

RETINAL FINDINGS IN A CASE OF PRESUMED CUTIS MARMORATA TELANGIECTATICA CONGENITA

Eman A. Taleb, MD, FICO, Manish P. Nagpal, DO, MS, FRCS,
Navneet S. Mehrotra, DNB, Kalyani Bhatt, DNB

Purposes: To correlate the clinical picture and fundus fluorescein angiography findings in case of presumed cutis marmorata telangiectatica congenita.

Methods: A 41-year-old woman with presumed cutis marmorata telangiectatica congenita who is a known case of hypothyroidism for the last 6 years and has bilateral peripheral retinal vascular abnormalities, peripheral retinal nonperfusion on fluorescein angiography and bilateral optic disk drusen.

Result: A patient presented with blurring of vision in both eyes for last 6 months. On examination, livedo reticularis skin lesions in both upper and lower extremities were noted. Best-corrected visual acuity in the right eye was 6/18 and left eye was 6/36. Slit-lamp examination revealed posterior subcapsular cataract in the right eye. Fundus examination showed bilateral optic disk drusen, tortuous blood vessels, and peripheral fan-shaped sclerosed neovascularization. Fundus fluorescein angiography showed peripheral retinal nonperfusion. She underwent peripheral laser photocoagulation of the left eye and cataract surgery in the right eye. After 2 years of follow-up, her best-corrected visual acuity was 6/9 in both eyes. Her fundus examination and fundus fluorescein angiography findings were stable in both eyes.

Conclusion: Presumed cutis marmorata telangiectatica congenita is a rare cutaneous vascular disorder that can manifest with nonprogressive retinal vascular abnormality and optic disk drusen.

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From the Retina Department, Retina Foundation, Ahmedabad, India.

Cutis marmorata telangiectatica congenita (CMTC) is a rare congenital vascular disorder with variable and rare ocular involvement. It has been reported in association with glaucoma usually congenital glaucoma,^{1,2} bilateral congenital retinal detachments,³ bilateral tractional retinal detachments secondary to proliferative vitreoretinopathy,⁴ and retinoblastoma.⁵

Cutis marmorata telangiectatica congenita was first recognized and described in 1922 by van Lohuizen.⁶ Fewer than 100 cases of CMTC have been published worldwide. Petrozzi et al⁷ reported the first case of CMTC in the United States in 1970.

The pathophysiology is still unclear, with most cases occurring sporadically, although rare cases were reported in families. Studies indicated the primary involvement of capillaries, venules, and veins, and possibly also that of arterioles and lymphatics.

Associated abnormalities include the following: 1) body asymmetry (extremities; macrocephaly), 2) glaucoma, 3) cutaneous atrophy, 4) neurological anomalies, 5) vascular anomalies (nevus flammeus/Sturge-Weber/Klippel-Trénauna Adams Oliver syndrome), 6) psychomotor and/or mental retardation, 7)

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Reprint requests: Manish P. Nagpal, DO, MS, FRCS, Retina Foundation, Asopalav Eye Hospital, Rajbhavan Road, Underbridge, Shahibaug, Ahmedabad 380 004, India; e-mail: drmanishnagpal@yahoo.com



Fig. 1. Livedo reticularis skin lesion found in both upper and lower extremities.

chronic ulceration that can complicate long-term CMTC, and 8) chronic urticaria.⁸

We describe a case of presumed CMTC who is a known case of hypothyroidism for last 6 years. She had upper and lower limbs livedo reticularis since birth with similar skin lesions in her daughter and grandmother. Ocular examination showed bilateral peripheral retinal vascular abnormalities, retinal

nonperfusion on fluorescein angiography, and bilateral optic disk drusen.

Case Report

A 41-year-old woman presented to our hospital on May 2012 with complaints of blurring of vision in both eyes for the last 6 months. She was a known case of hypothyroidism, with no known cause, and was taking tab thyroxin for last 6 years. On examination, we noted livedo reticularis skin lesions in both upper and lower extremities (Figure 1). Her best-corrected visual acuity in the right eye was 6/18 and left eye was 6/36. Slit-lamp examination revealed posterior subcapsular cataract in the right eye. Fundus examination showed bilateral optic disk drusen, tortuous blood vessels, and peripheral fan-shaped sclerosed neovascularization (Figure 2).

