

Case Report

A case of acrogeria-a rare premature ageing syndrome

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Abstract A case of acrogeria of a 9-month-old boy is reported here who presented with dry, scaly skin, birdlike facies, prominent frontal tuberosities, recessed chin, dry, thin skin with generalized subcutaneous fat loss, prominent veins, prominent eyes, thin and brittle nails, poorly developed musculature and hair loss at the back of scalp.

Key words

Premature ageing syndromes, progeria, acrogeria, pangeria

Introduction

Increasing age causes many anatomical and functional changes in human skin, but some of these may be the result of cumulative damage due to sun exposure, etc. There are more than 150 diseases which manifest one or more features of apparent premature ageing.

All the premature ageing syndromes¹ are probably inherited disorders although they may not present in the first few years of life. Cutaneous changes which may be a sign of a premature ageing syndrome include atrophy, loss of cutaneous fat, wrinkling, greying and loss of hair, nail dystrophy, defective pigmentation, poikiloderma, sclerosis and ulceration.

The conditions associated with cutaneous signs of premature ageing are shown in **Table 1**.

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Table 1 Conditions associated with premature ageing

1. Classical inherited premature ageing syndromes:
 - a. Pangeria² (Werner's syndrome)
 - b. Progeria³ (Hutchinson- Gilford syndrome)
 - c. Acrogeria (Gottron's syndrome)
2. Other congenital progeroid⁴ syndromes:
 - a. Trisomy 21 (Down's syndrome)
 - b. Neonatal pseudohydrocephalic progeroid syndrome (Weidman-Rauchenstrauch)
 - c. Osteodysplastic geroderma
 - d. Wrinkly skin syndrome
 - e. Familial mandibulo-acral dysplasia
3. Photosensitivity e.g. Poikiloderma congenitale, xeroderma pigmentosum
4. Cutix laxa
5. Diabetic cheiroarthropathy
6. Prolidase deficiency
7. Generalized lipodystrophy

Case report

A 9-month-old boy reported to the Dermatology department of CMH, Hyderabad with dry, scaly skin since 2 months of age. Baby was born of a consanguineous marriage, having an elder sister. There was no evidence of premature

aging either in the parents or in the sibling. On examination there was birdlike facies, prominent frontal tuberosities, recessed chin (**Figure 1**), dry, thin skin with generalized subcutaneous fat loss and prominent veins, prominent eyes (**Figure 2**), poorly developed musculature, thin and brittle nails (**Figure 3**), and hair loss at the back of scalp. Examination of the cardiovascular, pulmonary and central nervous systems revealed no abnormalities. There were no grossly abnormal laboratory parameters. A skin biopsy performed on the hand revealed a relatively normal looking epidermis with swollen and disorganized collagen fibers 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 was first described in the year 1941 by Gottron⁵ 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. There has also been a preponderance of female patients in the literature.⁶

The term 'acrogeria' refers to premature ageing of the extremities. It begins at birth or soon after birth. Most cases occur without a family history but both autosomal recessive as well as autosomal dominant inheritance has been reported. COL 3 mutations⁷ cause variable phenotype, including Gottron-type acrogeria.

It is characterized by cutaneous atrophy and loss of subcutaneous fat, particularly over the distal extremities. The skin becomes dry,



Figure 1 birdlike facies, prominent frontal tuberosities, and recessed chin



Figure 2 Poorly developed musculature and loss of subcutaneous fat.



Figure 3 Thin and brittle nails.

thin, transparent and wrinkled, especially over the hands and feet, although the trunk and face may be affected to a lesser extent. The veins are prominent, and there may be easy bruising, poikiloderma and telangiectasia. The nails may be atrophic or thickened. The face appears pinched with a hollow-cheeked 'owl eyed' appearance, a beaked nose and thin lips. Micrognathism may be present. The lack of subcutaneous fat accentuates the appearance of premature senility. Some patients have low birth weight and persistent short stature, but the general health and life expectancy are normal. The hands and feet may be very small.

All the premature ageing syndromes show marked reduction in fibroblast growth potential, therefore fibroblast culture⁸ is required to confirm the diagnosis of premature ageing syndromes, unfortunately this facility is not available in Pakistan.

Cases are occasionally described which do not fit easily into any of the recognized categories and have been termed metageria and acrometageria.

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