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Clinical-Medical Image

Albinism a Paradigmatic Image

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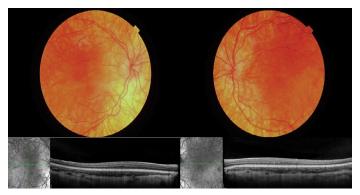


Figure 1: Fundus and OCT structural imaging.

Clinical-Medical Image

A 4 years old girl referenced to the Paediatrics Ophthalmology department due to low vision and suspected albinism due to hair and skin hypopigmentation. Personal and family history was otherwise unremarkable. Visual acuity was 5/10 in both eyes. Ocular motility and alignment were normal. Biomicroscopy was unremarkable without retro illumination defects. Fundus and OCT structural imaging (Figure 1) showed marked fundus hypopigmentation and absence of foveal depression, compatible with oculocutaneous albinism. Electrophysiology and genetic study later confirmed the diagnosis [1].

Keywords: Albinism; Retina

References

[1] Gronskov K, Ek J, & Brondum-Nielsen K (2007) Orphanet journal of rare diseases. Orphanet J Rare Dis 2: 43-51.

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