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Familial acrogeria in a brother and sister

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ABSTRACT

Acrogeria is one of the premature aging syndromes with an unknown mode of inheritance. Familial cases are quite rare. A report of the disease in a brother and sister demonstrating a possible autosomal recessive inheritance is described. An unusual finding was bilateral corneal opacities in both the patients.

Key Words: Pangeria, Atrophy, Poikiloderma

INTRODUCTION

Acrogeria is a very rare genetic disease characterized by atrophy of skin with loss of subcutaneous fat most prominently over the acral parts giving rise to a prematurely aged appearance.[1] The disease was first described in the year 1941 by Gottron who reported the disease in two siblings. After the original report most of the subsequent cases described have been sporadic in nature with no family history.[1,2] There has also been a preponderance of female patients in the literature, with 17 females and only 4 males reported till 1980.[3] The mode of inheritance of the disease has not been established clearly to date. A brother and a sister affected with the disease and born of a consanguineous marriage are reported herein.

CASE REPORT

Two siblings, a sister and a brother aged 20 years and 24 years respectively, reported to our department with complaints of premature graying of hair and excessive wrinkling of skin over the face, hands and feet. They were born of a consanguineous marriage and were the second and the third in birth order in their family. There was no evidence of premature aging either in the parents or in the three other siblings.

On examination, they were found to have an emaciated and prematurely aged appearance with a remarkable facies, having a thin, pinched nose and peculiar pattern of wrinkling around the mouth and eyes. The skin of the face, hands, feet and neck showed gross atrophy with loss of subcutaneous fat. Similar changes, though of a less severe degree, were found over the forearms and legs, but the proximal parts of limbs and the trunk were relatively normal

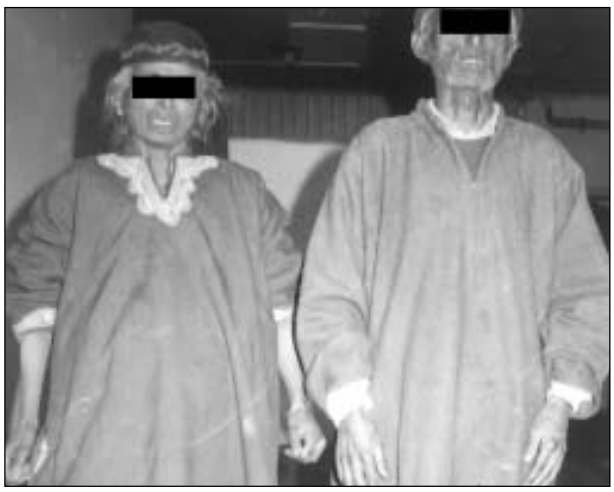

Figure 1: Brother and sister with acrogeria

(Figure 1). The height of both the patients was within normal limits (156 cm and 167 cm for the sister and brother respectively). However, both were grossly underweight, 43 kgs and 49 kgs respectively. This was below the third percentile as calculated for the respective ages and heights. Examination of the cardiovascular, pulmonary and central nervous systems revealed no abnormalities. The muscle mass was well preserved and there was no evidence of any systemic abnormalities.

Examination of the teeth, mucosae and nails was within normal limits. Examination of the scalp hair revealed premature canities in both patients but the sister was more severely affected than her male sibling. There was no evidence of any bruising, poikiloderma or telangiectasias, but the veins over the face and extremities appeared unduly prominent. Examination of the eyes revealed bilateral corneal opacities in both the siblings, with the eyes involved unequally on both sides. There was no evidence of cataract formation.

There were no grossly abnormal laboratory parameters. A skin biopsy performed on the hand in the male sibling revealed a relatively normal looking epidermis with swollen and disorganized collagen fibres in the dermis. The subcutaneous fat layer was almost invisible and it had been replaced almost entirely by connective tissue indistinguishable from that in the dermis.

DISCUSSION

Acrogeria is a genetically determined skin disorder with an unclear mode of inheritance.[3] The disease has been reported mainly in females and almost all the cases have been sporadic ones.[3] In fact, occurrence of acrogeria in two generations of a single family has been reported only once, i.e. in the original description by Gottron.[3] We are reporting here the occurrence of the disease in two siblings, viz. a brother and sister, who had a classical presentation of acrogeria with additional features of bilateral corneal opacities. The history of consanguinity and presence of three normal siblings in the family point towards a possible autosomal recessive mode of inheritance. The same pattern of inheritance has been reported in the original case report by Gottron, but a later report by W P DeGroot et al[3] mentioned the possibility of a dominant mode of inheritance. A number of non-dermatological findings have been documented in the literature,[1-3] but we did not find any mention of bilateral corneal opacities in any of these reports.

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