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Case Report on Hereditary Lymphedema Associated Pleural Effusion

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Abstract

According to the National organization for rare disorder (NORD) defines Hereditary lymphedema as a genetic developmental disorder affecting the lymphatic system. Lymphatic system, commonly consist of vessels, ducts and nodes that distribute a protein -rich fluid called lymph. In lymphedema patients, the entire lymphatic system is disrupted, making a fluid buildup in the subcutaneous tissue resulting in swelling. It can affects limbs, legs as well as any parts of the body. Here in we report a case of Hereditary lymphedema with complaints of swelling in face as well as bilateral lower limbs along with scrotal oedema. Surprisingly, there is significant family history of consanguinity in his parents and first degree relatives.

Keywords

Hereditary Lymphedema, NORD, pleural effusion

Introduction

Hereditary lymphedema or primary lymphedema is a chronic disease characterized by abnormalities in the lymphatic system affecting the extremities especially upper or lower or both[1].Hereditary lymphedema can be classified into three types mainly congenital hereditary lymphedema or Milroy disease ; lymphedema precox or mei-ge's disease, which occurs at puberty and lymphedema tarda[2,3].In congenital lymphedema, swelling occurs in unilateral lower extremities. Oedema of multiple limbs ,genitalia as well as face can also be observed.[1]Lymphedema precox may be defined as lymphedema typically occurs after 35 years of age[2]. Here,swelling usually confined to foot and calf in majority of the patients[2].Secondary lymphedema occurs as result of any obstruction in the lymphatic pathways, trauma,inflammation ,neoplastic disruption

or surgery and is more common than primary form[2].Prevalence describes 1 in 6,000 individual with general population and therefore, is rare[3].The pathogenesis may be more likely to be consequence of poorly functioning lymphatic system with accumulation of protein and cellular metabolites such as molecular protein and hyaluronian leads to increased interstitial hydraulic pressure.Chronic lymphedema associated with increase in fibroblasts,adipocytes and keratinocytes in the edematous tissues ,ultimately leads to subcutaneous fibrosis[6].Complication associated with type 1 lymphedema include papillomatosis, urethral abnormalities and hydrocele (development of fluid filled sac along with spermatic cord of the scrotum in males.Type ii lymphedema (meige disease) ,some patient present with yellow nails.In some cases,symptoms may improve overtime.However ,obesity makes it difficult to manage the lymphedema and therefore, isproblematic. Affected individuals are more prone to develop infections such as lymphangitis affecting lymphatic system,cellulitis etc. They present with fever spikes, chills even headache. If left untreated, leads to bacteremia,ulceration and finally tissue damage.Diagnosis is confirmed by various imaging tests such as ultrasound ,lymphoscintigraphy, ultrasound and Magnetic resonance imaging.MRI is commonly used to detect hereditary lymphedema. One of the classic features of lymphedema distinguished from other extremities is stemmer sign which is inability to pinch the skin of the dorsum of the foot or hand.

Case Report

65 year old male patient, with medical history of lymphedema was admitted to a teritiary care hospital with complaints of breathlessness MMRC-3 with gradual onset, progressive orthopnea(+).He was presented with scrotal oedema, swelling in face as well as bilateral lower limbs. At the age of 15 year, the patient was diagnosed with hereditary lymphedema. His son told the patient neither took any medications nor any supportive measures in alleviating the symptoms associated with lymphedema, unless any hospitalization needed. Surprisingly, there is significant family history of consanguinity in his parents and first degree relatives. Physical examination showed bilateral upper and lower oedema,facial and scrotal oedema.Complete blood count, blood sugar level, renal function test,serum electrolytes ,serum albumin,serum globulin , albumin/Globulin ratio(A/G ratio) were normal during the course of illness. Mantourx test were negative. Arterial blood gas(ABG) ,ECG, USG-Abdomen, ECHO , CT-Thorax were done. ABG were done in alternative days, showed type 2 respiratory failure. Chest Roentgenography showed right blunting of costophrenic angle & bilateral infiltration. Thoracentesis was done in which 150 ml of pleural fluid aspirated which was straw in colour revealed transudative pleural effusion. Pleural fluid analysis showed turbid blood stained, rich in LDH. In addition, pleural fluid culture & sensitivity was performed & revealed sterile after 48 hrs of incubation. USG-Abdomen and pelvis showed bilateral mild to moderate pleural effusion. Moreover, fatty changes in liver was found. CECT -Thorax revealed moderate right pleural effusion with encystment in the upper coastal pleura noted causing complete atelectasis of the right lower lobe and posterior segment of the right upper lobe.

