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An unusual initial presentation of Proteus syndrome: macrosyndactyly

Proteus sendromunun nadir görülen formu: Makrosindaktili

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Proteus syndrome is a complex disorder characterized by a wide variety of deformities including macrodactyly. In the present report, we present a case with complex macrosyndactyly in his hand. The patient was surgically treated. However, he admitted with lymphangiomas on his body during the follow-up period, leading to the diagnosis of Proteus syndrome. This article discusses the differential diagnosis of Proteus syndrome and the treatment methods used for macrodactyly along with the presentation of the case.

Key words: Hand; lymphangioma; macrodactyly; Proteus syndrome; syndactyly.

Proteus sendromu makrodaktilinin de içinde bulunduğu çok çeşitli deformitelerle karakterize kompleks bir hastalıktır. Bu yazıda elinde kompleks makrosindaktili bulunan bir olgu sunuldu. Hasta cerrahi olarak tedavi edildi. Ancak daha sonraki takip döneminde vücudundaki lenfanjiyomlar ile başvurdu ve bu bulgu ile Proteus sendromu tanısı konuldu. Bu yazıda, olgu sunumuyla birlikte Proteus sendromunun ayırıcı tanısı ve makrosindaktilide uygulanan tedavi yöntemleri tartışıldı.

Anahtar sözcükler: El; lenfanjiyom; makrodaktili; Proteus sendromu; sindaktili.

Proteus syndrome is a rare congenital hamartomatous disorder manifesting with gigantism of hands and feet, lymphangiomatous hamartomas and other varying features which were first described by Wiedemann.[1]

The reports on macrodactyly in association with Proteus syndrome and its treatment are quite rare.[2,3] Our literature search did not reveal any other case of Proteus syndrome initially presenting only with isolated macrosyndactyly and receiving surgical treatment for its correction.

This report presents a case of Proteus syndrome initially manifesting with macrosyndactyly, and discusses its differential diagnosis and treatment.

CASE REPORT

A four-month-old male infant was admitted to our institution with a deformity in his left hand which was recognized at birth. The patient was the first child of non-consanguinally related parents. There was no family history of a similar condition.

Physical examination was unremarkable except for macrosyndactyly between the 3rd and 4th digits of the left hand (Figure 1). In addition to X-rays of the thorax, cranium and whole upper and lower extremities, magnetic resonance images (MRI) of the central nervous system (brain, medulla spinalis) and the left hand were also obtained. Magnetic resonance images of the central nervous system were normal. Magnetic resonance images of the left hand only showed a diffuse fat tissue increase with normal digital nerves. Chromosomal examination did not reveal any abnormality. Based on these findings, the patient was diagnosed with isolated macrosyndactyly and follow-up was scheduled.

The patient's 3rd and 4th digits with macrosyndactyly had complete active and passive range of motion (ROM) only at the metacarpophalangeal joint.

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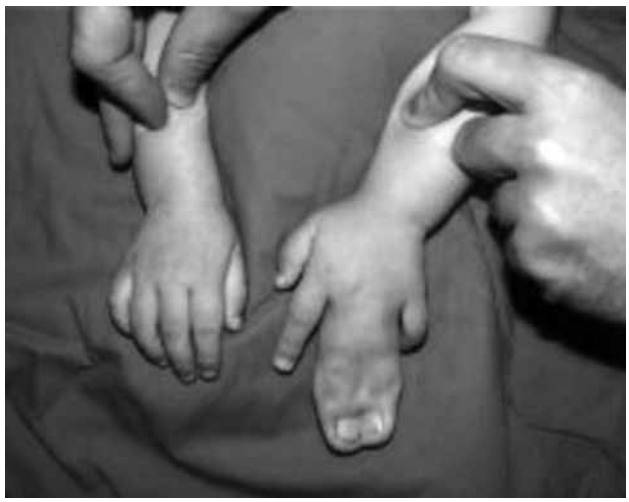

Figure 1. Appearance of both hands at the time of his initial admission at an age of four months.

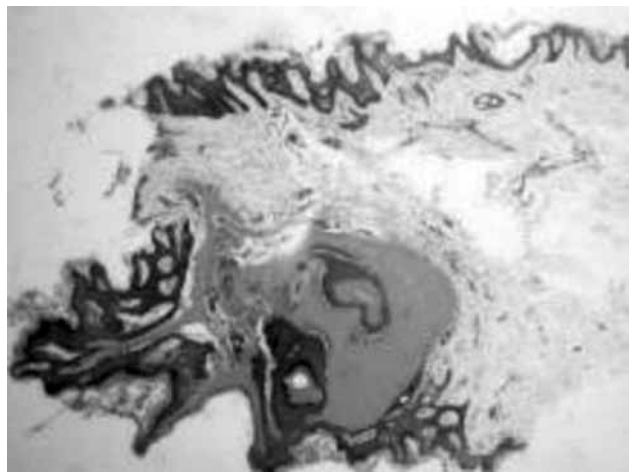

Figure 2. Microscopic appearance of lymphangioma. Located beneath the epidermis, enlarged lymphatics are seen with lymphatic fluid in their lumen (H-E x 40).

Proximal interphalangeal (PIP) and distal interphalangeal (DIP) joints did not have either active or passive motion. Longitudinal growth was monitored without any intervention on the digits with macrodactyly. During one-year follow-up, the digits with macrosyndactyly developed far more rapidly compared the corresponding digits of the right side. When the child was 16 months old, the distance between the metacarpophalangeal joint and pulp for his third and fourth digits was similar to the corresponding distance of his father's hand, and epiphysiodesis of the related digits was performed. The syndactyly between the 3rd and 4th digits was separated surgically when the patient was 21 months old. Debulking of the radial and ulnar sides of the digits was performed at the age of three and four. Thereafter, no further surgical operation was performed and the patient was followed.

