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Evaluation of Balance and Improvement of Proprioception by Repetitive Muscle Vibration in a 15-Year-Old Girl with Joint Hypermobility Syndrome

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Joint hypermobility syndrome (JHS), alternatively termed hypermobility syndrome or benign joint hypermobility syndrome, is a relatively common rheumatologic condition most frequently diagnosed in women showing chronic pain and other musculoskeletal complaints. There is a significant clinical overlap with Ehlers-Danlos syndrome (EDS), which comprises a clinically variable and genetically heterogeneous group of inherited connective tissue disorders, mainly featuring joint hypermobility, skin hyperextensibility and tissue fragility (1). The revised classification identifies 6 major and other rarer forms (2). Among them, the most common is the hypermobility type (EDS-HT), which is dominated by joint laxity and lacks most cutaneous features observed in other variants (3). Recently, an international group of experts proposed that JHS and EDS-HT are the same condition (4), consisting in a wide clinical spectrum ranging from severe phenotypes with early manifestations to milder forms which are diagnosed in adulthood or, at worst, remain undiagnosed.

Proprioception refers to the sense of static position and movement of the limbs and body without using vision. It usually comprises the sense of stationary position of the limbs (limb-position sense) and the sense of limb movement (kinesthesia). These sensations are important for controlling limb movements, manipulating objects that differ in shape and mass, and maintaining an upright posture (5). Lack of proprioception seems a prominent feature in JHS (6,7). Although its relationship with the underlying joint hypermobility is still largely obscure, a handful of studies suggest that improvement of proprioception may be effective for ameliorating both the functional status, including balance, and chronic pain (8). However, as JHS is a systemic, progressive and chronic condition, physical therapy very probably suffers of major limitations, such as time consumption and short-lasting effects. Recently, repetitive muscle vibration (rMV) has been outlined as a non-invasive method that can significantly and persistently improve muscle performance by acting as an important proprioceptive input (9). Although accumulated data are preliminary, this technique is promising for treating postural dysfunction correlated with chronic rheumatologic conditions, such as JHS or EDS-HT.

We studied a 15-year-old girl with JHS, which was inherited from her mother. She presented with severe joint instability and consequent balance impairment. The patient was treated by rMV following a standard protocol and its effects on balance were measured by the evaluation of displacement of the centre of pressure (COP) in time domain, obtained using a force platform.

Case Report

The participant was a 15-year-old Caucasian girl who requested evaluation by our dedicated rehabilitation outpatient service. She was referred after the diagnosis of JHS/EDS-HT by a clinical geneticist. The diagnosis was established combining the Villefranche and Brighton criteria for the EDS-HT and JHS, respectively (Table I) (2,10). Since early infancy, the patient was "clumsy", particularly lax and naturally predisposed to contortionism. She practised classic dance until her teens, when she started to suffer from recurrent poliarticular pain, mainly involving feet, ankles, knees and the temporomandibular joint. Since 13 years of age, she complained of recurrent falls due to ankle sprains (> 1/week) and anterior knee pain with patellar-femoral subluxations. She also referred myalgias and limb pain, recurring joint effusions, fatigue, recurrent headaches, chronic gastritis and easy bruising. Psychomotor development and mental abilities were otherwise normal. Complete screening for inflammatory arthritis was negative. The same condition was present in her mother and maternal grandmother (directly evaluated). During examination, the patient displayed generalized joint hypermobility (Beighton score: 8/9), limitation of spine movements due to back pain, bilateral *genus recurvatum*, lumbar hyperlordosis, velvety, smooth but not hyperextensible skin, multiple piezogenic papules at the right heel and marked kinesiophobia. Complete neurologic examination was unremarkable except for positive Romberg sign (see below). For several months before our evaluation, she underwent physiotherapy (including proprioceptive and strength exercises) by various specialists without a specific training in joint hypermobility twice/week without significant improvement in the mid- and long-term in terms of quality of life based on the parents'report. Patient's parents signed an informed consent for the following experimental treatment program.

