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Current Understanding of Minor Cardiac Anomalies in Children with Undifferentiated Connective Tissue Dysplasia

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Abstract This review presents current data on minor cardiac anomalies (MCAs) in children, which include congenital or early acquired morphological features of the cardiovascular system that do not lead to significant hemodynamic disturbances. The main types of MCAs are discussed, including mitral valve prolapse, patent foramen ovale, accessory chordae, and others. Information is provided on the frequency of detection and factors influencing their formation, including hereditary predisposition and connective tissue dysplasia. Clinical manifestations are described, ranging from asymptomatic courses to mild cardialgia and autonomic disorders. Particular attention is paid to diagnostic possibilities, primarily echocardiography, as well as the role of electrocardiography and other research methods. The necessity of an individualized approach to the management and dynamic follow-up of children with MCAs is emphasized: most cases do not require active treatment; however, when several anomalies are combined, in cases of significant valvular regurgitation or rhythm disturbances, medical correction and closer monitoring may be required. The review concludes with a discussion of prevention and healthy lifestyle issues, as well as prospects for studying molecular-genetic mechanisms and developing personalized management recommendations.

Keywords Minor cardiac anomalies, Mitral valve prolapse, Patent foramen ovale, Accessory chordae, Children, Connective tissue dysplasia, Diagnostics, Autonomic disorders, Prevention, Echocardiography

1. Introduction

Minor cardiac anomalies (MCAs) in children represent a group of morphological changes in the cardiovascular system that are not accompanied by significant hemodynamic disturbances but may influence prognosis, physical development, and the overall health status of the child in the long term [1,2,5,6]. The term "minor cardiac anomalies" (sometimes referred to as "minor developmental anomalies of the heart") encompasses various types of minor defects in the anatomical structure of the valvular apparatus, atria, ventricles, major vessels, and their combinations. As imaging techniques such as echocardiography and cardiac MRI continue to improve, the relevance of studying this pathology is increasing because the diagnostic detection rate of MCAs is growing, requiring appropriate approaches to the management of such children [3,7].

In recent decades, this group has included several borderline conditions that may be considered variants of normal but acquire clinical significance under certain circumstances (physical stress, growth, concomitant diseases). Although MCAs generally do not lead to severe circulatory disorders, some variants may serve as markers of genetic or dysplastic pathology and may increase the risk of arrhythmias or other complications [4,8].

The aim of this review article is to summarize and systematize the current state of knowledge regarding minor cardiac anomalies in children, including their definition, classification, prevalence, etiological factors, pathogenesis, clinical presentation, diagnostic methods, management principles, and dynamic follow-up.

Definition and Classification of Minor Cardiac Anomalies

Minor cardiac anomalies are understood as congenital or early postnatal structural and anatomical features that do not lead to pronounced hemodynamic disturbances and usually do not require surgical correction [5,11]. Many MCAs may be regarded as individual morphological variants without clinically significant impact; however, in some situations they may be associated with pathological processes or become complicated when combined with other cardiovascular changes.

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Several classifications of MCAs exist, differing in scope and inclusion criteria. One of the most common is the functional-anatomical classification, according to which MCAs are divided into:

1. **Anomalies of the valvular apparatus:** mitral valve prolapse (MVP), tricuspid valve prolapse, accessory and abnormal chordae in the ventricles, abnormal attachment of chordae.
2. **Interatrial septal anomalies:** patent foramen ovale (PFO).
3. **Anomalies of the location and structure of major vessels:** elongated aortic arch, tortuous course of the subclavian arteries without hemodynamic disturbances.
4. **Myocardial tissue anomalies:** excessive trabeculation (incomplete noncompaction), hypertrophy of small myocardial regions without signs of cardiomyopathy.

A separate group may include MCAs associated with hereditary connective tissue dysplasia syndromes (for example, Marfan syndrome), in which multiple manifestations of dysplasia are detected, including elongated limbs, chest deformities, changes in the valvular apparatus and major vessels, as well as MCAs [6].

Epidemiology

Accurate data on the prevalence of MCAs in the pediatric population are difficult to obtain because of heterogeneous diagnostic criteria. It is believed that signs of MCAs are detected in 5–10% of newborns, and the detection rate directly depends on the quality and availability of ultrasound diagnostic methods [7]. Among older children, the prevalence of MCAs ranges from 4–12%. Some authors note that due to the widespread implementation of screening echocardiography, the number of diagnosed MCA cases is steadily increasing [8,10,20].

