ANSWER TO PHOTO QUIZ (PAGE 441)

TWO SIBLINGS WITH HEPATOSPLENOMEGALY AND PULMONARY RETICULATION

DIAGNOSIS

The results of the hepatic biopsy of the older patient were consistent with lysosomal storage disease and suggested Niemann-Pick disease. Neurological examination and respiratory function test results were normal. White blood cell sphingomyelinase activity was reduced, confirming the diagnosis of Niemann-Pick disease type B.

Niemann-Pick disease is a rare recessive and autosomal hereditary lysosomal storage disease caused primarily by an absence or deficiency of the enzyme acid sphingomyelinase. It is characterised by the intracellular accumulation of sphingomyelin in the liver, spleen, lungs, bone marrow, and/or brain, resulting in the presence of lipid-laden 'foamy' macrophages (Niemann-Pick cells) in these organs.[1-4]

Niemann-Pick disease presents with three subtypes. Type A is a severe neurodegenerative disorder that results in death in early childhood.[1,4,5] Type B is less severe, with little or no neurological involvement.[1,5] Type C is caused by a different genetic mutation.[4] Niemann-Pick disease type B is characterised by hepatosplenomegaly and pulmonary and bone marrow involvement, with an insidious course and a more benign prognosis.[1,2,4] Pulmonary involvement ranges from a lack of symptoms to respiratory failure with oxygen dependence.[1,2] High-resolution computed tomography (HRCT) findings consist of smooth thickening of the interlobular septa mainly involving the lower lung zones, intralobular lines, and patchy ground-glass opacities throughout both lungs, sometimes with a crazy-paving pattern.[2-5]

The diagnosis of Niemann-Pick disease is made by bone marrow biopsy or sphingomyelinase enzyme assays in leukocytes, lymphocytes, or fibroblast skin cultures.[3,5] On histopathological examination, the lungs show alveolar filling by lipid-laden macrophages, which causes a type of endogenous lipoid pneumonia. Macrophages also infiltrate the alveolar walls and interstitium, but the lung architecture is generally preserved. Fibrosis is generally absent or mild.[1,3,5]

In conclusion, Niemann-Pick disease type B must be considered in patients presenting with dyspnoea and HRCT findings of interlobular and intralobular septal thickening in association with small foci of ground-glass opacity, especially when associated with a long-term history of hepatosplenomegaly or when a patient has family members with comparable problems.

REFERENCES

1. Guillemot N, Troadec C, de Villemeur TB, Clément A, Fauroux B. Lung disease in Niemann-Pick disease. Pediatr Pulmonol. 2007;42:1207-14.
2. Baldi BG, Santana AN, Takagaki TY, Fujita C, Kairalla RA, Carvalho CR. Lung cyst: an unusual manifestation of Niemann-Pick disease. Respirology. 2009;14:134-6.
3. Rodrigues R, Marchiori E, Müller NL. Niemann-Pick disease: high-resolution CT findings in two siblings. J Comput Assist Tomogr. 2004;28:52-4.
4. Ahuja J, Kanne JP, Meyer CA, et al. Histiocytic disorders of the chest: imaging findings. Radiographics. 2015;35:357-70.
5. Mendelson DS, Wasserstein MP, Desnick RJ, et al. Type B Niemann-Pick disease: findings at chest radiography, thin-section CT, and pulmonary function testing. Radiology. 2006;238:339-45.