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Clinical and Functional Significance of Associated Lesions in Combined Anomalies of Internal Organs in Children with Connective Tissue Dysplasia

Khojaeva Nigora Abdurashidovna, Takhirova Rohatoy Narmatovna

Tashkent Pediatric Medical Institute, Uzbekistan

Abstract Examination of children aged 6 months - 16 years was carried out with the definition of signs of connective tissue dysplasia. The study showed a high prevalence of signs of undifferentiated connective tissue dysplasia with damage to the cardiovascular system, biliary tract. Diseases in children with minor connective tissue abnormalities are more severe and are characterized by a longer course and a period of convalescence, and have a risk of developing chronic diseases.

Keywords Undifferentiated connective tissue dysplasia, Minor heart anomalies, Biliary tract anomalies

1. Introduction

Connective tissue is an integral physiological system that is widespread in the body everywhere in the form of numerous specialized varieties. The structural and functional role of connective tissue is represented by metabolic plastic, musculoskeletal and morphological properties.

The metabolic function of connective tissue is determined by the fact that being an integral part of the internal environment of the body, with blood vessels passing through it, it provides other tissues with nutrients. The plastic function of connective tissue is to eliminate tissue defects. This is the healing of wounds, ulcers, replacement regeneration of parenchymal organs. The musculoskeletal function is due to the formation of the skeleton (bone) of internal organs (stroma), muscles, blood vessels. A very important property of connective tissue, as strength is due to the presence of collagen fibers and structural glycoproteins. Tissue elasticity is associated with elastic properties, viscosity and elastoplastic properties are associated with proteoglycans and glycoproteins, and contractility is associated with the presence of myofibroblasts. [10,16]

Such is the significant role of connective tissue for the human body, the presence of which is associated with the formation of the morpho-functional structure of all organs and systems.

Pathological changes in connective tissue are manifested by common systemic lesions. They can be primary and secondary.

Primary systemic lesions of connective tissue are congenital and hereditary in nature, associated with malformations and metabolic disorders of the tissue itself.

Secondary lesions of connective tissue in pathology are manifested by damage to internal organs. These are fibrous growths, cirrhosis of organs, granulomatosis and amyloidosis.

Congenital pathology of connective tissue, manifested by a decrease in its strength, is commonly referred to by the term "dysplasia". [7,8]

Connective tissue dysplasia is a genetically determined anomaly in the development of the mesenchymal matrix of the body, leading to a decrease in the strength of the connective tissue of many organs and systems. She represents It is a complex process associated with many quantitative and qualitative changes in its basic structures. In the development of the clinical picture of connective tissue dysplasia, mutations of genes encoding the synthesis and spatial organization of collagen are of leading importance. [2,6]

2. Materials and Methods

Thus, hereditary disorders of connective tissue development associated with abnormalities in the synthesis and metabolism of collagen, as well as other proteins, form the basis for the formation of pathology of various organs and systems. [7,8,10]

In clinical practice, there are no sufficiently clear diagnostic criteria and laboratory tests that allow the diagnosis of connective tissue dysplasia syndrome with the isolation of individual nosological forms. Given the complexity of the molecular genetic diagnosis of the

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syndrome, the diversity and uncertainty of clinical manifestations, it was proposed to consider connective tissue dysplasia syndrome as a heterogeneous group of monofactorial disease. Multifactorial diseases are considered as a disease caused by the influence of an unfavorable environmental environment, inadequate nutrition, stresses that affected the body during ontogenesis. [1,2,3,13]

Thus, it was found that the unique morpho-functional structure of connective tissue creates certain conditions for the formation of a plurality of its anomalies and diseases caused by chromosomal and gene defects. [1,10]

We conducted dynamic monitoring of 45 children with connective tissue dysplasia syndrome who were hospitalized with cardiac complaints. In a certain group of patients, along with cardiac complaints – pain in the heart, palpitations, abdominal pain was constantly noted, nausea periodically and very rarely vomiting, decreased appetite and body weight.

A comprehensive clinical study was based on the establishment of external phenotypic manifestations of SDS, anamnestic data (collection of hereditary and family history). The instrumental studies carried out included EchoCG, ECG at rest and after exercise, Holter monitoring, blood pressure monitoring (BP), ultrasound examination of the abdominal cavity and kidneys. In a clinical study, the majority of the observed (88%) showed signs of connective tissue dysplasia, which were characterized by impaired posture, funnel-shaped deformation of the chest, thin skin, high arched palate, overextension of the joints. One child was found to have incomplete syndactyly.

