The Echocardiographic Characteristics of Cardiac Involvement of Adult Mitochondrial Encephalomyopathy, Lactic Acidosis and Stroke-like Episodes Syndrome

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Abstract

Purpose: Diagnosis of mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes (MELAS) syndrome is often difficult in adult patients, especially those with mild clinical manifestations. The myocardium is susceptible to mitochondrial abnormalities, and therefore, echocardiography might play a role in the early diagnosis of MELAS. However, details of the echocardiographic findings have not yet been fully investigated. Accordingly, the aim of this study was to clarify the characteristic echocardiographic findings in adult patients with MELAS.

Subjects and Methods: Eight patients with MELAS who were undergoing echocardiography were included. Clinical symptoms, laboratory test results, and echocardiographic findings were investigated.

Results and Discussion: Among the 8 patients, neuromuscular symptoms were present in 6, hearing loss in all, diabetes in 6, and short stature in 4 patients Additionally, abnormalities were observed in lactic acid level in 7, lactic acid to pyruvic acid ratio in all, and BNP in 6 patients. Echocardiographic findings showed preserved left ventricular ejection fraction in all patients, apparent left ventricular hypertrophy and abnormal global longitudinal strain in 6, and impaired diastolic function in 5 patients. A fine-particle bright-echo ultrasonic appearance in the hypertrophied myocardium, so-called "granular sparkling echo", which is seen in glycogen storage disease or amyloidosis, was subjectively observed in 5 patients. Accordingly, in patients with left ventricular hypertrophy with abnormally bright echo accompanied by deafness, diabetes, or short stature, it is important that mitochondrial disease be considered in the differential diagnosis.

Conclusion: Echocardiographic findings of adult patients with MELAS were characterized by apparent left ventricular hypertrophy with fine-particle bright-echo appearance accompanied by left ventricular global longitudinal strain dysfunction and relaxation delay.

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