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Scleroderma

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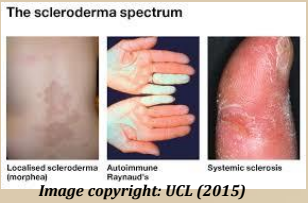

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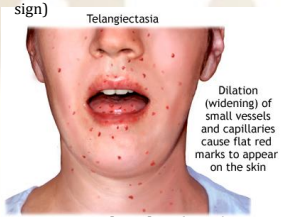
Figure 1: Image of sclerodactyly (Slideshare, 2014)

Signs & Symptoms

Signs and symptoms of scleroderma vary based on the subset of the disease, but according to Dixit, Kalkur, Sattur, Bornstein, & Melton (2016), they most commonly include:

- Raynaud's Phenomenon (one of the first signs)
- Finger swelling, stiffness, or digital ulcers
- Nailfold capillary changes
- Gastrointestinal reflux
- Fatigue
- Weight loss
- Musculoskeletal pain
- Extremity edema
- Skin thickening (usually most obvious sign)



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Pathophysiology

Scleroderma is not an inherited disease; however, a history of familial genetic autoimmune disorders is thought to predispose individuals to the development of the disease. Environmental factors such as exposure to vinyl chloride, silica, and some viral and bacterial infections are thought to trigger the onset of the disease.

Scleroderma is an autoimmune disease related to highly specific autoantibodies. Anti-centromere antibodies are associated with limited cutaneous scleroderma. Antitopoisomerase antibodies are a characteristic of the diffuse cutaneous subset. While anti-RNA polymerase III antibodies are also found in diffuse cutaneous disease, they are associated with the added risk of renal involvement. Anti-Th/To antibodies are linked to the limited cutaneous form with pulmonary fibrosis and PM-SCL antibodies specify scleroderma with polymyositis and severe calcinosis. The autoantibodies are used in confirming diagnosis, predicting disease subset, and potential organ involvement (Chighizola, Raschi, Cesana, Borghi, & Meroni, 2015).

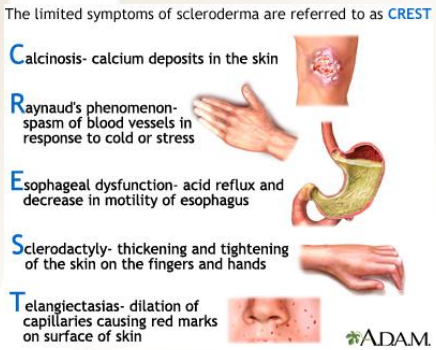
Antibody specificity is thought to affect organ manifestations and phenotype expression (Caetano, Nihtyanova, Harvey, Ong, & Denton, 2016). According to Mitra, et al. (2015), mediators in the pathophysiological process of scleroderma include:

- Transforming growth factor-β (TGF-β): contributes to overproduction of extracellular matrix proteins (ECM) by fibroblasts and promotes differentiation of dermal fibroblasts to myofibroblasts.
- Platelet derived growth factor (PDGF): helps in overproduction of ECM by fibroblasts

Microvascular damage has been found to be an early indicator of the disease process. Endothelial cells induce vascular cell adhesion molecule (VCAM)-1 expression, produce multiple cytokines and chemokines, release vasoconstrictor endothelin-1, and participate in the remodeling of vascular structures (Eckes, Moinzadeh, Sengle, Hunzelmann, & Kreig, 2014). Basement membrane thickening and duplication create narrowing of the lumen of capillaries which leads to loss of microvasculature. A cascade of events triggers the inflammatory response with fibrosis often a result.

Significance of Pathophysiology

Understanding the pathophysiological underlaying of scleroderma has significantly helped to identify individuals effected by the disease and label subsets. It has also aided in the development of possible treatments that could potentially delay the progression of the disease. Most research focuses on the blockage of pathways and mediators that promote formation of fibroblasts. Although experiments with mice have been successful, the same success has not been seen in humans.



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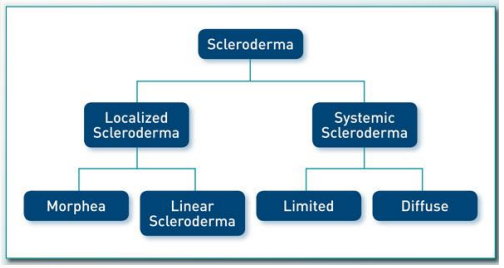
Scleroderma: What is it and why discuss it?

Caring for a patient struggling with the diagnosis of scleroderma sparks interest into the subject for those unfamiliar with the disease. This presentation is provided to further increase knowledge on the topic.