Certain physical data during auscultation of the heart were found in 80% of children. Most of them (50%) had a relatively short systolic murmur detected at the base of the heart and at the V point. Systolic noise detected at the base of the heart was regarded as wired. In 10% of children, systolic noise was heard in the form of a click. Percussion in all examined patients, the boundaries of relative dullness were within the age norm. Small cardiac anomalies detected during echocardiography were regarded as manifestations of connective tissue dysplasia. In 15% of cases, children were diagnosed with an open oval opening (O.O.O). This is a small anomaly of the heart with partial or complete preservation of atrial communication, which is associated with postnatal non-infection of the left atrial valve flap of the heart. [12,15]

In school-age children, % of cases were diagnosed with a combination of LLC with an atrial septal aneurysm, which was characterized by bulging of the septum in the area of the oval fossa towards the right atrium. Clinically, auscultative symptoms were not often determined.

Of the total number of children under observation with echocardiography of the heart, four were diagnosed with grade 1 tricuspid valve prolapse. It was manifested by prolapse of the anterior flap of the valve with regurgitation of the 1st degree. Taking into account the absence of clinical physical manifestations, this variant of valve development anomaly was regarded as a physiological norm. Echocardiography revealed tricuspid valve insufficiency in one child - tricuspid insufficiency. During auscultation, the I cardiac tone was normal, the II tone was relatively louder compared to the I tone, the systolic noise was not strong, the electrocardiography data were normal. The chest X-ray did not reveal any special changes, it was normal.

The determined auscultative changes in this group of observed patients were different. A systolic noise of varying intensity, a systolic click was heard. Metabolic disorders in the myocardium, according to the conclusion of the electrocardiogram, were characterized by changes in the form of incomplete blockade of the right leg of the Gis beam, a violation of repolarization processes and single supraventricular extrasystoles (according to Holterev monitoring), sinus arrhythmia, and a shift of the electrical axis to the right.

Thus, signs of connective tissue dysplasia were observed in all observed children. The results of the study revealed that small anomalies of the development of the heart in the form of established variants are much more common in children of older school age. [5,6,11]

The study of diseases of the digestive system in connective tissue dysplasia syndrome is not very numerous. There are reports in the literature on the diagnosis of gastritis, duodenitis, gastric ulcer and duodenal ulcer, intestinal pathology and chronic cholecystitis in adult patients with connective tissue dysplasia syndrome. In these studies, connective tissue dysplasia syndrome is considered as a factor provoking and aggravating the severity of clinical manifestations of gastrointestinal pathology. According to a number of reports, the frequency of diagnosis of gastrointestinal tract pathology with connective tissue dysplasia syndrome has increased significantly in recent years. Moreover, a significant percentage of this pathology is diagnosed in school-age children.

Biliary tract diseases are one of the most common pathologies in childhood (3-14 years). Especially often, the pathology of the biliary tract is associated with congenital anomalies of the development of the gallbladder, determined by examining children with abdominal pain. Anomalies are congenital defects and malformations with impaired drainage function of the biliary tract with the progression of pathology and specific symptoms. [4,14]

In the observed contingent of children, along with cardiological manifestations of dysplasia syndrome, an anomaly of the shape of the gallbladder was diagnosed in 23x. The anomaly of the gallbladder was represented by its constrictions, kinks and S-shaped congenital character. Definitely, the presence of congenital anomalies of the gallbladder leads to violations of the passage of bile, the development of dyskinesia is possible and the addition of an inflammatory process in it. [1,3]

3. Result and Discussion

In our study, the nature of clinical manifestations varied from the presence of a combination of gallbladder lesions. In

the group of patients with an anomaly of the biliary system, pain syndrome prevailed. Moreover, the nature of the pain was cramping, short-term, often associated with physical stress and negative emotions. In the intervals between bouts of pain, patients did not make any special complaints. In 12 (51%) children with functional disorders of the gallbladder on the background of congenital malformations, clinical manifestations depended on the type of dyskinesia. In the hypokinetic variant of dyskinesia, dull pains, feelings of heaviness in the right hypochondrium were noted and sometimes had a cramping character.

The hypertensive variant of dyskinesia is clinically often manifested by acute pain. Dyspeptic phenomena were characteristic of all patients, they were constant, manifested by a decrease in appetite, nausea, bitterness in the mouth, and vomiting sharply. It should be noted that in children with abnormalities of the development of the form of the gallbladder – inflection and constriction, complicated by a hypokinetic variant of biliary dyskinesia, ultrasound examination revealed sludge syndrome in isolated cases. In a small group of children (4-17%) of older school age 15-16 years, chronic stone-free cholecystitis was diagnosed against the background of revealed abnormalities in the development of the gallbladder. The functional state of the gallbladder was characterized by a decrease in its contractile function. In the clinic of chronic cholecystitis, pain syndrome prevailed, constant dyspeptic manifestations (nausea, a sharp decrease in appetite, sometimes vomiting associated with heavy meals, constipation, heartburn). Clinical manifestations of chronic cholecystitis on the background of abnormal development of the biliary tract, are characterized by frequent exacerbations against the background of diet disorders, exacerbations having chronic diseases (in children especially with chronic tonsillitis) and after acute inflammatory diseases of the respiratory tract. With the exacerbation of the disease, signs of intoxication were noted in this group of patients. During the period of remission, patients practically did not make any complaints. Of the total number of observed children with abdominal pain, reactive hepatitis was detected in 30.4% of cases against the background of an anomaly in the shape of the biliary tract. Clinical manifestations of liver damage were characterized by an increase in the size of the liver of varying severity from 1.5 to 3 cm. The severity of the lesion was characterized not only by an increase in its size, but by a change in its consistency to moderate compaction. During ultrasound examination, the echogenicity of the liver was increased with changes in the intrahepatic bile ducts. The pain syndrome was manifested by the indefinite nature of pain in the upper abdominal cavity. It was often combined with a feeling of constant heaviness and aching pains in the right hypochondrium. Dyspeptic (nausea, decreased appetite, bitterness in the mouth) and asthenic (weakness, fatigue, mood lability) syndromes were quite constant. Biochemical parameters were transient in relation to liver enzymes with a low increase in them, the parameters of the thymol sample were 1.5 times higher than normal.

