CONGENITAL LYMPHEDEMA WITH LYMPHANGIOMA CIRCUMSCRIPTUM ASSOCIATED WITH

Multiple Congenital Cardiac Defects and Lymphangiectatic Gastro-esophageal Reflux

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***Abstract*:**

Lymphedema is the swelling, usually of an extremity, resulting from poor drainage of fluid out of the body's tissues.

Primary lymphedema can be present from birth and it has been estimated to occur in about one in six thousand people , more often in females than in males[1].

Lymphangioma circumscriptum and lymphangiosarcoma has been reported to occur in cases of chronic lymphedema[2,3].

We report here two patients with clinico-pathologically typical primary lymphedema developing lymphangioma circumcsriptum, one of them associated with multiple congenital cardiac defects and the second one associated with intestinal lymphangiectasia with gastro-esophageal reflux.

To our knowledge, this is the fifth report of primary lymphedema with lymphangioma circumscriptum and associated with multiple congenital cardiac defects .

In our cases Magnetic Resonance Imaging(MRI) was done to determine the possible cause of the lymhedema and the extent of lymphangioma circumscriptum.

The clinical, microscopic, MRI illustration and literature are fully reviewed[4,18].

Introduction:

A normal lymphatic system, consists of blind-end vessels which collect the lymph that bathes and nourish the tissues.

In normal system, lymph is derived from the arterial side of capillaries, and is returned to the circulation via veins near the neck.

The purpose of the lymphatic system is to help the body to maintain fluid balance while filtering out waste products [1].

Aplasia, hypoplasia or hyperplasia of the lymphatic system is thought to cause a primary lymphedema, while secondary lymphedema is the result of a damaged or blocked lymphatic system caused by trauma, infection or injury[2].

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Lymphangiosarcoma is known to be associated with chronic lymphedema after a mastectomy(Stewart–Treves syndrome), also lymphangiosarcoma develops within other forms of congenital and acquired lymphedma as well[3].

Lymphangioma circumscriptum has been reported to occur in cases of chronic lymphedema[4] , while Donald and Duffy(1979) reported a case of lymphangiosarcoma developed at the site of a preexisting lyphmangioma circumscriptum[5].

Recently MRI introduced as a useful tool to demonstrate the true extent of the underlying cisterns in cases of lymphangioma circumscriptum and to clarify the possible cause of lymphedema .

Report of cases:

Case - 1.

A 14- year- old girl, first noted to have swelling of her left leg at the age of six years, at age of eleven the right leg started to swell gradually, the lymphedema steadily increased in size to involve the entire legs from tip of the fingers to five cm below the knee joints.

At the age of thirteen ,clusters of flesh-colored 1-2mm papules had appeared overlying her left poplitial fossa, the papules were asymptomatic. Her mother stated that these multiple lesions were of six months duration at time of presentation.

Her family history was unremarkable. The patient past history revealed that she had congenital Atrial Septal Defect(ASD), pulmonary stenosis and mitral valve prolapse.

Operative treatment of the ASD was done in India 11th of July 2001.

Physical examination showed a well-developed young girl with fair general condition.

Lower extremities showed bilateral, pale cool skin , non-pitting edema extending from the tip of the fingers to five cm below the knee joints.(fig.1). At the left poplitial fossa there were multiple elevated grouped translucent vesicles. These lesions ranged in size from 0.3 to 1 cm in diameter (Fig.2& 3).On puncture, the fluid in the lesions is mostly straw-colored, but some others appeared to be bloody.

The eruption was not present in any other part of the body, and no regional lymph nodes enlargement were

palpable.

There are no consistently abnormal laboratory finding in this patient apart from that of the cardiac illness .

Lymphoscintography to the lower extremities of the patient to determined the level of obstruction of the lymphatic system showed an occlusion to the level of the aorta.

A skin biopsy specimen of two of the vesicles from the left poplitial fossa was taken, both section demonstrate the papillary dermis to be occupied by wide and distended cavities that are lined by endothelial cells. All of the vessels contain fluid consistent with lymph and do not contain RBCs. These vascular channels did not extend below the middermis. In the overlying epidermis there is some degree of acanthosis and hyperkeratosis and the surrounding stroma shows scattered lymphocytes. Fig (4).

lymphedema was subsequently managed with the use of elastic stockings.

An MRI to evaluate the depth and the extent of the underlying cisterns in the site of the lymphangioma circumscriptum before surgical excision was recommended, but her family refused the surgical treatment, and the alternative treatment suggested is flahlamp pumped dye laser.

