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Aniridia With Dome-Shaped Maculopathy and Choroidal Excavation: Unveiling A Rare Ocular Association

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Abstract: ***A 66-year-old male presented with bilateral diminution of vision since childhood which was gradual and progressive. Both eye (BE) anterior segment examination revealed aniridia with peripheral iris remnants and pseudophakia. Fundus examination was unremarkable, but optical coherence tomography (OCT) detected dome-shaped maculopathy (DSM) in right eye (RE) and focal choroidal excavation (FCE) in the left eye (LE). The patient was counseled regarding his condition and prognosis. This case highlights a rare association of aniridia with DSM and FCE, emphasizing the need for comprehensive retinal evaluation in patients with aniridia.***

Keywords: Aniridia, Focal choroidal excavation, dome shaped maculopathy, iris, myopia, foveal hypoplasia

1. Case Report

A 66-year-old male presented with complains of gradual, painless diminution of vision in BE since childhood and intolerance to light. Despite undergoing bilateral uneventful cataract surgery two months prior, the patient's best-corrected visual acuity (BCVA) was 6/18, N8 in both eyes with the current prescription of +1.25 Dsph / +1.25 Dcyl in RE and +1.00 Dsph / +1.25 Dcyl in LE. Color vision and Amsler's grid test were. There was no history of ocular trauma, any systemic illness or similar complains in the family. BE anterior segment examination exhibited incomplete aniridia with iris tissue remnants in the periphery. **[Fig.1]** Intraocular pressure with Goldmann applanation tonometer in BE was 14mmHg and gonioscopy revealed open angles. No nystagmus was noted. On fundus examination, a fine epiretinal membrane (ERM) with macular puckering was noted in BE with presence of foveal reflex thereby, ruling out foveal hypoplasia. **[Fig.2]** OCT further revealed distinct macular pathologies alongside ERM. The RE showed elevation of the macula and fovea with loss of foveal contour without any sub-retinal fluid, corresponding to DSM, while the LE demonstrated juxta-foveal (superonasal) concave choroid suggestive of FCE. **[Fig.3]** The patient was counseled regarding his condition, and the visual prognosis was explained.

