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Current Issues of Diagnostics and Treatment of Patients with Dilated Cardiomyopathy

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Abstract: Dilated cardiomyopathy (DCMP) is a diffuse myocardial lesion with dilatation (dilation) of the heart cavities and a sharp decrease in its contractile function. DCMP is a common pathology and occurs in the world with a frequency of 5-8 cases per 100000 population. In particular, 36 cases per 100000 population are registered in the USA. Male persons are more often affected (2:10), clinical manifestations of the disease occur at the age of 10 to 30 years. The incidence of DCMP is 2.5 times higher in negroid males than in Caucasians and females. The prognosis of the disease becomes unfavorable when patients have heart failure, which in turn makes this pathology threatening to the life of the patient. The lethality rate ranges from 25% to 30% per year. In 5 years from the onset of the disease, 60-75% of patients with DCMP practically die. The classification of DCMP approved by the American Heart Association in 2006 is most often used in physician practice. In this classification, the AHA proposed the definition of CMP as "a heterogeneous group of myocardial diseases associated with mechanical or electrical dysfunction, which usually manifest with inadequate hypertrophy or dilatation and arise from a variety of causes that are often genetic. CMP is confined to the heart or is part of generalized systemic disorders, always leading to cardiovascular death or progression of heart failure.".

Keywords: Heart failure, genetic factors, arterial hypertension, ischemic heart disease, phenotypic classes.

Introduction. Cardiovascular diseases are the most frequent cause of morbidity and mortality worldwide. The continuous accumulation of information on the pathogenesis of cardiovascular diseases has led to the realization of how significant a role genetic factors play in their development. Today there are practically no diseases left, in the formation of which no hereditary component has been established. The most frequent diseases coronary heart disease (CHD), atherosclerosis and arterial hypertension (AH) are multifactorial. Heredity and environment contribute approximately equally to the formation of the clinical phenotype in these diseases [1,5,8]. For each disease, there is a sufficiently large number of genes whose different allelic

forms influence the probability of disease development, the rate of progression, and the severity of clinical symptoms. As a rule, predisposition genes are those genes whose protein products are directly or indirectly involved in the pathogenesis of the disease [2,10,14].

In addition to multifactorial, there is a large number of monogenic diseases, for the development of which the presence of a mutation in a single gene is sufficient. Currently, about 2.5 thousand monogenic hereditary syndromes are described, in which the heart is involved in the pathologic process [4,6,17]. About a hundred hereditary diseases are known, in which heart and vascular lesions are leading in the clinical picture. Even from the general structure of such classical multifactorial diseases as CHD and AH, an increasing number of monogenic forms are being isolated, which are inherited according to the Mendelian type [8,13,21].

Recently, ideas about the etiology of many cardiovascular diseases have undergone a significant evolution. This is particularly true for conditions for which no convincing data have been obtained that unequivocally testify in favor of infectious and/or inflammatory processes underlying them [16,22,24]. Typically, these conditions were described as "idiopathic". The rapid development of modern genetic methods has made it possible in many cases to resolve the question of the hereditary nature of the disease, to establish the primary biochemical defect, to develop DNA diagnostics, and to seek approaches to etiologic therapy. Cardiomyopathies are an extensive class of diseases characterized by structural changes in the myocardium. There are 3 phenotypic classes of cardiomyopathies: hypertrophic, dilative, and restrictive. Each class is characterized by its morphological, physiological, and clinical endpoints, reflecting different pathogenetic mechanisms leading to the disease [19,20]. These diseases are the most frequent cause of cardiogenic sudden death (CVD) in the young. In some cases, the cause of myocardial dysfunction can be elucidated, but more often the etiology of the disease remains unknown. Recently, new data have been obtained that expand our understanding of gene and protein changes underlying cardiomyopathies, a significant proportion of which are monogenic inherited diseases [2,4,7]

In 1995, the World Health Organization proposed a classification that divides cardiomyopathies into dilated, hypertrophic, restrictive, specific (metabolic, inflammatory, ischemic, valve, diabetic, alcoholic cardiomyopathy), arrhythmogenic right ventricular cardiomyopathy and unclassifiable cardiomyopathies (fibroelastosis, etc.).