Treatment description

Vibratory stimulation was applied simultaneously to both quadriceps as previously described (8). During vibratory stimulation, the participant was supine and she contracted the quadriceps. The transducer, applied perpendicularly to on the distal end of the *vastus medialis* and the common tendon of rectus and *intermedius femoris* at about 2 cm from the medial edge of the patella, generated a 0.2-mm to 0.5-mm peak to-peak sinusoidal displacement. Vibration frequency was set at 100Hz. Mechanical stimulation was applied over 3 consecutive days. Each application lasted for 30 minutes. For every 10 minutes of vibration, there was a 1-minute interval during which the mechanical application was interrupted and the subject relaxed the quadriceps. This investigation was performed in conformity with the ethical and humane principles of research. Researchers explained purpose, procedures, risks, and benefits of the study to parents who gave their informed consent.

Outcome evaluation

The equipment utilized for data acquisition consisted of a force platform (Kistler Instruments, Winterthur, Switzerland), used to obtain the COP displacement values, and two TV cameras (Video Controller, BTS, Milan, Italy), which was synchronised with the force platform for video recording of the subject and used to ascertain if any undesired behaviour occurred. The COP is the position of the location of the ground reaction force during the standing position. The subject was instructed to maintain an upright standing position for 30 s with arms along their sides and feet positioned over sketches representing the foot with an angle of 30° respect to the anterior-posterior (AP) direction. For each evaluation, data were collected in two consecutive trials. In the first trial, the participant was asked to maintain an upright standing position with eyes open (OE), looking at a black target 1.5 m far away (a circle with a diameter of 6 cm) which was positioned vertically to be in the patient's direct line of sight. In the second trial, the participant was requested to keep her eyes closed (CE). The subject was requested to sit for a period of about 120 s after the completion of each trial in order to give her time to rest.

Quantitative evaluation of posture was performed in 3 different phases: a pre-treatment session (pre) and two post-treatment sessions, after 10 days (post-10) and 40 days (post-40), respectively. The output data provided from the force platform is a series of COP values in time for both the anterior-posterior (AP) direction (Py) and the medio-lateral (ML) direction (Px) (Figure 1). A 2D-representation of body balance was obtained using the sway, plotting the Py as a function of Px COP displacements. The first 10 s interval was discarded in order to exclude the transition phase needed to reach a postural steady state (11). From each 2D components of COP displacements, the ML-COP (ROM Px) and AP-COP excursions (ROM Py) were computed as the difference between absolute maximum and absolute minimum value of Px and Py. The trajectory length (TL) of the COP was computed as previously described (11):

$$TL = \sum_{n=1,N-1} \left[\left(Py[n+1] - Py[n]\right)^2 + \left(Px[n+1] - Px[n]\right)^2 \right]^{\frac{1}{2}}$$

Results of ROM Px, ROM Py and TL were compared with data from a control group (CG) of 15 healthy teenagers (mean age:18.03 years; range: 13-20 years; body weight: 66.7±10.3 kg) evaluated at the same laboratory. Clinically, balance of the patient was evaluated using the standard and sensitized Romberg test as well as the Berg Balance Scale (12) applied to all phases of the study.

Results

In the pre session, we noted marked postural instability in our patient by evaluating the displacement of COP. In order to test the effectiveness of rMV application in our patient, we calculated ROM Px, ROM Py and TL in the pre session, as well as in the post-10 and post-40 session sessions, by comparing our results with the CG (Figure 1). Table II shows variations of the computer parameters (in percentage) between the OE and CE conditions. In the pre session, a significant difference emerged between the patient and CG for all parameters, which was confirmatory for the results obtained by the computing of Px and COP displacement. After treatment, values of ROM Px, ROM Py and TL reduced significantly thus indicating a persistent amelioration of the postural control in our patient (Figure 1). Effects lasted also after 40 days although with a slight decrement of postural control.

When comparing the results at the OE and CE phases, in the pre session there is a significant detrimental effect of the eye closing to the postural control in our patient. This difference is not yet visible after treatment especially for the TL, probably due to the increased ability of the proprioceptive system after muscle vibration (Figure 1; Table II).

