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# ANALYSIS OF THE EFFECTIVENESS OF GAIT TRAINING IN A PEDIATRIC PATIENT WITH MOTOR SEQUELAE FROM A STROKE AND A DIAGNOSIS OF DIGEORGE SYNDROME

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**Abstract**: DiGeorge Syndrome (DSG) is a genetic alteration correlated to a multisystemic expression, with a wide clinical manifestation, including more than 180 signs. The etiology is related to losses in specific regions of the genome. Therefore, it is essential to know and identify it, and multidisciplinary intervention is necessary due to its complexity. Cerebrovascular accident (CVA) is a pathology in which blood flow to the brain is altered, causing permanent sequelae. It is classified into two types, ischemic stroke and hemorrhagic stroke, and can affect individuals under the age of 18 causing, among other changes, motor sequelae. The relevance of this study is to demonstrate the fundamental role of physiotherapy in improving and enhancing the walking ability, functionality and quality of life of affected individuals, since the neuroplasticity process in children is greater than in adults and the elderly. The aim of this study was to analyze the effectiveness of gait training in a pediatric patient with motor sequelae from a stroke and a diagnosis of DiGeorge Syndrome. The methodology of this research is a clinical case study with a qualitative approach, with a participant, male, black, living in Porciúncula-RJ, aged 9, who underwent weekly physiotherapy sessions lasting 50 minutes from March to October 2024. The following techniques were applied passive stretching, passive and active joint mobilization with emphasis on the lower limbs, assisted active, free active and resisted active kinesiotherapy, orthostatic training, sensory motor training and gait, as well as functional range exercises integrating physical and cognitive training. The results were satisfactory in terms of improving the patient's overall posture, trunk control, gaining overall muscle strength with an emphasis on the lower limbs, adjusting muscle tone, improving static and dynamic proprioception, improving gait and greater confidence and independence in walking, even though the patient still needed support to do so. It can therefore be concluded that there is an important correlation between muscle strength, proprioception and gait. According to the above, gait training has a positive impact on preventing and reducing the rate of falls, providing functional independence and greater safety and confidence for individuals in carrying out their ADLs.
**Keywords:** DiGeorge Syndrome, Stroke, Gait, Neuroplasticity

## INTRODUCTION

DiGeorge Syndrome (DSG), which is also recognized as Velocardiofacial Syndrome (SVCF) or 22q11.2 deletion syndrome (DS 22q11.2), is a genetic alteration correlated to a multisystemic expression, being the most common microdeletion found in living beings, with a wide manifestation, including more than 180 clinical signs and with an incidence of approximately 1 in every 4000 live births. Therefore, it is essential to investigate and identify the alterations found in individuals (FREIRE et al., 2015).

The 22q11.2 deletion syndrome originates from modifications and displacements of neuroectodermal corpuscles from the neural crest to the 3rd and 4th pharyngeal arches and pouches during the embryogenesis process in humans. However, clinical traces have also been found indicating deformities in the 1st and 6th branchial arches and pharyngeal pouches, with the Tbx1 gene associated with the defect in this cell migration process. It is located on the long arm of chromosome 22 (q), being a critical region, at position 11.2, causing a gene haploinsufficiency that is also related to cardiac and coronary malformations (ARAGÓN et al., 2020).

In terms of diagnosis, although there is a wide clinical range, there are a few main alterations that serve as screening: heart defects congenital congenital heart defects hypoparathyroidism e immune dysfunction. There-

fore, efficient identification at an early stage guarantees a better treatment prognosis, and multidisciplinary intervention is necessary due to the complexity and scope of this pathology, with the aim of improving quality of life (SANTOS et al., 2023). O stroke stroke stroke (CVA) which can also can also the nomenclature of cerebrovascular accident (CVA) and stroke, is defined as a neurological pathology in which there is an alteration in the blood flow to the brain, causing necrosis of the neurons in the affected brain region, generating permanent sequelae. It can be classified into two types: ischemic stroke, in which there is an obstruction of blood flow, or hemorrhagic stroke, in which there is a rupture of blood vessels, with a high risk of stroke. morbidity and mortality rate and strong social impact among the diseases that affect the nervous system (JARDIM et al., 2023).

