CLINICAL AND INSTRUMENTAL CHANGES IN CONNECTIVE TISSUE DYSPLASIA SYNDROME. CLINICAL CASE

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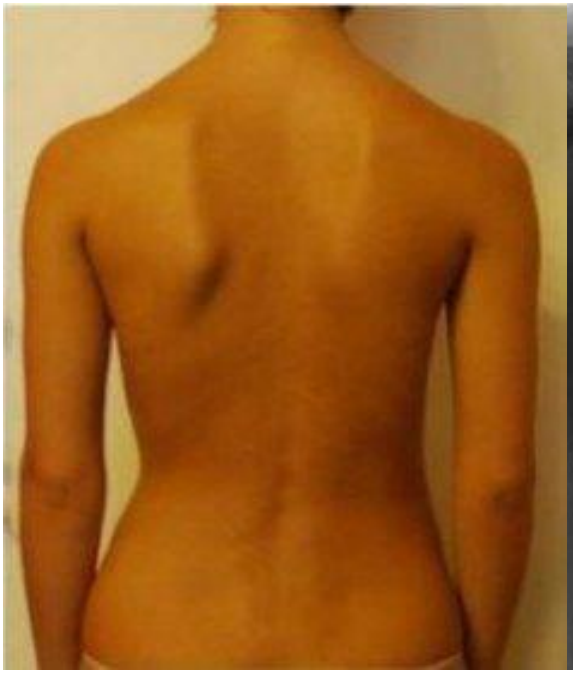
Abstract. *Patient N., 16 years old, was admitted to the clinic with complaints of rapid heartbeat, interruptions in heart function, stabbing pain in the heart area, and a feeling of lack of air. During the examination, electrocardiographic (ECG) changes were revealed in the form of horizontal ST segment depression of up to 3 mm, on a stress ECG without clinical manifestations of coronary and heart failure. During daily ECG monitoring at the height of the load, T wave inversion and ST segment depression occurred. There was no history of high blood pressure. A comprehensive examination revealed signs of connective tissue dysplasia syndrome (CTDS). The patient's constitution had curvatures of the chest and moderate kyphoscoliosis of the cervicothoracic spine. An echocardiographic (EchoCG) examination revealed signs of mitral and tricuspid valve prolapse with grade I regurgitation, accessory chords of the left ventricle, dilatation of the aortic root up to 3.6 cm, ejection fraction was 63%, myocardial mass index - 95 g/m^2. When monitoring the ECG, signs of sinus node dysfunction were recorded (rhythm reduction to 39 beats per minute at night). According to coronary angiography, there are no stenoses in the coronary arteries. Laboratory tests and other instrumental studies revealed no pathological abnormalities.*

Keywords: *dysplasia, connective tissue, electrocardiographic changes, depression of the ST segment, dysplastic heart, child.*

Introduction. Over the past decade, connective tissue dysplasia in children has attracted the attention of doctors of various specialties – cardiologists, pulmonologists, pediatricians, gastroenterologists, ultrasound diagnostics doctors. This is due to both an increase in the prevalence and improved diagnosis of connective tissue dysplasia in the population, and the risk of possible complications [1]. Connective tissue dysplasia syndrome is characterized by damage to the cardiovascular system in the form of various anomalies in the structure of the chambers of the heart, valvular apparatus, dilation of the aortic root, rhythm disturbances and cardiac conduction [2-4]. These disorders are manifested in the form of sinus tachycardia, arrhythmia, and sinus bradycardia. Changes on the ECG require dynamic monitoring, as they can be the initial manifestations of the emerging pathology [9].

A clinical case. Patient N., 16 years old, a student of secondary school, was admitted to the clinic with complaints of palpitations, heart failure, pain in the area of the heart of a stabbing nature, a feeling of lack of air. The girl has been practicing acrobatic gymnastics since the age of 4. From the anamnesis, tachycardia was noted from childhood. At the age of 10, fainting first occurred, and she was hospitalized for it. The patient was diagnosed with vegetative vascular dystonia syndrome. She was treated with sedatives and metabolic drugs. Deterioration of the condition during the last three years: palpitations persisted, interruptions in the heart function. She was treated with B-blockers and tranquilizers without any significant effect. Upon admission, the

condition is relatively satisfactory. On objective examination: a child of asthenic physique, satisfactory nutrition, body mass index according to Ketla - 22. Asymmetry was noted in the chest: left-sided bulge and moderate thoracic kyphoscoliosis of the spine (fig. 1, 2).



