Heliyon 8 (2022) e11702

Contents lists available at ScienceDirect

Heliyon

journal homepage: www.cell.com/heliyon

Research article

Energy analysis of gait in patients with down syndrome

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ARTICLE INFO

Keywords:
Energy analysis
Energy index
Down syndrome
Sport
Gait analysis

ABSTRACT

Background: the primary aim of this study is to analyse the energy parameters of patients with Down syndrome compared to a control group and secondly to verify whether the sport activity leads to differences in energy expenditure.

Methods: 3 groups of subjects were identified: 8 healthy subjects and 147 subjects with Down syndrome, of whom 14 played sports at least once a week. An energy index was calculated, given by the ratio between potential and kinetic energy. Next, kinetic ad potential energy parameters were extrapolated at 60% of the gait cycle (propulsion phase).

Findings: Down syndrome group was compared with the control group and emerged that the energy index was higher in the first one. No changes were found between Down syndrome and Down syndrome Sport groups. The analysis of the energy parameters showed that all parameters, except the medio-lateral kinetic energy, were higher in the control than in the Down syndrome groups. The potential energy, medio-lateral kinetic energy, and vertical were higher in the Down syndrome Sport group than in the Down syndrome group. The kinetic energy and the mean velocity were higher in the control group than in Down syndrome Sport group while the medio-lateral kinetic energy was lower.

Interpretation: sport modified the parameter of potential energy but not that of kinetic energy, which continued to be different compared to the healthy group and increased the oscillations in the medio-lateral plane, which were double compared to Down syndrome group. The increase in potential energy, found to be almost equal to that of control group, indicates an increase in vertical oscillations. This could be because subjects who practise sports have stronger muscles that allow a greater push-off ability, which therefore increases their potential energy.

1. Introduction

Trisomy 21 is a chromosomal malformation that leads to a difference in the size of specific areas of the brain, in the number and morphology of neurons and in the different connectivity that are established (Pinter et al., 2001). By means of imaging techniques and autopsies, reduced brain dimensions were found: a brachycephaly with a greater development of the cranium in width than in length; a smaller volume of the brain and, in particular, of the cerebellum; a hypoplasia of the hippocampus, cerebral cortex and white matter; and a reduction in the number of neuronal cells (Lott and Dierssen, 2010). It also shows how a volumetric reduction of the cerebellum, typical of patients with DS, leads to problems of motor control and proprioceptive as well as a non-motor involvement fronto-cerebellar and cerebellar limbic: emotion, attention, working memory and impaired language learning (Gunbey et al., 2017).

Numerous studies have been conducted to evaluate energy parameters starting from the analysis of the gait of patients with Down syndrome (DS) in order to identify the relationships between the typical motor characteristics of these patients and the impairment of functional performance (Kubo and Ulrich, 2006; Rigoldi et al., 2012; Salami et al., 2014; Wu et al., 2014). The main findings are an altered pattern of kinetic and potential energy in the different planes during walking, also as a result of energy indices calculated specifically for the study (Bennett et al., 2005; Salami et al., 2014).

Regarding movement, and in particular walking, in subjects with DS there is a difficulty in motor coordination, which makes these patients appear "clumsy" (Latash, 2007). The delay in neuro-psychomotor development present since birth and the reduction in the volume of the cerebellum mean that these patients present patterns and compensatory strategies different from healthy subjects: longer movement times and

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https://doi.org/10.1016/j.heliyon.2022.e11702
Received 30 April 2022; Received in revised form 24 June 2022; Accepted 11 November 2022

adaptation to changes, postural and balance deficits, cocontraction of agonist and antagonist muscles, abnormalities of the spine and joint instability in particular of the hip, knee and ankle (Galli et al., 2008; Zago et al., 2019; Cimolin et al., 2010). Typical characteristics of these patients are muscular hypotonia, joint stiffness and ligamentous laxity that lead to an incorrect postural control, a reduction in walking speed and step length, an inadequate static balance with antero-posterior and medio-lateral oscillations and a greater step width (Agiovlasitis et al., 2011; Horvat et al., 2012). All this inevitably leads to a greater energy expenditure due to an altered pattern of kinetic and potential energy of these subjects and the adoption of compensatory strategies, which often result in an abnormal gait (Salami et al., 2014; Webber et al., 2004; Zago et al., 2020). Improvement in gait and postural control after several training sessions, in terms of kinematics and kinetics, has been observed in several studies, demonstrating how physical activity programs can encourage a better lifestyle and slow down the development of age-related and sedentary diseases (Carmeli et al., 2002; Maki et al., 1994; Rigoldi et al., 2011; Winter et al., 2003), particularly if training and rehabilitation are carried out in childhood, before children acquire walking patterns (Zago et al., 2020).

