Case Report

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An unusual coexistence of iris mammillations and optic disc pit with keratoconus: A case report and literature review

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Abstract:

Iris mammillations are distinctive uniform nipple-like elevations that cover the anterior surface of the iris partially or totally. It is a rare finding and may coexist with other ocular and extraocular manifestations. Optic nerve pit (ONP), also known as optic disc pit (ODP) or optic hole, is a congenital defect resulting from the failure of fetal fissure closure during the embryonic development. It belongs to the congenital cavitary anomalies spectrum. This case presents a 19-year-old female patient who complained of a gradual decrease in visual acuity in both eyes for 4 years. Slit-lamp and fundus examinations revealed iris mammillations and ODP in the left eye. Corneal topography revealed bilateral keratoconus, which was managed with cross-linking. Iris mammillations and ODP are poorly understood ocular anomalies that are not reported frequently and have never been reported previously both combined with keratoconus. Thus, ophthalmologists should be aware of these conditions, their differential diagnosis, and their possible association with other disorders. This is the first reported case of the combined coexistence of iris mammillations and ODP with keratoconus.

Keywords:

Heterochromia iridum, iris mammillations, iris nodules, keratoconus, optic disc pit

Introduction

Iris mammillations are distinctive villiform elevations (teat- or nipple-like) that can cover the anterior surface of the iris, partially or totally, and are usually located on the anterior surface of brown irides or iris nevi. In 1912, coats described the mammillated appearance of the iris surface for the first time in a patient with ocular melanosis.[1] It is an ocular condition most commonly associated with ocular melanosis, with or without periocular skin involvement (nevus of Ota), and more prevalent among highly pigmented ethnic groups.[1-3] It is sporadic and usually unilateral. However, bilateral and familial cases have been described. Iris mammillations present with heterochromia iridis or iridum and can be associated with an elevated intraocular pressure and intraocular malignancies.[1-3] Optic nerve pit (ONP), also known as optic disc pit (ODP) or optic hole, is a congenital defect resulting from the failure of fetal fissure closure during the embryonic development. It belongs to the congenital cavitary anomalies spectrum, including morning glory, optic disc coloboma, and extrapapillary cavitation.[4] The ODP is typically a unilateral and sporadic anomaly. However, bilateral involvement (up to 15%) has been reported, and autosomal inheritance has been suggested in families with several affected individuals.[5,6] The estimated prevalence of ODP is 2 in 10,000 individuals with no gender predilection.[7] Fundus examination reveals typically single (can be more than one pit) oval or round grayish depression at any segment of the optic disc (central segment can be involved)

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but most commonly at the inferotemporal part.[5,6] Histopathologically, the OPD is described as herniation of dysplastic retina into a collagen-rich excavation that is able to stretch or extend into the subarachnoid space through a lamina cribrosa defect.[4,7]

Case Report

A 19-year-old female patient without a significant medical history presented with a gradual bilateral decrease in visual acuity for the past 4 years. She denied having any skin lesion, systemic disease, or a family history of any ocular or systemic disorder. Upon examination, she had heterochromia iridum (left iris darker than the right) and bilateral myopic astigmatism (OD: −4.25/−1.00 × 45°, OS: −0.50/−1.25 × 30°) with corrected distance visual acuity 20/40 in the right eye and 20/32 in the left eye. The slit-lamp examination was unremarkable in both eyes apart from the presence of smooth, thin, villiform, brown, diffuse, uniformly elevated, and regularly distributed iris nodules overlying the entire anterior surface of the iris in the left eye only [Figure 1b]. The bases of the nodules showed tiny radiations, and the nodular elevation tended to increase on approaching the pupillary margin. The corrected intraocular pressure was 16 mmHg in the right eye and 15 mmHg in the left eye. The dilated fundus exam showed a single oval grayish depression at the inferotemporal part of the left optic disc [Figure 2b]. The right fundus was normal [Figure 2a]. Figure 2 shows the optical coherence tomography (OCT) of both eyes. ODP maculopathy (ODP-M) was noticed in the left eye [Figure 2c].

