Editorial

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Joint hypermobility syndrome/Ehlers–Danlos syndrome hypermobility type: constructing a rehabilitative approach

"...the rehabilitative approach may consider the single subject with the unique and complex involvement presented and a particular attention may be due to the proprioceptive system always involved."

Keywords: fatigue • joint hypermobility syndrome • pain • proprioception • rehabilitation

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Ehlers–Danlos syndrome hypermobility type (EDS-HT) is a heritable connective tissue disorder mainly characterized by joint hypermobility, chronic/recurrent pain, joint instability complications and minor skin changes [1]. Recently, a group of international experts considered EDS-HT one and the same with joint hypermobility syndrome (JHS), thanks to the impossibility to distinguish them on clinical grounds only in most cases [2].

The epidemiology of JHS/EDS-HT has not been properly established. Nevertheless, clinical practice suggests a very high prevalence of JHS/EDS-HT with a proposed frequency of 0.75–2% in the general population [3].

Recognizing JHS/EDS-HT is, at the moment, an exclusion diagnosis based on adherence to either the Brighton [4] and Villefranche criteria [5], which comprise sets of major and minor criteria. Musculoskeletal pain and orthopedic complications, such as myalgia, tendonitis, recurrent back pain and polyarticular arthralgias, have a major role in diagnosing, assessing and managing JHS/EDS-HT patients. In addition, a wide spectrum of chronic complaints and complications, not yet included in the Brighton and Villefranche criteria include: instability; proprioceptive impairment; fatigue; kinesiophobia; motor deconditioning; cephalalgia; postural orthostatic tachycardia syndrome; abdominal pain; and gastroesophageal reflux, among others. A rehabilitative approach may consider all these issues that may contribute to a failure treatment [6].

Rehabilitative key point

Patients with hypermobility are characterized by a poor posture: false flat foot; recurvatum knee; anterior rotation of the pelvis; lumbar hyperlordosis; dorsal cifosis; and shoulder intrarotation [6]. This posture seems related to congenital widespread hypotonia and ligamentous laxity that determine reduced joint stiffness. It is also possible to observe, during walking, a reduced plantar flexion at initial contact and reduced dorsiflexion ability; this is probably related to tibialis anterior weakness and reduced calf muscle strength of the triceps surae. A gait analysis study showed that the ground reaction force has been correlated with fatigue and showed correlation between vertical force and fatigue. This suggests that muscle fatigue may be associated with a loss of proprioceptive acuity in lower limb muscles. Fatigue is considered one of the major determinants for such a severe deterioration of quality of life in these individuals [7].

Furthermore, JHS/EDS-HT patients present postural instability with a significant increase of center of pressure excursion during standing with closed eyes [8]. It may be due to an impairment of the

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proprioceptive system. Proprioception is a term that means the perception of one's self as a result of our own actions; this is a sensory modality based on receptors densely packed in the muscles, in tendons and the skin. JHS/EDS-HT may be described as the 'proprioceptive illness'; in fact, in this condition, it is possible that proprioceptors are unable to give the right feedback caused by the tissue laxity of the soft tissue. In other words, there is a continuous mismatch between the expected signal and those actually generated. The term proprioception includes the sense of limb position and movement, the sense of heaviness or force, the sense of effort and the sense of balance; proprioception has been restricted to receptors concerned with conscious sensations, but muscle spindles and tendon organs also play important roles in the unconscious, spinal reflex action [9]. During movement, new evidence has shown that an additional cue is provided by centrally generated motor control signals; this involvement may have implications for clinical conditions.

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Moreover, most patients report chronic generalized pain. Quite often the pharmacologic approach is not useful and a not clearly identification of the origin of the pain is frequently observed. Some authors have indicated a nociceptive origin, others have observed neuropathic features in the description of patients' pain. Pain is not likely attributable to one of these two types, but is probably a widespread pain, similar to one observed in patients with fibromyalgia and chronic pain syndrome [10]. It has been recently supposed in different studies that repeated noxious stimulation may lead to habituation (decreased response) or sensitization (increased response) as a form of plasticity in the nervous systems. Prolonged or strong activity of dorsal horn neurons caused by repeated or sustained noxious stimulation may subsequently lead to increased neuronal responsiveness or central sensitization. Chronic widespread pain can be the consequence of central sensitization. Central sensitization is known as an increased central neuronal responsiveness and causes hyperalgesia, allodynia, and referred pain and hyperalgesia across multiple spinal segments, leading to chronic widespread pain.

