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Case Report

Sparkle in the eye: A rare case of Bietti's crystalline dystrophy

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ABSTRACT

Background: Bietti's Crystalline Dystrophy is an uncommon autosomal recessive tapetoretinal dystrophy resulting from a mutation in the CYP4V2 gene. It is defined by the existence of sub-epithelial corneal deposits at the limbus and small sized, crystalline looking deposits in the retina, especially at the posterior pole. This is bolstered by retinal pigment epithelial atrophy and the choriocapillaris.

Case Report: In this report, we present the case of a 55-year-old male exhibiting reduced visual acuity, nyctalopia, impaired color vision, and yellow-white crystal deposition in the posterior pole of both eyes. Additionally, areas of retinal pigment epithelium atrophy and pigment clumping were observed. Spectral-domain optical coherence tomography (SD-OCT) of the macula revealed hyperreflective dots in the outer retinal layers, retinal pigment epithelium, and choroid, along with outer retinal tubulations. Electroretinography (ERG) results were subnormal, and visual field assessment indicated paracentral scotomas in both eyes. The patient has been prescribed spectacles and is under regular follow-up.

Conclusion: This case is highlighted due to its rarity in the lack of genetic association and underscores the significance of employing SD-OCT and electrophysiological studies for early disease diagnosis. Additionally, it emphasizes the role of genetic counselling in preventing the inheritance of this condition.

1. Introduction

A rare form of tapetoretinal dystrophy, Bietti's Crystalline Dystrophy is characterized by the deposition of yellow-white crystals in the cornea and retina.[1,2] This autosomal recessive disorder is mainly associated with changes in CYP4V2 gene[3,4] that normally helps in the metabolism of lipids in retina.[5] The end result of this disease is retinal tissue degeneration and choroidal vessel sclerosis leading to a variety of visual impairments including blurred vision, difficulty adapting to darkness, decreased ability to see different colours and reduced field of vision.[6]

Although BCD is a rare condition, it has a significant impact on vision and quality of life. Affected individuals usually suffer progressive visual deterioration that starts during early adulthood. The characteristic presence of crystals on the retina[7] and cornea greatly aids diagnosis for patients showing these symptoms. Nevertheless, there are many variations observed within its clinical manifestations and progression thus necessitating complete ophthalmic assessment involving fundus examination, optical coherence tomography (OCT), as well as electroretinography (ERG).The management strategies for BCD remain largely supportive encompassing visual rehabilitation and adaptive techniques for optimizing residual sight. Genetic counselling forms an essential part if one wants to avoid having children with this condition or

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its carrier status.

2. Case Report

A 55-year-old male presented at our outpatient department with a gradual onset of painless bilateral vision loss over the preceding 2 years. He reported experiencing night blindness for the past year. Notably, the patient had a 20-year history of alcoholism and was undergoing treatment for Depressive order with Tablet. Olanzapine 5 mg HS for the last 6 months. There were no reports of consanguinity in his parents, and no similar complaints existed among other family members. Best corrected visual acuity was measured at 6/12 in the right eye and 6/18 in the left eye, with near vision measured at N12 in both eyes. Anterior segment examination revealed Grade 1 nuclear sclerosis in both eyes, with a clear cornea lacking evidence of crystal deposition (Figures 1 and 2).

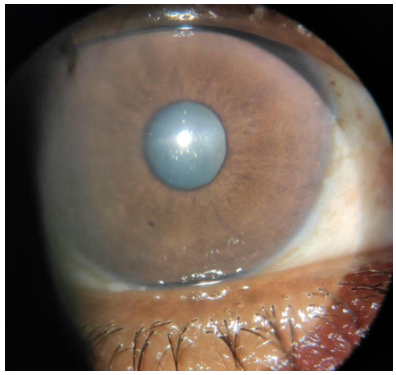

Figure 1: Anterior segment image of the right eye

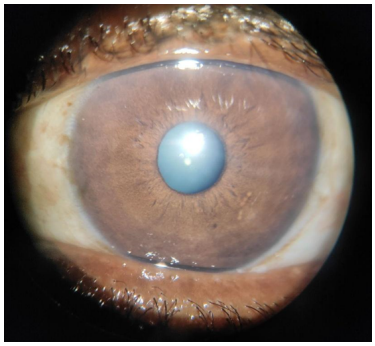

Figure 2: Anterior segment image of the right eye

Degeneration of the retinal pigment epithelium was noted in both eyes, and fundus examination revealed numerous sparkling crystals of yellow-white color dispersed across the posterior pole (Figures 3, 4, 5 and 6). Colour vision assessment revealed defects in both eyes (1/18 according to Ishihara's pseudo chromatic chart), while Automated Perimetry displayed few absolute scotomas in the nasal field bilaterally (Figures 7 and 8).

