

# The Spotted Iris: Unveiling a Case of Bilateral Iris Mammillations in a Type 1 Diabetic Adolescent

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## ABSTRACT

Bilateral iris mammillations are an exceedingly rare ocular anomaly characterised by small, papilliform nodules that cover part or the entire surface of the iris. Although often associated with systemic conditions such as Neurofibromatosis (NF), oculodermal melanocytosis, or phakomatosis pigmentovascularis, isolated bilateral cases are infrequently reported. This report describes a case of a 14-year-old male who presented for a routine dilated fundus examination at a tertiary care centre. The patient, a known case of Type 1 Diabetes Mellitus (DM) for 11 years, was maintained on subcutaneous human Actrapid insulin. He reported no ocular complaints. Examination revealed a best-corrected visual acuity of 6/6 in both eyes with intact colour vision. Slit lamp examination identified multiple, diffuse, small, round, nodular elevations over the surface of both irides without evidence of iris neovascularisation. Gonioscopy revealed open angles without trabecular hyperpigmentation. Posterior segment evaluation, including Optical Coherence Tomography (OCT) and B-scan ultrasonography, was unremarkable. Ultrasound biomicroscopy confirmed the absence of ciliary body involvement. Systemic examination showed no evidence of NF, oculodermal melanocytosis, or phakomatosis pigmentovascularis. Genetic testing for NF type 1 returned negative. The presence of isolated bilateral iris mammillations without systemic association is exceedingly rare. This case adds valuable data to the limited literature on such presentations and highlights the importance of thorough ocular and systemic evaluation to differentiate these nodules from other ocular anomalies. Continued surveillance and long-term follow-up are crucial, given the potential but rare association with secondary glaucoma and ocular malignancy.

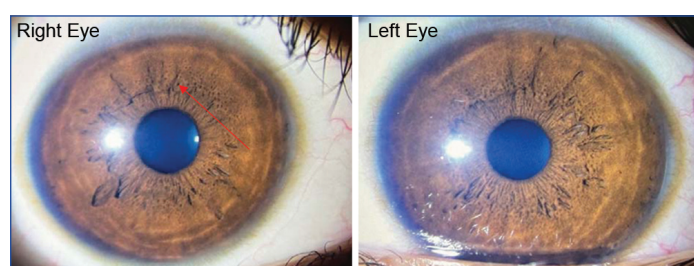
**Keywords:** Actrapid insulin, Nodular elevations, Papilliform nodules

## CASE REPORT

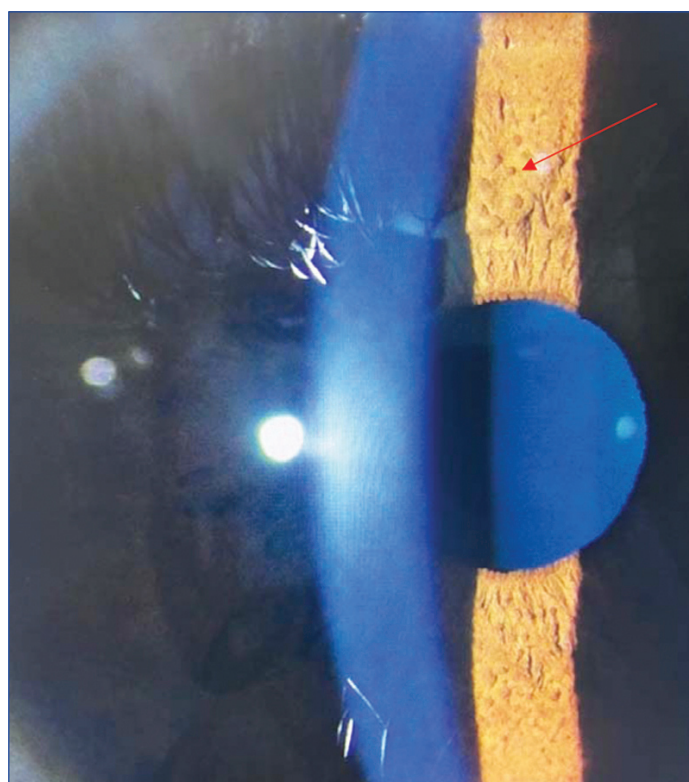
A 14-year-old male, residing in an urban area, presented to the tertiary hospital in Pune for a routine dilated fundus examination. The patient was diagnosed with Type 1 DM at the age of 3 years and was on subcutaneous human Actrapid insulin therapy since his diagnosis. He has been wearing spectacles for distance vision for the past three years. The patient denied any ocular pain, redness, or visual disturbances.

