Bilateral Optic Neuropathy in a middle age female in association with Diffuse Systemic Sclerosis

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INTRODUCTION

Systemic Sclerosis is a chronic connective tissue autoimmune disorder involving multiple systems in a progressive manner, characterized by fibrosis of the skin and internal organs, and vasculopathy. It affects mainly musculoskeletal, gastrointestinal, renal and cardiopulmonary systems. It is a sporadic disease with worldwide distribution. The hallmark of systemic sclerosis is thickening and fibrosis of the skin which is due to excessive collagen production. In the early stages, evidence of inflammation, autoimmunity and altered microvascular function are common. Over time, progressive and irreversible structural changes in small blood vessels and fibrosis in multiple organs are seen. Ocular involvement is rare which mainly results as a complication of the disease. It may involve anterior to posterior segment, orbit and extraocular muscles.

CASE ILLUSTRATION

A 37 year old female who is a known case of diffuse systemic sclerosis with interstitial lung disease since seven years and is on irregular treatment for the same, presented with painless loss of vision in her both eyes since two months. Initially there was diminution of vision in her both eyes for one and half months, which then progressed to loss of vision and now she cannot even perceive light. She also has a history of multiple joints pain. There was no history of hypertension, diabetes, tuberculosis or asthma. Menstrual history was normal. There were no similar complaints or any chronic illness in any of the family members.

General physical examination revealed tightening of skin over face, eyelids, trunk and extremities, thickening of the skin over hands and fingers, hypopigmented macules over forehead, hyperpigmented lesion over hands, contractures of the digits and narrowing of oral aperture (figure 1-4). Ocular examination revealed no perception of light in either eye. Pupil of both eyes were mid-dilated sluggishly reacting to light. Rest anterior segment was normal. Intraocular pressure was 17.3 mmHg in both eyes.

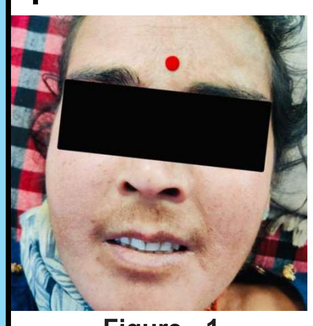


Figure - 1

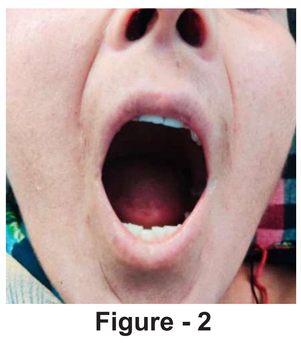


Figure - 2

Figure - 3

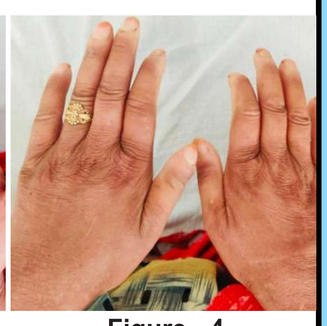


Figure - 4

Fundus examination of both eyes showed pale disc with blurred margins, with obscuration of cup and disc ratio, foveal reflex was present, suggestive of bilateral optic atrophy (figure 5-6).

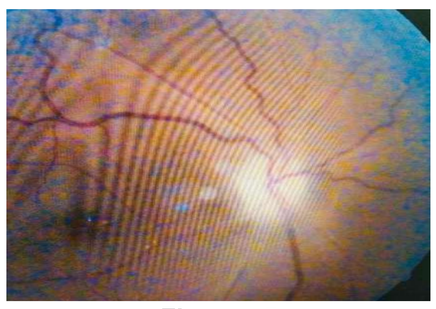


Figure - 5

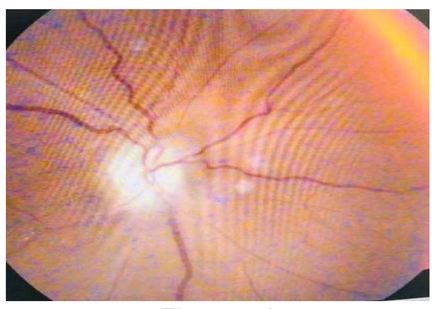


Figure - 6

INVESTIGATION

Systemic investigation revealed normal hemoglobin, total and differential leucocyte count, sugar level and renal function test. C-reactive protein and ESR was raised four fold. ECG was within physiological limits. X-ray chest PA view showed multiple ill-defined opacities in bilateral lung parenchyma suggestive of inflammatory pathology (figure 7). HRCT Chest revealed inter and intra lobular septal thickening with cystic lesions giving honey combing appearance and focal areas of tractional bronchiectasis changes in bilateral lungs suggestive of interstitial lung disease (figure 8). MRI Brain was normal (figure 9) and MRI orbits showed kinking of left optic nerve (figure 10).

