Hereditary Sclerosing Poikiloderma

Hyo Jin Lee, Dong Hoon Shin, Jong Soo Choi, and Ki Hong Kim

Department of Dermatology, College of Medicine, Yeungnam University, Daegu, Korea

Received: 11 May 2011
Accepted: 1 November 2011

Address for Correspondence:
Dong Hoon Shin, MD
Department of Dermatology, College of Medicine, Yeungnam University, 170 Hyoemchung-ro, Nam-gu, Daegu 705-802, Korea
Tel: +82.53-620-3160, Fax: +82.53-622-2216, E-mail: dhshin@med.yu.ac.kr

INTRODUCTION

Hereditary sclerosing poikiloderma (HSP) is a very rare autosomal dominant genetic disease which was first described by Weary et al. (1) in 1969. Clinical manifestations consist of 1) widespread poikiloderma with accentuation in flexural areas and over extensor bony prominences, 2) linear or reticular shaped hyperkeratotic and sclerotic bands on the axillae, antecubital and popliteal fossae, 3) sclerosis of the palms and soles, 4) clubbing fingers, and 5) calcinosis of the tissues. HSP may be also accompanied with musculoskeletal and cardiovascular abnormalities. Therefore, it is necessary to evaluate the existence of other combined abnormalities.

Few cases of HSP have been reported in medical literature, but no Korean case of HSP has been reported yet. Here, we report the first Korean case of HSP presented with poikiloderma and sclerotic bands on both popliteal and antecubital fossae.

CASE DESCRIPTION

A 18-yr-old Korean male visited the Department of Dermatology of the Yeungnam University Hospital due to progressively generalized pigmented lesions on November 2, 2005. He was normal at birth. Reticular hyperpigmented lesions were first observed at 2-yr-old on the popliteal and antecubital fossae. Similar changes gradually became on the axillae and trunk. Linear sclerosing bands appeared on both of the antecubital and popliteal fossae after yr. Most lesions had been progressively increased in size and number with time, but he was asymptomatic.

In family history, his father had similar symptoms and died in his fourties due to cardiovascular disease. Also, his younger brother had similar poikilodermatous lesions. The lesions appeared at the age of 3 yr and hyperpigmented and sclerotic changes had been progressed.

Hereditary sclerosing poikiloderma (HSP) is a very rare disease. The clinical features are principally widespread poikiloderma and linear hyperkeratotic and sclerotic bands. We report an 18-yr-old male who presented reticular hyperpigmented lesions on the trunk and extremities since 2-yr-old. Also, linear sclerosing bands appeared on both antecubital and popliteal fossae after yr. Histopathologic finding showed dense sclerotic collagen fibers with telangiectasia in the upper dermis and fragmentations of damaged elastic fibers in the elastic stain, consistent with HSP. We report the first Korean case of HSP.

Key Words: Hereditary Sclerosing Poikiloderma; Korean

DISCUSSION

The diagnosis of HSP is based on the clinical findings and the family history, because no specific tests for the diagnosis have
been established (2). The five major clinical manifestations of HSP were proposed by Weary et al. (1). Of these, the most important features for diagnosis for HSP are widespread poikiloderma and sclerotic bands. Previously reported cases suggest that the diagnosis for HSP requires the presence of the above two features and other clinical features are not essential to diagnosis (3). Our patient had generalized poikiloderma, linear sclerotic bands in flexural areas, sclerosis of the palms and soles, and clubbing of the fingers as other reported cases, but tissue calcinosis was not found. In addition, micrognathia was noted. Also, his height is 155 cm and body weight 46 kg, these are similar to an average of sixth grade of elementary school in Korean. Therefore, his growth had lagged behind the industry at least six yr. Thus, our patient was able to diagnose as a HSP.

The widespread poikiloderma characterized by speckled hypopigmentation and hyperpigmentation, slight atrophy, and minimal telangiectasia mainly involves in the flexural areas and over bony prominences (2). It is absent at birth and first appears within the second to fourth yr of life without prior vesiculation or eczema and progressively worsens with increasing time. The sclerotic bands are observed in the skin of the flexural areas of the axillae and antecubital and popliteal fossae after poikilodermatous changes which, in addition to marked poikiloderma, exhibits formation of extraordinary reticulated and linear, hyperkeratotic and sclerotic bands which extend across the flexures.

HSP may be inherited as autosomal dominant trait with incomplete penetrance and is most severe in males (1). In our patient, the family history showed that the patient’s father and younger brother had poikilodermaous skin, but other members were not affected including grandfather and grandmother. The fact that his father is the first affected member of the family suggests that the disease in this male may represent a new mutation.

HSP may be also accompanied with cardiovascular abnormalities. Though cardiac involvement was reported in only two previously described patients (1, 3), several members of reported patient’s family had cardiac valvular diseases or impending valvular diseases (1, 3, 4). Also, our patient’s father died at 40 yr as a result of cardiovascular disease. These facts suggest that cardiac abnormalities may represent an important element in HSP. Therefore, it is necessary to evaluate regularly the exis-
tence of cardiovascular disease.

Clubbing is a physical sign characterized by bulbous enlargement of the ends of one or more fingers or toes. It usually acquired and is associated with cardiopulmonary and gastrointestinal diseases (5). Though it was observed in most HSP patients, no cardiopulmonary abnormalities were found to explain the clubbing in most previously reported patients except two above described patients who had cardiovascular diseases.

Other combined abnormalities included tissue calciosis (1), micrognathia (2), maxillary bossing (2), mandibuloacral dysplasia (4), Raynaud’s phenomenon (3) and growth retardation were also reported. Our case had micrognathia and growth retardation, but had no other above described diseases.

