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SHORT COMMUNICATION

Long-term follow-up in Bietti crystalline dystrophy

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PURPOSE. *To present a long-term follow-up of Bietti crystalline dystrophy.*
METHODS. *Two brothers are presented including the clinical findings, fluorescein angiography, electrophysiology (electroretinography [ERG], electrooculography [EOG], adaptometry), optical coherence tomography (OCT), and transmission electron microscopy of bulbar conjunctiva and peripheral blood lymphocytes. The clinical findings were documented over a period of 25 years in one brother and 5 years in the other.*
RESULTS. *The most striking features were deposits in the retina that were formed de novo with old ones replaced by choroidal atrophy in advanced stage of the disease. The light rise (EOG), rod- and cone-driven responses (ERG), and visual fields were affected progressively during the course. These changes of the retinal pigment epithelium and choriocapillaris were observed in the second decade and worsened gradually. OCT demonstrated preferential crystal accumulation in the inner retina. Cytoplasmic lipid crystalline inclusions were found in lymphocytes and conjunctival fibroblasts by transmission electron microscopy.*
CONCLUSIONS. *Bietti crystalline retinopathy is a progressive retinal disease characterized by retinal crystals gradually replaced by atrophy of the retinal pigment epithelium and gradual constriction of visual fields. (Eur J Ophthalmol 2007; 17: 680-2)*

KEY WORDS. *Bietti crystalline dystrophy, Crystalline retinopathy, Retinal crystals, Retinal dystrophy*

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INTRODUCTION

In 1937, Bietti (1-5) reported a tapetoretinal degeneration with associated corneal deposits at the limbus. The hallmark of the disease was crystals in the paracentral and peripapillary retina with or without marginal cornea. Bagolini and Ioli-Spada (1) presented a 30-year follow-up on one of Bietti's cases. We present long-term fundus photographic documentation (21 years) and clinical follow-up (24 years) of one of two brothers with Bietti crystalline dystrophy (BCD).

Case report

A 24-year-old man presented intraretinal refractile crystals (Fig. 1), paracentral scotomas, abnormal electrooculogram (light peak-to-dark trough ratio of 1.3 right eye and 1.4 left eye), and no corneal crystals. He complained of decreased night vision at age 17. He was examined at age 21 elsewhere and reported as having BCD (4). Then, he had a generalized disturbance of the retinal pigment epithelium by fluorescein angiography, normal color vision, normal electroretinogram scotopic responses, de-

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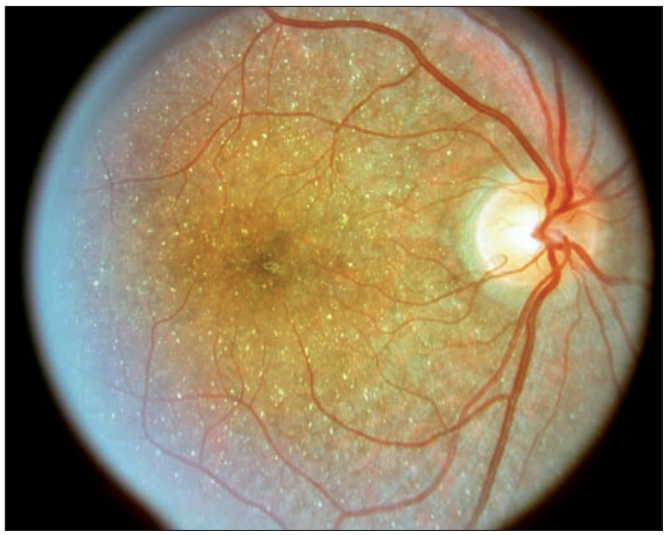

Fig. 1 - *Color photograph of the posterior pole of the right eye (March 1983): tiny refractile yellowish crystals were noted throughout the posterior pole (hypofluorescent on fluorescein angiography) with absence of corneal crystals and with 6/6 Snellen visual acuity.*

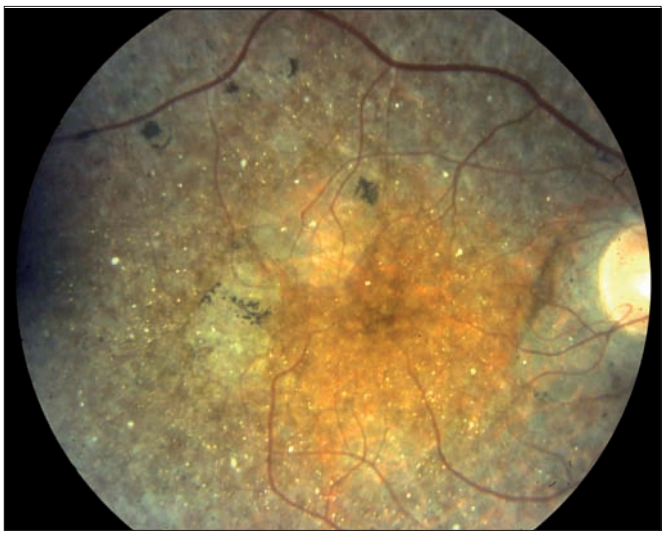

Fig. 2 - *Color photograph of the posterior pole of the right eye (February 2004) revealed scarcity of retinal crystals in areas of chorioretinal atrophy. The crystals were located mainly in the inner retina by optical coherence tomography.*

