

SYSTEMIC SCLEROSIS IN THE AFRICAN-AMERICAN PATIENT

By Virginia Steen, M.D.

Systemic sclerosis is an uncommon connective tissue disease, which is seen in all races, but does have an increased incidence and prevalence in African-Americans. It develops at a younger age in African-Americans than in Caucasians. The peak age of onset in Caucasian females is between 55 and 64. In African-American females, there is a group with very early onset at age 15 to 24, with the main peak between 45 and 54 years old. In African-American males, there is also increased incidence and prevalence, with an earlier onset than in Caucasians, 35 to 54 years old in African-Americans compared to age 45 to 64 in Caucasian males.

In addition to being more frequent and occurring at a younger age, African-Americans with scleroderma have increased morbidity and mortality than Caucasians. This is not a unique finding. In many diseases, such as hypertension and diabetes, African-Americans have more severe disease. For scleroderma, the death rate is 50% higher in females and 100% higher in males who are African-American compared to Caucasians. We will review some of the reasons why this may be happening, but first we will describe some of the ways in which scleroderma is different in African-Americans than Caucasians.

First, the initial presentation is often different. Although Raynaud Phenomenon is the most common first symptom of scleroderma, African-Americans often do not easily recognize it. The blue, pale fingers associated with cold-induced Raynaud's may not be as dramatic as in Caucasians. Also, many African-Americans first notice changes in skin pigmentation. It is quite common for African-American patients to notice marked darkening of the hands arms or face, (or hyperpigmentation) even before they have Raynaud's or swollen fingers. They may also

develop decreased or absence of pigmentation of the skin, often around the hairline or on the chest or arms (hypopigmentation). The disease affects the melanocytes, which are cells that make pigment, resulting in these increased and decreased pigmented areas. These changes in pigmentation are often very distressing to African-American patients. Although there are no good treatments, fortunately, the pigment changes tend to fade with time and can be covered with make-up.

We recently summarized our experience of scleroderma in African-Americans from the University of Pittsburgh Scleroderma Program. Comparing 202 African-Americans with 2,945 Caucasian patients with scleroderma, African-Americans were younger at onset; mean age was 38 compared to Caucasians, who were 43 when they developed scleroderma. Also, African-Americans more frequently had diffuse cutaneous disease. More than half of the African-Americans (51%) had diffuse disease, whereas only 43% of the Caucasian did. Since diffuse cutaneous disease is associated with more severe internal organ involvement, that explains, at least in part, why African-Americans have more severe disease. We also found that African Americans were more likely to have severe muscle, gastrointestinal and lung involvement.

Muscle involvement in scleroderma can be from either inflammation or from fibrosis (or scar tissue). The types of muscle problems that are seen in African-American patients often are subtle. While there may be some inflammation, much of the problem is from scar tissue. Low-dose prednisone may be helpful, but the most important treatment in these patients is aggressive exercise. Because the weakness is less obvious to the patient, they often just don't raise their arms as easily and then develop

decreased motion to the point that they can't raise their arms at all. We call this "contractures," and once they develop, it is much harder to regain motion. I regularly prescribe physical therapy for all patients early in their illness to make sure they know how to do the stretching exercises to prevent these contractures.

Gastrointestinal involvement is present in almost all patients with scleroderma. Reflux, difficulty swallowing and heartburn are the most common symptoms. African Americans have been shown to have more small intestine involvement, which includes more bloating, diarrhea and malnutrition. So watching for weight loss, vomiting, abdominal distension is particularly important to identify and treat aggressively. This is the only gastrointestinal problem that is associated with any subgroup.

One of the major reasons for poor outcomes in African Americans with scleroderma is because of increased frequency of lung involvement. This is, in part, because the antibodies associated with severe lung disease are seen with increased frequency in African-Americans. But we have shown that even in patients with the same autoantibody, that African Americans have more severe disease than Caucasians. **Thus, it is absolutely critical that all African American scleroderma patients are carefully monitored for lung involvement.** Pulmonary function tests, high-resolution CT scans and echo cardiograms should be done in all African American scleroderma patients at baseline and then every 6 to 12 months to determine if there is interstitial lung disease. Treatment for interstitial lung disease, with either cyclophosphamide or mycophenolate mofetil, can stabilize disease if started early enough. There are more than 10 medications approved for the treatment of pulmonary arterial hypertension (PAH), but it is critical to identify the patients as early as possible. A marked decrease in the DLCO (an abnormality on the pulmonary function test) is an excellent marker that identifies patients at increased risk for PAH. Patients with a low DLCO should definitely have

yearly screening with echocardiogram and symptoms for evidence of PAH.

Over all, scleroderma in African-Americans occurs at a younger age, in greater frequency associated with diffuse scleroderma, more severe small bowel involvement and more severe pulmonary fibrosis. Also, pulmonary arterial hypertension occurs earlier, in younger patients with diffuse scleroderma and thus, African-Americans with scleroderma have worse prognoses. While these differences may be from socioeconomic disparities or because of access to care, we believe that most of it is because of the difference of the disease in African-Americans. Genetic differences may play a major role. An exciting study, GRASP, (Genome Research in African American Scleroderma Patients) is looking carefully at genetic factors that may be making the disease different in African Americans than in Caucasians. ■

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