



## Physical Activity and Indices of Physical Well-Being in Children and Adolescents with Trisomy 21



Prosiniklis Polymeris <sup>a</sup>, Kallistratos Ilias <sup>b</sup>, Xristara Papadopoulou Alexandra <sup>c</sup>, Tsigaras Georgios <sup>d</sup>, Anna Chalkia <sup>e</sup> Efthymia Vargiami <sup>f</sup> Dimitra Katsantoni <sup>g</sup>

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### Corresponding Author <sup>a</sup>



### Keywords

dynamometer;  
Jamar;  
MicroFet 4;  
Pediatric Balance Scale;  
time up and go test;  
Trisomy 21;

### Abstract

**Introduction:** Trisomy 21 (Down syndrome) is the most common genetic disorder, which is caused by a chromosomal abnormality and presents various deviations in areas such as mental and motor levels. The presence of extra information corresponding in whole or in part to chromosome 21 affects the development of children with trisomy 21, and abnormal gene expression leads to changes in brain function. The overall prevalence of the syndrome is one (1) to two (2) per thousand (1,000) live births, although the prevalence may have variability among racial or ethnic groups. The purpose of this research is to highlight the deviations in the balance ability, the strength of the upper limbs and to what extent they affect the quality of life of children and adolescents with trisomy 21. **Methods:** Eighteen (18) children and adolescents with trisomy 21 participated in the research, with three (3) being excluded due to exclusion criteria. Fifteen (15) children and adolescents with trisomy 21 were finally included, of which four (4) were boys and eleven (11) were girls. Measurements were performed through standardized balance tests (Pediatric Balance Scale, Time Up and Go test), with standardized upper limb strength measuring tools [hydraulic handgrip dynamometer (Jamar) and electronic handgrip dynamometer (Micro Fet 4)]. The statistical analysis was performed with the spreadsheet (Excel). A study protocol was drawn up in the pediatric department of the Apostolos Fokas Center for Developmental Pediatrics of G.H.Th. Hippocrates in the pediatric population with trisomy 21. **Results:** The recording of strength results demonstrates the most important deviations in the total of children and adolescents with trisomy 21 compared to the general population, for handgrip strength. The balance ability of all children with

<sup>a</sup> Physiotherapist PT, MSc, Department of Physiotherapy, International Hellenic University, Greece

<sup>b</sup> Professor, Department of Physiotherapy, School of Health Sciences, International Hellenic University, Greece

<sup>c</sup> Former Professor, Department of Physiotherapy, School of Health Sciences, International Hellenic University, Greece

<sup>d</sup> Academic Scholar, Doctor of Medicine School of A.U.Th., Department of Physiotherapy, International Hellenic University, Greece

<sup>e</sup> Professor, Department of Physiotherapy, School of Health Sciences, International Hellenic University, Greece

<sup>f</sup> MD, PhD Professor of Child Neurology Pediatrics 1st Department of Pediatrics, Aristotle University of Thessaloniki Hippokratio General Hospital, Greece

<sup>g</sup> Pt, Graduate, Department of Physiotherapy, International Hellenic University of Thessaloniki, Greece

trisomy 21 who participated in the research, shows deviations related to postural control compared to typically developing children. Discussion and Conclusion: Discrepancies in the scoring of tools such as the Pediatric Balance Scale demonstrate problems in functional balance and movement disorders and more specifically posture and balance as categorized by the ICF model. This research is one of the first in the world literature to examine the correlation of upper limb strength with the balance ability of children and adolescents with trisomy 21.

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## 1 Introduction

### *Trisomy 21*

Trisomy 21 (Down syndrome) is the most common genetic disorder and is caused by a chromosomal abnormality and presents various deviations in areas such as mental and motor (Aly & Abonour, 2016; Eid, 2015; Pinero-Pinto et al. 2020). The presence of extra information corresponding in whole or in part to chromosome 21 affects the development of children with trisomy 21, and abnormal gene expression leads to changes in brain function (Aly & Abonour, 2016; Jung et al., 2017).

The overall prevalence of the syndrome is one (1) to two (2) in one thousand (1,000) live births, although it may have variability among racial or ethnic groups (Gupta et al., 2011; John et al., 2016; Ijezie, 2023). In the USA, an estimated six thousand (6,000) children with trisomy 21 are born annually, with an estimated prevalence of thirteen (13) per ten thousand (10,000) live births (Antonarakis et al. 2020).

Regarding the pathophysiology of the syndrome, the most common cause is the presence of an extra copy of chromosome 21, which is caused by a failure to segregate during the development of the ovum or sperm and ultimately leads to trisomy [Asim, A. et. al. 2015]. About 95% of cases are caused by maternal meiotic nondisjunction, which results in an extra copy of chromosome 21 (Asim et al., 2015; Holz et al., 2019). The other 5% of trisomy 21 causes occur from Robertsonian translocation (2–4%) or mosaicism (1–2%) (Holz et al. 2019; Starbuck et al., 2013). The phenotype includes intellectual disability, short stature, cardiac malformations with specific facial features such as almond-shaped brachycephaly, eyes with epicanthic folds, flat nasal bridge, and prominent tongue (Sherman et al., 2007; Starbuck et al., 2017).

