Long term follow up of Bietti's Crystalline Dystrophy with Multi Modal Imaging

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Bietti crystalline dystrophy is a rare autosomal recessive disease with chorio-retinal degeneration characterized by the presence of yellow-white crystals in the retina[1], The patient presents with reduced visual acuity, poor night vision and visual field loss with onset of disease during the second or third decade of life. The crystalline deposits have been observed to diminish or even disappear in areas of severe chorio-retinal atrophy, as the disease progresses to later stages[2,3].

Fluorescein angiography reveals patchy hypofluorescent areas of retinal pigment epithelium (RPE) atrophy. Optical coherence tomography (OCT) showing degeneration is most prominent in the outer retina, including the photoreceptor layer. In addition to the crystalline deposits, other reflective spots like outer retinal tubulation (ORT) can be seen on OCT[4,5]. Visual field loss may manifest in different individuals as peripheral field loss (ring, paracentral, or central scotoma) or central or pericentral scotomas.

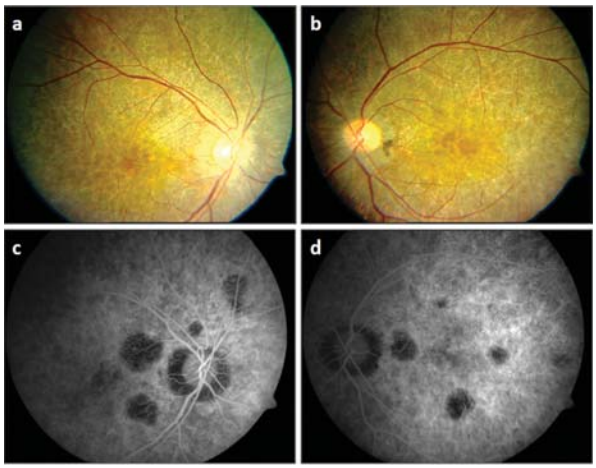

Figure 1: *Fundus photo shows numerous small white-yellowish crystals on the posterior pole and extending till mid periphery (1a and 1b); FFA reveals patchy hypofluorescent areas of RPE and choriocapillaries atrophy with generalized disturbance of RPE (1c and 1d).*

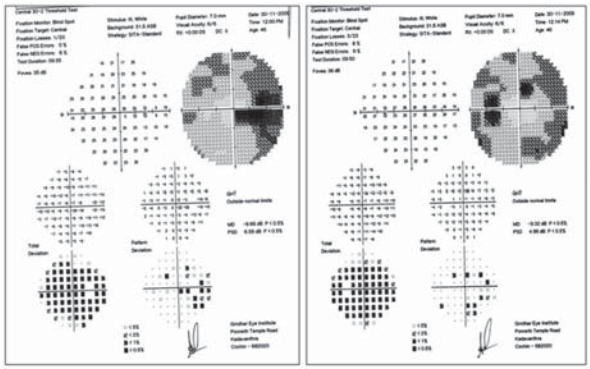

Figure 2: *Humphrey perimetry shows bilateral paracentral scotoma with enlarged blind spot on 30-2.*

CASE

The authors present one of the case where a 43 year old female presented with history of night blindness and best corrected visual acuity of 6/6 N6. Fundus examinations of each eye showed numerous small white-yellowish crystals on the posterior pole (Figure 1a and 1b). Fundus fluorescein angiography (FFA) confirmed posterior pole atrophy of the RPE and the underlying choriocapillaris (Figure 1c and 1d). Sparing of the peripheral retina was also noticeable on FFA. There were no important changes in the major retinal vessels or in the optic disc. Visual field revealed bilateral paracentral scotoma (Figure 2). Based on multi modal imaging patient was diagnosed of Bietti crystalline dystrophy.

