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DIAGNOSTIC CHALLENGES IN SYSTEMIC SCLEROSIS

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Systemic sclerosis, often known as scleroderma, is a chronic autoimmune connective tissue disease characterized by excessive collagen deposition leading to skin fibrosis and various internal organ manifestations, small vessel vasculopathy. It is an uncommon disease with an estimated global prevalence of 17.6 per 100,000 persons and an annual incidence rate of 1.4 per 100,000 people. Research advancements have improved our knowledge of the pathophysiology and clinical presentation of the disease, as well as our possibilities for therapy for what was once believed to be an unpredictable and incurable disorder. Immune response dysregulation is a hallmark of the autoimmune connective tissue disease systemic sclerosis (SSc). Massive tissue fibrosis and peripheral tissue hypoxia are the outcomes of autoantibody formation, alterations in T- and B-lymphocyte activity, and injury to the vascular bed. Thus, increased endothelial damage and widespread collagen deposition in the skin and internal organs cause patients with systemic sclerosis to experience final organ failure. Diagnostics for SSc have evolved significantly in recent years, driven by advancements in serological markers and imaging techniques that help in early detection and targeted interventions to improve patient outcomes and quality of life. The existence of serum autoantibodies directed against various intracellular antigens, these autoantibodies, which can be useful diagnostic markers for SSc, are present in over 95% of people . Although the presence of autoantibodies is one of the characteristics of SSc, there is no single laboratory test to diagnose SSc. In the absence of a diagnostic test proving absence or presence for SSc, several sets of classification criteria have been developed, one of the well known classification is the The European League Against Rheumatism and the American College of Rheumatology (ACR/EULAR) classification criteria which was published in 2013. It was determined that skin thickening of the fingers extending proximal to the metacarpophalangeal joints is sufficient for to classify a patient as systemic sclerosis. In the absence of this finding, seven findings should be identified and score: skin thickening of the fingers, fingertip lesions, telangiectasia, abnormal nailfold capillaries, interstitial lung disease or pulmonary arterial hypertension, Raynaud's phenomenon, and SSc-related autoantibodies. Patients with a score ≥9 are considered to have definite systemic sclerosis (scleroderma). Sensitivity was 91% and specificity was 92% during the validation .Other methods like Nailfold capillaroscopy is used for the evaluation of microcirculation in patients with Raynauds phenomenon. This imaging technique is easy to repeat, non-invasive and inexpensive. It has become a gold standard in diagnostic process of scleroderma. Nailfold capillaroscopy and immunological tests together with physical examination allow distinguishing primary Raynaud’s phenomenon from secondary since Raynaud’s phenomenon remains a typical manifestation of vascular involvement in scleroderma . The final diagnosis of patients suffering from scleroderma can take years to diagnose. As the disease progresses ,the skin manifestations worsen and show typical signs of scleroderma. Since the stages of SSc, endothelial cell (EC) death and damage can result in tissue hypoxia, oxidative stress, and perivascular inflammation , Raynaud's phenomenon, edematous puffy hands, digital ulcers, pulmonary artery hypertension, erectile dysfunction, scleroderma renal crisis, and heart involvement all have a significant impact on survival and quality of life. The most fatal rheumatologic condition accounts for the cause of death in over half of SSc patients.