Case Report

Proteus Syndrome with Arteriovenous Malformation

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Abstract

Proteus syndrome is a rare sporadic disorder that appears with localized macrosomia, congenital lipomatosis, and slow flow vascular malformations, connective tissue nevus, and epidermal nevus. There are usually some manifestations at birth. The vascular abnormalities that have been reported in Proteus syndrome are capillary and slow flow venous malformation. We report a case of a 10-year-old boy with confirmed Proteus syndrome characterized by high flow vascular malformation (arteriovenous [AV] malformation) unlike the usual vascular malformations seen in this syndrome. This case adds a new perspective to the established clinical findings of the Proteus syndrome.

Keywords: *Capillary malformation, Proteus syndrome, vascular malformation*

Introduction

Proteus syndrome, a rare sporadic disorder as asymmetrical overgrowth of body parts, was first described in 1983.[1] It has been called a hamartoneoplastic disease and presented with exostosis, cranial hyperostosis, verrucous epidermal nevus, vascular malformations, and lipoma. The cardinal features are enlarged hands and feet, raised pigmented nevus, hemihypertrophy, subcutaneous masses, skull abnormalities and visceral abnormalities, and accelerated growth. The most characteristic signs are cerebriform overgrowth of the soft tissues of the palms or soles. The vascular lesions may be extensive capillary malformations or more complex combined capillary-venous-lymphatic malformations.[1-7] Since Proteus syndrome is characterized by a constellation of signs and symptoms, for the clinical diagnosis, general and specific criteria have been proposed [Table 1].[8,9] Few reports are available in the literature for the presentation of arteriovenous (AV) malformation in this syndrome. We report a case of Proteus syndrome in a patient presented with AV malformation.

Case Report

We report a case of a 10-year-old boy who was referred to Alzahra hospital, supervised by Isfahan University of Medical Sciences in July 2010 due to vesiculobulous lesions [Figure 1]. This patient was born as a full term neonate through vaginal delivery without any perinatal problems. On the first pediatric visit, there was no significant abnormality. Except an episode of hospitalization due to gastroenteritis and repair of inguinal herniation he was otherwise healthy until the 14th month after birth. At this time, his parents found the evolution of vesiculobollous lesions on his right buttock. The hitopathologic examination of the lesion following surgical resection was compatible with hemangiolymghanioma. However, the lesions had recurred on his right buttock and right thigh as hemorrhagic vesiculobollous lesions when he was presented to our dermatology clinic. The lesions tore easily on palpation and the scar formation was seen in his buttock. The biopsy from the lesions demonstrated lymphangioma circumscriptum. On physical examination, two other giant purple palpable plaques in the left hemi trunk and the ipsilateral flank [Figure 2] of which magnetic resonance imaging (MRI) showed AV malformation that were discovered. Apart from these vascular lesions, macrodactyly of the third toe on both feet [Figure 3a and b] and kyphoscoliosis was obvious. On detailed inquiry, it was revealed that at 4 years of age, the third toes had been growing. This local gigantism caused problems in his wearing shoes and walking when he was 8 years old.

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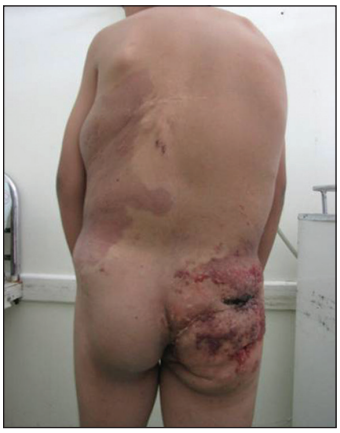