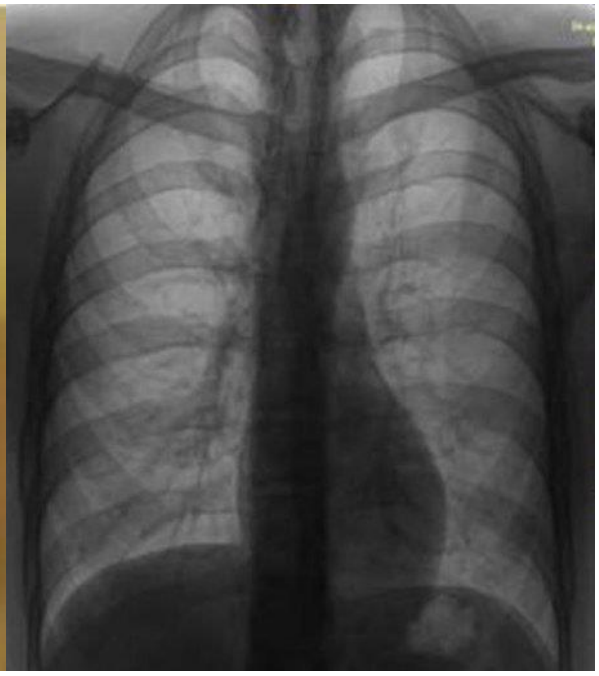


Fig. 1. The constitution of patient N., 16 years old ***Fig. 2. Chest X-ray***

The skin is pale and dry. Pain on palpation of paravertebral points in the lower thoracic spine. BR 20 in min. In the lungs, breathing is vesicular, there is no wheezing. Heart sounds are rhythmic, and a systolic click is heard at the apex. Pathological noises in the precordial region, on the vessels of the neck and abdominal aorta were not recorded. Heart rate 105 in min. Blood pressure is 120/70 mm Hg. The liver and spleen are not enlarged, the lower pole of the right kidney is palpated.

Results of the conducted research.

ECG registration at admission: sinus rhythm, heart rate (HR) 105 beats /min.; P-0.10 ms; QRS-0.36 ms; elongation and depression of the ST segment in I, II, AVF, VV4 – VV6 leads up to 0.5 mm; the T-wave is smoothed in all leads, SV1 > SV2. Conclusion: enlargement of the left atrium and moderate load on the left ventricle. Diffuse myocardial changes (fig. 3).

Veloergometry (load test): achieved a load of 85% heart rate (165 beats/min.). At the 3rd stage of the load (150 W), a horizontal depression of the ST segment to 1.0–3.0 mm appeared in combination with (-/+) T wave in II, III, aVF, V3 – V6 leads. There are no complaints, the ECG recovered at 3 minutes of rest, the type of blood pressure response to exercise is dystonic, and the tolerance to physical activity is high. The load test was evaluated as positive (fig. 4).

Holter ECG monitoring (HM ECG): a sinus rhythm with a heart rate of 39-135 beats/min. was recorded. The average heart rate during the day was 63 beats/min.; the average heart rate at night was 47 beats/min., and the minimum heart rate at night was 39 beats/min. The circadian index is 1,34. 1767 pauses of more than 1,5 seconds were registered: during the day - 3 (less than 1 per hour), at night – 1764 (less than 196 per hour). The maximum pause of up to 1832 msec. was at 02:42:37 hours. Three physical activities were performed with a maximum heart rate of up to 135 beats/min. Depression of the ST segment in the I and II standard leads up to 3

mm and inversion of the T wave in the III lead were recorded against the background of a maximum increase in heart rate (fig. 5).

