FACT SHEET

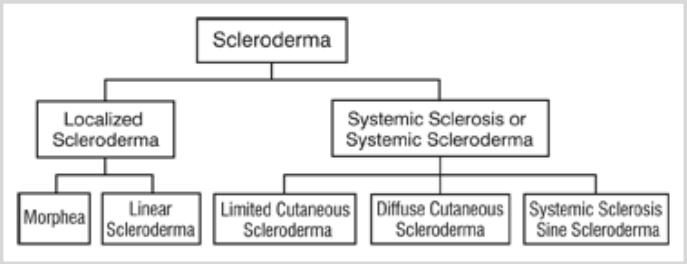
Scleroderma

NATIONAL INSTITUTES OF HEALTH

Scleroderma is a disabling disease characterized by fibrosis, an excessive production of connective tissue proteins, such as collagen. Its name is derived from the Greek words “sklerosis” (which means hardness) and “derma” (which means skin), because a major symptom is hard skin due to fibrosis. The cause of the disease is not known, but one of the reasons is believed to be autoimmunity (a condition in which the body mistakenly detects its own tissue as foreign and attacks itself), which may trigger molecular events leading to fibrosis.

Types of Scleroderma

Scleroderma has many different forms that are placed in two major categories: localized and systemic. Localized scleroderma is generally limited to skin, underlying tissues, and, in some cases, underlying muscle. Systemic sclerosis affects many parts of the body, such as skin, internal organs, and blood vessels.

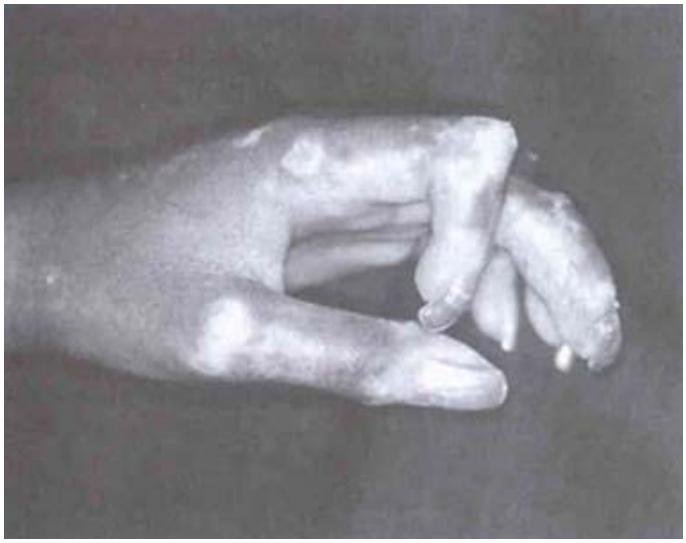


Approximately 50,000 Americans are affected by systemic sclerosis and it is 2-3 times more common in women than men. Although it strikes patients of all ages, including children, incidence is most likely between the ages of 40-60. There is a higher prevalence in some Native American populations.

Raynaud’s phenomenon—an intensified reaction to cold or anxiety, including numbness in fingers, toes, and sometimes other extremities—affects approximately 90% of people with scleroderma at some point in their life. By contrast, only about 3% of the general population has Raynaud’s phenomenon.

Yesterday

- In the past, scleroderma frequently went undiagnosed because of its rarity. Patients were usually referred to orthopedic surgeons to treat shortening of tendons and muscles (a permanent condition known as “contracture”), as well as complications from infections that might require an amputation or the removal of dead or damaged tissue.



Photograph, from Primer on the Rheumatic Diseases, 11th edition (courtesy of Springer) shows severe scleroderma of the hand, with depigmentation, ulceration, and finger contractures due to skin fibrosis.

- Although there were no known treatments, a published description from 18th century Italy did report the successful resolution of symptoms following 11 months of warm milk and vapor baths, bleeding from the foot, and small doses of mercury.

Today

- We have learned that scleroderma is not contagious. Patients are usually referred to dermatologists and rheumatologists who have expertise in autoimmune diseases and musculoskeletal disorders.
- Patients are still treated by orthopaedic surgeons to manage health issues related to the disease, such as those associated with infections. While the amputation of fingers and toes is sometimes still unavoidable, these procedures are rarely necessary because of the availability of many new treatments that target specific tissues and nerves.

- In order to manage the activities of daily living and to intervene in problems of contracture, occupational and physical therapists are also often involved in a patient's care. In addition, other health specialists become involved if particular organs are affected: for example, cardiologists for heart problems, pulmonologists for lung complications, and gastroenterologists for digestive tract issues.
- The majority of treatments address symptoms in specific, affected organs and not the underlying cause of scleroderma, which is the excessive production of connective tissue proteins.
- NIH-funded research has shown that the immunosuppressive drug cyclophosphamide may improve lung function and quality-of-life in scleroderma patients.
- Genetic factors that may contribute to the disease have been identified and are an important focus of NIH investigation.
- The NIH-funded National Family Registry for Scleroderma is collecting biological samples from patients and, when possible, their parents, so that genetic differences between patients and healthy individuals can be detected and potentially traced to a parent. Further research based on this registry may reveal genes that contribute to the development of scleroderma.

Tomorrow

- Categorizing the severity of the disease by distinct patterns of gene expression, or "gene signatures," may guide personalized treatment for scleroderma patients.
- Antibodies are molecules that attack the body's invaders, such as bacteria or viruses. In the case of scleroderma, autoantibodies (antibodies that attack a person's own tissues) may carry out some of the tissue damage seen in this disease. Therefore, monitoring these scleroderma-associated autoantibodies may help characterize patients' conditions and predict the future course of their disease.
- New therapies may prevent the development of scleroderma by interrupting the process of fibrosis or immune system dysfunction that leads to attacks on a patient's own tissues. NIH-supported researchers have developed several mouse models that mimic these biological pathways in humans. Additional insights from these models could be pivotal to the development of new scleroderma treatments.
- Scleroderma susceptibility genes discovered by the National Family Registry may identify new targets for treatment.
- Participation of patients in clinical research is one of the best ways to advance new knowledge and contribute to the development of new treatments.

Contact: NIAMS Information Clearinghouse toll free: 877-22-NIAMS (226-4267); email: NIAMSInfo@mail.nih.gov

National Institute on Arthritis and Musculoskeletal and Skin Diseases (NIAMS):

www.niams.nih.gov