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EXTENSIVE PELVIC AND RETROPERITONEAL LYMPHANGIECTASIA WITH T AND B CELL IMMUNODEFICIENCY IN AN ADOLESCENT GIRL: A RARE CASE REPORT

Dr. Shruti Appaswamy*	Assistant Professor, Pediatrics, GGMC And JJH *Corresponding Author
Dr. Bela Verma	Head Of Department And Professor, Pediatrics, GGMC And JJH
Dr. Ananya Mathur	Junior Resident. Pediatrics, GGMC And JJH

ABSTRACT Lymphangiectasis is defined by widespread, abnormal enlargement of the lymphatic vessels without developing excess lymphatic tissue, large fluid pockets, septations, or notable stromal components. This condition can be either primary (congenital) or secondary (acquired). The symptoms can vary widely depending on the severity and location of the affected lymphatic vessels- such as abdominal pain, diarrhoea, protein losing enteropathy, oedema, persistent vaginal discharge which can lead to failure to thrive, micronutrient deficiencies, immunocompromised state and increased susceptibility to infections. In this case report we have an adolescent girl, presenting with a rare form of lymphangiectasia- pelvic and retroperitoneal in origin, along with T and B cell immunodeficiency. After overcoming diagnostic challenges, a precise treatment was planned for the patient. Treatment options include both medical and surgical modalities. Given the scarcity of published reports on paediatric lymphangiectasia, this case serves to enrich the limited data available and underscores the need for further research and clinical awareness in this area.

KEYWORDS : pediatrics, lymphangiectasia, immunodeficiency, protein-losing enteropathy, sclerotherapy

INTRODUCTION

Lymphangiectasis is defined by widespread, abnormal enlargement of the lymphatic vessels without developing excess lymphatic tissue, large fluid pockets, septations, or notable stromal components. (1) This condition can be either primary (congenital) or secondary (acquired), with the primary form sometimes occurring on its own or as part of various congenital syndromes, such as Noonan, Ullrich-Turner, Ehlers-Danlos, and Down syndromes. Primary lymphangiectasis can affect multiple organs, including the lungs, liver, spleen, retroperitoneal structures, and the gastrointestinal tract.(2) Secondary forms can result from conditions such as retroperitoneal fibrosis and tumours, intestinal scleroderma, pancreatitis, constrictive pericarditis, congestive heart failure, regional enteritis, radiation- induced enteritis, mesenteric and diffuse lymphomas, and abdominal tuberculosis.(3) In this case report, we have a female patient, aged eleven years, presenting with vulvar swelling and a copious mucoid vaginal discharge. Examination was suspicious of ambiguous genitalia, with a Sexual Maturity Rating (SMR) of I. A comprehensive clinical and diagnostic evaluation was performed to investigate the underlying cause. MRI imaging of the abdomen and pelvis along with lymphangiography identified a rare form of lymphangiectasia. In view of recurrent infections requiring intravenous and oral antibiotics, an immunodeficiency workup was performed which revealed T cell and Memory B cell deficiency.

Managing lymphangiectasia can indeed be challenging. Along with medical therapies such as high protein diet, albumin infusions, octreotide and anti-inflammatory drugs, CO_2 laser and sclerotherapy have been documented as a treatment modality in several cases, offering precise targeting of lymphatic lesions to reduce swelling and discharge. However, both treatments require careful consideration of the patient's specific condition and long-term monitoring due to potential complications.

Case Study

An 11-year-old girl, presented with complaints of persistent copious vaginal discharge since 3 years of age. The discharge was whitish coloured, mucoid and foul-smelling, occurring every 2-3 months and lasting for a few days. On probing, there was no history of trauma, sexual assault, fever, burning micturition, loose stools, urinary complaints, abdominal distension or pain. Patient was birth and developmentally normal, attending school which was however being hindered by the vaginal discharge. On examination, the patient was vitally stable but was however severely wasted and pale. On local examination, there was copious whitish vaginal discharge with multiple rugosities and localised swelling around the vulva (Fig 1 and 2). Sexual Maturity Rating of the patient was 1. In view of possible ambiguous genitalia, an endocrinology reference was done for the patient.

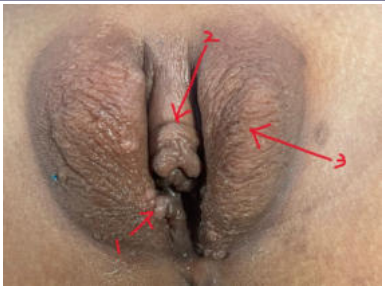

Fig 1 showing 1) cutaneous lymphangiectasia 2) apparent clitoromegaly 3) vulval oedema

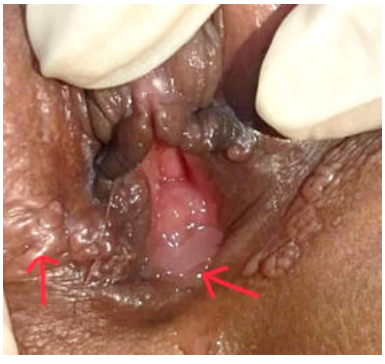

Fig 2 showing cutaneous lymphangiectasia (left) and vaginal discharge (centre)

Subsequently a battery of tests was planned for the diagnostic evaluation.

