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Kamelia Rifai, Salma Moutamani

Corresponding author: Kamelia Rifai, Department A of Ophthalmology, Mohammed V University Souissi, Rabat, Morocco. rifai.camelia@gmail.com

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Pediatric patient with oculocutaneous albinism

Kamelia Rifai[1,&], Salma Moutamani[1]

[1]Department A of Ophthalmology, Mohammed V University Souissi, Rabat, Morocco

[&]**Corresponding author**

Kamelia Rifai, Department A of Ophthalmology, Mohammed V University Souissi, Rabat, Morocco

Image in medicine

Oculocutaneous albinism (OCA) is a heterogeneous autosomal recessive disorder characterized by a reduced or absent melanin synthesis. OCA causes a complete or partial absence of pigment in the skin, hair, and eyes. Reduction of melanin in the eyes results in reduced visual acuity, photophobia, and nystagmus. A 6-year-old boy, the first child of a related couple, was born following a normal gestation and delivery. At term, he weighed 3800 g, and his length was 50 cm. OCA was noted at birth; there was total depigmentation of the skin, hair, and iris. He presented with photophobia and bilateral strabismus. The patient had white hair, eyebrows, eyelashes, and depigmented skin (A). Ophthalmologic examination revealed characteristic iris depigmentation and

translucency (B), retinal depigmentation (C), misrouting of the optic nerve fibers and reduced visual acuity (5/10). He had a normal mental development with no other congenital deformities. He did not have any abnormalities of the internal organs. The diagnosis of OCA is based on clinical findings of hypopigmentation of the skin and hair, in addition to the characteristic ocular symptoms. However, due to the clinical overlap between the OCA subtypes, molecular diagnosis is necessary in order to establish the gene defect and thus the OCA subtype. Reduced visual acuity can be helped in various ways. Glasses, possibly bifocals, may often be of sufficient help. Oculocutaneous albinism is an autosomal recessive disorder that presents with heterogeneous expressivity.

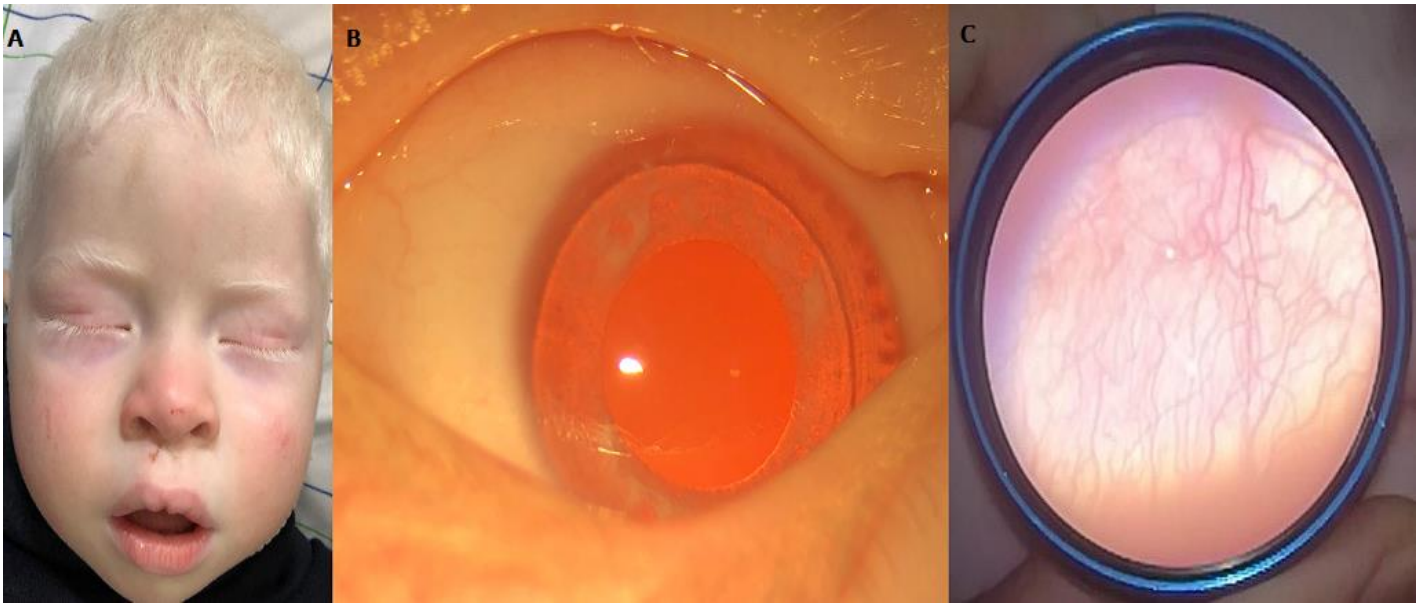

Figure 1: (A, B, C) oculocutaneous albinism