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ЕЖЕМЕСЯЧНЫЙ НАУЧНЫЙ ЖУРНАЛ

Медицинские новости Грузии

საქართველოს სამედიცინო სიახლენი

GEORGIAN MEDICAL NEWS

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GMN: Georgian Medical News is peer-reviewed, published monthly journal committed to promoting the science and art of medicine and the betterment of public health, published by the GMN Editorial Board since 1994. GMN carries original scientific articles on medicine, biology and pharmacy, which are of experimental, theoretical and practical character; publishes original research, reviews, commentaries, editorials, essays, medical news, and correspondence in English and Russian.

GMN is indexed in MEDLINE, SCOPUS, PubMed and VINITI Russian Academy of Sciences. The full text content is available through EBSCO databases.

GMN: Медицинские новости Грузии - ежемесячный рецензируемый научный журнал, издаётся Редакционной коллегией с 1994 года на русском и английском языках в целях поддержки медицинской науки и улучшения здравоохранения. В журнале публикуются оригинальные научные статьи в области медицины, биологии и фармации, статьи обзорного характера, научные сообщения, новости медицины и здравоохранения. Журнал индексируется в MEDLINE, отражён в базе данных SCOPUS, PubMed и ВИНИТИ РАН. Полнотекстовые статьи журнала доступны через БД EBSCO.

GMN: Georgian Medical News – საქართველოს სამედიცინო სიახლენი – არის ყოველთვიური სამეცნიერო სამედიცინო რეცენზირებადი ჟურნალი, გამოიცემა 1994 წლიდან, წარმოადგენს სარედაქციო კოლეგიისა და აშშ-ის მეცნიერების, განათლების, ინდუსტრიის, ხელოვნებისა და ბუნებისმეტყველების საერთაშორისო აკადემიის ერთობლივ გამოცემას. GMN-ში რუსულ და ინგლისურ ენებზე ქვეყნდება ექსპერიმენტული, თეორიული და პრაქტიკული ხასიათის ორიგინალური სამეცნიერო სტატიები მედიცინის, ბიოლოგიისა და ფარმაციის სფეროში, მიმოხილვითი ხასიათის სტატიები.

ჟურნალი ინდექსირებულია MEDLINE-ის საერთაშორისო სისტემაში, ასახულია SCOPUS-ის, PubMed-ის და ВИНИТИ РАН-ის მონაცემთა ბაზებში. სტატიების სრული ტექსტი ხელმისაწვდომია EBSCO-ს მონაცემთა ბაზებიდან.

WEBSITE

www.geomednews.com

К СВЕДЕНИЮ АВТОРОВ!

При направлении статьи в редакцию необходимо соблюдать следующие правила:

1. Статья должна быть представлена в двух экземплярах, на русском или английском языках, напечатанная через **полтора интервала на одной стороне стандартного листа с шириной левого поля в три сантиметра**. Используемый компьютерный шрифт для текста на русском и английском языках - **Times New Roman** (**Кириллица**), для текста на грузинском языке следует использовать **AcadNusx.** Размер шрифта - **12**. К рукописи, напечатанной на компьютере, должен быть приложен CD со статьей.

2. Размер статьи должен быть не менее десяти и не более двадцати страниц машинописи, включая указатель литературы и резюме на английском, русском и грузинском языках.

3. В статье должны быть освещены актуальность данного материала, методы и результаты исследования и их обсуждение.

При представлении в печать научных экспериментальных работ авторы должны указывать вид и количество экспериментальных животных, применявшиеся методы обезболивания и усыпления (в ходе острых опытов).

4. К статье должны быть приложены краткое (на полстраницы) резюме на английском, русском и грузинском языках (включающее следующие разделы: цель исследования, материал и методы, результаты и заключение) и список ключевых слов (key words).

5. Таблицы необходимо представлять в печатной форме. Фотокопии не принимаются. **Все цифровые, итоговые и процентные данные в таблицах должны соответствовать таковым в тексте статьи**. Таблицы и графики должны быть озаглавлены.

6. Фотографии должны быть контрастными, фотокопии с рентгенограмм - в позитивном изображении. Рисунки, чертежи и диаграммы следует озаглавить, пронумеровать и вставить в соответствующее место текста **в tiff формате**.