The pathological incidence of stroke ranges from 2 to 13 individuals in 100,000 occurrences in children under 18 years of age, of which 50% are ischemic strokes. Thus, among the most recurrent etiologies are congenital heart disease, sickle cell anemia, vasculopathies, hematological diseases, blood vessel malformations and aneurysms, among others. Thus, causing motor, cognitive and sensory sequelae, significantly affecting the quality of life of individuals affected by this disease (GERZSON et al., 2015).

Diagnosis is based on clinical evidence, which varies depending on the location and extent of the injury. This can take the form of paresis, plegia, paresthesia, absence of homolateral sensory recognition, cognitive and psychological alterations, postural instability, imbalance, among others. When the event occurs, the affected brain region shows edema, tissue degradation and also deterioration of the smaller caliber arteries, so the severity and degree of mortality are greater depending on the extent of the injury and the affected region (SANTOS et al., 2020).

Among the main motor alterations caused by stroke, loss of balance, altered tone, reduced muscle strength in the lower limbs (LL) and reduced aerobic capacity directly and potentially interfere with gait speed and quality, as well as being important predictors of falls. Therefore, the role of physiotherapy in improving functionality, quality of life, performing activities of daily living (ADLs) and minimizing the deleterious effects of pathology on affected individuals is essential (ASA et al., 2021).

Improving walking ability is essential in individuals with stroke sequelae, since gait is analyzed as a complex movement from a kinetic-functional point of view. It is defined as an alternating movement of the lower limbs and is divided into phases, classified as: swing or oscillation and support, which are carried out in a single movement. continuous integration of the upper limbs (upper limbs) and lower limbs (lower limbs). Proprioception during gait is divided into the ability to sustain the center of mass on the surface and simultaneously reduce the sway in the lower and upper regions of the body during the movement and the ability to concentrate the center of mass on top of the base of support during walking.

Thus, both static and dynamic balance are essential in the functional rehabilitation process (RHYU & RHI, 2021).

This study is justified by the fact that it provides a significant improvement in the functional capacity and gait of individuals affected by stroke while still in childhood, since pediatric patients have a better prognosis due to the fact that the brain's neuroplasticity mechanism is greater in children when compared to adults and the elderly. In this process, the central nervous system (CNS) reorganizes itself based on environmental changes and stimuli.

DiGeorge syndrome is a pathology with numerous clinical manifestations, making its diagnosis quite rare. Among the clinical findings are congenital heart defects that can affect fundamental structures of the circulatory system; it is an important risk factor for strokes in childhood, since they arise from specific causes, often an underlying disease. Knowledge about this condition, its influence and its consequences throughout human development is therefore essential.

The main objective of this study was to analyze the effectiveness of gait training in a pediatric patient with motor sequelae from a stroke and a diagnosis of DiGeorge Syndrome.

Figure 1- Passive joint mobilization.

Figure 2 -Calf pump exercise passively associated with stretching the posterior muscle chain of the lower limbs. Source: personal archive.

Figure 3 -Abdominal exercise with Peanut Ball. Source: personal archive.

Figure 4 -Orthostatism with partial weight suspension in the Therasuit Cage. Source: personal archive.

Figure 5 -Sit and stand exercise on the steep. Source: personal archive.

Figure 6 -Patient performing sitting and standing exercises on the steep without the help of the physiotherapist. Source: personal archive

Figure 7 -Patient performing gait education training on a neurofunctional treadmill with the help of a physiotherapist. Source: personal archive

Figure 8 -Gait training on the ground combined with strength training. Source: personal archive.

Figure 9 -Functional reach training. Source: personal archive.