People with trisomy 21 present motor clumsiness, as they are characterized by generalized hypotonia and joint laxity, strongly affecting their fine and gross motor skills (Pinero-Pinto et al., 2020; Foley & Killeen, 2019). Due to ligamentous instability and loose joints, there is a high probability of dislocation of the hip joint, approximately (6%) (Bull, 2011), while the most frequent problem, 10-30% probability is the dislocation of

the atlantoaxial joint, which is caused by hypermobility of the 1st and 2nd vertebra and there is a high risk of damage to the spine. In the first months of life, the motor development of children with trisomy 21 is slow and lags behind compared to typically developing children.

The delay in motor development from four (4) to five (5) months in the first twelve (12) months of their life becomes approximately two (2) years at the age of five (5). As a consequence of their delayed and reduced motor activities, the onset of walking is delayed by an average of twelve (12) months (Dellasudas, 2005).

Children with developmental disorders, such as trisomy 21, show impaired reactive control of posture and stability. This has the result that almost all interventions, to improve the motor functions of people with special needs, place great emphasis on the constant practice of balance (Shumway-Cook & Woollacott, 1985). They also present a reduced level of muscle strength compared to individuals of typical growth (Aly & Abonour, 2016; Eid et al., 2017, Shields et al., 2008). Muscle weakness is associated with reduced cardiovascular fitness and increased incidence of osteoporosis (Shields et al., 2008). For this reason, the evaluation of the function and muscle strength of the upper limbs is essential for planning the therapeutic program for children and adolescents with trisomy 21 (John et al., 2016).

## 2 Materials and Methods

### *Subjects*

A study protocol was drawn up in the pediatric department of the Apostolos Fokas Center for Developmental Pediatrics of G.H.Th. Hippocrates in the pediatric population with trisomy 21. Random sampling was performed with inclusion criteria including: 1) ability to balance and walk independently, 2) normal muscle tone, including mild hypotonia, 3) mild mental retardation, 4) absence of serious vision and hearing problems, 5) sufficient knowledge of the orders given and 6) upper limb grasping ability. Exclusion criteria included: 1) severe intellectual disability, 2) inability to execute orders, 3) inability to grasp upper limbs, 4) inability to balance, 5) existence of a cardiac or musculoskeletal problem and 6) receiving medical treatment that prevented participation in the research. Eighteen (18) children and adolescents with trisomy 21 took part in the research, with three (3) being excluded due to exclusion criteria. Fifteen (15) children and adolescents with trisomy 21 were finally included, 33% boys and 66% girls [five (5) boys, ten (10) girls].

### *Measurements*

Since for the present study it was necessary to choose valid and reliable assessment tools, the **Jamar** hydraulic dynamometer and the **MicroFet 4** electronic dynamometer were chosen to measure hand grip strength, while the **Timed Up and Go Test (TUG)** and the **Pediatric Balance Scale (PBS)** were chosen to assess balance ability. Dynamometer measurement (**Jamar**) was performed using a standard handgrip test protocol (Jamar, Creative Health Products, Plymouth, MI) with the elbow flexed at 90°. The measurement with the **MicroFet 4** electronic dynamometer was performed according to the manufacturer's specifications, with the elbow flexed at 90°. **TUG** is widely used to determine fall risk, to assess balance, standing, and walking. A chair (with back support), a tape measure, a cone, and a timer are required to perform the activity. The execution is carried out by order of the therapist (Ng & Hui-Chan, 2005; Hiengkaew et al., 2012). The examinee gets up from the chair, which is the starting position, walks parallel to the tape [three (3) meters], goes around the cone, comes back, and sits in the chair. Timing starts from the moment the examinee is asked to stand up after the appropriate instructions have been given. The **PBS** is a qualitative assessment test of the motor behavior of children aged four (4) to eighteen (18) years. It consists of fourteen (14) elements based on daily life activities starting from the easiest to the most difficult. The scale score ranges from 0-4 for each test, with 0 representing the lowest level of performance and 4 the highest level of performance (Laspa et al., 2020).

### *Intervention*

In the final sample of 15 children and adolescents with Trisomy 21, an assessment was made that included assessment of gait, identification of unequilateral, presence of valgismo, ability to use upper limbs, grasping

ability, and shape of hand and phalanges. Weight was measured with a precision scale and height with a precision tape measure. Mid-arm circumference was measured, first at the participant's preferred limb and then at the other upper limb. Standing waist circumference was also measured after removing the participant's shirt (Bezalel et al., 2010).

Handgrip strength measurement was performed on both upper limbs bilaterally. It was performed with two dynamometers, the first with the Jamar hydraulic dynamometer and the second with the MicroFet 4 electronic dynamometer. For the dynamometer, the participant was placed in a wooden chair specific to the dimensions of the participants without arms, while having a back support.

