Restrictive cardiomyopathy

Restrictive cardiomyopathy is a rare type of cardiomyopathy characterized by reduced compliance and increased ventricular myocardial stiffness. This leads to the development of diastolic dysfunction primarily of the left ventricle, without the presence of dilatation but of atrial dilatation. Ventricular systolic function may also be affected in later stages of the disease. Signs of chronic heart failure and conduction heart disorders predominate in the clinical picture. The etiology of restrictive cardiomyopathy is diverse and includes both genetic mutations and some infiltrative, storage and inflammatory diseases (e.g amyloidosis, hemochromatosis and endomyocardial fibrosis).

In differential diagnosis, restrictive cardiomyopathy must be distinguished from restrictive pericarditis. The issue of amyloidosis is addressed in a separate article on cardiac amyloidosis

Etiopathogenesis

In some cases, the cause is unknown and cardiomyopathy is therefore idiopathic. The most common causes of restrictive cardiomyopathy include genetic mutations, inflammatory diseases (endomyocardial fibrosis, Loeffler endocarditis), infiltrative and storage diseases (primary amyloidosis, sarcoidosis, hemochromatosis, Fabry disease), toxic damage of myocardium (from anthracyclines), and possible post radiation damage or carcinoid.

Genetic mutations

Genetic mutations associated with restrictive cardiomyopathy most commonly involve sarcomeric proteins. Typically, these are mutations such of cardiac troponin-I (TNNI3) or T (TNNI2), β-myosin heavy chain (MYH7) and desmin (DES). In this regard, restrictive cardiomyopathy is quite similar to the genetic mutations associated with hypertrophic cardiomyopathy. The inheritance of these mutations is usually autosomal dominant. [1]

Sarcoidosis

Sarcoidosis is a multi-organ granulomatous inflammatory disease of unknown cause. Autoimmune and genetic influences and various infectious and non-infectious agents are considered in the etiopathogenesis. Isolated cardiac sarcoidosis may also occur. Histopathologically, sarcoidosis is characterized by the finding of well-defined granulomas accompanied by more extensive fibrosis changes and a lower incidence of necrosis.

Hemochromatosis

Hemochromatosis is a disease associated with increased absorption of iron, the accumulation of which in various organs (especially the liver and heart) leads to the formation of free oxygen radicals and damage to mitochondrial function. This in turn leads to dysfunction of the organs.

Genetic mutations may be the cause. Secondary hemochromatosis may develop due to increased iron resorption due to thalassemia, sideroblastic anemia or excessive blood transfusions. In the early stages, like other types of restrictive cardiomyopathy, it manifests itself in the presence of diastolic dysfunction of the undilated left ventricle. Later, the disease progresses in left ventricular dilatation and systolic dysfunction of both ventricles. In addition, both supraventricular and ventricular arrhythmias may occur.

Epidemiology

The incidence of one form of amyloidosis, AL amyloidosis, in the population is approximately 3-9 per million inhabitants per year. For sarcoidosis in the Czech Republic the prevalence is 60 per 100.000 inhabitants. It is reported that the heart is affected in about 2-5% cases, but the finding of sarcoidosis, whether demonstrated by biopsy or magnetic resonance imaging of the heart, is confirmed in up to 25% of cases.

The clinical picture

Sarcoidosis

The clinical manifestation is very diverse, due to the multi-organ granulomatous involvement. Cardiac manifestations are dominated by heart failure in both ventricles with images of dilated or restrictive cardiomyopathy, AV conduction disorders and other arrhythmias.

Hemochromatosis

Clinical manifestations of hemochromatosis most often include hepatopathy (elevation of liver enzymes, hepatomegaly), hyperpigmentation (bronze skin), diabetes mellitus and other pathologies of the endocrine glands. Cardiac involvement occurs in up to 20% of cases and manifests itself in conduction defects and as restrictive or dilated cardiomyopathy.

Diagnosis

Sarcoidosis

Transmitter flow recording with restrictive left ventricular filling (E/A wave ratio more than 2)

Higher grade AV block, Tawar arm block, and ventricular tachycardia or fibrillation predominate from ECG findings. Decreased left ventricular ejection fraction is often described at ECHO. Magnetic resonance imaging of the heart plays an important role in the diagnosis, including examination of late gadolinium saturation. PET examination with evidence of accumulation of 18-fluorodeoxyglucose (18-FDG) in the area of inflammatory activity in the myocardium is also used. Endomyocardial biopsy is often indicated, but with low specificity due to the local occurrence of granulomatous inflammation in the myocardium.

Hemochromatosis

In hemochromatosis, magnetic resonance imaging (not just the heart) is of great importance, which has even reduced the importance of biopsy examination of the affected tissue to confirm the disease. The suspicion of this diagnosis arises from the fact that a patient with heart failure and conversion disorders develops another extracardiac disorder characteristic of hemochromatosis. However, an isolated form of cardiac hemochromatosis may be present. Elevated ferritin and transferrin levels also support suspicion . Genetic analysis and magnetic resonance imaging are at the forefront of diagnostics .

Treatment

Sarcoidosis

The main treatment is immunosuppressive therapy in monotherapy or in combination with other drugs. In terms of the possible risk of arrhythmic death, an ICD (implantable cardioverter-defibrillator) is implanted in indicated cases. Heart failure therapy consists mainly of ACE-inhibitors, β-blockers, diuretics, etc. In some cases, heart transplantation is necessary.

Hemochromatosis

In non-anemic patients, phlebotomy is the therapeutic option. Therapy chelates is considered in patients with significant anemia or low tolerance for phlebotomy hypotension or hypovolemia. Cardiac function usually improves with lower iron levels. Of the regime measures, it is important to limit the intake of alcohol and multivitamin preparations containing iron. Treatment with calcium channel antagonists and antioxidants is being investigated in clinical trials. In exceptional cases, heart or liver transplantation is required

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