Mitral valve prolapse is the most common minor cardiac anomaly in children. According to some data, the prevalence of MVP in adolescents may reach 2–6%, although only one-third of these cases demonstrate clinical manifestations or clinically significant signs [9,12]. The next most frequent MCA is patent foramen ovale, which can be detected in 20–25% of healthy individuals, and in some of them it remains open throughout life without any complications [10,22].

Etiology and Pathogenesis

The formation of minor cardiac anomalies is generally associated with the peculiarities of embryogenesis and postnatal development of the cardiovascular system. The following factors and mechanisms are most commonly identified:

1. **Hereditary factors.** Predisposition to dysplastic connective tissue changes, one of the common causes of such conditions, may be due to genetic mutations (for example, mutations of the FBN1 gene in Marfan syndrome) [6,11].
2. **Disturbances of embryonic development.** Any teratogenic factors (maternal infectious diseases, medications with potentially negative effects on the fetus, intrauterine hypoxia, etc.) may lead to underdevelopment or abnormal formation of individual cardiac structures [12,15].
3. **Connective tissue dysplasia.** This term implies various structural and functional changes in connective tissue elements affecting the valvular apparatus, vessels, and stromal myocardial components [13,17].

The pathogenesis of MCAs often involves mechanisms of excessive connective tissue extensibility, changes in collagen synthesis, and alterations in neurohumoral regulation of the cardiovascular system. For example, in mitral valve prolapse, elongation of the valve leaflets may occur due to an abnormal ratio of collagen types I and III, contributing to excessive valve mobility [9,14,16].

Clinical Manifestations

Most MCAs are characterized by an asymptomatic course or minimal nonspecific symptoms that often do not attract the attention of parents or pediatricians. However, when several minor anomalies coexist, especially against the background of connective tissue dysplasia, the clinical picture may become more pronounced. Possible manifestations include:

1. **Cardialgia.** Mild stabbing or pressing pain in the cardiac region, more often associated with autonomic nervous system imbalance than with organic myocardial pathology [14,19].
2. **Cardiac rhythm disturbances.** Extrasystole, sinus tachycardia or bradycardia, supraventricular tachyarrhythmias, and in rare cases ventricular rhythm disturbances [3,18,21].
3. **Autonomic dysfunction.** Complaints of sweating, labile blood pressure, dizziness, and fainting upon rapid standing [15].
4. **Syndromes associated with connective tissue dysplasia.** Such patients may demonstrate a characteristic phenotype: tall stature, elongated limbs, arachnodactyly, musculoskeletal disorders (scoliosis, flat feet, chest deformities) [16].

MCAs are often detected incidentally during routine examinations or investigations for other diseases. During physical examination, the physician may identify functional murmurs (for example, a systolic click in MVP), which do not always correlate with the severity of pathology. Such children frequently demonstrate unstable pulse and blood pressure associated with peculiarities of autonomic regulation.

Diagnostic Methods

The most informative, accessible, and safe method for diagnosing minor cardiac anomalies is echocardiography (EchoCG) [3,17,22]. It enables the detection of valvular prolapse, accessory chordae, patent foramen ovale, assessment of cardiac chamber dimensions, myocardial contractile function, and much more.

However, it is important to note that a critical interpretation of echocardiographic findings is mandatory. Minimal valvular prolapse or insignificant accessory

chordae identified during EchoCG should be interpreted in the context of the clinical picture and the overall assessment of the child's condition.

Other methods include:

1. **Electrocardiography (ECG).** Helps detect possible rhythm disturbances and signs of overload in specific cardiac chambers. In MCAs, ECG changes may be absent or minimal; however, long-term (24-hour) Holter ECG monitoring is important for excluding arrhythmias.
2. **Exercise testing (bicycle ergometry, treadmill test).** Allows identification of latent rhythm disturbances or impaired adaptation of the cardiovascular system to physical activity.
3. **Cardiac MRI.** May be prescribed in complex cases or to clarify anatomy. It is more commonly used for evaluating structures difficult to visualize by ultrasound or when combined developmental defects are suspected.
4. **Doppler examination.** Assesses the speed and direction of blood flow in the cardiac chambers and major vessels and helps evaluate the degree of regurgitation in valvular prolapse and other anomalies.

Thus, the diagnosis of MCAs is based on a comprehensive approach in which noninvasive methods are prioritized. MCAs are often detected incidentally in children, and in such cases the choice of further management depends on the results of additional examinations, particularly the degree of regurgitation according to Doppler studies and the presence or absence of rhythm disturbances.