On examination, his best-corrected visual acuity was 6/6 in both eyes, and his colour vision was intact. The ocular adnexa appeared normal, with no hyperpigmentation in the ocular or periocular areas. Extraocular movements were full, free, painless, and no strabismus was noted. On slit lamp examination, multiple, diffuse, small, round, nodular elevations were observed on the iris bilaterally [Table/Fig-1,2]. No neovascularisation of the iris was noted. Anterior segment structures showed no evidence of hyperpigmentation. The pupils were equal, circular, and reactive to direct and consensual light reflexes.

Intraocular Pressure (IOP) measurements were 12 mmHg in the right eye and 14 mmHg in the left eye, obtained using a Goldmann applanation tonometer. Gonioscopy revealed open angles with no heavy pigmentation of the trabecular meshwork.

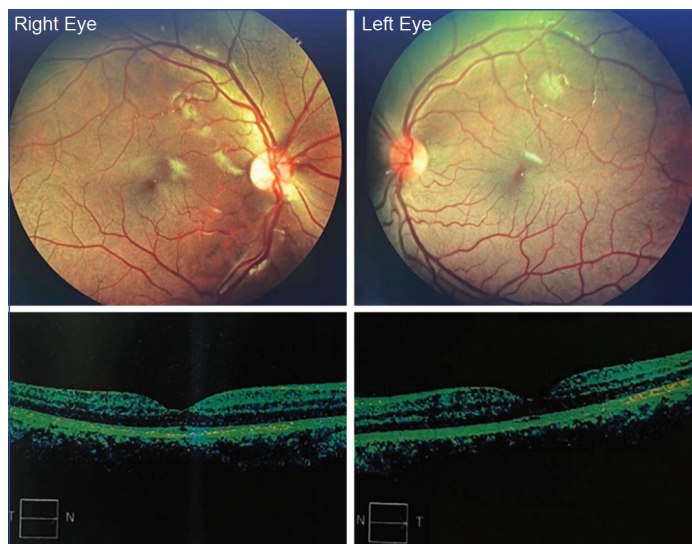


**[Table/Fig-1]:** Slit lamp photographs of anterior segment of both eyes showing iris mammillations.

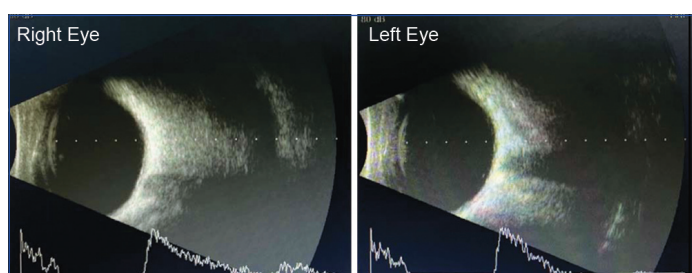


**[Table/Fig-2]:** Slit section focusing on iris mammillations.

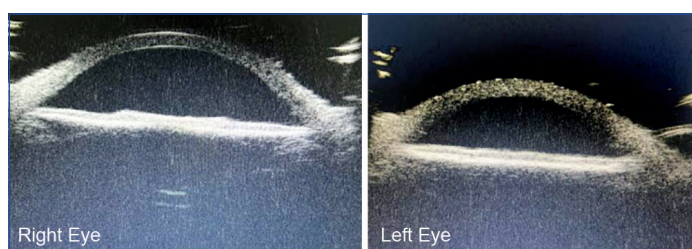
Indirect ophthalmoscopy using a 20 D lens showed a normal fundus in both eyes. OCT demonstrated normal macular and optic disc morphology [Table/Fig-3]. B-scan ultrasonography was unremarkable [Table/Fig-4], and ultrasound biomicroscopy showed no ciliary body involvement [Table/Fig-5]. Differential diagnoses considered included Lisch nodules associated with NF type 1 (NF1), iris nevi, melanocytosis, and iris melanoma.