Dilated cardiomyopathy (DCMP) is the most common and occurs in all countries of the world. Until recently, disagreements on the definition of cardiomyopathies and the lack of clear diagnostic criteria for DCMP cause difficulties in epidemiological studies in this area, and therefore, to date, there are no accurate data on the prevalence of DCMP and morbidity of the population, since most studies are retrospective and based on the analysis of only accurately established diagnoses without taking into account the early stages of the disease [12,21].

The specific weight of DCMP among other cardiomyopathies is 60%. In this respect, the statement of N.M.Mukharlyamov has not lost its significance: "Serious epidemiologic studies are needed to find out the true state of affairs. The importance of this problem is emphasized by the fact that patients with DCMP become permanently disabled faster than in other non-coronary myocardial diseases.

In DCMP, the main function of the heart muscle - contractile - is disturbed. It is weakened. In this case, the heart chambers are diffusely (in all directions) dilated. Much less often, only one of the heart chambers is affected. According to statistics, dilated cardiomyopathies account for 9% of all cases of heart failure. The incidence of dilated cardiomyopathy is from 3 to 10 cases per 100,000 people [1,3].

The causes of this type of cardiomyopathy are not fully understood. The hereditary nature of the disease is thought to be possible, as it is not uncommon to find several cases of the disease in close relatives. Perhaps contribute to the disease contribute to disorders in the immune system, but confirmation of this has not been found in all patients. Many researchers are convinced of the

viral origin of dilated cardiomyopathy, but no one has yet been able to isolate the virus that causes it. The disease usually develops gradually. Sometimes a person doesn't notice the disease for several years. And dilated contours of the left ventricle are detected accidentally during X-ray examination [5,6].

The first signs of the disease: they are not specific, i.e. it is impossible to immediately guess that it is cardiomyopathy. This is increased fatigue with physical activity, weakness. Later, there is shortness of breath, first on exertion then it increases and in the midst of the disease appears cardiac asthma. Very often there are pains in the heart of different character: aching, stabbing, squeezing. But angina pectoris is rare. From the fact that the internal chambers of the heart are enlarged and for other reasons not completely clarified often in the heart cavity and in large veins there are thrombi. These clots can break off and with the flow of blood into the lungs. Then there is a dangerous complication - pulmonary embolism (TELA). In 33% of cases of sudden death, TELA is the culprit [11,13].

Examination of the patient reveals cold skin, dilated heart borders. Listening reveals a systolic murmur, various rhythm disturbances. With a significant expansion of the left ventricular cavity and a decrease in the volume of blood ejected by it due to weakness of the heart muscle - the patient's prognosis.

The diagnosis of dilated cardiomyopathy is established on the basis of X-ray examination, electrocardiogram, angiography. But the main method of diagnosing cardiomyopathy is echocardiography. Echocardiographic study reveals enlargement of the left ventricle, enlargement of the other heart chambers, normal or reduced thickness of the left ventricular wall is determined. If dilated cardiomyopathy is suspected, echocardiography is mandatory. Already at the first examination, the diagnosis is established. Subsequent examinations are carried out to monitor treatment and the condition of the heart. A biopsy may be performed. Examination under a microscope of a small piece of heart muscle. Extensive changes are found in it - dead heart muscle cells, replacement of muscle tissue by connective tissue (sclerosis of heart muscle) [12,15].

At present, DCMPs of unidentified etiology, the so-called idiopathic DCMPs, are of great interest. Numerous studies of the last decade are focused on the study of their etiopathogenesis, and in this aspect the hypotheses of chronic viral infection, autoimmune influence and genetic determinism are considered [14,17]. Molecular biological technologies (including polymerase chain reaction) have become available, which have revealed the role of enteroviruses, particularly coxsackieviruses B group, in the pathogenesis of DCMP. Despite the high sensitivity and specificity of these techniques, the frequency of virus detection varies from 0 to 40%. In children aged 1 day to 19 years with rapidly developing left ventricular dilatation and dysfunction, the viral genome was detected in 68% of cases, with enterovirus in 30%, adenovirus in 58%, herpesvirus in 8%, and cytomegalovirus in 4.