4. Conclusions

1. Anomaly of the biliary tract due to connective tissue dysplasia is a risk factor for the formation of functional and inflammatory diseases of the gallbladder associated with liver damage in children.
2. Clinical manifestations depended on the type and conjugacy of the lesion, were characterized by the presence of pain syndrome, dyspeptic disorders.
3. Children with abnormalities of the biliary tract with connective tissue dysplasia can be considered at risk for possible further development of cholelithiasis.

REFERENCES

[1] Arsentiev V.G., Baranov V.S., Shabalov N.P. Multi-organ disorders in connective tissue dysplasia in children. // Pediatrics 2009 T. 88 No. 1. C 135-138.

[2] Arsentiev V.G. connective tissue dysplasia as the constitutional basis of poly-organ disorders in children: Author. diss. Doctor of Medical Sciences. SPB., 2012; 44c.

[3] Arsentiev V.G., Baranov V.S., Shabalov N.P. Hereditary connective tissue diseases as a constitutional cause of multiple organ disorders in children. – St. Petersburg.: Spec. Lit. 2014. – 188s.

[4] Arseityev V.G., Mozhaiko A.G., Staroverov Yu.I., Gnusaev O.K., Shabalov N.P. Results of gastroenterological examination of children with connective tissue dysplasia. // Pediatrics. -2012; 91(1): 149.

[5] Akhrarova F.M. Clinical and pathological substantiation of cardiovascular pathology of connective tissue dysplasia. Author. diss. Doctor of Philosophy (PhD) in Medical Sciences. 2020; p.

[6] Zemtsovsky E.V., Malev E.L. Small anomalies of the heart. // Russian Journal of Cardiology. 2012; 1(93): 78-81.

[7] Kadurina T.N. // Defeat of the cardiovascular system in children with various variants of hereditary connective tissue diseases. Conduct arrhythmology. 200. No.18. From 87.

[8] Kildiyarova R.R., Ivanova I.L., Chernysheva T.E. Connective tissue exchange in adolescents with connective tissue dysplasia. In: Pediatric aspects of connective tissue dysplasia: achievements and prospects. Issue 2 Under. Edited by S.V. Gusaeva, T.I. Kadurina; A.N. Semyachkina. M. -Tver. -San. -Petersburg: RG. <<PE 100>> 2011, 52-57.

[9] Kotovskaya E.S., Mazaev V.P., Zhdanova S.M. // Cardiovascular diseases and connective tissue dysfunction. Dep. in CMB. 1993. from 1-8.

[10] Metabolic aspects of undifferentiated connective tissue dysplasia in children / I.I. Ivanova, S.D. Gnusaev, N.Yu. Koval, N.A. Gerasimov, I.A. Soldatova, Ros Vest Perinatal Pediatr 2012. 4(1): 103-111.

[11] Muratkhodzhaeva A.V., Akhrarova F.M. Echocardiographic characteristics of small heart development abnormalities in children with clinical manifestations of connective tissue dysplasia // Journal of Pediatrics No. 3, 2021; pp.179-186.

[12] Mutafyan O.A. Heart defects and minor anomalies in children and adolescents, St. Petersburg, 2005.

[13] Hereditary and multifactorial disorders of connective tissue in children. Diagnostic algorithm, management tactics. The draft of Russian recommendations was developed by the committee of experts of the pediatric group "Connective tissue Dysplasia" at the Russian Scientific Society of Therapists – 2014. – T93 No. 5-40s.

[14] Samokhina A.V. // Modern ideas about variants of the gallbladder and bile ducts when using modern methods of instrumental research. Journal of Grodno State University No. 3(35) – 2011-From 3-6.

[15] Edward A. Gill Definitions and pathophysiology of the patent foramen owale: broad overview // Cardiology Clinics, 2005- vol. 23 iss. 1 pp. 1-6.

[16] Human physiology. Pokrovsky V.M., Korotko G.F. Textbook. twothousandeight.