Echocardiographic role in the detection and monitoring of Anderson-Fabry disease

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Anderson-Fabry disease is a hereditary defect of the enzyme alpha-galactosidase, characterized by the accumulation of glycosphingolipids in lysosomes. Patients with Fabry disease are not able to catabolize the membrane neutral glycosphingolipids, especially globotriaosylceramide, which therefore accumulates mainly in the heart, skin, kidneys, blood vessels and central nervous system. The disease is recessive and sex-linked, it is being transmitted with the X chromosome. Characteristic symptoms and signs of Fabry disease are the presence of angiokeratomas (the occurrence of vascular skin lesions), acroparesthesias (periodic painful crises in the extremities), hypohidrosis (the inability to sweat) and the characteristic blurring of the cornea. The disease starts at birth, during childhood it is usually without clinical significance, organ damages become manifest during the fourth decade of life in males and during fifth decade in females. The heart manifestations of the disease are a result of accumulated globotriaosylceramides in cardiomyocytes, conduction system cells and valves, with the main consequence being hypertrophic cardiomyopathy of the left or both ventricles. In an advanced stage of the disease, the cardiac muscle changes can lead to heart attack, cardiomyopathy and conduction disturbances. Echocardiography is an excellent noninvasive diagnostic method for the detection of Fabry disease (left ventricular hypertrophy, state of the heart valves and dimension of cardiac chambers). Doctors' and nurse technicians' coordination is obligatory for optimal echocardiographic examination, detection and further follow up of the disease. In patients with Fabry disease suspicion, fingerstick or venous blood samples were taken. After the drying process, samples were sent for enzymatic activity and genetic testing. The follow-up of the disease is carried out by the echo controls, determination of biomarkers (GL3, Lyso GL3) and antibodies during the application of enzymatic replacement therapy.¹⁻³

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LITERATURE

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