Case report
Clin Ter 2020; 171 (6):e471-475. doi: 10.7417/CT.2020.2259

A novel microgravity postural rehabilitation protocol in Beckwith Wiedemann syndrome: a case report

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Abstract

Background. Beckwith-Wiedemann syndrome (BWS) is a genetic overgrowth syndrome. The excess growth can manifest as whole-body or regional overgrowth in approximately 25% of patients.

Purpose. The aim of this case report is to show an original rehabilitation protocol in BWS for improving the recovery of postural control in asymmetric body growth of the body.

Case Description. An 11-year-old male with BWS performed a rehabilitation program for 12 months, once a week (every session lasted 45 minutes), composed of postural exercises under microgravity conditions using the Dynamic-Antigravity-Postural-System (SPAD) and proprioceptive insoles.

Conclusions. The results showed that: SPAD improved the asymmetry of the morphology and body dynamics and could prevent further postural alterations; the proprioceptive insoles increased the postural stability of the patient with improved walking. Future research will be needed to expand these results. ***Clin Ter 2020; 171 (6):e471-475. doi: 10.7417/CT.2020.2259***

Key words: *exercises, posture, body, stabilometry*

Introduction

Beckwith-Wiedemann syndrome (BWS) is a genetic overgrowth syndrome, defined classically by omphalocele, macroglossia, and macrosomia. Other common features include hemihyperplasia, ear lobe creases, posterior helical pits, umbilical hernia, visceromegaly, adrenocortical cytomegaly, renal abnormalities, and neonatal hypoglycemia. The diagnosis of the disease is clinical and is based on criteria that have been refined (1); none of the components of the classical triad (*omphalocele, macroglossia, and macrosomia*) is a diagnostic requisite, because a genetic test is necessary. The phenotype of BWS can vary significantly, and a high index of suspicion must be maintained for children who present with fewer findings to identify those who are at risk for tumor development (2). Macroglossia and macrosomia are generally present at birth but might have postnatal onset.

Beckwith-Wiedemann syndrome has an approximate incidence of 1 in 13,700 births (3) and affects both sexes equally, except in monozygotic twins (significant female preponderance). The estimated mean prevalence of classical Beckwith-Wiedemann syndrome in Europe is 3.8 per 100,000 births or 1 in every 26,000 births (4).

The molecular basis of the syndrome is complex and heterogeneous, being a pathological model due to a lack of genomic imprinting, with several molecular defects in the 2 zones that are subjected to imprinting of the 11p15.5 chromosomal region. Abnormalities in this region are found in approximately 85% of subjects who are affected and include genomic, genetic, and epigenetic changes, which result in altered physiological allelic contribution of parental chromosomes (5,6).

Medical treatments, surgery, and rehabilitation have been proposed for certain signs and symptoms that are related to BWS, such as oromyofunctional dysfunction (macroglossia) (7), but for hemihyperplasia, there is no scientific evidence of rehabilitation. Hemihyperplasia can affect segmental regions of the body or select organs and tissues (8) and can cause serious postural instability under static and dynamic conditions during growth.

Moreover, the excess growth in BWS can manifest as whole-body or regional overgrowth, due to cellular hyperplasia, which occurs in approximately 25% of patients but might not be apparent at birth (2). This kind of hypergrowth can involve half body side or be localized (frequently to a limb but also the tongue). It can be accompanied by hyperplasia of the organs (visceromegaly), possibly with a component of asymmetry, characterized by dissimilar maximum renal diameters between the 2 organs, corresponding to the ipsilateral hyperplasia of the body or lower limb (1,9). In addition to possible aesthetic problems, this hyperplasia can result in complications, depending on the site of onset. When arising in the lower limbs, it can cause varying degrees of asymmetry, which can negatively affect walking and the

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ISSN 1972-6007

development of scoliosis and incorrect postural attitudes (10). There are no studies on physiotherapy and rehabilitation programs that support motor development with regard to postural asymmetry of children with BWS. To this end, our retrospective observational case report aimed to examine the efficacy of an early rehabilitation approach to improve postural alignment and postural balance.

