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Original Article

Prognostic research of changes in the phenotypic features in the musculoskeletal apparatus in children with joint hypermobility that require physical rehabilitation

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Abstract:
The study explores the specifics of local involvement of structures of musculoskeletal apparatus in dysplastic process. The researchers estimated the significance of external and visceral phenotypic features for further development of pathologic process in children with joint hypermobility as one of the main manifestations of undifferentiated connective tissue dysplasia. It is established that the examined cohort have physiologic joint mobility as well as mild, medium or severe degree of joint hypermobility.
The research of prognostic significance of correlation patterns of certain features of joint hypermobility in children enabled constructing conceptual contextual and abstracted mathematical model.
It is established that most indicators of physical development are significant factors of phenotypic manifestation of joint hypermobility in children aged 4–6, but the most dominant are: body length, chest circumference, body mass, foot length and arm span. While the influence of such factors as platypodia, hand length, postural kyphosis and scoliosis is less significant compared to that of the indicators mentioned above.
Key words: joint hypermobility, phenotypic feature, musculoskeletal apparatus, undifferentiated connective tissue dysplasia.

Introduction

Currently, studying the state of health of child population has become one of the most important tasks for researchers. The results of the research indicate the increase in chronic pathologies and negative tendencies in morphofunctional development of children [3, 5, 9, 13, 22].

At the present time, the problem of musculoskeletal apparatus deformities in children becomes more and more exacerbated. It is established that 79–86% of children with deformities of musculoskeletal apparatus have undifferentiated connective tissue dysplasia, caused by deficiency of collagen synthesis and abnormality of connective tissues of human body. These types of defects were first diagnosed during neonatal period but prominent changes in musculoskeletal apparatus appeared at the age of 3. It is acknowledged that the difficulties of studying pathogenesis of undifferentiated connective tissue dysplasia caused by complex mechanism of defects at tissue, organ and organism levels in polysystemic pathology [1, 4, 8, 11, 18].

Unique feature of connective tissue lies in its mesenchymal origin, it makes up for half of the body mass and functions as an integrative system. Professionals in the field of Pediatrics, Physiology and Orthopedics choose rehabilitation and prevention of dysplasia based on specifics of its corresponding type. Variety of the approaches to the problem of undifferentiated connective tissue dysplasia in children that professionals use complicates the diagnostic process and decreases the effectiveness of applying complex preventive and rehabilitation activities [2, 6, 12, 20, 25].

Children with different phenotypes of undifferentiated dysplasia have apparent polymorphism of clinical-morphological deformities. At that morphologic changes in connective tissue are mostly stereotypical. It is worth noting that, undifferentiated connective tissue dysplasia includes a series of pathological, intermediate and background states, which is defined as genetic-heterogenous syndrome of polygenous-multifactor nature, with external phenotypic features of dysplastic defects in connective tissue and clinically significant dysfunction of one or several internal organs. The mentioned states are often characterized by autosome-dominant type of inheritance, as well as different set of qualitative and quantitative symptoms [7, 10, 14, 21].

The following types of undifferentiated connective tissue dysplasia are singled out: MASS-phenotype, Marphanoid, Ehlers-Danlos-like phenotypes. For Maphanoid phenotype the following characteristics are common: asthenic constitution type, dolichostenomelia, arachnodactyly, pathological changes in musculoskeletal apparatus, valvular heart disease and vision disorders. The case of undifferentiated dysplasia of Ehlers-Danlos-like phenotype is characterized by a combination of numerous symptoms of damage with manifestations of joint hypermobility [14, 19, 21, 23].

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However, the scholars all over the world find the moderate manifestations of joint connective tissue dysplasia the most interesting, namely joint hypermobility, which is the main phenotypic feature of undifferentiated connective tissue dysplasia and is mostly characterized by increased mobility volume in one or several joints compared to average norm and is encountered with 50–72.2% frequency among child population [1, 2, 20, 24].

It is generally known that morphofunctional state of children's organism is an integral indicant of health. Specifics of functional properties of organism determine the degree of prepathologic changes. For this reason, prognostic analysis of phenotypic features, which characterize morphofunctional state of organisms in children with joint hypermobility is an important factor in the process of designing programs in physical rehabilitation [16, 22, 24, 26].