Scleroderma, which generically means hardening of the skin, is a rare disease that causes fibrosis of connective tissue resulting in loss of effectiveness. Sclerosis is a heterogenous, multisystem, multistage disorder noted by variable expression in its forms of presentation and progression of disease (Masi & Medsger, 2015).

Scleroderma has two main subsets: localized and systemic. Both subsets have different patterns in which the disease can follow. Localized scleroderma is mostly mild, does not involve internal organs, and generally consists of localized areas of skin or muscle. It can be morphea, which consists of waxy patches of skin; or linear, which involve a line of hardened waxy skin either on limbs or forehead. Systemic sclerosis (SSc) can be diffuse or limited. SSc can involve muscle, joints, skin, blood vessels, and internal organs such as heart, lungs, gastrointestinal tract, and the esophagus.

Skin and organ fibrosis are key manifestations with SSc and the fingers are always effected (Distler & Cozzio, 2016). The diffuse form of SSc is a more rapidly progressing subset of the disease in which it is more likely for internal organs to be involved. Limited scleroderma is sometimes referred to as CREST syndrome, the name being an acronym for the five common features of the disease: calcinosis, Raynaud's phenomenon, esophageal dysfunction, sclerodactyly, and telangiectasia. SSc predominately affects women ages 20-50 years of age and is associated with increased morbidity and mortality rates (Baron, 2016). Scleroderma has no known specific cause; however, it does involve an overproduction of collagen. Localized scleroderma is more frequent in children while systemic scleroderma is seen more in adults. Although there are treatments for symptoms of sclerosis, there is no cure.



Presentation of Case

Contact with a patient in a medical office demonstrates the following data:

- 25-year-old Caucasian female, no co-morbidities known to date
- Complaining of joint swelling, decreased hand grip, nausea, and heartburn
- Familial history of hypertension, diabetes, and maternal rheumatoid arthritis
- Further investigation reveals: involuntary weight loss of 20 lbs. over last 2-3 months, cold/numb feeling in fingers when in air-conditioned environment, and shortness of breath with exertion
- Physical exam reveals: palpable hard deposits around upper extremity joints, early developing finger flexion contractures, and noted flat red marks to areas on face

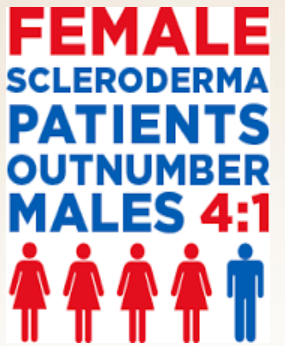
Tests ordered with suspicion of scleroderma diagnosis:

- Complete blood count (CBC)
- Antinuclear antibodies (ANA)
- Scl-70 antibody
- Comprehensive metabolic panel (CMP)
- Centromere antibody (ACA)
- Pulmonary function tests (PFTs)
- Chest x-ray

Follow-up tests show:

- CBC and CMP within normal limits
- Positive ANA, ACA, and Scl-70
- Interstitial fibrosis noted on x-ray
- PFTs suggestive of fibrosis

Diagnosis: Limited scleroderma with pulmonary fibrosis



Implications for Nursing Care

While pathological evidence and research continues to focus on remission of the disease, nursing care must continue to focus on management of symptoms for those currently effected. Symptom management generally focuses on vasculopathy, autoimmunity, and tissue fibrosis. According to McMahan and Wigley (2013), rituximab, tocilizumab and IVIG are common drugs used now; along with newer agents that have shown promise, including bortezomib, LPA-1 antagonists, anti-CCN2 therapy, anti-IL-13 and thrombin antagonists. Vasodilators, corticosteroids, proton pump inhibitors, and angiotension-converting enzyme inhibitors are also used; along with pain management and physical therapy, depending on specific organ involvement. Although medicinal treatments have several options, non-pharmacological interventions are lacking. Research into non-pharmacological supportive therapy is being promoted by committees, such as Scleroderma Patient-Centered Intervention Network (SPIN), to develop patient centered care programs (Thombs, et al., 2012).

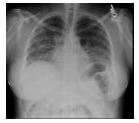


Figure 2: Pulmonary fibrosis on chest x-ray

Conclusion

In conclusion, scleroderma is a rare disease but one associated with multiple complications and comorbidities. Increased knowledge of the signs and symptoms, familial history of autoimmune disorders, subset identification, pathophysiological process, and current treatment options are vital in the diagnosis and management of scleroderma. Ongoing research into better symptom management, improved prognosis, and possible cure are grounded in the pathological foundation of the disease. Healthcare providers should be abreast on the latest research and best practices when delivering care to their patients effected by the disease.

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