The aim of this study is therefore to define an energy characterization of patients with DS, in terms of kinetic and potential energy in the different directions, compared to the healthy control group, and secondly to evaluate the effect of sports activity.

2. Methods

The locomotion can be summarized as an alternate transfer between gravitational-potential energy and kinetic energy within each step (Figure 1) (Cavagna et al., 1977). This mechanism is the same that happens considering an inverse pendulum. To shape the body as a pendulum it should be considered that the body is supported by the legs and rotated around the ankle joint. The inverted pendulum is a passive system, as opposed to the real walk that depends on the gravitational force and the moments of the body (Omer et al., 2014). These concepts were used for the calculation of the potential and kinetic energy during the walk.

2.1. Energy analysis – experimental protocol

First, the energy expenditure during the gait of patients with DS was evaluated starting from the kinematic quantities acquired using the optoelectronic system and spherical retro-reflective passive markers, placed on the individuals' skin at specific landmarks according to the protocol proposed by Davis et al. (1991). The data have been acquired in the laboratory of motion analysis of the institute IRCCS San Raffaele Pisana of Rome. The digital optoelectronic system consists of twelve cameras

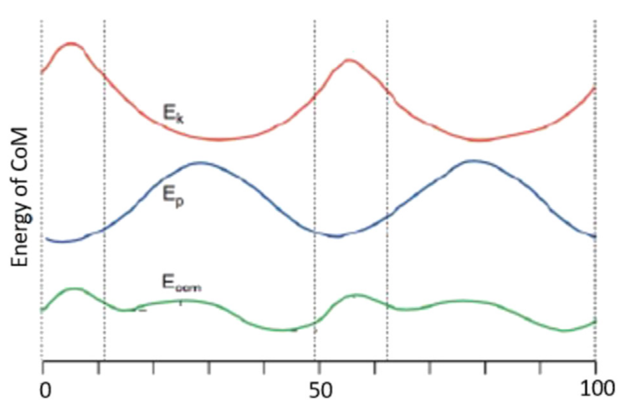

Figure 1. Representation of kinetic, potential and total energy trends of the center of mass (CoM). Top: mean kinetic energy (Ek). Middle: mean potential energy (Ep). Bottom: sum of kinetic and potential energies (Ecom).

Smart DX system (BTS Spa, IT) with a sampling frequency of 100 Hz, appropriately fixed to the laboratory walls. The acquired signal was interpolated and filtered using a low-pass Butterworth filter with a cutting frequency of 10 Hz. The subjects were involved in standardize gait analysis trials consisting of walking barefooted along a 5 m walkway at a self-selected comfortable speed. The data was analysed using BTS SMART Analyzer.

The main parameters analysed were the kinetic energy and potential energy of the CoM of the subjects at 60% of the walk, i.e. during the propulsion phase. In particular, using Smart Analyzer, the following were extracted for this phase of the cycle: maximum and minimum kinetic energy, maximum and minimum kinetic energy in the antero-posterior, vertical and medio-lateral directions, maximum and minimum potential energy and average walking velocity. All of these values were considered for both the right and left lower limb to test whether motor impairment was symmetrical across limbs.

The general formulation of the kinetic (E_k, Eq. (1)) and potential energy (E_p, Eq. (2)) were calculated as follows:

$$E_k = \frac{1}{2}mv^2 \quad [J] \tag{1}$$

$$E_p = mgh \quad [J] \tag{2}$$

where h is the average vertical position of the centre of mass (CoM) - approximated with the marker on the sacrum - during the test, m is the body mass of the subjects, g is the acceleration of gravity and v is the velocity of travel.