Figure 10 -Patient performing a dual task (motor and cognitive activity). Source: personal archive

Figure 11 -Patient in orthostatic position without physiotherapist assistance. Source: personal archive.

Figure 12 -Patient walking with minimal support from the physiotherapist. Source: personal archive.

## METHODOLOGY

The methodology of this research is a case study, in which the male patient, belonging to the black ethnic group, currently 10 years old and living in the city of Porciúncula in the state of Rio de Janeiro, was diagnosed with DiGeorge Syndrome and because of it, and its other complications and disorders, he suffered a stroke, causing motor impairment.

The site of the research was the Physiotherapy School Clinic of the Universidade Iguaçu - Campus V, Itaperuna-RJ. At the aforementioned institution, data was collected on the patients and their pathologies, and a pediatric physiotherapy assessment form was also applied. The sessions took place regularly on Wednesdays at 2:30 p.m. from February to November 2024, with two assessment periods, an initial assessment in May 2024 and a reassessment in August of the same year.

During the sessions, passive stretching techniques were applied, passive and active mobilization of the joints with an emphasis on the lower limbs, assisted active, free active and resisted active kinesiotherapy, orthostatic training, sensory motor training and walking with a neuropsychomotor circuit, as well as functional range exercises integrating physical and cognitive training. These techniques were aided by a number of instruments, such as: bobath ball, peaunut ball (bean ball), 0.5kg and 1kg shin guards, steep, elastic bands, cones, mini rings, 25cm over ball (milk ball), stick, therasuit cage and neurofunctional treadmill.

The physiotherapy treatment selected in this case study was planned and prescribed individually according to the patient's kinetic-functional and cognitive limitations, with the aim of enabling functional independence through gait training in a pediatric patient with motor sequelae from a stroke and a diagnosis of DiGeorge Syndrome.

The approaches selected focused on strength training with an emphasis on the quadriceps muscle group, made up of the muscles: rectus femoris, vastus lateralis, intermedius and medialis, located in the anterior region of the thigh involving the upper portion of the femur; Stretching of the muscles that make up the posterior muscular chain of the MMII's, with emphasis on the hamstrings, made up of the biceps femoris, semitendinosus and semimembranosus muscles and the triceps suralis made up of the gastrocnemius and soleus muscles; Static and dynamic balance training, varying base changes (stable and unstable) and gait training using the neurofunctional treadmill and the floor.

In addition, passive mobilization of the joints of the lower limbs was also carried out: hip (hip joint), knee (tibiofemoral joint) and ankle (tibiotarsal joint); passive calf pump exercise for stretching and activation of the sural triceps; strengthening of the abdominal muscles was carried out to improve trunk control and overall posture, which is essential for the orthostatic position; Orthostatic training with the aid of the Therasuit cage and partial weight suspension and later with unloading of the patient's body weight; In conjunction with the aforementioned procedures, psychomotricity was used, integrating psycho-emotional and motor activities with the aim of promoting the child's overall development, improving movements, motor coordination, balance, rhythm, improving cognition and integrating the body's systems.

The patient was not cooperative with the appointments, showing psychomotor agitation, diverting attention and concentration easily and not responding to verbal commands, so it was often necessary to use playful resources to improve adherence to the treatment, such as the use of toys related to his hyperfocus, music therapy and other audiovisual resources.

Joint mobilization, which is one of the manual therapy techniques, was used with the aim of increasing mobility and gaining range of motion in the aforementioned joints. the patient in dorsal decubitus and passively in the hip, tibiofemoral and tibiotarsal joints, maintaining stability and control of the movement.

A calf pump exercise was performed with the patient in the supine position, combined with stretching of the posterior muscle chain (hamstrings and triceps suralis), since the constant use of the AFO orthosis prevents activation of the triceps suralis muscles, keeping the ankle in plantar dorsiflexion at all times, which hinders venous return and blood circulation. The patient also uses a wheelchair, where he remains in a sitting position for long periods, causing shortening of the ischitibials, making it impossible to properly extend the lower limbs while walking. These techniques are therefore of great importance.

