

# A Case of Diffuse Cutaneous Systemic Sclerosis in a 6-year-old Filipino Male

Mary Rose M. Maballo MD<sup>1</sup>, Benedicto dL Carpio MD<sup>2</sup>, Eileen Regalado-Morales MD<sup>2</sup>, Amelita Tanglao-de Guzman MD<sup>2</sup>, Armelia Lapitan-Torres MD<sup>2</sup>

<sup>1</sup>Resident, Department of Dermatology, Ospital ng Maynila Medical Center, Manila, Philippines

<sup>2</sup>Consultant, Department of Dermatology, Ospital ng Maynila Medical Center, Manila, Philippines

**Abstract:** Systemic sclerosis, also known as Systemic Scleroderma is a multisystem disorder characterized by fibrosis of the dermis and vascular abnormalities. It is rare disorder, with an incidence of about 2-8 per 1,000,000 population, and is even more unusual during childhood. It belongs under the spectrum of Scleroderma, a group of disorders that present with indurated patches or plaques on the skin. We are presenting a case of a 6-year-old male with diffuse indurated hidebound skin, Raynaud's phenomenon, flexion deformity of the digits, microstomia and a beak like nose with onset at 3 years of age. Biopsy revealed prominent sclerosis of the papillary and deep reticular dermis and few to absent adnexal structures in the dermis. Clinical and histological findings of the case were compatible with a diagnosis of Scleroderma. However, testing of autoantibodies such as anti-nuclear antibodies and anti-Scl70 yielded negative results. Additionally, systemic manifestations like pulmonary or renal involvement, which are often encountered in patients with Systemic Sclerosis, were notably absent in our case. Despite heterogeneity of its presentation, careful medical history, thorough dermatological evaluation and histopathological correlation still remains the cornerstone of diagnosis of Systemic Sclerosis. Finally, a multidisciplinary approach to management cannot be overemphasized in these patients.

**Keywords:** Systemic sclerosis, scleroderma, autoantibodies.

## 1. INTRODUCTION

Systemic sclerosis, also known as systemic scleroderma, is a rare multisystem disorder characterized by vascular abnormalities, connective tissue sclerosis and atrophy and autoantibodies. It is a complex and multifactorial disease whose exact pathogenesis and etiology remains unknown. It is a variant within the spectrum of scleroderma.

## 2. CASE HISTORY

A case of a 6-year-old male presented with a 3-year history of progressive, symmetric induration of skin. This was associated on history with episodes of Raynaud's phenomenon, hypohidrosis and joint stiffness. Persistence prompted consult at the Department of Dermatology of Ospital ng Maynila Medical Center.



Figure 1a and b. Diffuse hypo and hyperpigmented indurated, shiny patches and plaques affecting the face, neck, trunk and extremities

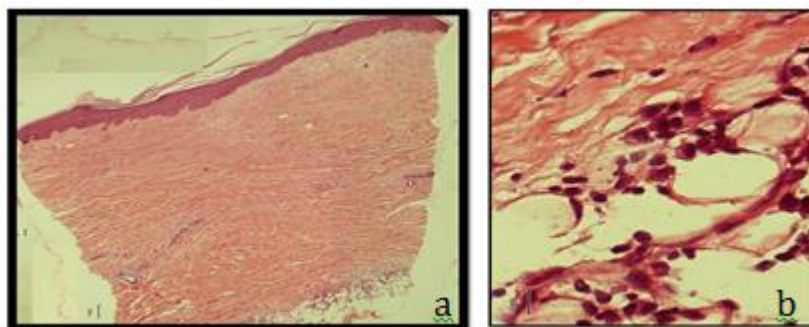


**Figure 2 (a) Close up view of patient's facial features (b) Restricted mouth opening (c) Pinched or beak like nose (d) flexion deformity of the fingers**

Cutaneous examination showed diffuse hypo- and hyperpigmented indurated, waxy, shiny hide-bound patches and plaques affecting the face, neck, trunk and both upper and lower extremities.

Birth history and review of systems were unremarkable. No other family members exhibited similar lesions. Patient was referred to a Pediatrician and was found to be clinically normal. Workups were normal.

Five-millimeter punch biopsy was done on an indurated plaque on the patient's lower back and revealed prominent sclerosis of the papillary and deep reticular dermis and few to absent adnexal structures in the dermis. Other findings included dilated and telangiectatic blood vessels in the papillary dermis and small clusters of lymphocytes at the border between the panniculus and reticular dermis.



**Figure 3: Histopathological findings on (a) low power view showing prominent sclerosis of the papillary and deep reticular dermis and absent adnexal structures in the dermis (b) oil immersion objective showing lymphocytes between the panniculus and reticular dermis**

Clinico-histopathological diagnosis revealed scleroderma. It is important to remember that the different clinical variations of scleroderma may be linked by certain histologic findings, however they are not a single disease. On the basis of microscopic findings alone, one cannot determine which variant afflicts a particular patient.