Mitral Valve Prolapse as the Most Common Minor Cardiac Anomaly

Special attention should be paid to mitral valve prolapse because it is one of the most extensively studied forms of MCA in children [9,13]. MVP may be primary (associated with connective tissue dysplasia) or secondary (arising as a consequence of other heart diseases such as myocarditis or rheumatism).

The degree of prolapse is usually classified according to the depth of leaflet displacement into the left atrial cavity:

- Grade I: less than 5 mm;
- Grade II: 5–9 mm;
- Grade III: more than 9 mm.

However, the most important prognostic factor is not so much the depth of prolapse as the degree of mitral regurgitation. Most children with Grade I–II MVP without significant regurgitation have no manifestations or complications. At the same time, the presence of moderate or severe regurgitation increases the risk of arrhythmias, progressive dilation of the left chambers, and heart failure development [1].

The question of the need for drug therapy in pediatric MVP is resolved individually. In asymptomatic cases, preventive observation is sufficient (echocardiography once a year, monitoring of blood pressure and ECG). If MVP is accompanied by rhythm disturbances, significant regurgitation, or autonomic disorders, beta-blockers, antiarrhythmic agents, or other medications may be prescribed [3,9].

Patent Foramen Ovale

Patent foramen ovale represents a residual opening in the interatrial septum that persists after birth. Normally, the foramen ovale functions during intrauterine life, providing blood redistribution bypassing the lungs. After birth, when the lungs expand, pressure in the left atrium increases, and the valve of the foramen ovale normally closes. In some children, complete anatomical obliteration may not occur, leading to persistent communication between the atria [10,24].

Most individuals with PFO (according to some data, up to 25% of the population) have no clinical manifestations. However, it is believed that in a small proportion of patients, PFO may increase the risk of paradoxical embolism and cryptogenic stroke, especially in the presence of concomitant risk factors such as deep vein thrombosis of the lower extremities [18,23]. In childhood, such complications are extremely rare.

The decision regarding the necessity of PFO closure in children is made individually, usually in the presence of obvious blood shunting and risk of paradoxical embolism. In many cases, PFO remains merely an anatomical finding that does not require intervention [10,18].

Other Minor Cardiac Anomalies

Less common but clinically significant MCAs include:

1. **Accessory and abnormal left ventricular chordae.** Frequently found in the population and usually without clinical significance [5,25]. However, transverse chordae may produce turbulent blood flow and sometimes provoke rhythm disturbances.
2. **Thickening or hypertrophy of individual myocardial regions** without signs of outflow tract obstruction. This may occur in connective tissue dysplasia and does not always indicate hypertrophic cardiomyopathy.
3. **Abnormal ventricular trabeculae.** Often incidental findings that do not affect hemodynamics.
4. **Tortuous vascular course.** Tortuous or loop-shaped segments of the aorta, subclavian arteries, pulmonary arteries, etc., usually do not require treatment in the absence of clinical symptoms [4].

Management Approaches and Dynamic Follow-Up

Management of children with MCAs should be based on a comprehensive assessment of the clinical picture, the presence or absence of symptoms, and the risk of complications. Most MCAs do not require specific therapy; however, systematic observation of such children is important because some cardiac features may become clinically apparent during periods of active growth or hormonal changes in adolescence [7].

The main management approaches include:

1. **Regular follow-up by a cardiologist:** frequency of visits (1–2 times per year) with EchoCG, ECG, and if necessary, 24-hour ECG monitoring.
2. **Assessment of physical activity:** moderate physical activity is generally not contraindicated for children with MCAs. On the contrary, dosed exercise may improve cardiovascular function. However, in cases of pronounced prolapse with regurgitation or frequent arrhythmias, the type and regimen of exercise are determined individually [2,15].
3. **Correction of autonomic disorders:** nonpharmacological methods are often indicated (physiotherapy, massage, therapeutic exercise, normalization of sleep and rest schedules). In cases of severe complaints such as tachycardia, extrasystole, or autonomic crises, medical therapy (beta-blockers, sedatives) may be prescribed [9,15].
4. **Prevention of infective endocarditis:** in some cases (especially valvular prolapse with regurgitation), physicians recommend preventive measures during dental procedures and other interventions [19].
5. **Drug therapy:** in severe forms of MVP or other MCAs with hemodynamic disturbances, specific treatment aimed at reducing regurgitation, improving contractility, and correcting arrhythmias may be prescribed (beta-blockers, antiarrhythmic drugs).
6. **Surgical intervention:** an extremely rare option for children with MCAs and applied only in cases of pronounced functional impairment and ineffective conservative management.

