PROTEUS SYNDROME: A CASE REPORT

HEYDARIAN FARHAD MD[1] ,
ASHRAFZADEH FARAH MD[2],
TAHERI HERAVI MAHMOUD MD[3]

1.Associate Professor of Pediatrics, Mashhad University of Medical Sciences, Mashhad, Iran
2. Pediatric Neurologist, Professor of pediatrics, Mashhad University of Medical Sciences, Mashhad, Iran
3. Associate Professor of Radiology, Mashhad University of Medical Sciences,Mashhad, Iran

Corresponding Author:
Heydarian F. MD
Pediatric ward,Ghaem hospital, Mashhad, Iran
Cell Phone: +98 915 5155126
Tel: +98 511 8012469
Email:heydarianf@mums.ac.ir

Received: 10-Jan-2009
Last Revised: 10-March-2009
Accepted: 24-Apr-2010

Abstract

Objective

Proteus Syndrome is an extremely rare, sporadic and progressive disorder. We describe a four-month-old male baby with central nervous system manifestations in this article.

Clinical presentation

A four-month-old boy was admitted into our hospital with three tonic - clonic generalized seizure attacks which started from the day before admission. Each seizure attack lasted less than 10 minutes and the baby was well between attacks. No fever was detected. On physical examination, abnormal facial features, macrocrania, a wide nasal bridge, overgrowth of the right lower limb, macrodactyly in the third left toe, epidermal nevus on the left side of the abdomen and skin vascular abnormalities were detected. His developmental status was normal. Brain Magnetic Resonance Imaging (MRI) revealed left sided hemihypertrophy, ventricle enlargement and macrocrania on the same side.

Keywords: hemihypertrophy, macrocrania, vascular abnormalities, seizure, hemimegalencephaly

Introduction

Proteus Syndrome is a very rare congenital syndrome and the reported number of cases does not reach 100 (1, 2).

Many various clinical features are described. The most important ones include asymmetric and disproportionate patchy overgrowth of different tissues especially lower limbs and digits, epidermal nevus, connective tissue nevi, vascular abnormalities and abnormal facial characters (1,2,3,4).

In some cases, central nervous system (CNS) manifestations are reported including hemimegalencephaly and the resultant hemicranial hyperplasia, convulsions and mental deficiency (5,6).

We hereby present an infant Proteus Syndrome case with CNS manifestations.

Case report

This four-month-old male was brought to our hospital for tonic –clonic generalized seizure attacks from the day before admission. He suffered from 3 brief seizure episodes lasting less than 10 minutes each time. He was well between attacks with no fever. His feeding status was normal. He was born to a 24-year-old primigravida mother with cesarean section.

His parents were not relatives. At birth, his weight was 3700 grams, his head circumference was 40 cm and his height was 50 cm. On physical examination, his weight was 7kg, his head circumference was 45 cm and his height was 65 cm. His vital signs including pulse rate, respiratory rate and blood pressure were normal and he had no fever. His developmental status was normal. He had an abnormal morphologic feature including an asymmetric large head, overgrowth of the left frontal side and a wide nasal bridge. Vascular lesions were seen just below his nose (Figure 1).

Patchy hyperpigmentations were seen on face and trunk. (Figure 1) There was a 0.5 × 0.5 cm pigmented raised nevus on his left side of the abdomen just under the ribs. An overgrowth was seen in the right lower limb. Macrodactyly was detected in the third left toe. (Figure 2) A vascular lesion was also seen on the dorsal aspect of the left foot (Figure 2). Other physical findings including heart and lungs examinations were normal. He had no organomegaly. Genital examination revealed a normal male appearance. Laboratory tests including Complete Blood Count (CBC), serum Blood Urea Nitrogen (BUN) and electrolytes were within normal range. Sonography of the abdomen (spleen, liver and urinary tract) was normal. Electroencephalography (EEG) revealed abnormal epileptic discharges. Brain MRI showed left sided hemihypertrophy, ventricle enlargement and macro crania on the same side (Figure 3).

Discussion

Proteus Syndrome is a very rare sporadic syndrome with an unknown etiology. Only less than 100 cases have fulfilled the diagnostic criteria so far (1,5,7,8,9). Main clinical presentations include asymmetrical and disproportionate overgrowth of various tissues such as limbs, especially the lower limbs, skull and viscera, cerebriform connective tissue nevi, epidermal nevi, vascular malformations and facial dysmorphia (6,10).

No definite mutation of special genes has been detected. Overgrowth is patchy in this syndrome (1, 4) which may be seen early after birth (5).

In our case, there was overgrowth of the right lower extremity and obvious macrodactyly in the third toe of his left foot. Also, there was overgrowth of the second toe of the right foot.

Vascular abnormalities may be seen in these patients. These lesions gradually increase in size and never disappear (7, 11). Our patient had these developmental vascular malformations in different parts of his body including the dorsum of the left foot and below his nose.

Central nervous system manifestations occur in about 40% of the patients (5). They include convulsions, hydrocephaly, mental deficiency, hemimegalencephaly and cortical thickening. Our patient suffered from some of these CNS problems including seizure attacks that brought the patient to the hospital and hemimegalencephaly and macrocrania.

Some patients have overgrowth of abdominal organs including spleen and liver. However, the size of spleen and liver were within normal limits in our patient. Since it is a progressive disorder, organomegaly may appear later (1).