Balance was assessed with TUG and PBS. The application of the TUG was carried out in a specially designed corridor of the clinic. In PBS, commands were given to the participant in order, as indicated by the PBS protocol.

### 3 Results and Discussions

#### 3.1 Results

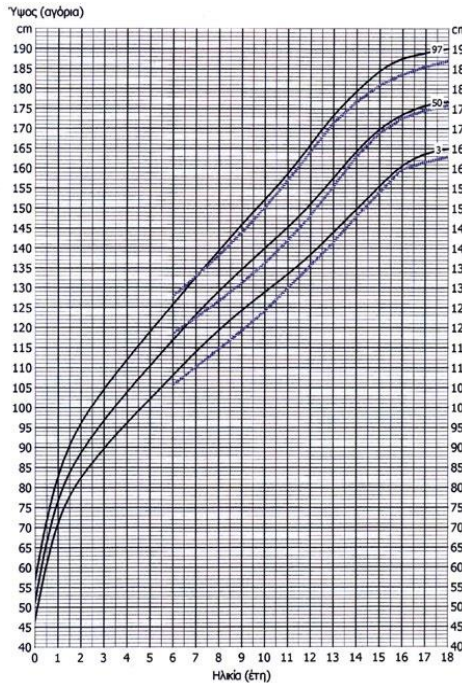
##### Demographics

Of these 15 patients, 5 were boys and 10 were girls. Patient measurements were performed under the same conditions. Baseline data, corresponding means, and standard deviations between patients are listed in Table 1.

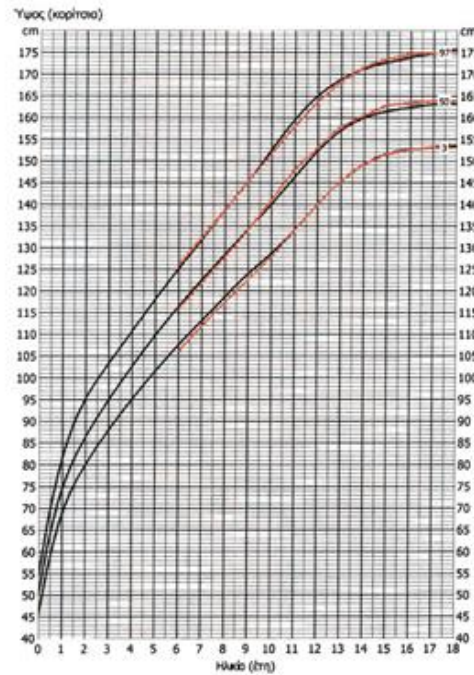
Table 1  
Somatometric characteristics of the sample

Variables	Male (n=5)	Female (n=10)	Total Sample (n=15)
Age	10,4 ± 5,6	8,4 ± 1,1	9 ± 7
High (cm)	1,36 ± 0,26	1,22 ± 0,8	1,26 ± 0,36
Weight (kg)	41,4 ± 28,3	30 ± 13,9	33,7 ± 36
Perimeter waist (cm)	59,2 ± 19,8	61,4 ± 7,6	60,66 ± 18,34
Perimeter humerus(cm)	21,86 ± 6,14	18,67 ± 4	19,73 ± 8,27
Valgus	4	9	13

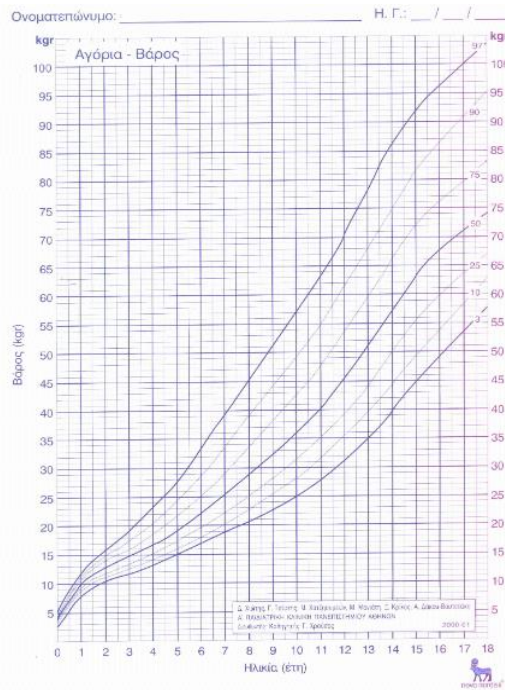
All of the children in the sample met the normal growth curves of somatometric elements of height, and weight according to age and sex. Relatively, the height parameter in boys and girls ranged from min=10n/100 to max=90n/100, while the weight parameter ranged from min=10n/100 to max=95n/100.



