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Radiographic findings of Proteus Syndrome

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The extremely rare Proteus Syndrome is a hamartomatous congenital syndrome with substantial variability between clinical patient presentations. The diagnostic criteria consist of a multitude of clinical findings including hemihypertrophy, macrodactyly, epidermal nevi, subcutaneous hamartomatous tumors, and bony abnormalities. These clinical findings correlate with striking radiographic findings.

Introduction

This case report radiographically presents many of the findings associated with the rare Proteus Syndrome, also known as Wiedemann syndrome. The Proteus Syndrome was first described in four patients in 1983 by Wiedeman (1). It is thought to affect roughly 1 out of a million, to 1 out of 10 million (2). The diagnosis is difficult due to the high variability of clinical presentations. General diagnostic criteria suggested as mandatory for diagnosis are mosaic distribution of lesions, progressive course, and sporadic occurrence. More specific clinical manifestations have also been suggested, including distorting, progressive overgrowth of the skeleton; cerebriform connective tissue nevi, which have been considered pathognomonic for the syndrome; linear verrucous epidermal nevus; adipose dysregulation; and several other cutaneous malformations (3, 4). We review several of the radiographic findings of the Proteus Syndrome in a 26-year-old Guatemalan male who presented with chronic foot and abdominal pain.

Case report

A 26-year-old male presented to Harbor-UCLA General Hospital with a chief complaint of chronic abdominal and foot pain. The patient had no previous diagnosis for his congenital deformities. He was referred to multiple subspecialties for further evaluation of enlarging feet since age 1, multiple congenital skin lesions, and chronic chest wall and abdominal pain.

Upon evaluation by the Dermatology service, the patient was found to have diffuse congenital epidermal nevi throughout the body and multiple malformations, including a left-flank lymphangioma circumscriptum and a right-flank dermatofibroma. There were also superficial hyperpigmented lesions overlying the anterior chest wall masses, seen on CT.

The patient had a history of congenital deformities including cryptorchism, for which he had an orchiopexy in 2002. The only laboratory abnormality found was low testosterone, thought to be secondary to cryptorchism. After evaluation by the GI-Oncology service, the chest wall

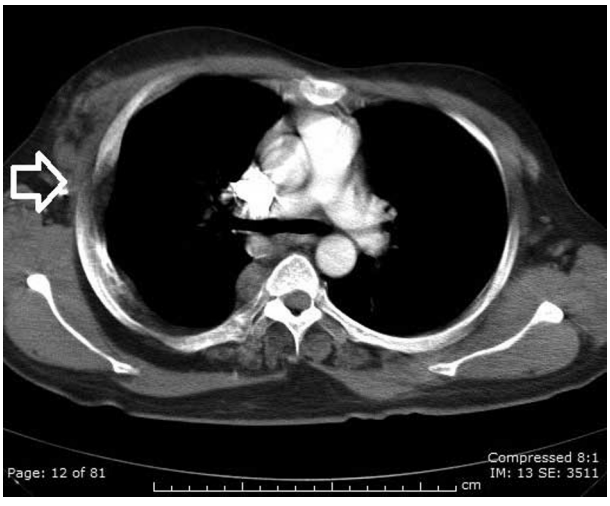

Figure 1. Proteus Syndrome in a 26-year-old male. Axial CT (All images were created with Siemens SOMATOM Sensation 64 Slice, mA 200, KVp 120). Note hemihypertrophy of the right chest, and a soft-tissue mass under the right crus (white open arrow).

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masses were followed radiographically, as they were thought to be part of the patient's broader syndrome. The patient claimed normal pubertal onset, and a negative family history.

Based on complete radiographic evaluation and physical exam findings, the diagnosis of the Proteus Syndrome was made.

Discussion

The rare Proteus Syndrome is difficult to diagnose due to its multisystem involvement and broad variability in both clinical presentation and radiographic findings. The radiographic findings can also be easily confused with neoplasm. Proteus Syndrome has been associated with mosaicism for a somatic activating mutation in the AKT1 gene on chromosome 14q32.3 (5). Genetic testing may be performed to make the diagnosis (2). The differential diagnosis can include Klippel-Trenaunay-Weber syndrome, Ollier disease, and Maffucci syndrome (4, 6).

