

# Acrogeria with Perforating Elastoma and Bony Abnormalities

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A case of acrogeria, a premature aging syndrome with acral distribution, is reported in association with perforating elastoma and bony abnormalities. *Key words: Premature aging syndrome; Atrophy of the skin; Hyperpigmentation.* (Received December 6, 1983.)

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Acrogeria is a rare developmental defect first described by Gottron in 1941 (1). Since then, more than 20 cases have been reported, pointing out interesting associations, including perforating elastoma and bony abnormalities.

## REPORT OF A CASE

A 14-year-old mentally retarded white boy was seen at the Mayo Clinic in 1959. He had a 3-year history of asymptomatic, grouped, horny, red papules with an annular configuration on the left side of the neck, just below the posterior hairline (Fig. 1).

On clinical examination, the skin was found to be diffusely atrophic and hyperpigmented, with atrophy most pronounced on the extremities. The hands were small and hyperpigmented, with spindle-shaped fingers, and the venous pattern was readily visible because of the atrophic skin (Fig. 2 A). The feet were small, with atrophic wrinkling of the skin, varus deformation of the right foot, and dystrophic toenails (Fig. 2 B). The nose was pinched. A deep venous pattern was seen on the anterior aspect of the upper trunk, and there was moderate gynecomastia. The fingernails, hair, and dentition were normal. No joint hypermobility or skin hyperextensibility was noted. The family history was negative for acrogeria, and there was no history of consanguinity. The personal history was difficult to obtain from this mentally retarded patient, but the mother had noted "thin skin" from birth and easy bruising of the skin. Bone roentgenograms showed varus deformity of the right foot, congenital dislocation and elongation of the right radial head at the elbow, deformity of the left radioulnar joint with shortening of the ulna, cervical and lumbar scoliosis, and spina bifida occulta. Results of an eye examination were normal. A hemogram, sedimentation rate, and blood chemistry values were normal. A biopsy specimen of a skin lesion in the neck showed elastosis of the papillary dermis and

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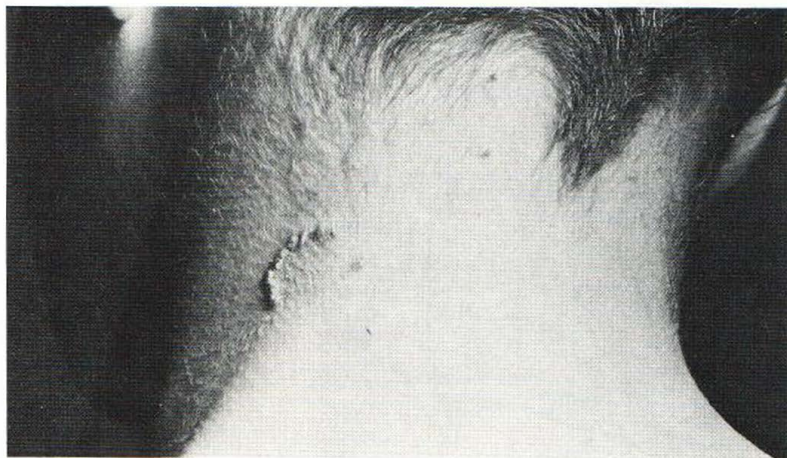


Fig. 1. Perforating elastoma on left side of neck.

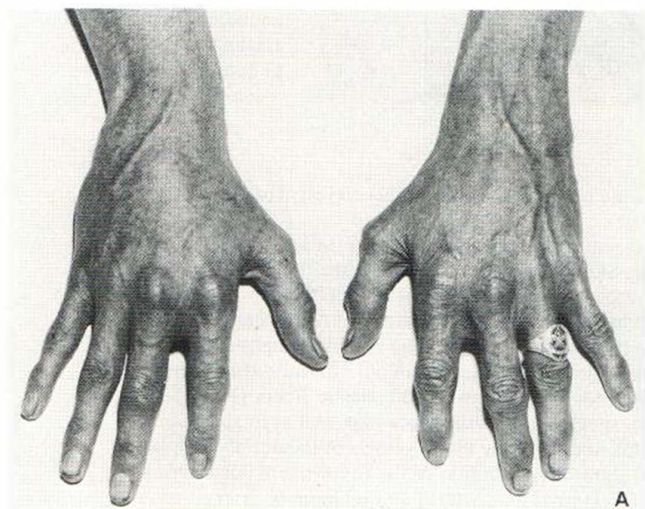


Fig. 2. (A) Small, atrophic, hyperpigmented hands with pronounced wrinkling and abnormal visibility of venous pattern. (From Whyte HJ, Winkelmann RK. Elastosis perforans [perforating elastosis]: the association of congenital anomalies, salient facts in the histology, studies of enzyme digestion and a report of necropsy in a case. *J Invest Dermatol* 1960; 35: 113-122. By permission of Williams & Wilkins Company.) (B) Atrophic skin of feet with varus deformity of right foot and dystrophic toenails.