Case-2 :

19-year-old female patient, born after a normal pregnancy and normal delivery.

At the age of one month the parents noticed that the right leg was larger than the left one. At age of one and half year she had a non-pitting edema on the right foot, by time the edema increased to involve the entire right leg up to the lower third of the thigh. The left leg also showed non-pitting edema but of lesser degree.

At the age of 16 the patient noticed on the right foot multiple gelatinous lesions which increased in number to involve the rest of the dorsum of the right leg .

Family history of the patient demonstrated that one of her four brothers had lymphedema of his lower extremities since the age of two years .

The patient medical history revealed that she has intestinal lymphangiectasia with gastro-esophageal reflux diagnosed by barium meal.

Physically: The patient is in good general condition . Lower extremities examination revealed the right leg is pale with non-pitting edema involving the foot, leg and lower third of the thigh. The left leg is also involved but to a lesser degree(Fig. 5).

At the dorsum of the right foot there were a multiple groups of flesh-colored papules containing straw-colored fluid (Fig. 6).

No other abnormalities were detected and the laboratory tests showed normal findings.

Biopsy specimen from the vesicular lesion affecting the dorsum of the right foot showed distended cavities filled with lymph in the papillary dermis while the epidermis shows some degree of acanthosis and hyperkeratosis (Fig. 7).

Magnetic Resonance Imaging(MRI) was done to clarify the possible cause of the lymphedema and to evaluate the depth and extent of the underling cisterns in the site of the lymphangioma circumsciptum demonstrated that : the number of lymph nodes in both inguinal regions was markedly decreased (Fig.8) suggesting hypoplastic development of the lymphatic system of the lower limbs as the cause of lymphatic obstruction and lymphatic edema. MRI also demonstrated multiple foci of cystic dilatation of lymphatic spaces found exclusively in subcutaneous region, indicating the depth of the lymphangioma circumsriptum (Fig.9).

The lymphedema of the patient was managed by pressure- supports, using elastic stockings and anti microbial treatment, while the lymphangioma circumsriptum was proposed to be treated by flashlamp pumped dye laser which has been used effectively in such cases.

Discussion and review of literature :

Lymphedema is a chronic swelling of a part due to obstruction or inadequate drainage of lymph outflow.

The lower limbs occasionally with the genitalia are most frequently involved [6]. There are two types of lymphedema:

Primary lymphedema is thought to result from an inherited abnormality of the lymphatic system.

It has been estimated to occur in about one in six thousands people, more often in females than in males[7]. Primary lymphedema can be present from birth (Congenital lymphedema) and symptoms can begin at the time of puberty (Lymphedema Praecox) or onset can occur in adulthood (Lymphedema tarda).

Lymphangiography and MRI in the primary lymphedema may reveal aplasia, hyperplasia or hypoplasia of the lymphatic system.

Secondary lymphedema is the result of a damaged or blocked lymphatic system caused by trauma, infection or injury.

