Motor Disorders Due to Collagen Diseases

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With 2 Figures

1. Progressive Systemic Sclerosis

1.1. Definition and General Data

Progressive systemic sclerosis is a generalized disease of unknown origin, involving many organs (GOETZ, 1945; TUFFANELLI and WINKELMANN, 1961). It produces hardness of the skin, fibrosis, loss of smooth muscle of internal organs and progressive loss of visceral and cutaneous functions (WINKELMANN, 1971). Muscles, bones, heart, lungs, mucous membranes and the gastrointestinal tract may be involved. Several types of systemic sclerosis have been described: (1) Acrosclerosis is the more common form of systemic scleroderma (95%). It is characterized by Raynaud’s phenomenon, which is often the initial symptom. The disease usually begins on the hands and later spreads to other structures. The incidence in women is 2.6 to 2.9 times that in men (LEINWAND et al., 1954; TUFFANELLI and WINKELMANN, 1961; D’ANGELO et al., 1969; POIRIER and RANKIN, 1972). (2) Acute diffuse scleroderma, or malignant scleroderma, is a rare condition (5% of the patients with systemic sclerosis), characterized by generalized cutaneous sclerosis and usually beginning centrally. It has a rapid onset and course, and a poor prognosis. Raynaud’s phenomenon is absent. Malignant scleroderma affects as many men as women.

About 75% of the cases of systemic sclerosis are diagnosed between the ages of 30 and 60 (POIRIER and RANKIN, 1972). The average annual age-specific mortality has been estimated at about 1 per million for males, 2.2 per million for white females and 6.6 per million for negro females (Masi and D’ANGELO, 1967). The characteristic pathological lesions include proliferation of collagen and vascular changes (LEINWAND et al., 1954). In contrast with lupus erythematosus or dermatomyositis there is no or only minimal inflammatory cell infiltration and no evidence of proliferation of fibroblasts (WINKELMANN, 1971). No abnormalities of collagen structure or metabolism have been found (FISHER and RODNAN, 1960; RASMUSSEN et al., 1964; WINKELMANN, 1971). The collagen in scleroderma contains more hexosamine than normal and the levels of hexosamine in blood and urine are elevated as well. This may account for the increased water binding and edema (FLEISCHMAIER and KROL, 1967; WINKELMANN, 1971).

The blood vessels of the affected organs show concentric intimal proliferation, hyperplasia of the media with reduplication of the elastic fibers and adventitial sclerosis (BANCHI et al., 1966). A characteristic feature is the widespread microangiopathy resulting in a disappearance of many capillaries. The remaining capillaries frequently show thickening and reduplication of the basement membrane, and thickening and degeneration of the endothelium (NORTON and NARDO, 1970).
Usually the symptoms begin insidiously. The cutaneous lesions generally precede the visceral symptoms and begin at the hands with subsequent involvement of face, neck and chest. After an initial period of edema a widespread symmetrical leathery induration develops which binds the skin tightly to the subcutaneous tissues. These lesions are followed by atrophy, pigmentation and ulcerations, especially of the hands and fingers; soft tissue calcification in the fingers and absorption of the terminal phalanges, shortening the fingers, also occur (KEMP HARPER and JACKSON, 1965; BIANCHI et al., 1966). Furthermore, calcium may be deposited in pressure areas such as elbows, knees and buttocks. The periodontal membrane is widened in 10 to 36% of the cases (FARMER et al., 1960; KEMP HARPER and JACKSON, 1965). Patchy reticulated pigmentation and telangiectasia of the skin are frequently observed, particularly on the face and upper trunk (CULLINAN, 1953). Arthralgias occur in the majority of cases at some time in the course of the disease (ORBONA and ALBANO, 1958). Systemic sclerosis involves mainly the small joints and may suggest rheumatoid arthritis, but there is no pannus formation in the synovial membrane (RODNAN, 1962) and the articular bone destruction typical of arthritis is lacking.

The esophagus is affected in 50 to 80% of the cases. Whereas gastric lesions are rare, radiological abnormalities of the small intestine, particularly the duodenum, are found in 42% of the patients (BLUESTONE et al., 1969; POIRIER and RANKIN, 1972). A decreased motility, thickening of the mucosal folds with "wire spring" appearances when the loops are not distended with barium, dilatation, segmentation and dilution or flocculation of barium are the most typical radiological images (KEMP HARPER and JACKSON, 1965; PEACHEY et al., 1968). Between 20 and 57% of the patients have symptoms referable to the small intestine (REINHARDT and BARRY, 1962; HEINZ et al., 1963; BLUESTONE et al., 1969). Steatorrhea, if present, is usually moderate and caused by bacterial overgrowth (GREENBERGER et al., 1967; BLUESTONE et al., 1969). The reported incidence of colon involvement varies from 10 to 50%, although most patients have no complaints related to the colon (HALE and SCHATZKI, 1944; KEMP HARPER, 1953; GONDOS, 1960; KAUFMANN et al., 1968). Large mouth square neck diverticula on the antimesenteric border give the colon a typical asymmetrical aspect (HALE and SCHATZKI, 1944; KEMP HARPER and JACKSON, 1965). These diverticula are interspersed with areas of rigidity. Later the entire colon wall becomes involved and atonic and the diverticula disappear.

Respiratory symptoms are due to alveolocapillary block and pulmonary fibrosis, especially in the lower half of the lungs (BIANCHI et al., 1966; D'ANGELO et al., 1969). Repeated infections, respiratory insufficiency, hypoxemia without CO₂-retention and pneumothorax can complicate the condition.

Heart failure is rarely due exclusively to replacement of myocardium with fibrous tissue. Pulmonary hypertension contributes to the right-sided, and systemic hypertension to the left-sided heart failure. Pericardial effusion is occasionally present.

Involvement of the kidneys is frequent and may lead to renal lesions which are indistinguishable from those of malignant hypertension. Malignant hypertension may complicate the course of the disease and rapidly progressing renal insufficiency is a cause of death in between 5 and 20% of the patients. Mixed collagenoses occur in 4% of the patients (POIRIER and RANKIN, 1972). Rare instances of combined scleroderma and systemic lupus erythematosus with a positive LE cell phenomenon have been described (TUFFANELLI and WINKELMANN, 1962; BIANCHI et al., 1966; CLARK et al., 1971b; WINKELMANN, 1971).