2.2. Participants

The analysis was carried out on a total of 147 patients with DS. This sample was further divided into two: patients with DS who play sports (DS Sport) (n = 14) and those who do not (n = 133). The DS Sport group was composed of subjects who perform various disciplines in an amateur way, including swimming, dance, basketball, karate, gymnastics, water polo, etc. The sports activity was practiced at least once a week. Patients with DS were selected among individuals from the IRCSS "San Raffaele Pisana" hospital (Rome, Italy). Exclusion criteria used in this study were: surgery or orthopaedic treatment that compromised the patient's ability to walk independently without an assisting device, presence of congenital cardiac abnormalities, presence of auditory or visual impairments, presence of clinical signs of dementia.

The approval of the study was carried out by two ethics committees of IRCCS San Raffaele Pisana: for patients who play sports, the approval was given by the ethics committee with protocol number SPOL-17/17–6/2017; for patients who do not play sports, the approval was given by the ethics committee with protocol number DOPLAGA-19/35–12/2019.

In this study, a group of non-sporting healthy subjects was also involved as a control group (CG) (n = 8). In this case the experimental procedure was explained in detail to the participants and the study was carried out in accordance with the ethical standards of the institutional and national research committee and with the 1964 Helsinki declaration and its later amendments.

Two gait tests were acquired for all subjects; in some cases, it was possible to keep both valid, in other cases one was discarded because the data acquired were not consistent, e.g., the subject stopped during the test, or the markers were not visible. An average was made for the subjects with two trials, so they affect the final results equally.

The main anthropometric parameters of these three groups were presented in Table 1, whose normality was first checked by applying the Kolmogorov-Smirnov test. If normal, Student's t-test was then used to compare the means; if not, the equivalent nonparametric Mann-Whitney test was used.

No statistically significant difference was found between the ages of the three groups (p values all higher than 0.05). It can be noted that statistically significant differences were found in the BMI of CG and DS

Table 1. Anthropometric data expressed as mean and (standard deviation). M, male; F, female; BMI, body mass index. * Indicates that there is a statistically significant difference between DS or DS Sport with the CG.

	Sex	Age	Height [cm]	Weight [kg]	BMI [kg/m^2]
DS	71M, 62F	21.07 (9.89)	144.00 (13.59)*	53.39 (16.49)	25.10 (5.55)*
DS Sport	8M, 6F	21.57 (6.44)	151.39 (7.57)*	62.01 (11.19)	27.05 (4.46)*
CG	1M, 7F	26.75 (10.24)	168.88 (12.73)	61.75 (9.17)	21.73 (2.98)

group (p = 0.016) and between CG and DS Sport group (p = 0.003), while there was no difference between DS and DS Sport groups (p = 0.147). The same results between the groups were found in height: there was a statistically significant difference between the DS and CG groups (p = 2,0 10^{-4}) and between DS Sport and CG groups (p = 0.002), while no difference was found between DS and DS Sport groups (p = 0.054). In addiction no statistically significant difference was found between the weight of the three groups (p values all higher than 0.05). These differences found between subjects with DS and the CG are precisely due to the pathology; in fact, it is typical for Down syndrome to have a shorter height, and consequently a higher BMI, than healthy subjects (Bertapelli et al., 2016; Rubin et al., 1998).

2.3. Statistical analysis

The Minitab software was used to perform all the statistical analysis. The aim was to verify the presence of any statistically significant differences in energy expenditure during walking between the different populations.

The first step involved the application of the Kolmogorov-Smirnov test to verify the normality of the sample. At this point, a nonparametric statistic was performed using the Mann-Whitney test.

A significance level of 0.05 with a 95% confidence interval was considered.

3. Results

First, the energy expenditure during the gait of patients with DS (n = 133) was evaluated starting from the signal acquired using the optoelectronic system. The energy index (i_E, Eq. (3)), considered was the ratio between potential energy amplitudes (E_p) and kinetic energy (E_k) (Bennett et al., 2005):

$$i_E(t) = \frac{E_p(t)}{E_k(t)} = \frac{h(t)*g}{0.5*v(t)^2} \tag{3}$$

where h is the average vertical position of the centre of mass (CoM) - approximated with the marker on the sacrum - during the test, g is the acceleration of gravity and v is the velocity of travel. This index was evaluated on the DS group, with the aim of verifying if there was a statistically significant difference between the index evaluated on the right and left lower limb. No statistical difference was found between the energy index of the right and left lower limb, with a p-value = 0.720. This result was explained considering that in the case of DS patients the motor impairment is symmetrical on all limbs. Since there was no statistically significant difference between the two classes, it can be considered right and left index as one population.