Recurrent episodes of lymphangitis can result in scle-

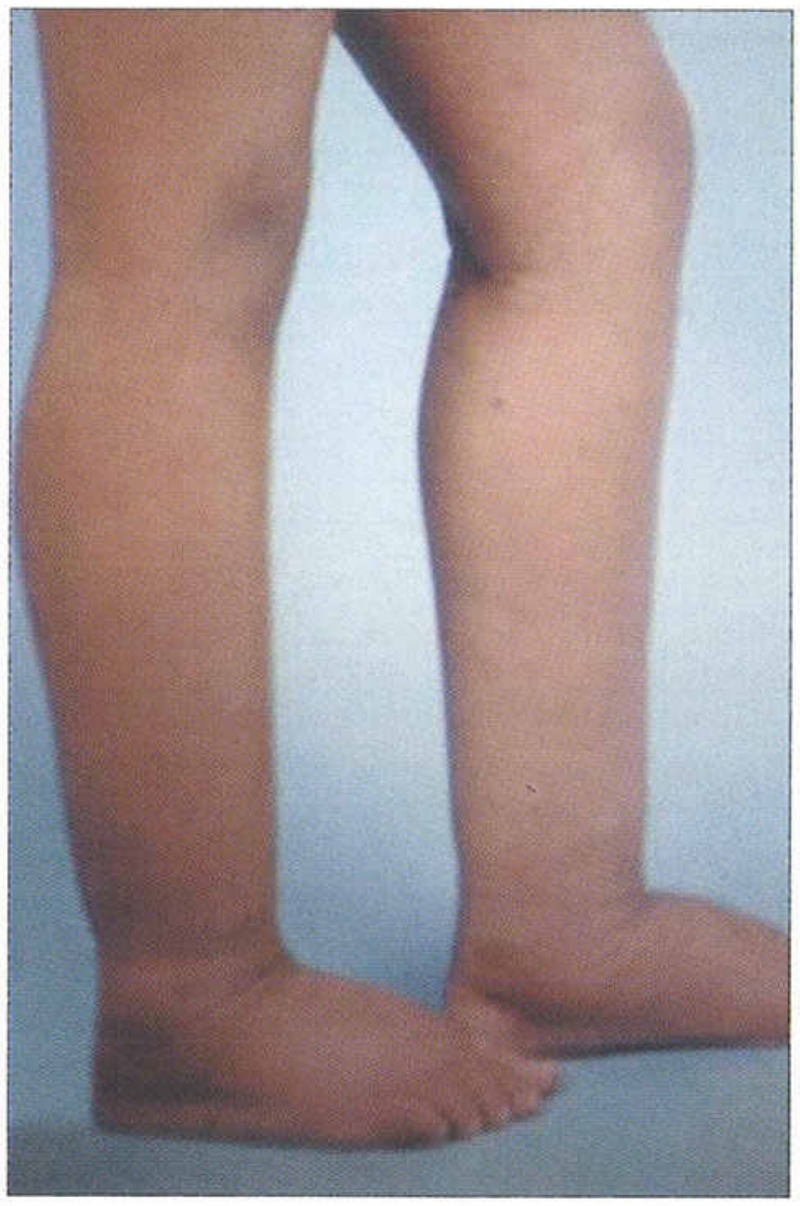


Fig.1 : Bilateral lymphedema

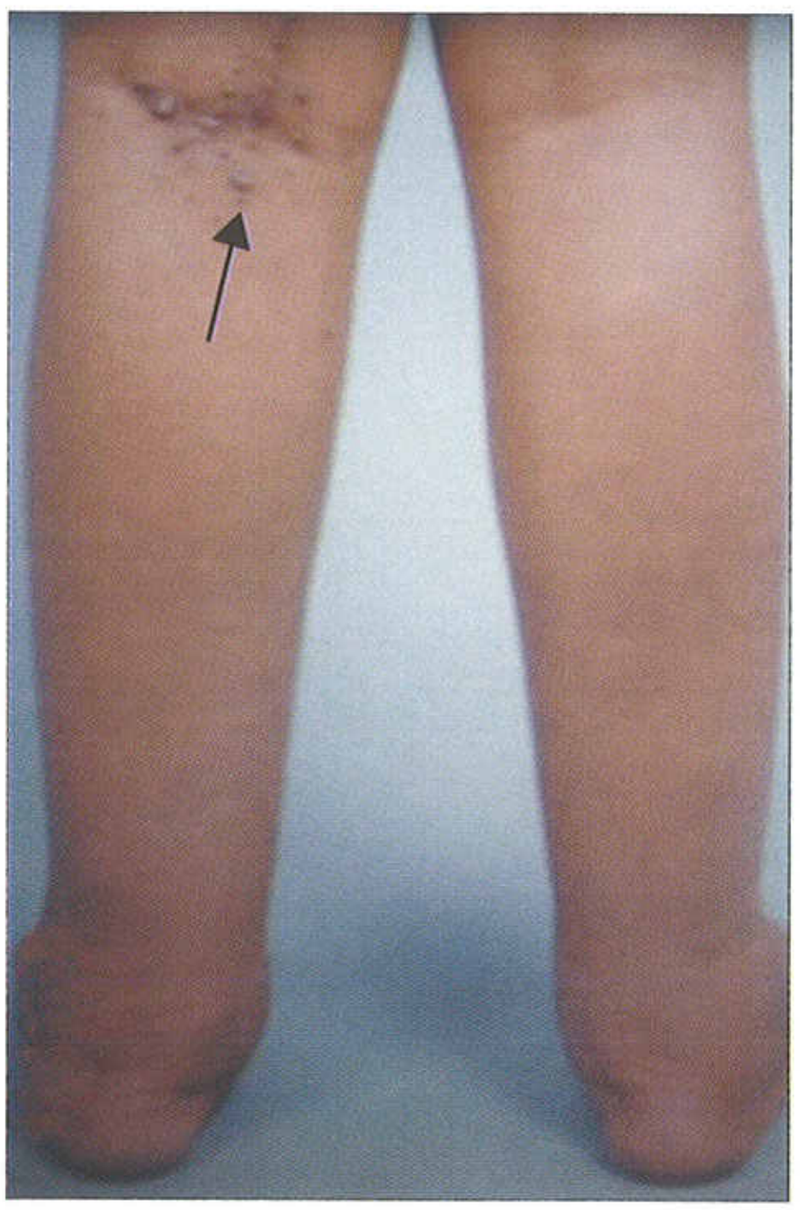


