DE SANCTIS CACCHIONE SYNDROME

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A case of de Sanctis Cacchione syndrome in a female infant is presented.

Key Words : De Sanctis Cacchione syndrome, Xeroderma pigmentosum

Introduction

De Sanctis Cacchione syndrome is an extremely rare autosomal recessive condition characterized by xeroderma pigmentosum, ocular, neurologic and somatic abnormalities.

Case Report

A 9-month-old female baby, the only child of 2^0 consanguineous parents, was seen in the department of Paediatrics with history of generalized tonic clonic seizures of 2 months duration. Her antenatal period was uneventful. She was an SGA (small for gestational age) baby and had asphyxia neonatorum. All mile stones of development were grossly delayed.

She developed multiple pigmented and depigmented spotty lesions on the face at the age of 3 months. The lesions gradually increased in number and also appeared on the trunk and extremities. She had photophobia also.

On examination she was a lethargic, sick looking baby weighing just 3 kg. She had microcephaly, head circumference being 34.5 cms. She also had hypotonia, hyporeflexia and splenomegaly.

Dermatological examination showed multiple freckles, depigmented macules and telangiectasia on the face, upper trunk and extremities. Scalp hair was brownish and eyebrows were hypopigmented and she had ichthyosis. Nails were normal. Skin biopsy was consistent with xeroderma pigmentosum.

In view of the skin lesions, photophobia, microcephaly, mental retardation, delayed milestones and seizures, a diagnosis of De-Sanctis Cacchione syndrome was made.

Discussion

In 1932, De Sanctis and Cacchione reported three brothers who had cutaneous and ocular manifestations of classic xeroderma pigmentosum and additional neurologic and somatic abnormalities. The De-Sanctis Cacchione syndrome includes microcephaly with progressive mental deterioration, hyporeflexia or areflexia, choreoathetosis, ataxia, spasticity, shortening of Achilles tendon with eventual quadriparesis, markedly retarded growth and immature sexual development. Progressive sensorineural deafness, abnormal electroencephalographic findings and epilepsy can also occur.

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The complete De Sanctis Cacchione syndrome has been reported in very few patients with xeroderma pigmentosum. However, many patients have one or more of its neurologic features.[2]

Of the 286 xeroderma pigmentosum patients reported before 1949,[3] 41 had severe neurologic complications. Six of 15 patients reported in 1974[4] had neurologic symptoms. The most severely affected patients have onset of neurologic symptoms in infancy, as in our patient.

Untreated patients with xeroderma pigmentosum often die at an early age from the consequences of multiple cutaneous neoplasms with local invasion, metastasis, or secondary infection. Patients with onset in infancy have a particularly poor prognosis. Affected siblings tend to have similar clinical disease including presence or absence of neurologic involvement.

References

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