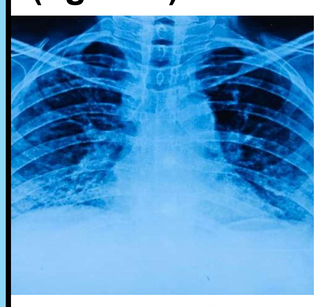
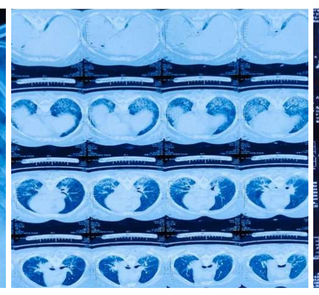


Figure - 7



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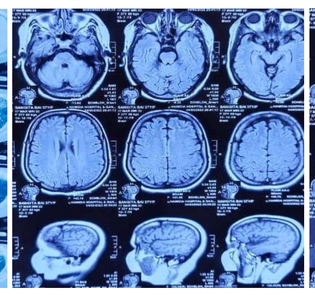

Figure - 9

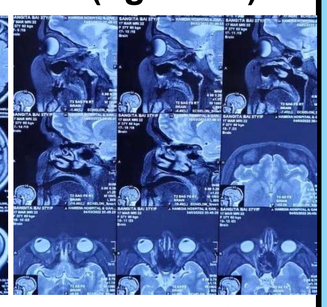

Figure - 10

TREATMENT

Systemic and local findings were suggestive of diffuse cutaneous systemic sclerosis with interstitial lung disease and bilateral optic neuropathy. Systemic steroid was started, prednisolone 60 mg once a day in a tapering dose for a month after consulting with rheumatologist . Along with that systemic immunosuppression with mycophenolate mofetil 500 mg twice a day and supportive treatment of vitamin B12 and folic acid supplements. One month after the treatment, her systemic manifestations were controlled, though she could not gain her vision.

DISCUSSION

Systemic sclerosis is a multi-system, chronic, autoimmune disease characterized by fibrosis of the skin and internal organs and vasculopathy. It affects mainly the women of childbearing age. There are two types of systemic sclerosis based on pattern of skin involvement, associated clinical and laboratory manifestations and natural history. The first one is limited cutaneous which slowly involves the skin of distal extremities and face. The second one is diffuse cutaneous which involves diffusely the skin of extremities, face and trunk and various organs progressively. The pathogenesis involves several genetic and environmental factors characterized by vascular injuries, autoimmune system, visceral and vascular fibrosis. Three abnormalities have been studied in its pathophysiology. A fibroblast dysfunction, which leads to increased deposition of extracellular matrix, tissue hypoxia as a result of vascular abnormality and an altered immune response resulting in an autoantibody production. Ocular manifestations have been reported, but rare. It is due to persistent inflammation leading to vasculopathy, which results in chronic ischemia and increased serum VEGF level. Eyelid skin fibrosis is the most common ocular finding, resulting from pathological collagen deposition in the dermis. In advanced cases, restriction of eyeball mobility may occur. Acute vision loss is rare which is due to autoimmune vasculopathy. This patient developed bilateral vision loss with features of optic neuropathy as evidenced by characteristic fundoscopy findings. Optic disc pallor indicates chronicity, degeneration of axons and loss of ganglion cells. The role of systemic sclerosis in the incidence of optic neuropathy has not been adequately studied but it has been proposed that microvascular changes and inner retinal perfusion abnormalities may play a major role, attributing to inflammatory or ischemic mechanism. The neural retinal tissue has the highest oxygen consumption per volume of the entire body, choroid has the highest blood flow per volume. This renders the retinal and choroidal vasculature susceptible to the vascular changes.

CONCLUSION

Rare ophthalmic manifestation can occur in systemic sclerosis in the form of bilateral optic neuropathy leading to grave irreversible visual loss. Routine ophthalmic and detailed fundus examination in patients with systemic sclerosis can help in characterization of less frequently reported posterior segment features of this disease. Ocular involvement is a sensitive marker for the onset or exacerbation of an immune reaction in systemic sclerosis. Hence clinicians must be aware of the diverse spectrum of ocular manifestations and accordingly include ocular investigation for the disease. Systemic sclerosis, though uncommon, is the most severe connective tissue disease with highest disease related mortality and morbidity. Early recognition and treatment can prevent sight threatening complications and help in maintaining the quality of life.

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