creased photopic amplitudes, subnormal dark adaptation, abnormal electrooculogram, and concentrically constricted visual fields (4). Both the electroretinogram (decreased photopic and scotopic responses) and the color tests (error score of 292 on Farnsworth-Munsell 100-hue [abnormal if above 100], deutan axis on panel D-15) became abnormal at age 29. At age 45, he maintained 6/6 visual acuities and developed bilaterally perifoveal chorioretinal atrophy (Figs. 2, 3) with central 10º visual fields. By optical coherence tomography (OCT), most of the crystals were located in the inner retina, were moderately reflective, were small, and did not cast any shadow on the outer retina. There were minimal cystic changes in the fovea with normal retinal thickness. His brother was diagnosed with BCD at age 16 and developed abnormal electroretinogram (decreased scotopic and photopic amplitudes) at age 26, with normal color tests (panel D-15 and Farnsworth-Munsell 100-hue). By transmission electron microscopy, both lymphocytes and conjunctival fibroblasts showed cytoplasmic lipid-crystalline complex inclusions.

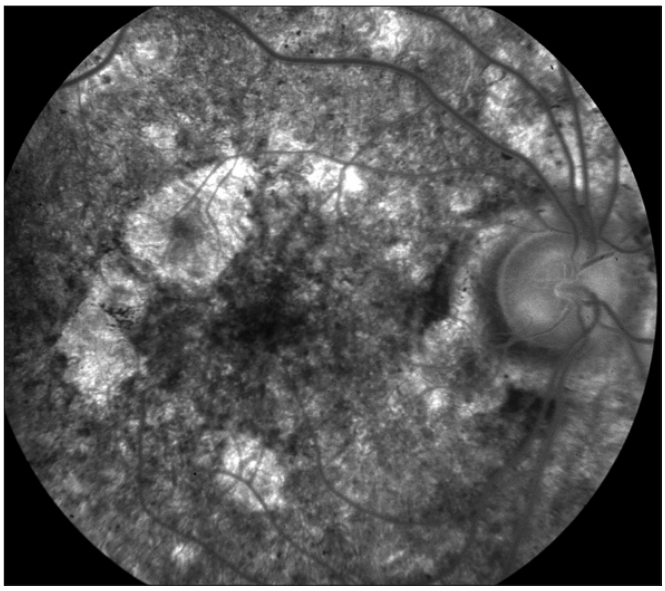

Fig. 3 - *Late fluorescein transit of the posterior pole of the right eye (February 2004) revealed focal perifoveal chorioretinal atrophy. Snellen visual acuity was 6/6.*

DISCUSSION

BCD is caused by mutations of the *CYP4V2* gene, a novel family member of the cytochrome P450 genes on chromosome 4q35. The crystalloid lysosomal inclusions resembling cholesterol esters have been detected in peripheral lymphocytes, skin, conjunctival, corneal, and choroidal fibroblasts. OCT demonstrated preferential crystal accumulation in the inner retina.

These crystals commonly disappear and are replaced by areas of chorioretinal atrophy.

There are three cases having long-term photographic documentation of 30 years (3), 16 years (2), and 5 years (5). In our case, crystals were apparent in the second decade with mild functional deficit. In the fourth decade, functional impairment becomes severe with marked chorioretinal degeneration.

Proprietary interest: None.

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REFERENCES

1. Bagolini B, Ioli-Spada G. Bietti's tapetoretinal degeneration with marginal corneal dystrophy. Am J Ophthalmol 1868; 65: 53-60.
2. Bernauer W, Daicker B. Bietti's corneal-retinal dystrophy. A16-year progression. Retina 1992; 12: 18-20.
3. Jurklies B, Jurklies C, Schmidt U, et al. Kristalline korneoretinale Dystrophie (Bietti)-Langzeitverlauf von einem Patienten von 30 Jahren und interindividuelle Variabilitat klinischer und elektrophysiologischer Befunde von zwei Patienten. Bietti's crystalline dystrophy of the retina and cornea. Klin Monatsbl Augenheilkd 2001; 218: 562-9.
4. Mauldin WM, O'Connor PS. Crystalline retinopathy (Bietti's tapetoretinal degeneration without marginal corneal dystrophy). Am J Ophthalmol 1981; 92: 640-6.
5. Sanchez Vicente JL, Herrador Montiel A, Diez-Garretas C, Guiote Linares JR. Distrofia cristalina de Bietti. Progresion durante 5 anos. Arch Soc Esp Oftalmol 2001; 76: 323-6.