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Case Report

Hereditary Lymphedema of the Leg – A Case Report

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Abstract

Primary of hereditary lymphedema is a rare but progressive disease. It is yet not curable. We present a 48-year-old male patient with hereditary lymphedema of his left leg, that was realised by minor trauma (able twist) when he was seven years old. He had never been treated for lymphedema but experienced multiple erysipelas during his life. After diagnostic procedures to exclude other causes of leg swelling, the diagnosis of hereditary lymphedema of the leg, stage III was confirmed. We initialized complex decongestive therapy. During two weeks of intensive treatment, the circumference of the left leg could be reduced by 10 cm. This case illustrates the "natural course" hereditary lymphedema. But it raises the hope that even after decades of ignorance, the patients benefits from complex decongestive treatment. Therapeutic nihilism is unnecessary and poses lymphedema patients to risks of infection and secondary malignancies like Stewart-Trewes syndrome.

Introduction

Primary or congenital lymphedema is a rare disorder. It occurs 1 in 100 000 persons affecting in < 90 % of cases the lower limbs [1]. Primary lymphedema may be present at birth or sometimes develop year's later - also called lymphedema tardum. Secondary lymphedema is seen more often after an injury to lymphatic vessels and lymph nodes – a common problem in certain cancer treatment protocols like in breast or endometrial cancer [2, 3].

Primary lymphedema is a progressive, not-curable disease. Subcutaneous enlargement of fibro-adipous tissue leads to progressive limb swelling, loss of functionality, mobility and quality of life. Lymphedematous skin is an immunocompromised district predisposing to infection and tumours [4].

Case report

A 48-year-old male patient presented with a swelling of his left leg. He had a medical history of multiple erysipelas of the same leg and an ankle twist when he was 7-year-old. Despite antibiosis, he had never had diagnostics or treatments before.

On examination, we observed men without any signs of cardiopulmonary decompensation. He had neither inguinal lymphadenopathy nor a total involvement of the oedema. There were no sensorimotoric deficiencies.

There was a collum-like pasty oedema of his left leg, pressable on the upper leg but impressable on the lower leg. The distal lower leg was further characterised by fibrosis, pachydermia, and hyperkeratosis left leg. There was a dewlap-like disfiguration of soft tissue in the ankle region (Fig. 1a). Foot and toes were edematous with a positive Stemmer sign. The interdigital skin was macerated.

Routine laboratory was unremarkable.

Imaging: Color Duplex sonography demonstrated normal venous and arterial blood flow without any signs of recent or previous thrombosis.

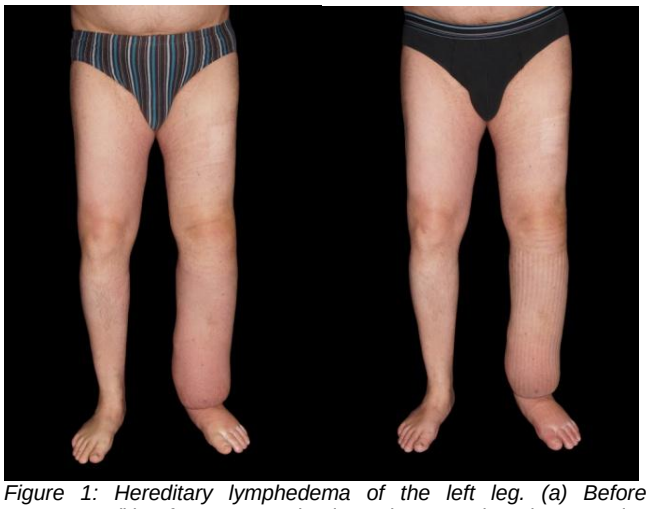

Figure 1: Hereditary lymphedema of the left leg. (a) Before treatment, (b) After two weeks intensive complex decongestive treatment

X-ray of feet disclosed degenerative changes of the proximal joint of digitus I on both feet and edematous soft tissue changes (Fig. 2a, b). Abdominal ultrasound remained unremarkable.