The main task of pediatricians and cardiologists is the timely identification of cases in which MCAs extend beyond the boundaries of "minor" anomalies or become markers of a more serious condition (genetic or progressive dysplastic process). In most situations, dynamic observation and maintenance of a healthy lifestyle are sufficient.

MCAs as a Manifestation of Connective Tissue Dysplasia

A separate issue is the problem of associated connective tissue disorders in children with MCAs. In some cases, the presence of several MCAs (for example, MVP, accessory chordae, PFO) combined with signs of generalized dysplasia (joint hypermobility, posture disorders, pathology of the dentofacial system) may indicate connective tissue dysplasia syndromes such as Marfan syndrome or Ehlers–Danlos syndrome [6,16].

Such patients typically develop an entire complex of clinical manifestations: cardiovascular (aortic aneurysm, valvular insufficiency), orthopedic (scoliosis, arachnodactyly), ophthalmologic (lens subluxation), and dermatological (excessive skin extensibility). If systemic dysplasia is suspected, consultation with a geneticist and comprehensive diagnostics including molecular-genetic testing are necessary [11].

Management of such patients requires a multidisciplinary approach: in addition to a cardiologist, the team may include an orthopedist, ophthalmologist, neurologist, physiotherapist, and other specialists.

Prognosis and Possible Complications

Although minor cardiac anomalies are generally considered benign conditions, they are not always completely harmless. The risk of complications depends on the type and severity of the anomaly. Possible adverse outcomes include:

1. **Progressive valvular regurgitation** (for example, in MVP). This may lead to an increase in regurgitation volume and remodeling of the left heart chambers [1].
2. **Rhythm disturbances.** Some patients with MCAs (especially with combinations of several anomalies) may develop a tendency toward extrasystole and paroxysmal tachycardia [20].
3. **Infective endocarditis.** The theoretical risk in valvular prolapse with regurgitation is slightly higher than in the general population, although the absolute probability remains low [19].
4. **Paradoxical embolism.** In the presence of PFO and certain concomitant factors (for example, deep vein thrombosis), embolism into the systemic circulation may occur. In childhood, such cases are extremely rare [18].

Overall, with adequate follow-up and timely detection of complications, the prognosis for children with MCAs is favorable: most of them live full lives and participate in physical education and sports without restrictions. Reducing psychoemotional stress is also important: excessive parental anxiety and overemphasis on regular examinations may negatively affect the child and contribute to the development of "cardiophobia."

Prevention and Healthy Lifestyle

The issue of prevention regarding minor cardiac anomalies is multifaceted. On the one hand, MCAs often have a genetic or intrauterine basis, complicating specific prevention. On the other hand, measures aimed at strengthening overall health can improve tolerance to possible MCA manifestations and reduce the risk of complications.

General preventive measures include:

1. **Balanced nutrition:** a diet with sufficient protein, vitamins, and micronutrients.
2. **Physical activity:** regular participation in accessible sports (swimming, walking, cycling) provided there are no contraindications.
3. **Prevention of infections:** timely vaccinations and hygiene measures; in cases of MVP with regurgitation, antibiotic prophylaxis may be recommended for certain procedures.
4. **Stress control:** psychoemotional well-being plays an important role in preventing autonomic disorders and rhythm disturbances.
5. **Regular preventive examinations:** dispensary observation including assessment of anthropometric parameters, blood pressure, and cardiac auscultation.

For children with already identified MCAs, individualized selection of physical activity levels is important, taking into

account the results of instrumental examinations. In most cases, moderate activity is not only permissible but also contributes to cardiovascular conditioning.

2. Conclusions

Minor cardiac anomalies in children represent a broad group of congenital or early acquired structural features of the heart that, in most cases, do not lead to significant hemodynamic disturbances and do not require surgical treatment. The main significance of MCAs lies in their potential association with connective tissue dysplasia and possible complications such as rhythm disturbances, progressive regurgitation, and the risk of infective endocarditis.

The modern approach to managing children with MCAs involves comprehensive diagnostics using noninvasive methods (EchoCG, ECG, 24-hour ECG monitoring) and dynamic observation aimed at timely detection of clinically significant changes. Most patients do not require drug therapy in the absence of rhythm disturbances or significant regurgitation. An important role is assigned to nonpharmacological preventive measures, rational daily regimen, and physical activity.

Future studies will focus on clarifying the molecular-genetic mechanisms underlying MCA formation, developing personalized approaches to management and prevention of possible complications, and optimizing clinical recommendations that take into account the individual characteristics of the growing organism.

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