Corneal topography (Pentacam) revealed keratoconus in both eyes [Figure 3]. Therefore, the patient underwent epi-off corneal cross-linking under topical anesthetic. The appearance of iris nodules and the absence of systemic manifestations were crucial for iris mammillations diagnosis. Findings in fundus examination revealed an ODP with subretinal fluid, which was not treated due to the patient's refusal. The treatment plan was to start with argon laser photocoagulation temporal to the optic disc and injection of intravitreal perfluoropropane gas, followed by regular follow-ups with best-corrected visual acuity (BCVA) and OCT. If the initial treatment failed, the next step was supposed to be pars plana vitrectomy, posterior vitreous detachment induction, and gas tamponade, with argon laser photocoagulation redo or not, according to the patient's situation.

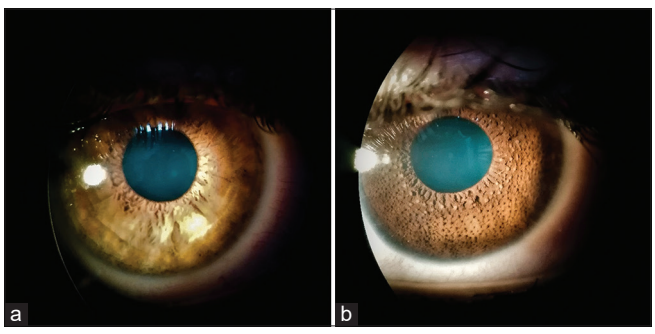

Figure 1: Shows heterochromia iridum. Normal bright iris in the right eye (a) and dark iris with iris mammillations in the left eye (b)

Discussion

In 2019, Antunes-Foschini *et al*. reported for the first time a possible association between bilateral iris mammillations and keratoconus.[8] From 1995 to 2018, the reported ocular conditions associated with iris mammillations included myopia, amblyopia, esotropia, congenital ptosis, eyelid trichilemmoma, ipsilateral ocular melanocytosis, oculodermal melanocytosis, scleral pigmentation, iris hamartomas, persistent pupillary membrane, congenital cataract, congenital glaucoma, calcified ciliary body mass, choroidal melanoma, choroidal hemangioma, retinal detachment, optic neuritis, and optic nerve damage.[1,2,9] Based on these previously reported conditions, keratoconus was reported only recently in 2019. The systemic disorders included congenital heart disease, congenital adrenal hyperplasia, tuberculosis frontal arachnoid cyst, cleft palate, marfanoid habitus, abnormal rib, seizures, ectopic Mongolian spot, preauricular skin tag, clitoris acne, labioscrotal hyperpigmentation, Axenfeld and Peters anomaly, neurofibromatosis Type 1 (NF1), phakomatosis pigmentovascularis Iib, Cowden syndrome, Sturge-Weber syndrome, and nevus flammeus of the face.[1,2,9] In addition, some cases were reported without other ocular or systemic disorders.[1,2]

Iris mammillations are commonly confused with iris nodules accompanying NF1, known as Lisch nodules.[1]

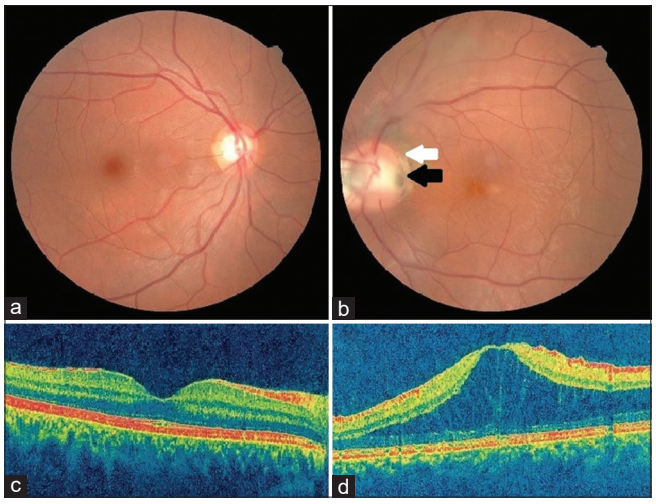

Figure 2: (a) Normal right optic disc and fundus. (b) Inferotemporal optic disc pit (ODP) (black arrow) with an area of peripapillary atrophy (white arrow). (c) Normal macular OCT in the right eye. (d) Left eye OCT shows optic disc pit maculopathy with subretinal fluid. OCT: Optical coherence tomography