During clinical evaluation, Romberg tests showed marked instability with an evident bent to fall in the pre session. After treatment, there was an increased stability with persistent normalization of the test (data not shown). The Berg Balance Scale demonstrated marked improvement of proprioception with a pre-treatment value of 37 and post-10 and post-40 values of 52 (improvement = 26.7%) and 50 (improvement = 26%), respectively. This amelioration is clinically significant, as in the original study by Berg et al. (12) a cut-off of 45 was used to allow calculation for fall relative risk, a value less than 40 was considered useful in predicting risk, and the risk of falling was inversely related to the score value (13). On a holistic approach, after the 40 day treatment period the general sense of good health and kinesiophobia of the patient improved significantly. Therapy was well tolerated and the patient manifested her intention to persevere with it.

Discussion

In the present work, we evaluated with a quantitative method the lack of postural stability in a 15-year-old girl with JHS and demonstrated improvement of proprioception by the evaluation of COP displacement after a standard treatment protocol of rMV.

Joint instability and chronic pain are major complaints of JHS/EDS-HT patients and they may manifest with a wide variety of presentations (14). Additional features, such as fatigue, anxiety and depression, are equally common in JHS/EDS-HT and, very probably, represent late consequences of an irreversible chronic pain syndrome. These complications fuels a detrimental downward spiral, which, in turn, causes severe and progressive deterioration of physical activity and quality of life (15). In JHS, lack of proprioception is a well-known feature, being demonstrated as affecting specific joints, including knee and proximal interphalangeal joint (6,7,16). Taken together, these data indicate a consistent perturbation of the regulation of the segmental posture (i.e., joint stability) in JHS. However, as JHS is a systemic disorder virtually affecting all joints, it seems logical that JHS patients also suffer from poor postural control and balance (i.e., postural equilibrium), as recently demonstrated by a qualitative scoring system in 35 patients with JHS (17). In line with this, a preliminary although consisting result has been obtained by Ferrell et al.(8), who showed improvement of balance after home-based physiotherapy regimen by using a computer-connected board measuring its oscillations.

For the first time, our study demonstrated lack of proprioception with a strictly quantitative approach and also pointed out that, before treatment, postural instability is strongly influenced by visual input (Figure 1). Roughly speaking, this may imply that, in our patient, exteroception (vision) made up for, in part, the lack of proprioception in maintaining an acceptable posture. The differences between the open eye and closed eye phases were no more perceived after treatment (Figure 1; Table II). And this could mean that rMV may improve proprioception, thus minimizing the consequences of an interruption of visual input on posture control.

Focal muscle vibration was demonstrated as a highly selective stimulus for Ia spindle afferents . In fact, vibratory stimulation with specific parameters (i.e., frequency 100Hz, peak-to-peak amplitude of 0.20 to 0.50 mm) may activate different mechanoceptors, in particular spindle afferents and Golgi tendon organs. Activation of peripheral contractile elements strongly influence the activity of the gamma-motoneuron system and therefore the muscle spindle in providing afferent information. The tonic spindle activation is able to induce a long-term primary motor cortex reorganization, characterized by enduring increase of intracortical and cortical reciprocal inhibition (18).

The ameliorative effects of physiotherapy on proprioception and quality of life in JHS is a relatively consolidated assumption in the clinical practice. However, besides anecdotal reports, at present, only two studies demonstrated amelioration of segmental posture by a program of specific exercises in JHS (6,8). The rationale of these studies was to increase joint stability by improving muscle strength in order to facilitate muscle cocontraction. In particular, it was hypothesized that the dynamic stabilization process of cocontraction may be inhibited by abnormal firing of afferent mechanoreceptors (6,8). This treatment strategy can be considered in patients with a significant residual stamina. However, many JHS patients suffer of kinesiophobia, that implies movement avoidance as a strategy to limit bodily pain. Kinesiophobia, in turn, aggravates muscle deconditioning thus determining a vicious downward spiral of declining function and loss of independence. In these subjects, physiotherapy is often not applicable and/or ineffective. rMV may be a good alternative for patients in an advanced stage of the disease in order to improve proprioception and, consequently, increase stamina. In fact, in our patient, the application of rMV significantly improved kinesiophobia and the sense of good health which permitted to progressively return to her daily activities, except for dance. In this patient, amelioration was observed without subsequent application of physical therapy. However, in more severely affected subjects, it could be speculated that rMV may be considered a pre-treatment which permits the following application of physical therapy in subjects who do not normally well tolerate it. The singular observation reported here and the age difference between our patient (15 year) and the young-elderly subjects studied by Farrell et al. (8) may represent a major bias in the attempt to generalize this assumption. Only further studies may shed more light on this point.