В подписях к микрофотографиям следует указывать степень увеличения через окуляр или объектив и метод окраски или импрегнации срезов.

7. Фамилии отечественных авторов приводятся в оригинальной транскрипции.

8. При оформлении и направлении статей в журнал МНГ просим авторов соблюдать правила, изложенные в «Единых требованиях к рукописям, представляемым в биомедицинские журналы», принятых Международным комитетом редакторов медицинских журналов - http://www.spinesurgery.ru/files/publish.pdf и http://www.nlm.nih.gov/bsd/uniform_requirements.html
В конце каждой оригинальной статьи приводится библиографический список. В список литературы включаются все материалы, на которые имеются ссылки в тексте. Список составляется в алфавитном порядке и нумеруется. Литературный источник приводится на языке оригинала. В списке литературы сначала приводятся работы, написанные знаками грузинского алфавита, затем кириллицей и латиницей. Ссылки на цитируемые работы в тексте статьи даются в квадратных скобках в виде номера, соответствующего номеру данной работы в списке литературы. Большинство цитированных источников должны быть за последние 5-7 лет.

9. Для получения права на публикацию статья должна иметь от руководителя работы или учреждения визу и сопроводительное отношение, написанные или напечатанные на бланке и заверенные подписью и печатью.

10. В конце статьи должны быть подписи всех авторов, полностью приведены их фамилии, имена и отчества, указаны служебный и домашний номера телефонов и адреса или иные координаты. Количество авторов (соавторов) не должно превышать пяти человек.

11. Редакция оставляет за собой право сокращать и исправлять статьи. Корректура авторам не высылается, вся работа и сверка проводится по авторскому оригиналу.

12. Недопустимо направление в редакцию работ, представленных к печати в иных издательствах или опубликованных в других изданиях.

При нарушении указанных правил статьи не рассматриваются.

REQUIREMENTS

Please note, materials submitted to the Editorial Office Staff are supposed to meet the following requirements:

1. Articles must be provided with a double copy, in English or Russian languages and typed or compu-ter-printed on a single side of standard typing paper, with the left margin of **3** centimeters width, and **1.5** spacing between the lines, typeface - **Times New Roman** (**Cyrillic**), print size - **12** (referring to Georgian and Russian materials). With computer-printed texts please enclose a CD carrying the same file titled with Latin symbols.

2. Size of the article, including index and resume in English, Russian and Georgian languages must be at least 10 pages and not exceed the limit of 20 pages of typed or computer-printed text.

3. Submitted material must include a coverage of a topical subject, research methods, results, and review.

Authors of the scientific-research works must indicate the number of experimental biological species drawn in, list the employed methods of anesthetization and soporific means used during acute tests.

4. Articles must have a short (half page) abstract in English, Russian and Georgian (including the following sections: aim of study, material and methods, results and conclusions) and a list of key words.

5. Tables must be presented in an original typed or computer-printed form, instead of a photocopied version. **Numbers, totals, percentile data on the tables must coincide with those in the texts of the articles.** Tables and graphs must be headed.

6. Photographs are required to be contrasted and must be submitted with doubles. Please number each photograph with a pencil on its back, indicate author's name, title of the article (short version), and mark out its top and bottom parts. Drawings must be accurate, drafts and diagrams drawn in Indian ink (or black ink). Photocopies of the X-ray photographs must be presented in a positive image in **tiff format**.

Accurately numbered subtitles for each illustration must be listed on a separate sheet of paper. In the subtitles for the microphotographs please indicate the ocular and objective lens magnification power, method of coloring or impregnation of the microscopic sections (preparations).

7. Please indicate last names, first and middle initials of the native authors, present names and initials of the foreign authors in the transcription of the original language, enclose in parenthesis corresponding number under which the author is listed in the reference materials.

8. Please follow guidance offered to authors by The International Committee of Medical Journal Editors guidance in its Uniform Requirements for Manuscripts Submitted to Biomedical Journals publication available online at: http://www.nlm.nih.gov/bsd/uniform_requirements.html
http://www.icmje.org/urm_full.pdf
In GMN style for each work cited in the text, a bibliographic reference is given, and this is located at the end of the article under the title "References". All references cited in the text must be listed. The list of references should be arranged alphabetically and then numbered. References are numbered in the text [numbers in square brackets] and in the reference list and numbers are repeated throughout the text as needed. The bibliographic description is given in the language of publication (citations in Georgian script are followed by Cyrillic and Latin).