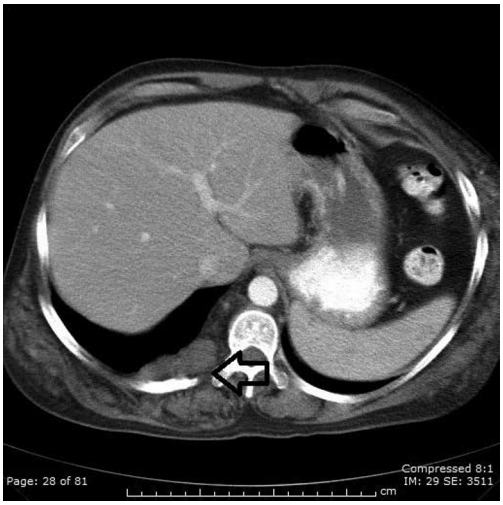

Figure 2. Proteus Syndrome in a 26-year-old male. Axial CT. An 8 x 2cm soft-tissue mass appears posterior to the right crus (black open arrow). Also note diffuse soft-tissue infiltration of the right flank musculature, thought to be secondary to subcutaneous nevi.

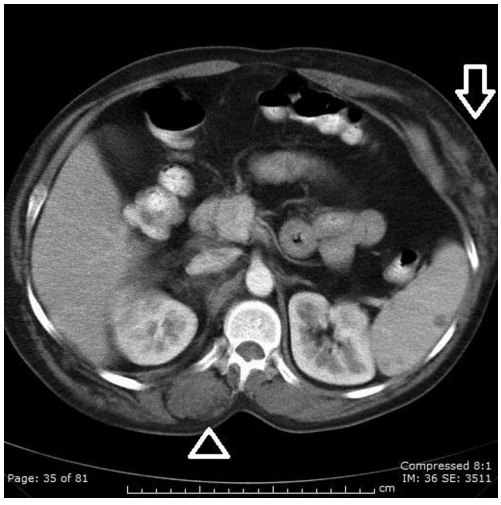

Figure 3. Proteus Syndrome in a 26-year-old male. Axial CT. The asymmetry of the paraspinous muscles is thought to be due to deposition within the muscles (open arrowhead). Note soft-tissue infiltration of the left anterior chest wall (white open arrow), as well as nonspecific, low-density lesions in the spleen.

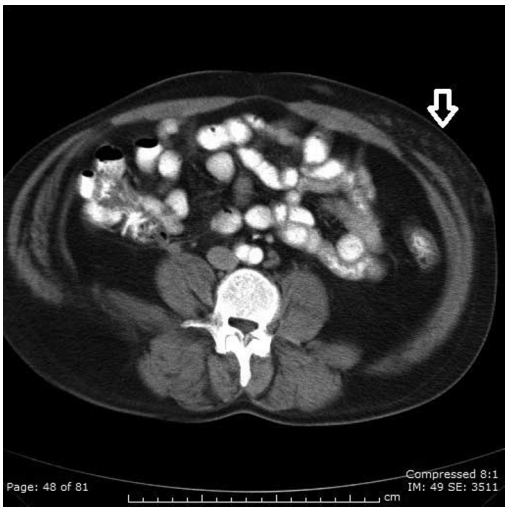

Figure 4. Proteus Syndrome in a 26-year-old male. Axial CT. Note soft-tissue infiltration of the left anterior abdominal wall fat (white open arrow).

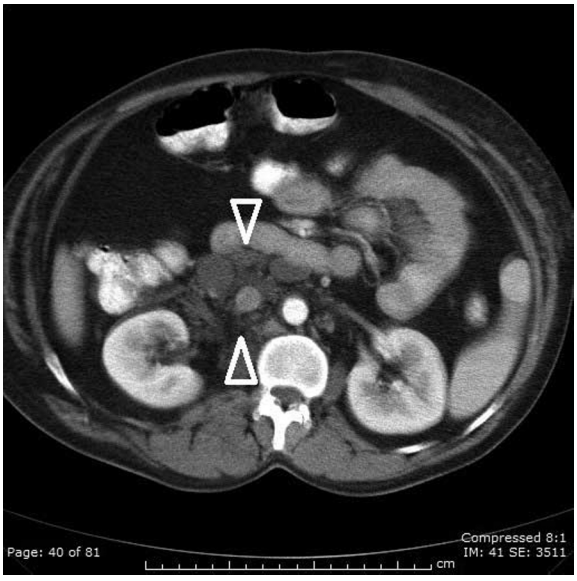

Figure 5. Proteus Syndrome in a 26-year-old male. Axial CT. Note striking para-aortic lymphadenopathy versus conglomerate soft-tissue mass (white open arrowheads).

Named after the Greek god Proteus—"the polymorphous"—the disorder combines a multitude of highly variable congenital features that include partial gigantism of