Characterization of Brazilian Children with Joint Hypermobility

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Abstract

Introduction: Joint hypermobility (JH) may be the extreme of normal range of motion or one condition in the polygenic group of inherited disorders of connective tissue. It is found in about 40% of the population depending on which variables are used for its classification. It may be associated with musculoskeletal and extra skeletal disorders in which case it is called Ehlers-Danlos Syndrome-hypermobility type.

Objective: To investigate JH in a Brazilian sample of school children using the variables of the Beighton score and possible correlations between these variables and with selected variables of the Brighton score. Additionally, to study the effect of gender, ethnicity, playing of musical instruments such as the piano, and regular physical activity on each of the analyzed parameters.

Method: In 2013, a study was performed of 1749 physically normal and apparently healthy 7 to 12 year old pupils in government schools, irrespective of gender. Variables of the Beighton score (JH of the fifth finger, wrist, elbow, knee and trunk) was used to identify children with JH.

Results: JH was detected in 41% of the children. Of the children with JH, about 60% were female and 55% were Caucasians. Hyperextension of the fifth finger was the most common feature, followed by apophyse of the thumb and hyperextension of the elbow; joint pain was reported by 34.6% and 10.6% used eyeglasses.

Conclusion: The prevalence of JH in Brazilian children is similar to a study in a Chilean population; most are female and Caucasians. There are high errates of JH of the upper limbs, which is important for children who play musical instruments. There is an association between hyperextension of the knee and apposition of the thumb and also between hyperextension of the knee, apophyse of the thumb, hyperextension of the 5th finger and hyperextension of the elbow. Further population studies of children in Latin America are needed.

Keywords

Hypermobility, Child, Flexibility, Ehlers-Danlos syndrome

Introduction

Joint hypermobility (JH) may be the extreme of the normal range of motion or one condition in a polygenic group of inherited disorders of connective tissue [1,2]. It is generally found in 9–40% of the population depending on the variables used for its classification [3–7]. An exception was found in the study of 1120 preschool-age children with 64.6% of the infants being classified as hypermobile [8].

JH is not an illness but a condition with the incidence depending on age, gender and ethnicity; it is more common among women and children [9] and is often transmitted vertically by an affected mother [10]. JH is also more frequent in Blacks [11] than in Caucasians [12].

According to Grahame, in Bravo [13], JH is unknown to most healthcare professionals, as is the occurrence of damage to multiple organs, because the clinical picture does not show signs of inflammation, and there are no laboratory tests nor specific radiological evidence to define the condition. Even so, this condition is considered the commonest cause of pain in rheumatic patients. Musculoskeletal pain is mentioned in the diagnostic criteria and described as debilitating in adults with Ehlers-Danlos Syndrome hypermobility-type (EDS-HT).

The identification of individuals with generalized JH is made using the Beighton score [9], but the clinical effects are poorly understood and often overlooked by rheumatologists worldwide [14] as are associations with other signs and symptoms that characterize EDS-HT. Also called Ehlers-Danlos Syndrome type III or JH Syndrome, EDS-HT is an under diagnosed disorder of connective tissues. This condition occurs due to genetic changes of collagen that generate musculoskeletal and extraskeletal lesions resulting in difficulty to transmit muscle strength [7,14] which leads to hernias, varicose veins, genital or rectal prolapse and myopia [7,15,16].

This condition is emerging as a generalized disorder with implications for virtually all organs and systems suggesting that the consequences are much more wide-ranging than just the clinical implications of loose joints; this is different to the classical idea that it is a benign and asymptomatic condition. EDS-HT is currently a major challenge for the public health system [17].

Clinical diagnostic criteria and tests define many hereditary disorders of connective tissue. However, the diagnosis of EDS-HT is based on a physical evaluation and family history [18]. The most commonly used criteria to define this syndrome are those of the Brighton score [19]; diagnosis is made by the presence of two major criteria or one major criterion and two minor criteria or four minor criteria; two minor criteria are sufficient, when a first-degree relative is unmistakably affected [19].
According to Bravo [7], individuals with EDS-HT can present with an atypical triangular face, irregular and a typical ears, thin nose, tapered chin, anti-mongoloid slant and blue sclera, and they can thus be recognized as one recognizes a person with Down syndrome.

It is known that string musicians and piano players with this condition have musculoskeletal problems [20], as do individuals participating in certain physical activities, such as dancers [21].

