Oculocutaneous albinism with iridofundal coloboma

Gunjan Saluja,[1] Shorya Vardhan Azad,[1] Amar Pujari,[2] Shreyas Temkar[3]

[1]Dr Rajendra Prasad Centre for Ophthalmic Sciences, All India Institute of Medical Sciences, New Delhi, India
[2]Department of Ophthalmology, Dr Rajendra Prasad Centre for Ophthalmic Sciences, All India Institute of Medical Sciences, New Delhi, India
[3]Department of Ophthalmology, Dr Rajendra Prasad Centre for Ophthalmic Sciences, All India Institute of Medical Sciences., New Delhi, India

Correspondence to
Dr Shorya Vardhan Azad, shoryaazad@hotmail.com

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DESCRIPTION

A 20-year-old woman presented to the retina clinic with complaints of diminution of vision, photophobia and involuntary movement of both eyes since birth. The patient had light-coloured skin complexion along with golden hair. Best corrected visual acuity was 1/60 and 4/60 in the right and left eyes, respectively. Ocular examination revealed manifest nystagmus, but there was no evidence of squint or head posture. The anterior segment had clear cornea and lens, with colobomatous light-coloured iris. Fundus examination in the right eye showed diffusely hypopigmented fundus with a large, well-defined excavated area along the inferior and nasal quadrant, extending well above the optic disc and the macula, suggestive of type I iridofundal coloboma (figure 1). Similarly, in the left eye, there was diffuse hypopigmentation except at the macula. Retinal and choroidal vessels were well appreciated along with their drainage into the vortex veins (figure 2). Based on these classical ocular and cutaneous findings, a diagnosis of oculocutaneous albinism with right-sided fundal coloboma was made. The patient was counselled about the nature of the disease and advised for refractive correction, dark-tinted glasses for photophobia and kept under follow-up.

Oculocutaneous albinism is a rare genetic disorder characterised by the complete or partial absence of melanin pigment from the skin, eyes and hairs. The worldwide prevalence has been estimated to be around 1 in 17 000.[1] There are various types and classifications of albinism, but broadly they can be divided into oculocutaneous and ocular albinism. The oculocutaneous type is further subclassified into tyrosine-positive and tyrosine-negative variants depending on the activity of tyrosinase enzyme, which is crucial for the synthesis of melanin.[2 3]

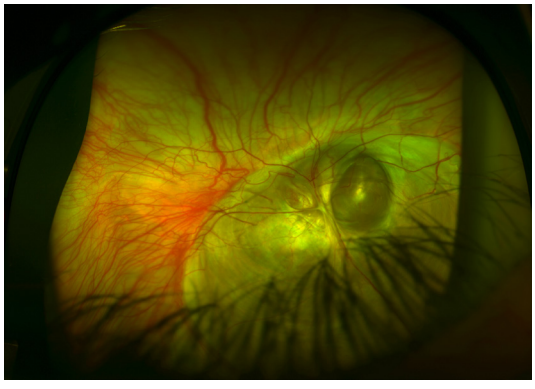

Figure 1 Right eye fundus showing diffuse hypopigmentation along with excavated lesion involving the inferior and nasal retina. The optic disc and macula were within the colobomatous area suggestive of type I iridofundal coloboma.

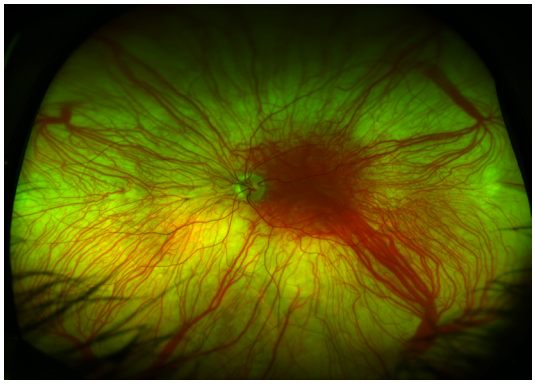

Figure 2 Left eye fundus showing similar kind of diffuse hypopigmentation sparing the macula with prominent retinochoroidal drainage into the vortex vein.

Coloboma of choroid occurs due to defective closure of the embryonic fissure of the developing optic vesicle. It can be an isolated disorder or associated with ocular or systemic abnormalities. Association with iridofundal coloboma has not been previously reported. Although the exact association between these two entities cannot be ascertained, their occurrence can either be coincidental or may be due to a defective genetic pathway. Genetic testing for specific genes may reveal any such association.

Learning points

- Oculocutaneous albinism is rarely associated with iridofundal coloboma.
- Patients with oculocutaneous albinism might possess significant posterior segment pathology. Thus a detailed ocular evaluation is necessary.

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