Generalized Pseudoxanthoma Elasticum

Ki Hong Lee, M.D., Ju Eun Lee, M.D., Ki Bum Myung, M.D.

Department of Dermatology, College of Medicine, Ewha Womans University, Seoul, Korea

We report a patient with a cutis laxa-like, generalized PXE without systemic involvement. A 28-year-old woman had loose, pendulous skin of the neck, axillae, thighs, trunk and body folds which resulted in a prematurely aged appearance. She had no family history of related diseases. Histological examination showed considerable accumulations of swollen and irregularly clumped fibers in the middle and lower dermis and von Kossa's stain revealed calcium deposits along the altered elastic fibers. (Ann Dermatol 5:(1) 38-40, 1993)

Key Words: Generalized pseudoxanthoma elasticum

Pseudoxanthoma elasticum (PXE) is a disorder of the elastic tissue involving the skin, eyes and cardiovascular system. Pope[1] divides PXE into four genetic types. He described two autosomal ant two autosomal recessive PXE. Both dominant and recessive type I show classic skin, eye and vascular diseases. Dominant type II has a macular pattern of skin lesions with hyperextensible skin and hypermotile joints. Recessive type II, the least common form, has generalized PXE without systemic involvement. Only three families of recessive type II were detected in Britain by Pope[2], and recently two cases of generalized PXE were reported. One was reported in Italy[3] and the other in Korea[4]. As a rare instance, we report a case of PXE presenting generalized cutaneous laxity without systemic involvement. We suspect this case is an autosomal recessive type II PXE.

REPORT OF A CASE

A 28-year-old woman seen at Ewha Womans University Hospital in 1990 with loose, pendulous skin of the neck, axillae, thighs, trunk and body folds, resulting in a prematurely aged appearance (Fig. 1). The skin changes began on the neck at the age of 22 and gradually got worse with age, especially after her first delivery, leading to marked laxity and diminished elasticity over the entire body. she had no family history of related diseases. On physical examination, the skin was easily pulled away from tissues below, returned very slowly to its former position and had a doughy feel. There were no alterations in sensation or increase in fragility and abnormalities of the joints. The laboratory tests including a complete blood count, erythrocyte sedimentation rate, urinalysis, electrolyte, liver function test; chest P-A, EKG, upper GI, barium enema and funduscopic examination were within normal limits. A skin biopsy taken from a lesion on the right axilla showed considerable accumulations of swollen and irregularly clumped fibers in the middle and lower dermis (Fig. 2). Weigert's stain revealed enlargement and fragmentation of elastic fibers (Fig. 3). Von Kossa's stain showed calcium deposits along the altered elastic fibers (Fig. 4). In Masson's trichrome stain, the collagen fibers were normal. Electron microscopic examination showed marked swelling and bizzare distorsions of the calcified elastic fibers.

DISCUSSION

Pseudoxanthoma elasticum is an inherited disorder of connective tissue charaterized by generalized elastorrhexis affecting the elastic tissue in the dermis, the blood vessels and Bruch's membrane of the eye[5].

Received May 15, 1992
Accepted for publication June 3, 1992
Reprint request to: Ki Hong Lee, M.D., Department of Dermatology, College of Medicine, Ewha Womans University, Seoul, Korea

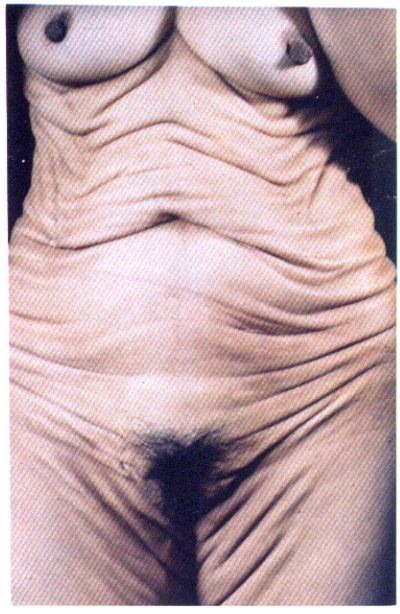

Fig. 1. Loose-fitting skin of the trunk, thighs and body folds.

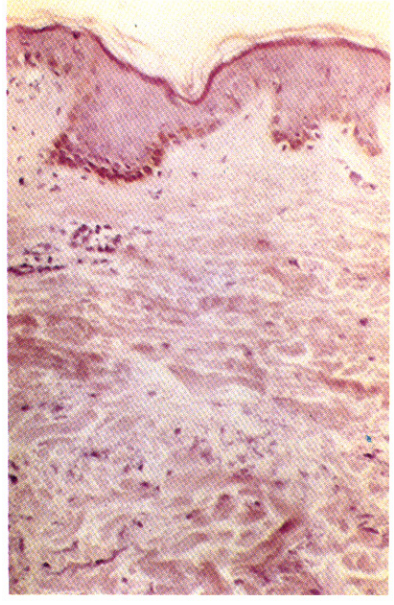

Fig. 2. Accumulations of swollen, clumped basophilic fibers in the middle and lower dermis (H&E stain, × 100).

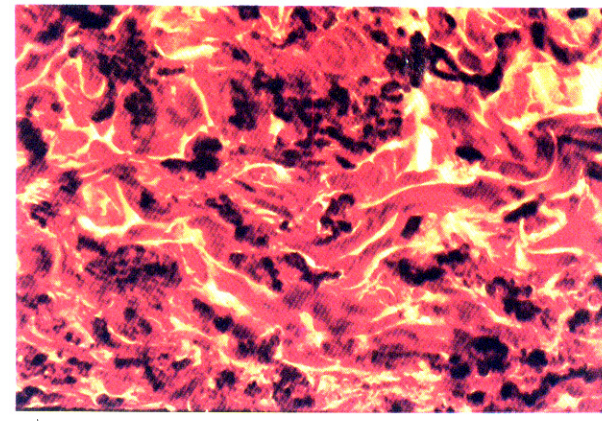

Fig. 3. Enlargement and fragmentation of elastic fibers in the dermis (Weigert's stain, × 400).

