Meanwhile, systemic evaluation revealed progression of pancytopenia to bone marrow failure during 12 months. Concurrently, patchy cutaneous hypopigmented maculae 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. 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. Retinal changes are rare and include hemorrhages, nerve fiber layer infarction, arteriosclerosis, macular edema, preretinal fibrosis, and optic atrophy. 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 vaso-occlusive 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. 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%. Tumor decalcification usually occurs spontaneously after many years. Laser photocoagulation for choroidal neovascularization can induce focal tumor decalcification. 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. We report photodynamic therapy to an extrafoveal choroidal osteoma that induced decalcification and resorption of bone, leaving a small area of sub-retinal fibrosis.

Report of a Case. An asymptomatic 25-year-old woman had a juxtapappillary 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-
orrhage from related choroidal neovascularization. In an analysis of 74 eyes with choroidal osteoma, good visual acuity (20/20-20/40) was found in 24 of 30 eyes (80%) without foveolar involvement of tumor compared with 20 of 44 eyes (45%) with foveolar involvement. In addition, tumors with decalcification showed little or no growth in the direction of the decalcification. Based on these observations, inhibition of tumor growth into the fovea by stimulating decalcification within the entire tumor or at least the portion of the tumor nearest the fovea could protect long-term visual acuity.

Decalcification of choroidal osteoma appears as an atrophic flat bed of retinal pigment epithelium and choroid. Decalcification typically occurs spontaneously and initiates in the central part (37%), peripheral part (48%), or diffusely (15%) within the osteoma. Progressive decalcification occurs slowly and occupies approximately 40% of the tumor by 10 years. Laser photocoagulation can induce focal tumor decalcification. In this report, we observed that photodynamic therapy caused decalcification and involution of the entire tumor. This finding is important because photodynamic therapy could be a therapeutic modality for choroidal neovascularization and induction of decalcification of extrafoveal osteoma to prevent tumor growth into the fovea.

These results should not be extrapolated to subfoveal choroidal osteoma. It is speculated that decalcification of subfoveal choroidal osteoma could result in worse visual acuity because of loss of retinal pigment epithelium and choroidal perfusion.

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Orbital Inflammatory Disease After Intravenous Infusion of Zoledronate for Treatment of Metastatic Renal Cell Carcinoma

Zoledronate, a bisphosphonate, is an inhibitor of osteoclastic bone resorption, indicated for treatment of osteolytic bone lesions from multiple myeloma and other solid tumors. The most common adverse effect of zoledronate is a transient flu-like syndrome. Ocular adverse effects of bisphosphonates include conjunctivitis, uveitis, episcleritis, and scleritis. Cases of orbital inflammatory disease have been reported after treatment with another bisphosphonate, pamidronate sodium. There have been no reported cases of orbital inflammatory disease after treatment with zoledronate. We describe a case of a man developing orbital inflammation after intravenous treatment with zoledronate for bone-involving metastases from renal cell carcinoma.

Figure 2. Findings 9 months after single photodynamic therapy: the hemorrhage and mass were completely resolved, leaving only subretinal fibrosis and retinal pigment epithelial hyperplasia (A); ultrasonography showed no evidence of calcium (B); and optical coherence tomography showed the subretinal tissue with shadowing from retinal pigment epithelial hyperplasia and transmission through fibrosis (between arrows) (C).