Next, the indices of the population of subjects with DS (DS Sport group was not considered in this analysis) were compared with the indices of the CG. These subjects consisted of 7 females and 1 male with a total of 27 acquired gait tests (54 i_E indices). The results are summarized in Table 2.

It can be observed that there was a statistically significant difference between CG and DS: DS population had a higher index than the CG, with a p value of 0.35 10^{-14}.

Table 2. Mean, median, and standard deviation of the indices of DS patients and the control group. The presence of the symbol "*" indicates that there is a statistically significant difference.

	N	i_E mean	i_E std	i_E median
DS *	266	0.943	0.243	0.900
CG *	54	0.633	0.195	0.656

A further investigation was performed between the indices of the population presenting DS and the DS Sport group (Table 3). The DS Sport group was composed of 14 patients and a total of 18 tests were acquired with therefore 36 indices (right and left).

In this case there was no statistically significant difference between the DS and DS Sport groups (p value = 0.485).

3.1. Energy analysis of the propulsion phase

After having carried out the analysis on the i_E energy index, the study continued by analysing the kinetic and potential energy of the CoM of the subjects at 60% of the walk, i.e., during the propulsion phase. In particular, using Smart Analyzer, the following were extracted for this phase of the cycle: maximum and minimum total kinetic energy, maximum and minimum kinetic energy in the antero-posterior, vertical and mid-lateral directions, maximum and minimum potential energy and speed. All these values were considered for the right lower limb only. Subsequently, kinetic and potential energy (ΔE) changes were calculated as the difference between the maxima and minima at the propulsion phase. The same statistical analysis was then performed on these parameters for the DS, DS Sport, and CG.

A statistical analysis of the energy parameters was carried out starting from the comparison between DS, DS Sport and CG (Table 4).

For all parameters considered, a statistically significant difference resulted between DS and CG. In particular, it was possible to observe a great difference in the values of ΔE_k (almost 13 J, p-value = 0.94*10^{-11}) and in the maximum and minimum peaks of kinetic energy (11 J and 19 J respectively with p-value = 0.33*10^{-13} and p-value = 0.58*10^{-13}), caused by the differences in speed of the two groups. Typical of Down syndrome is a reduced gait velocity, which in fact shows a statistically significant difference with CG with a p-value = 0.29*10^{-14}. However, the variation of potential energy ΔE_p, was greater in the CG than in the DS group with a p-value = 0.48*10^{-4}. On the other hand, the mid-lateral

Table 3. Mean, median, and standard deviation of indices of DS patients and DS group playing sports.

	N	i_E mean	i_E std	i_E median
DS	266	0.943	0.243	0.900
DS Sport	36	1.011	0.330	1.045

Table 4. Mean and standard deviation of energy parameters of DS, DS Sport and CG. * indicates that there is a statistically significant difference between DS or DS Sport with the CG; ° indicates that there is a statistically significant difference between DS with the DS Sport; ML, medio-lateral; V, vertical.

Variable	DS	DS Sport	CG
ΔE_k [J]	11.93 (5.26) *°	19.35 (9.25)	24.03 (5.85)
ΔE_p [J]	10.27 (5.09) *°	16.48 (8.67)	16.23 (6.22)
$E_{k\ max}$ [J]	24.16 (11.82) *	32.00 (17.34) *	55.85 (16.70)
$E_{k\ min}$ [J]	12.23 (7.24) *	12.65 (9.29) *	31.82 (11.84)
$E_{p\ max}$ [J]	439.2 (160.6) *°	549.7 (96.6)	547.7 (107.3)
$E_{p\ min}$ [J]	428.9 (157.1) *°	533.2 (92.2)	531.5 (104.1)
$E_{k\ ML\ max}$ [J]	1.86 (2.78) *°	3.84 (2.93) *	0.93 (0.64)
$E_{k\ V\ max}$ [J]	0.38 (0.33) *°	0.88 (0.83)	0.88 (0.52)
Mean Velocity [m/s]	0.79 (0.16) *	0.79 (0.26) *	1.18 (0.15)

component of the kinetic energy $\Delta E_{k\ ML\ max}$ (p-value = 0.009) was much larger in the DS than in the CG.