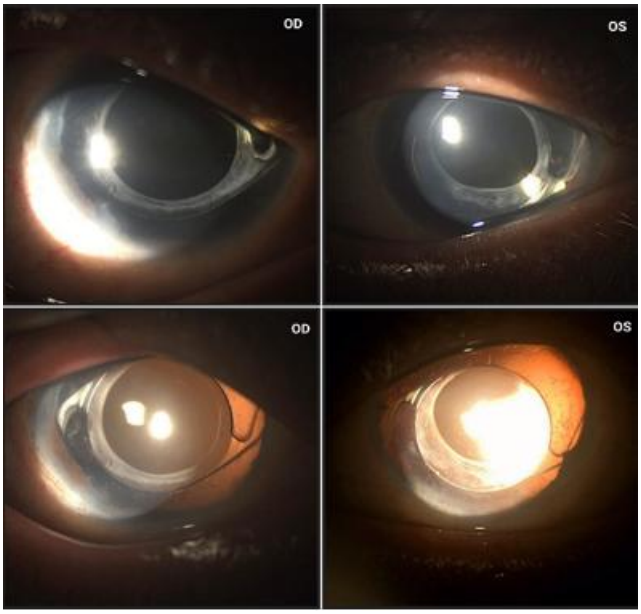

Figure 1

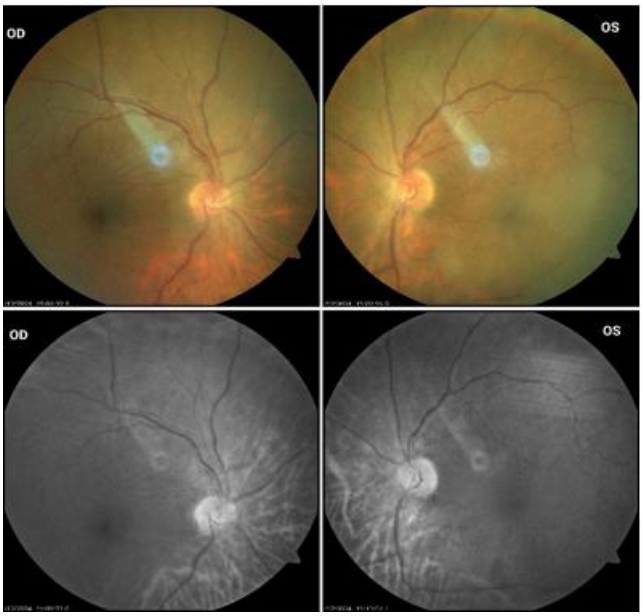

Figure 2

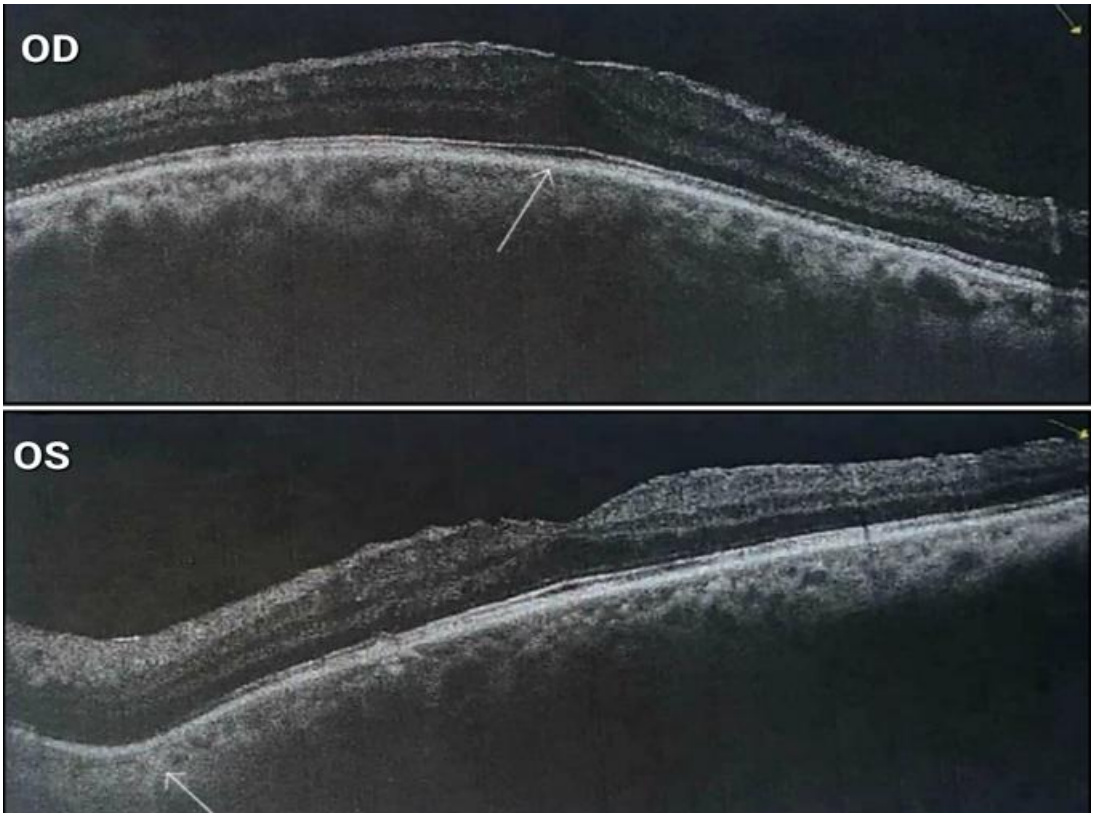

Figure 3

2. Introduction

Aniridia is a panocular disorder primarily associated with variable degrees of iris hypoplasia or absence of iris, foveal hypoplasia, reduced visual acuity, and nystagmus. Additional ocular features such as cataract, glaucoma, aniridic keratopathy, and optic disc hypoplasia are common.[1] It is seen in approximately 1.8/100 000 live births while the incidence ranges from 1:40 000 to 1:100.000 with no significant racial or gender predilection.[2]

