GALAXY INTERNATIONAL INTERDISCIPLINARY RESEARCH JOURNAL (GIIRJ)
ISSN (E): 2347-6915
Vol. 12, Issue 2, February (2024)

DIAGNOSTICS AND FEATURES OF THE FLOW OF HYPERMOBILITY SYNDROME IN CHILDREN

Dzhuraev Akhrorbek Makhmudovich
Dr. M.Sc., Professor, Head of the Department of General Orthopaedics

Alimukhamedova Feruza Shavkatovna,
Phd, Junior Researcher, Traumatologist-Orthopaedist, Author
Republican Specialised Scientific and Practical Medical Centre of Traumatology and Orthopaedics, Tashkent, Uzbekistan

ABSTRACT

Scientific and practical interest in joint hypermobility dates back to the late nineteenth century, when hereditary syndromes were described in the clinical picture of which, joint hypermobility was one of the leading symptoms. In 1967, Kirk (J. H. Kirk), Ansell (B. M. Ansell) and Bywaters (E. G. Bywaters) proposed the term "hypermobile syndrome" to characterize the pathology in patients with hypermobile joints and persistent musculoskeletal complaints in the absence of any other rheumatic disease. Since then, systematic study of this pathology within the framework of rheumatologically and orthopedic syndromes has begun

Keywords: joint, hypermobility, connective tissue, dysplasia, epidemiology.

INTRODUCTION

Hypermobility syndrome (HS) is a systemic connective tissue disease characterised by joint hypermobility (JHM), combined with musculoskeletal complaints or internal and external phenotypic signs of connective tissue dysplasia, in the absence of any other rheumatic disease. Today, thanks to advances in genetics, more than 200 hereditary connective tissue and skeletal diseases have been described and classified. The study of this pathology of the musculoskeletal apparatus has determined a number of unresolved issues concerning age and sex boundaries of hypermobility, the state of the musculoskeletal apparatus in persons with hypermobility of joints, clarification of criteria of hypermobility syndrome and its prevalence, direct causes of joint pain in hypermobility syndrome, as well as practically unexplored social significance of the problem as a whole, determine the urgency of the problem. importance of the problem as a whole, determine the relevance of the undertaken epidemiological and clinical-instrumental study.

RELEVANCE

Joint hypermobility syndrome is a condition of connective tissue explaining its increased elasticity and manifested by a combination of several musculoskeletal conditions to a large symptom complex affecting different systems of the human body. The term "hypermobile syndrome" reflected the phenomenon of joint hypermobility combined with musculoskeletal dysfunction (subluxations, arthralgias).

Later, due to the absence of life-threatening complications in joint hypermobility syndrome, the term "benign joint hypermobility syndrome" was used, and then the term "joint

hypermobility syndrome" became established. It is believed that joint hypermobility is a genetically determined condition with dominant inheritance, which is predominantly transmitted through the female line (Belenky A.G. 2004, Grahame, R. Rheumatol.J. 2000). The morphological substrate underlying the pathological features in the above syndrome is a change in the structure and ratio of different types of collagen and, as a consequence, its greater extensibility than in the norm (Grahame, R. 2000.).)., "hereditary collagenopathy" (Kadurina, T.I. 2000) growth pain syndrome (given the peak incidence at the age of 13-16 years) and simply arthralgia's of unclear genesis, which reflect the systemic involvement of connective tissue.