*Fig. 3.* Skin biopsy specimen from trunk, showing abundant irregular "pseudo-elastin" material. (Aldehyde-fuchsin-Giemsa;  $\times 160$ .)

central plugging with basophilic necrotic material and eosinophilic elastic fibers, typical of perforating elastoma (2).

The patient died from a fracture of the cervical segment of the spinal column in a farm accident 3 months later. Autopsy showed no visceral disease. Skin biopsy specimens from the thorax, abdomen, right ankle, and right leg were obtained at the autopsy. Hematoxylin-eosin staining showed limited areas of epidermal atrophy and hypertrophy with hyperkeratosis and a patchy hypermelanosis of the basal cell layer. The blood vessels were dilated in the papillary dermis. The aldehyde-fuchsin-Giemsa stain for elastic tissue was normal in the papillary dermis, but in the middle dermis, there was a striking increase of elastic staining material in smooth sheets between the elastic fibers (Fig. 3). A mild atrophy of the dermis was present in the skin specimen from the thorax, as well as in specimens from the extremities. The elastic tissue staining was normal in the blood vessels of the dermis and the subcutaneous tissue. The elastic staining did not show any alteration of the following major arteries sampled at autopsy: aorta, right renal artery, right external iliac artery, and pulmonary artery.

## COMMENT

Acrogeria is a well-defined clinical entity involving mainly the face, hands, and feet and giving the patient an appearance of premature aging. As in the present case, the family history is usually negative, but two familial cases have been reported (1, 3), with a different mode of transmission in each case. The female sex is preponderant, but several male patients have been described (1, 3, 4). Premature birth is frequent (4, 5-11), and the defect is usually noticed at or soon after birth (4, 5, 9-11). The term "acrogeria" does not account for the abnormal visibility of the venous pattern, mainly on the anterior upper trunk, which is a common feature of the disease (3, 7-9, 11) and which was seen in our patient. This, together with the atrophy and the pigmentation, led us originally to classify this patient as having a congenital poikiloderma (2). The association of perforating elastoma with acrogeria is frequent (1, 3, 7), although perforating elastoma also has been found in other connective tissue diseases, such as the Ehlers-Danlos syndrome. Nail abnormalities consist predominantly of onychogryphosis (3). Various skeletal or orthopedic (or both) abnormalities, including spina bifida occulta, congenital dislocation of hips (9, 10), multiple fractures (4), clubfoot, pedes plani, and diaphyseal thinning of long bones (7), have



been seen in these patients. Micrognathia is also a frequent feature (1, 7, 11), and easy bruisability or formation of hypertrophic scars (or both) has been previously reported (3, 5, 6, 8, 9).

The histologic findings in the present case are consistent with those in previous cases. The epidermal changes include focal areas of atrophy, hypertrophy, hyperkeratosis, and patchy hypermelanosis of the basal cell layer, while in the dermis there is degeneration of the collagen fibers and presence of a "pseudo-elastic" material in the middle dermis which is not limited to an acral distribution (3, 4), as well as atrophy of the dermis or the subcutis (3, 4, 9), which is not very well specified in most cases (3). The electron microscopic findings reported by De Groot et al. (3) include a decreased number of normal elastic fibers, the presence in the middle dermis of "pseudo-elastic" material consisting of granular and fibrillar material, immaturity of collagen fibers, and large abnormal fibroblasts with many vacuoles and widened endoplasmic reticulum. The sheets of elastic staining material in the dermis of our patient are the equivalent of the "pseudo-elastic".

The clinical appearance of acrogeria is different from that in other premature aging syndromes, such as progeria, metageria (6), and mandibuloacral dysplasia. There was no evidence of Ehlers-Danlos syndrome Type IV in this case, including the autopsy findings.

However, from a nosologic point of view, further studies of this kind of patient are needed to determine if acrogeria might represent the benign end of a large collagen III defect spectrum, as has been suggested previously (12).

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