**[Table/Fig-3]:** Fundus photographs and optical coherence tomographic macula image of both eyes reveal normal posterior segment structures without any remarkable abnormalities.



**[Table/Fig-4]:** B-scan of both eyes reveal a normal posterior segment with an attached retina, clear vitreous cavity, and no evidence of intraocular mass, retinal detachment, or choroidal thickening. Overall, the B-scan findings are unremarkable in both eyes.



**[Table/Fig-5]:** Ultrasound biomicroscopy of both eyes reveal no ciliary body involvement.

No abnormal pigmented spots or patches were observed anywhere on the skin. No bluish-grey pigmentation on the face or ocular structures was noted [Table/Fig-6]. The family history was negative for similar ocular findings. To confirm or exclude a diagnosis of NF type 1, genetic testing was conducted, which revealed no pathogenic variant in the NF1 gene. Systemic examination revealed a heart rate of 80 bpm and a blood pressure of 120/80 mmHg. Laboratory investigations showed an HbA1c level of 7.0%. Additionally, the patient was evaluated by a multidisciplinary team, with no significant abnormalities identified.



**[Table/Fig-6]:** Clinical pictures- No abnormal pigmentation noted.

Based on the clinical and imaging findings, the final diagnosis was bilateral iris mammillations. No specific treatment was required

in this case, as the patient had no associated systemic or ocular complications. Regular annual ophthalmologic examinations were recommended to monitor for any new clinical signs, the development of secondary glaucoma, or other associated ocular conditions.

## DISCUSSION

Bilateral iris mammillations are an exceedingly rare ocular anomaly characterised by small, papilliform nodules covering part or the entire surface of both irides. These elevations are often deep brown, smooth, and conical in shape, and may increase in height towards the pupil margin. While unilateral cases are relatively more common, bilateral iris mammillations have been reported infrequently in the literature, often associated with conditions such as NF, melanosis oculi, or phakomatosis pigmentovascularis [1,2].

A recent case report by Usui S et al., highlighted the association of iris mammillations with uveitic glaucoma, where the patient presented with multiple nodules scattered over the iris surface. After trabeculectomy, long-term follow-up revealed stability of Intraocular Pressure (IOP) and resolution of nodular prominence without signs of malignancy [3]. This underscores the importance of ruling out malignancy in cases of iris mammillations associated with glaucoma to ensure appropriate management.

In addition to their association with glaucoma, iris mammillations have been linked to ocular melanocytosis and phakomatosis pigmentovascularis type IIb, which can predispose individuals to melanoma development. Monitoring of these patients is crucial to detect any malignant transformation early [4].

In clinical practice, iris mammillations are frequently mistaken for Lisch nodules seen in NF1. However, iris mammillations differ significantly as they are typically uniform in size and colour, matching the underlying iris pigment, while Lisch nodules vary in size and colour and are typically found on lightly pigmented areas of the iris [5]. Furthermore, while Lisch nodules are often associated with other signs of NF, iris mammillations may occur independently or with other non-neurofibromatous conditions [5].

Reports have shown that iris mammillations are usually unilateral and sporadic, though familial cases with dominant inheritance have been noted [1,6]. They are predominantly seen in individuals of more pigmented ethnic backgrounds. Although often asymptomatic, their detection warrants thorough evaluation to rule out associated systemic conditions and ocular hypertension [1,6]. Similar cases have been reported in the literature. Yamamoto M et al., described bilateral iris mammillations in amblyopic eyes without any systemic association [2]. Antunes-Foschini RMS et al., documented iris mammillations associated with keratoconus, suggesting a possible link to anterior segment anomalies [7]. Usui S et al., reported iris mammillations associated with uveitic glaucoma, emphasising the need for long-term monitoring to rule out complications [3].