Fig.2 : Lymphangioma cirumscriptum at the left poplitial fossa

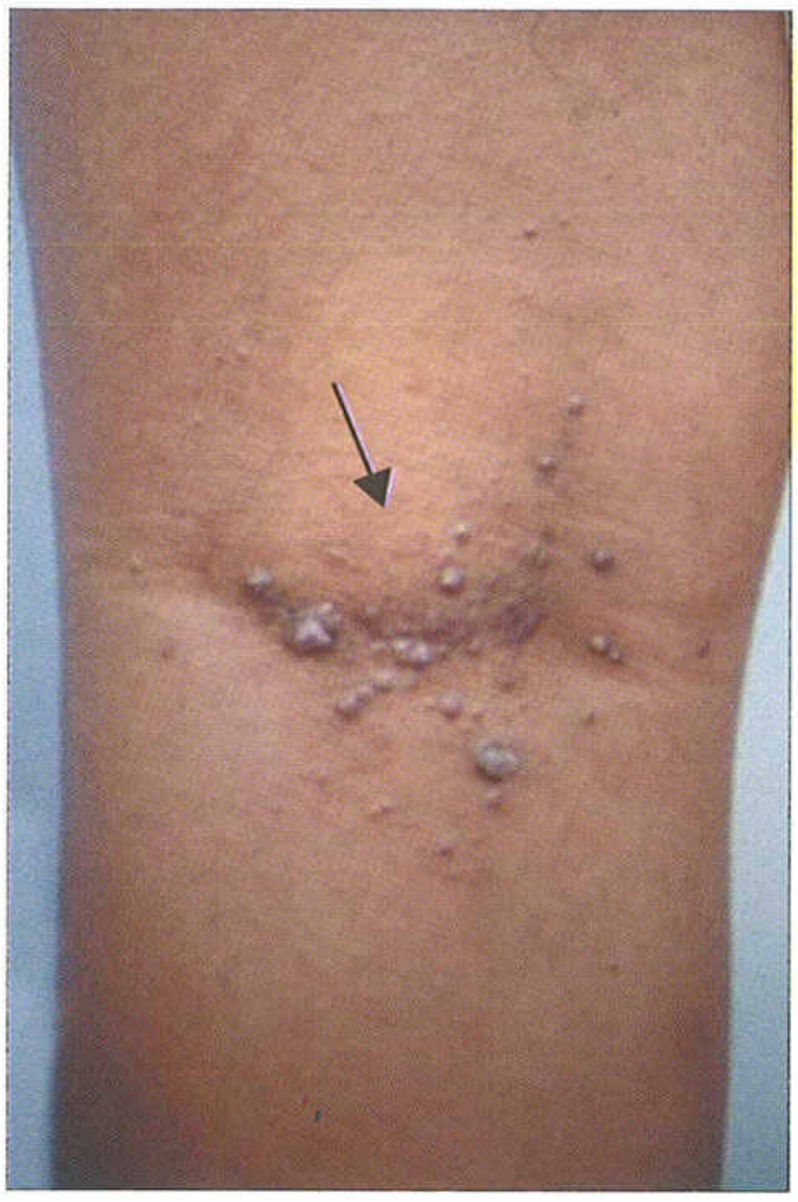


Fig.3 : Lymphangioma cirumscriptum at the left poplitial fossa (close - up)

