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Retinal Vasculitis and Choroidopathy in Pediatric-Onset Mixed Connective Tissue Disease

To the Editor

Mixed connective tissue disease (MCTD) is a distinct clinical entity in which subgroups of patients have overlap of systemic lupus erythematosus, systemic sclerosis, or dermatomyositis/polymyositis.^{1,2} Retinal vasculitis has only rarely been reported in MCTD. We present a case MCTD with bilateral retinal vasculitis and choroidopathy.

An 18-year-old African American young woman was diagnosed as having MCTD at the age of 13 years, based on the Kasukawa criteria³ after initially presenting with chest pain, dyspnea, pericarditis, pulmonary hypertension, and the following laboratory values: antinuclear antibody of 1:1640, ribonucleoprotein of 97.5 ELISA unit (EU)/mL (reference range, 0–15.9 EU/mL), SSA/Ro >100 EU/mL (reference range, 0–15.9 EU/mL), and SSB/La >100 EU/mL (reference range, 0–15.9 EU/mL). The double-stranded DNA Smith and antiphospholipid were negative. Five years after diagnosis, she developed dizziness, blurred vision with floaters, left-sided hearing loss, and tinnitus. Repeat laboratory tests included an elevated Western erythrocyte sedimentation rate of 60 mm/h, with negative infectious titers (herpesvirus, human immunodeficiency virus, and toxoplasmosis). On examination, she had no light perception in the right eye and 20/100 visual

acuity in the left eye. An afferent pupillary defect was noted in the right eye, with clear vitreous, whereas vitritis was noted in the left eye. Bilateral optic nerve head edema with peripapillary hemorrhages was detected on dilated fundus examination. There was a pale macula in the right eye and diffuse retinal hemorrhages throughout the retina consistent with a combined central retinal artery occlusion and central retinal vein occlusion. In both eyes, retinal vessels had perivascular inflammation, sheathing, and leakage. Bilateral inferior serous retinal detachments were confirmed by B-scan ultrasound with bilateral choroidal thickening. Magnetic resonance imaging/magnetic resonance angiography of the brain and neck showed a right-sided retinal detachment and labyrinthitis. A vitreous biopsy was negative for infectious material. She was diagnosed as having

MCTD-associated retinal vasculitis with choroidopathy (Fig. 1) and was treated with methylprednisolone 1000 mg intravenously administered daily and azathioprine for 5 days. Because of her lack of clinical improvement, cyclophosphamide was initiated (0.5 g/m² with escalation of dose to 1 g/m²) for a total of 6 doses and methylprednisolone 30 mg/kg with a maximum dose of 1000 mg intravenously once monthly. Two weeks after initiation of cyclophosphamide, her vision in the left eye had improved to 20/20 with resolution of the retinal vasculitis and choroidopathy (Fig. 2).

Mixed connective tissue disease is rare in children and can have a variable presentation at onset.⁴ Retinal vasculitis has been described in MCTD, but it is more commonly seen with systemic lupus erythematosus (SLE).⁵ Several cases of retinal vasculitis associated with combined central retinal artery occlusion and central retinal vein occlusion in SLE have been reported.⁶ Combined central retinal artery and vein occlusion with choroidopathy has been reported in a single patient with SLE but has not been described in MCTD.⁷ To our knowledge, this is the first report of retinal vasculitis and choroidopathy in a patient with pediatric-onset MCTD. A high index of suspicion and urgent ophthalmologic evaluation and treatment are critical to optimize visual outcomes in MCTD.

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FIGURE 2. Retinal montage of the patient's left eye 2 weeks after initiation of cyclophosphamide showing resolution of retinal vasculitis and choroidopathy.

FIGURE 1. Retinal montage of the patient's left eye 1 week after onset of symptoms showing peripheral retinal vasculitis (asterisk) and choroidopathy indicated by resolving macular edema with exudates in a star pattern (arrow).

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