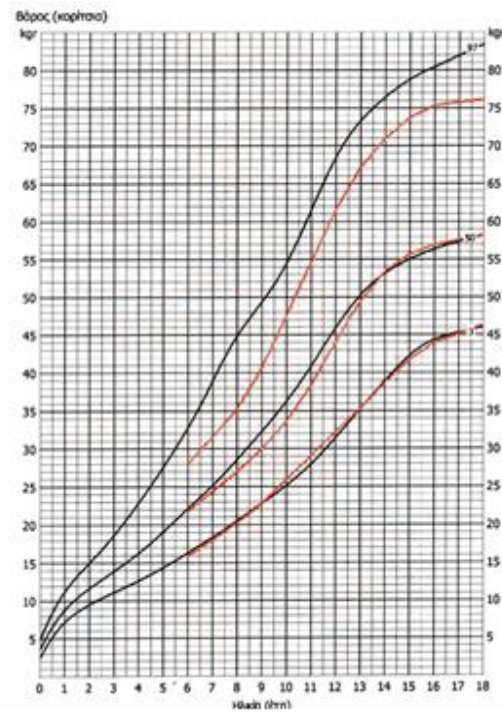
Graph 1. Height for boys



Graph 2. Height for girls



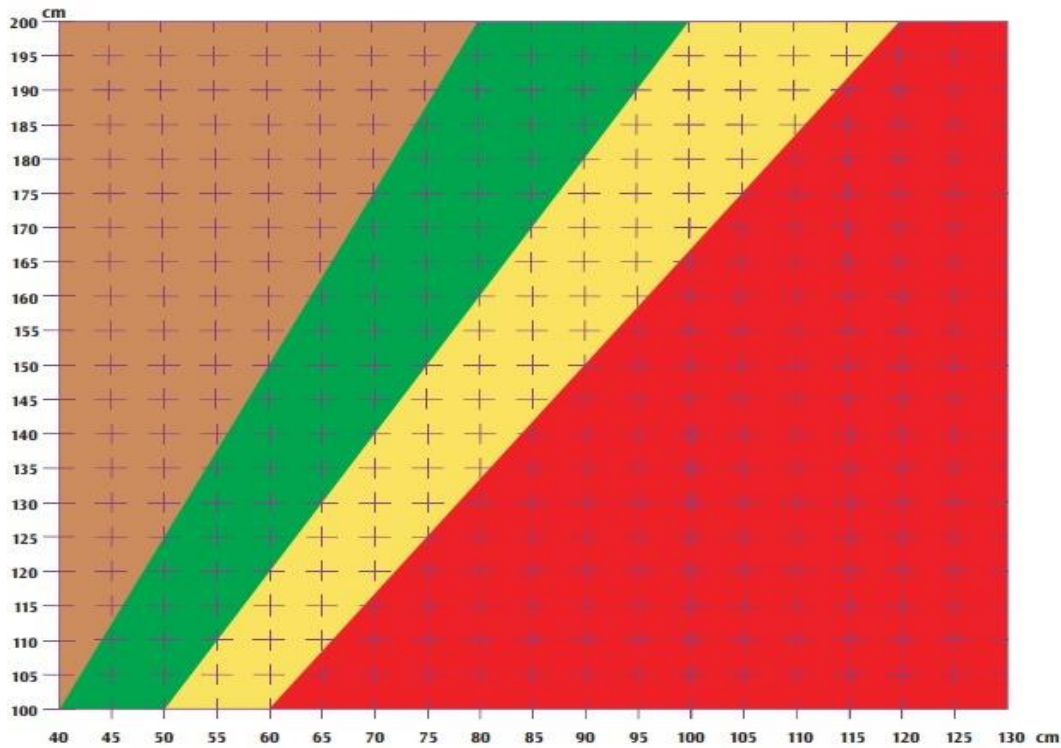
Graph 3. Weight for boys



Graph 4. Weight for girls

The average waist circumference was 60.66cm. in the total sample, with the average for boys being 59.2cm. and for girls 61.4cm. The average circumference of the humerus was 19.73cm. in the total sample, with the average for boys being 21.86cm. and for girls 18.67cm. In the total sample, as well as each individual participant was within the normal limits of waist and upper arm circumference to height. Out of the total

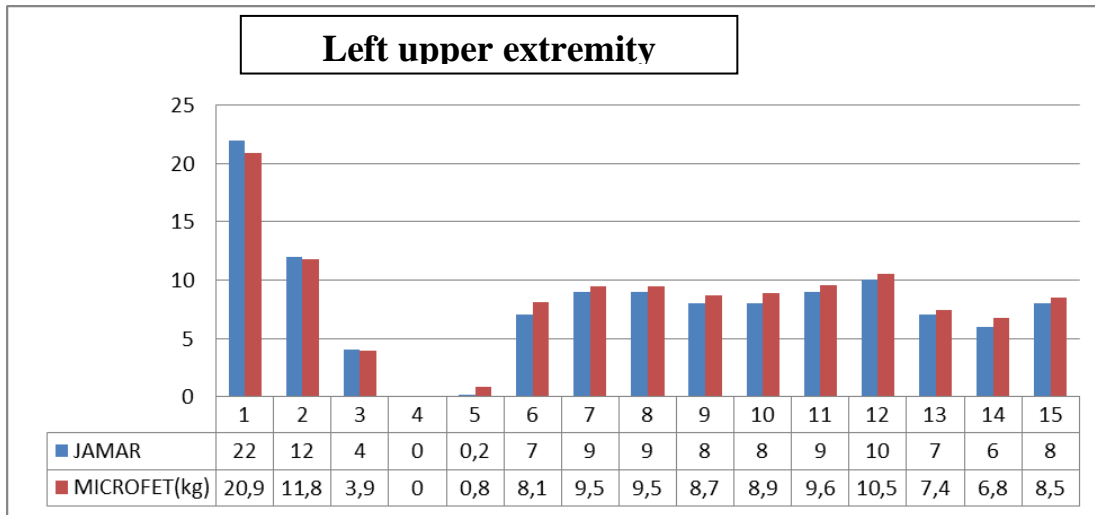
sample, thirteen (13) participants had valgus. A characteristic of children with trisomy 21 is that they show high rates of valgus, with the research sample presenting 86.6% of valgus.