Aniridia may be congenital or acquired. Approximately two-thirds of cases follow an autosomal dominant inheritance pattern with complete penetrance and variable expressivity. One-third occur sporadically and are often associated with systemic conditions such as WAGR syndrome (Wilms tumor, Aniridia, Genital anomalies, and Mental retardation). Additionally, around 2% of cases exhibit autosomal recessive inheritance, commonly linked to Gillespie syndrome.[3]

The PAX6 gene on chromosome 11p13 plays a crucial role in aniridia pathogenesis, affecting the development of multiple organs, including the eye.[1] Etiopathogenesis is explained by various theories, including the neuroectodermal theory, which associates aniridia with neuroectodermal defects like foveal hypoplasia, and the mesodermal theory, which links it to optic nerve head hypoplasia but does not explain foveal abnormalities.[4]

3. Discussion

While aniridia is predominantly associated with foveal hypoplasia, our case presented a unique and previously unreported combination of DSM and FCE in the fellow eye.[5] This rare finding suggests a novel variation in aniridia-related retinal and choroidal changes, emphasizing the need of comprehensive retinal evaluation in patients with aniridia.

DSM is characterized by a convex elevation of the macula, typically observed in high myopia due to localized choroidal and scleral thickening.[6] Theories for DSM in myopic eyes include posterior eye-wall collapse, vitreomacular traction, or focal scleral thickening. Additionally, a defect in Bruch's membrane could allow localized relaxation of the posterior sclera, leading to anterior bulging and dome formation.[7]

However, DSM has also been reported in emmetropic and hypermetropic eyes, suggesting an alternative, yet unidentified, underlying mechanism, as observed in our case.[7]

FCE is another rare structural anomaly with unknown etiopathogenesis, characterized by a localized depression in the choroid without associated staphyloma or scleral ectasia.[8] It is usually congenital, unilateral and associated with conditions like central serous chorioretinopathy, choroidal neovascularization, age-related macular degeneration, high myopia, and pachychoroid spectrum disorders.[9] FCE is detected using OCT based and is classified into conforming (where retinal layers follow the excavation contour) and non-conforming (where the photoreceptor layer separates from the excavation, leading to potential visual disturbances).[8]

While FCE has been observed in both myopic and non-myopic individuals, its co-occurrence with DSM in a non-myopic aniridia patient suggests a distinct pathophysiological link that warrants further investigation.

The exact mechanism underlying the coexistence of DSM and FCE in an aniridia patient remains unclear, but the neuroectodermal theory may provide a plausible explanation. Given that PAX6 mutations affect multiple ocular structures during embryonic development, it is possible that abnormal neuroectodermal differentiation contributes to both DSM and FCE.

Structural alterations in the choroid and sclera, coupled with disruptions in Bruch's membrane, may play a role in this unique presentation.

Diagnosis of DSM and FCE relies primarily on OCT, which reveals characteristic structural changes. Advanced imaging techniques such as Swept-Source OCT (SS-OCT) and Enhanced Depth Imaging OCT (EDI-OCT) allow for a more detailed assessment of choroidal and scleral thickness, aiding in a better understanding of disease mechanisms. While many cases of DSM are asymptomatic, potential complications include central serous retinal detachment and choroidal neovascularization (CNV), which may require interventions such as anti-VEGF therapy or photodynamic therapy (PDT). Asymptomatic cases, however, necessitate long-term monitoring due to the risk of progressive changes.

Our patient likely had autosomal dominant aniridia with variable expressivity, presenting with vertical DSM in the RE without evidence of subretinal fluid and juxtafoveal, conforming FCE in the LE. The patient was counseled and scheduled for follow-up to monitor for potential complications such as CNV and central serous chorioretinopathy (CSCR).

4. Conclusion

The rare co-occurrence of DSM and FCE in a non-myopic individual with aniridia expands the spectrum of retinal and choroidal anomalies associated with PAX6 mutations. Further longitudinal studies and genetic analyses are needed to elucidate the underlying mechanisms and their implications for visual prognosis and management strategies.

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