In this case, the presence of bilateral iris mammillations without neurocutaneous systemic associations such as NF or oculodermal melanocytosis is unusual. This highlights the importance of thorough ocular examination and accurate differentiation from other iris anomalies. While their exact pathogenesis remains unclear, the occurrence in the absence of other ocular or systemic conditions is particularly noteworthy. This case adds to the limited number of reports documenting isolated bilateral iris mammillations and underscores the rarity of this ocular finding [8].

## CONCLUSION(S)

This case highlights a rare presentation of bilateral iris mammillations in a young adolescent without associated neurocutaneous systemic disorders. The absence of typical associations such as NF, oculodermal melanocytosis, or phakomatosis pigmentovascularis makes this case particularly unique. Despite the lack of clear aetiological factors, the identification of bilateral iris mammillations



warrants careful ophthalmologic evaluation to differentiate them from other similar presentations, such as Lisch nodules.

The importance of distinguishing iris mammillations from other iris anomalies lies in their potential association with ocular and systemic pathologies. While most cases are benign and asymptomatic, rare instances of association with glaucoma and increased IOP have been reported. Therefore, long-term follow-up is crucial to monitor any potential changes in IOP or the development of secondary glaucoma. Additionally, the risk of malignant transformation, although low, necessitates periodic evaluation, especially in patients with concurrent ocular melanocytosis.

This case underscores the need for comprehensive ocular examination and awareness among clinicians regarding the rarity and potential implications of bilateral iris mammillations. Further studies are necessary to elucidate the pathophysiology and long-term outcomes associated with this unique presentation, particularly in cases without associations.

## REFERENCES

- [1] Ragge NK, Acheson J, Murphree AL. Iris mammillations: Significance and associations. *Eye*. 1996;10(1):86-91.
- [2] Yamamoto M, Mimura T, Matsumoto K, Hamano S, Nanba H, Ubukata S, et al. Bilateral iris mammillations in amblyopic eyes without oculodermal melanocytosis or neurofibromatosis. *Case Rep Ophthalmol Med*. 2018;2018:2534042.
- [3] Usui S, Okazaki T, Fujino T, Kawashima R, Hashida N, Matsushita K, et al. Long-term course with iris changes after trabeculectomy for uveitic glaucoma associated with iris mammillation: A case report. *BMC Ophthalmol* [Internet]. 2023;23(1):01-05. Available from: <https://doi.org/10.1186/s12886-023-02854-z>.
- [4] Gündüz K, Shields CL, Shields JA, Eagle RC, Singh AD. Iris mammillations as the only sign of ocular melanocytosis in a child with choroidal melanoma. *Arch Ophthalmol*. 2000;118(5):716-17.
- [5] Jain N, Johnson MW. Pathogenesis and treatment of maculopathy associated with cavitory optic disc anomalies. *Am J Ophthalmol*. 2014;158(3):423-35.
- [6] Kim BZ, Lim JZ, McGhee CNJ. Cysts and tumours of the iris: Diagnostic tools and key management considerations- A review. *Clin Exp Ophthalmol*. 2024;52(6):665-83.
- [7] Antunes-Foschini RMS, Costa RMS, Faria-e-Sousa SJ, Rocha EM. Are iris mammillations correlated with keratoconus? *Am J Ophthalmol Case Reports* [Internet]. 2019;14:16-18. Available from: <https://doi.org/10.1016/j.ajoc.2019.01.011>.
- [8] Marugán BS, García MA, Hinojosa JG, Cabello ML, Valino MC. Iris mammillations. Three case reports. *Archivos de la Sociedad Española de Oftalmología (English Edition)*. 2014;89(7):279-81.

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