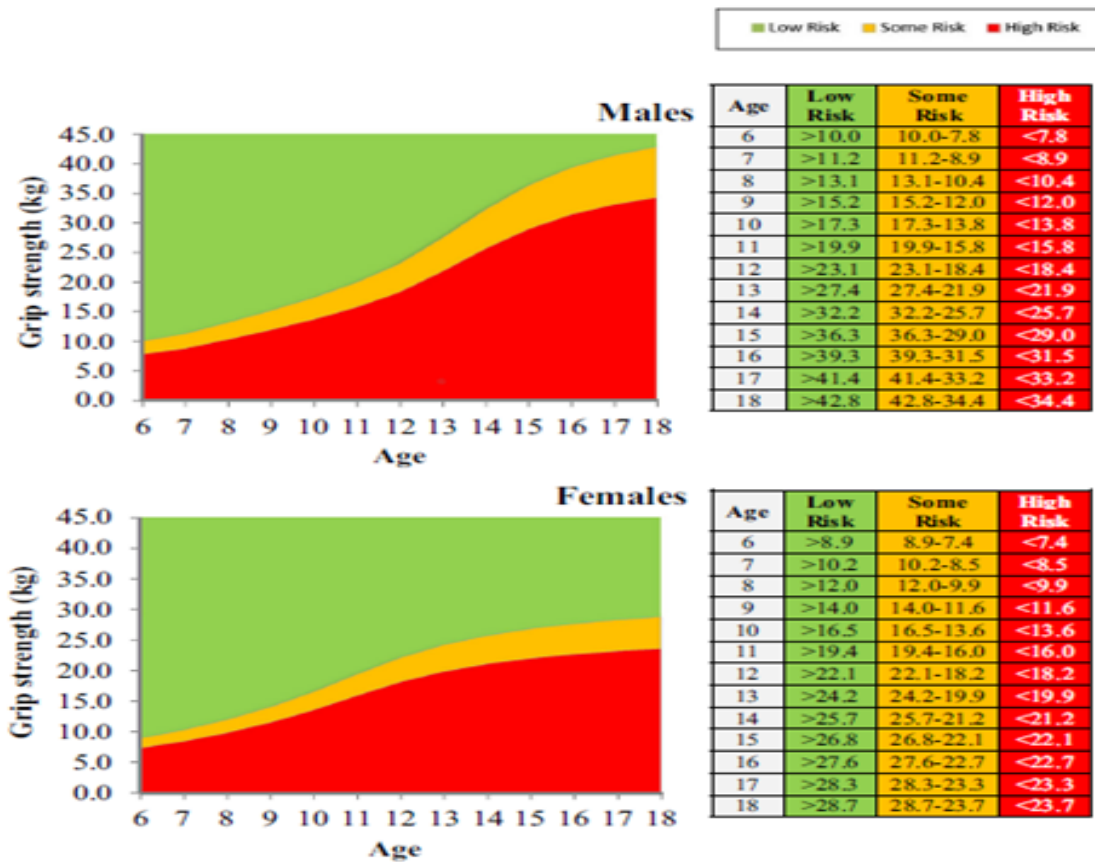
Graph 5. The normal ratio of waist perimeter to height