Description of the Case

This study presents the results of a 12-month physiotherapy and rehabilitation program in a case with BWS. We obtained informed consent from the parent of the minor for participation in the publication of this case report but not for publication of photographic material. The therapeutic protocol complied in accordance with the Declaration of Helsinki ethical standards.

We examined an 11-year-old male with BWS that was diagnosed at birth who had never undergone rehabilitation and who was treated with motor postural exercises in rehabilitation unit at G. D'Annunzio University of Chieti (Italy). His clinical history was unremarkable except for partial glossectomy (surgical technique not known). On admission to the outpatient rehabilitation unit, the anthropometric values of weight and height were measured at baseline (T0) and after 12 months of rehabilitative therapy (T1a and T1b), and BMI was calculated by dividing body weight by the square of the height (kg/m^2); no pharmacological treatment has been reported. A spinal x-ray revealed reduced physiological sagittal alignment, mild convex right lumbar scoliotic misalignment (Cobb= 15°) (11), and lower limb asymmetry with left lower limb shorter than right one of 3 centimeters; abdominal ultrasonography in the past year detected hepatomegaly with moderate steatosis and splenomegaly (12). A neurological examination showed no evidence of cognitive deficits.

Before starting the rehabilitative treatment, the patient did not report any accidental falls and had a sedentary lifestyle, occasionally (once per week) playing football.

Description of Rehabilitative Intervention

The aim of the rehabilitation treatment was to improve the symmetry of the body and the alignment of the spine with respect to the frontal and sagittal planes and increase postural stability. Our patient underwent the initial rehabilitation for 12 months, once per week, comprising postural exercises under microgravity conditions using the Dynamic Antigravity Postural System (SPAD): the SPAD is a device for bodyweight relief, consisting of a machine that is designed to reduce, modify, and condition the force of gravity that acts on the body structures of movement during rectilinear motion. This system is based on the rationale that gait training can combine a motor task with sensory feedback, consistent with the multisensory approach to postural control.

The system is composed of a treadmill on which the patient trains, supporting his body weight, and a structure to which he is harnessed with a pneumatic belt that us placed between the iliac crests and costal arches, connected to a lifting system with 4 tie-rods that attached to the body and pelvic girdle this equipment is completed by 4 front pads (2 on the humeral heads for the shoulder girdle and 2 on the anterior superior iliac spine for the pelvis) that act as stabilizers, preventing twisting of the pelvis or shoulder during movement on the treadmill, and as proprioceptive informants and 2 rear pads, one placed on between scapulae region and one on the sacral apex (13). Proprioceptive insoles were prescribed to correct the lower limb asymmetry, with 2.5 cm left heel elevation, because the correct distribution of loads and pressures on the surface of the foot is fundamental in ensuring correct walking and maintenance of balance under static and dynamic conditions (14).

Description of Outcomes

At baseline, no pathological values emerged from the assessment using the Pediatric Balance Scale (PBS)(15) (Tot= 52); also, on the timed up and go test (TUG)(16), the values were at the lower end of the normal range (Tot= 14 seconds). The patient underwent an appropriate postural evaluation before rehabilitative treatment (T0) and after 12 months of follow-up (T1 particularly T1A with insoles, T1B without insoles) using digital biometric measurements to allow us to observe various structural characteristics of his body by stabilometry under open eyes and closed eyes conditions (OE and CE) and by static and dynamic baropodometry (BioMetricSoftware® - Diasu - Italy). These body characteristics were analyzed using frontal (anterior and posterior) and sagittal (right and left side) projections using the 3D Body Analysis Kaptur (BAK3d) system (17). We noted a good response to SPAD treatment for static and dynamic posture, associated with greater postural balance. In the static baropodometric test, the symmetrical distribution of foot load between the left and right feet improved significantly compared with the pretreatment condition (Table 1). Also, in the dynamic baropodometric test, walking speed increased, the time of contact between the foot and ground declined, and the length of the step between the right and left feet became symmetrical, indicating symmetry of the body after rehabilitative treatment (Table 1).