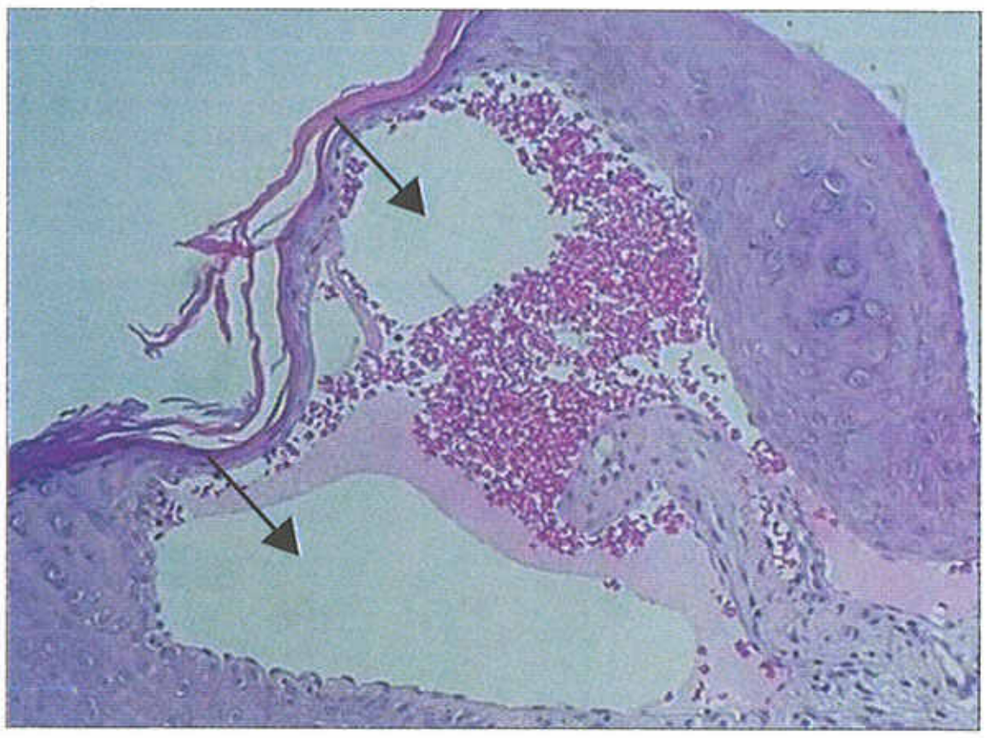


Fig.4 : Photomicrograph of the lymphangioma circumscriptum

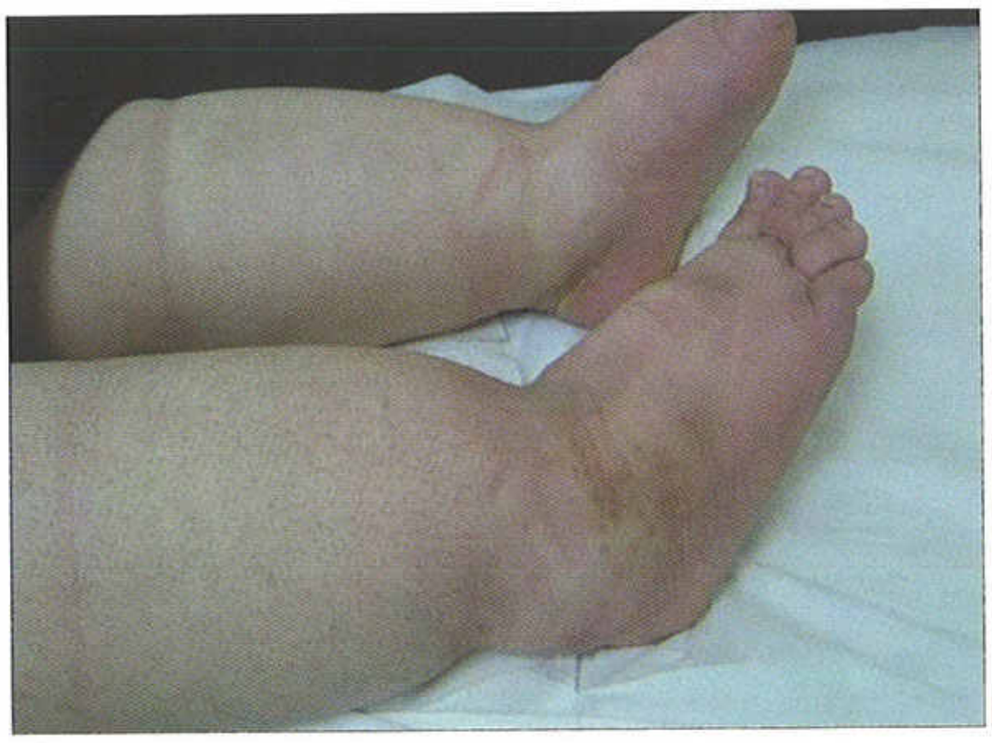


Fig.(5). Lymphedema of both legs.