On long term follow up of eight years there was disappearance of crystals on fundus examination (Figure 3a and 3c). Fundus auto fluorescence (FAF) shows hypoautofluorescence corresponding to area of RPE atrophy with a hyperautofluorescent edge representing the lipofuscin-laden cells that may atrophy imminently shown in (Figure 3b and 3d). OCT shows loss of outer retinal layers with presence of ORT and crystals located on the RPE-Bruch's membrane

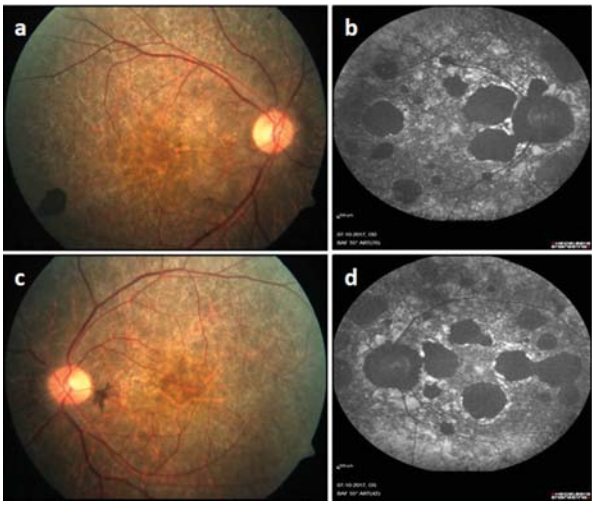

Figure 3: *Fundus photo shows disappearance of crystals on long term follow up of eight years (1a and 1c); FAF shows hypoautofluorescence corresponding to area of RPE atrophy with a hyperautofluorescent edge (1b and 1d).*

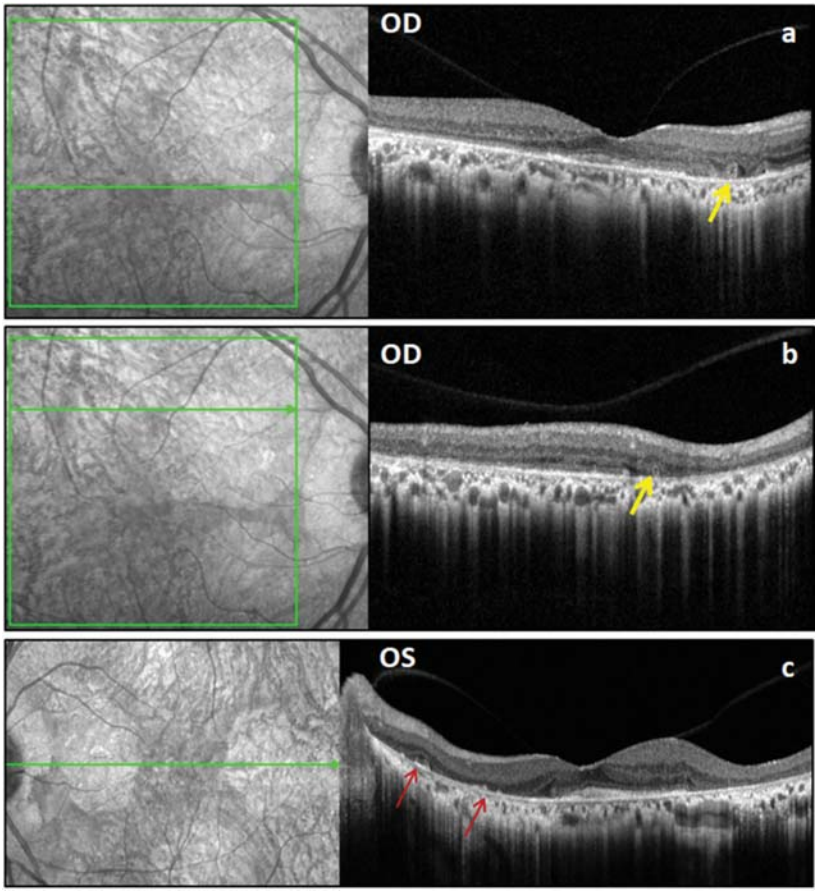

Figure 4: *SD-OCT shows loss of outer retinal layers with presence of ORT (yellow arrow) and crystals located on the RPE-Bruch's membrane complex (red arrow).*

complex (Figure 4). Cornea was not involved in our case.

COMMENT

This case is unique as it shows the long term follow up of Bietti crystalline dystrophy with disappearance of crystals as it has been reported previously that as the severity of disease progressed crystals deposits disappeared as the number of formation of crystals lag behind the number of formation.

REFERENCES

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