Ocular Findings of Revesz Syndrome in a 12-Year-Old Girl

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A 12-year old girl presented with gradually progressive diminution of vision in both eyes and the classic triad of findings associated with dyskeratosis congenita (DC), a rare inherited condition with progressive bone marrow failure: reticulate skin pigmentation over the trunk and neck (Figure 1), nail dystrophy (Figure 2) and white plaques over the tongue and oral mucosa (Figure 3). Best corrected visual acuity in both eyes was 6/60. Fundus examination showed optic atrophy, retinal fibrosis, retinal nerve fiber layer hemorrhages, exudative retinopathy over the posterior pole (Figures 4-5), and periphery, along with pigmentary changes (Figures 6-7) resembling those seen in retinitis pigmentosa. No abnormalities were detected on neuroimaging. Revesz syndrome is a variant of DC characterized by exudative retinopathy and intracranial calcification among other systemic anomalies. A mutation in the DC gene 1 (DKC1) at Xq28 results in dysfunction of dyskerin, a protein that is involved in telomere maintenance and ribosomal biogenesis. Other ocular findings may include nasolacrimal duct obstruction, retinal detachment, ectropion, entropion, and trichiasis. Features of retinitis pigmentosa have also been previously described in Revesz syndrome.

REFERENCES