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Proteus Syndrome

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ABSTRACT. Proteus Syndrome (PS) is a rare hamartomatous syndrome with a variety of abnormalities. We report a case of a 14 year-old male with PS affected with bilateral glaucoma. This is the second case to be reported in Saudi Arabia.

Keywords: Proteus syndrome.

Introduction

Proteus Syndrome (PS) is a hamartomatous disorder. It is characterized by multifocal overgrowth of many tissues including epidermis, connective tissue, endothelium, adipose tissue and bone. This is reflected on the features seen in the patient as hemihypertrophy, macrodactyly, skin thickening of the palms and soles, lipomas, verrucous epidermal nevus, skull and limb bone abnormalities, seizures and ophthalmologic abnormality.

The name "Proteus Syndrome" was suggested by Wiedemann in 1983[1]. He derived the name from the Greek god "Proteus" which means polymorph, for his ability to change his shape at will to avoid capture. This is to emphasize the wide range of clinical manifestations seen in this syndrome. The cause of this syndrome is unknown. It is regarded as a complex developmental abnormality. Happle suggested that a dominant lethal gene that survives by mosaicism as a result of an early somatic mutation causes this syndrome[2].

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Case Report

A 14 year-old male was seen in our clinic. He has asymmetrical hemihypertrophy of the right side of his body, a port-wine stain nevus on his arm, multiple subcutaneous soft lipoma masses on the back of the right thigh and gynecomastia (Figs. 1 and 2). Thickening of the palms and soles were noted. The patient had limping gait where the X-ray showed hypertrophy of the bones on the right side. Ophthalmologic examination revealed the presence of bilateral glaucoma.

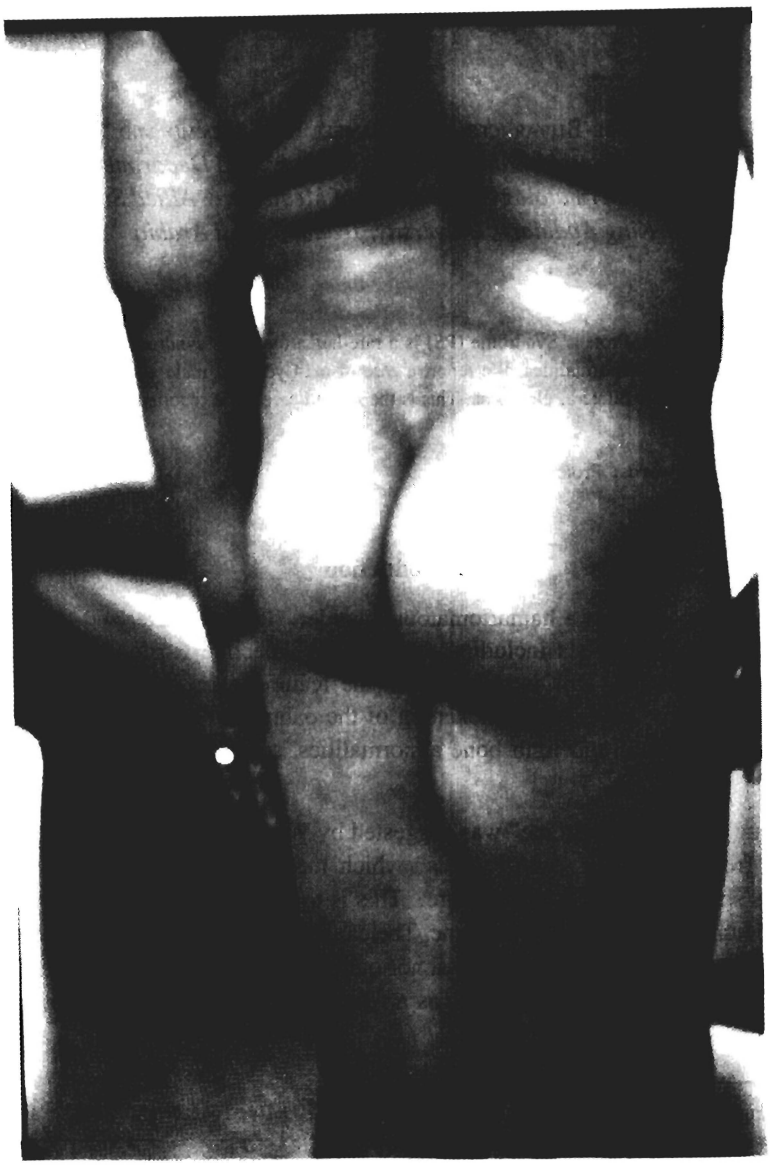

FIG. 1. Asymmetrical hemihypertrophy of right side of body, Lipoma of right thigh.