- **CBC**: Anaemia with lymphopenia
- **LFT**: Hypoalbuminemia
- **Vaginal Swab**: Mixed growth of commensals
- **Serum LH, FSH, TFT, Testosterone, DHEA**: all within normal limits
- **Karyotyping**: 46XX
- **Ultrasonography Abdomen**: Mild bulky cervix with hypoechoic endocervical canal with minimal fluid in the vaginal canal? cervicitis. Minimal free fluid in the abdomen and pelvis
- **MRI Pelvis**: moderate free fluid in abdomen and pelvis with mesenteric fat stranding. Submucosal oedema in the rectum. Extensive T2W1 and STIR hyperintense areas noted within entire pelvis, encasing the rectum and other pelvic organs. Similar hyperintense areas noted in entire retroperitoneum, peripancreatic

areas and along the descending colon. Hyperintensities also noted in wall of vagina, cervix, rectum, sigmoid colon- consistent with involvement. STIR hyperintense lymphatic channels underneath the vulval region noted. Enlarged reactive bilateral inguinal and external iliac nodes. Mild ascites noted. On dynamic contrast, no early enhancement noted. Gradual persistent enhancement seen on delayed image, suggestive of slow flow/lymphatic origin. (Fig 3 and 4)

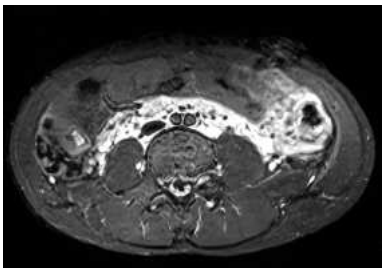

Fig 3: MRI showing uptake in the retroperitoneal region

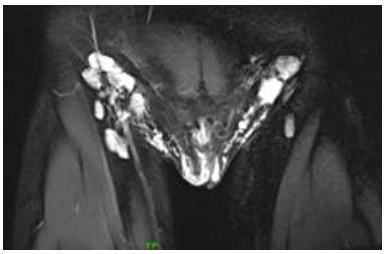

Fig 4: MRI showing uptake in the rectal and pelvic regions

- **Lymphoscintigraphy**: Abnormal collection of tracers in the pelvis above the urinary bladder favouring lymphatic collection and lymphangiectasia. (Fig 5)

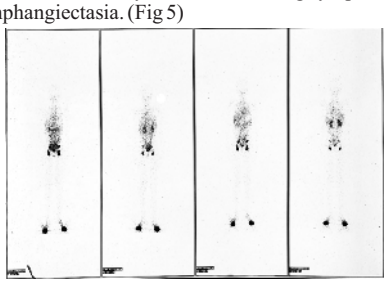

Fig 5: Lymphoscintigraphy showing tracer in the pelvic region

A diagnosis of extensive pelvic and retroperitoneal lymphangiectasia was made. Following subsequent hospital admissions, the patient was diagnosed with pleuropulmonary tuberculosis and multidrug-resistant organism urosepsis, prompting a workup for immunodeficiency:

- **Decreased Levels Of Absolute T Lymphocytes (CD3+/CD19-):** 891 cells/µl (Normal range 1000-2200).
- **Decreased Levels Of Absolute TH Lymphocytes (CD3+/CD4+):** 389 cells/µl (Normal range 530-1300).
- **Decreased Levels Of Tc Lymphocytes (CD3+/CD8+):** 233 cells/µl (Normal range 330-920)
- **Decreased Levels Of Absolute NK Cells (CD3-/CD16+/CD56+):** 40 cells/µl (Normal range 70-480)
- **Decreased Memory B Cells (CD19+/CD27+):** 6.75% (Normal range 7-29%)

The patient was started on appropriate antibiotics and anti-tubercular treatment. She also needed multiple albumin infusions for hypoalbuminemia. Post stabilization, the patient was initiated on a high-protein, low-fat diet, along with iron and medium-chain triglyceride supplementation. An interventional radiology consultation was obtained, and local vulval sclerotherapy was planned. The patient remains under close follow-up and is scheduled to undergo sclerotherapy.

DISCUSSION

The causes of lymphangiectasia can be classified into two broad categories: primary and secondary.

