Camerota F, Celletti C./Senses Sci 2015; 2 (1):8 -10
doi: 10.14616/sands-2015-1-0810

The impaired proprioception in Ehlers-Danlos Syndrome-Hypermobility Type/Joint hypermobility Syndrome: the rehabilitation role

Filippo Camerota, Claudia Celletti

Physical Medicine and Rehabilitation Division, Umberto I Hospital, Sapienza University of Rome, P.le Aldo Moro, 00185 Rome, Italy

***Corresponding author** Dr. Filippo Camerota, Physical Medicine and Rehabilitation Division, Umberto I Hospital, Sapienza University of Rome, P.le Aldo Moro, 00185 Rome, Italy; Tel. +39 06 49975947; e-mail filippo.camerota@uniroma1.it

Article history
Received: February 28, 2015
Accepted: March 27, 2015
Published: March 31, 2015

Editorial

Ehlers-Danlos syndrome (EDS) is an umbrella term for a growing group of hereditary disorders of the connective tissue mainly manifesting with generalized joint hypermobility, skin hyperextensibility, and vascular and internal organ fragility. The heritable disorders of connective tissue (HDCTs) comprise four principal disorders: Marfan syndrome (MFS), Ehlers-Danlos syndrome (EDS), Oesteogenesis imperfecta (OI), Benign joint hypermobility syndrome (BJHS). [1]

The last Ehlers-Danlos syndrome (EDS) classification identifies six major variants, including classic (cEDS), hypermobility (EDS-HT), vascular (vEDS), kyphoscoliotic, arthrochalasis and dermatosparaxis subtypes, which are distinguished on the basis of specific diagnostic criteria, i.e., Villefranche criteria [2]. In the clinical practice, adhesion to such criteria help in selecting patients for confirmatory laboratory tests, which are now available for all major EDS subtypes except EDS-HT [3]. As a whole, EDSs have a presumed cumulative frequency of 1/5,000 [4], with cEDS and EDS-HT being the most common variants [5]. Recent observations support with evidence the concept that EDS-HT and JHS may be indeed one and the same condition (i.e., JHS/EDS-HT) also at the genetic level [6].

In the JHS/EDS-HT like in the others connective tissue disorders, there is an involvement of the connective tissue matrix proteins. The proteins concerned include collagens, elastins, fibrillins and almost certainly others that have yet to be discovered. Because of the widespread distribution of these molecules throughout the body, particularly in the joints, skeleton, eyes and vasculature JHS relates to the faulty collagen proteins that affect the body systemically. This implies that symptoms may be varied and may interest not only the musculoskeletal system [7].

Collagen alterations may influence the quality of movement but also movement has a role for the collagen quality: motion has a prevention role in the formation of contractures and adhesions. The immobilization and the reduction of motion (also present in the kinesiophobia) is able to make qualitative change of the tissue. The deficit of movement is able to change the length of the connective tissue which adapt to the shortest distance between the insertion points. This mechanism involves the progressive loss of functioning and the establishment of pain [8].

The collagen alterations involves also the fascial body system, a structural and functional unifying body system. The fascial body seems to be one large networking organ, with many bags and hundreds of rope-like local densifications, and thousands of pockets within pockets, all interconnected by sturdy septa as well as by looser connective tissue layers. Based on this background, a more encompassing definition of the term fascia was proposed as a basis for the first Fascia Research Congress [9] and was further developed [10]for the following two congresses. The term fascia here describes the 'soft tissue component of the connective tissue system that permeates the human body'. One could also describe them as fibrous collagenous tissues that are part of a body wide tensional force transmission system. The complete fascial net then includes not only dense planar tissue sheets (like septa, muscle envelopes, joint capsules, organ capsules and retinacula), which might also be called "proper fascia", but it also encompasses local densifications of this network in the form of ligaments and tendons. Additionally it includes softer collagenous

connective tissues like the superficial fascia or the innermost intramuscular layer of the endomysium.

Commonly neural coordination is seen as the key factor in and the main origin of our motor behavior: the neural system generates impulses to the motor units in the skeletal muscle fibers, and they in turn respond by pulling the origin and insertion of the muscles towards each other thereby exerting forces on the skeleton. Recently some authors have put attention to the role of the fibrous collagenous connective tissues in the body; these are seen as providing a tensional network throughout the whole body, the biomechanical properties of which provide the framework for muscular force transmission as well as for the haptic sensory system [11]. They also suggest that the lack of proprioceptive perception may be more than an adaptation of neural tissues in response to a change in fascial tissue properties. The changes in sensory dynamics maybe themselves driven by the alteration in mechanical connective tissue properties [11].