Considering the group DS vs DS Sport, a statistically significant difference was shown in ΔE_k (p-value = 0.004), ΔE_p (p-value = 0.001), $E_{p\ max}$ (p-value = $0.25*10^{-3}$) and $E_{p\ min}$ (p-value = $0.29*10^{-3}$). Velocity and $E_{k\ max}$ did not undergo sport-induced improvements. Moreover, it was possible to observe that in the medio-lateral plane there was an increase of oscillations in fact $E_{k\ ML\ max}$ was greater in the DS Sport group than in the DS with a p-value = $0.42*10^{-3}$. However, the physical activity seems to have improved the values of potential energy, indicating that there were more vertical oscillations. This was confirmed by the value of kinetic energy in the vertical direction ($E_{k\ V\ max}$) which was greater in those who practice sports.

Considering the DS Sport and CG groups there was a statistically significant difference in the $E_{k\ max}$ (p-value = $0.56*10^{-4}$) and $E_{k\ min}$ (p-value = $0.17*10^{-5}$), in the $E_{k\ ML\ max}$ (p-value = $0.11*10^{-4}$), and between the mean velocities (p-value = $0.53*10^{-5}$).

3.2. *Temporal trends of energy parameters*

Temporal energy trends in the step cycle were then evaluated; graphs were made using Smart Analyzer (Figures 2 and 3). For the potential energy, the subtracted curve of the mean value was reported. X represents the antero-posterior direction, Y the vertical direction, and Z the mid-lateral direction.

It can be observed that potential energy and kinetic energy are in phase opposition, as is reported in the literature and explained previously. The graphs represent the results obtained by statistical analysis of the energy parameters at 60% of the step cycle.

Between DS and CG, not only there are differences between the maximum and minimum of kinetic energy, but it is evident, as already reported in the statistical analysis, a great difference in the variation of this: the graph is not only shifted towards lower values but is also flattened.

Finally, it is possible to see a greater medio-lateral component of kinetic energy in the DS as previously described.

4. Discussion

The aim of this work was to analyse the energy expenditure during walking of a population with Down syndrome and compare it with a healthy control group and with a group of DS subjects but practicing amateur sports, in order to verify whether this physical activity improves the strategies adopted during locomotion.

As already widely discussed, this pathology presents typical characteristics, including joint stiffness, muscular hypotonia, ligamentous laxity, poor balance and motor coordination, low push-off ability, greater CoM oscillations in the medio-lateral direction (Rigoldi et al., 2011; Zago et al., 2020).

The starting point was to analyse the i_E index using Smart Analyzer with the aim of quantifying energy expenditure and therefore the degree

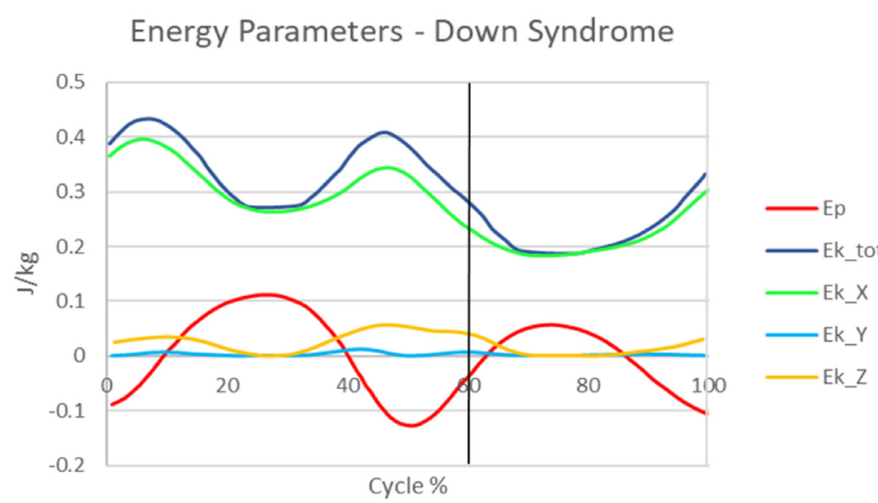

Figure 2. Energy parameters of a patient with Down syndrome.