Figure 1: Vesiculobulous lesions in right buttock

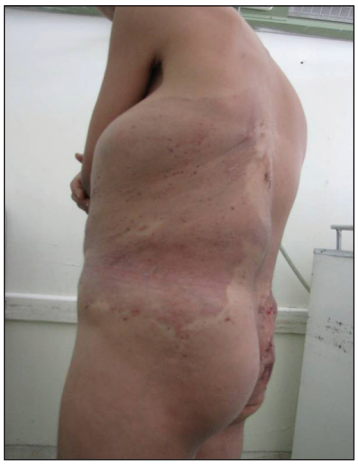

Figure 2: Ateriovenous malformation in left flank

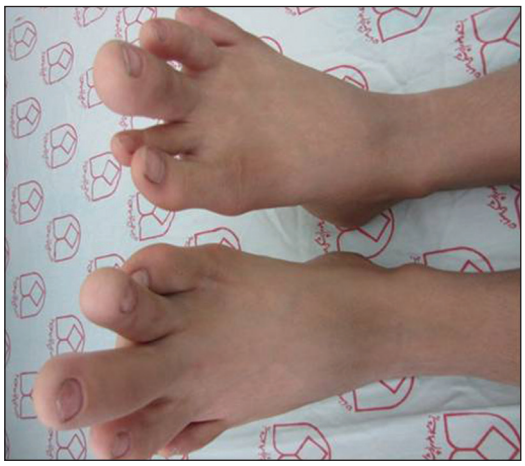

Figure 3a: Big toe

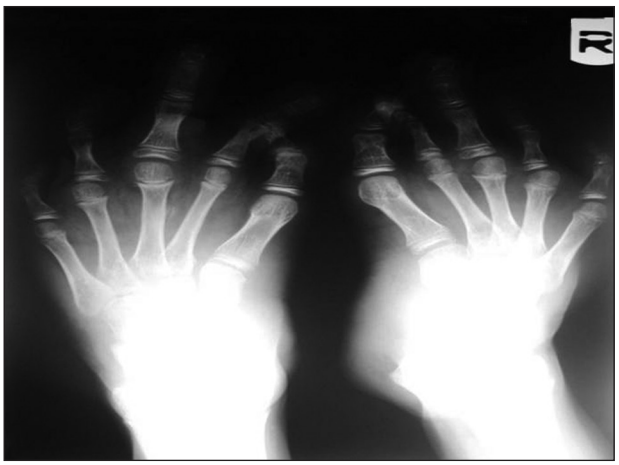

Figure 3b: Its radiographic picture

Table 1: Criteria for diagnosis of Proteus syndrome

General criterion
Mosaic distribution
Progressive course and
Sporadic occurrence
Specific criterion*
Connective tissue nevus or
Two or more of the following criteria: Epidermal nevus, either bilateral ovarian cystadenoma or parotid adenoma, disproportionate overgrowth, lipoma, capillary, venous or lymphatic malformation, and lung cysts

*If connective tissue nevus is not presented, diagnose is made by presentation of three mandatory general criteria

Discussion

Proteus syndrome is a rare sporadic disease characterized by asymmetrical overgrowth of any part of the body, exostosis, cranial hyperostosis, verrucous epidermal nevi, vascular malformations, and lipoma-like hamartomas. The vascular lesions are capillary malformations or combined capillary-venous-lymphatic malformations.[1,2] There is no definite genetic inheritance, although tumor suppressor gene phosphatase and tensin homolog (PTEN) tumor suppressor gene abnormalities have been described in cases of the syndrome.[10] The somatic motation of AKT1 was detected in 90% of patients. There are usually some manifestations at birth.[1] In our case, the patient had disproportionate overgrowth of limbs, skeletal abnormalities (feet), and vascular malformations (AV malformation and lymphangioma). Taken together, our case fulfilled the general criterion of harboring Proteus syndrome. The vascular abnormalities that have been reported in Proteus syndrome are capillary and slow flow venous malformation; however, no AV malformation was observed in our patient. Our case had had inguinal herniation in infancy not reported in Proteus syndrome. By comparison, exostosis, cranial hyperstosis, verrucous epidermal nevus, connective tissue nevus, lipoma, visceral, dental, and eye abnormalities were not seen in our patient. Due to the fact that Proteus syndrome is a sporadic disease in the category of mosaicsism disorders, attempts at discovering other possible mutations or making bridges between clinical findings and known gene mutations (PTEN) continue. In other words, this case adds a new perspective to the established clinical findings of Proteus syndrome. Due to the varying features of this syndrome, a multidisciplinary approach by consultation with a vascular surgeon and an orthopedist was requested.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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