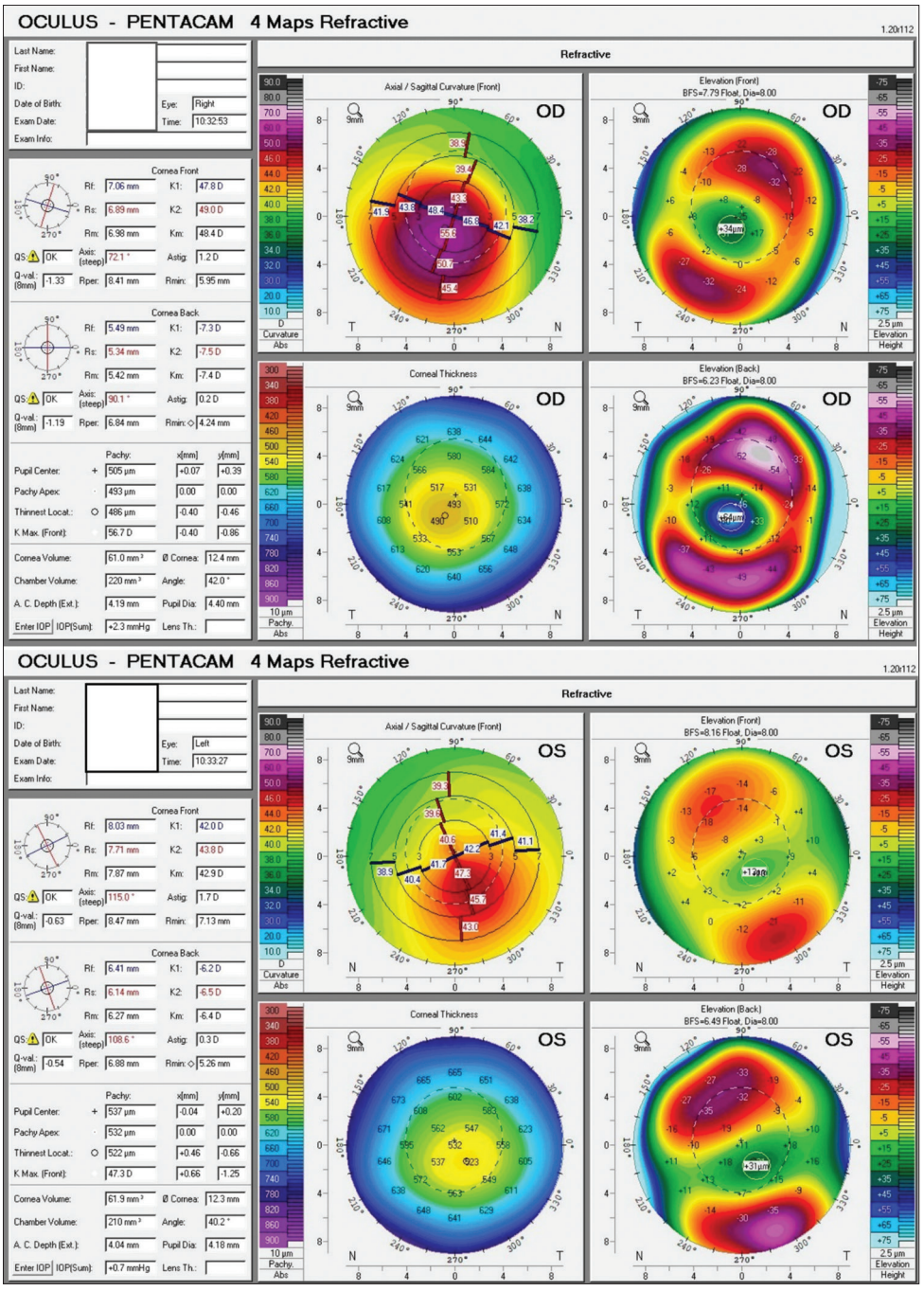

Figure 3: Corneal topography (Pentacam) for the right (OD) and left (OS) eyes indicates the presence of keratoconus in both eyes

In addition to Lisch nodules, the differential diagnosis includes Brushfield flecks, iris melanoma, granulomatous iridocyclitis, iris nevi, Cogan-Reese iridocorneal endothelial syndrome, juvenile xanthogranuloma, and retinoblastoma.[1,2,8,10] In this case, the typical appearance of mammiform iris protuberances in addition to the absence of iris melanosis and systemic disorders were significant to diagnose this extremely rare ocular condition [Figure 1]. Iris mammillations are distinctive, uniformly elevated, and regularly spaced smooth conical and villiform nodules, which sometimes stellate-like with tiny radiations emanating from the nodules bases, overlying areas of iris hyperpigmentation (naevi or iris melanosis), but not a pigmentary condition *per se*. In contrast, Lisch nodules are fluffier, brown pigmented, and irregularly separated hamartomatous nodules that do not overlie areas of iris hyperpigmentation and coexisted with ocular hypertension, intraocular malignancies, or/and extraocular disorders.[1,2]

The pathogenesis of ODP is not fully known yet, and no associations with other ocular or systemic disorders were found.[5,6] In 2008, Fasciani *et al*. reported bilateral ODP and keratoconus for the first time followed by

Aslankurt *et al*. in 2015.[11,12] Patients with ODP may present with visual field defects (enlarged blind spot or paracentral arcuate scotoma), but usually, this condition is found incidentally.[6,13] A term known as ODP ODP-M is used to describe retinal problems resulting from the ODP, including serous detachment and retinoschisis, which can affect vision significantly (BCVA 20/70 or lower) and has been estimated to affect 25%–75% of ODP patients during their life.[6,14] Long-standing cases of ODP-M can lead to irreversible vision loss with BCVA worse than 20/200.[6] Patients with ODP-M can be managed with laser photocoagulation, pars plana vitrectomy, pneumatic tamponade with or without laser photocoagulation, macular buckling, autologous fibrin, glial tissue removal, and inner retinal fenestration.[6]

