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Lung Involvement in Gaucher Disease

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ABSTRACT

Introduction: Gaucher Disease is an autosomal recessive lysosomal storage disease. Pulmonary involvement in Gaucher Disease is rare and often seen in the severe form of the disease with the worst outcome.

Case Presentation: A 30-year-old man and known case of Gaucher Disease presented to our clinic with history of progressive dyspnea since 8 months ago. Pulmonary function test showed restrictive pattern. Chest CT scan revealed diffuse bilateral interlobular septal thickening and small interstitial nodules with ground glass opacities in lower lobes.

Conclusion: Patients with Gaucher Disease that present with progressive dyspnea may have a manifestation of interstitial or alveolar lung disease.

Introduction

A 30-year-old man with Gaucher Disease (GD), confirmed by his bone marrow finding from 20 years ago, presented to the clinic with a history of progressive dyspnea since 8 months ago. He had a history of splenectomy due to huge splenomegaly and severe thrombocytopenia. On examination, he had fine rales in the base of both lungs and severe hepatomegaly. His chest x-ray (Figure 1) showed bilateral diffuse reticulonodular opacities. High resolution Computed Tomography of the chest (Figure 2) revealed diffuse bilateral interlobular septal thickening and small interstitial nodules with ground glass opacities in lower lobes. Pulmonary function test showed restrictive pattern as follows: FVC= 2.63 (65%), FEV1= 2.09 (60%), and FEV1/FVC= 82%. Echocardiography showed an appropriate systolic

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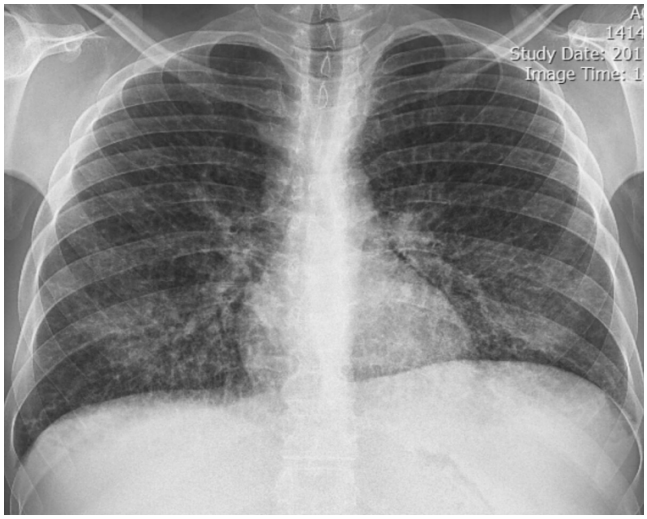

Figure 1. Chest-x-ray diffuse reticulonodular opacities in both lungs are visible

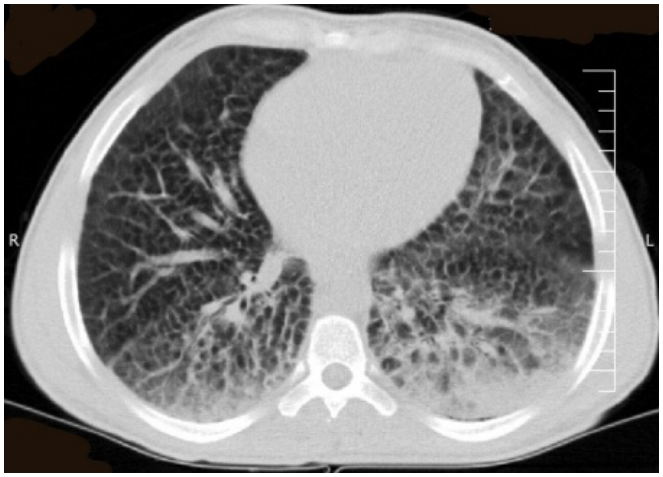

Figure 2. Chest CT scan showing diffuse bilateral interlobular septal thickening and small interstitial nodules with ground glass opacities in the lower lobes

function (ejection fraction=55%) with calcified aortic valve and pulmonary arterial pressure of 30 mm Hg. Video bronchoscopy revealed no proliferative masses in the evaluated areas. The Bronchoalveolar Lavage (BAL) was negative for Koch's bacilus.

GD is an autosomal recessive lysosomal storage disease. Pulmonary involvement in GD is rare and often seen in the severe form of the disease with the worst outcome [1]. Deficiency of beta glucocerebrosidase enzyme leads to the accumulation of glucocerebroside in the reticuloendothelial system, lung, and heart [2].

Pulmonary involvement ranges from clinically asymptomatic with normal imaging to severe respiratory findings due to infiltration of gaucher cells in alveolar, interstitial space, and peribronchial vascular tissue [3]. Lung involvement is diagnosed with chest x-ray, high resolution CT of the chest and pulmonary function test. The radiologic investigation is often normal, but may present with typical reticular, nodular, or reticulonodular interstitial involvement [4].

Enzyme replacement therapy and substrate reduction therapy are two approaches in patients with complications, such as anemia, thrombocytopenia, organomegaly, skeletal disease, liver and lung involvement [5]. As a result, the patient with GD who presented with progressive dyspnea may have a manifestation of interstitial or alveolar lung disease.

Ethical Considerations

Compliance with ethical guidelines

All ethical principles were considered in this article.

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Conflict of interest

The authors declared no conflicts of interests.

References

[1] Tofolean D, Adam T, Mazilu L, Parepa IR. A case of late diagnosis of Gaucher disease with lung involvement (case study). ARS Medica Tomitana. 2015; 21(2):63-6. [DOI:10.1515/arsm-2015-0022]

[2] Mehta S. Severe pulmonary involvement with cor pulmonale; the initial presenting feature of non-neuronopathic form of Gaucher's disease. Journal of Pediatric Sciences. 2015; 7:e245. [DOI:10.17334/jps.45559]

[3] Sherwani P, Vire A, Anand R, Gupta R. Lung lysed: A case of Gaucher disease with pulmonary involvement. Lung India. 2016; 33(1):108-10. [DOI:10.4103/0970-2113.173086] [PMID] [PMCID]

[4] Ceyhan M, Celik F, Elmali M, Gurmen N. An unusual form of Gaucher's disease: Pulmonary and cardiovascular involvement and cholelitiasis. Central European Journal of Medicine. 2010; 5(4):495-8. [DOI:10.2478/s11536-009-0062-1]

[5] Linari S, Castaman G. Clinical manifestations and management of Gaucher disease. Clinical Cases in Mineral and Bone Metabolism. 2015; 12(2):157-64. [DOI:10.11138/ccmbm/2015.12.2.157] [PMID] [PMCID]