9. To obtain the rights of publication articles must be accompanied by a visa from the project instructor or the establishment, where the work has been performed, and a reference letter, both written or typed on a special signed form, certified by a stamp or a seal.

10. Articles must be signed by all of the authors at the end, and they must be provided with a list of full names, office and home phone numbers and addresses or other non-office locations where the authors could be reached. The number of the authors (co-authors) must not exceed the limit of 5 people.

11. Editorial Staff reserves the rights to cut down in size and correct the articles. Proof-sheets are not sent out to the authors. The entire editorial and collation work is performed according to the author's original text.

12. Sending in the works that have already been assigned to the press by other Editorial Staffs or have been printed by other publishers is not permissible.

Articles that Fail to Meet the Aforementioned Requirements are not Assigned to be Reviewed.

ავტორთა საყურადღებოდ!

რედაქციაში სტატიის წარმოდგენისას საჭიროა დავიცვათ შემდეგი წესები:

1. სტატია უნდა წარმოადგინოთ 2 ცალად, რუსულ ან ინგლისურ ენებზე,დაბეჭდილი სტანდარტული ფურცლის 1 გვერდზე, 3 სმ სიგანის მარცხენა ველისა და სტრიქონებს შორის 1,5 ინტერვალის დაცვით. გამოყენებული კომპიუტერული შრიფტი რუსულ და ინგლისურენოვან ტექსტებში - **Times New Roman (Кириллица)**, ხოლო ქართულენოვან ტექსტში საჭიროა გამოვიყენოთ **AcadNusx.** შრიფტის ზომა – 12. სტატიას თან უნდა ახლდეს CD სტატიით.

2. სტატიის მოცულობა არ უნდა შეადგენდეს 10 გვერდზე ნაკლებს და 20 გვერდზე მეტს ლიტერატურის სიის და რეზიუმეების (ინგლისურ, რუსულ და ქართულ ენებზე) ჩათვლით.

3. სტატიაში საჭიროა გაშუქდეს: საკითხის აქტუალობა; კვლევის მიზანი; საკვლევი მასალა და გამოყენებული მეთოდები; მიღებული შედეგები და მათი განსჯა. ექსპერიმენტული ხასიათის სტატიების წარმოდგენისას ავტორებმა უნდა მიუთითონ საექსპერიმენტო ცხოველების სახეობა და რაოდენობა; გაუტკივარებისა და დაძინების მეთოდები (მწვავე ცდების პირობებში).

4. სტატიას თან უნდა ახლდეს რეზიუმე ინგლისურ, რუსულ და ქართულ ენებზე არანაკლებ ნახევარი გვერდის მოცულობისა (სათაურის, ავტორების, დაწესებულების მითითებით და უნდა შეიცავდეს შემდეგ განყოფილებებს: მიზანი, მასალა და მეთოდები, შედეგები და დასკვნები; ტექსტუალური ნაწილი არ უნდა იყოს 15 სტრიქონზე ნაკლები) და საკვანძო სიტყვების ჩამონათვალი (key words).

5. ცხრილები საჭიროა წარმოადგინოთ ნაბეჭდი სახით. ყველა ციფრული, შემაჯამებელი და პროცენტული მონაცემები უნდა შეესაბამებოდეს ტექსტში მოყვანილს.

6. ფოტოსურათები უნდა იყოს კონტრასტული; სურათები, ნახაზები, დიაგრამები - დასათაურებული, დანომრილი და სათანადო ადგილას ჩასმული. რენტგენოგრამების ფოტოასლები წარმოადგინეთ პოზიტიური გამოსახულებით **tiff** ფორმატში. მიკროფოტოსურათების წარწერებში საჭიროა მიუთითოთ ოკულარის ან ობიექტივის საშუალებით გადიდების ხარისხი, ანათალების შეღებვის ან იმპრეგნაციის მეთოდი და აღნიშნოთ სურათის ზედა და ქვედა ნაწილები.