#### *Dynamometer*

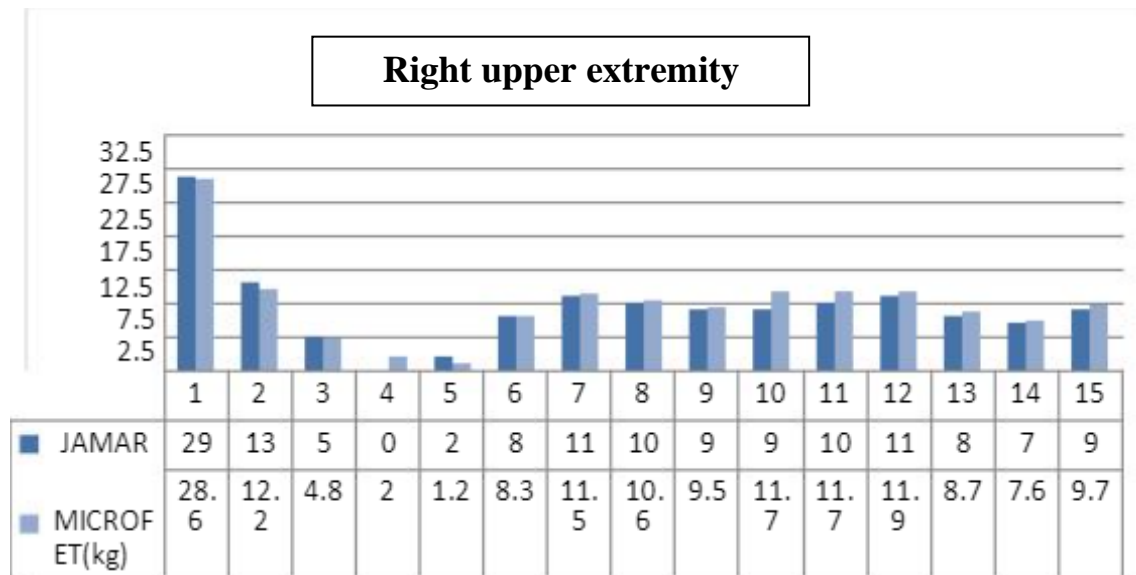
In the results of the measurements of the strength of the upper limbs (Hand Grip) according to the general population, increased deviations are recorded in the entire sample. Specifically, based on the results of the measurements, 100% of the participants in boys and girls, in all upper limb handgrip strength measurements with both dynamometers, register a high degree of difficulty and risk (High Risk). It appears that the handgrip strength in these children does not keep pace with the normal limits of the healthy population. In all measurements, the strength with both dynamometers used was below the lower limits of the general population.



Graph 6. Jamar and MicroFet 4 left upper limb dynamometry for each participant



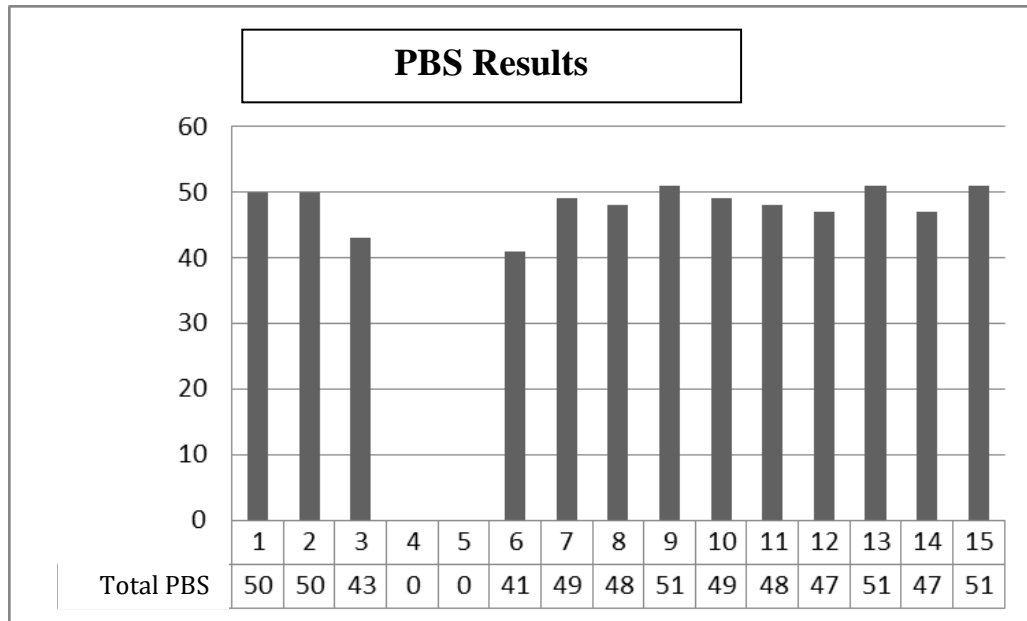
Graph 7. Jamar and MicroFet 4 right upper limb dynamometry for each participant



Graph 8. Normal upper limb strength values of the general population of children and adolescents aged six (6) to eighteen (18) years. Low, medium, and high risk in boys and girls in relation to age

