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ORIGINAL ARTICLE

Congenital chylothorax and primary lymphoedema: a case report

Beatriz Maria Vilaça[1], João Arthur Pazello[1], Adria Karina Farias[2], Leilane de-Oliveira[2], Miguel Angelo Stremel Andrade[2], Andre Ivan Bradley dos Santos Dias[2], Miguel Angelo Agulham[2], Camila Girardi Fachin[2]

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Abstract

Objective: To report the case of a child diagnosed with congenital chylothorax and primary lymphedema without chromosomal changes. **Methods:** The patient's medical records were reviewed and the literature was reviewed in the Pubmed database. **Results:** Female patient with prenatal ultrasound presenting bilateral pleural effusion, subcutaneous edema and right pyelectasis. At birth, at term, she evolved with groaning and tachypnea, and bilateral chylothorax, laryngomalacia and generalized lymphedema were found. Upon genotypic evaluation, the karyotype was normal (46, XX). At 6 months of age, we opted for treatment with sildenafil, a low-fat diet and semi-elemental formula. During her first year of life, the patient was hospitalized several times for respiratory failure and underwent multiple chest drainage. She had severe pneumonia and at 1 year of age had an infected chylothorax. The patient developed refractory septic shock and acute renal failure with renal necrosis, dying at 1 year and 22 days of life. **Conclusion:** In this report, there is a rare association between congenital chylothorax and primary lymphedema in the absence of genetic syndrome.

[1] Federal University of Paraná, Medical School - Curitiba - Paraná - Brazil.
[2] Federal University of Paraná, Pediatric Surgery Department - Curitiba - Paraná - Brazil.

Correspondence to:
Beatriz Maria Vilaça.
Universidade Federal do Paraná - Curso de Medicina. Rua Padre Camargo, nº 285, Alto da Glória.
Curitiba - PR. Brasil. CEP: 80.060-240. E-mail: bea.vilaca@gmail.com

INTRODUCTION

Congenital chylothorax may be the result of malformation, acquired obstruction, or rupture of the thoracic duct[2]; rare causes include lymphangioma and lymphangiectasia[1]. It is often associated with chromosomal abnormalities such as Down, Turner, and Noonan syndromes[11]. Currently recommended treatments include fluid drainage, hemodynamic support and total parenteral nutrition followed by nutrition with medium chain triglyceride-enriched formula[7]. Sildenafil and octreotide have shown benefits in some reports[2,5].

Lymphedema is a rare condition, affecting approximately 1 in 6,000 children[4]. The buildup of lymph in certain tissues and regions of the body due to failure or overload of drainage mechanisms characterize it. Lymphedema may be primary or secondary to other conditions such as tumors and infections. When it is primary, it can be isolated or part of a syndromic condition - such as Down, Turner and Noonan Syndromes, Trisomy 13 and 18[3]. It`s causes may be: lymphatic duct aplasia or hypoplasia, thoracic or abdominal lymphatic trunk abnormalities or valvar incompetence of the lymphatic system[3].

No cases were found in the literature associating primary lymphedema and congenital chylothorax in the absence of genetic syndromes.

METHODS

We reviewed the patient`s chart and carried out a literature review in Pubmed.

RESULTS

Female patient, born at full term, weight 3355g, Apgar 8/9. At the 32nd gestational week, ultrasonography revealed bilateral pleural effusion, subcutaneous edema and right-side pyelectasis. At birth, she evolved with groaning and tachypnea, and bilateral chylothorax, laryngomalacia and generalized lymphedema were diagnosed. She was referred to the Neonatal ICU of the University Hospital. Upon genotypic evaluation, the karyotype was normal (46, XX). Chest computed tomography revealed bilateral pleural effusion, collapse of the lower lobes, and irregularity of the pleural surface, with no changes suggestive of lymphangiectasia or lymphangiomatosis. Lymphoscintigraphy showed no significant drainage through the lymphatic pathways in the hands, feet and liver. After clinical recovery, the patient was discharged from the hospital at 14 days of life, and an outpatient return was scheduled in 7 days. After three days, the patient was brought in by her parents and readmitted with perioral cyanosis, tachypnea and choking after breastfeeding. The patient received supplemental oxygen and total parenteral nutrition until clinical improvement and discharge at 1 month of life.

At 3 months of age, her parents to the emergency room with fever, tachypnea and drowsiness again brought her in. She was diagnosed with infected chylothorax and underwent videothoracoscopy with decortication on the right and pleural drainage closed by massive chylous effusion on the left. Due to edema accompanied by oliguria, it was necessary to introduce diuretics (Spironolactone and Furosemide) and albumin replacement.