Proprioception is a term coined by Charles Sherrington in 1906 that means the perception of one's self; this is sensory modality based on receptors densely packed in the muscles and in tendons and called the "secret sixth sense". Proprioceptors precisely measure physical proprieties, such as muscle length, tendon tension, joint angle and deep pressure. Signals from this sensory orchestra are sent by afferent nerves through the spinal cord to the somatosensory, motor and parietal cortices of the brain, where the continuously feed and update the sensory-motor maps of the body [12]. A poor sense of proprioception could explain why people with JHS/EDS-HT become injured, having a lack of sensation of the joint at the end of the range. In hypermobile people the proprioceptors are basically not given the right feedback. The reason underlying why JHS/EDS-HT patients have poor proprioception may be the tissue laxity of the soft tissue; proprioceptors are unable to give the right feedback [13] and the body is continuously getting the "wrong" motor information about where it should be. Impaired proprioception has been described in various joints of JHS/EDS-HT patients [14,15,16].

Moreover in JHS/EDS-HT patients kinesiophobia manifestations has been observed, associated to chronic widespread pain and fatigue [17]. Kinesiophobia has been described as an excessive, irrational and debilitating fear of physical movement and activity resulting from a feeling of vulnerability to painful injury or reinjury [18]. Kinesiophobia, in turn, aggravates muscle deconditioning, thus determining a vicious downward spiral of declining function and loss of independence.

All these features seems to confirm the ameliorative effects due to physiotherapy treatment on proprioception in particular and for quality of life secondarily [19]

In addition to a manual work specific for the fascia [8] an approach to improve proprioception may be the use of repetitive muscle vibration [20] that 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. In fact, vibratory stimulation with specific parameters (i.e., frequency of 100 Hz, peak-to-peak amplitude of 0.20–0.50 mm) may activate different mechanoceptors, in particular spindle afferents and Golgi tendon organs. Activation of peripheral contractile elements strongly influences the activity of the γ-motoneuron system, and therefore the muscle spindle in providing afferent information. The tonic spindle activation is able to induce long-term primary motor cortex reorganization, characterized by an enduring increase of intracortical and cortical reciprocal inhibition [7,19]. Also if evaluated in a single case, the application of focal muscle vibration shows to be able 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 co-contraction may be inhibited by abnormal firing of afferent mechanoreceptors. Repetitive muscle vibration may be a good alternative also for patients in an advanced stage of the disease in order to improve proprioception and, consequently, increase stamina[7,19].

Another possible approach may be the use of Neuromuscular Taping (NMT) that has been proved to be effective in various musculoskeletal conditions [21,22,23]. NMT may play a role as a sensitive input [24] that is integrated by the central nervous system and used for assisting motor program execution process known as sensorimotor integration. Mechanoreceptor stimulation induce by NMT may improve proprioceptive inputs for muscles to perform task-specific functions [25].

References

1. Grahame R. Heritable disorders of connective tissue. Baillieres Best Pract Res Clin Rheumatol. 2000;14:345-61.
2. Beighton P1, De Paepe A, Steinmann B, Tsipouras P, Wenstrup RJ. Ehlers-Danlos syndromes: revised nosology, Villefranche, 1997. Ehlers-Danlos National Foundation (USA) and Ehlers-Danlos Support Group (UK). Am J Med Genet. 1998 ;77:31-7.
3. Mayer K, Kennerknecht I, Steinmann B. 2013. Clinical utility gene card for: Ehlers-Danlos syndrome types I-VII and variants – update. Eur J Hum 21:doi:10.1038/ejhg.2012.162
4. Steinmann B, Royce PM, Superti-Furga A. 2002. The Ehlers-Danlos syndrome. In: Royce PM, Steinmann B, editors. Connective tissue and its heritable disorders, 2nd edition. New York: Wiley-Liss pp. 431–524.
5. De Paepe A, Malfait F. 2012. The Ehlers-Danlos syndrome, a disorder with many faces. Clin Genet 82:1–11.
6. Hermanns-Lê T, Reginster MA, Piérard-Franchimont C, Delvenne P, Piérard GE, Manicourt D. 2012. Dermal ultrastructure in low Beighton score members of 17 families with hypermobile-type Ehlers-