It is not uncommon for persons with hypermobile joints to have no musculoskeletal complaints for a long time (sometimes their whole life). This condition is defined as constitutional joint hypermobility and is a variant of the norm (Bird H., 1986). Such symptomatic individuals often successfully utilise their innate characteristics in sports (gymnastics) or professions (dance, ballet). The quality of life of most individuals with joint hypermobility is often far from satisfactory (Viktorova I.A., 2008, Grahame R., 2000, Hakim A.J., 2002). In most of them at a certain stage of life appear pathologies of musculoskeletal system, to a pronounced degree forming the clinical picture of hypermobility syndrome. Appearance and intensification of symptoms (usually arthralgia) is possible at any age (Belenky A.G. 2004, Grahame, R. Rheumatol.J. 2000.), which depends on both subjective (L.I. Benevolenskaya, O.M. Lesniak, 2005) and objective factors, among which the most frequent is the violation of the established balance between the load and initially reduced threshold of "mechanical strength" of the musculoskeletal system (Belenky A.G. 2004, Grahame, R. Rheumatol.J. 2000). Some researchers point to the important role of environmental disadvantage in the occurrence of this pathology (V.G. Rebrov, O.A. Gromova, 2003).

The most frequent reason for seeking medical help for persons with hypermobility is problems with the musculoskeletal system (Kadurina, T.I. 2000, Yakovlev, V.M. 2001), which led to the spread among rheumatologists (mainly in Great Britain) of a synonym for hypermobility syndrome - "syndrome of benign familial hypermobility of joints". But it is obvious that this definition does not fully reflect the essence of the described pathology. Manifestations of hypermobility syndrome are by no means limited to problems with the musculoskeletal system. Hypermobility of joints is naturally associated with extra-articular signs of "failure" of connective-tissue structures: prolapse of heart valves, skin hyperextensibility, traumatism, hernia, vein pathology, ptosis of internal organs, and others. Extra-articular signs (including phenotypic) of connective tissue lesions in persons with hypermobility are so typical that in international practice the definition "hypermobile syndrome" is accepted, which according to the modern classification belongs to the group of non-inflammatory diseases of connective tissue (code M 35.7. according to ICD-10) (ICD 10., 1995). In addition, according to the literature, predisposition to excessive joint flexibility is inherited, often from the mother. Carriers are gene mutations that interfere with collagen and tenaxin X formation.

Collagen and tenaxin are the building blocks of the ligament-tendon apparatus and synovial capsule, the excess or deficiency of which leads to the development of connective tissue dysplasia. However, not every parent with a gene defect has a child with hypermobile joints, as connective tissue dysplasia develops under the influence of many other genes. In addition,

unique combinations of alleles of both parents are of significance. (Belenky A.G. 2004, Kadurina, T.I. 2000, Yakovlev, V.M. 2001)

Since the description of hypermobile syndrome a third of a century ago, there has been a significant evolution in the view of the syndrome - from the view of the syndrome as a curiosity rather than a disease to a recognised nosological form (ICD 10., 1995, Belenky A.G. 2004, Grahame, R. Rheumatol.J. 2000). The initial attitude to the syndrome as a benign condition, often not requiring treatment, has changed markedly (Belenky A.G. 2004, Grahame, R. 2000.). The development of a unified clinical method for assessing the degree of joint hypermobility Carter, Wilkinson (1964), later modified by P.Beighton (1973), is an undoubted achievement in the study of joint hypermobility syndrome. The limits of the average (normal) degree of joint mobility for the European population have been determined, joint and extra-articular manifestations of the syndrome have been described (Viktorova I.A., 2008, Grahame R., 2000, Hakim A.J., 2002). Epidemiological studies have revealed sex, age and ethnic differences in the degree of joint mobility. Russian researchers Korshunov N.I., Gauert V.R. (1996,1997), Maslova E.S. (2002), made a significant contribution to the development of knowledge about hypermobility syndrome in Russia and determined the relevance of this problem for Russian rheumatology.

Diagnostic criteria for hypermobile syndrome have been proposed, which allow to establish this diagnosis with a certain degree of reliability,

However, taking into account the frequency of revision of the diagnostic criteria and the absence of pathogonomous symptoms among the manifestations of hypermobility syndrome, the process of refining the diagnostic criteria of the syndrome cannot be considered complete. In particular, the latest "Brighton" criteria do not suggest a differentiated approach to individuals of different ages and sexes, whose differences are obvious, and do not take into account the phenotypic features of connective tissue dysplasia associated with joint hypermobility.