Optic disk drusen were shown in the auto fluorescence image (Figure 3). Fundus fluorescein angiography of both eyes showed bilateral multiple fan-shaped sclerosed neovascularization with leakage of the dye in the late phase and extensive areas of peripheral retinal nonperfusion (Figures 4 and 5). Optical coherence tomography of the macula in both eyes was within normal limits. Intraocular pressure was 18 mmHg in both eyes with open angles on gonioscopy.

The patient gave a history of similar skin lesions in her grandmother and her daughter since birth. She was a known case of hypothyroidism for 6 years. Two brothers and four sisters also had hypothyroidism with no definite cause (idiopathic hypothyroidism).

From the clinical findings and the fundus picture with family history of the same skin lesion in different generations of the family, we diagnosed the patient as a case of presumed CMTC, and we advised her for left eye peripheral laser and right eye cataract surgery.

She underwent left eye peripheral laser photocoagulation in April 2013 and cataract surgery in the right eye in June 2014. On August 2014, her best-corrected visual acuity was 6/9 in both eyes. Her fundus examination and fundus fluorescein angiography findings were stable (Figure 1). We advised the patient for regular 6 months' follow-up.

Discussion

Ocular involvement in CMTC is rare and variable. Glaucoma,^{1,2} bilateral congenital retinal detachments,³

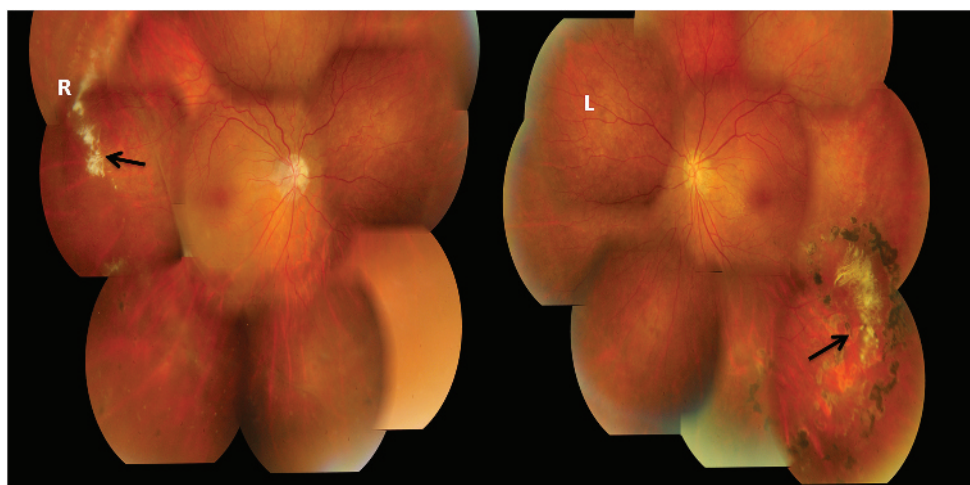


Fig. 2. Fundus photograph of the right and left eyes showing peripheral fan-shaped sclerosed neovascularization with laser marks in the left eye (black arrows).



Fig. 3. Auto fluorescence image of the optic nerve drusen.

bilateral tractional retinal detachments secondary to proliferative vitreoretinopathy,⁴ and retinoblastoma have been reported. Soohoo et al⁹ reported a case of vascular abnormalities and retinal nonperfusion in fundus fluorescein angiography findings, and they treated with laser photocoagulation.⁹ In our case, we have treated one eye with laser photocoagulation, and we just observed the other eye, and during patient follow-up from May 2012 to August 2014, the best-corrected visual acuity and ocular examination were stable in both treated and nontreated eyes. Genetic testing may help to further describe any correlation of the ocular and systemic disease. We are the first to report optic disk drusen in case of presumed CMTC.

Key words: cutis marmorata telangiectatica, peripheral neovascularization, optic disk drusen.

