

Case Report

Proteus syndrome: a case report

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Abstract Proteus syndrome (PS) is a congenital hamartomatous dysplasia. This sporadic disorder involves the skeletal system, soft tissues, skin and vascular system. It is a rare disease and frequently over or under diagnosed. The clinical picture mimics Klippel-Trenaunay-Weber syndrome. Only the presence of full blown syndrome should prompt the diagnosis. We present a typical case of PS who was mishandled by general practitioners and physicians and was treated as a case of acromegaly.

Key words

Proteus syndrome

Introduction

Proteus syndrome (PS) is a rare disorder of patchy or mosaic postnatal overgrowth of unknown etiology. The onset of overgrowth typically occurs in infancy and can involve any tissue of the body. Commonly involved tissues include connective tissue and bone, skin, central nervous system and eye, but it apparently can affect any tissue. Diagnosing PS is difficult and the diagnostic criteria are controversial. Effective management requires knowledge of the wide array of manifestations and complications of the disorder and a team approach that includes the geneticist, surgeons, and other specialists.¹

Our patient presented with epilepsy as the only symptom. This complaint although described in patients of PS is an extremely rare finding as compared to other neurocutaneous syndromes.

Case report

Our patient a 22-year-old male presented to

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medical out-patient department with complaints of grand mal seizures since 2 years of age. He had an unusually large face with jaw protrusion, large feet and hands. Due to his clinical appearance and symptoms he was previously labeled as acromegaly and was treated accordingly. Later on the case was referred to dermatology OPD for review.

On detailed history it was found that the patient was the youngest of 4 brothers and 3 sisters who were all healthy and normal looking. The patient was born normally but after 2 years there was an unusually increased growth of left side of face and left upper limb and both lower limbs. Gradually his tongue increased in size and he had difficulty in talking. The patient was mentally slow of his age for which reason he left school at an early age. He started having fits at 2 years of age which gradually increased in severity in the passing years.

On examination the patient was well-oriented but slow in response to questions. He had a characteristic overgrowth of left side of the body. There was left hemi-facial hypertrophy with jaw protrusion on left side (**Figure 1**). There were multiple lipomas, sebaceous cysts and comedones on left side of face and



Figure 1 Left hemi-facial hypertrophy with jaw protrusion on left side.



Figure 4 Left hand significantly larger than the right.



Figure 2 Multiple sebaceous cysts and comedones on left side of face and verrucous epidermal naevus on left side of the neck



Figure 5 Huge feet with relative overgrowth of second and third toes of both feet. His father's foot photographed for comparison.

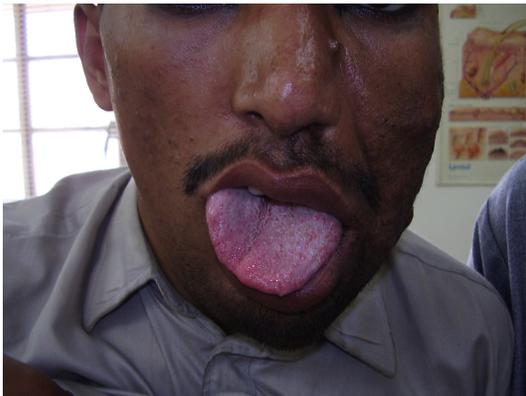


Figure 3 Left side of tongue larger than right.

verrucous epidermal naevus on left side of the neck (**Figure 2**). The left side of tongue was larger than right (**Figure 3**). There was a large port-wine stain on left forearm and hand, and the left limb and hand was significantly larger in size and girth as compared to right side (**Figure 4**). The legs were however, symmetrically enlarged. He had huge feet with



Figure 6 Large port-wine stains, starting from thighs down to the both feet.