Objective

The objective of this study was to characterize a Brazilian sample of school children in respect to JH using the variables of the Beighton score and to investigate possible correlations between these characteristics and with selected characteristics of the Brighton score. Additionally, to study the effect of gender, ethnicity, playing of musical instruments and regular physical activity on each of the analyzed parameters.

Patients and Methods

The study sample consisted of 1749 physically normal and apparently, healthy pupils of government schools in São José do Rio Preto, São Paulo, irrespective of gender. The ages of the children ranged from seven to 15-years-old with a mean age of 9.7 years (SD ± 1.8) and 976 (55.8%) were girls.

Data were collected in the months of May through September 2013. All participants were informed about the nature of the study and signed informed consent forms before being interviewed and participating in the survey.

JM was assessed for the fifth finger, wrist, elbow, knee and trunk and the Beighton score (1973) was used to characterize generalized JM. The angular values and bilateral analyses were obtained using a goniometer except for anterior trunk flexion. One point was assigned for each hypermobile joint thus the maximum score was nine with individuals with a score of four points or more being classified as hypermobile.

The children were also assessed using specific criteria of Brighton (2000) for EDS-HT including joint pain, marfanoid habitus (height/ arm span ratio < 0.89 or > 1.04), and their use of eyeglasses. Moreover, whether the children played string instruments and performed regular physical exercises was investigated, as were possible correlations between the variables of the Beighton score, and correlations between the variables of the Beighton score and the specific variables of the Brighton score.

This study used inferential statistical analysis, and the chi-square and Spearman correlation tests. Categorical variables are presented as absolute frequencies and percentages. Data were analyzed using the GraphPad INSTAT (version 3.0) and Prism (version 6.01) computer programs. The study was approved by the local Research Ethics Committee (n° 3757/2011).

Results

The results of this study show that of the total sample, 719 (41%) children had generalized JH (p-value < 0.0001). Of those with JM, 426 (59%) were female, 395 (55%) were Caucasians, 233 (32%) mulattos, 60 (8%) Blacks, 10 (1.5%) Asian descendants and 6 (0.8%) were Amerindians with the ethnicity of 15 individuals (2.7%) unknown.

An analysis of the variables of the Brighton score showed a higher prevalence of hyperextension of the fifth finger (986 cases-56.4%), followed by opposition of the thumb (834 cases-47.7%), hyperextension of the elbow (612 cases-35%), hyperextension of the knee (426 cases-24.4%) and anterior trunk flexion (121 cases-6.9%).

Of the total sample, 185 (11%) children had specific diagnostic criteria of the Brighton score for EDS-HT, including Beighton scores greater than or equal to four, joint pain, marfanoid habitus and the use of eyeglasses (p-value < 0.0001). Joint pain was reported by 64 (34.6%) of the individuals who fulfilled the selected criteria of Brighton (p-value < 0.0001), 186 (10.6%) wore eyeglasses (p-value < 0.0001) and 226 (12.9%) individuals had marfanoid habitus (p-value < 0.0001). No significant association was found between JH and wearing eyeglasses (p-value = 0.7934) or marfanoid habitus (p-value = 0.6683). Of the affected individuals, 110 (59.5%) were female, 97 (52.5%) were Caucasians, 56 (30.3%) mulattos, 21 (11.5%) Blacks, 4 (2%) Asian descendants and 2 (1%) were Amerindians; the ethnicity of 5 (2.7%) cases was unknown.

There was a significant association between apposition of the thumb and hyperextension of the fifth finger (p-value < 0.0001), and between the selected variables used in the Brighton score with apposition of the thumb, and hyperextension of the fifth finger (p-value < 0.0001). Also there was a tendency of a correlation involving apposition of the thumb with hyperextension the fifth finger, hyperextension of the elbow and the selected variables of the criterion of the Brighton score (p-value < 0.0001).

There was an association for children who played musical instruments (364 - 20.8% cases), with apposition of the thumb, hyperextension of the fifth finger and hyperextension of the elbow (p-value < 0.0001). A total of 642 (36.7%) children had regular physical activities, giving a positive, albeit non-significant, association with JH (p-value = 0.1929). There was no association between this variable and the selected variables of the criterion of Brighton (p-value = 0.1604).