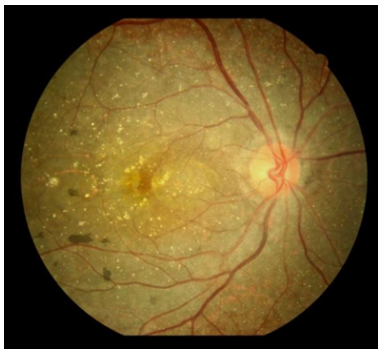

Figure 3: Fundus photograph of right eye showing yellow white crystalline deposits

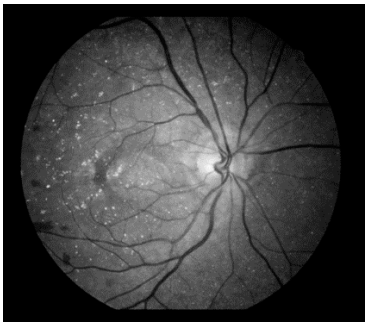

Figure 4: Fundus autofluorescence photograph of right eye

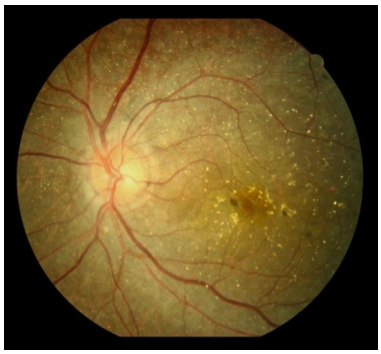

Figure 5: Fundus photograph of left eye showing yellow white crystalline deposits