As is known, when studying hereditary traits, one can use the genealogical method, in the broad sense of the word - the study of pedigrees. Genealogical method is a method of pedigrees, i.e. tracing a disease or a family tree with indication of the type of kinship between the members of the pedigree. In medical genetics, this method is often called clinical-genealogical method because it is the study of pathologic traits in a family using clinical examination techniques. When compiling a pedigree, the initial person is the proband whose pedigree is being studied. This is usually the patient or carrier of the trait whose inheritance is to be studied. The pedigree may be collected on one or several traits. In the latter case, the linked nature of their inheritance can be revealed, which is used in the preparation of chromosome maps. Depending on the purpose of research pedigree can be complete or limited. It is necessary all the same to aspire to the most complete drawing up of pedigrees, and for this purpose data not less than about 3-4 generations of family of the proband are necessary. Compilation of pedigree is accompanied by a brief record about each member of pedigree with exact characterization of his relation to the proband (legend of pedigree). It is also necessary to note examined and unexamined for the

presence of the investigated trait. Obtaining information about relatives is not an easy task. Patients are often unaware of their relatives' diseases or provide incorrect information. To obtain more accurate information, questionnaires are used, and sometimes complete clinical and laboratory genetic examination of relatives. When analyzing pedigrees, first of all it is necessary to establish the hereditary character of a trait. If one and the same pathologic trait occurs several times (over several generations) in a pedigree, it is likely to be hereditary in nature. Next, it is necessary to establish the type of inheritance (autosomal dominant, autosomal recessive, X-linked dominant or recessive, Y-linked). Determining the type of inheritance in a particular pedigree is a serious genetic problem, and the physician must have specialized training to solve it. Genealogical method belongs to the most universal methods in human genetics. It is widely used in solving theoretical applied problems, to establish the hereditary nature of a trait, in determining the type of gene inheritance. Empirical observations on pedigrees, in which the inheritance of pathological traits was noted, have been known for a long time. The application of family analysis to the study of human pathology in the XVIII-XIX centuries can be considered as a prerequisite for the formation of the genealogical method. The essence of the genealogical method is reduced to the elucidation of kinship relations and tracing of a trait or disease among close and distant and indirect relatives. Technically, it consists of two stages, compilation of pedigrees and genealogical analysis. Collection of information about the family begins with a proband, which is the person who first came to the researcher's attention. Most often it is a patient or a carrier of the trait under study, but not necessarily. Children of one parental couple are called sibs (brothers - sisters). Family in the narrow sense refers to a parental couple and their children, but sometimes also to a wider circle of blood relatives, although in the latter case it is better to use the term "clan".

Depending on the purpose of the study, a pedigree may be complete or limited. It is desirable, of course, to strive for the most complete compilation of pedigrees along ascending, descending, and lateral lines. This task is not as easy as it may seem at first glance. The more generations involved in a pedigree, the more extensive it is. This entails inaccuracy of the information obtained and, consequently, inaccuracy of the pedigree as a whole. Sometimes people do not even know the number of their cousins, let alone any traits in them or their children. The compilation of a pedigree is accompanied by a brief record of each member of the pedigree with a precise characterization of his kinship in relation to the proband.

Conclusions: Thus; in the future, a graphic representation of the pedigree is prepared for clarity (or for publications). The first task in pedigree analysis is to establish the hereditary nature of a trait. If one and the same trait (or disease) occurs several times in a pedigree, it is possible that it is hereditary in nature. Thus, the role of genetic factors is undisputed in the etiopathogenesis of idiopathic DCMP. However, in the literature available to us in the literature, there are almost no data on the study of the role of genetic factors in individuals of local ethnicity. Studies in this area are necessary to assess the genetic risk of disease development. And it is understandable, as there is an increase in morbidity and mortality from this pathology.

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