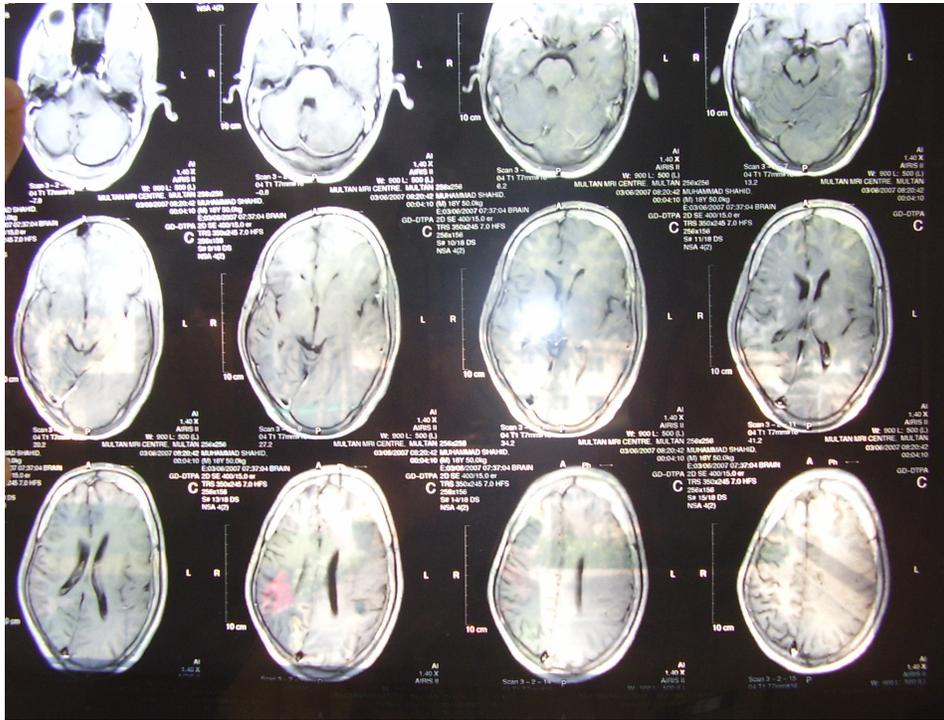


Figure 7 CT Scan brain- 211 hemi-cerebral hypertrophy and shift of mid-line structures to right.

relative overgrowth of second and third toes of both feet (**Figure 5**). There were large port-wine stains, starting from thighs down to the feet (**Figure 6**).

A skin biopsy was performed on one of the lesions on face suspected to be lipoma and histopathology confirmed the diagnosis. Cat scan of brain revealed hemi-cerebral hypertrophy pushing the mid-line structures to right (**Figure 7**).

The family was counseled in detail about the disease and explained that little can be done as far as the physical abnormalities are concerned. They were advised to observe any potential complication like deep venous thrombosis, pulmonary embolism or gastrointestinal bleeding. He was advised carbamazepine 200 mg twice daily for fits.

Discussion

Proteus syndrome is characterized by asymmetrical overgrowth of any part of the

body, verrucous epidermal naevus, vascular malformations and lipoma-like subcutaneous hamartomas. It affects males more than the females. The disorder reflects a mosaicism for a genetic mutation and regarded as a complex development abnormality. There is no specific histopathological feature. The individual lesions have their own characteristic histological features e.g. lipomas, verrucous epidermal naevus and collagenoma.

There are general and specific criteria to diagnose a patient as PS. The general criteria include (1) mosaic distribution of the lesions, (2) progressive course and (3) sporadic occurrence. Specific criteria are (1) epidermal nevus, (2) disproportionate overgrowth of limbs and (3) vascular malformations.²

The most striking feature of the syndrome is overgrowth of any body part. There may be asymmetrical hypertrophy of face, part or whole of one or both limbs, the trunk or any combination of these. Macrodactyly has been regarded as characteristic but is not absolutely

essential. Three main types of skin lesions are described. These are (1) epidermal naevi, (2) vascular malformation and (3) soft subcutaneous lipoma-like masses. Among 24 patients of PS who fulfilled the criteria 22 (92%) had lipomas, 21 (88%) had vascular malformations, 20 (83%) had cerebriform connective tissue nevi on the soles of the feet, 16 (67%) had epidermal nevi and 9 (38%) had partial lipohypoplasia. Some patients had localized alterations in skin pigmentation and hair or nail growth. Patients with a greater number of skin abnormalities tended to have a greater number of extracutaneous abnormalities. The number of abnormalities tended to increase with age up to 8 years.³

As far as vascular malformation is concerned this syndrome is associated with slow-flow vascular anomaly characterized by a triad of a port-wine stain, anomalous veins and overgrowth of the limb.⁴ The overgrowth in PS is progressive and can be difficult to manage.¹ The progressive overgrowth of limbs results in severe orthopaedic complications like scoliosis, megaspondyly, irregular and disorganized bones, hyperostoses, hyperproliferation of osteoid with variable calcification, calcified connective tissue, and elongation of long bones with abnormal thinning.⁵ The common complications in patients with PS are deep venous thrombosis and pulmonary embolism, which can cause premature death.¹ Others include cystic lung disease, central nervous system abnormalities (hemimegacephaly and migrational disorders), various neoplasms, otolaryngologic complications, pulmonary cystic

malformations, and dental and ophthalmologic complications.⁵

The aim of treatment is to minimize the disability of the patient. Substantial contributions can be made by orthopaedic surgeons, ophthalmologists and physiotherapists. It is possible to stop bone overgrowth in childhood by destruction of growth plate. Lipomatous swellings can sometimes be reduced by liposuction.

In the end it may be summarized that; (1) PS should be diagnosed only if the patients fulfill the diagnostic criteria, (2) it is useful for clinical care and research; (3) individuals with PS were more likely to have serious complications which should be closely monitored.⁵

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