7. სამამულო ავტორების გვარები სტატიაში აღინიშნება ინიციალების თანდართვით, უცხოურისა – უცხოური ტრანსკრიპციით.

8. სტატიას თან უნდა ახლდეს ავტორის მიერ გამოყენებული სამამულო და უცხოური შრომების ბიბლიოგრაფიული სია (ბოლო 5-8 წლის სიღრმით). ანბანური წყობით წარმოდგენილ ბიბლიოგრაფიულ სიაში მიუთითეთ ჯერ სამამულო, შემდეგ უცხოელი ავტორები (გვარი, ინიციალები, სტატიის სათაური, ჟურნალის დასახელება, გამოცემის ადგილი, წელი, ჟურნალის №, პირველი და ბოლო გვერდები). მონოგრაფიის შემთხვევაში მიუთითეთ გამოცემის წელი, ადგილი და გვერდების საერთო რაოდენობა. ტექსტში კვადრატულ ფჩხილებში უნდა მიუთითოთ ავტორის შესაბამისი N ლიტერატურის სიის მიხედვით. მიზანშეწონილია, რომ ციტირებული წყაროების უმეტესი ნაწილი იყოს 5-6 წლის სიღრმის.

9. სტატიას თან უნდა ახლდეს: ა) დაწესებულების ან სამეცნიერო ხელმძღვანელის წარდგინება, დამოწმებული ხელმოწერითა და ბეჭდით; ბ) დარგის სპეციალისტის დამოწმებული რეცენზია, რომელშიც მითითებული იქნება საკითხის აქტუალობა, მასალის საკმაობა, მეთოდის სანდოობა, შედეგების სამეცნიერო-პრაქტიკული მნიშვნელობა.

10. სტატიის ბოლოს საჭიროა ყველა ავტორის ხელმოწერა, რომელთა რაოდენობა არ უნდა აღემატებოდეს 5-ს.

11. რედაქცია იტოვებს უფლებას შეასწოროს სტატია. ტექსტზე მუშაობა და შეჯერება ხდება საავტორო ორიგინალის მიხედვით.

12. დაუშვებელია რედაქციაში ისეთი სტატიის წარდგენა, რომელიც დასაბეჭდად წარდგენილი იყო სხვა რედაქციაში ან გამოქვეყნებული იყო სხვა გამოცემებში.

აღნიშნული წესების დარღვევის შემთხვევაში სტატიები არ განიხილება.

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FREQUENCY AND CLINICAL MANIFESTATIONS OF CONNECTIVE TISSUE DYSPLASIA IN CHILDREN IN THE CITY OF SEMEY

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Abstract.

Introduction: Connective tissue dysplasia (CTD) is a common condition in children and is characterized by a variety of manifestations of the musculoskeletal and cardiovascular systems. The aim of this study was to assess the frequency, age distribution, and clinical features of CTD in children aged 3–16 years.

Materials and methods: A total of 375 children (3–16 years) were examined for signs of CTD. Clinical evaluation included analysis of musculoskeletal signs (joint hypermobility, scoliosis, flat feet, chest deformities, muscle hypotonia), phenotypic features (asthenic body type, auricular anomalies, arachnodactyly, dolichostenomelia), and cardiovascular disorders (mitral valve prolapse, conduction disturbances, accessory chords and trabeculae). Children were divided into age groups (3–7, 8–10, 11–16 years) and CTD severity (grades 1–3) to analyze the prevalence and characteristics of clinical manifestations.

Results: Signs of CTD were identified in 75 children (20%). Most cases were mild forms (grade 1- 12.8%), moderate forms (grade 2) were less common (5.9%), and severe forms (grade 3) were extremely rare (1.3%). The most common musculoskeletal manifestations were joint hypermobility (78.7%), asthenic body type (61.3%), scoliosis (54.7%), and flat feet (42.7%). Cardiovascular disorders, including mitral valve prolapse (84%) and conduction abnormalities (65.3%), were observed in all age groups. The severity of manifestations increased with the degree of CTD, whereas age differences were more pronounced for scoliosis and cardiac changes.

