Two cases of scleroderma

Early recognition, referral and treatment for scleroderma are important to help maintain and preserve quality of life, writes Rachel Quigley

SCLERODERMA, ALSO KNOWN AS SYSTEMIC SCLEROSIS is an autoimmune condition resulting in abnormal growth of connective tissue, causing fibrosis and vascular damage. It affects more women than men and the usual age of onset is 25-55 years, but it can affect any age group. About 10% of Irish people have Raynaud’s disease and of these some 2% go on to develop scleroderma. It is rare in children and is evenly distributed among all ethnic groups.

There are two types of scleroderma: limited cutaneous systemic sclerosis (lcSSc or limited scleroderma) and diffuse cutaneous systemic sclerosis (dcSSc).

Limited cutaneous systemic sclerosis accounts for 70% of systemic sclerosis cases. It affects only the face, forearms and lower legs up to the knee. It is also known as CREST syndrome (calcinosis, Raynaud’s disease, oesophageal dysmotility, sclerodactyly, telangiectasia). Diffuse cutaneous systemic sclerosis accounts for the remainder and can affect the upper trunk and thighs.

Common features of scleroderma

Common features include Raynaud’s phenomenon, skin thickening and induration, telangiectasia, hypo or hyper-pigmentation and digital ulcers. The skin appears hard and thickened, with sausage-shaped digits. This may restrict joint movement and cause a contracted appearance. The facial skin may be also tightened with perioral tightening causing microstomia. Calcinosis may also be present.

Other features include dyspepsia, shortness of breath, dry eyes and mouth, myalgia, arthralgia, generalised fatigue, weight loss, constipation, bacterial overgrowth causing bloating, malabsorption and diarrhoea.

Case 1

A 65-year-old previously well lady presented with symptoms suggestive of UTI. During the consultation she happened to mention she constantly suffered from dry gritty eyes. She was subsequently asked about dry mouth or cold hands to which she agreed she complained of both. An autoantibody screen demonstrated she was ANA (antinuclear antibody) positive, in particular anticentromere positive. At a follow-up visit she had mild sclerodactyly, a few telangiectasia and complained of upper GI discomfort. She was referred to rheumatology and commenced on mycophenolate mofetil and a calcium channel blocker.

Case 2

A 50-year-old lady attended the emergency department complaining of difficulty swallowing and skin-tightening, especially of the fingers. She was advised to obtain autoantibody screening via her GP. She was noted to be ANA and anticentromere antibody positive. On examination she had digital swelling and skin tightening distal to her MCP joints. She did not have calcinosis or telangiectasia. She was reviewed by a rheumatologist, and commenced on a proton pump inhibitor and a calcium channel blocker.

Discussion

Investigations that can be performed in general practice include an FBC, ESR and biochemistry, including baseline renal function. Autoantibody screen will be positive for antinuclear antibody (ANA). Autoantibody subsets predict the likelihood of limited and cutaneous disease with or without organ involvement.

Antinuclear antibodies are present in about 90-95% patients, usually with a speckled or centromere pattern. Topoisomerase I antibodies (also known as Scl-70) are present in approximately 30% of patients with diffuse disease (absent in limited disease) and are often associated with pulmonary fibrosis. Anticentromere antibodies are present in 45-50% of patients with limited disease and are rare in diffuse disease.

Anti-RNA polymerase I and III antibody is associated with dcSSc and especially with kidney involvement. Antifibrillarin (U3RNP) antibody is associated with heart involvement, pulmonary hypertension, kidney involvement and myositis. Anti-U1RNP (nRNP) antibody is associated with joint involvement and overlap syndromes.

Treatments and complications

All patients should be referred to a rheumatologist for regular monitoring and treatment. Recent emphasis has been on early recognition of the symptoms via the Very Early Diagnoses clinics for Systemic Sclerosis (VEDOSS) initiative in Europe. Treatments include physiotherapy, occupational therapy, home exercises, emollients to skin, PPIs to relieve gastrointestinal symptoms, calcium channel blockers to relieve the symptoms of Raynaud’s disease along with warm or heated clothing.

No treatments have been shown to delay the disease process although mycophenolate, cyclophosphamide and penicillamine are being currently used. The use of these medications will require regular blood monitoring in the community. The disease is associated with complications including pulmonary hypertension, pulmonary fibrosis, scleroderma crisis, GI complications and Sjogren’s syndrome. See www.irishraynauds.com for patient support.

Although rare, this is an important condition which has a negative impact on the lives of patients and families. Early recognition, referral and treatment are therefore important to help maintain and preserve quality of life. Prognosis is variable. For patients with limited involvement, 10-year survival rates are roughly 60-70%. For patients with diffuse disease, 10-year survival rates are 20%.

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