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Case report
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A rare presentation of systemic lupus erythematosus in a paediatric patient: a case report

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Abstract

Background: Systemic lupus erythematosus (SLE) is the prototype of a multisystem autoimmune disease. We report a rare case of severe vaso-occlusive retinopathy as the first presenting feature of SLE in a child.
Case presentation: An 11-year-old girl presented with sudden blurred vision in the right eye for 1 day. Visual acuity in the right eye was counting fingers. The patient was diagnosed with combined central retinal artery and vein occlusion in the right eye. Fundus examination of the left eye was normal. On day 2, vision in the right eye deteriorated to no light perception and the left eye's fundus developed Roth spots. Laboratory findings were suggestive of SLE. The patient was comanaged by a multidisciplinary team consisting of paediatrics, rheumatology, and ophthalmology.
Conclusion: We highlight the significance of early detection and multidisciplinary approach in the management of this patient to protect the fellow eye and for systemic control of inflammation.

Keywords: central retinal artery occlusion, central retinal vein occlusion, paediatric systemic lupus erythematosus

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Penyakit sistemik lupus eritematosus pada pesakit pediatrik: laporan kes

Abstrak

Latar belakang: Sistemik lupus eritematosus (SLE) adalah sejenis penyakit autoimun yang melibatkan pelbagai sistem di dalam badan. Kami melaporkan kes retinopati vaso oklusif yang teruk sebagai tanda pertama SLE yang jarang berlaku pada kanak-kanak.

Laporan kes: Seorang kanak-kanak perempuan berusia 11 tahun mengalami kekaburan penglihatan mata kanan secara tiba-tiba dalam tempoh masa satu hari. Tahap penglihatan mata kanan pesakit tersebut adalah hanya dapat mengira jari (counting fingers). Diagnosis oklusi arteri dan vena retina utama pada mata kanan telah dibuat. Pemeriksaan mata kiri adalah normal. Pada hari kedua, penglihatan mata kanan merosot kepada tiada persepsi terhadap cahaya dan retina mata kiri muncul tanda baru bintik Roth. Keputusan ujian makmal condong ke arah SLE. Rawatan pesakit ini diuruskan oleh pelbagai disiplin yang terdiri daripada pasukan pediatrik, reumatologi dan oftalmologi.

Kesimpulan: Kami menekankan kepentingan pengesanan awal dan kerjasama dengan pelbagai disiplin dalam pengurusan rawatan pesakit untuk melindungi penglihatan yang sedia ada pada pesakit dan untuk kawalan radangan sistemik.

Kata kunci: lupus eritematosus sistemik kanak-kanak, oklusi arteri utama retina, oklusi vena utama retina

Introduction

Systemic lupus erythematosus (SLE) is the prototype of a multisystem disease of autoimmune origin, characterized by a vast array of autoantibodies, particularly antinuclear antibodies. Although the cause remains unknown, a defect in the mechanism of immunological self-tolerance has been inferred due to the presence of auto antibodies in all patients.[1]

SLE has a prevalence as high as 1 in 2,500 in certain populations. It predominantly affects females, with a frequency of 1 in 700 among women of childbearing age and a female-to-male ratio of 9:1. When the disease develops during childhood, the female-to-male ratio is 2:1.

Other than antinuclear antibodies, antiphospholipid antibodies are present in 40% to 50% of SLE patients, and these are directed against epitopes of plasma proteins when the proteins are in complex with phospholipids: prothrombin, annexin V, B_2-glycoprotein I, protein S, and protein C. These patients have complica-

tions associated with a hypercoagulable state, *i.e.*, venous and arterial thrombosis which may be associated with ocular ischemia. The most common clinical manifestation of SLE is haematological (100%), while the least common is ocular (15%).

The ocular manifestations of SLE can range from mild (*e.g.*, conjunctivitis, scleritis, anterior uveitis) to vision-threatening (*e.g.*, severe vaso-occlusive retinopathy, vitreous haemorrhage secondary to proliferative retinopathy, lupus choroidopathy, choroidal effusion, choroidal infarction, optic neuritis, anterior or posterior optic neuropathy, cortical infarcts). The incidence of SLE-associated retinopathy has been reported to be up to 29% among adult patients.[2] Severe occlusive retinopathy has been found in less than 1% of patients.[3]

We have found 2 cases in the literature reporting on paediatric patients with combined central retinal artery occlusion (CRAO) and central retinal vein occlusion (CRVO).[4,5] However, in both these cases reported, the patients had other systemic symptoms on first presentation, such as chest pain, fever, arthritis, and ulcers. We report a case of combined CRAO and CRVO in a paediatric patient as the first presenting feature of SLE.