The aim of the research is to study and investigate phenotypic features in order to make prognoses of local involvement of structures of musculoskeletal apparatus associated with joint hypermobility in children.

Material & methods

The study involved 446 children between four and six years of age (43.60 ± 2.81% boys and 56.40 ± 2.80% girls). The study plan included: 1) copying data from a comprehensive medical examination; 2) estimation of joint hypermobility by goniometry method and evaluating results according to the criteria for diagnosing joint hypermobility and its degree by the method of S. Carter, J. Wilkinson in modification of R. Beighon using a medical goniometer. In the role of criterion components, the following properties were examined: passive bending of both thumbs to the forearm, passive hyperextension of little fingers at 90º, hyperextension of elbow joints at 10º, hyperextension of knee joints at 10º, and touching the ground while bending forward with straight knee joints, where each of the properties were valued at 1 point. The total of 0–2 points indicated the absence of joint hypermobility, 3–4 points indicated its mild degree, 5–8 – the apparent (medium) degree of joint hypermobility, 9 points – the generalized form (evident degree) of joint hypermobility.

Anthropometric examination of children involved using classic methods and standard instruments for determining length, body mass and chest circumference.

The length of lower segment of body was determined in initial standing position as the distance between pubic symphisis and foot base along the midline. The length of the upper segment was defined as the difference between the body length and lower segment of body. Measuring arm span was carried out in the initial standing position. The distance between the fingertips of right and left hands was measured using centimeter ribbon. The hand length was measured as the distance between the tip of middle finger and distal epiphyses of redial bone. The foot length was measured in initial standing position as the distance between the edge of heel bone and phalanges of the longest finger. The main part of mathematical processing was carried out using standard package STATISTICA 8.0. Mathematical processing of numeric data was carried out by methods of differential statistics with calculating arithmetic mean ($\bar{x}$) and mean error (m). The difference between sample groups in case of comparison to their average, that are divided according to normal law, were assessed according to parametric criteria of Student (t). The probability of differences was assessed according to the level of significance (p) at the level $p<0.05$. The correlation of anthropomorphic parameters as active factors in manifestation of dysplastic abnormalities and joint hypermobility was established using regressive analysis.

The prognosis of functional changes in musculoskeletal apparatus in children with joint hypermobility was made using discriminant analysis, the researchers determined the informative variables that may influence further complication of musculoskeletal apparatus on the background of joint hypermobility and the classification functions with calculation of specificity and sensitivity of each parameter for every variant of results separately. The research was carried out in accordance with the plan of research work of the Chair of Medical and Biological Foundations of Physical Culture at Sumy State Pedagogical University named after A.S. Makarenko "Physiologic and Hygienic Support of Health-Preserving Activity of Educational Establishments" Number of state registration 0113U004662.

Results And Discussion

Upon careful examination the following results were obtained: 14.45±3.76% of the examined cohort have physiological joint mobility, mild joint hypermobility was detected in 27.73±3.46% of children, medium joint hypermobility was found in 45.18±3.01% of subjects and 12.62±3.80% of children possess severe joint hypermobility.

One of the specific features of morphofunctional development of children with manifestation of undifferentiated connective tissue dysplasia are external and visceral phenotypic characteristics, determining which enables to diagnose local involvement of structures of musculoskeletal apparatus in dysplastic process. The dolichostenomelia is one of the main features that determine proportionality of correlation between the length of body segments. According to the obtained results, it is established that disharmonic proportions in hand length, foot length and body length ratio in children with joint hypermobility were the most common (51.16±2.03% and 59.47±2.01% respectively ($\bar{x}$ ±m)) (Table 1).

The researchers observed age specific differences in disharmonic hand length to body length ratio, that was found in 42.12 ± 3.42% (± m) of children aged 5 and in 34.02 ± 2.96% of children aged 6 with apparent degree

of hypermobility irrespective of sex. The abnormalities in proportions of upper and lower body segments were found in 24.42 ± 1.75% (± m) of children with articular manifestations, among them the highest proportion of children aged 6 (38.94 ± 2.99%), mainly boys, 1.5 times more as compared to girls (59.97 ± 4.04% and 40.03 ± 4.04%, respectively, p <0.05). The change in the ratio of arm span to body length in 13.62 ± 1.39% (± m) of the examined children confirms the fact that children with dysplastic disorders have a predisposition to disproportionate development of musculoskeletal system.