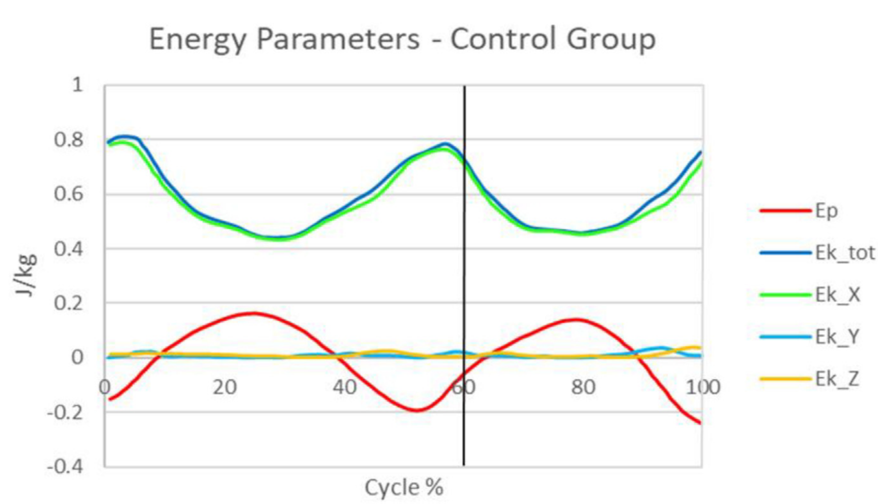

Figure 3. Energy parameters of a healthy subject.

of physical impairment. The results obtained showed a statistically significant difference between DS groups and CG: this was due to the reduced speed that characterizes patients with Down syndrome. In fact, at the denominator of the index there was the kinetic energy that is directly proportional to the velocity, so lower values of velocity determine a higher index. In general, the index was higher as the degree of physical disability increased, as the lower speed of gait at the denominator increases its value. No statistically significant difference between the DS and DS Sport groups were found. This could be due to the fact that sport practiced in an amateur way does not affect patients enough to generate a modification of energy expenditure.

The i_E index was evaluated by considering the maxima and minima of energy at the push-off phase. This term refers to the push-off that the ankle exerts at the end of the stance phase due to the plantar flexor muscles generating positive power in the joint. From an energetic point of view, it represents an important moment for locomotion, as it contributes to the acceleration of the limb that is about to enter the swing phase and to the acceleration of the CoM. Although, in fact, this energy change was localized in a single limb, it is included in the CoM calculations and plays the role of redirecting it during the period between one step and another (Zelik and Adamczyk, 2016). As seen earlier in energy analysis, there are energy losses when the foot impact with the ground and these are restored through muscle work. An adequate push-off reduces these losses and for this reason represents an important moment within the stride cycle, from an energetic point of view. For these reasons, this parameter has been chosen as a time interval in which to study the analysed parameters.

The data of the three populations (DS, DS Sport and CG) were then analysed; the three groups did not present statistically significant differences in the average age and are therefore well comparable. The only differences were those concerning height and BMI between the DS and DS Sport groups with the group of healthy subjects. This is not surprising as Down syndrome presents with a shorter stature than the average. From this derives a lower walking speed that in fact was statistically different between the subjects with DS (sports and non-sporting) and the subjects of the control group.

Summarizing these main results were obtained:

(i) DS vs CG: statistically significant difference for all parameters. In particular, there was a large difference in kinetic energy, caused by the reduced walking speed.
(ii) DS Sport vs CG: also for DS Sport patients, as for DS, the kinetic energy continued to be lower than for those not affected by the syndrome, indicating that there was no improvement determined by sport. The medio-lateral oscillations of DS Sport patients are high (greater than those of DS patients), subjects who practice sport therefore present greater vertical excursions of the CoM. Potential energy did not differ between the two groups, suggesting a sport-induced improvement.
(iii) DS vs DS Sport: there were differences between potential energies, which patients who do motor activity have improved as previously described, and between kinetic energy in the ML direction, which was almost double in DS Sport.