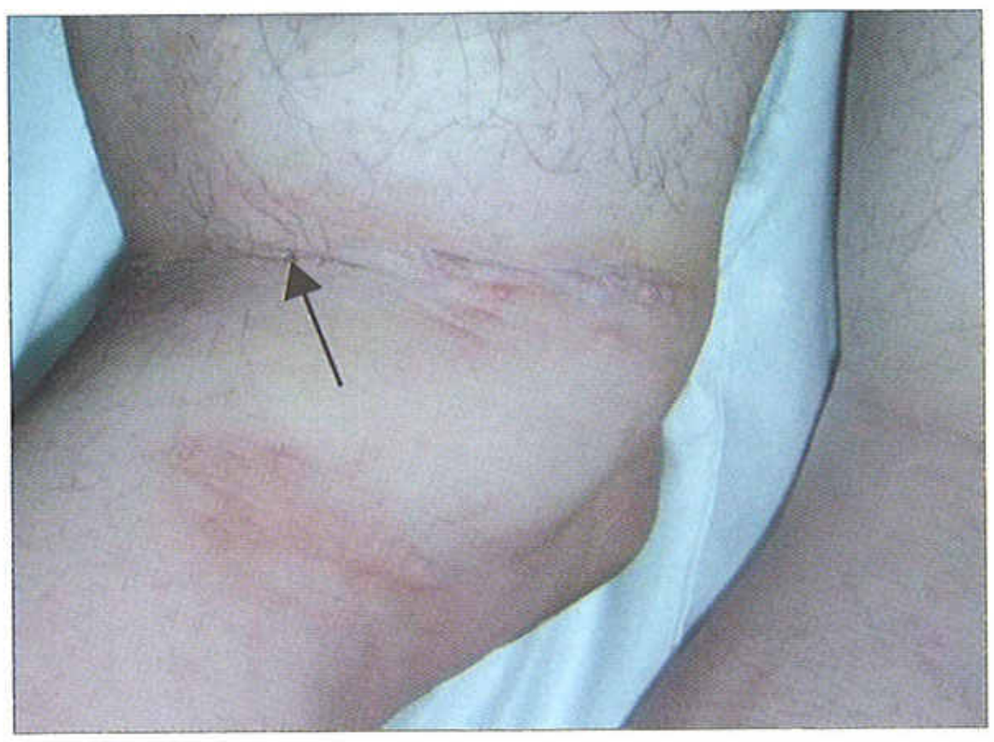
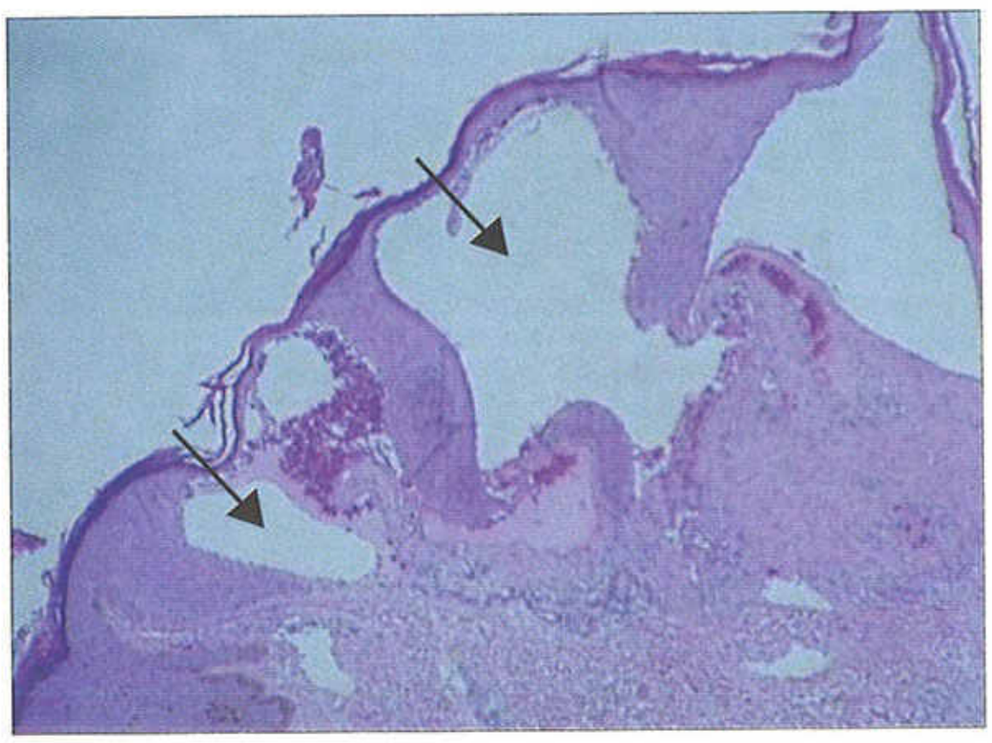


Fig.(6). Lymphangioma circumscriptum at the dorsum of the right leg .