Table 1

Distribution of Children with Joint Hypermobility Based on Signs of Dolichostenomelia

Signs of dolichostenomelia	Proportion of children with joint hypermobility and dolichostenomelia, %									
	physiological joint mobility *n*=65		mild joint hypermobility *n*=124		medium joint hypermobility *n*=199		severe joint hypermobility *n*=58		Total *n*=446	
	$\bar{x}$	*S*	$\bar{x}$	*S*	$\bar{x}$	*S*	$\bar{x}$	*S*	$\bar{x}$	*S*
Arm span / Body length	0.17	0.44*	1.66	0.98*	10.63	1.87	1.16	1.22	13.62	1.39
Hand length/ Body length	1.51	1.31	2.49	1.21	40.70	2.98 **	6.48	2.82 **	51.16	2.03
Foot lenght/ body length	1.33	1.23	5.98	1.83●	46.51	3.02●	5.65	2.64	59.47	2.01
Upper body segment/ lower body segment	0.83	0.97	2.66	1.24	18.60	2.36	2.33	1.73	24.42	1.75

Note:

* probable difference in disharmonic arm span to body length ratio in children with physiologic and mild degree of joint hypermobility t=2.03 (p<0.01);

** probable difference in disharmonic hand length to body length ratio in children with medium and severe degree of joint hypermobility *t*=3.01 (p<0.05);

• probable difference in disharmonic foot length to body length ratio in children with mild and medium degree of joint hypermobility *t*=2.04 (p<0.05).

The research of prevalence rate of osteoarticular manifestations of connective tissue changes in musculoskeletal apparatus in children helped to identify the following peculiarities: firstly, chest deformation was detected in 5.98±0.96% *($\bar{x}$ ±m)* of children with joint hypermobility and only 0.66±0.86% of children with physiological joint mobility (*p*<0.05); secondly, postural kyphosis was detected in 35.88±1.24% of children with joint hypermobility, and 0.17±0.16% of examined children without joint hypermobility (*p*<0,05); thirdly, similar tendency was observed in children with postural scoliosis (14.29±1.42% and 0.18±0.17%, respectively, *p*<0.05); fourthly, platypodia was detected in 64,46±% and dental and jaw abnormalities in 58,81±2.01% of examined children with joint hypermobility of different degrees (Figure 1).

To prevent further progressing of pathologic changes it is necessary to study prognostic meaning of change in ratio of singular parameters by means of mathematical construction that adequately reflects features and specifics as well as creates schematic in content and abstracted model, the calculation will enable scholars to determine the specter and impact of each and every factor on development of main pathologic change. For this reason, using regressive analysis enabled constructing of the model in several stages.

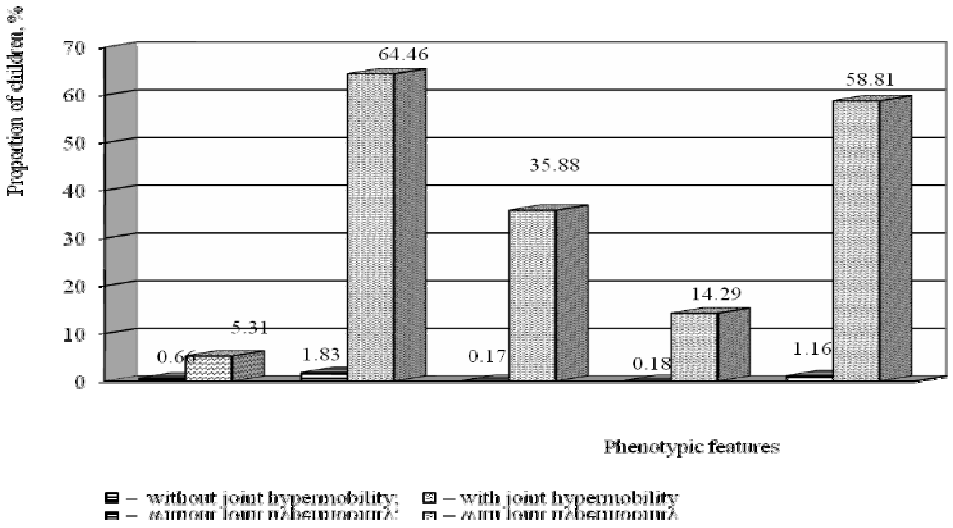

Figure 1. Percentage of Children with Apparent Osteoarticular Phenotypic Features, % (*n*=446)