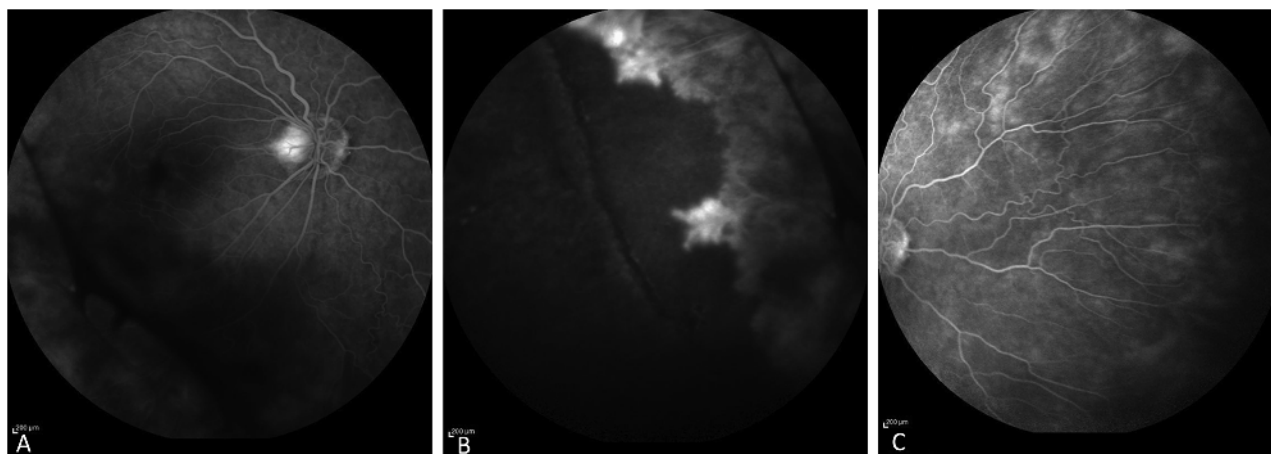


Fig. 4. Fundus fluorescein angiography of the right eye showing multiple fan-shaped sclerosed neovascularization with leakage of the dye in the late phase and extensive areas of peripheral retinal nonperfusion.

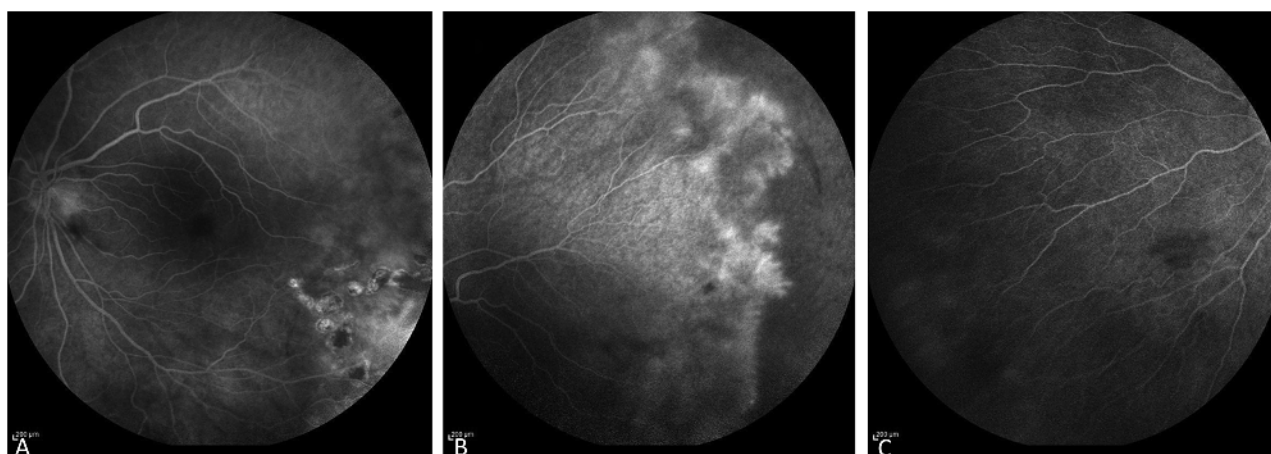


Fig. 5. Fundus fluorescein angiography of the left eye showing multiple fan-shaped sclerosed neovascularization with leakage of the dye in the late phase surrounded with laser marks and extensive areas of peripheral retinal nonperfusion.

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