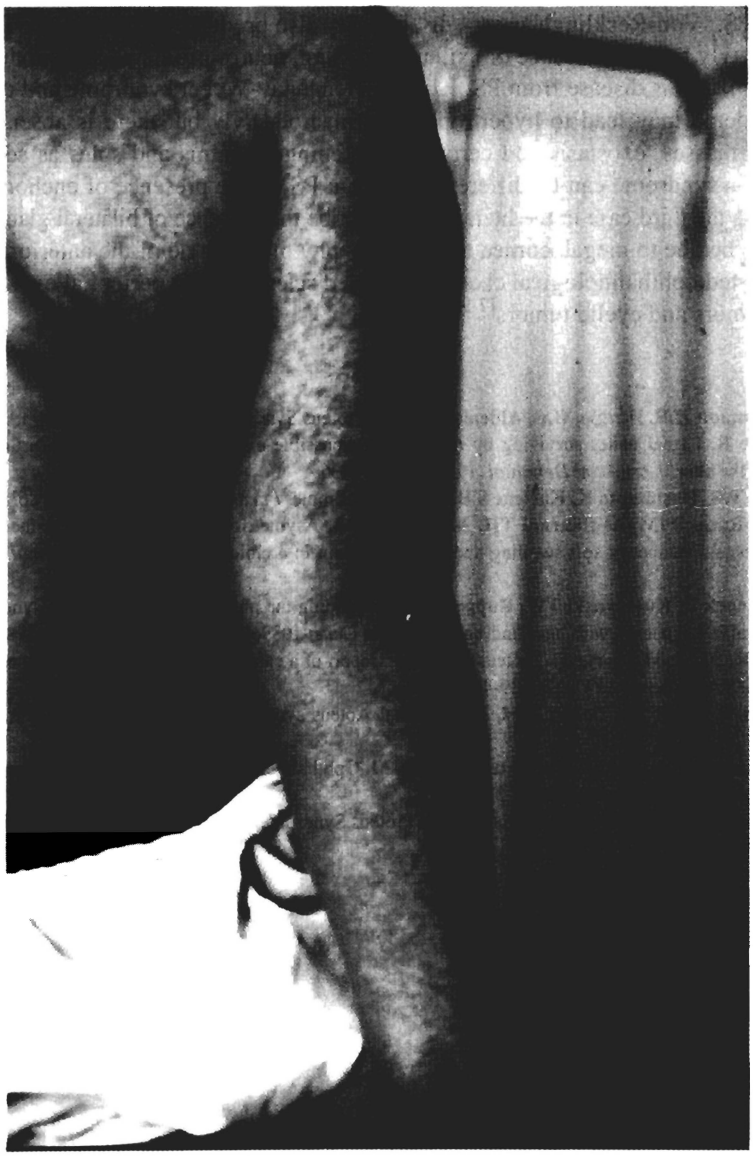

FIG. 2. Port wine stain nevus on arm.

Discussion

PS is rare and fewer than 70 cases have been described in the literature[3]. This is the second case seen in Saudi Arabia[4]. Because of the wide range of manifestations, it is difficult to differentiate this condition from a variety of other conditions. The lipome-like masses seen in the present case can be seen in Banayan Zonana Syndrome but the lack of intracranial tumors, cavernous hemangioma and macrocephaly help to

exclude PS. Von-Recklinghausen's disease may also produce asymmetric body growth, macrodactyly but the presence of axillary freckling, neurofibroma and iris nodules helps to distinguish this disease from PS. Klippel-Trenaunay-Weber syndrome and congenital lymphoedema may lead to hypertrophy and macrodactyly but there is absence of lipoma-like masses, exostasis and cerbroid thickening of palms and soles as seen in PS. Maffucci's syndrome can be differentiated from PS by the presence of enchondromatosis. This is the third case in the literature to describe the presence of bilateral glaucoma[5,6]. This may be due to megalocornea and hypertrophy of the angle of the anterior chamber. The reported ophthalmological changes included scleral tumor, periobital exostosis, epibulbar tumors and eyelid tumors[7-9].

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