) suffer from glaucoma.

We conclude that the juvenile open-angle glaucoma in this pedigree has an autosomal dominant mode of inheritance, since the 6 patients (male: 3, female: 3) were found to belong to three continuing generations (1st, 2nd, 3rd) and the patients of the 3rd generation are daughters of the same father, patient of the 2nd generation. Our findings agree with the current data in the literature [1-5].

According to the follow-up results of our genealogic tree, the insidious juvenile open-angle glaucoma presents a progressive course. For that, all the patients remain under close observation because a filtering operation will become necessary.

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