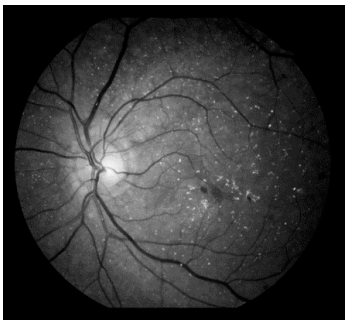

Figure 6: Fundus autofluorescence photograph of left eye

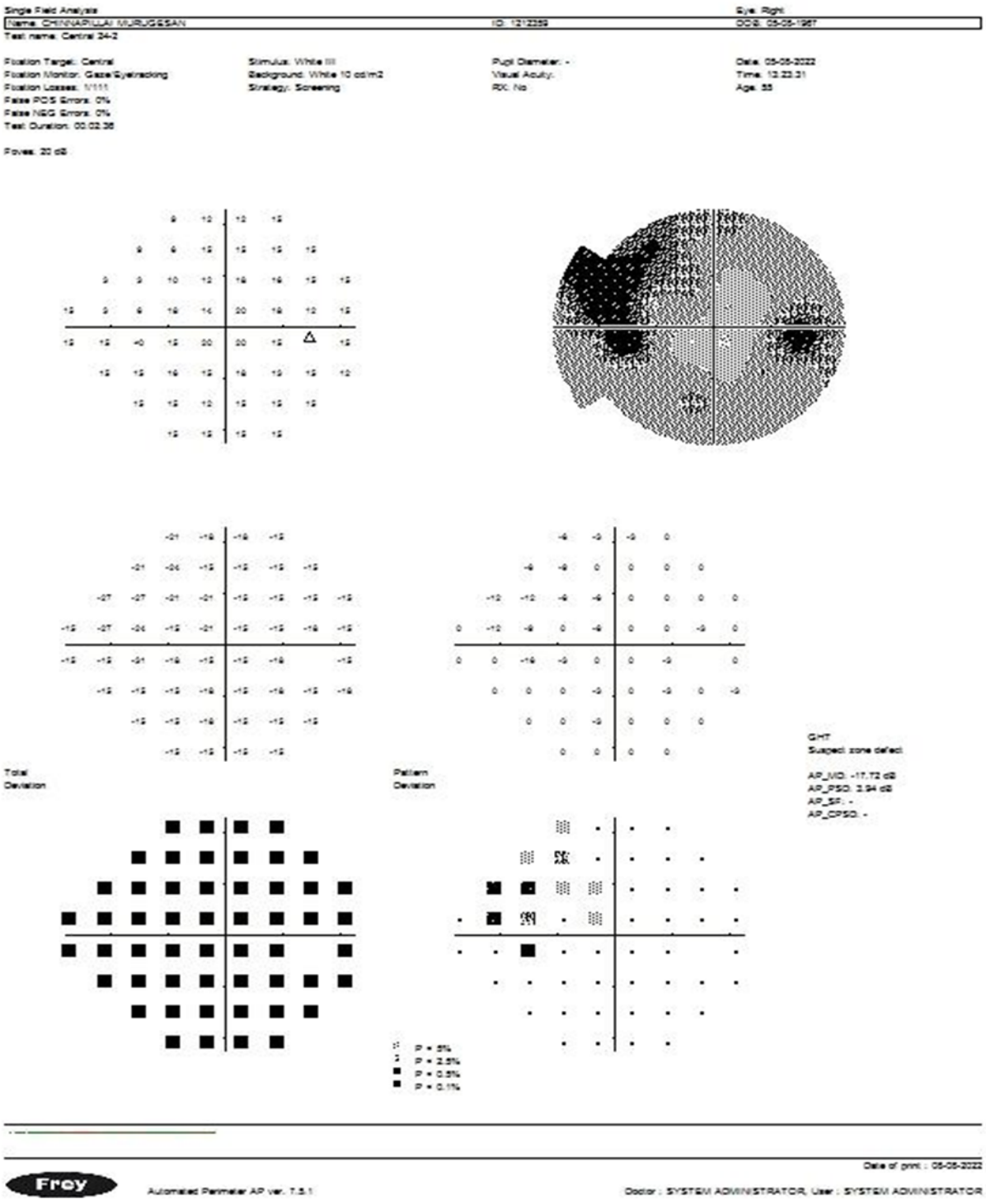

Figure 7: Automated perimetry right eye

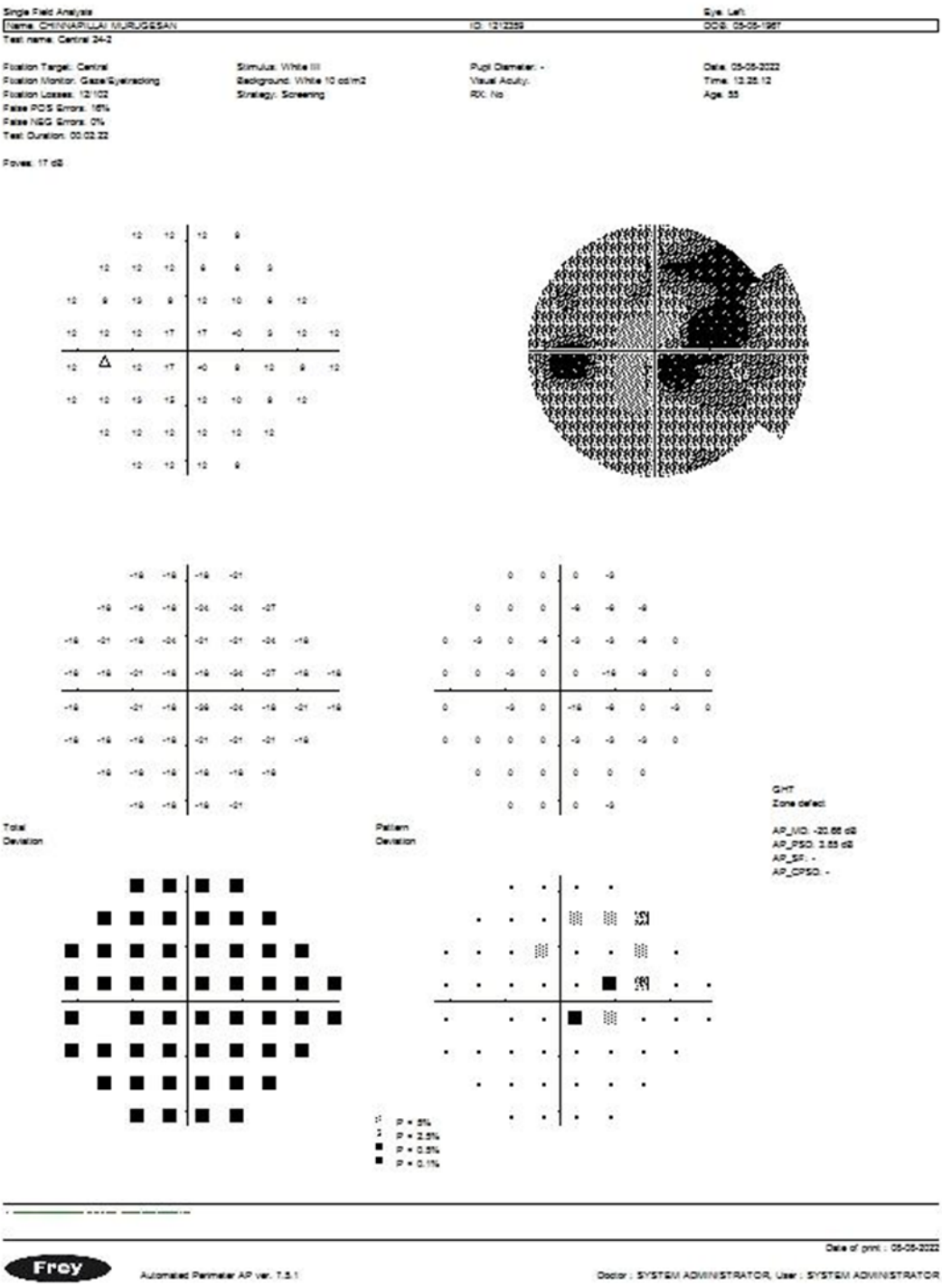

Figure 8: Automated perimetry left eye

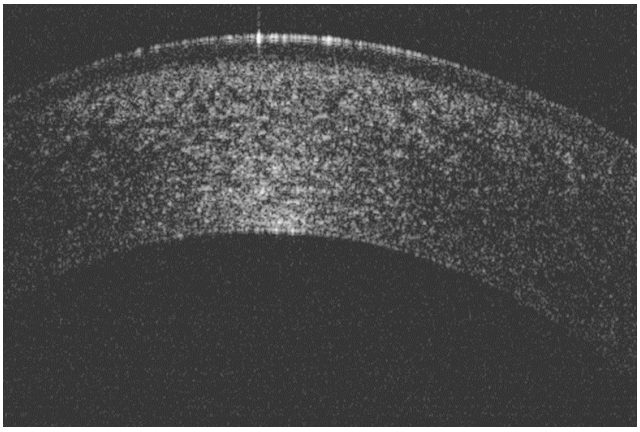

Figure 9: Anterior segment OCT right eye

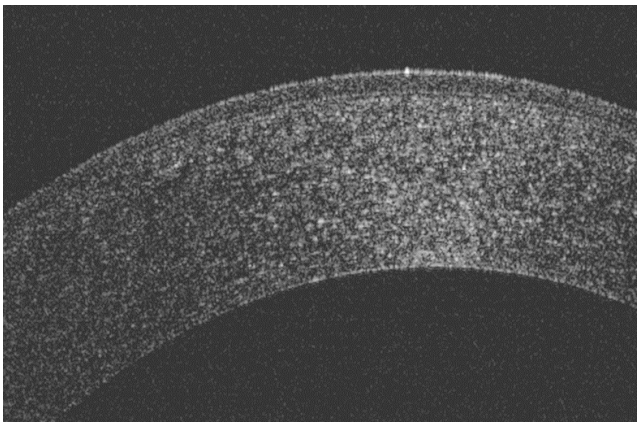

Figure 10: Anterior segment OCT left eye

A Spectral-Domain Optical Coherence Tomography (SD-OCT) scan of the macula revealed numerous hyperreflective dot-like lesions in the outer retinal tubulations, choroid, and neurosensory retina.[8] (Figures 11 and 12). Electroretinography (ERG) demonstrated reduced scotopic and photopic responses bilaterally (Figure 13).

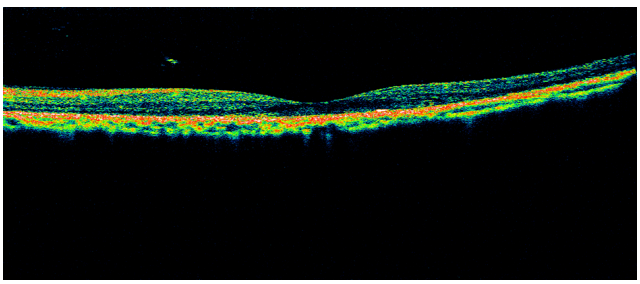

Figure 11: SD OCT right eye

Systemic investigations, including complete blood counts, random blood sugar, lipid profile, renal function tests, liver function tests, and urine analysis, yielded normal results. The patient was prescribed spectacles and low vision aids, with regular follow-up scheduled.

