Case Report

Atypical presentation of congenital lymphedema

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ABSTRACT

Lymphedema, lymphatic fluid is due to an abnormal accumulation in the body in a regional body edema. Congenital lymphedema represents all forms that are clinically evident at birth and accounts for 10-25% of all primary lymphedema cases. The patient was born from consanguineous parents as the first child after two abortion and history of a child death at 14 month of age. On examination the patient had pitting edema at all extremities and abdomen. Lymphangiosintigraphy was performed and no lymph nodes were seen so hereditary primary lymphedema diagnosis was confirmed. Primarily, often caused by a congenital anomaly or dysfunction. As a secondary, acquired disorder is caused by lymphatic flow. Congenital lymphedema, occurs in the first weeks of life. After resolution of the edema the patient was malnourished and had feeding problems. In English literature, congenital lymphedema case with organomegaly, ascites and pitting generalized edema is rarely defined. We report this case because of its unique presentation.

KEY WORDS: Hereditary, Lymphedema, Organomegaly.

Pak J Med Sci October - December 2011 Vol. 27 No. 5 1194-1195

How to cite this article:

Silfeler Y, Genens M, Sumengen D, Guler S, Acar Y. Atypical presentation of congenital lymphedema. Pak J Med Sci 2011;27(5):1194-1195

INTRODUCTION

Hereditary primary lymphedema is a rare disorder presenting with persistent edema of lower extremities at birth. It is characterized by the dysfunction of lymphatic vessels due to swelling of the soft tissue secondary to obstruction of lymphatic drainage.[1] Primary lymphedema is divided into 3 groups: Congenital, Praecox and Tarda.[1] The pattern of inheritance is autosomal dominant with variable penetrance and expressivity.[1] The first chromosomal locus of the gene has been lately identified in the long arm of chromosome 5.[2] Congenital lymphedema represents all forms that are clinically evident at birth and accounts for 10-25% of all primary lymphedema cases. Females are affected twice as often as males, and the lower extremity is involved 3 times more frequently than the upper extremity.[3] Two thirds of patients have bilateral lymphedema, and this form may improve spontaneously with increasing age.

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* Received for Publication: May 5, 2011
* Revision Received: September 20, 2011
* Revision Accepted: September 22, 2011

CASE REPORT

The patient was born from consanguineous parents as the first child after two abortion and history of a child death at 14 month of age. At the delivery room generalized edema had been noted. On examination the patient had pitting edema at all extremities and abdomen. Complete blood count

and urinalysis were normal, protein levels were decreased. On the abdominal ultrasound massive ascites and hepatosplenomegaly were present. Paracentesis repeated four times, benign serous effusion and elevated triglycerides were found. On echocardiography patent foramen ovale and enlarged left atrium were found. Lymphangiosintigraphy was performed and no lymph nodes were seen so hereditary primary lymphedema diagnosis was confirmed.

DISCUSSION

Lymphedema, lymphatic fluid, due to an abnormal accumulation in the body is a regional body edema. It is seen in both sexes equally, even if advised in literature it is more common in women in practice.[4] Primarily, it is often caused by a congenital anomaly or dysfunction. Lymphedema, the lymphatic circulation may occur secondary to obstruction. Congenital lymphedema, occurs in the first weeks of life. Congenital lymphedema, may be associated with Noonan's and Turner's syndrome.[5,6] Hereditary primary lymphedema is characterized by swelling of the soft tissue because of the lymphatic obstruction. It is usually associated with edema at the lower extremities.[1,3] The diagnosis is usually made with history and physical examination.

All 3 forms of primary lymphedema likely originate from a developmental abnormality that is present, but not always clinically evident, at birth.[7] Congenital lymphedema is found in patients with edema present at birth or shortly afterwards.[8] Some cases may become evident later in life when a triggering event or worsening of the condition causes the lymphatic transport capacity to be exceeded by the volume of interstitial fluid formation, causing the patient to be unable to maintain normal lymphatic flow. In differential diagnosis, edema secondary to congestive heart failure, renal insufficiency, hepatic insufficiency, or venous stasis disease should be considered.[9]

In our case lymphedema was atypical, involving all body. The patient did not have any other anomalies and the lymphedema resolved in 3 months. In most cases congenital lymphedema affects only upper/lower limb.[1,3,10] It is also different that the patient did not have any other anomalies. After resolution of the edema the patient was malnourished and had feeding anomalies. In english literature, congenital lymphedema case with organomegaly, ascites and pitting generalized edema is rarely defined. We report this case because of its unique presentation.

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Contribution of Authors:

Ibrahim Silfeler, Mikayir Genens and Dilek Sumengen: Literature search and collection of data.
Salih Guler: Design and writing of the manuscript.
Salih Guler and Yesim Acar: Helped the main author in preparing the final manucript.