Fig(7)Photomicrograph of lymphangioma circumscriptum at the dorsum of the right leg .

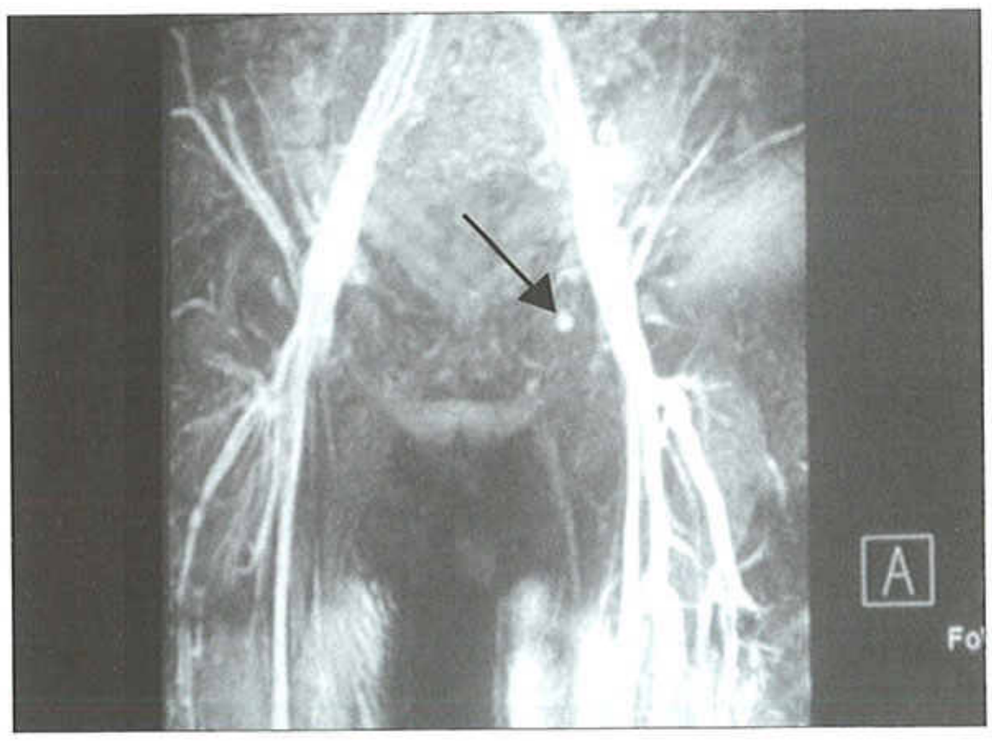


Fig.(8)MRI.The number of the lymph nodes in both inguinal regions markedly decreased.

