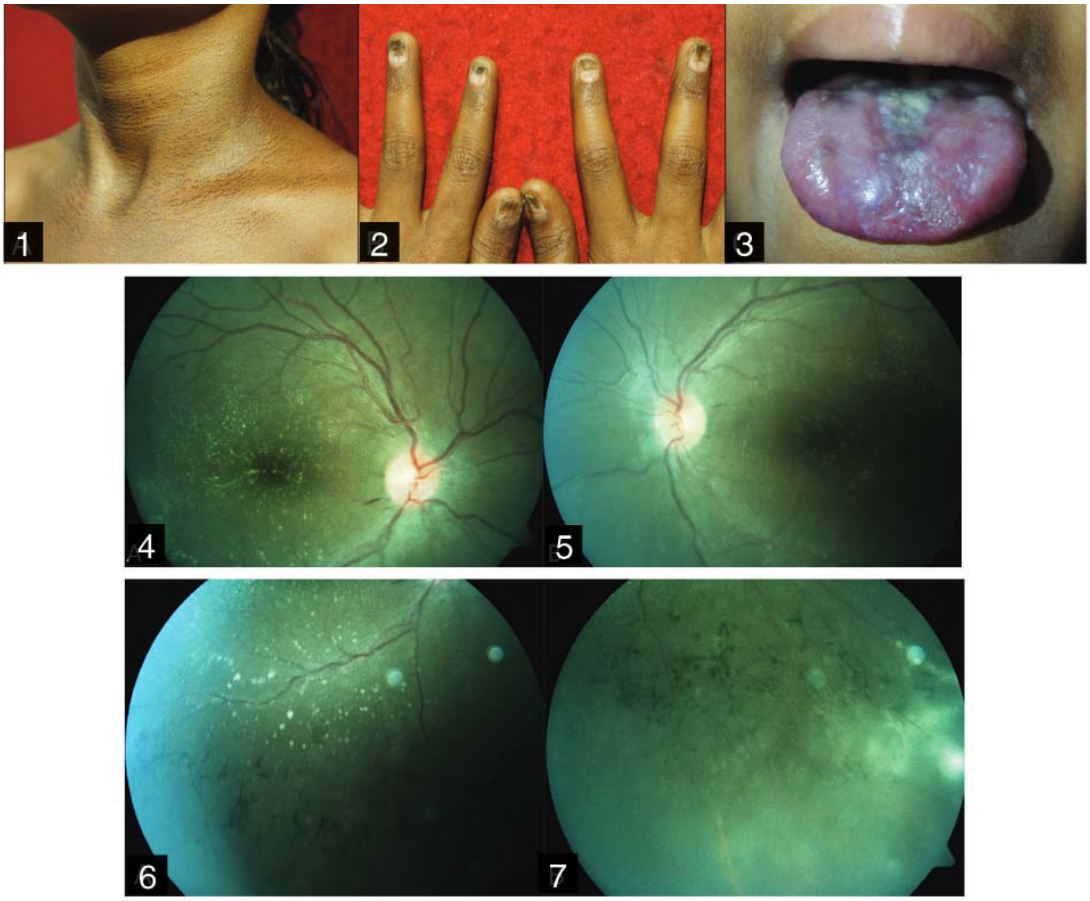
Ocular Findings of Revesz Syndrome in a 12-Year-Old Girl

Mihir G. Trivedi, MS; Poonam J. Rai, MS; Shruti P. Shirwadkar, DNB; Harsha S. Pagad, DNB, FMRF; Nayana A. Potdar, MS, FAICO; Chhaya A. Shinde, MS; Akshay Gopinathan Nair, DNB



A 12-year old girl presented with gradually progressive diminuition 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.[1] 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.[2] Other ocular findings may include nasolacrimal duct obstruction, retinal detachment, ectropion, entropion, and trichiasis.[3] Features of retinitis pigmentosa have also been previously described in Revesz syndrome.[4]

REFERENCES

1. Revesz T, Fletcher S, al-Gazali LI, DeBuse P. Bilateral retinopathy, aplastic anaemia, and central nervous system abnormalities: a new syndrome? *J Med Gen.* 1992;29:673-675.
2. Marrone A,Walne AJ, Dokal I. Dyskeratosis congenita telomerase, telomeres and anticipation. *Curr Opin Genet Dev.* 2005;15:249-257.
3. McElnea EM, van der Spek N, Smith O, Fitzsimon S, Patel CK, O'Marcaigh A. Revesz syndrome masquerading as bilateral cicatricial retinopathy of prematurity. *J AAPOS.* 2013;17:634-636.
4. Riyaz A, Riyaz N, Jayakrishnan MP, Mohamed Shiras PT, Ajith Kumar VT, Ajith BS. Revesz syndrome. *Indian J Pediatr.* 2007;74:862-863.

From the Department of Ophthalmology, Lokmanya Tilak Municipal Medical College & General Hospital, Mumbai, India (MGT, PJR, SPS, HSP, NAP, CAS); Advanced Eye Hospital & Institute, Mumbai, India (AGN); and Aditya Jyot Eye Hospital, Mumbai, India (AGN).

The authors have no financial or proprietary interest in the materials presented herein.

Correspondence: Akshay Gopinathan Nair, DNB, Department of Ophthalmology, Lokmanya Tilak Municipal Medical College & General Hospital, Dr. Babasaheb Ambedkar Road, Sion West, Mumbai, Maharashtra 400022, India. E-mail: akshaygn@gmail.com

doi:10.3928/01913913-20160122-04