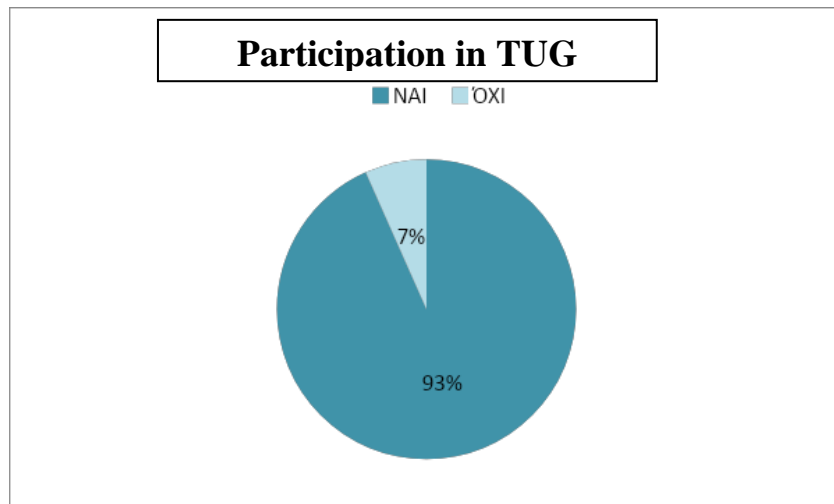
Percentages of deviations also refer to the balancing ability of children with trisomy 21. In the PBS results, the largest percentage of 20% of the sample recorded the best performance 51/56, followed by 13% 50/56, 13% 49/56, 13% 48/56, 13% 47/56, 6.6% 43/56, and the smallest percentage of 6.6% with 41/56. Two people could not run the PBS. The entire sample recorded difficulties in balancing ability with grade deviations. The lowest score was min=41/56 and the highest max=51/56. 20% with 51/56 and 13% with 50/56 have a small balance deviation, 39% with a score from 47/56 to 49/56 register greater balance problems while 13.2% with 43/56 and 41/56 record a large deviation and significant difficulties in balancing ability.

PBS scores show that children and adolescents with trisomy 21 have balance problems compared to the general population. With the highest result recorded at 51/56 and the lowest at 41/56, it seems that these children lag behind the healthy population. Children and adolescents with typical development at the respective ages have higher percentages with the majority recording 56/56.

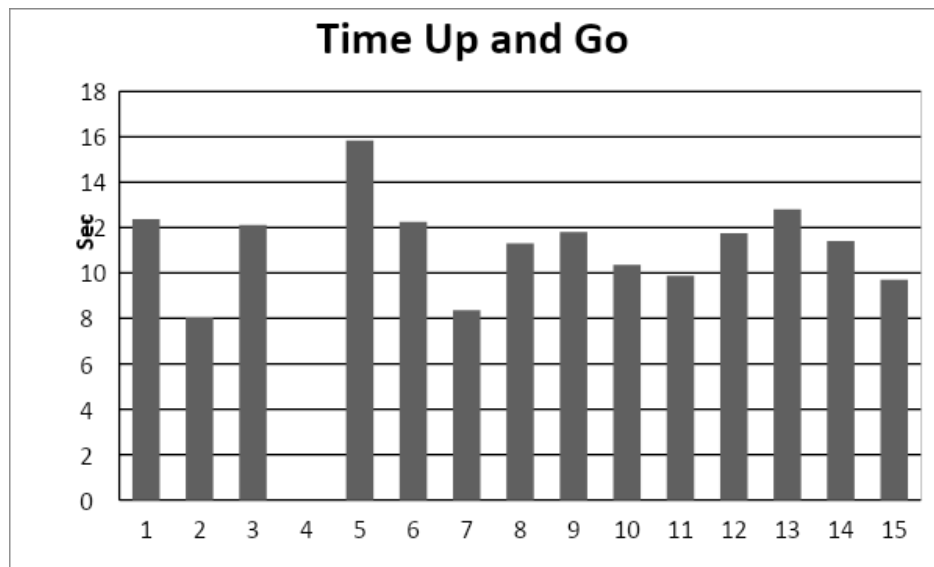


Graph 9. Pediatric Balance Scale scores for each participant

The TUG was performed by all but one participant. All participants who ran it completed it within the allowed time limits, except for one who clocked 15.8 seconds, with the shortest time being 8.04 seconds. and the longest 15.83sec. According to [MacNee \(2009\)](#), the normal time limit for completing the TUG for children aged six (6) to sixteen (16) years of typical development is from 4.9sec. up to 6.3 sec., while according to [Katz-Leurer et al. \(2010\)](#), the normal time limit for performing the TUG for children aged seven (7) to thirteen (13) years of typical development, is from 5.3 sec. up to 6.7 sec. It thus appears that despite correctly completing the TUG within the allowed time limits, 100% of participants who completed the procedure appear to deviate greatly from the general population's time limits and achieve high times relative to the normal average.



Graph 10. TUG participation rate



Graph 11. TUG results for each participant

### 3.2 Discussion

To measure and assess indicators of physical well-being, upper extremity functional complexity, and balance ability in children and adolescents with trisomy 21, various methods have been introduced to develop an objective assessment system. According to the somatometric data recorded in this research, it seems that children and adolescents with trisomy 21 follow the normal values of their typically developing peers in height and weight, while as mentioned in the literature, in these children, vulnerability prevails (Kurtieva et al., 2021).

The upper extremity strength measure has been described as the force exerted by the combined contraction of all the muscle groups that flex the extremity. Accurate measurement of upper limb muscle strength provides insight into children's development. In this study force measurement with both the Jamar hydraulic dynamometer and the MicroFet 4 electronic dynamometer can be applied to the entire trisomy 21 population. The results showed that all the children who performed the measurement were classified in the (High-risk category compared to the general population. This shows that the development of upper limb strength is deficient in children with trisomy 21 compared to age-matched individuals with typical development (Ehrich et al., 2011; Ashoor et al., 2012).