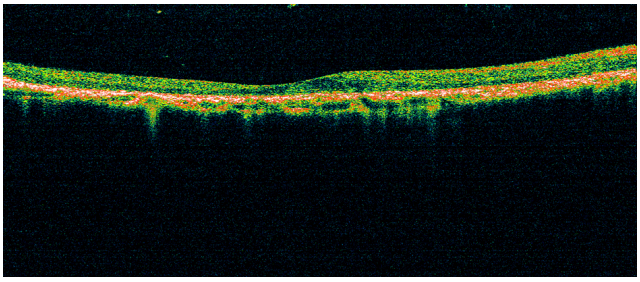

Figure 12: SD OCT right eye

3. Discussion

Bietti crystal dystrophy (BCD) poses special problems in its diagnosis and treatment due to its rare and complex pathology.[9] Our report covers all aspects of BCD, including its clinical manifestations, genetic basis, diagnosis, and treatment.

Two subtypes of Bietti's crystalline dystrophy have been proposed: a localised type with findings confined to the posterior pole and a diffuse type with widespread retinal findings.[10] Visual field examination may reveal paracentral scotomas that may progress to loss of peripheral vision in later stages. Colour vision abnormalities are common.

Clinical manifestations: BCD usually occurs in people of Asian descent between the ages of 20 and 40 years. It is characterized by three symptoms: decreased color vision, night blindness, and reduction in visual acuity.[11] These symptoms are often progressive and lead to progressive vision loss over time. Hallmarks of BCD include bright yellow-white crystals in the retina with pigment changes and corneal subepithelial deposits. These diseases also include colour vision abnormalities and blindness, leading to functional impairment.

Genetic basis: Pathogenic mutations in the CYP4V2 gene, which is involved in lipid metabolism,[10,12] constitute the etiological basis of BCD. Histological analysis of bone marrow provides insight into the lipid dysregulation associated with crystal-like cholesterol or cholesterol ester deposits seen in BCD. Understanding the genetics of BCD is important for future accurate diagnosis, genetic counselling, and therapeutic interventions.

3.1. Diagnostic approach

An optimal approach is essential to accurately identify BCD. Light bio-microscopy can be performed. Imaging studies like fluorescein angiography (FFA) and optical coherence tomography (OCT)[13] can evaluate the extent of the illness and track its development. Electrophysiological studies, including electroretinography (ERG) and electrooculography (EOG) provide insight into ocular