Conclusion: CTD in children is characterized by a persistent set of musculoskeletal and cardiovascular manifestations across various age groups, with the severity of symptoms correlating with the degree of dysplasia. Early detection and systematic clinical evaluation are essential for identifying risk groups and implementing preventive strategies for potential complications.

Key words. Connective tissue dysplasia, children, musculoskeletal manifestations, cardiovascular abnormalities, age distribution, clinical severity.

Introduction.

The prevalence of various diseases in children is formed under the influence of both genetic factors and environmental conditions, as well as lifestyle characteristics [1-3]. In recent years, there has been an increase in the number of chronic musculoskeletal disorders in children, which is primarily associated with decreased physical activity, postural disorders, unbalanced nutrition, and increased spinal load at an early age [4-6]. However, the presence of genetically determined disorders of connective tissue structure significantly enhances the impact of external factors and creates a higher risk of developing functional disorders and morphological changes.

One of the most common variants of such pathology is undifferentiated connective tissue dysplasia (CTD), characterized by multiple phenotypic features and the involvement of various organs and systems [7]. CTD is widespread among the pediatric population; however, its clinical manifestations vary depending on age, which often complicates timely diagnosis. Of particular importance in children with CTD are musculoskeletal disorders, as well as the high frequency of cardiovascular changes, including mitral valve prolapse, conduction abnormalities, and minor cardiac anomalies [8,9].

Despite the substantial amount of research devoted to hereditary connective tissue disorders, data on the age-related characteristics of the structure of clinical manifestations of CTD, as well as on the distribution of its severity, remain limited. This determines the need for a detailed analysis of the frequency and spectrum of CTD symptoms in children of different age groups.

It is important to note that in international practice, the conditions described as undifferentiated CTD largely overlap with the concept of Hypermobility Spectrum Disorders (HSD) and hypermobile Ehlers-Danlos syndrome (hEDS), according to the 2017 International Classification of the Ehlers-Danlos syndromes [10]. While the term CTD is traditionally used in the studied region to describe a heterogeneous group of connective tissue anomalies that do not fit specific syndromic definitions, the phenotypic spectrum observed—including joint hypermobility, skin hyperextensibility, and tissue fragility—closely mirrors the criteria for HSD/hEDS [11-14].

In this regard, the study of the prevalence of connective tissue dysplasia signs in children, as well as the identification of features of the clinical picture depending on age and the severity of the pathological process, appears to be relevant.

Aim. The aim of the study was to determine the frequency and clinical features of connective tissue dysplasia in children depending on age and the severity of its expression.

Materials and Methods.

The study included 375 children aged 3 to 16 years living in Semey. All children underwent a comprehensive clinical and instrumental examination according to the approved protocol, followed by an assessment of connective tissue dysplasia (CTD) signs.

Inclusion criteria: Children aged 3–16 years.

· Presence of phenotypic and/or functional signs corresponding to the diagnostic criteria for undifferentiated connective tissue dysplasia according to the Russian clinical guidelines "Hereditary disorders of connective tissue structure and function".

· Completion of a full clinical examination, anthropometry, physical assessment, and instrumental evaluation of the cardiovascular system.

· Obtaining informed consent from parents (legal representatives), as well as from adolescents over 14 years old.

Exclusion criteria:

· Presence of chronic inflammatory or autoimmune diseases affecting connective tissue (e.g., juvenile idiopathic arthritis, systemic lupus erythematosus), malignancies, confirmed genetic syndromes; differentiated forms of CTD; congenital heart defects not classified as minor developmental anomalies.

· Refusal of participation by parents or children, incomplete examinations, or low adherence to further follow-up.

Clinical assessment of CTD.

The diagnosis of CTD severity (grades 1-3) was based on a set of external and systemic manifestations, including: asthenic body type; joint hypermobility (according to Beighton criteria); chest deformities; scoliotic posture; dolichostenomelia, arachnodactyly; flat feet; ear pinna anomalies; bite abnormalities and high-arched palate; hernias of various locations.

The severity was graded based on the cumulative sum of stigmatization scores and visceral involvement. Grade 1 (mild) was defined by the presence of minor phenotypic anomalies and subjective complaints without organ dysfunction. Grade 2 (moderate) included a combination of >5 phenotypic signs and functional changes in one or more systems (e.g., mitral valve prolapse with regurgitation). Grade 3 (severe) was characterized by multiple severe phenotypic stigmas (>10) and clinically significant involvement of the musculoskeletal or cardiovascular systems requiring therapeutic intervention.