The lower performance of children with trisomy 21 compared to the healthy population, for grip, strength, and manual dexterity, is probably influenced by some factors such as intellectual deficit, hypotonia, and physical growth, which stops early. Atypical grip patterns in children and adolescents with trisomy 21, such as using fewer fingers and overextending them when grasping objects, may compromise manipulative skills (Priosti et al., 2013).

Also, body mass, body fat percentage, and nutritional status are additional factors that affect strength measurements (Priosti et al., 2013). Regarding gender, the results showed that grip strength was similar for girls and boys.

The balance ability of children with trisomy 21 who participated in this study, shows deviations related to postural control compared to typically developing children. The highest score recorded on the Pediatric Balance Scale is 51/56, while the completion of the TUG by 100% of the participants achieving higher times than the normal average demonstrates the balance deviations of these children in relation to the general population. Discrepancies in scoring tools such as the Pediatric Balance Scale assess functional balance and movement disorders and more specifically posture and balance as categorized by the ICF model (Chen et al., 2013; Jantakat et al., 2015).

## 4 Conclusion

The present study is one of the first studies in the world literature to correlate handgrip strength with balance ability in children and adolescents with trisomy 21. The development of handgrip strength is delayed in children with trisomy 21, compared to the general population. The balance ability of children with trisomy 21 also shows deviations related to postural control compared to typically developing children.

Discrepancies in handgrip dynamometry and PBS show:

- Difficulty in achieving motor milestones quantitatively and qualitatively,
- Fatigue easily,
- Increased falls,
- Difficulty in balancing and coordinating movements.

### *Limitations*

A limitation of the study is the small sample of participants. The study needs further investigation in a larger sample of children for the optimization and reliability of the results, to validly delineate the normal handgrip strength values of children and adolescents with trisomy 21 about their age.

### *Acknowledgments*






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## Biography of Authors

	<p><b>Polymeris Prosiniklis</b>          Enrolled in the Department of Physiotherapy at the University of Thessaloniki in Greece in September 2016 and graduated in December 2020. During his academic journey, he actively attended various educational conferences. Additionally, he completed his undergraduate thesis focusing on neurological physiotherapy for children, and in the summer of 2024, he finished his bachelor's in Pediatric Physiotherapy.  <i>Email: <a href="mailto:polysprosiniklis@gmail.com">polysprosiniklis@gmail.com</a></i></p>
	<p><b>Kallistratos A. Ilias</b>          PT, PhD Professor Vice President of Physiotherapy Department Faculty of Health Sciences International Hellenic University Lab of Basic and Applied Research in Physiotherapy Rehabilitation, Director Dept. Physiotherapy.  <i>Email: <a href="mailto:elikall@hotmail.gr">elikall@hotmail.gr</a></i></p>
	<p><b>Alexandra Xristara-Papadopoulou</b>          She is a former teacher in the International Hellenic University. In 1978 she got her degree in physical therapy. In 2006 Ph.D title studies from the University National Sport Academy &lt;&lt; Vasil Levsky &gt;&gt; of Sofia from a physical therapy department, recognized by DOATAP, with theme &lt;&lt; Comparative study of effectiveness of three physical therapy methods in the treatment of cystic fibrosis &gt;&gt; , &lt;&lt; Comparative study of the efficiency of three physiotherapy methods in the treatment of Cystic Fibrosis &gt;&gt;.  <i>Email: <a href="mailto:hristara2@hotmail.com">hristara2@hotmail.com</a></i></p>
	<p><b>George Tsigaras</b>          PT PhD, MSc, NDT, SI 1st Department of Pediatrics, Aristotle University Thessaloniki Hippokratio General Hospital Adress: 49 Konstantinoupoleos str, Thessaloniki 54642, Greece,  <i>Email: <a href="mailto:tsigarasphysiopediatric@gmail.com">tsigarasphysiopediatric@gmail.com</a> , <a href="mailto:tsigaras@auth.gr">tsigaras@auth.gr</a></i></p>
	<p><b>Anna Chalkia</b>          She is an Assistant Professor at the International Hellenic University of Thessaloniki, Department of Physiotherapy.  <i>Email: <a href="mailto:annachalkia18@gmail.com">annachalkia18@gmail.com</a></i></p>

	<p><b>Efthymia Vargiami</b> MD,PhD Professor of Child Neurology Pediatrics 1st Department of Pediatrics Aristotle University of Thessaloniki Hippokratio General Hospital 49 Konstantinoupoleos str, Thessaloniki 54642, Greece <i>Email: <a href="mailto:evargiam@auth.gr">evargiam@auth.gr</a></i></p>
	<p><b>Dimitra Katsantoni</b> Enrolled in the Department of Physiotherapy at the University of Thessaloniki in Greece in September 2018 and graduated in December 2022. During her academic journey, she actively attended various educational conferences and participated in the university's journal club. Additionally, she completed her undergraduate thesis focusing on neurological physiotherapy for children. <i>Email: <a href="mailto:dimkatsantoni1@gmail.com">dimkatsantoni1@gmail.com</a></i></p>