



Postural strategies in Prader–Willi and Down syndrome patients

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ABSTRACT

Patients affected by Down (DS) and Prader–Willi syndrome (PWS) are characterised by some common clinical and functional features including gait disorders and reduced postural control. The aim of our study was to quantitatively compare postural control in adult PWS and DS.

We studied 12 PWS and 19 DS adult patients matched for age, height, weight and body mass index.

They were instructed to maintain an upright standing position on a force platform for 30 s with open eyes (OE) and we calculated the range of center of pressure (CoP) displacement in the A/P direction ($RANGE_{AP}$) and in the M/L direction ($RANGE_{ML}$) and the total CoP trajectory length during quiet stance (Sway Path, SP).

The range of oscillations in PWS and DS in both AP and ML direction were higher than in controls. PWS and DS were statistically different for $RANGE_{AP}$, with PWS showing higher mean values.

Our results confirm a reduced capacity of both PWS and DS in maintaining postural stability. This appears to be in some respect different in PWS and DS, with PWS showing poorer control in AP. DS and, particularly, PWS should be encouraged to undergo specific balance training and strengthening of the ankle muscles as part of a comprehensive rehabilitation program to enhance daily functioning and quality of life.

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1. Introduction

Prader–Willi (PWS) and Down syndrome (DS) are two different chromosomal disorders characterised by some common clinical features, such as obesity, muscular hypotonia, ligament laxity and mental retardation. They also share some functional features: gait and postural disorders are common in both syndromes and tend to progressively worsen as the clinical picture advances, severely limiting the patients' quality of life. In particular, muscular hypotonia seems to be directly related to the impaired postural control and gait alterations that characterise these two pathological states (Cowie, 1970; Davis & Kelso, 1982). Whether the biomechanical determinants of such motor limitations are the same is still unknown and needs further investigations at various levels.

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Gait analysis has focused mainly on DS, with special reference to the specific orthopaedic conditions and biomechanical limitations associated with the syndrome (Caselli, Cohen-Sobel, Thompson, Adler, & Gonzalez, 1991; Galli, Rigoldi, Brunner, Virji-Babul, & Albertini, 2008; Parker & Bronks, 1980; Roizen & Patterson, 2003; Vismara et al., 2007). One study compared the spatiotemporal, kinematic and kinetic parameters of gait in PWS and DS outlining significant differences in terms of cadence, anterior step length and progression velocity. PWS were in fact characterised by values closer to normal than DS and were able to walk with a more “stable” strategy (Cimolin et al., 2010).

Postural analysis has also been investigated mainly in DS (Carvalho & Almeida, 2009; Galli, Rigoldi, Mainardi et al., 2008; Shumway-Cook & Woollacott, 1985; Webber, Virji-Babul, Edwards, & Lesperance, 2004) and, to our knowledge, only one study was conducted on PWS patients (Capodaglio et al., 2010). According to this literature, both DS and PWS are characterised by a reduced postural control, as measured by center of pressure displacement (CoP) and postural sway velocity. Despite scanty scientific evidence, postural stability in PWS is a clinically relevant issue: fracture risk is approximately 50% in children (Kroonen, Herman, Pizzutillo, & MacEwen, 2006) and more than 30% in adults (Butler et al., 2002). The high rate of fractures in these patients may be related to acromicria, reduced bone mineral density, early obesity and dysfunctional behaviors, sarcopenia and muscular hypotonia. Reduced strength of knee flexors and extensors may account for reduced balance in adult PWS (Capodaglio et al., 2009). In general, postural control appears a relevant issue in all individuals with excessive body weight (Menegoni et al., 2009). Obese persons appear to be at greater risk of fall than normal-weight subjects under daily postural stresses and perturbations (Teasdale et al., 2007).

Rehabilitation specialists are increasingly aware of motor disability in PWS and DS patients, but they fail to provide evidence-based specific treatment modalities. Due to their common clinical and functional features, rehabilitation strategies aimed at reducing postural disability in these two genetic conditions may possibly share some common bases. A deeper understanding of the causes of their postural abnormalities may improve the definition of rehabilitation planning and treatment. No studies have so far investigated possible quantitative differences in postural strategy between PWS and DS. The aim of the present study was therefore to quantitatively compare postural control in adult PWS and DS.