In the stabilometry test under open eyes (OE) and closed eyes (CE) conditions, the ellipse surface (ES) and path length (PL) decreased at T1B, demonstrating a reduction in energy expenditure for posture balance control (Table 2), improved further by wearing proprioceptive insoles at T1A.

The postural analysis by 3D Body Analysis Kaptur, with regard to the anterior and posterior frontal lines and the right and left sagittal lines, allowed us to evaluate and quantify possible asymmetries in our patient with respect to the reference ideal position (18,19). The position of the head and spine curvature with respect to the plumb line was recorded considering the distance from the plumb-line at the spinous processes of the C7, T12, and L3 (according to Stagnara, the values at the level of C7 and L3 must be between 25 and 40 mm). Pelvic tilt was calculated as the angle of inclination of the straight line that passed through the cutaneous reference in the external anatomical region

Table 1. Baropodometry values.

Evaluation Times			
Parameters	**T0**	**T1 A**	**T1 B**
FSL right (%)	45,51	47,32	46,13
FSL left (%)	54,49	52,68	53,87
CoP X (mm)	15,51	5,6	10,58
CoP Y (mm)	16,95	23,11	20,55
FDL right (%)	53,2	49,2	48,03
FDL left (%)	46,8	50,8	51,97
FPA right (°)	25,56	21,51	20,1
FPA left (°)	17,04	17,08	17,08
SL right (mm)	118,89	135,49	130,59
SL left (mm)	121,98	130,86	126,98
WS right (m/sec)	36,72	42,64	40,55
WS left (m/sec)	46,79	42,37	41,84

Legend: FSL=foot static load; CoP Y= the antero-posterior direction of the body's center of pressure; FDL=foot dynamic load; WS= walking speed, m/s.; SL= the length of the step; FPA= foot progression angle.

that corresponded to the posterior superior iliac spine (PSIS) and the skin reference on the upper front iliac spine (ASIS) according to normal reference values between 12 and 18 degrees. Mandibular angle, normally evaluated as the insertion of the plane of Frankfurt, can be between 15° and 60°, with an average value of approximately 35°. Shoulders symmetry was calculated as the inclination, expressed in degrees, of the line that passes through the 2 cutaneous references on the apex of the right and left acromion, with respect to the absolute horizontal. This indicator quantifies structural misalignments of the humeral shoulder girdle and consequently of the spine's dorsal tract. ASIS symmetry was defined as the inclination, expressed in degrees, of the line that passes through the 2 skin references on the upper right and left iliac spine, compared with the absolute horizontal. This indicator quantifies any misalignments of the pelvis, on the frontal plane. Patellar symmetry was calculated as the inclination, expressed in degrees, of the line that passes through the 2 cutaneous references at the center of the right and left patella, versus the absolute horizontal, and quantifies misalignments of the lower limbs. Malleolar symmetry was considered the inclination, expressed in degrees, of the line that passes through the 2 cutaneous references on the internal malleolus on the right and left compared with the absolute horizontal, quantitating misalignments of the lower limbs and, in particular, the ankle. Scapular symmetry was calculated as the inclination, expressed in degrees, of the line that passes through the 2 skin references on the lower apex of the right and left scapulae, compared with the absolute horizontal, quantifying any paramorphism of the back. PSIS symmetry was defined as the inclination, expressed in degrees, of the line that passes the 2 cutaneous references on the right and left upper posterior iliac spine, with respect to the absolute horizontal. This indicator quantifies any misalignments of the pelvis. Normally, all values for the body symmetries above must not exceed 2/3 degrees.

After rehabilitative treatment, as shown in Table 3, the relationship between the distance from the plumb line in the spinous processes of C7 and L3 improved, with good alignment with respect to the sagittal plane, and the asymmetry of the posterior superior iliac spine (PSIS) decreased, with better harmony of the body.

