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PROTEUS SYNDROME

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A case of proteus syndrome in a 20 year old male is reported. Hemihypertrophy, asymmetric megalodactyly, linear epidermal naevus, naevus flammeus, angiokeratoma, lymphangioma circumscriptum, thickening of the palms and soles, scoliosis and varicose veins were present. There are only few reports of these cases in adults. The syndrome has not been reported from India.

Key Words : Hemihypertorphy, Proteus syndrome

Introduction

Proteus syndrome is a hamartomatous disorder described by Wiedemann et al 1983.[1] Since then about 50 cases have been reported in World literature, only 4 being in adults.[2] The typical clinical features include progressive and asymmetric megalodactyly, hemihypertrophy, subcutaneous masses, localised cerebroid thickening of the palms and soles and linear skin lesions. The occurrence is sporadic.[2] In this report we describe a case of proteus syndrome in a 20 year adult male from Bihar. There are no reports to the best of our knowledge of a similar case from India.

Case Roport

A 20-year-old male, born full term following a normal delivery to non-consanguinous parents was noticed to have asymmetric enlargement of the fingers and toes at birth and large red patches on the trunk, upper limbs and the left lower limb. There was progressive asymmetrical enlargement of the left half of the trunk, left lower limb and the fingers and toes as he grew up. During early childhood he developed raised linear lesions on the upper and lower limbs on the hypertrophied side. His developmental milestones were normal and scholastic performance good. He had no associated systemic symptoms except for bleeding per rectum. Mother and father had no congenital anomalies. He had no siblings.

Patient was thin built, height 160cm, arm span 162cm. There was partial hemihypertrophy of the left half of the trunk, left upper and lower limbs (Fig.1).

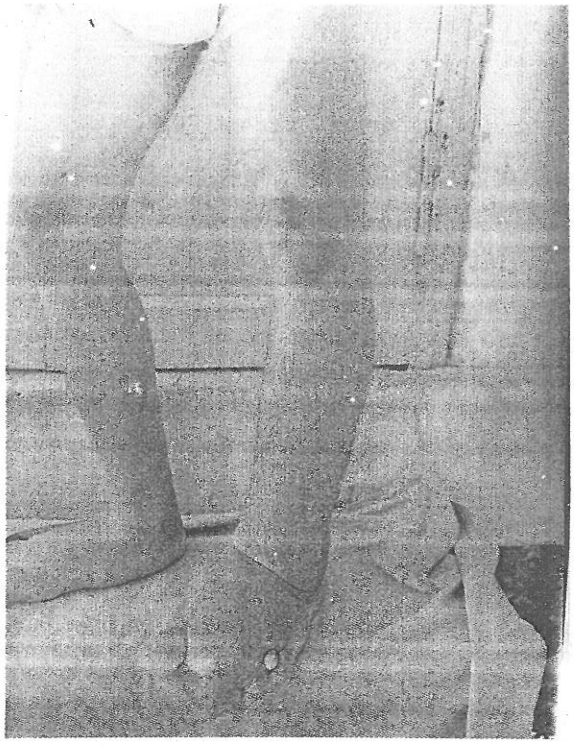

Fig.1. Hypertrophy of the left lower limb and enlarged 1st and 2nd toes

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The difference in girth was 1 cm in the arms, 2 cms in the thighs and 5 cms in the legs. There was asymmetric macrodactyly affecting the hands and feet. The palms and loles were grossly thickened with mild cerebroid hyperplasia.

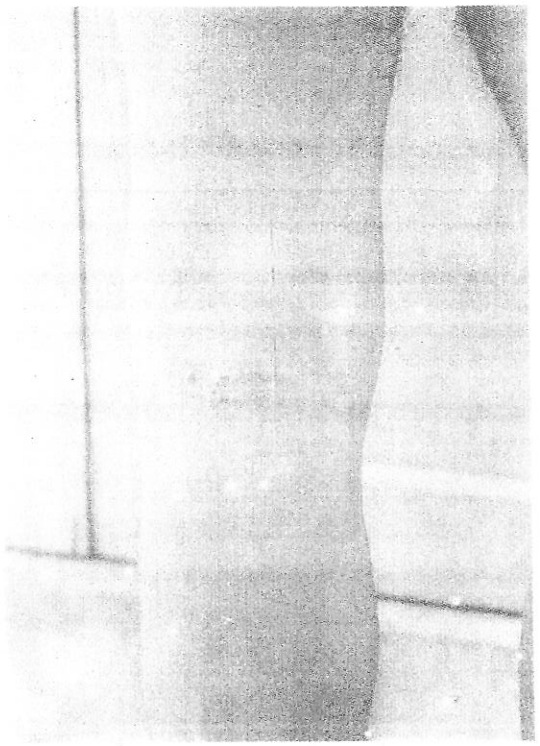

Fig.2. Naevus flammeus on the left lower limb. Angiokeratoma are seen in the popliteal fossa.

The following lesions were also present:

(1) There was a large naevus flammeus involving the whole of the upper trunk and left upper and lower limb (Fig.2) and proximal half of right upper limb. He also had a naevus flammeus over the rights soft palate.

(2) Linear epidermal naevi on the left upper and lower limb.

(3) Angiokeratoma were present on the left posterior axillary fold, penile and scrotal skin and left popliteal fossae.

(4) Lymphangioma cirumscriptum in the right cubital fossa.

There were varicose veins in the hypertrophied left lower limb. He had scoliosis. There were no bony exostoses. There were no obvious defects in the eye. Per rectal examination showed grade I haemorrhoids.

Biopsies taken from linear epidermal naevus and lymphangioma circumscriptum were confirmatory. X-ray spine showed scoliosis. Chest X-ray and ultrasound abdomen and pelvis were normal.

Comments

Patient presented with several of the classical features of proteus syndrome.[2,3] The linear epidermal naevus, varicose veins were present only on the hypertrophied side. Although the naevus flammeus was present on both sides of the trunk, the lesion was more prominent on the hypertrophied side. Lymphangioma circumscriptum was noted on the opposite upper limb. The palms and soles were grossly thickened but cerebroid appearance was not striking. Palmar or plantar masses have been noted in 64.7% (22/34) of patients reviewed in one series and have been described as fairly typical of this syndrome.[3] Subcutaneous massese, a fairly common occurrence in patients with proteus syndrome, were not seen in our patient.[3] Ultrasound of the abdomen and pelvis also did not reveal any masses. Pelvic lipomas has been reported in patients with proteus syndrome.[4] There were no clinically evident exostoses in our patient. The common sites of exostoses are on the skull, fingers and toes. Among the unusual features noted were penile hypertorphy reported only in 1 earlier case.[3]

As there are only few reports of proteus syndrome in adults the long term prognosis is not known. Serious complications evolve slowly depending on the location of tissue over growth. Testicular tumours have been reported in 2 cases, having developed at age 5 and 14 years.[5] Our patient had no evidence of malignancy at the time of examination. A close follow up of these patients is necessary because of the reported occurence of non-epithelial neoplasms and also mechanical problems arising from tissue overgrowth.[5]

As proteus syndrome has only been recently recongnised it is possible that the apparent rarity of the syndrome from India is probably because it has been reported under different names or grouped with other conditions.

References

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