Pervasive pain and chronic fatigue are probably the major factors influencing the onset or amplification of kinesiophobia [11]. If not properly and promptly approached, these patients may develop sever, long-lasting motor deconditioning. This is one of the major problems that a rehabilitator may face in managing JHS/EDS-HT patients.

Role of the conventional rehabilitative approach

JHS/EDS-HT requires a holistic approach in terms of assessment and management, which may involve a multidisciplinary team [1,12]. Management of JHS frequently include the variable and nonstandardized application of: education and lifestyle advice; behavior modification; manual therapy; taping and bracing; electrotherapy; exercise prescription; functional rehabilitation; and collaborative working with a range of medical, health and fitness professionals [13].

> "Proprioceptive information in motor control may be achieved with motor programs adjusted to accommodate unexpected perturbations and with planning and modifications of internally generated motor commands."

Recently, a systematic review of a clinical trial that showed an insufficient research exploring the clinical outcomes of a number of interventions, including sensory integration, positioning and posture management, concerning occupational therapy and physiotherapy has been published [14]. The only evidence-based founded support clinician's use of proprioceptive-based exercises. Anyhow, there is insufficient evidence to determine what type, frequency or dosage of exercise or the means of delivering such exercise interventions. Some proposal in the exercise progression has been indicated [6].

The new proposal

Although, at the moment, reported efficacious in a single patient, the use of repetitive muscle vibration may have a role to improve proprioception and may minimize 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. Tonic spindle activation is able to induce long-term primary motor cortex reorganization, characterized by an enduring increase of intracortical and cortical reciprocal inhibition and the application of focal muscle vibration in a JHS/EDS-HT young patient shown to be able to increase joint stability by improving muscle strength [15]. A mechanoceptor activation able to interact with neuromuscular function is possible to obtain with the application of the neuromuscular taping [16].

Exercise has an important role working in the sensory afferent implementation. Enhance muscle stiffness, of which muscle spindles are crucial

elements, is argued to be an important characteristic for dynamic joint stability [17]. Proprioceptive information in motor control may be achieved with motor programs adjusted to accommodate unexpected perturbations and with planning and modifications of internally generated motor commands. The sources of input that can be used making the exercise are tactile, visual and proprioceptive ones in a multisensory comparison. In addition, an incongruence between sensory input and central motor output (i.e., sensory–motor incongruence) reflecting discrepancies between the motor and sensory cortex may lead to generalized and unfocused pain [18]. In JHS/EDS-HT patients, it has been hypothesized that defective sensory input coupled with disinhibition of the motor output may correspond to the well-known impairment of proprioception. Exercise working on the perception of different tactile stimuli in order to activate skin mechanoceptor and subsequently on kinesthetic information in order to activate muscle receptors [19], may be proposed to JHS/EDS-HT patients for various joints and lower spine that are quite often affected. With these, may be associated proprioceptive exercises to enhance core stability.

If we want to consider different time phases in the syndrome, during the subacute episodes of musculoskeletal pain, dislocation and subluxation the rehabilitative approach may consider a manual mobilization of the fascia. As a connective tissue disorder, the EDS-HT/JHS may present an increased compliance of fascia; fascia, or body's connective tissue matrix, forms a continuous tensional network throughout the human body, covering and connecting every single organ, muscle, nerve and tiny muscle fibers. In this syndrome, it is possible to observe an increased compliance of the fascia, which means the inverse of stiffness (i.e., actual change in force for a given change in length) as a possible cause of the multisystem disorders [20].

During a steady state, the rehabilitative approach may consider a global postural treatment with the assistance of the breathing; breathing should not be paradoxical, but should be characterized by a prolonged expiration in order to reduce the diaphragm muscle tone and contribute to the restoration of sympathetic/parasympathetic systems. The repetitive postural treatment is able to create long-term memory that may stably influence posture.

In conclusion, the rehabilitative approach may consider the single subject with the unique and complex involvement presented and a particular attention may be due to the proprioceptive system always involved.

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