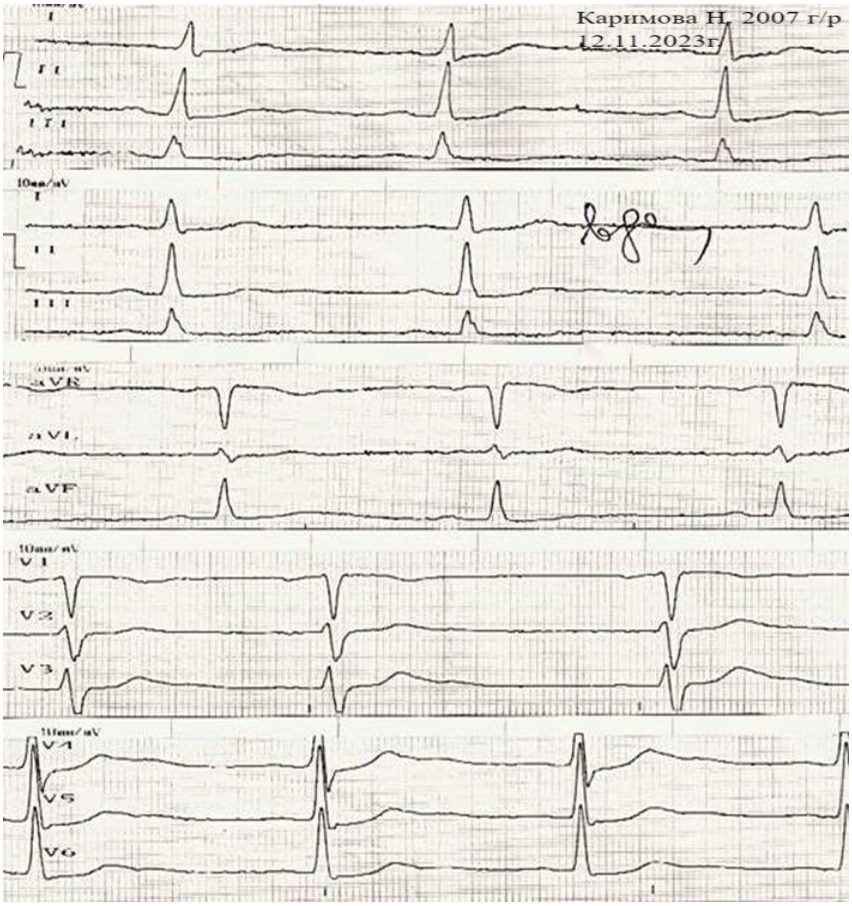


Fig. 3. Resting ECG of patient N., 16 years old

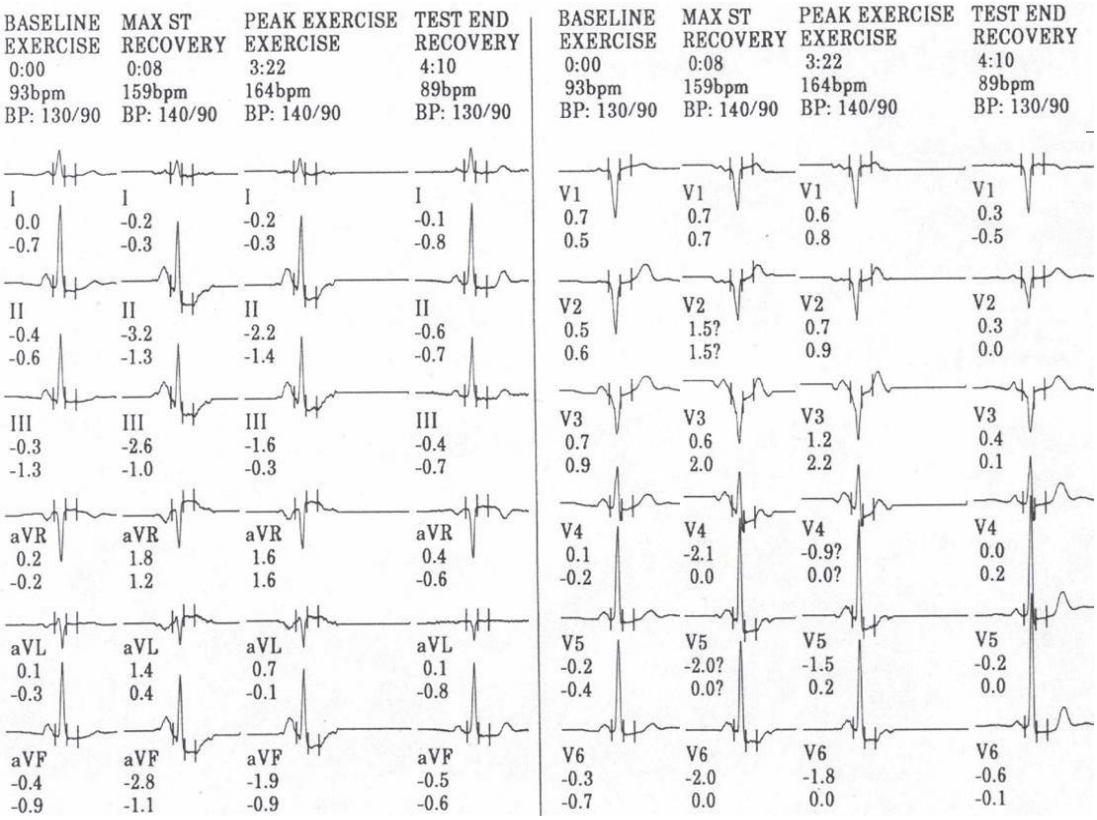


Fig.4. ECG dynamics during the exercise test of patient N., 16 years old

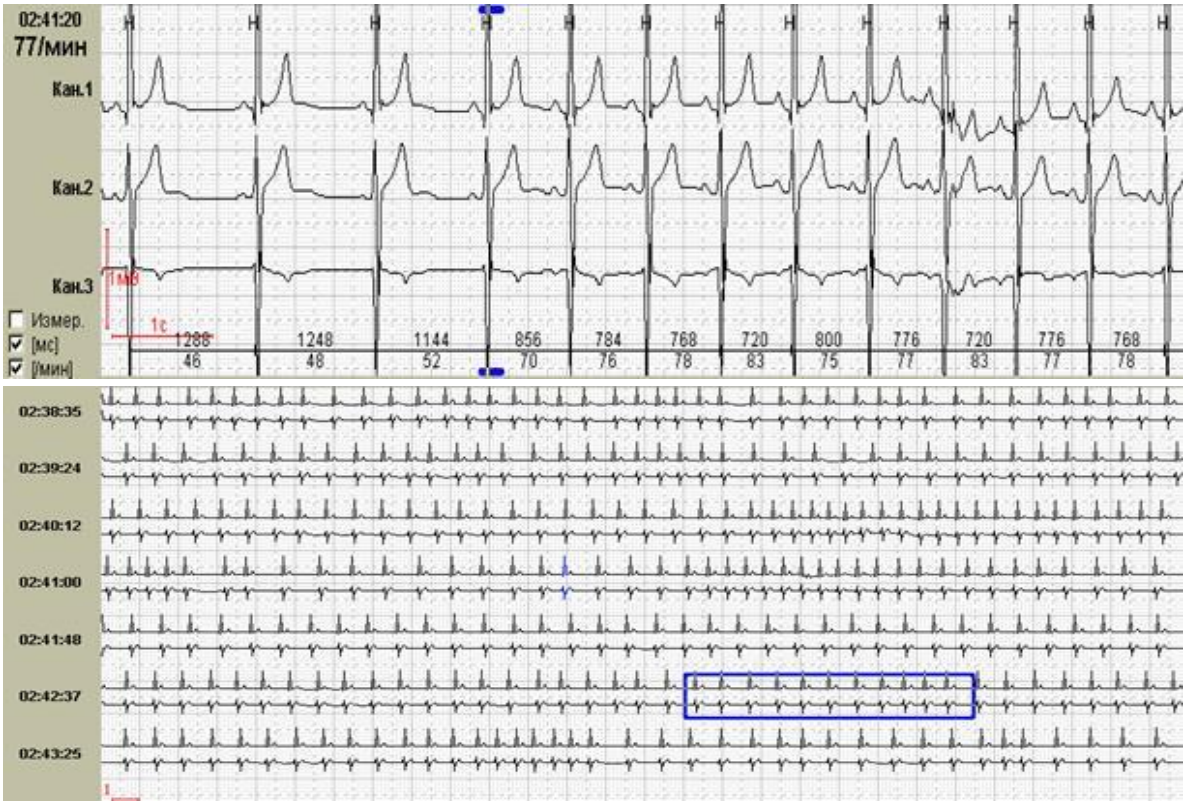


Fig.5. Fragment of the XM ECG of patient N., 16 years old

Protocol of echocardiography (EchoCG): final diastolic size of the left ventricle (LV) - 4,9 sm; final diastolic volume of the left ventricle - 133 ml, hypo- and akinesia zones were not detected. The Simpson's ejection fraction is 63%. The myocardial mass index is 95 g/m^2, the left ventricular myocardial mass is 198 g. The volume of the left atrium is 42 sm^3. There are median and apical additional chords in the left ventricular cavity. The aortic lumen at the level of the Valsalva sinus was 3,6 sm. The basal size of the right ventricle was 3,2 sm. Tricuspid and mitral regurgitation of the I degree (up to 0,3 mm). Systolic pressure in the pulmonary artery is 23 mmHg. Conclusion: LV function is preserved. Grade I mitral valve prolapse with grade I regurgitation. Tricuspid regurgitation of the first degree. Moderate aortic root dilation. In the cavity of the left ventricle there are additional (median and apical) chords.

Coronary angiography (CAG): Myocardial blood supply according to the right type. There are no stenoses in the coronary arteries. No additional turbulent flows were detected (fig. 6).

