

Case Report : Proteus syndrome - an unusual hamartomatous disorder.

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Introduction

Proteus syndrome is a congenital hamartomatous disorder of neuroectodermal origin distinct from other neurocutaneous disorder such as Neurofibromatosis or Klippel Trenauney Weber Syndrome. It was first reported in 1979 and was named Proteus Syndrome after the Greek God proteus who could change his shape at will to avoid capture.

Case report

A two year old female child presented with history of mental retardation, seizures, enlarged head, protrusion of the right eye, a depressed nasal bridge and swelling over the right side of nose and increasing abdominal girth.



Figure 2. Photograph of hands showing macrodactyly of the right index and middle fingers



Figure 1. Clinical Photograph showing macrocrania, depressed nasal bridge, left lower limb hypertrophy.



Figure 3. Skull Radiograph - Macrocrania with depressed nasal bridge

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On examination she was found to have macrocrania and achromic naevus on the right side of the nose (Fig-1), macrodactyly of the right index and middle finger (Figure 2) and an obvious soft tissue bulge over left side of abdomen which was doughy on palpation. Her left lower limb was grossly enlarged and pigmented. Gyriform hyperkeratosis of left sole was observed. A skeletal survey was performed and contrast enhanced CT scan of head, abdomen and left lower limb was performed on Hitachi W-2000 Spiral CT Scanner. The skeletal survey revealed macrocrania (Figure 3). Malformed vertebral bodies, enlarged metacarpals and phalanges of right middle and index finger and uniform enlargement of bones of left lower limb (Figure 4).



Figure 4. Lower Limbs Radiograph - Enlargement of bones of left lower limb

Discussion:

Macrocrania, hemihypertrophy, subcutaneous lipoma, macrodactyly and epidermal naevi are classical manifestation of Proteus Syndrome (5). This syndrome shares significant overlap with other hamartomatous disorders like Klippel Trenauney Weber Syndrome

Neurofibromatosis, Riley-Smith Syndrome, etc. Cutaneous manifestations such as epidermal naevi, hyperkeratosis and subcutaneous mass (3) in addition to the features described above permits a confident diagnosis. Other features which is typical of this syndrome are calvarial exostosis, spectrum of ocular anomalies such as macrophthalmia, heterochromia iridis, cataract and chorioretinitis. Hypertrophy of scapula and ribs, megaspondylodysplasia, kyphoscoliosis, macroorchidism and penile hypertrophy (4,5). Involvement of both kidneys by this disorder with altered RFT has been described (2).

Given the protean manifestation of this disorder we believe that diagnosis of this syndrome must always be considered in patients, with features described above.

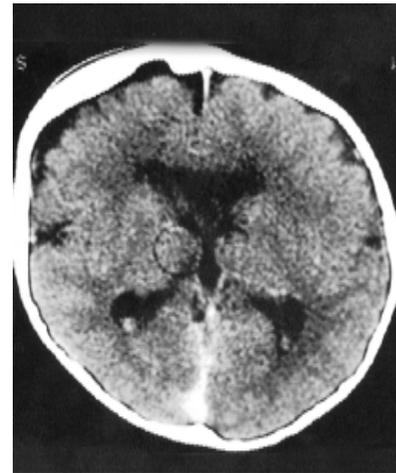


Figure 5. CT Brain - Ventriculomegaly with cortical atrophy

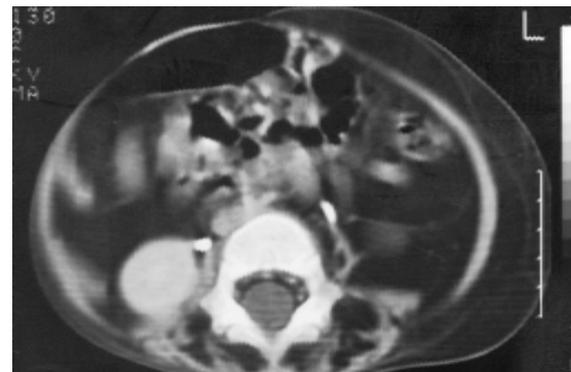


Figure 6. CT Abdomen - Abundant fat deposition in Anterolateral aspect of left anterior abdominal wall, mesentery and retroperitoneum.

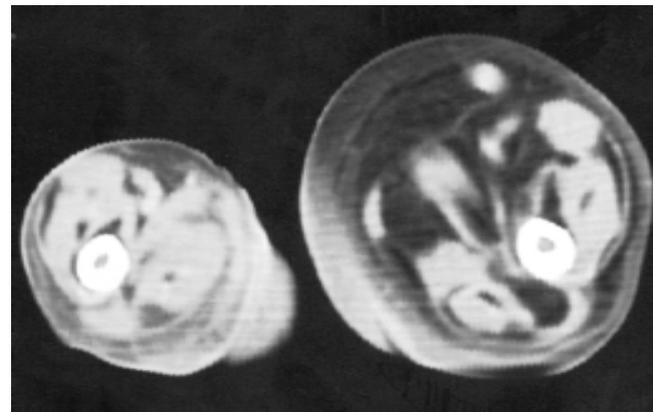


Figure 7. CT lower limbs - Lipodermal hyperplasia of left lower limb

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