Interstitial Lung Disease Associated with Adult Niemann-Pick Disease Type B

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A 66-year-old female presented with chronic dry cough and mild dyspnea. She had no medical history but reported Niemann-Pick disease (NPD) type B in her sister. Physical examination was normal except for hepatosplenomegaly. Pulmonary function tests showed a decrease in carbon monoxide diffusion capacity without obstructive or restrictive ventilatory pattern. Thoracic

computed tomography showed bilateral ground-glass opacities and interlobular septal thickening in the upper and lower lung zones (Fig. 1). Bronchoalveolar lavage showed predominance of “foamy” macrophages (93%) (Fig. 2). Blood acid sphingomyelinase activity was decreased, and chitotriosidase activity was increased, as found in NPD type B. Genetic testing found a homozy-

Fig. 1. Thoracic computed tomography showing ground-glass attenuation predominantly in the left lung, with interlobular septal thickening, compatible with interstitial lung disease in Niemann-Pick disease type B.

Fig. 2. Cytological analysis of bronchoalveolar lavage (×40) after fixation with methanol and Papanicolaou stain, showing “foamy” macrophages (round cells with a large cytoplasm) containing multiple lipid-laden vacuoles.

gous mutation (p.R610del) in the *SMPD1* (sphingomyelin phosphodiesterase 1) gene, confirming NPD type B disease. Screening for autoantibodies was negative. No specific treatment was initiated. The patient's respiratory condition is stable after a 3-year follow-up.

NPD is a rare genetic lysosomal lipid storage disease, characterized by a defect in the sphingomyelinase enzyme activity. Pulmonary involvement is common in NPD type B, and mainly consists in progressive interstitial lung disease [1]. Radiological findings include ground-glass opacities, thickened interlobular septa, intralobular reticulation, nodules, and cysts [2]. Respiratory outcome is generally favorable, although some cases may present with severe interstitial lung disease.

Financial Disclosure and Conflicts of Interest

The authors declare that there are no conflicts of interest concerning this paper.

Author Contributions

N.C., F.T.-B. and V.C. all contributed in the conception, design, analysis, and interpretation of the manuscript.

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