Although there were reported cases of isolated iris mammillations with keratoconus and ODP with keratoconus, no association was found between these anomalies and keratoconus. Therefore, this coexistence is most probably an incidental finding. This report highlighted a rare case of unilateral iris mammillations and ODP that coexisted with keratoconus, which was not reported previously in one patient.

Conclusion

Iris mammillations and ODP are rarely reported ocular conditions that can be isolated or associated with other ocular and extraocular manifestations. Iris mammillations may be associated with ocular hypertension and intraocular malignancies. Thus, due to their similarity to other iris nodules, patients should be evaluated thoroughly. Most probably, iris mammillations and ODP were incidental findings presented with keratoconus. However, studies are needed in the future to look for a correlation among iris mammillations, ODP, and keratoconus. These two anomalies are poorly understood and not reported frequently in the literature. Ophthalmologists should be aware of these rare anomalies, their differential diagnosis, and their possible coexistence with other ocular or/and extraocular manifestations.

Declaration of patient consent

The author certify that they have obtained all appropriate patient consent forms. In the form, the patient has given her consent for her images and other clinical information to be reported in the journal. The patient understands that her name and initials will not be published, and due efforts will be made to conceal her identity, but anonymity cannot be guaranteed.

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Conflicts of interest

The author declares that there are no conflicts of interests of this paper.

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