Danlos syndrome. J Biomed Biotechnol2012:878107.
7. Celletti C, Camerota F. The multifaceted and complex hypermobility syndrome (a.k.a. Ehlers-Danlos Syndrome Hypermobility Type): evaluation and management through a rehabilitative approach. Clin Ter. 2013;164:e325-35.
8. Pilat A. Terapia miofasciale: induzione miofasciale. Aspetti teorici e applicazioni cliniche. Marrapese Editore Roma 2006.
9. Findley, T.W., 2012. Fascia science and clinical applications: a clinician/researcher's perspectives. Journal of Bodywork and Movement Therapies 16 (1), 64e66.
10. Huijing, P.O., Langevin, H.M., 2009. Communicating about fascia: history, pitfalls and recommendations. International Journal of Therapeutic Massage and Bodywork 2 (4), 3e8.
11. Turvey, M. T., & Fonesca, S. T. (2014). The medium of haptic perception: A tensegrity hypothesis. Journal of Motor Behavior, 46, 143–187.
12. Rigoldi C, Cimolin V, Camerota F, et al. Measuring regularity of human postural sway using approximate entropy and sample entropy in patients with Ehlers-Danlos syndrome hypermobility type. Res Dev Disabil 2013; 34:840-6.
13. Ferrell WR, Tennant N, Sturrock RD, et al. Amelioration of symptoms by enhancement of proprioception in patients with joint hypermobility syndrome. Arthritis Rheum 2004; 50:3323-8
14. Hall MG, Ferrell WR, Sturrock RD, et al. The effect of the hypermobility syndrome on knee joint proprioception. Br J Rheumatol 1995; 34:121-5
15. Mallik AK, Ferrell WR, McDonald AG, et al. Impaired proprioceptive acuity at the proximal interphalangeal joint in patients with the hypermobility syndrome. Br J Rheumatol 1994; 33:631-750.
16. Rombaut L, De Paepe A, Malfait F, et al. Joint position sense and vibratory perception sense in patients with Ehlers-Danlos syndrome type III (hypermobility type). Clin Rheumatol 2010; 29:289-95
17. Celletti C, Castori M, La Torre G, Camerota F. Evaluation of kinesiophobia and its correlations with pain and fatigue in joint hypermobility syndrome/Ehlers-Danlos syndrome hypermobility type. Biomed Res Int. 2013;2013:580460
18. M. Leeuw, M. E. J. B. Goossens, S. J. Linton, G. Crombez, K. Boersma,. Vlaeyen J. W. S. The fear-avoidance model of musculoskeletal pain: current state of scientific evidence. Journal of Behavioral Medicine 2007; 30: 77–94.
19. Celletti C, Castori M, Galli M, Rigoldi C, Grammatico P, Albertini G, Camerota F. Evaluation of balance and improvement of proprioception by repetitive muscle vibration in a 15-year-old girl with joint hypermobility syndrome. Arthritis Care Res (Hoboken) 2011;63:775-779.
20. Camerota F, Celletti C, Bini F, Marinozzi F. Focal muscle vibration: evaluation of physical properties and his applications. Senses Sci 2014; 1:23-28
21. Camerota F, Celletti C . The Touch, the sense of body and the sense of action. Senses Sci 2014; 1:3-4.
22. Camerota F, Galli M, Cimolin V, Celletti C, Ancillao A, Blow D, Albertini G. The effects of neuromuscular taping on gait walking strategy in a patient with joint hypermobility syndrome/Ehlers-Danlos syndrome hypermobility type. Ther Adv Musculoskelet Dis. 2015;7(1):3-10.
23. Camerota F, Galli M, Cimolin V, Celletti C, Ancillao A, Blow D, Albertini G. Neuromuscular taping for the upper limb in Cerebral Palsy: A case study in a patient with hemiplegia. Dev Neurorehabil. 2014;17:384-387.
24. Celletti C, Blow D, Masala D, Camerota F. The hand as an instrument of cerebral plasticity. Senses Sci 2014; 1:84-86.
25. Kaya Kara, O. Atasavun Uysal, S. Turker D. Karayazgan S. Gunel, M. and Baltaci, G. The effects of kinesio taping on body functions and activity in unilateral spastic cerebral palsy: a single-blind randomized controlled trial. Dev Med Child Neurol. 2014.