Several workups were requested for the patient, namely a complete blood count, a urinalysis and a chest radiograph. However, results were normal. The presence of autoantibodies were also tested, namely quantitative ANA and Anti-Sc170. Results were however negative (ANA 0.5 and anti Sc170 0.4 U/mL). Patient was given multivitamins and emollients. Relatives of the patient were advised regarding the patient's condition and prognosis. Currently, the patient is being referred to a Pediatric Rheumatologist.

### 3. DISCUSSION

Scleroderma is a spectrum of disorders characterized clinically by thickening of the skin caused by accumulation of connective tissue. It is characterized by the appearance of circumscribed or diffuse, hard, smooth, ivory colored areas that are immobile and give the appearance of hidebound skin. There are two ends on the spectrum of scleroderma. On one end is localized scleroderma or morphea and on the other end is systemic sclerosis. They differ in the extent of skin involvement and the presence or absence of clinically detectable systemic involvement. Systemic sclerosis is compatible with the case that had a very notable presence of Raynaud's phenomenon, symmetrical induration and sclerodactyly.

Systemic sclerosis is a multisystem disorder characterized by vascular abnormalities, connective tissue sclerosis and atrophy and the presence of autoantibodies. It is a rare disorder with an incidence of 2-8 per 1,000,000. The ratio of males to females is 1 to 3 with a peak onset in the 4<sup>th</sup> decade in females and later in males. Systemic sclerosis results in a significant reduction of lifespan with an overall 20 year survival of under 70%. It has no known racial predilection.

Systemic sclerosis is a complex, multifactorial disease and the exact pathogenesis and etiology remains unknown. It is well established that there are 3 key pathogenic abnormalities in the skin and internal organs: endothelial cell damage, inflammation and excessive tissue deposition of collagen and other matrix proteins. However the pathogenic steps leading to these abnormalities are not clear.

Scleroderma presents difficulties in diagnosis and management due to the heterogeneity of its manifestations. And the diagnosis of scleroderma relies heavily on clinical judgment. However, the American Rheumatism Association (1980) established criteria for diagnosis. The single major criterion was scleroderma proximal to the digits, affecting limbs, face, neck or trunk, usually in bilateral and symmetrical pattern. Minor criteria were sclerodactyly, digital pitting scars and bilateral pulmonary fibrosis. The diagnosis of systemic sclerosis requires the presence of the major criterion or two of the three minor criteria. Patient was able to fulfill the one major criterion and one minor criteria, which was sclerodactyly.

Although the clinical overlap may be substantial, most clinicians have accepted the separation of systemic sclerosis into 2 major clinical categories: limited and diffuse disease. The distinction is based principally on the basis of the extent of cutaneous involvement. On this case, we are more leaning towards a case of diffuse cutaneous systemic sclerosis due to the involvement of the truncal and peripheral skin involvement.

Other manifestations that were evident in this case were Raynaud's phenomenon, limited extension of fingers, lost of sweat glands, microstomia, and pinched or beak like nose. Notably however systemic manifestations, which are often encountered in patients with the same condition, were absent in our patient.

The management of systemic sclerosis is as complicated as the disease itself. However it is important to emphasize, especially to our patients that there is no specific treatment and no therapy is known to alter the course of the disease. But even though SSc cannot be cured, treatment of involved organ systems can relieve symptoms and improve overall function. For this patient, it was opted that patient will be best managed by a Pediatric Rheumatologist.

### 4. CONCLUSION

Systemic Sclerosis is a complex disease with heterogeneous clinical manifestations. Despite this, a thorough dermatologic evaluation supplemented by histopathologic examination remains the mainstay of diagnosis. And though ancillary procedures exist, they should guide rather than dictate a clinician's decision making. Finally, a multidisciplinary approach to management cannot be overemphasized in these patients.

### REFERENCES

- [1] Bologna et al. *Dermatology* 2<sup>nd</sup> edition. 2003.
- [2] Burns et al. *Rook's Textbook of Dermatology* 7<sup>th</sup> edition, 2004.
- [3] Folderi et al. Systemic sclerosis in childhood. *Rheumatology* 45:3 28-29. 2006.
- [4] James et. al. *Andrew's Diseases of the Skin, Clinical Dermatology*, 10<sup>th</sup> edition. 2005.
- [5] Masi et. al. Preliminary criteria for the classification of systemic sclerosis. *Arthritis and Rheumatism* 23:5 581-590. 1980.
- [6] Steen et al. Clinical Correlations and prognosis based on serum autoantibodies in patients with systemic sclerosis. *Arthritis and Rheumatism* 31:2 196-203. 1988.
- [7] Wolff et al. *Fitzpatrick's Dermatology in General Medicine* 7<sup>th</sup> edition. 2008.