Case presentation

An 11-year-old girl presented with sudden, painless blurred of vision in the right eye for a day, which worsened on the next day. Otherwise, she denied other systemic symptoms. There was no significant medical or surgical history, nor any family history of malignancy.

On arrival, examination of the right eye showed visual acuity of counting fingers with positive relative afferent pupillary defect, normal anterior segment, and a swollen optic disc with generalized retinal haemorrhages on a pale retina. The retinal arteries were also attenuated with cattle-track sign and the retinal veins were dilated and tortuous. Optical coherence tomography of the macula showed thickened inner retina layers with intraretinal fluids (Fig. 1). Examination of the left eye was unremarkable. The right eye was diagnosed with combined CRAO and CRVO.

Even though more than 48 hours had passed from the onset of symptoms, we performed the emergency manoeuvres indicated for the management of acute CRAO, consisting of ocular massage, rebreathing into a plastic bag, and a 500-mg dose of oral acetazolamide. However, there was no improvement in visual acuity. Vision in the right eye deteriorated to no light perception in all 4 quadrants on the next day with similar fundus findings (Fig. 1). Vision in the left eye was 6/9, but 2 Roth spots were found in the superotemporal and inferonasal arcades near the optic disc (Fig. 1).

Laboratory results returned pancytopenia (white cell count 2×10^9/L, haemoglobin 9.1 g/dL, platelets 50×10^9/L), deranged coagulation profile, hypalbuminaemia, transaminitis, increased erythrocyte sedimentation rate (118 mm/hr), increased

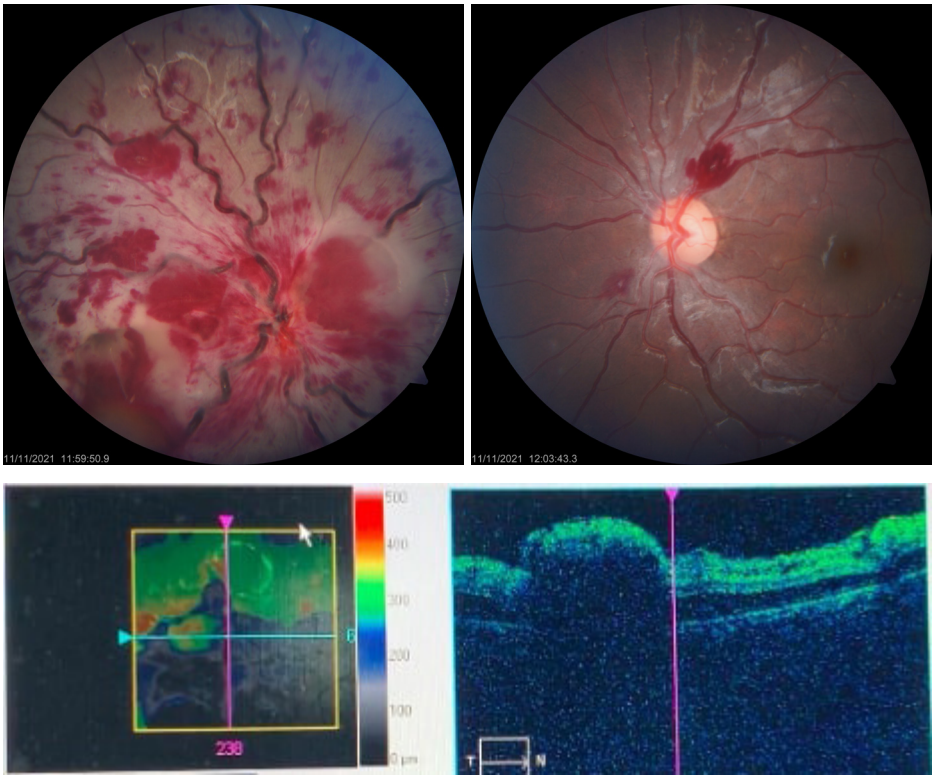

Fig. 1. Fundus photographs on day 2 of presentation of the *(a)* right eye and *(b)* left eye. *(c)* Optical coherence tomography of the right eye's macula on first presentation

C-reactive protein (15.9 mg/L), high ferritin levels, and elevated fibrinogen. In addition, there was also heavy proteinuria of 528 mg/day. Peripheral blood film revealed normochromic normocytic red cells with occasional polychromatic cells. Direct Coombs test was positive. Infective screening consisting of hepatitis B and C, venereal disease research laboratory test (VDRL), human immunodeficiency virus (HIV), toxoplasma, rubella, cytomegalovirus, and parvovirus B19 returned negative. Fasting lipid profile was normal.

