Sarcomeric and Non-sarcomeric forms of hypertrophic cardiomyopathy

Hypertrophic Cardiomyopathy is one of the most common types of Cardiomyopathy with a frequency of **1:500** [1] [2] and also one of the most frequent causes of sudden death of young individuals and athletes . It is characterized by thickening (hypertrophy) of the myocardium without the presence of left ventricular dilatation, while other pathologies that could explain myocardial hypertrophy (e.g arterial hypertension , aortic valve stenosis , ischemic heart disease , etc.) are excluded .[1]

The most common cause of hypertrophic cardiomyopathy is mutations in genes encoding sarcomeric components of cardiomyocytes **.**[1] **However, the situation is complicated by the fact that a specific mutation is identified in only about 50% of patients, as well as by the occurrence of mutations in which their association with hypertrophic cardiomyopathy is still unclear (variation of unclear significance).**[1]**. Hypertrophic cardiomyopathy also occurs in some cases in metabolic and neuromuscular disorders (Fabry disease , Friedreich's ataxia).**[1][2]

The article on sarcomeric and non-sarcomeric forms of hypertrophic cardiomyopathy is a more detailed supplement to the subchapter etiopathogenesis in the article on hypertrophic cardiomyopathy . You can find more information about this disease in the relevant article.

Sarcomeric form

The mutations most commonly affect the genes for contractile myofilaments of cardiomyocyte sarcolemma. Mutations in the heavy chain beta myosin (MYH7) and myosin binding protein C (MYBPC3) genes are present in up to 30% of patients with hypertrophic cardiomyopathy who have been screened.[1] In about 5-7% [1] The mutation affects the genes for troponin T (*TNNT2*) or alpha-tropomyosin (*TPM1*).

Non-sarcomeric forms

These forms of hypertrophic cardiomyopathy make up about 10% of all forms of this cardiomyopathy and include various metabolic and storage diseases. The development of cardiomyopathy is an important prognostic factor in them.

Fabry disease

Fabry disease is a genetically determined disease linked to the **X chromosome** belonging to the group of storage lysosomal diseases.[3] The pathophysiological basis is a **defect in the activity of the enzyme alpha-galactosidase A** . This results in the **intracellular accumulation of glycosphingolipids** , which are stored, inter alia, in cardiomyocytes and in the walls of small blood vessels, leading to their insufficiency.[1][3] The disease is most common in men, but women's disability is no exception.[1][3] Early clinical manifestations include anhydrosis, acroparesthesia, and specific skin lesions.[1][3] Other manifestations are cerebrovascular, renal and cardiovascular disorders, which are dominated by more or less pronounced left ventricular hypertrophy.[1][3]

Danon Disease

This disease also belong among the lysosomal genetic disease linked to chromosome X. pathophysiological substantial disability gene is a **lysosomal membrane protein 2** (LAMP-2)..[3][4] The clinical picture of the disease is dominated by **cardiomyopathy, mental retardation and skeletal myopathy** .[3] The prognosis of patients, especially men, is very unfavorable. In women, it is usually less progressive. There is sometimes an extreme thickening of the left ventricular walls and a gradual development of ventricular dilatation, severe systolic heart failure and malignant arrhythmias.[3]

Friedreich's ataxia

The disease is genetically determined and has an autosomal recessive type of inheritance. The basis is in the vast majority of cases the amplification of genetic information on chromosome 9.[3] in the region encoding the protein **frataxin** , which plays a role in the assembly of Fe / S proteins in the respiratory chain. The main manifestation is a **neurodegenerative disease** with the development of ataxia, sensory disorders and eye problems. **Cardiovascular disease** is also typical, with the development of milder left ventricular hypertrophy, as well as heart failure , which is the predominant cause of patient mortality.[3]

Other

There are a variety of other etiologies called. Nesarkomerické form of hypertrophic cardiomyopathy involving e.g. Pompe disease, **PRKAG2 syndrome** (defect in the gene for adenosine monosfátem activated protein active in energy processes cells dominate the clinical picture conduction disorders), **mitochondrial cardiomyopathy**

(defective oxidative phosphorylation, multiorgan disability, myocardial infarction with a very poor prognosis), **mucopolysaccharidosis , glycogenosis** , etc. In general, the diagnosis of these forms is based on **genetic** , laboratory or biopsy examination of patients. Treatment is usually supportive and symptomatic.[3][4]

Links

Related articles

- Hypertrophic Cardiomyopathy

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