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COCKAYNE'S SYNDROME

K Krishna

A 10-year-old boy, a product of consanguineous marriage was diagnosed clinically as a case of Cockayne's syndrome because of delayed milestones, deaf mutism with spastic paraplegia, dwarfism, salt and pepper fundus, typical facies and a photosensitive rash on the butterfly area of the face.

Key Word: Cockayne's syndrome

Introduction

Cockayne, in 1936, first described a 7-ear-old English girl and her 6-year-old brother with dwarfism, retinal atrophy and deafness.[1] On the basis of neuropathologic findings,Cockayne's syndrome is regarded as a sudanophilic leukodystrophy of the symptomatic type and as a subgroup of the Pelizaeus-Merzbacher disease.Also, it has been viewed as a calcifying vasopathy.[2]

Clinical onset is in infancy after a normal first year of life, and death in the 2nd to 3rd decades.[3] It differs from other diseases of radiation sensitivity and from those of increased chromosomal breakage in that it is not associated with an increased rate of carcinogenesis.[4] However, pneumonitis, renal disease, hypertension and advanced atherosclerosis may occur.[5]

Case Report

A 10-year-old male cachectic dwarf,a product of consanguinous marriage, presented with inability to hear and talk since birth and a photosensitive skin rash since the age of one year. There were delayed milestones. He was 96 cm tall had a head circumference of 45.6 cm and weighed 12 kg.Theere was deaf mitism with spastic paraplegia and ataxic gait. Dermatological examination revealed photosensitivity, diffuse scaly hyperpigmentation in the butterfly area of the face and a wizened appearance with bird head facies.

Other findings included salt and pepper fundus with mottled machula, kyphoscoliosis and caries teeth. Haematological parameters and radiography of chest, hands and skull were within normal limits.

Discussion

Cockayne's syndrome is an autosomal recessive, degenerative disease with cutaneous, ocular, neurologic and somatic abnormalities. It is characterised by cachectic dwarfism, deafness and pigmentary retinal degeneration with a characteristic "Salt and pepper" appearance of the retina. Variable eye signs are loss of macular reflex,cataracts, nystagmus, lack of tearing and poor response to mydriatics.3 The skin has photosensitivity and diffuse hyperpigmentation without the excessive pigmentary abnormalities as seen in exeroderma pigmentosum.There is marked loss of subcutaneous fat resulting in a "wizened" facies. Neurologic abnormalities include deafness, peripheral neuropathy, normal pressure hydrocephalus and microcephaly. Pathologically, there is loss of neurons without inflammatory reactionor deposition of material. The present case did not show some features like long extremities, joint contractures, loss of

From the Department of Skin and VD, Rural Medical College, Pravara Medical Trust, Loni-413736, India.

Address correspondence to : Dr K Krishna

subcutaneous tissue, and cold blue extremities.[4]

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ADDENDUM

The names of authors of article"Phentyon Therapy in Resessive Dystrophic Epidermolysis Bullosa" it found wl 61 No. 3 pages 170-170 have given the name of single author due to oversight. The authors of the above are :

V R Rege

M F Mascarenhas

V Coelho

K Singh

A Kudchadkar

Editor