Abstract: We report the clinical and spectral domain optical coherence tomography findings in a rare case of Bietti crystalline dystrophy. A 23 year old male patient presented with defective vision, night blindness and paracentral scotoma for a duration of 9 months. Fundus examination showed characteristic yellowish white crystals scattered over the posterior pole with areas of patchy retinal pigment epithelial degeneration. The typical age of onset during 2nd or 3rd decade of life, progressive visual impairment, night blindness, paracentral scotoma, asymmetry of presentation between the two eyes were suggestive of Bietti's dystrophy. Spectral domain optical coherence tomography (SD-OCT) is of value in diagnosis and during follow up. SD-OCT showed deposition of crystals in the retinal pigment epithelium associated with atrophy.

Keyword: crystalline retinopathy, night blindness, SD-OCT

INTRODUCTION: Crystalline corneo-retinal dystrophy is a very rare autosomal recessive condition first reported by Bietti in 1937, as a corneo-retinal degeneration. The disease usually occurs in the 2nd or 3rd decade of life and is characterised by the presence of yellow white crystals in the retina, atrophy of retinal pigment epithelium (RPE) and choroid, and sparkling yellow cysts in the superficial paralimbal cornea. The characteristic retinal crystals are observed in all cases but the associated corneal dystrophy is found to occur in one-third to one-fourth of the patients. Bietti's crystalline retinopathy has been reported from different parts of the world, though majority are of East Asian origin.

CASE REPORT: A 23 year old male patient presented to us with complaints of defective vision in both his eyes for the past 9 months for which he had been prescribed glasses. Further deterioration of vision and diminished brightness was noticed in the left eye for the past one month. A history of night blindness and metamorphopsia was present. No similar complaints in parents or siblings. On presentation, the best corrected visual acuity (BCVA) was 6/6p in the right eye and 6/9p in the left eye. Slit lamp examination of the anterior segment showed clear corneas in both eyes. Fundus examination showed yellowish white refractile crystals scattered all over the posterior pole with patchy RPE degeneration in both eyes. The disc and vessels were normal. (figure 1 and 2)

FUNDUS OF RIGHT EYE  FUNDUS OF LEFT EYE

Colour fundus photographs of right and left eyes shows glistening yellow white crystals scattered throughout the posterior pole, patchy areas of RPE atrophy. Subretinal scars seen in left eye. (fig 1,2)

Central visual field testing using Bjerrum's screen showed paracentral scotomas in the left eye. The colour vision tested with Ishihara's pseudoisochromatic plates were normal. Multifocal electroretinogram (mf ERG) testing was done which showed diminished responses to scotopic and photopic stimulation. Spectral domain Optical Coherence Tomography (SD-OCT) revealed juxta and parafoveal hyper reflective dots, irregular RPE with inner segment/outer segment disruption in both eyes. Parafoveal RPE atrophy was seen in left eye. (figure 3 and 4)

As there are no specific treatments available, the patient was prescribed glasses with best correction and was asked to come for a regular follow up to detect disease progression. His parents and siblings were also asked to come for fundus examination.

DISCUSSION: The characteristic crystalline deposits over the posterior pole associated with atrophy of RPE is a distinctive clinical feature of Bietti's dystrophy in our case. Fluorescein angiography reports and histopathologic studies have revealed panchoroidal atrophy, sclerosis of choriocapillaries and severe retinal degeneration. Crystals and lipid inclusions were detected within lysosomes of choroidal fibroblasts.
biopsy specimens have demonstrated cholesterol like crystals and lipid inclusions in the corneal and conjunctival fibroblasts. Similar inclusions were also seen in circulating lymphocytes, suggesting an abnormality of lipid metabolism as a cause of Bietti’s dystrophy. However, biochemical studies have failed to characterise these deposits.

Visual field defects correspond to areas of RPE and choriocapillary atrophy, manifesting as paracentral scotoma or peripheral field loss depending on the severity of disease. The full field ERG and multifocal ERG testing shows decreases in amplitude of scotopic and photopic responses in all patients. The SD-OCT is of value for both diagnosis and prognosis of disease progression. Deposition of crystals in RPE – choriocapillaries complex and degeneration of outer retina have been reported in OCT studies. Potential retinal complications such as choroidal neovascular membrane (CNVM) and macular hole can be detected using OCT. The clinical symptoms and findings of electrophysiological testing in Bietti’s dystrophy are similar to those of other forms of retinal degeneration that fall under the category of Retinitis pigmentosa and allied disorders. The differential diagnosis of crystalline retinopathy are: Primary hyperoxaluria type 1 and 2, cystinosis, Sjogren – Larsson syndrome, drug toxicity (tamoxifen use, canthaxanthine use), talc retinopathy, CYP4V2 (mapped to chr 4q35) is the only gene in which mutations are known to cause Bietti’s crystalline dystrophy. Molecular genetic testing showing mutations in CYP4V2 gene have been reported in a high percentage of patients with this condition. In our case, the age of onset of presenting symptoms, progressive visual impairment, night blindness, paracentral scotoma and asymmetry of presentation between the two eyes were suggestive of Bietti’s dystrophy.

REFERENCES: