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CASE REPORT

Pulmonary thromboembolism in children with Duchenne Muscular Dystrophy: A Case Report

Daniel Raylander da Silva Rodrigues[1], Paulo Sucasas Costa[1], Patrícia Marques Fortes[1], Dégila da Costa Cruz[2], Flávia Medeiros Fonseca[2], Deborah Cristina Fogaça[2]

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Pulmonary embolism,
Muscular dystrophy,
Duchenne,
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Abstract

Objectives: To describe a clinical case of pulmonary thromboembolism (PE) in a child with Duchenne muscular dystrophy. **Methods:** A detailed description of the clinical picture and a review of the patients medical record were carried out, in addition to a discussion based on a literature review. **Discussion:** PTE is not common in children and in 95% of cases it is secondary to an underlying cause or disease, with Duchennes disease being a predisposing condition due to the hypercoagulable state. Diagnosis in the pediatric population is not supported by tests validated as for adults; in this case, CT angiography was the exam of choice with sensitivity and specificity above 90%. In addition to diagnosis, a risk classification is essential, and supports the therapeutic choice. Therefore, low-risk patients receive anticoagulation, while high-risk patients receive thrombolytic therapy; in patients with contraindications to these drugs, surgical intervention may be necessary. Correct identification and early management are essential to delay disease progression and prevent sequelae, as well as avoid unnecessary invasive approaches. **Conclusion:** Due to the difficulty in diagnosing pulmonary thromboembolism in children, a detailed clinical investigation is essential, through the discussion of differential diagnoses and assessment of risk factors, since other diseases can mimic this clinical condition, which can present with signs and nonspecific symptoms.

[1] Hospital das Clínicas UFG, Departamento de Pediatria - Goiânia - Goiás - Brazil
[2] Universidade de Rio Verde, Faculdade de Medicina - Aparecida de Goiânia - Goiás - Brazil

Correspondence to:
Daniel Raylander da Silva Rodrigues.
Hospital das Clínicas UFG. Rua 235 QD. 68 Lote Área, Nº 285, s/nº - Setor Leste Universitário, Goiânia, GO, Brazil. CEP: 74605-050. E-mail: danielraylander@gmail.com

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INTRODUCTION

Pulmonary Thromboembolism (PTE) is not a common condition among children, having been a diagnostic challenge for pediatricians since its first report by Von Löschner, almost two centuries ago. The wide clinical spectrum, the limitation of complementary exams and the lack of scientific evidence in the pediatric population mean that a high index of suspicion is necessary for its identification and appropriate treatment[1].

Venous thrombosis occurs when erythrocytes, platelets, fibrin and leukocytes form a mass inside a vessel. This phenomenon arises from the interaction of Virchow's Triad factors (endothelial injury, vascular stasis and hypercoagulability), and in up to 95% of cases there is some underlying cause, such as central venous access (the umbilical catheter being the main factor), cancer, congenital heart disease, trauma, surgery or immobilization, infection, nephrotic syndrome, systemic lupus erythematosus, sickle cell anemia, hypovolemia, obesity, oral hormonal contraceptives and previous venous thrombosis[2].

Among the predisposing causes are muscular dystrophies, such as Duchenne Disease, in which susceptibility is due to a state of hypercoagulability, without a direct relationship with age, respiratory or cardiac function, in addition to chronic vascular obstructions and endothelial hyperplasias[3,4]. Despite the high prevalence of venous thrombosis in this group, the occurrence of pulmonary thromboembolic events remains uncommon, resulting in worse outcomes when present[3].

The objective of this paper is to describe the case of PTE in a child with Duchenne muscular dystrophy, helping to identify and treat other cases.

CASE REPORT

This is an 11-year-old, male patient, previously diagnosed with Duchenne Dystrophy, obese, bed-ridden, unable to walk, complaining of falling from a wheelchair three days ago and beginning of prostration, hyporexia, dyspnea and chest pain since then. He had no fever or cough. Upon physical examination, he was pale 1+/4, afebrile, tachydyspneic, conscious and oriented. Cardiac auscultation showed no change in rhythm, heart rate was 144 beats per minute, blood pressure of 100 x 60 mmHg in the right upper limb. Reduced vesicular murmur on the right pulmonary base with fine local rales, respiratory rate of 50 breaths per minute, peripheral saturation of 85% in room air. Paretic lower limbs with bilateral edema, greater in the right calf, with pain when moving the ipsilateral knee.

Admitted for investigation with empirical antibiotic treatment for treatment of community-acquired pneumonia and oxygen therapy through a nasal catheter, with a good response through saturation.

CBC revealed hemoglobin 11.5 g/dL, 14,600 leukocytes/mm^3 (predominance of segmented cells without shift to the left), 269,000 platelets/mm^3, sodium 139 mmol/L, potassium 2.8 mmol/L, magnesium 2.2 mg/dL, C-reactive protein 5 mg /dL (10 times above the reference value), urea 12 mg/dL, creatinine 0.3 mg/dL, CK 3455 U/L, CK-MB 157 U/L, negative troponin. Arterial blood gas analysis with pH 7.44, pO2 89 mmHg, pCO2 25.4 mmHg, bicarbonate 17.1 mEq/L and base excess (BE) -5.4. Chest X-ray with discrete bilateral perihilar infiltrate, without condensation or effusions.

DISCUSSION

With scientific advances and the increase in the survival rate of critical and chronic patients, the number of PTE cases has increased. In the pediatric population, its incidence varies from 8.6 to 57 cases for every 100,000 hospitalized, while in those without hospitalization the rate is no more than 0.9. There is a predominance in black children (2.38 times higher than in whites) and a bimodal incidence with a peak in infants and adolescents[1].

Unlike the adult population, this clinical condition can be silent with patients even being asymptomatic, which delays diagnosis by up to seven days. The classic presentation includes respiratory discomfort, pleuritic chest pain (especially in adolescents), hemoptysis, cough, syncope and desaturation. However, tachycardia, tachypnea, limb edema and signs of heart failure are also common[1,2].

The complaint of dyspnea may be based on the difficulty in obtaining air or the feeling of suffocation, and sometimes, like other subjective conditions, it is difficult to diagnose. The list of differential diagnoses ranges from anatomical causes (vascular rings, stenoses and atresia) to external causes (extrinsic compressions due to adenopathy, masses or hematomas, foreign bodies in the airway and trauma with direct injuries) and infectious (pneumonia, bronchiolitis, epiglottitis and laryngitis). The mode of installation, associated symptoms, comorbidities and other risk factors contribute to the diagnostic conclusion[5].

While probability tests are used in the adult population – with emphasis on the Wells Criteria, Geneva score and PERC (Pulmonary Embolism Rule-out Criteria) – the pediatric population lacks similar validated tests, with current studies only bringing reasonable results[1] . In this way, the importance of diagnostic suspicion is highlighted when there is the presence of risk factors and non-specific symptoms[6].

In the clinical case presented herein, in which the patient had a basic condition with a known state of hypercoagulability, in addition to physical restriction, associated with an acute case of chest pain, tachydyspnea and desaturation in the absence of fever and diagnostic radiographic image, the hypothesis of pulmonary thromboembolism was raised.

The diagnostic assessment of PTE can be divided into two groups: specific ones and those that identify the severity of the condition. Regarding the diagnosis of thromboembolism, CT angiography, magnetic resonance imaging (MRI), ventilation/perfusion (V/Q) testing and pulmonary arteriography stand out[2].

CT angiography is the test of choice due to availability, time to perform and interpret, as well as low cost and high accuracy (90% sensitivity, 95% specificity, 9.3% false-positive and 2.4% false-negative). On the other hand, it uses ionizing radiation, is contraindicated in renal failure and is of low quality in cases of small peripheral embolisms[1].

Based on data extrapolated from the adult population, severity assessment looks for signs of cardiac overload resulting from increased pressure in the pulmonary vasculature to classify patients and guide therapy. High-risk patients are those with signs of cardiovascular collapse, while those at intermediate risk are normotensive, but with evidence of cardiac overload on the electrocardiogram, echocardiogram or elevated markers of muscle damage. Low-risk patients do not show signs of cardiac or hemodynamic involvement[1].

The investigation of the reported case began with an electrocardiogram that showed an S1Q3T3 type change: S wave in DI, Q wave in DIII and T wave inversion in DIII (Figure 1). Ultrasonography of the lower limbs with Doppler showed no changes. With chest angio-CT, the diagnosis was concluded by showing filling failure with intraluminal thrombus and increased caliber in the segmental and subsegmental branches of the lateral and posterior basal segments of the right lower lobe (Figure 2). As a sequential assessment, an echocardiogram was performed with signs of increased pulmonary arterial pressure and dilation of the right heart chambers, in order to characterize the condition as being of moderate risk.

Identification and correct management promote, in addition to stopping the progression of the disease, the prevention of sequelae and unnecessary invasive approaches. Due to the lack of specific algorithms for the pediatric population, the recommended treatment is based on studies and recommendations made to the adult population[1].

Treatment is divided into a pharmacological (anticoagulants and thrombolytics) and mechanical approach, with the indication varying according to the risk classification. Low-risk patients will receive anticoagulation, while high-risk patients will receive thrombolytic therapy. Those at intermediate risk should be evaluated for the use of thrombolytics[1].

Anticoagulants aim to interrupt the thrombus formation cascade, being represented by Unfractionated Heparin (UFH) and Low Molecular Weight Heparin (LMWH). UFH is indicated in patients without clinical stability or those on a

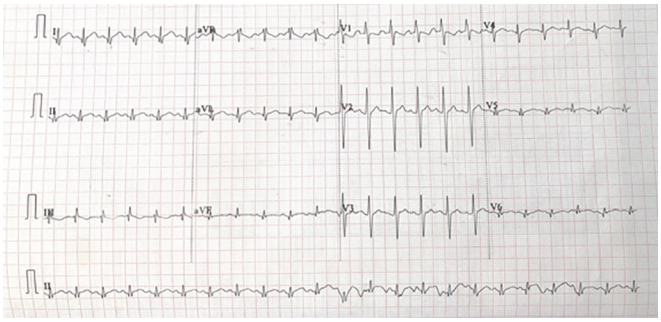


Figure 1. Electrocardiogram.

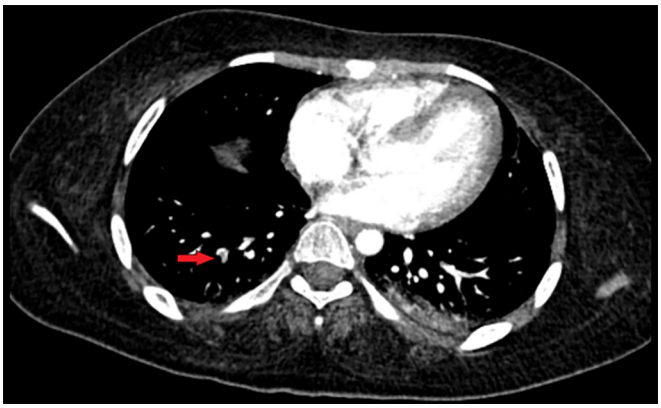


Figure 2. Chest angio-CT; red arrow shows : obstruction to contrast flow in the lower segment of the right lung.

higher risk of bleeding (given the shorter half-life and possibility of an antidote) at a dose of 75 U/kg intravenously and an infusion rate that varies according to age: in those > 1 year infuse at 20 U/kg/h; in < 1 year at 28 U/kg/h[1].

LMWH ranges from 1 mg/kg, in those over 2 months of age, to 1.5 mg/kg/day in younger children, without the need for venous access as it is administered subcutaneously. Maintenance treatment is carried out with LMWH or vitamin K antagonist (warfarin) lasting three months to one year[1].

Thrombolytics can be used in peripheral access or close to the clot through catheterization of the pulmonary circulation, aiming for faster resolution, thus reducing right ventricular overload. Recombinant tissue plasminogen activator (rtPA) is preferable to streptokinase due to its lower immunogenicity, greater activity on clots and greater specificity to fibrin. Its dose varies in the literature, with two current protocols: one uses higher doses (0.1 to 0.6 mg/kg/h for 6 hours) with a greater probability of clot resolution, but greater risk of bleeding, while the second presents lower doses (0.03 to 0.06 mg/kg/h, maximum 2 mg/h), with similar efficacy, but lower risk of adverse effects[1].

Mechanical thrombolysis and surgical thromboendarterectomy, despite the limited experience in pediatric patients of the latter, are indicated in those patients at high risk of bleeding or contraindications to thrombolytics and

anticoagulants, in those who have failed pharmacological therapy or in cases of hemodynamic instability[1].

The patient in this case in question responded well to oxygen therapy and, due to hemodynamic stability, we opted for anticoagulation with a vitamin K inhibitor associated with low molecular weight heparin until obtaining an International Normalized Ratio (INR) of 2.0, when suspension of the second and discharged from hospital for outpatient follow-up, which happened after seven days of treatment. Currently, the patient continues to use an oral vitamin K antagonist, with good INR control, without episodes of bleeding or thrombotic events.

CONCLUSION

Pulmonary thromboembolism in children can present subjective signs and symptoms that mimic other conditions, making diagnosis difficult and late, which makes detailed clinical investigation necessary due to the presence of risk factors for the development of this condition. This identification of PTE associated with correct management promotes interruption in the progression of the disease, prevention of sequelae and avoids unnecessary invasive approaches.

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