SLE diagnosis was confirmed from positive activated partial thromboplastin time-lupus anticoagulant (36.1 seconds), dilute Russell viper venom test (41.2 seconds), ribonucleoprotein antibodies, anti-Sm antibodies, high ANA (1:1280), and ds-DNA antibodies with low C3, C4 complement (0.24 g/L, 0.05 g/L, respectively).

Fundus fluorescein angiography of the right eye confirmed prolonged choroidal phase of 24 seconds, delayed filling of the central retinal artery, complete CRVO, and extensive area of capillary dropout seen in the posterior pole. There was no angiographic evidence of vasculitis or capillary fallout in the left eye. Magnetic resonance

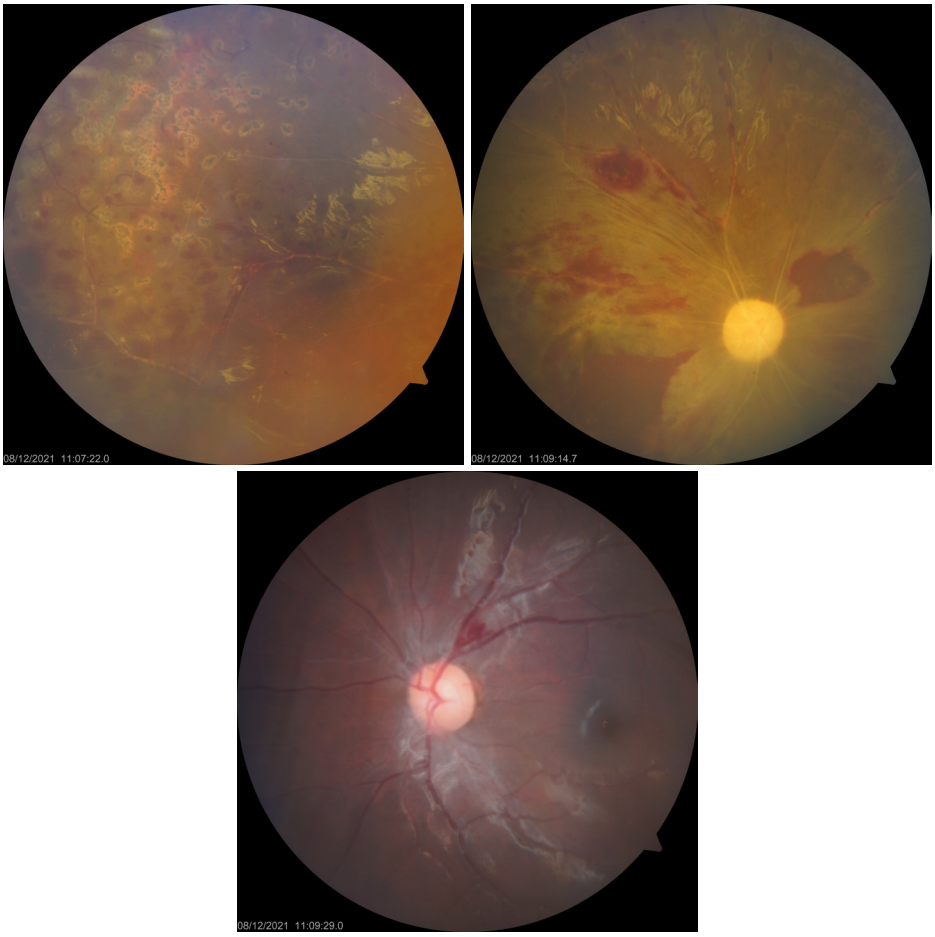

Fig. 2. Fundus photographs *(a)* after 1 month; *(b)* right eye after full PRP; *(c)* resolving flame-shaped haemorrhage with no new bleeding in the left eye.

imaging, angiography, and venography of the brain showed no evidence of intracranial bleed, space-occupying lesions, or thrombosis. Ultrasonography of the carotid arteries was also normal.