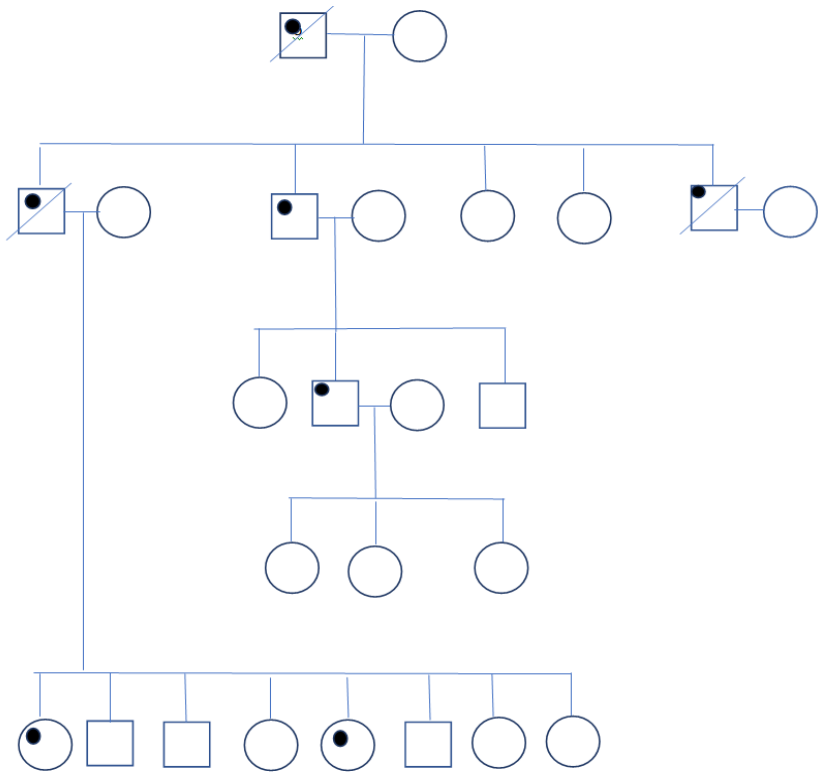

Figure 1: Patient's Family Tree is Consistent with Autosomal Dominant Inheritance of Hereditary Lymphedema.

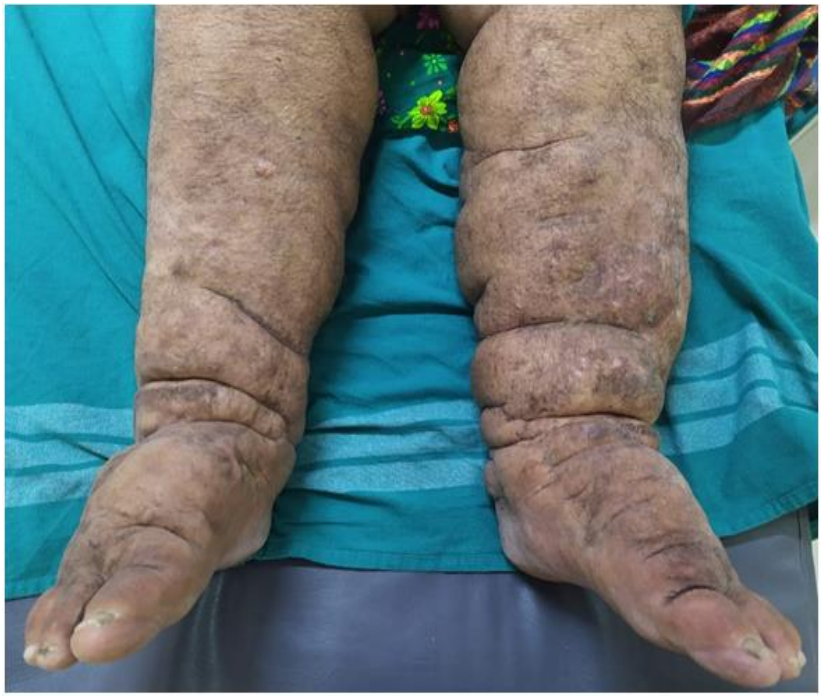

Figure 2: this picture depicts bilateral leg edema, which is the classic feature of hereditary lymph edema

