Autosomal dominant juvenile vitreoretinal degeneration and retinal detachment

K.M. Saari
Tampere, Finland

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Abstract

To study the inheritance and clinical picture of a new form of vitreoretinal dystrophy I examined 18 family members of a family with six generations. Seven patients, three male and four female, in three consecutive generations were observed to be affected indicating autosomal dominant inheritance. The disease was characterized by juvenile degeneration of the vitreous with detachment of the vitreous body and some floating vitreous opacities, cystoid degeneration of the peripheral retina with whitish glistening stippled areas of superficial retinal degeneration, spotty hyperpigmentation, patches of retinal atrophy with pigmentations, occasional atrophic retinal holes, and in four family members at the age of 4 to 12 years, unilateral or bilateral retinal detachment with breaks in the peripheral retina. Most patients had hyperopia with or without astigmatism. In eyes without detached retina, the disease did not show any marked progression, the lens was clear, the posterior fundus and the retina and choroidal vessels were normal, and the visual acuity, visual fields, dark adaptation, colour vision, electroretinograms, and visually evoked response findings were normal.

Vitreoretinal dystrophies are genetically determined degenerations of the vitreous body and the retina (5, 6). They include X-linked juvenile retinoschisis (4, 9, 15), Goldmann-Favres macrofibrillar vitreoretinal dystrophy (8), Wagner’s disease (25), clefting syndromes (7, 11, 20, 24), familial exudative vitreoretinopathy (3, 14), autosomal dominant vitreotapeto-retino-choroidal degeneration (26), autosomal dominant vitreoretinocchoriodopathy (19), lattice degeneration with and without myopia (12), snail-track degeneration (1), snowflake degeneration (16), and inherited high myopia (10).

In 1979, I initially treated a 10-year-old boy due to bilateral retinal detachment. During examination and follow-up studies of this patient and his family members, it became apparent that I was dealing with a specific and, to my knowledge, undescribed type of vitreoretinal dystrophy.

Material and methods

The inheritance pattern of the disease was examined by active family history and genealogical studies. I examined 18 family members of the last three generations of this family. The proband and his mother underwent a careful ophthalmological
examination and follow-up including Haag-Streit 900 slitlamp biomicroscopy, three-mirror contact lens examination, photography and fluorescein angiography of the fundus with a Canon CF60Z fundus camera, examination of the visual fields with the aid of a Goldmann perimeter using the kinetic method, noncorneal electroretinography and visually evoked response recordings, dark adaptation using the Goldmann-Weekers adaptometer, and examination of colour vision with the Ishihara test and desaturated Panel D-15. Sixteen relatives underwent a routine ophthalmological examination including slit-lamp biomicroscopy and a three-mirror contact lens examination. Affected family members underwent systemic general examinations that included skull and chest X-ray roentgenograms, and in children pediatric evaluation.

Results

Family study

The pedigree of the family with six generations is shown in Fig. 1. The affected family members were descended from a family living during the first half of the eighteenth hundreds in Kitee parish in eastern part of Finland (Fig. 1, I and II). Altogether 10 family members in five successive generations were affected including seven cases with verified disease and three other family members with reported blindness since early childhood. No consanguinity was known.

Clinical findings

No systemic manifestations were noted in any affected family member. None of the family members had a history of neonatal oxygen administration, facial bone hypoplasia, clefted palates, joint abnormalities, the Pierre Robin malformation complex, dwarfism or hearing loss. Skull and chest roentgenograms showed normal findings in all affected family members.

Table 1 shows the summary of ocular findings in the examined affected family members. Retinal detachment occurred in four family members (Cases 1–4) at the age of 4 to 12 years. All of them and three other blood relatives showed vitreous detachment with degenerative vitreous opacities, and degeneration of the peripheral retina.

Visual acuity was 1.0 or better in 7 eyes including the postoperative visual acuity of both eyes in a patient with bilateral retinal detachment (Table 1, Case 1). In two patients the retina was not reattached after electrocoagulation operations and these eyes remained blind (Case 2, LE; Case 3, LE). In one patient with retinal detachment and anisometropia the retina was reattached after ocular surgery but the eye developed amblyopia (Case 4, LE). One patient with anisometropia and amblyopia of the right eye since early childhood has visual acuity of 1.0 in the left eye at the age of 42 but at the age of 71 visual acuity of the left eye had decreased to 0.5 due to senile incipient cataract (Case 6). In his sister (Case 5) visual acuity was 1.0 without glasses at the age of 39 but at the age of 63 visual acuity had decreased to 0.6 in both eyes due to senile incipient cataract.

Refraction. All affected family members had hyperopia or emmetropia with or without astigmatism (Table 1). The refraction of the left eye of Cases 2 and 3 could not be determined due to total