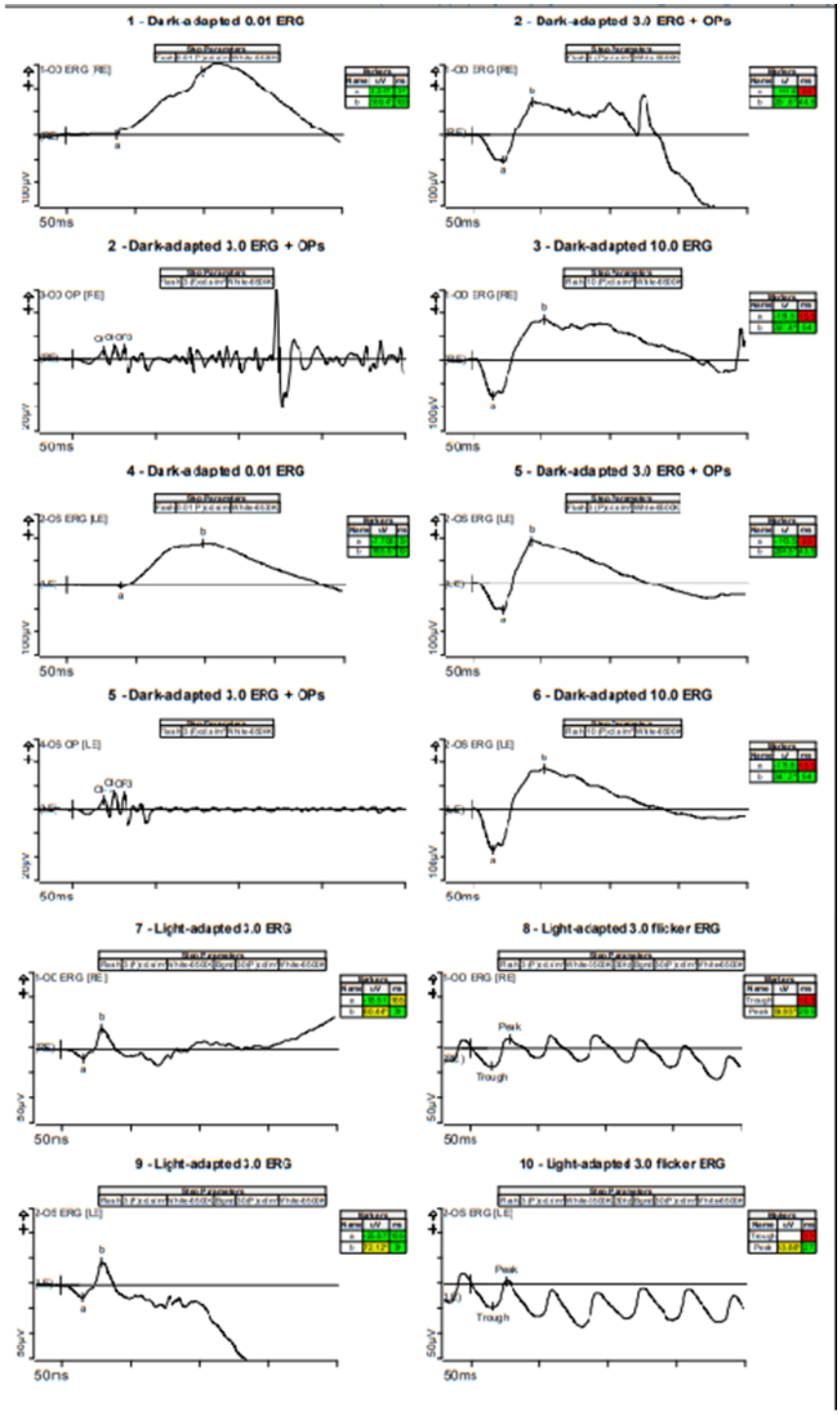

Figure 13: Electroretinogram

activity and the resting electric potential and help distinguish serious diseases with dysfunction.

3.2. Treatment options

Currently, evidence-based treatments for BCD are limited and management generally involves supportive measures. Low vision aids and vision care strategies can help restore vision and improve the quality of life of affected individuals. Genetic counselling is important for families to understand the inheritance patterns of BCD and make informed decisions about family planning and genetic testing. Research is ongoing to define disease processes and investigate therapeutic interventions in the lipid metabolic pathway or gene therapy.

Role of detection and electrophysiology research: Advanced imaging techniques, especially spectral domain OCT, help in the differential diagnosis and monitoring of diseases by providing detailed information about changes occurring in the retina.[14] Electrophysiological studies play an important role in assessing retinal function and correlating abnormal activity with clinical outcomes. These models facilitate early diagnosis, prognosis, and clinical monitoring of BCD.

In summary, our study highlights the importance of a multidisciplinary approach involving ophthalmologists, geneticists, and medical specialists in the diagnosis and treatment of BCD. Advanced imaging techniques and electrophysiological studies play an important role in the evaluation and care of the disease. Genetic counselling is important for families and highlights the need for continued research to address the mechanisms underlying BCD and improve treatment plans.[15]

3.3. Differential diagnosis

1. Retinitis Pigmentosa (RP)
2. Cystinosis
3. Sjögren-Larsson Syndrome
4. Gyrate Atrophy
5. Talc Retinopathy
6. Canthaxanthin Retinopathy
7. Ocular Toxocariasis
8. Hyperoxaluria
9. Chronic Solvent Exposure
10. Methoxyflurane Toxicity
11. Crystalline Maculopathy
12. Idiopathic Retinal Vasculitis, Aneurysms, and Neuroretinitis (IRVAN).

4. Conclusion

This particular case report is notable because it is uncommon, as there is no genetic link, and it emphasizes the importance of using SD-OCT and electrophysiological examinations to diagnose the disease early. Furthermore, it underscores the importance of genetic counseling in preventing the transmission of this condition.

5. Declaration of Patient Consent

All necessary patient consent documents have been received, as the authors attest. With this form, the patient or patients have granted permission for their photos and other clinical data to be published in a peer-reviewed journal. Patients are informed that their identities will be kept confidential and that every effort would be taken to avoid publication of their names and initials; however, complete anonymity cannot be assured.

6. Source of Funding

None.

7. Conflicts of Interest

There are no conflicts of interest.

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