At the initial stage the factors, which probably have no influence on joint manifestation in children were eliminated. After calculating multiple changes, namely body mass (BM), body length (BL), chest circumference (CC), postural kyphosis (PK), postural scoliosis (PS), platypodia (P), foot length (FL), hand length (HL), arm span (AS), their accuracy was verified (*p*-level), error index (Sβ) and the sign coefficient were defined (*a*). Besides, using Fisher's criterion the adequacy of the equation was defined, and ranging and comparing different parameters was based on percentage of impact.

The next stage included constructing regressive model presented below:

$y=a_0+a_1\text{BM}+a_2\text{BL}-a_3\text{CC}-a_4\text{PK}+a_5\text{PS}-a_6\text{P}+a_7\text{FL}-a_8\text{HL}+a_9\text{AS}$,

The obtained numeric characteristics of coefficients $(a_0, a_1,\ldots,a_9)$ reflect laws of correlation of various factors.

y=4,52+0,07BM+0,07BL-0,12CC-0,003PK+0,24PS-0,19P+0,23FL-0,29HL+0,005AS,

(F=4,41, p<0,001).

Upon analysis of the main parameters of regressive model the percentage of contribution of each factor was calculated, thus, the impact of each factor, which influences parameters of physical development of children with joint hypermobility, was analyzed. According to the results of data processing, it was established that the most important indicators of physical development are the main factors of phenotypic manifestation of joint hypermobility in children, but the most prevalent are: body length (41.71%), chest circumference (29.27%), body mass (10.08%), foot length (8.75%), arm span (6.45%) (Figure 2).

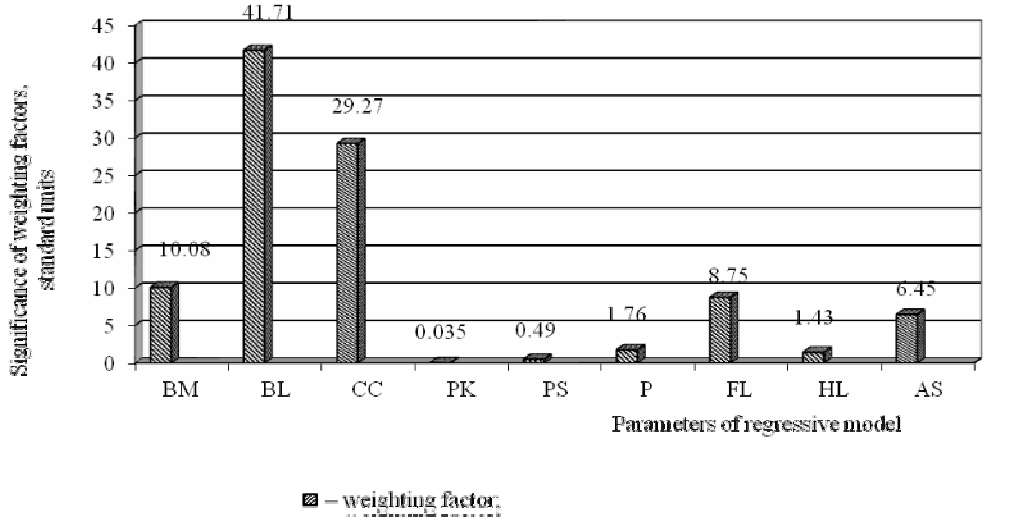

Figure 2. Significance of Weighting Factors of Characteristics of Regressive Model, standard units:
BM – body mass, BL – body length, CC – chest circumference, PK – postural kyphosis, PS – postural scoliosis, P – platypodia, FL – foot length, HL – hand length, AS – arm span.

Conclusions

The influence of such factors as platypodia, hand length, postural kyphosis and scoliosis on manifestation of joint hypermobility in children is less significant, but they too have prognostic significance, since the level of the accuracy of the regressive analysis results is relatively high.

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