Table 2. Stabilometry Evaluation

Evaluation Times				
Parameters		T0	T1A	T1B
OA	ES	188.99	49.69	52,26
	PL	833.77	108.09	110,15
	CoP X	0.32	3.28	4,56
	CoP Y	0.08	-20	-20,56
OC	ES	155.63	36.32	6,47
	PL	748.54	109.89	125,1
	CoP X	1.52	4.46	5,01
	CoP Y	-3.05	-19.78	-22,5
Romberg index		82.34	73.09	117,62

Legend: ES= ellipse surface; PL= path length; CoP X= the latero-lateral direction of the body's center of pressure; CoP Y= the antero-posterior direction of the body's center of pressure; Romberg Index= represents the relationship between the surface of the confidence ellipse with closed eyes and the surface of the confidence with the eyes open, multiplied by 100.

Discussion

Although BWS is a well-known congenital overgrowth syndrome (20) with regard to its clinical aspects, the literature on postural rehabilitation is scant. BWS has wide phenotypic variability that correlates with several etiological mechanisms, from genomic, genetic, and epigenetic alterations that cause clinical profiles with overlapping phenotypic characteristics (10).

The aim of our study was to develop an effective rehabilitation program that positively influences the recovery of postural control and establishes a more stable scheme of walking and better postural balance.

Table 3. Postural Evaluation

Evaluation Times			
Parameters	T0	T1A	T1B
Cervical arrow (mm)	93.4	43.3	61,4
Lumbar arrow (mm)	81.9	43.6	57,3
Pelvic tilt (°)	22.9	18.5	20,4
Mandibular angle (°)	59.6	7.05	15,6
Shoulder symmetry (°)	-3.4	-2.9	-3,6
SIAS symmetry (°)	-5.3	-3.7	-4,1
Patellar symmetry (°)	19.2	6.1	15,4
Malleolar symmetry (°)	21.9	-2.3	5,3
Scapular symmetry (°)	3.7	-0.2	1,1
SIPS symmetry (°)	3.3	2.8	3,1

Legend: SIAS= Anterior superior iliac spines; SIPS= Posterior superior iliac spine;
The value "–" is indicative of a symmetry shifted from the left side.

SPAD treatment improves the asymmetry of the morphology and body dynamics of the syndrome and prevents further postural alterations, especially considering the pubertal growth period of our young patients. During puberty, correct postural alignment on the sagittal plane helps prevent future paramorphism or dysmorphism of the spine (21).

The SPAD system has dual functions: mechanical, allowing neuromotor retraining with corticosubcortical forms of learning to acquire new body schema in a balance that minimizes the energy consumption that is needed to maintain posture, and proprioceptive, maintaining the adaptations that are induced over time relative to walking (13,22,23). SPAD improves postural stability in patients with biomechanical diseases of the spine, such as low back pain and postural biomechanical syndrome (24). The rehabilitative treatment was well tolerated, the patient was able to complete the entire exercise program.

Moreover, the use of proprioceptive insoles increased the postural stability of the patient (at T1A in Table 1, 2, 3), with improved walking, implicating a re-learning process and plastic re-adaptation of a walking generator that allows a more physiological and efficient walking cycle. The stabilizing action on the body also has beneficial effects on the control of balance through greater synergy of the antigravity muscles and a faster response of the support apparatus, leading to quicker repositioning of the body's center of gravity (CoP) (25). As in overweight children compared to normal-weight peers, the distance from the plumb line to L3 is significantly correlated with the area of the ellipse in the condition of open eyes: as the hypotonia of the abdominal muscle, which is frequent in the phase puberty, and the slight paramorphism of the spine during growth in the absence of scoliosis can disturb postural control, even in a quiet position (26).

The results of our study show that this novel rehabilitation protocol for BWS, composed of SPAD and proprioceptive insoles, improves posture and walking quality. The patient is currently being treated, and we await for the follow-up.

What This Case Adds to Evidence-Based Practice

Patients with BWS should be followed systematically during the period of growth, even with regard to rehabilitation, concerning the characteristic body hemi-hyperplasia (involving a single body extremity to an entire half body), which, if it is not promptly and adequately treated, can cause serious morphological and dynamic alterations to the whole-body scheme that realize a function reduction.

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