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Gait strategy in patients with Ehlers-Danlos syndrome hypermobility type and Down syndrome

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ABSTRACT

People suffering from Ehlers-Danlos syndrome (EDS) hypermobility type present a severe ligament laxity that results in difficulties in muscle force transmission. The same condition is present in people suffering from Down syndrome (DS) even if their clumsy movements are due to cerebral and cognitive impairments. The aim of this study was to quantify the gait patterns of subjects with EDS and with DS using Gait Analysis (GA). We quantified the gait strategy in 12 EDS individuals and in 16 participants with DS. Both pathological groups were compared to 20 age-matched healthy controls in terms of kinematics and kinetics. Results showed that DS individuals are characterized by a more compromised gait pattern than EDS participants, even if both groups are characterized by joint hypermobility. All the patients showed significant decreased of ankle stiffness probably due to congenital hypotonia and ligament laxity, while different values of hip stiffness. These findings help to elucidate the complex biomechanical changes due to joint hypermobility and may have a major role in the multidimensional evaluation and tailored management of these patients.

1. Introduction

Ehlers-Danlos syndrome (EDS) is a relatively common rheumatologic condition which comprises a clinically variable and genetically heterogeneous group of inherited connective tissue disorders, mainly featuring joint hypermobility, skin hyperextensibility and tissue fragility (Callewaert, Malfait, Loeys, & De Paepe 2008). The various forms of EDS are characterized by abnormalities in the chemical structure of the body's connective tissues (for example, skin, muscles, tendons and ligaments) (Voermans et al., 2009). EDS results in weakness and/or excessive flexibility of the connective tissues of the body: as a result, skin may become fragile and joints unstable. People suffering from EDS type III present a severe ligament laxity that results in difficulties in muscle force transmission, showing muscle hypotonia, and in movement instability.

Recently, Galli et al. (2011) pointed out the typical features of gait pattern in a population with EDS: a non-physiological gait was observed by the authors. In particular, moreover the spatio-temporal parameters, all the differences between physiological and non-physiological gait pattern could be summarized in pathological kinematic and kinetic of the ankle joint, in terms of sustained plantarflexion and lower value of absorbed and generated work. They also investigated ankle and

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hip stiffness: the results evidenced a stiffness reduction in both joints compared to Control Group. As reported in the literature (Carr, 1970; Ferrell et al., 2004), the key feature of EDS gait is characterized by hypermobility that results in a decrease of joint stiffness and in the lack of a correct force transmission, and consequently in muscle hypotonia: these features, as well documented in literature (Dowdy-Sanders, & Wenger, 2006; Dyer, Gunn, Rauh, & Bery, 1990; Galli, Rigoldi, Brunner, Virji-Babul, & Albertini, 2008; Morris, Vaughan, & Vaccaro, 1990; Vieregge, Schulze-Rava, & Wessel, 1996; Weeks, Chua, & Elliott, 2000), are similar to the ones that afflict the movements of Down syndrome (DS) people.

Motor disability is widespread among individuals with DS. It includes longer motion and reaction times, balance and postural deficits, and cocontraction of agonist and antagonist muscles (Aruin, Almeida, & Latash, 1996; Shumway-Cook & Woollacott, 1985). The motor dysfunction in individuals with DS involves impaired muscle control, which is frequently referred to as "clumsiness" by parents and health professionals (Latash & Corcos, 1991) and Carr (1970) referred that the delay in motor development in DS is linked to the generalized muscle hypotonia and ligament laxity that is characteristic of the condition. Galli et al. (2008) documented the gait characteristics of children with DS and quantified the hip and ankle joint stiffness that characterize the gait pattern in individuals with DS: in their work pathological subjects pointed out higher values of hip joint stiffness as compensatory strategies in order to lower the numbers of degrees of freedom, and lower ankle joint stiffness as consequence of joint laxity and muscle hypotonia, which cause functional weakness.

As the common feature that characterized EDS and DS syndrome, ligament laxity and muscle hypotonia, aim of this work is to study the relationship between these and the gait pattern alterations in EDS and DS adult patients in order to characterize from a motor point of view the two syndrome.

2. Materials and methods

2.1. Participants

The gait pattern was investigated in a group of 12 participants (EDSG: Ehlers-Danlos Syndrome Group) with a diagnosis of EDS type III (mean age: 43.08 years, sd 6.78; range: 36–59 years) and in a group of 16 participants (DSG: Down Syndrome Group) with Down syndrome (mean age: 35.60 years, sd 4.43; range: 31–45 years): data were collected in the Posture and Motion Analysis Lab of IRCCS "San Raffaele-Pisana", TOSINVEST Sanità, Rome, Italy. All participants gave their informed consent to participate in the study and all investigations were performed in conformity with the ethical and humane principles of research. The researchers explained the purpose, procedures, risks, and benefits of the study to parents who gave their informed consent.

A group of 20 healthy adults was included as controls (CG: Control Group) (ten male and ten female; mean age: 37.23 years; sd 8.91; range 30–50 years): exclusion criteria for the CG included prior history of cardiovascular, neurological or musculoskeletal disorders. They showed normal flexibility and muscle strength and no obvious gait abnormalities.

2.2. Instrumentation and data acquisition

The equipment utilized for data acquisition during the gait trials consisted of a 12-camera optoelectronic system (ELITE2002, BTS, Milan, Italy) with a sampling rate of 100 Hz, two force platforms (Kistler, CH) with a sampling rate of 500 Hz and 2 TV camera Video system (BTS, Italy) synchronized with the system and the platforms for videorecording.

To evaluate the kinematics of each body segment, passive markers were positioned on the participants' body, as described by Davis, Ounpuu, Tyburski, and Gage (1991).

After placement of the markers, subjects were asked to walk barefoot at their own natural pace (self-selected speed) along a walkway (6 m) containing the force platforms at the mid-point. Three acquisitions comprehensive of kinematic and kinetic data were collected for each participant in order to guarantee data reproducibility: once the consistency was verified, the kinematic and kinetic data of one trial for each patient was considered for the analysis.

2.3. Data analysis

Starting from the markers coordinates, using a specific software, the kinematics of the lower limb joints during gait (i.e. pelvis, hip, knee and ankle 3D movements) were computed and represented as % of gait cycle. From the data recorded by force platforms, also the kinetic of hip, knee and ankle was analysed. From the graph representing these variables, some punctual indexes were computed.

In particular, concerning the spatio-temporal parameters, we computed the % stance (%ST), that is the percentage of stance phase of gait cycle; the mean velocity (MV), that is the mean velocity during the gait cycle, normalised to the individual's height (1/s); the anterior step length (ASL), normalised to individual's height.

Considering the kinematics, we calculated the mean value (MPT index) of pelvis tilt on sagittal plane and the mean value of foot progression (MFP) during the gait cycle; the values of the angle of ankle (AIC index), knee (KIC index) and hip joint (HIC index) at the Initial Contact (IC); the values of maximum of the hip flexion (HM index) during the gait cycle, the value of the maximum of the ankle dorsiflexion during stance phase (AMSt index) and the maximum of the flexion of the knee (KMSw index) during swing phase; the value of the minimum of ankle dorsiflexion in stance phase (AmSt index), the value of the minimum of the knee flexion in stance phase (KmSt index), and the value of the minimum of the hip flexion (HmSt index) in