The patient performed active-assisted abdominal exercises to strengthen the abdominal muscles that make up the CORE (rectus abdominis, internal and external oblique and transverse abdominis) using a peaunut ball to improve muscle activation. This exercise helps to improve the strength of the abdominal muscles, posture and trunk control, essential elements for walking.

The patient performed orthostasis using partial suspension of body weight in the Therasuit Cage, with the help of the physiotherapist. The use of the orthostatic position is extremely important for the better overall systemic functioning of the body, as it is one of the ways of training static balance and a preparatory position for walking.

The patient performed the sit and stand exercise on the steep using a closed elastic band and a 25 cm over-ball to better activate the quadriceps muscles, especially the external (lateral) muscles and prevent the patient from making compensatory movements (dynamic valgus). The muscle group worked in this exercise is essential for maintaining orthostasis and walking.

The patient performed the quadriceps strengthening exercise (sitting and standing) on the steep independently, without the help of the physiotherapist.

The image shows the patient performing gait training on a neurofunctional treadmill with the help of a physiotherapist who teaches the steps and stride. The equipment offers resistance and speed on a stable, controlled surface, where the patient must adapt to the pace imposed, and obstacles can be added, which is important in preparing for gait training on the ground on different surfaces, where static and dynamic balance are required.

The patient walked independently, leaning on a stick, associated with strength training using a 0.5kg shin guard. The gain in muscle strength in the lower limbs is fundamental and directly related to gait quality and the acquisition of proprioception.

The patient underwent functional range training associated with psychomotor activity, developing attention, concentration, precision and fine motor coordination skills, and responding to verbal commands. These skills are important for the orientation of body segments in space, the avoidance of obstacles, the perception of visuospatial changes and the prevention of possible falls due to visomotor improvement and refinement.

The patient performed a double task of functional reaching associated with orthostatic training, maintaining static balance.

The patient remained in an orthostatic position for 25 minutes and did not need any help to remain orthostatic, maintaining static balance.

The patient gait adapts independently to their condition, requiring minimal support and adopting characteristic compensations

due to their neuromotor condition as a result of the stroke. This motor compensation is important for functional autonomy and gait without high energy expenditure, due to the prognosis of individuals affected by neurological injuries, where the rehabilitation of the patient's motor skills is essential.physiological and referential gait pattern is not possible due to neuronal damage.

There is a broad and intimate relationship between physiotherapeutic procedures and brain plasticity, whereby by performing new exercises and activities the individual is able to carry out the process of motor neurolearning, in which the brain reorganizes itself and adapts to new neuropsychomotor stimuli. This process, called cerebral neuroplasticity, is greater in children than in adults and the elderly, making it essential to carry out physiotherapy early on, especially in cases of childhood stroke diagnosis.

## RESULTS AND DISCUSSIONS

This case study achieved satisfactory results in terms of improving the patient's overall posture, trunk control, gaining overall muscle strength with an emphasis on the lower limbs, adjusting muscle tone, improving static and dynamic proprioception, improving gait and greater confidence and independence in walking, even though the patient still needed support to do so, based on the procedures proposed and carried out during the physiotherapy treatment.

The patient progressed and did not need aids such as the closed elastic band and the over ball in the medial quadriceps region, in order to prevent movement compensations and correct muscle activation, showing a gain in muscle strength in the lower limbs, increasing the CRM for 56 with strength grade 4 according to the reassessment, performing the movement in an independent, cadenced manner. These gains highlight the importance of kinesiotherapy, especially active resistance, in the physiotherapeutic treatment of the participant in this case study.

The progress of the patient, who at the start of treatment was unable to remain in an orthostatic position due to reduced muscle strength. The act of walking on the neurofunctional treadmill with the addition of resistance from the use of the device at different speeds indicates a gain in muscle strength in relation to the adequacy of their body weight, not needing any partial weight support (previously used for better education of the gait pattern) and the acquisition of both static and dynamic balance.

