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 7 kg, 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).

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 neurofibromatosis 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.
PROTEUS SYNDROME: A CASE REPORT

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

References