Instrumental Evaluation.

Echocardiography was performed to assess cardiovascular anomalies. To avoid overdiagnosis of mitral valve prolapse (MVP), strict diagnostic criteria were applied: MVP was defined as a systolic displacement of one or both mitral leaflets by >2 mm beyond the mitral annulus in the parasternal long-axis view, conforming to standard echocardiographic recommendations.

Signs were recorded in standard protocols. Their frequency and severity were assessed according to the degree of detected dysplasia.

Statistical analysis of the results was performed using SPSS version 20 for Windows.

The data processing system included the Romanovsky test to identify random sampling errors and group data by analyzed parameters. To determine the significance of differences in numerical series, the Student t-test or nonparametric methods (Mann-Whitney for independent groups, Wilcoxon for indicator dynamics within a single group) were used. Pearson's $\chi 2$ test [16] was used to analyze contingency tables.

To reject the null hypothesis, the critical significance level was set at $p < 0.05$.

Results.

Table 1 presents data on the overall frequency and age distribution of CTD in the examined cohort.

Overall, this pathology was identified in 75 out of 375 examined children (20.0%). The highest frequency of CTD was observed in the 3-7 years subgroup (21.6%), and the lowest in the 8-10 years subgroup (17.7%), although there were no significant differences between the identified subgroups.

When CTD was distributed by severity, as expected, grade 1 was diagnosed most frequently (12.8% of the total examined), grade 2 was twice as rare (5.9%), and only in isolated cases did CTD reach grade 3 (1.3%).

Table 2 presents the main clinical manifestations of CTD, including their distribution according to severity.

Among the physical signs in children, both on average across the group and by CTD severity, the most frequently observed were joint hypermobility, asthenic body type, ear pinna anomalies, scoliosis (more than 50%), flat feet, and muscle hypotonia (over 40%). Chest deformities, bite abnormalities, arachnodactyly, dolichostenomelia, and hernias of various locations were observed in more than 25% of cases.

In addition to physical signs, cardiovascular abnormalities were studied as the most common and clinically significant. Among these abnormalities, mitral valve prolapses, various intraventricular conduction disturbances, accessory trabeculae, and an accessory chord of the left ventricle predominated. Other manifestations were significantly rarer. For almost all of the above-mentioned signs, the highest frequency was observed in children with grade 3 CTD, and, accordingly, the lowest frequency in grade 1 CTD.

Table 3 presents a similar distribution of CTD symptoms by age of the examined children.

Table 1. *Frequency of CTD in the examined children depending on age.*

Age	CTD					
	1st degree		2nd degree		3rd degree	
	abs. no.	%	abs. no.	%	abs. no.	%
3-7 years, n=116	16	13,8	7	6,0	2	1,7
8-10 years, n=113	13	11,5	6	5,3	1	0,9
11-16 years, n=146	19	13,0	9	6,2	2	1,4
Total, n=375	48	12,8	22	5,9	5	1,3

Table 2. *Main clinical manifestations of CTD and their frequency depending on the severity of dysplasia.*