2. Methods

2.1. Participants

In this study we considered 12 PWS and 19 DS adult patients matched for age, height, weight and body mass index (kg/m²: BMI) (Table 1). PWS patients had been periodically hospitalised at the San Giuseppe Hospital, Istituto Auxologico Italiano, Piancavallo (VB), Italy. All patients showed the typical PWS clinical phenotype (Holm et al., 1993). Cytogenetic analysis was performed in all participants; 10 had interstitial deletion of the proximal long arm of chromosome 15 (del15q11–q13). Moreover, uniparental maternal disomy for chromosome 15 (UPD15) was found in 1 female. All PWS participants showed mild mental retardation. In this respect, one of the requirements for participating in the study was a score over the cut-off value of 24 in the Mini Mental State Examination (MMSE) Italian version (Neri, Andermacher, Spanó, Salvio, & Cipolli, 1992). Scores over the MMSE cut-off are recognized as suggesting the absence of widespread acquired cognitive disorders in adult people. DS patients were all referred to the IRCCS “San Raffaele Pisana”, Tosinvest Sanità, Roma, Italy. The distribution of chromosomal anomalies is pure trisomy 21 in all of the DS patients. All PWS and DS patients were able to understand and complete the test.

Ten age-matched individuals were included as controls (control group: CG). Exclusion criteria for the control group included prior history of cardiovascular, neurological or musculoskeletal disorders.

All participants were free from conditions associated with impaired balance and individuals with vision loss/alteration, vestibular impairments, neuropathy, as detected by the clinical examination and those who reported symptoms related to intracranial hypertension, were excluded. The study was approved by the Ethics Committees of the two Institutes for PWS and DS patients. Written informed consent was obtained by the parents or, when applicable, by the patients.

2.2. Experimental set-up

The PWS patients were evaluated at the Movement Analysis Lab of the San Giuseppe Hospital, Istituto Auxologico Italiano, Piancavallo (VB) and DS patients were assessed at the Movement Analysis Lab of the IRCCS “San Raffaele Pisana”,

Table 1

Clinical characteristics of the study groups (PWS: Prader–Willi syndrome; DS: Down syndrome; CG: control group). Data are expressed as mean (standard deviation).

	PWS patients	DS patients	Control group
Participants (M/F)	12 (5/7)	19 (10/9)	20 (10/10)
Age (years)	25.7 ± 6.1	29.1 ± 8.5	31.4 ± 9.6
Height (cm)	152.1 ± 9.9 [*]	150.6 ± 10.2 [*]	173.3 ± 5.01
Weight (kg)	93.9 ± 17.1 [*]	82.5 ± 10.9 [*]	626.9 ± 8.5
BMI (kg/m ²)	41.6 ± 6.4 [*]	36.7 ± 5.9 [*]	22.8 ± 3.2
Foot length (mm)	208.6 ± 12.9 [*]	219.8 ± 14.7 [*]	239.9 ± 11.4

^{*} $p < 0.05$ compared with control group.

Table 2

Postural parameters of the study groups (PWS: Prader–Willi syndrome; DS: Down syndrome; CG: control group). Data are expressed as mean values (standard deviation). The values are normalised for the subject's height and foot length.

	PWS	DS	CG
RANGE _{AP} (1/m)	0.07 (0.02) ^{*,*}	0.05 (0.02) [*]	0.02 (0.01)
RANGE _{ML} (1/m)	0.07 (0.03) [*]	0.05 (0.02) [*]	0.03 (0.02)
SP (mm/m)	2.56 (0.63) ^{*,*}	4.32 (0.74) [*]	0.51 (0.98)

^{*} $p < 0.05$, PWS versus DS group.

^{*} $p < 0.05$ compared with control group.

Tosinvest Sanità, Roma, Italy, equipped of the same instrument for postural assessment. In particular, the postural evaluation was conducted in both Labs with a force platform (Kistler, CH; acquisition frequency: 500 Hz) integrated with a video system.

The individuals were instructed to maintain an upright standing position for 30 s with open eyes (OE) focusing on a 6 cm black circle positioned at the individual line of vision at a distance of 1.5 m. Arms were at their sides and feet were positioned at an angle of 30° with respect to the A/P direction. To avoid any kind of learning or fatigue effect only one trial was acquired in this study (Tarantola, Nardone, Tacchini, & Schieppati, 1997).

2.3. Data analysis

The outputs of the force platform allowed us to compute the CoP time series in the A/P direction (CoP^{AP}) and the M/L direction (CoP^{ML}). The first 10-s interval was discarded in order to avoid the transition phase in reaching the postural steady state (Galli, Rigoldi, Mainardi et al., 2008).

Using the specific software (SMARTAnalyser, BTS, Italy) from platform data we identified and calculated the RANGE_{AP} and RANGE_{ML} indices, that are the range of CoP displacement respectively in the A/P direction and in the M/L direction (expressed in mm) and the Sway Path (SP), that is the total CoP trajectory length (expressed in mm) during quiet stance.