Ultrasound examination of the abdominal cavity (liver, kidneys, adrenal glands, pancreas) is a variant of the norm.

Laboratory studies: hemoglobin – 134 g/l; hematocrit – 42%; leukocytes – 5,4X10^9l; platelets – 220X10^9l; ESR -10 mm/hour; total cholesterol – 4,2 mmol/l; kalium - 4,6 mmol/l; creatinine - 92 mkmol/l; C-reactive protein – 0,2 mg/l; alanine aminotransferase - 19 U/l; aspartate aminotransferase - 21 U/l; total bilirubin - 18 mkmol/l; glucose - 4,1 mmol/l; markers of hepatitis HBsAg (screening) and anti-HCV Ig M, G (screening) – not detected, RW IEA – negative. Urine analysis without features.

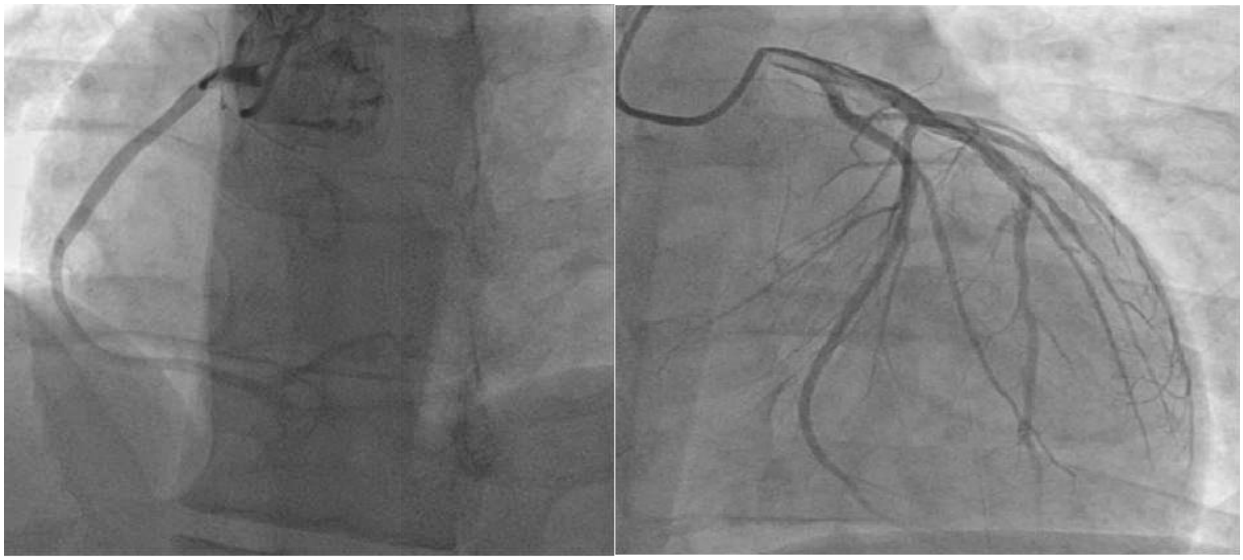


Fig.6. Coronary angiographic picture of patient N., 16 years old

Discussion. Patient N., 16 years old, was diagnosed with CTDS, which was confirmed by the presence of additional chords of the left ventricle of the heart and valve prolapse; dilation of the aortic root, autonomic dysfunction of the sinus node and deformation of the chest; diffuse myocardial changes were detected on the resting ECG and pronounced depression of the ST-segment at the height of physical activity. This made it difficult to interpret the obtained ischemic ECG criteria and required an additional CAG study, which revealed the absence of stenoses in the coronary arteries and confirmed non-coronary myocardial damage. This study made it possible to diagnose dysplastic heart caused by one of the types of metabolic cardiomyopathy with nonspecific ECG changes. In this syndrome, ECG changes are most often manifested by QT prolongation or metabolic disorders of the myocardium [5; 6]. Ischemic ECG criteria in individuals with undifferentiated connective tissue dysplasia are quite rare, which causes diagnostic difficulties and incorrect management tactics for such patients.

In this case, the girl's fitness for physical activity was assessed as questionable for medical reasons, since small cardiac abnormalities increase the risk of sudden cardiac death [7]. Metabolic therapy [8] (magnesium, ranolazine, mexidol) and the need for follow -up by a cardiologist were recommended.

Conclusion. The description of this clinical case shows that at present, further clinical observations of patients with dysplastic heart and continued study of the features of pathomorphological changes in the myocardium in connective tissue dysplasia syndrome are necessary for risk stratification.

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