We diagnosed hereditary lymphedema of the left leg, stage III.

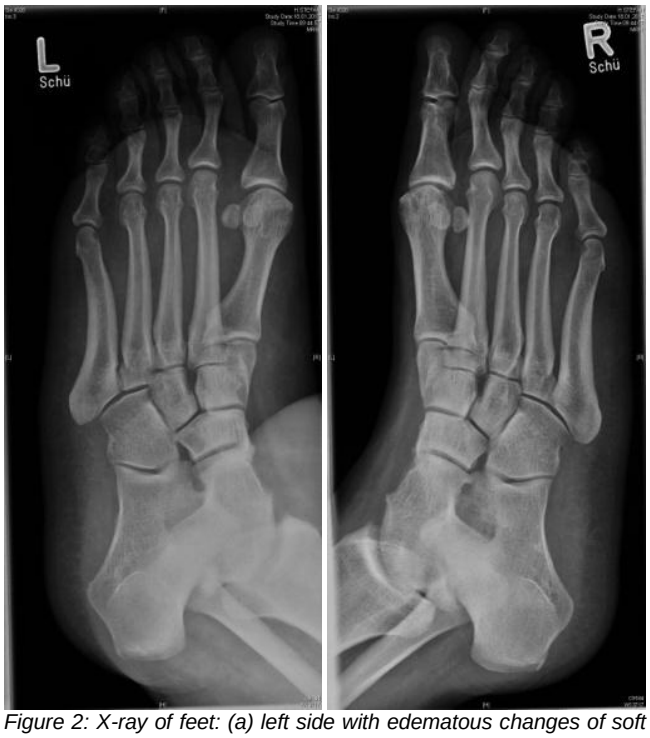

Figure 2: X-ray of feet: (a) left side with edematous changes of soft tissue, (b) right side. Arthrosis of Dig. I proximal joints on both sides

Treatment and course: We started with manual decongestive therapy (manual lymph drainage for the left leg combined with padded short stretch bandages and manual mobilisation of the left ankle. Topical interdigital treatment consisted of nystatin zinc-oil. During 15 days of inpatient complex decongestive therapy, there was a reduction of leg circumferences of 10 cm (Fig. 1b). Outpatient treatment was continued. Bandaging was replaced by flat knitted socks for the upper leg (compression class II) and flat knitted socks for lower leg and foot (compression class III).

Discussion

The patient suffered from an ankle twist at the age of seven. After that a persistent swelling oft he left leg developed. The trauma caused an overload for his lymphatic vessels. Usually, no lymphedema develops, but in the case of a predisposition with congenital malformation of lymphatic vessels, any trauma may realise the genetic disposition. His course was characterised by ignorance of lymphedema and absence of any treatment. This predisposed him to an increased risk of streptococcal soft tissue infections (erysipelas). The possible enters forte for germs is the intertriginous dermatitis of the interdigital space.

In primary (congenital) lymphedema germline mutations of at least 20 different genes have been identified which are enoding for protein interacting with vascular endothelial growth factor (VEGF) receptor-3 or with other tyrosin kinases. These mutations act on RAS/MAPK or PI3/AKT signal transduction [5].

There is yeat no cure for hereditary lymphedema. Complex decongestive therapy is considered the major approach. However, sufficient muscular-joint interactions are important for decongestion as well [6].

Our case report demonstrates that (a) therapeutic nihilism is contra productive and (b) even in longstanding lymphedema complex decongestive therapy is an effective treatment [7]. Surgical treatments include liposuction, lymphodemous anastomosis, vascularized lymph node transfer, and combined/multiple approaches. These methods are limited to a minority of patients [8].

Debulking soft tissue resection can become necessary in case of major fibrosis according to elephantiasis nostras with formation of immature lacune-like lymphatic vessels and massive fibrosis [9]

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