All parameters were normalised to the participant's height (expressed in meters) and to their foot length (expressed in millimeters). In fact, short feet are one of the typical features of PWS (Hudgins, McKillop, & Cassidy, 1991) and DS patients (Diamond, Lynne, & Sigman, 1981). It has been shown that the base of support, as calculated from the foot length, is one of the most relevant biomechanical variables in postural analysis (Chiari, Rocchi, & Cappello, 2002).

2.4. Statistical analysis

All the previously defined parameters were computed for each participant and then the mean values and standard deviations of all indexes were calculated for each group. PWS and DS data were compared using Mann–Whitney *U* tests, in order to detect significant differences. The patients' and the controls' data were compared with Mann–Whitney *U* tests. Null hypotheses were rejected when probabilities were below 0.05.

3. Results

In Table 1 the clinical characteristics of PWS, DS and CG are reported. Age was not significantly different among groups. BMI, weight and height were similar in PWS and DS but significantly different from CG. In Table 2 the mean values and standard deviations of the postural parameters considered in this study for PWS, DS and CG are shown. The reported values were normalised for individual height (expressed in meters) and foot length (expressed in millimeters).

From our data it is evident that both PWS and DS individuals were characterised by higher RANGE_{ML}, RANGE_{AP} and SP as compared to CG. No differences were present between PWS and DS in terms of RANGE_{ML}, but PWS and DS were statistically different for RANGE_{AP}, with PWS showing higher mean values.

4. Discussion

In this study we aimed to quantify postural control in PWS and DS and compare their strategies with the evaluation of the 2D CoP trajectories and of some parameters based on the analysis in the time domain, such as CoP excursion in ML and AP directions and CoP trajectory length. The range of oscillations in PWS and DS in both AP and ML direction were higher than CG. PWS and DS differed only in AP displacement, leading to significant disparities in the length of CoP displacement. Our results confirm previous reports on reduced capacity of both PWS and DS in maintaining CoP inside their base of support. In particular, this non-physiological strategy was more evident in PWS in the AP direction. Postural stability is maintained by a combination of ankle and hip strategies, which act synergically. These strategies are used in anticipation of a voluntary movement that is potentially destabilizing.

Since both PWS and DS are characterised by muscle weakness and hypotonia, their joint stability decrease by increasing range of motion. In these patients, joint stiffness during gait varies according to the level: at hip level, PWS show values close to normal and DS increased stiffness values; at ankle level, a significant reduction is evident in both groups as compared to

controls (Cimolin et al., 2010). This different strategy may account for our results: the lower value of CoP AP oscillations in DS may be related to their higher hip stiffness as compared to PWS.

Interestingly, even if DS showed CoP oscillations close to PWS in ML direction and reduced CoP oscillations in AP, the CoP trajectory length in DS was statistically higher than PWS. We can therefore hypothesise that the two syndromes are characterised by different CoP velocities, faster in DS than PWS, but further studies are needed to back this hypothesis. Galli, Rigoldi, Mainardi et al. (2008) using a frequency-domain approach to postural analysis demonstrated that DS were characterised by instability also in the AP direction, which was not evident from the analysis in the time domain. A similar approach could be used possibly in PWS.

Postural strategy appears to be in some respect different in PWS and DS, with PWS showing poorer control in AP. PWS show a reduced muscle tonus already during the first year of life and develop obesity at 2–3 years of age. It is well known that these two factors affect the development of motor and functional skills that usually children learn at that age. On the other hand, DS show an increased stiffness at hip level. This may act as a limiting factor on gait (Cimolin et al., 2010) but at the same time as a stabilizing factor on posture. Higher CoP displacement in AP are in line with the low muscle strength values observed in PWS (Capodaglio et al., 2009) and are also supported by the improvement in gait after training of the key muscles for the ankle strategy (Vismara et al., 2007). While in a previous study we demonstrated that DS patients walked with a less physiological strategy (Cimolin et al., 2010), as concerns postural strategy DS individuals seemed to be characterised by a better postural scheme. For this reason improving balance and decreasing risk of fall is critical to independence in daily life activities and appears particularly relevant in PWS.

From a clinical point of view, quantitative characterisation of postural strategy in PWS and DS is important to develop, differentiate and enhance the rehabilitative options. Common goals in the two populations are to improve hypotonia, muscle strength, motor control and reduce body weight. In addition, both PWS and DS patients need specific balance exercise training as part of a comprehensive rehabilitation program and particularly PWS should be encouraged to undergo strengthening of the ankle muscles to counteract AP instability. Evidence-based rehabilitation programs aimed at improving postural control would contribute to enhance daily functioning and quality of life in those patients.

Competing interest

All authors have no conflicts of interest or any financial interest. All authors attest and affirm that the material within has not been and will not be submitted for publication elsewhere.

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