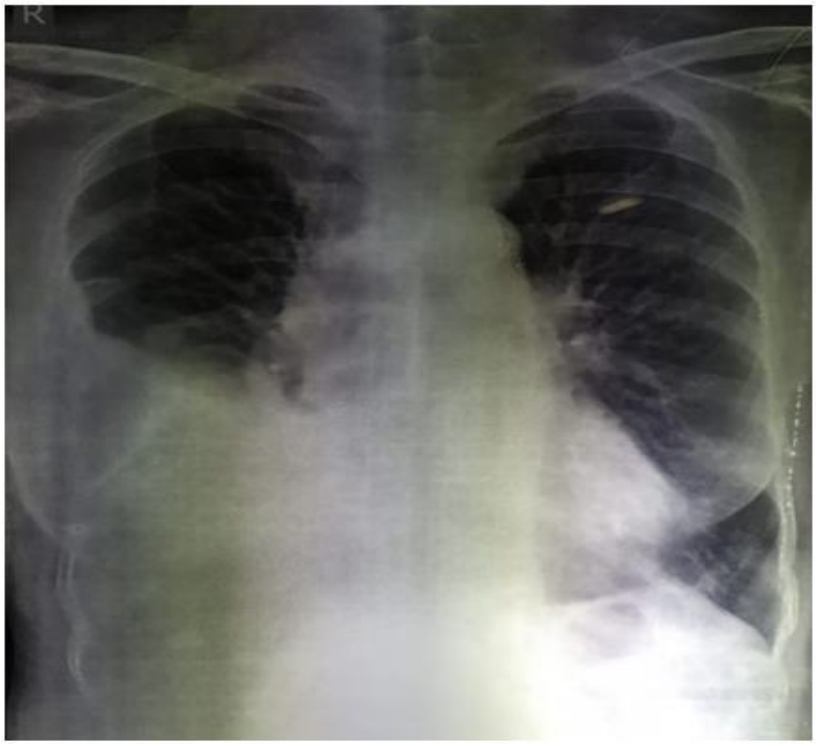

Figure 3: Chest Roentgenography of the Patient Demonstrating the Right Moderate Pleural Effusion

Discussion

Hereditary lymphedema ,a form of primary lymphedema , is a chronic illness in which lymphatic vessels fails to transport lymphatic fluid, resulting in swelling of the extremities[7].The patient had a non-pitting edema for last 50 years. Non pitting edema is the main characteristic feature of lymphedema. The patient was evaluated for cellulitis and septicemia. In addition, there were no blistering or breaks in the skin. Since the patient diagnosed with moderate right pleural effusion ,heart failure and liver disease was also ruled out. The tuberculosis were excluded after the proper laboratory testing . Patient was advised not to use tourniquet in affected limbs. Meticulous skin as well as nail care is necessary and advised to use skin moisturizer. This helps to control further skin infections. If not properly treated, leads to lack of focusing on daily activities. Bitterly, there is no ultimate drug found beneficial for lymphedema[7].Patient was advised for cardiac consultation to rule out pulmonary edema,CCF since D-Dimer was elevated and started LMW heparin for 5 days, T.rivaroxaban 20mg for 2 weeks. Furthermore, dermatology consultation was also advised. The treatment for lymphedema remains unclear ,with a goal of controlling symptoms rather than a cure[8] .Preventative measures advised for patient include massage therapy , not to use elastic bandages or stockings. Bilateral mild to moderate pleural effusion made this patient needed for hospitalization with symptom of breathlessness. Pleural fluid analysis and proper physiotherapy can alleviate symptoms in patient with pleural effusion.

Conclusion

Hereditary lymphedema is rare, longstanding condition affecting the lymphatic system.The development of pleural effusion is remarkable in these patients.Long term complication associated with hereditary lymphedema decreases the quality of life and persistence of well-being.So, this case report provides important insights into a rare disease and also educating the healthcare professionals .Hence ,large follow up is necessary for the prevention of complications.

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