Primary Lymphangiectasia: The congenital form typically manifests in infancy or early childhood and results from developmental abnormalities of the lymphatic system. While genetic factors may contribute, the exact aetiology often remains unclear. Primary lymphangiectasis may occur in isolation or in association with various congenital syndromes, including Noonan syndrome, Ullrich-Turner syndrome, Ehlers-Danlos syndrome, and Down syndrome. It can involve multiple organ systems, such as the lungs, liver, spleen, retroperitoneum, and gastrointestinal tract. (2)

Secondary Lymphangiectasia: Develops later in life and is typically caused by an underlying condition that disrupts normal lymphatic function. Contributing factors may include infections, inflammation, neoplasms, or iatrogenic damage to the lymphatic system. Management is often more complex, as it requires addressing the primary condition. Reported causes of secondary lymphangiectasia include retroperitoneal fibrosis and tumours, intestinal scleroderma, pancreatitis, constrictive pericarditis, congestive heart failure, regional enteritis, radiation-induced enteritis, mesenteric and diffuse lymphomas, lymphatic trauma, surgical disruption of lymphatic channels, and abdominal tuberculosis. (3)

Although the patient was later diagnosed with tuberculosis, the onset of clinical symptoms at the age of three years cannot be fully explained by this condition alone. Given the early age of presentation and the persistence of features beyond what is typically attributable to tuberculosis, this case is more appropriately classified as primary lymphangiectasia. Genetic testing could not be done due to financial constraints.

The symptoms of lymphangiectasia can vary widely depending on the severity and location of the affected lymphatic vessels. Common presentations include oedema, abdominal pain, diarrhoea, chylothorax, vaginal discharge, recurrent infection and failure to thrive. (4) A secondary form of immunodeficiency due to chronic loss of lymph through dilated intestinal lymphatic vessels may occur. This results in depletion of both T and B lymphocytes, contributing to impaired cellular and humoral immunity. The loss of immunoglobulins (IgG, IgA, and IgM) through the gastrointestinal tract further weakens the adaptive immune response, rendering patients susceptible to recurrent bacterial, viral, and fungal infections. T-cell dysfunction compromises cell-mediated immunity, while reduced B-cell counts and hypogammaglobulinemia impair antibody production and long-term immune memory. Consequently, affected individuals often present with frequent respiratory and gastrointestinal infections, poor vaccine responses, and delayed recovery from common illnesses, which is evident in our patient. (5)

Imaging plays a vital role in visualizing the lymphatic system and detecting structural abnormalities associated with lymphangiectasia. Commonly used imaging modalities include lymphoscintigraphy (6), MRI, CT scan (7) and genetic studies in the setting of a suspicion of primary lymphangiectasia.

Supportive evidences include anaemia, lymphopenia, hypoalbuminemia, hypogammaglobulinemia, elevated stool alpla-1 antitrypsin levels, fat-soluble vitamin deficiencies, increased triglyceride levels in the fluid with lymphocytic predominance (>80%) and dilated lymphatic vessels on histopathology. (8)

Treatment for lymphangiectasia aims to manage symptoms and address the underlying cause. The approach varies depending on whether the condition is primary or secondary. Dietary modifications being one of the first steps in managing lymphangiectasia aims at reducing the burden on the lymphatic system which includes low fat diet, medium-chain triglycerides, high protein and fat-soluble vitamin supplementations (A, D, E, K). (9) Several medications can help manage symptoms and complications of lymphangiectasia. These include diuretics (help reduce oedema), anti-Inflammatory drugs like steroids (reduce inflammation in the lymphatic vessels), albumin Infusions and octreotide (causes splanchnic vasoconstriction) (9)

In Severe Cases, Minimally-invasive And Surgical Interventions May Be Necessary. These Can Include:

- **Electrodesiccation** uses high-frequency electric currents to destroy superficial lymphangiectatic lesions and is primarily used for small, localized cutaneous lymphatic malformations. (10)
- **Laser therapy** using CO_2, Nd:YAG, or diode lasers targets and seals off superficial dilated lymphatic vessels and is particularly

useful for treating lymphangiectasia of the vulva, oral cavity, or skin. It is less invasive, allows precise targeting, and can be repeated if needed. (11)

- **Cryotherapy** involves applying extremely cold liquid nitrogen to destroy abnormal lymphatic tissue, suitable for small, superficial lesions and can be performed under local anaesthesia. However, recurrence is possible and multiple sessions may be required. (12)
- **Sclerotherapy** involves injecting sclerosant agents such as doxycycline, bleomycin, or ethanol directly into dilated lymphatic vessels to induce inflammation and fibrosis, leading to obliteration of the abnormal channels. It is minimally invasive and particularly effective for localized lymphatic malformations, including vulvar or retroperitoneal lymphangiectasia. (13)
- **Lymphatic Bypass Surgery/ lympho-venous anastomosis** is a microsurgical technique connecting lymphatic vessels to nearby veins, allowing lymph to drain directly into the venous system, bypassing the obstructed segments. It is indicated in refractory cases of lymphangiectasia with significant lymphatic stasis. Success depends on the availability of functional lymphatic channels and patient selection. (14)
- **Debulking Surgery** is done in advanced or disfiguring cases to remove hypertrophic or fibrotic tissues that compress lymphatic channels and contribute to lymph accumulation and is typically reserved for severe lymphoedematous or elephantiasis-like presentations and may be combined with reconstructive procedures. (15)

CONCLUSIONS

Lymphangiectasia is an uncommon disorder that necessitates both medical and surgical interventions. The loss of lymphocytes associated with this condition results in immunodeficiency, thereby predisposing patients to recurrent infections that require comprehensive management. However, definitive treatment ultimately depends on addressing the underlying aetiology.

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