Bilateral Retinal Vasculopathy in a Patient With Dyskeratosis Congenita

Dyskeratosis congenita is a syndrome of progressive bone marrow failure associated with patchy cutaneous pigmented abnormalities, leukoplakia, and nail dystrophy. We report the case of a boy who had progressive bilateral retinal vasculopathy, evolving pancytopenia, and skin and nail changes, symptoms indicating dyskeratosis congenita.

Report of a Case. An 11-year-old white boy noticed decreased vision in his left eye during 2 months. His ocular and family history were unremarkable, but he had been undergoing medical evaluation for mild pancytopenia for 6 months. He was referred with the diagnosis of possible Coats disease.

Visual acuity was 20/20 OD and 20/200 OS. The anterior segment was normal in both eyes and the right fundus was normal. The left eye displayed retinal vascular occlusion, with telangiectasia, intraretinal edema, and hard exudates in the posterior pole. The patient had intraretinal and preretinal macular hemorrhages (Figure 1A). Fluorescein angiography (Figure 1B) and optical coherence tomography supported the ophthalmoscopic findings.

Treatment included 2 sessions of argon green laser photocoagulation to the region of retinal nonperfusion and telangiectasia and a single intravitreal injection of triamcinolone (4 mg/0.1 mL) for the macular edema in the left eye. On follow-up 1 year later, both eyes were found to have peripheral retinal ischemia with vascular sheathing, mild telangiectasia, and intraretinal hemorrhages (Figure 1C). Argon laser photocoagulation was directed to the areas of retinal nonperfusion in both eyes.

Figure 1. An 11-year-old boy had poor vision in his left eye and was found to have unilateral retinal vasculopathy. This progressed to involve the fellow eye one year later. A. The left eye showed retinal ischemia with telangiectasia, subretinal fluid, intraretinal edema, and hard exudates in the macula, with preretinal macular hemorrhage. B. Fluorescein angiography of the left eye confirmed retinal microvascular occlusion with “light bulb” telangiectasia. C. One year later, the right eye developed peripheral retinal ischemia and vascular sheathing in the temporal periphery. D. Fluorescein angiography demonstrated peripheral nonperfusion, retinal vascular staining, and minimal telangiectasia.


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Additional Contributions: J. Raymond Buncie, MD, FRCS, referred 1 patient and David Smith, MD, FRCS, referred 2.
Meanwhile, systemic evaluation revealed progression of pancytopenia to bone marrow failure during 12 months. Concurrently, patchy cutaneous hypopigmented macules on the back, ridged finger nails, and longitudinal furrows on the ventral surface of the hands and feet suggested the diagnosis of dyskeratosis congenita (Figure 2A and B). Genetic analysis revealed abnormality in chromosome Xq28, confirming the diagnosis. Allogeneic stem cell transplantation was performed for the bone marrow failure, with recovery of bone marrow function at 6-months’ follow-up.

Comment. Dyskeratosis congenita is a multisystem disorder classically inherited as an X-linked recessive trait, occasionally as an autosomal dominant trait. Mutation in the dyskeratosis congenita gene 1 (DKC1) at Xq28 results in dysfunction of dyskerin, a protein involved in telomere maintenance and ribosomal biogenesis.2 Poor telomere function affects rapidly dividing cells in the epithelium, bone marrow, and skin and nails, resulting in the multisystem manifestations.

Most clinical abnormalities in dyskeratosis congenita appear during infancy or childhood. The most common manifestations are cutaneous alterations and bone marrow failure.1 Retinal changes are rare and include hemorrhages, nerve fiber layer infarction, arteriosclerosis, macular edema, preretinal fibrosis, and optic atrophy.3,4

Our patient initially had unilateral posterior pole retinal vasculopathy that was originally considered to be possible Coats disease. However, the unilateral retinal vasculopathy progressed to bilateral peripheral vasoocclusive retinopathy with only minimal telangiectasia, findings quite different from typical Coats disease. This case illustrates that retinal involvement can be an early manifestation of dyskeratosis congenita and that the course of retinal vasculopathy progresses parallel to progressive pancytopenia and bone marrow failure.

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**Regression of Extrafoveal Choroidal Osteoma Following Photodynamic Therapy**

Choroidal osteoma is a rare intraocular tumor, composed of mature calcified bone and typically found in young adult women.1,2 Long-term findings of this benign tumor include tumor growth in 51%, tumor decalcification in 46%, development of choroidal neovascularization in 31%, and visual acuity of 20/200 or worse in 56%.3 Tumor decalcification usually occurs spontaneously after many years.3 Laser photocoagulation for choroidal neovascularization can induce focal tumor decalcification.4 Decalcification is important because this minimizes further growth of the tumor at that site and could protect the fovea from tumor involvement and visual loss.5 We report photodynamic therapy to an extrafoveal choroidal osteoma that induced decalcification and resorption of bone, leaving a small area of subretinal fibrosis.

**Report of a Case.** An asymptomatic 25-year-old woman had a juxtapapillary choroidal osteoma in her right eye, measuring 3 mm in diameter. Her visual acuity was 20/20 in each eye. An overlying subretinal hemorrhage was present (Figure 1). Ultrasonography showed an echodense mass consistent with calcium. Observation was advised.

On follow-up at 6 and 18 months, the stable mass showed persistent hemorrhage, suggesting choroidal neovascularization. Photodynamic therapy to the entire osteoma was performed using a single 83-second laser spot at 689 nm (50 J/cm²) coupled with intravenous verteporfin, 6 mg/m². The hemorrhage resolved by 1 month. The bone remained intact until 9 months, when there was complete disappearance of the osteoma, leaving a 2-mm region of subretinal fibrosis and retinal pigment epithelial hyperplasia (Figure 2).

**Comment.** The reasons for vision loss from choroidal osteoma include photoreceptor degeneration, subfoveal fluid, and subfoveal hem-