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Case Report

A rare case of acrogeria

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Introduction

Acrogeria is an extremely rare genetic syndrome characterized by nonprogressive form of skin atrophy involving mainly the distal parts of the extremities. The other features include characteristic facies with pinched face and hollow cheek, 'owl eyed' appearance, beaked nose, thin lips and skeletal defects. The disease was first described in the year 1941 by Gottron who reported the disease in two siblings.¹ After the original case report, most of the subsequent cases described have been sporadic in nature with no family history.² Although most cases are sporadic, both autosomal recessive and autosomal dominant inheritance have been reported with a female predominance.³ Prognosis of these patients is good as they have no tendency to develop athetosis or diabetes mellitus. We report a case of acrogeria for its rarity.

Case report

A 24-year old male patient, a product of second degree consanguineous marriage presented with history of thin fragile skin over the distal portion of all four extremities since birth. There was history of redness and photophobia in both eyes. There was no history of delayed healing of wounds, history of bruising, chest pain, palpitations, recurrent infections or appearance of fluid filled lesions on sun exposure. He was born as a full term normal delivery, second of four siblings with no complications at the time of delivery. There were no similar complaints in his family or siblings.

General examination revealed height of 160 cm and a weight of 40 kg (BMI: 15.63). General and systemic examination revealed no abnormalities. Musculoskeletal examination revealed flexion deformities at the proximal and distal interphalangeal joints of the fourth and fifth toes of both feet. However no joint contractures were present and movements were normal. Dermatological examination revealed characteristic facies with beaked nose, pinched hollowed out cheeks with maxillary hypoplasia, thin lips and micrognathia. 'Owl eyed' appearance was present and ears were prominent [Figs. 1 and 2]. Skin over the whole body appeared dry with no evidence of ichthyosis. The skin over the distal third of all four extremities was extremely thin and wrinkled with loss of subcutaneous fat and prominent dilated veins [Figs. 3 and 4]. Areas of atrophic scarring were present over both knees. Both eyes showed conjunctival suffusion but vision was normal. Examination of hair, nails, teeth and genitalia was normal.

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Fig. 1 – Characteristic facies of acrogeria: Front view.

There were no abnormal laboratory parameters. X-ray of dorsolumbar spine and extremities were normal while ECG & echocardiography did not show any abnormalities. Skin biopsy revealed replacement of subcutaneous tissue almost entirely by connective tissue with associated epidermal atrophy.

Discussion

Acrogeria is one of the classic congenital premature ageing syndromes which include Werner's syndrome, progeria and acrogeria. These are rare genetic diseases associated with accelerated ageing of the skin and other tissues.⁴

Acrogeria is an extremely rare genetic syndrome which begins at birth or soon afterwards characterized by mild, nonprogressive form of skin atrophy involving mainly the distal parts of the extremities. Most cases are sporadic: however both autosomal recessive and autosomal dominant inheritance have been reported with a female predominance.

Fig. 2 – Characteristic facies of acrogeria: Lateral view.

Fig. 3 – Prominent veins over distal extremities.

Mutations in the COL3A1 gene were identified in varied phenotypes.⁵ In fibroblast culture, a reduction of RNA messenger cells in collagen types I and II with reduced life expectancy of fibroblasts and morphological alterations typical of premature ageing have been found. This is perfectly compatible with the patients' aged phenotype.⁶

The essential feature is atrophy of the skin and subcutaneous tissue, giving an aged appearance. Predominant cutaneous features include dry, thin, transparent and wrinkled skin, especially over the hands and feet and patients may have easy bruising, poikiloderma and telangiectasia. All such patients have characteristic facies similar to our case with pinched face and hollow cheeks, 'owl eyed' appearance, beaked nose and thin lips.⁷ Our patient did not have features of poikiloderma or telangiectasia. Nail changes include atrophy and thickening which were absent in our patient.⁸

In the skin histopathology, there is atrophy of the dermis and subcutis. The collagen fibres are loose and dispersed, while the elastic fibres are always fragmented. The epidermis is spared. All these features were present in our patient.

Fig. 4 – Thin acral areas.

A number of non-dermatological features have been documented in literature including acro-osteolysis of the distal phalanges, delayed cranial suture closure with wormian bones, linear lucent defects of metaphyses, antegonial notching of the mandible, avascular necrosis of the femoral heads, soft tissue calcification, osteoporosis and coxa valga,^{9,10} but no skeletal defects were present in our patient.

Prognosis of these patients is good as they have no tendency to develop atheroma or diabetes mellitus. The most important appears to be the relationship with Ehlers–Danlos syndrome type IV, i.e. a variant characterized by only slight skin hyperextensibility and joint hypermobility. This variant differs, however, from acrogeria clinically by a high incidence of rupture of great vessels and bowel, and biochemically by a total or partial lack of type III collagen, resulting from a structural defect in the alpha 1(III) chain, which enhances the susceptibility to proteinases.¹¹ Our patient had no features of EDS.

Various cases have been published, most in the European literature and predominantly affecting women. This case has been reported due to its rarity in this part of the world.

Conflicts of interest

All authors have none to declare.

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