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Case Report

A Rare Case of ACROGERIA from Odisha

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Abstract
Acrogeria, Gottron type is a rare premature aging syndrome. Characteristic signs include fragile, thin skin on the hands and feet. Other parts of the body (e.g., face, forearms, and lower legs) are variably affected. It is generally considered to be a mild, non progressive skin atrophy due to the loss of the fatty tissue directly under the skin. Although most cases are sporadic, both autosomal recessive and autosomal dominant inheritance have been reported with a female predominance. Prognosis of these patients is good as they have no tendency to develop atheroma or diabetes mellitus. We report a case 11 year old female child of acrogeria for its rarity.

Introduction

Acrogeria is an extremely rare genetic syndrome characterized by non progressive 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] Approximately forty cases have been reported in the medical literature.[4]

Case Report

Here we report a girl of 11 years who came to the pediatric department of M.K.C.G medical college with complains of loosening of skin and failure to gain weight for last two years. Dermatological examination revealed characteristic facies with beaked nose, pinched hollowed out cheeks with maxillary hypoplasia, thin lips and micrognathia. [Figs. 1]. Skin over the whole body appeared dry with no evidence of ichthyosis. Examination of hair, nails, teeth and genitalia was normal. Weight of the child was 26kg and the height was 133cm.She was born out of non consanguinous marriage through normal vaginal delivery and had attained all her developmental milestones at the appropriate age. There was no similar family history and her siblings were normal. She is performing very well at a English medium school. For last two years her classmates are teasing because of her looks. Due to the typical physical

characteristics, the diagnosis of Acrogeria, Gottron type was made. All routine investigations including thyroid profile and lipid profile were normal.

The parents were counseled about the disease and its non progression.

Fig 1 showing characteristic facies

Discussion

The mode of genetic inheritance is not accurately known. It has been considered autosomal dominant and autosomal recessive, though most reported cases own a positive family background. Mutations in the COL3A1 gene, located at chromosome 2q31–q32, have been reported in varied phenotypes, including acrogeria and vascular rupture in Ehlers-Danlos' syndrome (more especially type IV).[5] In the fibroblast culture, a reduction of RNA messenger cells in collagen types I and II was found, as well as reduced life expectancy of the fibroblasts most prematurely showing morphological alterations typical of aging. This seems perfectly compatible with the patients' aged phenotype. The differential diagnosis include Werner's syndrome and progeria. In Werner's syndrome there is abnormally slow growth rate, and growth ends at puberty. As a result, affected individuals have unusually short stature and low weight even relative to height. By age 25, those with the disorder typically experience early graying (canities) and premature loss of scalp hair. Children with progeria usually develop the first symptoms during their first few months of life. The earliest symptoms may include a failure to thrive and a localized scleroderma-like skin condition. As a child ages past infancy, additional conditions become apparent usually around 18–24 months. Limited growth, full-body alopecia, and a distinctive appearance are all characteristics of progeria.

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 hyper extensibility and joint hyper mobility. 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[6].

Conclusion

There is currently no specific treatment available for either of these so-called progeroid syndromes. Though the facial appearance looks like progeria, but absence of alopecia gives a clue towards acrogeria. The prognosis appears to be far better in acrogeria.

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