The significance of phenotypic signs of connective tissue dysplasia for pathology (mainly neurological) was developed in the concept of "dysraphic status" (Viktorova I.A., 2008, Hakim A.J., 2002). Although the list of signs of dysraphic status did not explicitly mention hypermobility, the frequent presence in the motor apparatus could certainly be related to hypermobility. Studies of dysraphic status, despite their obvious practical significance, did not lead to the creation of a coherent system of views on the problem and to the development of generally recognised criteria for the disease (or group of diseases). The reason for this situation was the uncertainty of the definition (different authors included in the list of signs of dysrhaphy status various manifestations in terms of severity - from gross malformations to paresthesias), which did not allow to give practical health care recommendations on the diagnosis of the described condition. Currently, the term "dysraphic status" is practically not used in the scientific medical literature.

PURPOSE OF THE STUDY

Analysis and synthesis of literature data, clinical and diagnostic features of hypermobile syndrome in children.

MATERIAL AND METHODS OF THE STUDY

The material for the study are 58 patients of children diagnosed with hypermobile syndrome, treated by RSNPMCTO in the department of consultative polyclinic and general orthopaedics from 2022 to 2023, aged from 1 to 18 years. Of which, boys 21 (36.0 %) and girls 37(64.0 %).
To diagnose hypermobility syndrome in children we applied Beighton's criteria:
- passive flexion of the metacarpal joint of the V finger by 90° to both sides (1-2 points, Fig. 1A);
- passive flexion of the I finger towards the forearm while flexing in the wrist joint (1-2 points);
- overextension of both elbow joints > 10 degrees (1-2 points);
- overextension of both knee joints > 10 degrees (1-2 points);
- when bending forward with the knee joints fixed, the planes of the patient's palms touch the floor (1 point).

Major criteria:
- Beighton's scale score of 4 out of 9 or more (at the time of examination or in the past).
- Arthralgia for more than 3 months in 4 joints or more.

Minor criteria:
- Beighton's scale score of 1-3 out of 9 (0-2 for people over 50 years old).
- Arthralgia in 1-3 joints or lumbalgia for more than 3 months,
- Presence of spondylolysis, spondylolisthesis.
- Dislocations/subluxations in more than 1 joint or recurrent dislocation in one joint.
- Periarticular lesions of more than 2 localisations (epicondylitis, tenosynovitis, bursitis, etc.).
- Marfanoidism (tall stature, thinness, arm span/height ratio more than 1.03, upper/lower body segment ratio less than 0.83, arachnodactyly).
- Abnormal skin: thinness, hyperextensibility, striae, atrophic scars.
- Ocular signs: overhanging eyelids or myopia.

Our observations have shown that the physical examination of Beighton, as well as other methods of examination complementary to each other helps to differentiate other diseases from hypermobility syndrome in children.

RESULTS AND DISCUSSION

Despite the current advances in the study of hypermobile syndrome, many questions relating to various clinical and epidemiological aspects of the syndrome remain unresolved. Thus, the direct cause of joint pain in hypermobility syndrome is unclear; mechanical causes, cytokine imbalance, altered proprioceptive sensitivity, autonomic nervous system dysfunction, hypermobility-associated anxiety and the common pathogenetic basis of pain syndrome with fibromyalgia are discussed. The fact that there is no direct relationship between the degree of joint hypermobility and the presence and clinical severity of pain syndrome, especially among children and adolescents, has not been explained.
The connection between orthopaedic anomalies (scoliosis, flat feet) and joint hypermobility is discussed in single works. There are no data on instrumental characterisation of the state of articular and near-articular soft tissues in hypermobility syndrome. There is still no unified

view on the problem of joint hypermobility and related organ pathology among specialists of different profiles.