At 6 months of age, we opted for treatment with Sildenafil and a low-fat diet associated with the semi-elemental formula. Five months later, she was brought back by her parents and admitted with the same symptoms of perioral cyanosis, tachypnea, fever, and lower limb edema. Despite a small left pleural effusion, conservative treatment with Ceftriaxone for the infection and Furosemide to reduce edema was implemented. Laboratory results revealed an important anemia, for which we prescribed blood transfusion. Fifteen days later, the patient was discharged after significant clinical recovery.

During her first year of life, the patient was hospitalized several times for respiratory failure and underwent four thoracic drainages. During this time a new lymphoscintigraphy was performed, which revealed significant hypoplasia of the lymphatic system in the upper and lower limbs, associated with drainage deficiency.

At 1 year of age, the patient was readmitted to the hospital with tachypnea, severe edema, fever and tachycardia. Left chest drainage (200 mL of exudate) was performed due to a large pleural effusion. The patient was referred to the ICU, evolved to treatment-refractory septic shock and acute renal failure with renal necrosis, and died at 1 year and 22 days of life.

For administrative reasons of the Hospital, no autopsy and other genetic investigations were performed. The family did not authorize images of the case.

DISCUSSION

Chylothorax is a rare condition, more common in males[7] and is often associated with chromosomal abnormalities.[3] It is characterized by the buildup of chylous fluid in the pleural cavity, with potential metabolic, immunological and nutritional complications, as well as respiratory failure, sometimes fatal. It is the most common type of pleural effusion in newborns, and the most commonly associated with respiratory failure. Cavity fluid buildup occurs when clearance is reduced, production is increased, or an association of both.[8]

The term congenital is usually reserved for abnormalities of the lymphatic system that are present during birth. It may be the result of malformation, acquired obstruction or due to rupture of the thoracic duct.[1] Rare causes include lymphangioma and lymphangiectasia.[2] In this case report, a potential cause could be drainage deficiency caused by the absence of functioning lymphatic ducts, as suggested by hypoplasia of the upper and lower limb lymphatic system, evidenced during lymphoscintigraphy.

This condition may be associated with a multitude of chromosomal abnormalities such as Down, Turner, and Noonan syndromes.[3] This patient, however, had a normal karyotype (46, XX). Most patients are diagnosed before birth, as was the case via ultrasound, and most are associated with fetal hydrops. [9] Respiratory manifestations such as tachypnea and dyspnea make up the clinical features of this condition. The buildup of chylous fluid in the pleural cavity can lead to protein, electrolyte, and white blood cell depletion, leading to dehydration, hypovolemia, malnutrition, electrolyte disturbances, metabolic acidosis, and lymphopenia immunosuppression.[10]

Postnatal treatments may include fluid drainage, hemodynamic and vascular support, total parenteral nutrition followed by the medium-chain triglyceride formula. The use of Sildenafil in pediatric patients with lymphatic malformations has been shown to be efficient.[5] In addition, some reports have indicated Octreotide as a beneficial drug in the treatment of such conditions.[1]

Overall mortality in patients suffering from pleural effusions in the neonatal period is 25%, ranging from 15 in isolated strokes to 95% in those associated with fetal hydrops.[11]

Lymphedema is also a rare condition, being present in 1 of 6,000 newborns.[12] It is characterized by lymph buildup in certain tissues and body parts due to failure or overload of drainage mechanisms. Lymphedema can be a primary disease or secondary to other abnormalities such as tumors and infections. When primary, it can be isolated (as in this report), or part of a syndromic disorder such as Down, Turner, and Noonan syndromes, as well as in the 13 and 18 trisomy.[6]

Lymphedema can present in three patterns: aplasia or hypoplasia of lymphatic ducts, as in this case, abnormalities of the thoracic and abdominal ducts, or due to valvar incompetence of the lymphatic system.[6] In addition, any condition that may develop fetal hydrops has the potential to develop primary lymphedema.[13] The diagnosis of this patient was during the prenatal period, at the 32nd gestational week, associated with fetal hydrops (a condition that has 95% mortality when associated with pleural effusion, as per described above).

Evaluation of the etiology may include lymphoscintigraphy, a test that captures fluid flow in the lymphatic system, especially in the limbs. It is done by injecting radiolabeled elements into the limbs, followed by sequential path images that follow through the lymphatic system. Drainage is considered impaired when delayed, absent or asymmetrical (comparing both limbs).

CONCLUSION

In the present case, despite the coexistence of congenital chylothorax and primary lymphedema, there were no chromosomal abnormalities detected by the patient's karyotype. This fact was considered unusual, since during the literature review no associations of these two conditions were found with the absence of chromosomal abnormalities.

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