This analysis confirms that sport brought improvements in the potential energy and vertical kinetic energy, while the other parameters remain unchanged.

In this regard it is worth making some considerations. Several studies (Pinter et al., 2001; Rigoldi et al., 2009) have shown, through magnetic resonance imaging (MRI) and autopsies, that individuals with Down syndrome are characterized by a smaller overall brain volume with significant reductions in the cerebellum. The cerebellum, which receives information from the vestibular system and the motor apparatus, plays an important role in the coordination of posture, movement and motor control (Gunbey et al., 2017). When alterations are present it is possible to observe irregularities in balance and hypotonia. The alteration of motor control is at the base of the typical “clumsy” attitude of these subjects and among the consequences of this behaviour there are the major oscillations of the CoM in the medio-lateral direction (Latash, 2007). Thanks to this study, it was possible to observe that patients with DS who practiced sports did not have an improvement but presented this attitude in a more accentuated way: the maximum kinetic energy in this plane was double that of the population that did not practice sports. This result demonstrates that the group practicing sports accentuates movement in the medio-lateral component, highlighting a deficit that is nonetheless persistent in terms of motor control and that is not influenced by motor activity. The increase in this parameter did not depend on the increase in speed, as the two populations did not show a difference in this variable.

Regarding, instead, the increase in E_p and vertical E_k, it is probably indicative of a compensatory strategy (Salami et al., 2014). As previously described, in order to have a good exchange of kinetic and potential energy and therefore decrease the muscular work, a deceleration of the CoM must correspond to an increase of it. This is what seems to happen in DS Sport: a low speed and kinetic energy in the antero-posterior direction was associated with an increase of CoM and therefore of E_p, confirmed by the increase of $E_{k\ V}$. This strategy, aimed at compensating a low E_k during walking, could positively affect energy expenditure.

4.1. Limitations

The main limitation of this study is the low numerosity of the CG, in which only one subject is male; in addition, the DS Sport group is less numerous than the DS group. Considering the physical activity carried out by the DS Sport group, it is non-competitive and patients practice it a few times a week. Further considerations could be made by analysing subjects practicing sports at a competitive level or considering disciplines that require more muscular effort or need more coordination and balance, which are poor in subjects with Down syndrome. Another interesting analysis could be a comparison of different sports to study which has a greater influence on patients' energy expenditure.

5. Conclusion

In conclusion, it is possible to summarize the results of this study as follows: (i) from the energetic point of view, a statistically significant difference emerged between DS and CG groups (i_E higher in DS subjects), while no difference was detected between DS and DS Sport groups; (ii) from the analysis of the energetic parameters listed in Table 4, statistically significant differences emerged of all parameters between CG and DS groups, of potential energy – ML kinetic energy and vertical between DS and DS Sport groups, of kinetic energy and ML kinetic energy between CG and DS Sport groups. It was also found that sport improved potential energy but not kinetics, which continued to be different from the healthy class and increased oscillations in the ML plane, which were twice as high as in the non-sporting population. However, it should be remembered that the population practicing sports is not very numerous, moreover, the physical activity is not competitive, and patients practice it few times a week. Future considerations could be made by analysing subjects who train at a competitive level. Finally, it would be interesting to verify whether sport practiced at a young age, when motor control is not yet mature, could affect it.

Although sport does not seem to be able to modify the motor control of DS subjects, it is still fundamental. In fact, sport can improve the cardio-circulatory system as well as increase the musculature, which is fundamental for these subjects who are characterised by overweight, ligamentous laxity and a high risk of cardio-circulatory diseases.

Declarations

Author contribution statement

CRISTINA FERRARIO: Analyzed and interpreted the data; Contributed reagents, materials, analysis tools or data; Wrote the paper.

Claudia Condoluci: Conceived and designed the experiments; Performed the experiments.

Marco Tarabini: Contributed reagents, materials, analysis tools or data.

Manuela Galli: Conceived and designed the experiments; Contributed reagents, materials, analysis tools or data.

Funding statement

This research did not receive any specific grant from funding agencies in the public, commercial, or not-for-profit sectors.

Data availability statement

Data included in article/supp. material/referenced in article.

Declaration of interest's statement

The authors declare no conflict of interest.

Additional information

No additional information is available for this paper.

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