Scleroderma Association B.C.

HOW TO DIAGNOSE SYSTEMIC SCLEROSIS

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The diagnosis of systemic sclerosis (SSc, systemic scleroderma) is usually based on the presence of a combination of symptoms and signs typical of systemic sclerosis:

- Raynaud's phenomenon;
- skin thickening or puffy "sausage-like" swelling of the fingers;
- autoantibodies associated with systemic sclerosis in a blood sample;
- abnormalities in small blood vessels at the base of the nails;
- other skin and internal organ involvement associated with systemic sclerosis.

RAYNAUD'S PHENOMENON

The most common and earliest problem observed in systemic sclerosis is Raynaud's phenomenon. This phenomenon is characterized by a change in color of the fingers (typically from a white discoloration to a blue then red color) and is most commonly provoked by exposure to cold. Raynaud's phenomenon can be "primary", meaning that it is isolated and not associated with a systemic autoimmune disease such as systemic sclerosis. However, when Raynaud's phenomenon occurs after the age of 40-50 years or is associated with ulcerations of the fingers (open lesions of the skin that heal very slowly) or other symptoms and signs of systemic sclerosis, a diagnosis of systemic sclerosis should be suspected and sought.

THICKENING OF THE SKIN OF THE FINGERS

Thickening of the skin of the fingers, especially when it extends to the back of the hand, or puffy swelling of the fingers with a sausage-like appearance in the earlier stages, are characteristic signs of systemic sclerosis.

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AUTOANTIBODIES

Systemic sclerosis is an autoimmune disease in which the immune system becomes dysfunctional and turns against its own cells. Evidence of this autoimmunity can be found by the presence of autoantibodies in the blood, i.e., antibodies directed against oneself. Several autoantibodies specific to systemic sclerosis have been identified, including anti-centromere, anti-topoisomerase I (Scl 70) and anti-RNA polymerase III. Their presence, detected by a blood test, supports the diagnosis of systemic sclerosis when it is associated with Raynaud's phenomenon and other symptoms and signs of systemic sclerosis.

NAILFOLD CAPILLARY ABNORMALITIES

Systemic sclerosis is also a disease of the small blood vessels. Abnormalities of these small vessels, or "capillaries", can be seen at the base of the nails. Specialized examination by high magnification microscopy of the capillaries in the nail bed, or "capillaroscopy", is often useful to support a diagnosis of systemic sclerosis.

OTHER SKIN AND INTERNAL ORGAN INVOLVEMENT

Systemic sclerosis can also present with skin ulcerations on the fingertips, telangiectasias (dilations of small blood vessels that form red or sometimes purple spots on the surface of the skin) and calcinosis (small, white bumps of calcium deposits under the skin). Systemic sclerosis can also affect the digestive, pulmonary, cardiac and renal systems. In the presence of other symptoms and signs suggestive of systemic sclerosis, these additional manifestations support the diagnosis of systemic sclerosis.

CLASSIFICATION CRITERIA

In scientific research, classification criteria are used to standardize the definition of systemic sclerosis (see Table below for the classification criteria issued jointly in 2013 by the American College of Rheumatology and the European League Against Rheumatism). Patients with a score of at least 9 points are classified as having systemic sclerosis. However, a diagnosis of systemic sclerosis (often at an early stage) can be made in a patient who does not meet the classification criteria.

SUMMARY

In summary, the diagnosis of systemic sclerosis is based on a constellation of symptoms and signs typical of systemic sclerosis, particularly Raynaud's phenomenon and thickening of the skin of the fingers, as well as the presence of specific autoantibodies in blood samples and characteristic abnormalities on examination of the small blood vessels (capillaries) at the base of the nail.

TABLE:

Classification criteria for systemic sclerosis, issued in 2013 by the American College of Rheumatology (ACR) and the European League Against Rheumatism (EULAR)

Critères	Points
Skin thickening of the fingers of both hands extending proximal to the metacarpophalangeal joints	9
Puffy fingers ***or***	2
Sclerodactyly (skin thickening of the fingers)	4
Digital tip ulcers ***or***	2
Fingertip pitting scars	3
Telangiectasia	2
Abnormal nailfold capillaries	2
Pulmonary arterial hypertension ***or***	2
Interstitial lung disease	2
Raynaud's phenomenon	3
Systemic sclerosis-related autoantibodies (anti-centromere, anti-topoisomerase I, anti-RNA polymerase III)	3