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Tables

Table I. Applied diagnostic criteria in our patient.

Brighton criteria (JHS)[15]	Villefranche criteria (EDS-HT)[1]
Major criteria	**Major criteria**
Beighton score ≥4/9	Beighton score ≥5/9
Arthralgia for >3 months in >4 joints	Skin involvement (hyperextensibility and/or smooth, velvety skin)
Minor criteria	**Minor criteria**
Beighton score of 1-3	Recurring joint dislocations
Arthralgia in 1-3 joints	Chronic joint/limb pain
History of joint dislocations	Positive family history
Soft tissue lesions >3	
Marfan-like habitus	
Skin striae, hyperextensibility, or scarring	
Eye signs, lid laxity	
History of varicose veins, hernia, visceral prolapse	
For the diagnosis: both major, or 1 major and 2 minor, or 4 minor criteria ***and*** *the exclusion of other connective tissue disorders*	

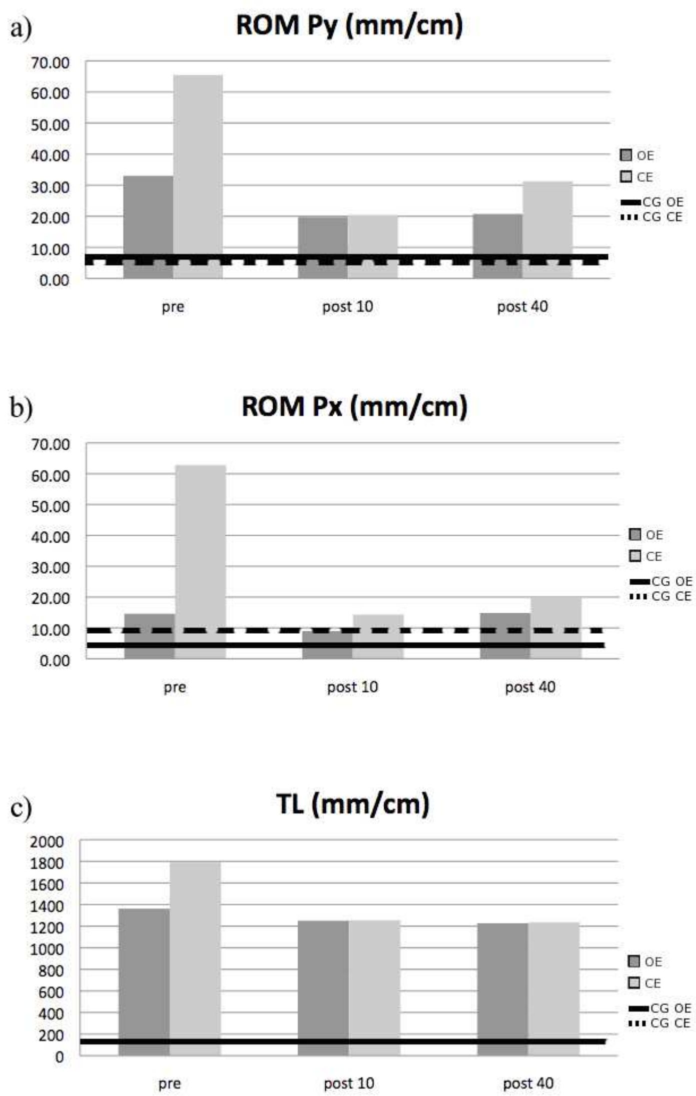
Table II. Variations of the computer parameters (in percentage = %) between the open eye (OE) and closed eye (CE) conditions.

Sessions	(Px) AP OE vs CE	(Py) ML OE vs CE	TL OE vs CE
Pre session	98,1	329,94	31,5
Post-10 session	3,5	59,36	0,28
Post-40 session	50,71	34,89	0,66

Legends to Figures

Figure 1

COP values for both the anterior-posterior direction (a) , the medio-lateral direction (b) and the trajectory length (c) at the open (OE) and close eyes (CE) conditions. Dark (OE) and light (CE) grey vertical bars indicate values for the proband. Dotted and continuous lines indicate values for the control group (CG).



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