

Features of the Clinical Manifestation of Juvenile Rheumatoid Arthritis Against the Background of Connective Tissue Dysplasia

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Abstract Juvenile arthritis is a complex medical problem. Diagnosis and treatment also require special attention from specialists. Connective tissue dysplasia is a genetic condition characterised by impaired collagen formation, which is involved in strengthening connective tissue. This condition is particularly important in the diagnosis of various diseases. This study was conducted among patients with juvenile arthritis with signs of connective tissue dysplasia. The study showed us that there are some features of juvenile arthritis against the background of connective tissue dysplasia.

Keywords Juvenile arthritis, Connective tissue dysplasia, Joint changes, C-reactive protein, Oligoarticular variant, Polyarticular variant, Joint signs, Visceral signs

1. Introduction

Juvenile arthritis is a common chronic inflammatory joint disease in children of multifactorial nature, characterised by a long progressive course leading to the development of contractures and loss of joint function. The disease is based on a chronic progressive inflammatory process of the inner layer of the joint capsule (synovial membrane), which leads to the destruction of cartilage and bone tissue. The prevalence of the disease in different countries ranges from 16 to 150 cases per 100,000 population [1].

Connective tissue dysplasia (CTD) is a genetically determined disease characterised by defects in the fibrous structures and ground substance of connective tissue, leading to abnormalities in the formation of organs and systems. It is progressive in nature, determining the characteristics of concomitant pathology, as well as the pharmacokinetics and pharmacodynamics of drugs. The formation of connective tissue dysplasia is based on hereditary mutations in genes encoding the synthesis and spatial organisation of collagen, structural proteins and protein-carbohydrate complexes, as well as mutations in genes encoding enzymes and their cofactors. Connective tissue dysplasia is characterised by impaired synthesis of type II collagen, which is caused by abnormalities in the structure of extracellular matrix components with progressive morphofunctional changes in

various organ systems. Indeed, connective tissue is responsible not only for maintaining the shape of organs, but also for trophic, metabolic, and immune functions, since in the human body it is represented not only in the form of connective tissue itself, but also in dense connective tissue (cartilage and bone), haematopoietic tissue, and blood cells. Connective tissue dysplasia (CTD) is a common condition which, according to current data, is found in approximately 80% of the population.

Most often (57-94% of cases), CTD is diagnosed based on skeletal pathology: deformation of the chest and spine, flat feet, joint hypermobility, juvenile osteochondrosis. In addition, there are some phenotypic and visceral signs of connective tissue dysplasia. Phenotypic signs include thin and stretchy skin, visible venous network, muscle hypotonia, diastasis, and others. Myopia, abnormalities of the biliary tract, abdominal hernias, mitral valve prolapse, vegetative-vascular dystonia, immune defence disorders, and abnormalities of the gastrointestinal tract. There are also laboratory indicators of connective tissue dysplasia. These include low magnesium ion levels in various substrates and high levels of proline and hydroxyproline in the blood [2].

2. Materials and Methods

The study was conducted at the 4th City Clinical Children's Hospital among 57 children. Of the 57 children, 32 had signs of connective tissue dysplasia and 25 had no signs of connective tissue dysplasia. Their ages ranged from 4 to 16 years with different variants of JRA and periods of activity.

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3. Results and Discussion

The most common type of juvenile arthritis was the oligoarticular variant in 57.6% of children. Preschool-aged patients (39.8%) and adolescents (42.1%) predominated in the study group. Girls outnumbered boys in the study group. Almost half of the children examined with JIA were patients with the first degree of inflammation activity, and 72% of them had the oligoarticular variant. In addition, among the patients we studied with the polyarticular variant of JIA, the second degree of inflammation activity was more common (68%). In many cases, boys had the first degree of activity (68.2%), while girls had the second (78.3%) and third (80.4%) degrees of activity. Among the study group, approximately 31% of children had no joint dysfunction, and these patients had the oligoarticular variant of JIA. Joint function limitation was observed in 47% of children, but they had no problems with self-care. 16.6% of children had difficulties with self-care and had a systemic variant of juvenile rheumatoid arthritis.

