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APPLICATION OF INNOVATIVE TECHNOLOGIES IN THE DIAGNOSIS OF CONNECTIVE TISSUE DYSPLASIA IN CHILDREN

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Annotation: Connective tissue dysplasia is one of the most urgent problems today. Currently, according to the literature, more than 200 phenotypic signs of connective tissue have been identified. The results showed that connective tissue dysplasia is more common in preschool and school-age children, with a predominance in girls. Further study of clinical and biochemical markers and special diet therapy is required, including a personalized approach with full-fledged micronutrient support, as well as the selection of medicines depending on the severity of the children's condition.
Key words: Connective tissue dysplasia, children, primary care.

The relevance of the work. Connective tissue dysplasia (CTD) has been known since the 19th century, thanks to advances in genetics. DST and genetic disorders were first identified in 1955 by the American geneticist Victor Mccastick, who combined some nosological forms: Marfan syndrome, osteogenesis imperfecta, Ehlers-Danlo syndrome. The researchers note that at the heart of every chronic disease lies the incorrect formation of connective tissue in the embryonic period of development.

DST is one of the most pressing issues today. The relevance of DST is expressed in the prevalence of morphogenesis markers. Currently, according to the literature, more than 200 phenotypic signs of connective tissue have been identified [2]. In recent years, the frequency of DST in different countries of the world among the child population has been increasing. For example, an initial cohort of patients with connective tissue diseases in Olmsted County, Minnesota, USA, from 1985 to 2014 reported an annual incidence of 1.9 per 100,000 population based on an individual review of medical records [3, 5].

Purpose of the work. Study of the prevalence of DST in children in hot climates, introduction of a computer program (OSTDST-VSKD.exe-VSKD-22,23,24,26,62) by primary care doctors in family polyclinics (no. -22,23,24,26,62) of Uchtepa district of Tashkent city.

Materials and methods of research. We conducted research in family polyclinics (№-22,23,24,26,62) of Uchtepa district. A total of 167 children aged from 1 to 18 years with various degrees of DST were identified. The study was conducted from July to December 2022. Detection selection was applied using a computer program (OSTDST-VSKD.exe-VSKD). The study program included children with more than 6 phenotypic traits and more than 2 dysembryogenesis stigmas.

Results and discussion. It should be noted that the specific structure and function of connective tissue leads to its defects of a certain specialized type or due to negative environmental factors (unfavorable environmental conditions, the presence of heavy metal salts

in the air, poor nutrition, stress, etc.) during pregnancy. As a result of the mutagenic effect on DST, it creates an opportunity for the development of anomalies and chronic diseases [1].

More than 70% of DST cases are differentiated DST. The incidence of undifferentiated CTD among the population is 10-30%. Using modern achievements of molecular genetics, we can distinguish 8 groups of congenital differential connective tissue dysplasia [5]:

1. Hereditary collagenopathies: incomplete osteogenesis, Ehlers-Danlos syndrome, various variants of chondrodysplasia, ophthalmopathies, nephropathies, abnormal cysts, organozrenia, myopathies, epidermolysis bullosa.

2. Hereditary fibrillopathies: Marfan syndrome; MASS syndrome; lens ectopia with mild skeletal manifestations of the marfanoid type without cardiovascular pathology; Marfan syndrome in combination with Sprintzen-Goldberg syndrome; marfanoid skeletal syndrome glandular cardiovascular and ocular anomalies; Weil-Marchesani syndrome; arachnodactyly contracture, congenital, or syndrome Bilsa.

3. Hereditary elastinopathies: supravalvular Eisenberg aortic stenosis; Williams Buren syndrome; cutis laxa, congenital, autosomal dominant

4. Hereditary fibulinopathies: cutis laha, congenital, autosomal dominant; congenital, autosomal recessive; patchy retinal degeneration, age -dependent; cellular retinal dystrophy.

5. Hereditary laminopathies: muscular dystrophy, congenital, merozindeficient, type 1A; autosomal recessive; bullous epidermolysis, linear, lethal; bullous epidermolysis, generalized, atrophic, benign; neonatal кожа lax skin with a marfanoid phenotype; Pearson's congenital nephrosis, perinatal lethal form.

6. Hereditary thrombospondinopathies: pseudochondroplasia, multiple epiphyseal dysplasia.

7. Hereditary proteoglycanopathies: various clinical variants of chondrodysplasia, joint abnormalities, ophthalmopathies, nephropathies and epidermolysis bullosa.

8. Hereditary DST caused by mutations in the gene of fibroblast growth factors, their receptors and antagonists: various forms of craniosynostosis, achondroplasia, chondrodysplasia, brachydactyly, symphalangism, multiple synostosis, ankylosis and sclerosis syndromes [4].

A peculiar pathology of the phenomenon of dysmorphogenesis is that the phenotypic signs of DST are pronounced from birth, and growth and development has a progrodient course, which leads to and aggravates the development and structure of organs and systems.

DST syndrome is characterized not only by changes in one or several organs, but also by varying degrees of changes in the entire body, depending on the penetrance of the gene titer and the severity of dysplasia in different degrees for each individual [5].

Using a computer program(OSTDST-VSKD.exe-VSKD"Assessment of the severity of connective tissue dysplasia in children based on the severity of symptom complexes" the following results were obtained in family polyclinics (SP) of Uchtepa district.

Table 1

Prevalence of DST in children in SP Uchtepa district

Degree Severity	of SP 22	SP 23	SP 24	SP 26	SP 62	Totalamount of it	percentage %
Normal	18	24	12	9	25	88	52.7%
Mild	13	11	8	5	19	56	33.5%
Moderate	4	6	3	1	7	21	12.6%
Severe	-1	1	--	-	1	2	1.2%
total	35	42	23	15	52	167	100%

A total of 167 children were examined: 79 of them were diagnosed with DST. According to the severity, they were divided into the following (Table 1): 0 degree or healthy: 88 children

(52.7%); 1 degree or mild: 56 children (33.5%); 2 medium degree: 21 children (12.6%); 3 severe degree: 2 children (1.2%).

Table 2

Prevalence of DST in children by age
in the SP Uchtepinsky district

Age	SP 22	SP 23	SP 24	SP 26	SP 62	Total	percentage %
1-3 years	2	3	1	3	5	14	17.7%
4-7 years	5	8	7	1	9	30	37,8%
8-14 years	4	6	2	1	9	22	27.8%
15-18 years	6	1	1	1	4	13	16.7%
Total	17	18	11	6	27	79	100%

Children with DST, depending on their age, were identified as follows: (2 tables)

The number of children aged 1-3 years was 14 (17.7%), at the age of 4-7 years it was 30 (37.8%), at the age of 8-14 years it was 22 (27.8%) and at the age of 15-18 years it was 13 (16.7%).

Among the identified 167 children with DST, 44 girls (55.7%), 35 boys (44.3%).

Conclusion. The results showed that DST occurs more frequently in children of preschool and school age, with a predominance in girls. Further study of clinical and biochemical markers and special diet therapy is required, including a personalized approach with full-fledged micronutrient support, as well as the selection of medications depending on the severity of the DST condition in children.

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