The patient was readmitted with dermatological lesions on his thigh, perineal and genital regions at the age of six. Dermatological lesions were scattered and vesicular with clear fluid. The patient consulted with the dermatology department and one of the gluteal lesions was biopsied confirming a diagnosis of lymphangioma (Figure 2).

Taking into account these late-onset findings and previous diagnosis of isolated macrosyndactyly that had been surgically treated, the patient was considered as a case of Proteus syndrome.

Currently, the patient is seven-years-old. At his latest visit, no growth was observed following epiphysiodesis. Although the metacarpophalangeal joints of the 3rd and 4th digits had full ROM, the proximal and distal interphalangeal joints of these digits had only 10 to 15 degrees of passive motion with no active motion (Figure 3a, b). The child does not hide and tries to use his hand when he is at home with his family. On the other hand, he hides his hand in his pocket and does not use it on other occasions. Amputation was recommended but the family refused.

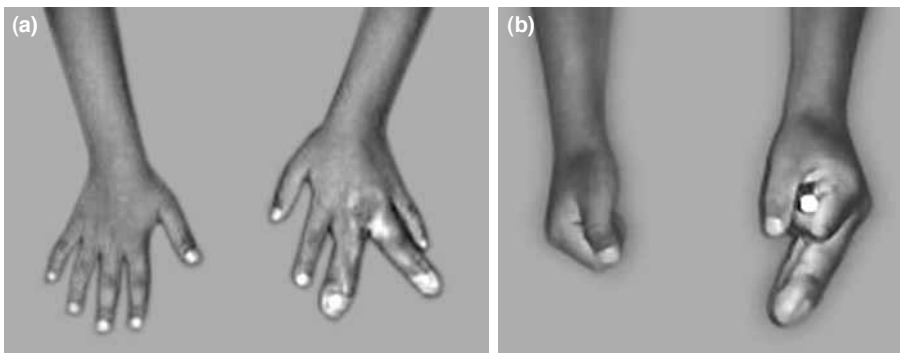

Figure 3. Appearance of both hands at age seven during the last available visit. **(a)** Front view and **(b)** side view.

DISCUSSION

Macrodactyly, if the term is used properly, refers to a rare congenital malformation characterized by an increase in the size of all the elements or stractures of a digit or digits.[4] The differential diagnosis of enlarged digits includes true macrodactyly, hemangioma and other vascular anomalies, tumors, multiple enchondromatosis, neurofibromatosis, Kilippel-Trenaunay-Weber syndrome and Proteus syndrome. All of these conditions have similar clinical manifestations and each should be differentiated from Proteus syndrome.[2]

Proteus syndrome may manifest itself at birth with major clinical findings such as partial gigantism involving hands and feet, pigmented nevi, hemihypertrophy, subcutaneous hamartomatous tumors (lymphangiomas, lipomas, lymphangiolipomas) and visceral abnormalities.[1] Alternatively, some clinical findings absent at birth may manifest later with advancing age.[5] Several large series-reporting patients with Proteus syndrome found dermatological findings in all patients.[6] The non-invasive and multiplanar imaging capabilities of MRI make it the imaging modality of choice in the assessment of patients with macrodactyly or macromelia when the diagnosis is unclear.[7]

Our case had macrosyndactyly at birth. Magnetic resonance images of the brain, medulla, spinalis and the part of left hand with macrodactyly were obtained, however the only finding was a diffuse increase in the amount of fat tissue at the site of macrodactyly. Magnetic resonance images of the body and lower extremities were not obtained, as there was no clinical finding at these sites.

In a previous study, the macrodactyly in Proteus syndrome was not associated with enlarged digital nerves or proliferation of subcutaneous fat.[3] But in another case report, the neurovascular bundles, subcutaneous tissues and the tendons in the hands were all enlarged in proportion to the enormous size of digits.[2] During debulking procedures, we explored the digital nerves under a microscope and did not find any remarkable proliferation or enlargement of these structures.

The management of macrodactyly should be conservative unless functional or cosmetic problems arise.[6] Factors that should be considered in the treatment include the type of macrodactyly, rate of progression and the age of the patient.[8] The most important point in the treatment of macrodactyly is to control the growth of the involved digit during the early stages while allowing the other fingers to grow so as to maintain proportionality of the fingers.[9]

Epiphysiodesis was found to be an effective method for prevention of longitudinal overgrowth of the digit.[8] Several debulking procedures may be required.[10] In the presence of unusually severe macrodactyly and involvement of only one or two digits, amputation is an alternative method to consider in management.[4,9,10]

Proteus syndrome should also be borne in mind in the differential diagnosis of cases with isolated congenital macrosyndactyly of the upper extremities. All signs and symptoms of Proteus syndrome may not be evident at the beginning and may develop later during follow-up. Therefore, macrodactyly patients should be closely followed and other emerging signs and symptoms should be re-evaluated together with relevant clinical disciplines.

Termination of the rapid and disproportionate growth compared to other digits by epiphysiodesis should be the priority of macrodactyly management. Correction of syndactyly and debulking of soft tissues should be considered later on. Although stopping the elongation of digits and the debulking procedure give good cosmetic results, functionality may not be satisfactory. Amputation may be considered, particularly when proximal and distal interphalangeal joints have no motion, in order to provide better functionality; however amputation has the potential to be refused by the patient and relatives.

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