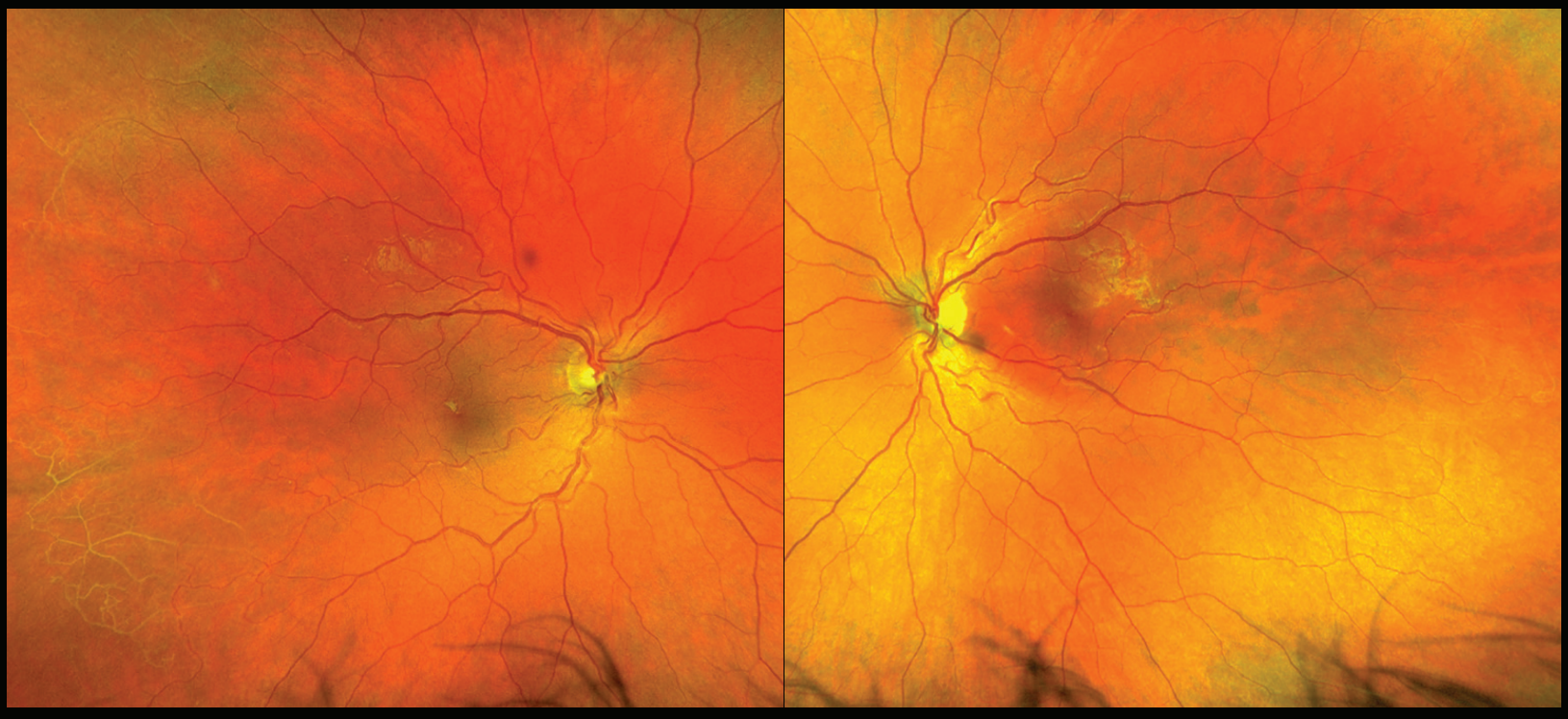

# DYSKERATOSIS CONGENITA RETINOPATHY


Understand the ocular signs of a rare genetic condition that poses serious systemic risk.

BY BOONTIP TIPSURIYAPORN, MD, AND YOSHIHIRO YONEKAWA, MD

A 20-year-old woman with a history of dyskeratosis congenita (DC) presented with retinal vascular abnormalities. VA was 20/20 OU, and she was experiencing trichiasis and dry eye symptoms. Fundus examination revealed peripheral nonperfusion with sclerotic vessels in each eye (Main Figure). Widefield fluorescein angiography (FA) demonstrated peripheral nonperfusion, telangiectasias, vascular shunting, and mild vascular leakage in each eye (Figure, next page). The nonperfusion was mild without signs of neovascularization, so close observation was recommended.

## DISCUSSION

Dyskeratosis congenita is a rare genetic condition characterized by telomere shortening that can lead to critical systemic sequelae, including bone marrow failure, pulmonary arteriovenous malformations and fibrosis, and gastrointestinal telangiectatic anomalies. Ophthalmic complications include trichiasis, punctual stenosis, exudative vitreoretinopathy, retinal neovascularization, and tractional/exudative retinal detachment.[1-5] Widefield FA is particularly useful for diagnosing and monitoring the associated retinopathy. Nonproliferative vasculopathies may be managed conservatively, but more significant vascular leakage and proliferative neovascular disease would benefit from treatment with laser photocoagulation. Advanced disease with retinal detachment may require surgical intervention. ■

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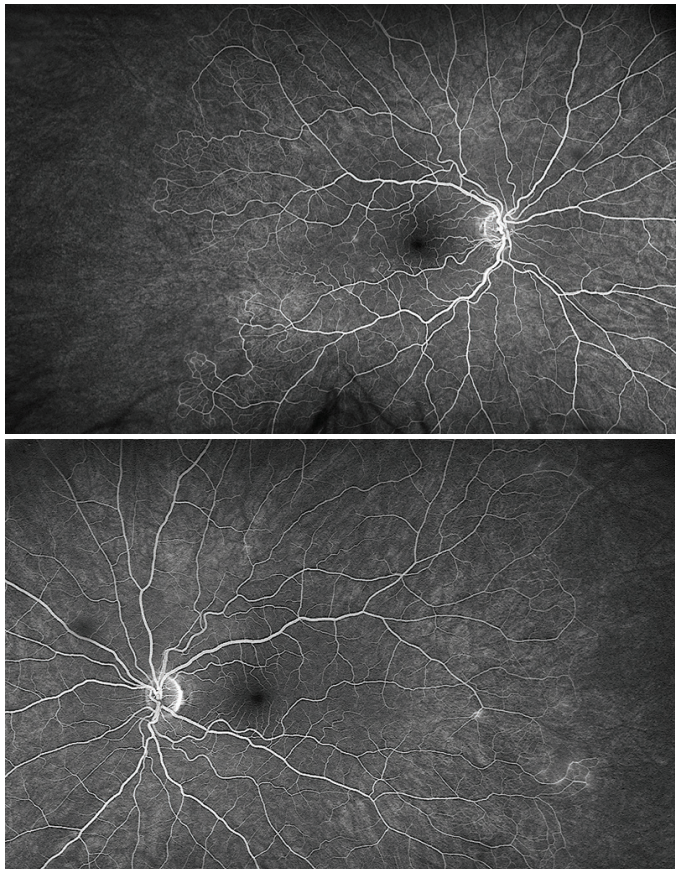

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