Epidermal black or brown nevi, which may be seen in flanks, abdomen, limbs and neck, are other important findings that may be detectable soon after birth (10,12). They were seen in our case in the left upper abdomen.

Other manifestations include fat tissue hyperplasia or tumors, ovarian or testicular tumors, meningioma and parotid adenoma. These, also, may appear later in life.

Some disorders should be considered in differential diagnosis; Klippel - trenaunay syndrome and neureofibromatosis are among them. In Klippel - Trenaunay, vascular malformations are seen in the hemihypertrophic limb and venous varicosities in the same side.

Patients with Proteus Syndrome may have early death resulting from pneumonia or lung embolism.

In conclusion, it should be kept in mind that seizure attacks along with macrocrania and hemimegalencephaly in children may be components of CNS abnormality in Proteus Syndrome.

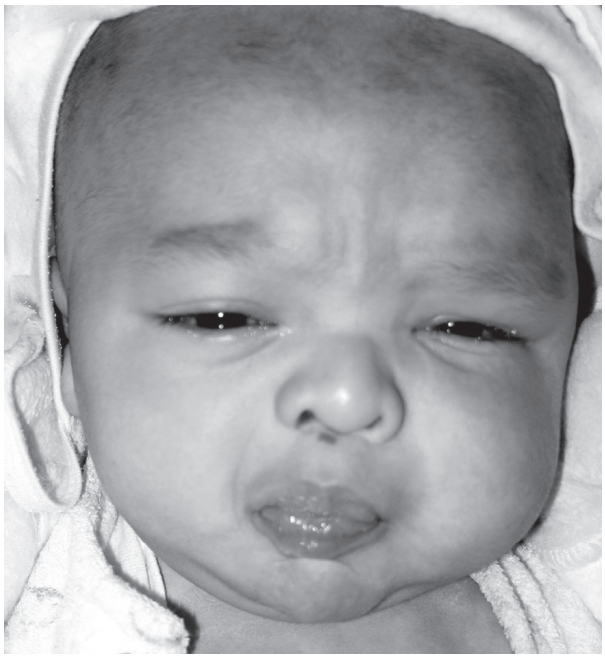

Fig 1. Abnormal facial morphology and hyperpigmented lesions.

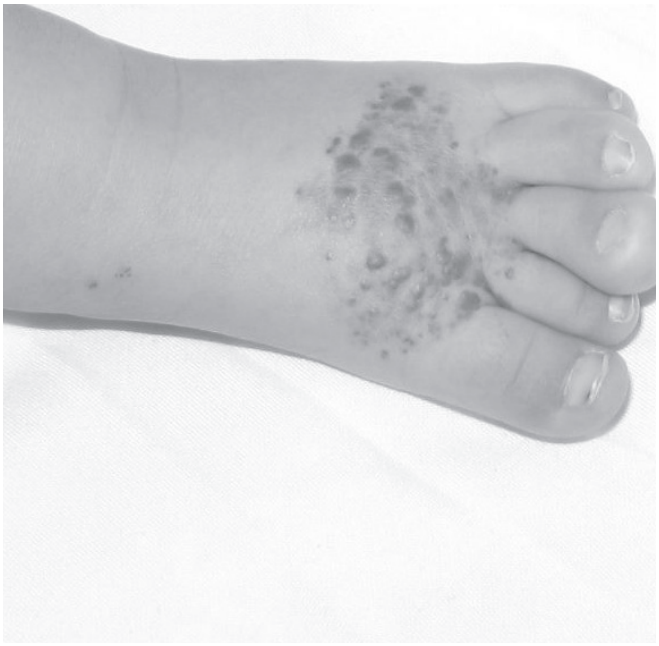

Fig 2. Macrodactyly and vascular lesions in left foot.

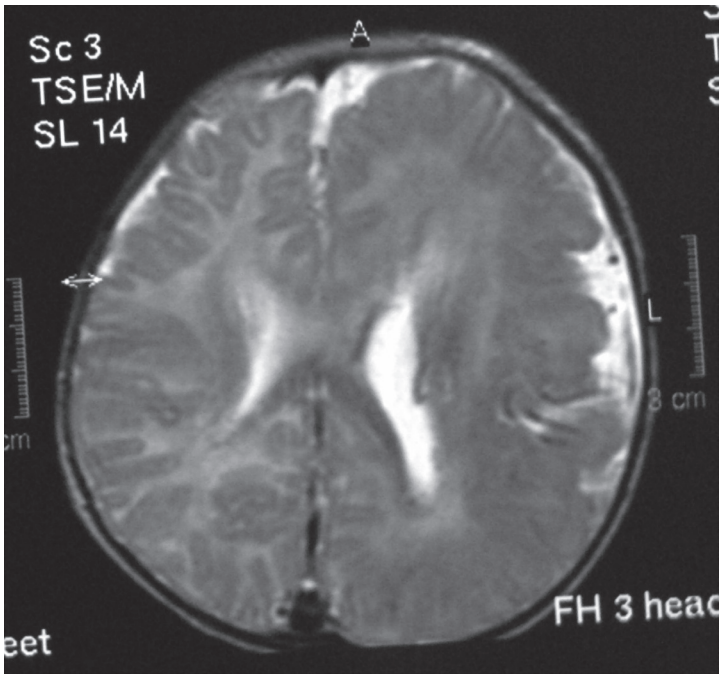

Fig 3. Left cerebral hemisphere hypertrophy and left ventricular enlargement

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