The prevalence of hypermobility syndrome (including in Uzbekistan) remains an unresolved issue, which until now could be judged only by indirect signs. For example, a practising rheumatologist in the UK diagnoses 30-40 first-time patients with hypermobile syndrome on average per year; unfortunately, this report does not provide data on the total number of patients treated. When analysing the reasons for admission to major rheumatology clinics, the diagnosis of hypermobile syndrome is established on examination in 3-9% of cases. Obviously, these data do not fully reflect the true prevalence of hypermobility syndrome, as many patients are not diagnosed; patients may be seen with extra-articular problems by other specialists (phlebologists, cardiologists), or with pronounced orthopaedic pathology become patients of operating surgeons.

Unresolved issues concerning the age and sex boundaries of hypermobility, the state of the musculoskeletal system in persons with joint hypermobility, clarification of the criteria of hypermobility syndrome and its prevalence, the immediate causes of joint pain in hypermobility syndrome, as well as the practically unexplored social significance of the problem as a whole, determine the relevance of the epidemiological and clinical-instrumental study of joint hypermobility and hypermobility syndrome in the children's contingent in the Republic of Sakha (Yakutia).

CONCLUSION

Data analysis from domestic and foreign literature sources showed that the incidence of hypermobile syndrome is not equal in different age groups of children. It should be noted that hypermobile syndrome is, as a rule, of progressive nature and underlies the formation of somatic pathology, which often comes to the forefront and determines the prognosis of the main disease. As a result of the study, the prevalence of hypermobile syndrome in children in the country was determined. Applying the Beighton criteria we determine the syndrome of this disease and the degree of hypermobility in patients. Timely determination of joint hypermobility plays an important role in the treatment and prevention of potential complications of this disease and its orthopaedic pathology.

REFERENCES

1.Abbakumova L.N., Kadurina T.I., Novik G.A., Filippova Y.N., Ghimbovschi S. Clinical polymorphism and frequency of occurrence of del30kb haplonedeficiency of the tenascin gene in children with joint hypermobility syndrome // Clinical Genetics in Paediatrics. - 2016. - C. 193-198.

2. Belenky A.G. Joint hypermobility and hypermobile syndrome: prevalence and clinical and instrumental characteristics. Avtoref. disc. doctor of medical sciences. - Moscow.- 2004. - c.1-50.

3.Verbenko V.A., Soiko V.V., Borzunov A.V. Features of mental and behavioural disorders in children with hypermobile syndrome // Psychiatry. - T.19. - № 3 (72). - 2015. - C. 22-26.

4. Viktorova I.A., Ivanova D.S., Konshu N.V. Syndrome of joint hypermobility: differential diagnosis with rheumatological diseases // Medical Bulletin of the North Caucasus. - 2016. - T. 11. - № 2. - Vyp. 2. - C. 312-315.
5. Glodeva V.S. Hypermobility syndrome: increased flexibility in gymnasts, Municipal budgetary institution of additional education "Children's and Youth Sports School No. 4" 2019 г.
6. Kadurina T.I., Abbakumova L.M. Dysplasia to connective tissue: Path to diagnosis // Vestn. of Ivanovo Medical Academy. - 2014. - T. 19. - № 3. - C. 5-11.
7.Спивак Е.М. Синдром гипермобильности суставов у детей и подростков. - Ярославль, 2003. - 128 с. Лесли Лоренда Николсон, Клифтон Чан, Луиза Тофтс, Верити Пэйси "Синдромы гипермобильности у детей и подростков: Оценка, диагностика и междисциплинарное ведение"// Королевский австралийский колледж врачей общей практики 2022/ стр 409-414;
8.Marco Castori, Brad Tinkle, Howard Levy, Rodney Grahame, Fransiska Malfait, and Alan Hakim, "A Framework for the Classification of Joint Hypermobility and Related Conditions", American Journal of Medical Genetics Part C (Seminars in Medical Genetics) 175C:148-157 (2017);
9.Франциска Мальфайт, Клер Франкомано, "Международная классификация синдромов Элерса-Данлоса 2017 года", Американский журнал медицинской генетики, часть С (Семинары по медицинской генетике) 175C:8-26 (2017).