Recent article about HSP was reported in 2006 (6). It showed poikilodermatous change of skin associated with tendon contracture and progressive pulmonary fibrosis. In previous reports, although there were cardiac involvement, no pulmonary involvement was described. Also, her family had diffuse interstitial pulmonary fibrosis. The authors suggested that HSP might be divided into the three categories: Weary form HSP, HSP with cardiac involvement, and HSP with tendon/pulmonary involvement. However, further research is needed for that.

The histopathologic findings showed homogeneous & dense sclerotic collagen fibers in the upper dermis and fragmentations of damaged elastic fibers in the dermis. In our patient, increased melanin pigments in the basal layer and sclerotic changes and telangiectasia in the upper dermis were found in the H&E stain. Also, Elastic stain showed fragmentations of elastic fibers in the dermis. According to Greer’s study (2), immunofluorescent studies revealed no evidence of immunoglobulin or complement deposition in the epidermis, basement membrane zone or other spaces in the dermis. Also, electron microscopic studies failed to demonstrate any specific abnormality of dermal connective tissues other than elastic fiber fragmentation.

The differential diagnosis of HSP consists of several diseases that are mandibuloacral dysplasia (4), hereditary acrokeratotic poikiloderma (7), poikiloderma congenital (8), xeroderma pigmentosa (9), dyskeratosis congenital (10), Werner’s syndrome (11), and poikiloderma vasculare atrophicans. Poikiloderma without prior vesiculation or eczema distinguishes HSP from hereditary acrokeratotic poikiloderma, and autosomal dominant trait does HSP from poikiloderma congenital and xeroderma pigmentosa. HSP must be also distinguished from mandibuloacral dysplasia and Werner syndrome. Mandibuloacral dysplasia has poikilodermatous appearance and linear sclerotic bands, but it has mandibular hypoplasia, delayed cranial suture closure, dysplastic clavicles, and acroosteolysis (12). Although cutaneous manifestations associated with mandibuloacral dysplasia were similar to those associated with HSP, the extracutaneous manifestations in our patient were not found in association with mandibuloacral dysplasia. Also, unlike autosomal recessive inheritance visible from mandibuloacral dysplasia, pedigree of our patient showed autosomal dominant inheritance. Werner syndrome becomes apparent later, typically in the third or fourth decade.

To the best of our knowledge, 11 cases of HSP have been reported in the medical literature (1, 2, 3, 6, 13). Of these previously described patients, eight patients were African-American (1, 2) and three were Caucasian (3, 6, 13). However, Korean patients have not been reported yet, therefore this is the first case of HSP occurred in Korea.

REFERENCES

1. Weary PE, Hsu YT, Richardson DR, Caravati CM, Wood BT. Hereditary sclerosing poikiloderma. Report of two families with an unusual and distinctive genodermatosis. Arch Dermatol 1969; 100: 413-22.
2. Greer KE, Weary PE, Nagy R, Robinow M. Hereditary sclerosing poikiloderma. Int J Dermatol 1978; 17: 316-22.
3. Grau Salvat C, Pont V, Cors JR, Aliaga A. Hereditary sclerosing poikiloderma of Weary: report of a new case. Br J Dermatol 1999; 140: 366-8.
4. Fryburg JS, Sidhu-Malik L. Long-term follow-up of cutaneous changes in siblings with mandibuloacral dysplasia who were originally considered to have hereditary sclerosing poikiloderma. J Am Acad Dermatol 1995; 33: 900-2.
5. Walk HK, Hall WD, Hurst JW, editors. Clinical method: the history, physical, and laboratory examinations. 3rd ed. Chapter 44 Clubbing. Boston: Butterworths, 1990.
6. Khumalo NP, Pillay K, Brightn P, Wainwright H, Walker B, Saxe N, Mayosi BM, Bateman ED. Poikiloderma, tendon contracture and pulmonary fibrosis: a new autosomal dominant syndrome? Br J Dermatol 2006; 155: 1057-61.
7. Weary PE, Manley WF Jr, Graham GF. Hereditary acrokeratotic poikiloderma. Arch Dermatol 1971; 103: 409-22.
8. Blinstein RS, Lehman R, Sternberg TH. Poikiloderma congenitale. Report of two cases. Arch Dermatol 1984; 89: 659-64.
9. Ramachandra L, Rajagopal Shenoi K, Santhosh Pai U. Xeroderma pigmentosa in siblings: cystosarcoma phylloides in a case of xeroderma pigmentosa. Indian J Dermatol Venereol Leprol 2002; 68: 168-70.
10. De Boeck K, Degreef H, Verwilghen R, Corbeel L, Casteels-Van Daele M. Thrombocytopenia: first symptom in a patient with dyskeratosis congenita. Pediatrics 1981; 67: 898-903.
11. Gottesmann T, Zala L, Vogel A, Mumenthaler M. The Werner syndrome. Schweiz Med Wochenschr 1980; 110: 246-50.
12. Young LW, Radebaugh JE, Rubin P, Sensenbrenner JA, Fiorelli G, McKusick VA. New syndrome manifested by mandibular hypoplasia, acroosteolysis, stiff joints and cutaneous atrophy (mandibuloacral dysplasia) in two unrelated boys. Birth Defects Orig Artic Ser 1971; 7: 291-7.
13. Fazio M, Lisi S, Amantea A, Maini A, Menaguthe G, Sacerdott G, Balus L. Weary hereditary sclerosing poikiloderma. Ann Dermatol Venereol 1995; 122: 618-20.