In addition to progressing to gait training on the ground with variations in different types of surfaces, the patient also showed progress when performing the gait with an additional load and still maintaining an adequate speed; previously, this same exercise was performed without an additional load, in addition to acquiring greater independence in walking by leaning on the stick; at the start of treatment, the patient only used wheelchairs.

The patient showed neurocognitive development when he voluntarily complied with the verbal command requested, performing the act of reaching and placing the ring on the cone. The development of these skills is extremely important during the act of marching, which goes beyond physical skills and acquisitions; it requires spatial orientation, attention, a visomotor component, motor coordination, confidence in the act and concentration on the movements performed.

The participant's development and progress at the end of treatment has been proven, with significant improvements in overall body posture, trunk control, muscle strength, proprioception and confidence in the act of walking. At the first appointments, he started with orthostatic training without full weight bearing, using partial support due to the muscle weakness present, walking requiring full support

and lacking confidence in his gait. At the end of the treatment, with sufficient muscle strength, he showed progress with orthostatic training with full body weight unloading, and began to walk independently, requiring minimal support, with improved speed (increase), rhythm and cadence of steps during the act of walking.

In addition, the participant showed confidence during ambulation with minimal support at the end of the treatment in this study of case, which is extremely important for the continuation of treatment afterwards.

In the final assessment of the treatment, carried out on October 23rd, 24th, the patient was reassessed using the Tinetti Balance and Gait Scale, obtaining a higher score than during the first assessment when the participant was admitted to the clinic to start treatment. The score obtained was 14 out of a total score of 28 points, still showing a high risk of falls.

The aim of this study is to demonstrate the importance of gait training and, consequently, functional independence in pediatric patients diagnosed with stroke who have suffered motor sequelae. According to the results observed and acquired, there was a gain in muscle strength, especially in the lower limbs, an improvement in static and dynamic proprioception, a gain in overall postural control and confidence and independence in walking.

According to Dutra et al (2024), who researched the effects of a kinesiotherapeutic intervention on the gait of hemiparetic individuals, including passive joint mobilization and stretching exercises; the studies showed that the effects of the intervention performed increased gait speed, knee range of motion increased after intervention on both the paretic and non-paretic sides. These results were confirmed in this study, since the patient had limited ROM and shortening of the posterior muscle chain, obtaining a significant improvement in gait and mobility of the lower limbs with the use of these techniques.

According to Camerin et al (2021), gaining muscle strength in the lower limbs is essential in the rehabilitation process to ensure the functionality and independence of stroke patients. He established a positive correlation between lower limb muscle strength and static balance in the affected lower limb with gait speed. Therefore, patients who have lower lower limb muscle strength and altered static balance have lower gait speed of . The focus of this study were exercises of strengthening and maintaining static balance, corroborating the author's statement. With the techniques applied and mentioned above in the description of the physiotherapy treatment, the patient gained muscle strength and static proprioception, thus increasing his walking speed.

According to Ferla et al (2015), good trunk control and effective balance play a fundamental role in the effectiveness of activities performed by the upper and lower limbs of the motor side affected by the stroke. Gait, for example, is closely associated not only with upper and lower limb muscle strength on the clinically affected side, but also with balance control. This statement attests to the results obtained in this study, given that as muscle strength increased, so did posture and gait independence.

## CONCLUSION

Physiotherapy is known to be extremely important in restoring the quality of life and functional independence of individuals affected mainly by neuropathies. The quality of gait is one of the main kinetic-functional losses that a stroke-affected individual of any age can suffer. This study therefore obtained satisfactory results in relation to the prescription and individualized application of physiotherapeutic techniques to achieve an efficient gait.

It can therefore be concluded that there is an important correlation between muscle strength, proprioception and gait. According to the above, gait training has a positive impact on preventing and reducing the rate of falls, providing functional independence, greater safety and confidence for individuals in carrying out their ADLs autonomously.

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