The study group had various types of joint syndrome, with joint deformation being the most common type (91.2%). Among patients with oligoarticular and polyarticular types, 49.3% complained of morning stiffness. Patients with grades 2 and 3 inflammation activity experienced pain, functional impairment, and local hyperthermia of the joints. Among the majority of patients with all types of juvenile arthritis, knee joint involvement was observed in 89.9% of cases. Cervical spine involvement was observed in 81.8% of children with the polyarticular variant. Among the majority of patients, we examined, the knee joint was primarily affected in 58.7% at the onset of the disease.

One of the extra-articular signs of lymphadenopathy was recorded in 57.8% of children. Patients with polyarticular and systemic juvenile rheumatoid arthritis experienced intoxication syndrome, manifested by weakness, lethargy, malaise, and loss of appetite. Almost half of the sick children had a fever. In the systemic variant of juvenile arthritis, the temperature was 39 degrees, while in patients with the oligo- and polyarticular variants, it was subfebrile and febrile. Among patients with the systemic variant of the disease, a maculopapular rash was observed. Two patients with juvenile arthritis had eye damage. Patients with the systemic variant of the disease had metabolic changes in the myocardium according to electrocardiography data. Patients with connective tissue dysplasia had conduction rhythm disturbances. In addition, patients with the systemic variant of juvenile arthritis had lung damage in the form of pneumonitis. Auscultation revealed harsh breathing, and X-rays showed increased pulmonary pattern. Patients with grades 2 and 3 had intoxication and hyperthermia syndromes.

According to laboratory analysis, half (52.4%) of patients with this disease had leukocytosis, a left shift in the leukocyte formula in 56.2% of patients, and accelerated ESR in 60.3% of patients. A biochemical blood test was also performed. It is important to pay attention to the increase in CRP, because it indicates the activity of inflammation in JIA,

and it is 88.3%. In patients with connective tissue dysplasia, a decrease in serum magnesium concentration was observed in 56.8% of patients.

The patients we examined underwent X-ray examination of the knee joints. In patients with a history of 6 months to 3 years, periarticular osteoporosis was observed in 76.7%. Joint space narrowing was detected in 20.6% of children with JIA. Some patients had joint subluxations.

Ultrasound examination revealed synovitis (thickening of the synovial membrane) in 94.2% of patients with JIA. In patients with the oligoarticular variant of JIA, 86.8% had a pathognomonic symptom of chronic arthritis – proliferation of the villi of the synovial membrane. In patients with signs of connective tissue dysplasia, thinning of the hyaline cartilage was noted.

Our study showed that there are some characteristics of juvenile arthritis against the background of connective tissue dysplasia. Among patients with signs of connective tissue dysplasia, it was more common in girls (72.8%) than in boys. Signs of connective tissue dysplasia were observed in preschool and school-age patients (58.4%) more often than in adolescents. Among patients with juvenile arthritis, the oligoarticular variant was more common in patients with signs of CTD (71.8%). The systemic variant was noted in patients without signs of connective tissue dysplasia (31.5%). A high degree of inflammation activity was noted in patients with connective tissue dysplasia (22.4%). Joint deformation was observed in patients with CTD, while patients without signs of dysplasia had arthralgia and joint restriction. In 89.8% of patients with signs of connective tissue dysplasia, frequent knee joint involvement was noted. According to laboratory studies among patients with juvenile arthritis, pronounced changes were observed in patients with signs of connective tissue dysplasia. These same patients had high ESR values of over 45 mm/h and CRP values above 60 mg/l. Radiographic changes in the joints were more common in patients with symptoms of connective tissue dysplasia (88%).

4. Conclusions

Patients with juvenile arthritis and connective tissue dysplasia have a higher degree of inflammation activity while maintaining functional joint capacity. Connective tissue dysplasia predisposes to earlier onset of degenerative and destructive changes in the joints.

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