



Depigmented Fundus and Fovea Plana in Hermansky-Pudlak Syndrome

A 17-year-old Emirati woman, born out of consanguineous marriage, was diagnosed with Hermansky-Pudlak syndrome using whole genome sequencing. Her medical history was significant for oculocutaneous albinism, myalgias, skin bruises, and polycystic ovarian syndrome. Best-corrected visual acuity was 20/200 in both eyes. She had a corneal pachymetry of 630 μm in both eyes. Ultrawidefield photography (A, B) showed hypopigmented fundus with visible choroidal vessels. OCT through the macula (C, D) showed fovea plana. She was monitored for glaucoma and myopia. (Magnified version of Figure A-D is available online at www.ophtalmologyretina.org)

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