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INCONTINENTIA PIGMENTI

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A 2-year-old female case of incontinentia pigmenti showed unusual pigmentation over the face and scalp and several other features associated with this disorder such as strabismus, nystagmus, nail dystrophy, delayed dentition and spastic paresis.

Key words : Incontinentia pigmenti.

Incontinentia pigmenti was first described by Bloch[1] in 1926 and later by Sulzberger[2] in 1928. It is an X-linked, dominantly-inherited disorder dominant in females and lethal in males. The disease is characterised clinically by three stages.[3] First stage consists of erythema and bullae which occur predominantly on the extremities and are present at birth or shortly after birth. In the second stage, linear, verrucous lesions are seen predominantly on the trunk and extremities. Each of these two stages usually lasts about 2 months.[4] In the third stage, brownish black pigmentation in bizarre, streaky, spidery, fountain-spray patterns and in whorls is seen on the trunk and extremities. This pigmentation slowly fades away and becomes imperceptible by the 2nd or the 3rd decade. But the sequence of these three stages is irregular and their duration is variable, and it is also possible that these stages occur in utero.[5] In addition to the skin changes, other ectodermal anomalies affecting eyes, teeth, CNS, skeletal and cutaneous appendages occur in about 60% of the patients.[6] The eye changes which occur in 35% of the cases according to Carney[7] include cataract, strabismus, nystagmus, optic atrophy, blue sclerae and exudative chorio-retinitis. The teeth may show delayed dentition, pegged or conical crowns, malformations and missing teeth. Neurological abnormalities include mental retardation, seizures and spastic paresis. Skeletal abnormalities such as dwarfism, syndactyly, microcephaly, spina bifida, extra ribs, and arm or leg length discrepancies may also occur. Cicatricial, patchy alopecia may appear in about 25% of the cases and nails may also show dystrophic changes.[8]

The present case is reported as it showed most of the features associated with this disorder, and in addition presented with unusual pigmentation over the face and scalp since birth.

Case Report

A 2-year-old female child born to non-consanguinous parents with an un-eventful normal vaginal delivery was brought with brownish black pigmentation over the body since birth and delayed developmental milestones. This child is the youngest of the three children (all females), the other two female children being healthy. Mother volunteered the history of recurrent seizures and cutaneous infections in the child. The child was marasmic and had physical and mental retardation. Spastic paresis was seen on the left side. Ophthalmological examination revealed strabismus and early optic atrophy in both the eyes. Delayed dentition was evident with only one pegged upper incisor. The nails over the left ring finger and right thumb showed dystrophic changes. The scalp hair was thin, scanty and lustreless. Brownish black pigmented patches in an irregular bizarre pattern in the form of streaks, whorls and waves were present on the trunk, extremities, chin, forehead and vertex.

Routine investigations on blood, urine and stools did not show any abnormality. Blood

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VDRL test in both the mother and the child was negative.

Histopathology of the skin lesion taken from the trunk revealed incontinence of melanin in the dermis with basal cell layer containing abundant melanin. Dopa staining could not be done due to lack of facilities.

Comments

In our case the pigmentation was evident since birth indicating that the earlier bullous and the verrucous stages might have occurred in utero. This is quite possible and has been recorded earlier also.[5] In addition, our case had unusual pigmentation over the face, chin, forehead and vertex, a feature which does not seem to have been recorded earlier.

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