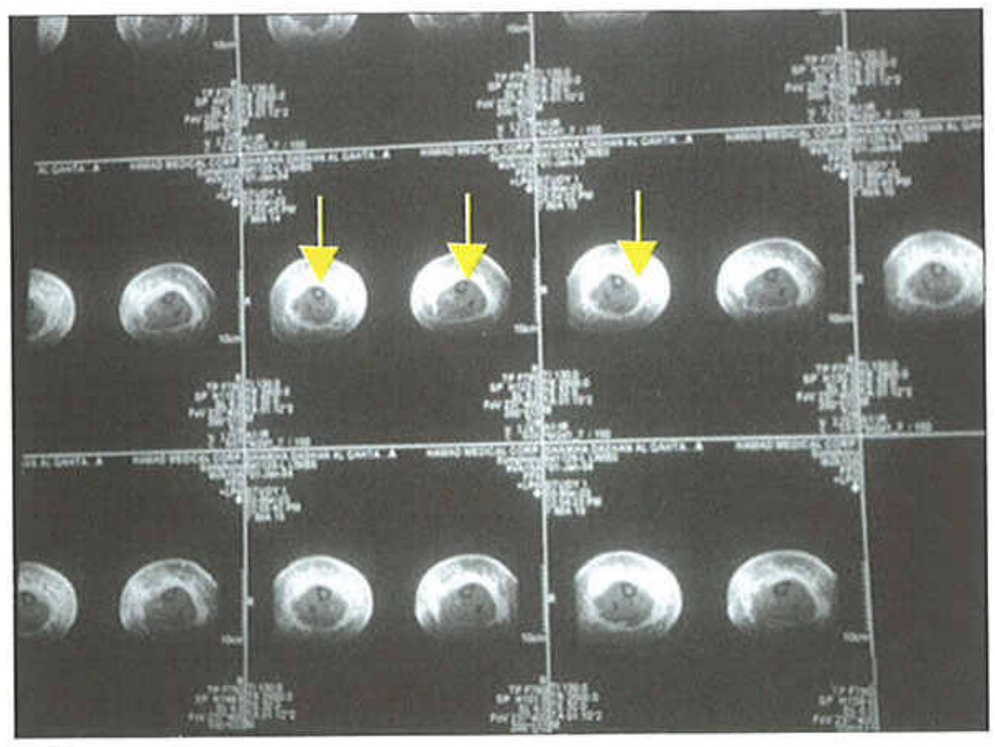


Fig.(9). MRI. Multiple foci of cystic dilatation of lymphatic spaces at the lymphangioma circumscriptum site..

rosis and great loss of lymphatic outflow. In roughly half of the cases of lymphedema the cause is unknown[8].

Clinically: swelling of the limb may be limited to the most distal part.

The affected limb may shows a pale swelling , pitting in pressure is present early but becomes minimal or absent late in the course while the skin is usually cool and normal looking[9].

The diagnosis of lymphyedema rests mainly on the relative persistence of the edema. In special cases lymphangiography or MRI may be necessary to define the diagnosis[10].

In our patient MRI demonstrate an aplasia of the lymph nodes in the inguinal region as cause of the primary lymphedema.

Lymphangiosarcoma is known to be associated with chronic lymphedema of an extremity, although most patients who develop Lymphangiosarcoma do so after a mastectomy (Stewart-Treves syndrome), it may also develops within other forms of congenital and acquired lymphyedema as well and the pathogenic mechanism by which lymphedema may induce lymphangiosarcoma is unknown[11].

The interval between the appearance of the lymphedema and the development of the lymphangiosarcoma ranges from one to thirty years[12]. In our patients in spite of the chronosity of the cases, there is no evidence of malignant changes demonstrated either clinically or by histology.

Whether the lymphedema is primary or secondary, treatment depends on reducing the edema as soon as possible and keep it under control with adequate pressure- gradient supports and prophylactic antimicrobial therapy.

Surgery has never solved the problem of lymphedema[13].'

Lymphangioma cicumsriptum has been reported to occur in cases of congenital lymphedema also[14]. Lymphangioma circumscriptum as a result of chronic lymphedema had been diagnosed in both of our cases.

Lymphangioma circumsriptum is a benign growth of lymphcontaining dilated lymphatic vessels lined by endothelium. it is most often located on the neck, upper trunk, extremities, perineum, glans penis, tongue and buccal mucosa.

Lymphangioma circumsriptum occurs world wide without sexual predilection or apparent genetic determination.

Most lesions are noted at birth or within first year of life, but some may have delayed onset [15].

The lesions of lymphangioma circumsriptum appear as a tense, grouped, thin-walled translucent vesicles of 1mm to 10mm in size, said to resemble "frog spawn"[16].

There are no characteristic laboratory changes in lymphangioma.

A superficial biopsy of lymphangioma circumsriptum will show only the dilated vessels situated in the papillary dermis.

The extent and depth of the lesion varies widely and cannot be adequately estimated from the cutaneous examination [17].

Magnetic Resonance Imaging (MRI) accurately demonstrated the true extent of involvement and prevent incomplete invasive surgical procedures from being performed [18]. In our patient MRI demonstrated multiple foci of cystic dilatation of lymphatic spaces found exclusively in subcutaneous region.

Lymphangioma circumsriptum may closely resemble a herpetic eruption, verruca vulgaris or linear verrucous epidermal nevi, the presence of serous fluid on aspiration will help clarify the differential diagnosis[19].

Reports document the occurrence of lymphangioma circumscriptum in the site of postmastectomy lymphedema.

Donald and Duffy(1997) reported a case of lymphangiosarcoma that developed at the site of a preexisting lymphangioma cicumsciptum on the lower part of the abdominal wall[20,5].

If treatment of lymphangioma circumscriptum is necessary, surgical excision is preferable.

The lesions are usually more extensive than they first appear and if inadequately excised they often recur, so deep excision including the cisterns is curative[21]

Flashlamp pumped dye laser can be used effectively to treat lymphangioma circumsciptum[22].

Our patients do not welcome surgery as a treatment option of their lymphangiomas, so we proposed flashlamp pumped pulsed dye laser as an alternative therapy.

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