Age	CTD							
	1st degree, n=48		2st degree, n=22		3st degree, n=5		Total, n=75	
	abs. no.	%	abs. no.	%	abs. no.	%	abs. no.	%
1	2	3	4	5	6	7	8	9
Asthenic body type	25	52,1	16	72,7	5	100,0	46	61,3
Chest deformities	17	35,4	8	36,4	2	40,0	27	36,0
Scoliosis / scoliotic posture	24	50,0	14	63,6	3	60,0	41	54,7
Dolichostenomelia	11	22,9	6	27,3	2	40,0	19	25,3
Arachnodactyly	9	18,8	10	45,5	1	20,0	20	26,7
Flat feet	19	39,6	10	45,5	3	60,0	32	42,7
Ear pinna anomalies	26	54,2	14	63,6	4	80,0	44	58,7
Visible venous network	5	10,4	3	13,6	2	40,0	10	13,3
Bite abnormalities	13	27,1	7	31,8	2	40,0	22	29,3
High-arched palate	7	14,6	5	22,7	1	20,0	13	17,3
Hernias	11	22,9	6	27,3	2	40,0	19	25,3
Joint hypermobility	36	75,0	18	81,8	5	100,0	59	78,7
Muscle hypotonia	20	41,7	10	45,5	3	60,0	33	44,0
Conduction abnormalities	28	58,3	17	77,3	4	80,0	49	65,3
Automaticity disorders	17	35,4	8	36,4	2	40,0	27	36,0
Signs of myocardial hypertrophy and/ or ventricular overload	15	31,3	7	31,8	2	40,0	24	32,0
Mitral valve prolapse	37	77,1	21	95,5	5	100,0	63	84,0
Patent foramen ovale	6	12,5	3	13,6	1	20,0	10	13,3
Accessory chord	28	58,3	14	63,6	3	60,0	45	60,0
Accessory trabeculae	30	62,5	17	77,3	4	80,0	51	68,0
Aneurysms of the interatrial septum	5	10,4	3	13,6	1	20,0	9	12,0
Dilation of the pulmonary artery trunk	3	6,3	3	13,6	0	0,0	6	8,0

Table 3. *Main clinical manifestations of CTD and their frequency depending on the age of the examined children.*

Age	CTD							
	3-7 years, n=25		8-10 years, n=20		11-16 years, n=30		Total, n=75	
	abs. no.	%	abs. no.	%	abs. no.	%	abs. no.	%
1	2	3	4	5	6	7	8	9
Asthenic body type	14	56,0	12	60,0	20	66,7	46	61,3
Chest deformities	8	32,0	8	40,0	11	36,7	27	36,0
Scoliosis / scoliotic posture	10	40,0	11	55,0	20	66,7	41	54,7
Dolichostenomelia	6	24,0	5	25,0	8	26,7	19	25,3
Arachnodactyly	6	24,0	6	30,0	8	26,7	20	26,7
Flat feet	10	40,0	8	40,0	14	46,7	32	42,7
Ear pinna anomalies	14	56,0	12	60,0	18	60,0	44	58,7
Visible venous network	3	12,0	3	15,0	4	13,3	10	13,3
Bite abnormalities	7	28,0	6	30,0	9	30,0	22	29,3
High-arched palate	5	20,0	4	20,0	4	13,3	13	17,3
Hernias	4	16,0	6	30,0	9	30,0	19	25,3
Joint hypermobility	20	80,0	17	85,0	22	73,3	59	78,7
Muscle hypotonia	11	44,0	9	45,0	13	43,3	33	44,0
Conduction abnormalities	14	56,0	12	60,0	23	76,7	49	65,3
Automaticity disorders	9	36,0	7	35,0	11	36,7	27	36,0
Signs of myocardial hypertrophy and/ or ventricular overload	4	16,0	7	35,0	13	43,3	24	32,0
Mitral valve prolapse	22	88,0	18	90,0	23	76,7	63	84,0
Patent foramen ovale	3	12,0	3	15,0	4	13,3	10	13,3
Accessory chord	16	64,0	12	60,0	17	56,7	45	60,0
Accessory trabeculae	18	72,0	11	55,0	22	73,3	51	68,0
Aneurysms of the interatrial septum	4	16,0	1	5,0	4	13,3	9	12,0
Dilation of the pulmonary artery trunk	2	8,0	1	5,0	3	10,0	6	8,0

Note: *P-values are not included as a separate column to maintain table clarity. Statistical significance for age-group comparisons (e.g., using the x2-test) was primarily reported within the results section. All differences not explicitly mentioned as statistically significant in the text were determined to be non-significant across age groups (p > 0.05).*

When distributing the clinical manifestations of CTD according to the age of the examined children, it was found that the average presence of these signs did not differ significantly. Only minor deviations were observed in the frequency of specific features. Asthenic body type was detected slightly more often in children over 10 years old, and chest deformities were more common at ages 8-10 years. The frequency of scoliosis increased significantly with age and was notably higher in children aged 11-16 years compared to those aged 3-7 years. The frequency of dolichostenomelia, arachnodactyly, flat feet, ear pinna anomalies, oral cavity changes, and hernias did not show significant age-related differences. Joint hypermobility was diagnosed slightly more often in the middle age group.