Of the total sample, 172 (9.8%) complained of joint pain (p-value < 0.0001). There were also significant associations for hyperextension of the knee and apposition of the thumb (p-value = 0.0099), and hyperextension of the knee, apposition of the thumb, hyperextension of the fifth finger and hyperextension of the elbow (p-value = 0.0358) and a correlation between the selected variables of the diagnostic criteria of Brighton (p-value = 0.0017). The correlation was not significant for hyperextension of the knee, apposition of the thumb and hyperextension of the fifth finger (p-value = 0.3927).

Discussion

The results show that there is a high incidence of children (41%) with JH. Bravo [7] found a similar result (40%) in a Chilean population however the results of a study of preschool-age children in Brazil gave an even higher frequency of 64.6% [8]. Of the children with JH, 426 (59%) were female there by corroborating the findings reported in the literature regarding the predominance of females [8-10].

Among the variables used for the Beighton score [9] the highest prevalence was hyperextension of the fifth finger in 986 (56.4%) cases, followed by apposition of the thumb in 834 (47.7%) cases, hyperextension of the elbow in 612 (35%), hyperextension of the knee in 426 (24.4%) and anterior trunk flexion in 121 (6.9%) cases.

The prevalence of JH was associated with hyperextension of the fifth finger (p-value < 0.0001) and hyperextension of the elbow (p-value < 0.0001), but not with hyperextension of the knee (p-value = 0.6683). Of the affected individuals, 110 (59.5%) were female, 97 (52.5%) were Caucasians, 56 (30.3%) mulattos, 21 (11.5%) Blacks, 4 (2%) Asian descendants and 2 (1%) were Amerindians; the ethnicity of 5 (2.7%) cases was unknown.

About one third (36.7%) of the children practiced regular physical activity. Briggs, et al. [21] observed that hyper extensibility of the skin and dislocation are the main clinical features reported in dancers with EDS-HT and hypermobility is commonly seen in ballet dancers. This suggests that EDS-HT may be associated with an increased risk of injury and longer post-injury recovery, which can have an adverse effect on the development of their career. Thus, the practice of physical activity among hypermobile individuals should be monitored, as both musculoskeletal complaints and physical activity are related to the quality of life of individuals with EDS-HT [22]. It is expected that a holistic approach to treat this condition will improve the long-term quality of life in respect to the various aspects of the disease [23].

At eight years of age, the motor and physical activity performances are not reduced in elementary school children with JH or EDS-HT. Hence, longitudinal studies of the negative influence of generalized JH on the musculoskeletal system over time is suggested [21]. There are studies that report that myopia occurs more frequently in first-
degree relatives of individuals with myopia indicating that genetic factors have a significant effect on the incidence of myopia. In this study, 10.6% of the children used eyeglasses and so an investigation of myopia in children with JH, their parents and their siblings is also suggested [15].

Pain is generally considered to be of musculoskeletal origin and mostly associated with recurrent joint damage however recent evidence indicates a broader spectrum of painful characteristics in EDS-HT [17]. Pain was reported in one or up to three joints by 34.6% of the children in this study. Musculoskeletal complaints, physical activity and health are factors linked to the quality of life of patients with EDS-HT [22].

For Lamari, et al. [24,25], the precocity of physical and functional diagnoses enables an investigation and guidance on risk situations with the possibility of preventive measures and physical therapy for postural reeducation. Early diagnosis also makes the monitoring of children and the investigation of complications possible. The true diagnosis is still neglected and without indication for treatment. The differentiation between hypermobile and non-hypermobile individuals is not yet well defined, nor is the optimal level of mobility that promotes physical health.

Population studies in schools are suggested to identify children who may have EDS-TH. Furthermore, strategies for inclusion and respect for current public policies that guarantee the right of and for diversity should be sought as well as a multidisciplinary approach to clinically assess and provide appropriate assistance to school age children with this syndrome [26]. It is important to study the peculiarities in Brazil with its geographical magnitude, different climates, different socio-cultural characteristics, and diverse colonization.

Conclusion

The percentage of Brazilian children in this study with characteristics of generalized JH is similar to a study of a Chilean population; most are female and Caucasians. There are higher rates of JH of the upper limbs in particular the finger, wrist and elbow joints, which is important for children who play musical instruments. There is an association between hyperextension of the knee and apposition of the thumb and also between hyperextension of the knee, apposition of the thumb, hyperextension of the 5th finger hyperextension and hyperextension of the elbow. Further population studies are needed in children from Latin America.

References