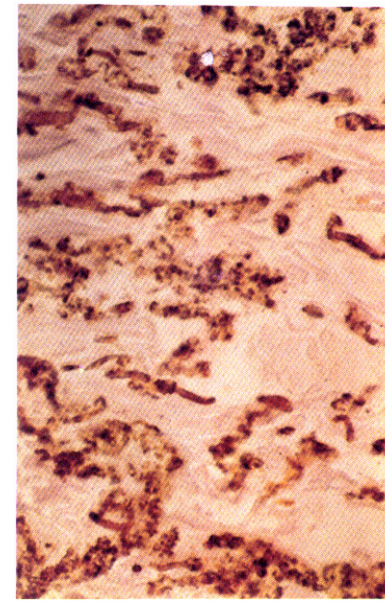

Fig. 4. Calcium deposits along the altered elastic fibers in the dermis (von Kossa's stain, × 400).

Most previous authors have suggested pseudoxanthoma elasticum as autosomal recessive types[6,7], but in a British survey, four types of pseudoxanthoma elasticum were clearly identified by Pope[1]. He described two forms of autosomal dominant and two forms of autosomal recessive. Dominant type I shows a flexurally distributed peau d'orange pseudoxanthomatous rash, severe vascular complications, such as, coronary artery disease and claudication, and a severe degenerative retinopathy, with early blindness. Dominant type II, which is much less severe, typically has a canary-yellow macular rash, minimal vascular symptoms and mild retinal changes with prominent choroidal vessels. Increased cutaneous extensibility, blue sclerae, a high arched palate and myopia are also signs. Recessive type I resembles the dominant type I group although vascular and retinal degeneration are much milder. Haematemeses are common, especially amongst affected females. Recessive type II, a unique and rare

variant, shows generalized cutaneous laxity and infiltration without systemic complications.

Although Pope reported that 53% of cases were of the autosomal dominant type, other studies reported a 75% to 90% autosomal recessive inheritance[3]. Also, among 12 cases of PXE described in Korea, one was classified as recessive type II[4] and the others as revessive type I.

In our case, the absence of any familial history and the patient's clinical features place her in the rare recessive type II PXE of Pope, which mimics cutis laxa without any vascular or ophthalmic involvement.

In the differential diagnosis, cutis laxa, Ehlers-Canlos syndrome and Marfan's syndrome must be considered. Especially, cutis laxa, which clinically resembles recessive type II PXE, must be ruled out. Cutis laxa is charaterized by loose, redundant skin, hanging in folds and drooping skin around the eyelids, cheeks and neck[8], but in recessive type II PXE the face is usually spared. On the basis of the charateristic histologic findings, it is possible to differenciate recessive type II PXE form cutis laxa without calcium deposits.

Histologic examination of the involved skin reveals in the middle and lower thirds of the dermis, considerable accumulations of swollen and irreguarly clumped fibers staining like elastic fibers; that is, they stain deeply black with orcein or the Verhoeff stain. Staining for calcium with the von Kossa method also shows these fibes well. In the vicinity of the altered elastic fibers, there may be accumulations of a slightly basophilic mucoid material, which stains strongly positive with the colloidal iron reaction or with alcian blue. Electron microscopic examination shows that with progression of the calcification, the elastic fibers ultimately become fully calcified, showing marked swelling and bizzare distorsions. The presence of calcified material outside of elastic fibers can be explained by the disintegration of completely calcified elastic fibers[9].

No definitive therapy is available. Usually the skin presents only cosmetic problems. Neldner et al[10] advise limiting dietary calcium and phosphorus to the minimal daily requirement level. Plastic surgery is helpful for loose folds.

REFERENCES

1. Pope FM: *Historical evidence for the genetic heterogeneity of pseudoxanthoma elasticum. Br J Dermatol 92:493-509, 1975.*
2. Pope FM: *Two types of autosomal recessive pseudoxanthoma elasticum. Arch Dermatol 110:209-212, 1974.*
3. Rongioletti F, Bertamino R, Rebora A, et al: *Generalized pseudoxanthoma elasticum with deficiency of vitamine K-dependent clotting factors. J Am Acad Dermatol 21:1150-1152, 1989.*
4. Kang IK, Chung BS, Kim YC, et al: *A case of pseudoxanthoma elasticum presenting as generalized cutaneous laxity. Kor J dermatol 29:423-427, 1991. (English abstract)*
5. Burton JL, Ebling FJG: *Disorders of connective tissue. In text book of Dermatology. Rook A, Wilkinson DS, Ebling FJG et al (eds). Vol 3, Blackwell Scientific Publications, Oxford, 1986, pp 1841-1844.*
6. Cockayne EA: *Inherited abnormalities of the skin and its appendages. Oxford University Press, London, 1933, pp 319-321, Cited from reference 1.*
7. McKusick VA: *Heritable disorders of connective tissue. 4th ed, C.V.Mosby Co., St. Louis, 1972, pp 475-520.*
8. Arnold HL, Odom RB, Jame WD: *Andrew's Diseases of the skin. 8th ed. WB Saunders Co, Philadelphia, 1990, pp 591-592.*
9. Lever WF, Schaumburg-Lever G: *Histopathology of the skin 7th ed, JB Lippincott Co, philadelphia, 1990, pp 84-86.*
10. Neldner KH, Reeve EB, Subryan V: *Studies on seudoxanthoma elasticum. J Invest Dermatol 69:425, 1977.*