Regarding the cardiovascular system, the frequency of conduction disturbances was higher in the older age group, with significant differences compared to the younger group. In contrast, the frequency of automaticity disorders did not differ across age groups. Signs of myocardial hypertrophy or ventricular overload were significantly more often detected in the middle ($\chi^2 = 7.25$, $p = 0.028$) and older ($\chi^2 = 9.71$, $p = 0.015$) age groups compared to the younger group. The frequency of mitral valve prolapse was high across all age categories, with the highest value observed in the middle age group. No significant differences were found in the frequency of congenital morphological heart changes, such as the presence of an accessory chord or accessory trabeculae. The lower number of children with aneurysms of the interatrial septum and dilation of the pulmonary artery trunk in the 8–10 years group can only be explained by fluctuation; differences between subgroups were also not significant.

Thus, it can be concluded that the studied clinical group of children with CTD is uniform in terms of the frequency and structure of the main features of this pathology.

Discussion.

The results of our study showed that connective tissue dysplasia (CTD) occurs in 20% of the examined children, with grade 1 severity being the most frequently diagnosed and grade 3 being rare. These data are consistent with the observations of other authors: for example, Kadurina et al. noted that mild and moderate forms of CTD dominate among pediatric populations, whereas severe forms occur in less than 5% of children [1,2].

Analysis of clinical manifestations showed a high frequency of joint hypermobility, asthenic body type, ear pinna anomalies, scoliosis, and flat feet, as well as cardiac changes (mitral valve prolapse, accessory chords and trabeculae, conduction disturbances). These results are consistent with the data presented by Braverman A. C. et al., where systemic manifestations, including orthopedic and cardiac changes, were noted in children with CTD [3].

Comparison of the distribution of signs across age groups revealed that most symptoms occur at all ages; however, the frequency of scoliosis and some cardiac abnormalities increases in children aged 11-16 years compared to younger groups. This trend can be explained by the rapid pubertal growth spurt, which places increased biomechanical stress on the compromised connective tissue structures of the spine. In adolescents with CTD, the imbalance between skeletal growth and the strength of the ligamentous-muscular apparatus significantly increases the risk of scoliotic deformities progression. Similar data have been reported in studies by Russian and international authors, indicating an increase in orthopedic and cardiological manifestations with age, despite the stable presence of mild CTD signs in younger age groups [4-6,15].

When comparing by CTD severity, it was found that children with grades 2 and 3 had a significantly higher frequency of systemic manifestations compared to grade 1, which is supported by the literature: pronounced forms of CTD are more often associated with cardiac and orthopedic abnormalities, while mild forms may remain unrecognized until older age [7,8].

Thus, the obtained results not only confirm previous research findings but also complement them, demonstrating the uniformity of the clinical picture of CTD in terms of the frequency and structure of signs across different ages, as well as showing the consistent increase in severity of manifestations with higher CTD grades. This underscores the need for early detection of mild forms of the pathology and systematic monitoring of all children with signs of dysplasia, regardless of age and symptom severity [9,10].

Comprehensive assessment and early diagnosis of CTD allow not only the optimization of monitoring for children with mild and moderate forms of the pathology but also the planning of preventive and therapeutic interventions, including correction of muscle hypotonia, support of the musculoskeletal system, and monitoring of cardiovascular changes [11-13].

This study has several limitations. First, it is a single-center cross-sectional study, which limits the assessment of causal relationships and long-term prognosis. Second, the diagnosis was primarily clinical and instrumental; genetic testing to exclude rare hereditary syndromes was not performed for all participants due to resource constraints. Finally, the absence of a healthy control group limits the ability to compare the prevalence of minor anomalies with the general population.

Conclusion.

Connective tissue dysplasia was identified in 20% of the examined children aged 3–16 years and is predominantly characterized by a mild grade. The clinical picture includes multiple orthopedic and cardiac signs, which are present in all age groups but increase with higher severity and age, highlighting the need for early diagnosis, systematic monitoring, and a comprehensive approach to treatment.

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None of the authors have any conflicts of interest to disclosure.

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