The patient was referred to a tertiary centre for management by a multidisciplinary team consisting of paediatrics, rheumatology, and ophthalmology. Intravenous (IV) pulse methylprednisolone 500 mg for 3 days was started immediately. In view of vision threatening vaso-occlusive vasculitis in the right eye, 2 cycles of IV cyclophosphamide 800 mg (500 mg/m^2) were completed, 1 month apart. In keeping with the suspicion of antiphospholipid syndrome, anticoagulation with subcutaneous clexane 60 mg twice a day was started and continued on discharge. Systemic immu-

nosuppression was continued with IV methylprednisolone maintenance of 75 mg twice a day and gradually tapered over a period of 1 week before converting to oral prednisolone 20 mg twice a day. Oral enalapril was commenced for its dual antiproteinuric and antihypertensive effect. Three sessions of full panretinal photocoagulation (PRP) were administered to ameliorate the ischemic complications of SLE retinopathy.

At the 1-month follow-up, visual acuity remained at no light perception in the right eye and 6/9 in the left eye. Anterior segments in both eyes were normal, no rubeosis iridis seen. The right eye's fundus showed a pale optic disc and improvement in previous areas of preretinal and intraretinal haemorrhages with generalised sclerosed vessels (Fig. 2a, b). In the left eye, intraretinal haemorrhage was resolving with no new bleeding or cotton wool spots (Fig. 2c).

Eight months later, although the left fundus remained normal, the patient developed uncontrolled intraocular pressure (IOP) between 29 and 34 mmHg secondary to steroid response in the left eye. She was prescribed maximal antiglaucoma therapy in eye drops in addition to 250 mg oral acetazolamide 4 times a day, as well as 30 ml syrup glycerol 3 times a day. She was not fit for trabeculectomy due to refractory thrombocytopenia (platelet 6 x × 10^9/L). Hence, trans-scleral diode cyclophotocoagulation was performed in the left eye. Nine months after diagnosis, the patient also developed iris neovascularization and increased IOP in the right eye, subsequently leading to tractional retinal detachment, microhyphaema, and vitreous haemorrhage secondary to refractory thrombocytopenia.

Discussion

Vasculitis is believed to be the single most important process and the starting point in the pathogenesis of tissue and organ damage in SLE. Perivascular inflammatory infiltrates have been demonstrated in eyes with SLE vasculitis. Immunoglobulin and complement deposits have been demonstrated in the retinal blood vessel walls, ciliary body, choroid, episclera, corneal basement membrane, and limbal vascular endothelial cells of SLE patients.[6] Luminal narrowing of vessel walls through intramural immune complex deposits with fibrinoid changes causing vaso-occlusion has been described in SLE vasculopathy.[7]

To the best of our knowledge, this is the first case report that describes combined CRAO and CRVO as the first presentation of SLE in a paediatric patient without any other systemic symptoms. A few similarities between our case and similar reported cases[4,5] include prompt commencement of IV steroid pulse therapy (methylprednisolone), panretinal photocoagulation, and maintenance of low-dose steroid therapy (methylprednisolone) and immunosuppressants (cyclophosphamide).

Visual prognosis is generally poor in combined CRAO and CRVO. The risk of iris neovascularization is approximately 75%, which our patient developed 9 months

after being diagnosed with combined CRAO and CRVO. Moreover, prolonged systemic steroid use may lead to ocular complications, which further complicated our patient's management. She developed uncontrolled IOP in the left eye despite maximal medical therapy for glaucoma and had to undergo trans-scleral diode cyclophotocoagulation.

Aggressive treatment with PRP is recommended.[8] In exceptional cases of combined CRAO with CRVO, a patient may manifest spontaneous improvement.[9] Unfortunately, our patient subsequently developed tractional retinal detachment, microhyphaema, and vitreous haemorrhage secondary to refractory thrombocytopenia in the right eye.

Conclusion

Prompt systemic and local treatment of underlying SLE successfully halted progression in the fellow eye in our patient.

Declarations

Informed consent for publication
The patient's mother provided informed consent for the publication of the clinical data and images contained in this case report.

Competing interests
None to declare.

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None to declare.

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