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## Motor Behavior in Down Syndrome: Atypical Sensorimotor Control

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### 1. Introduction

In addition to the phenotype characteristics, the Down syndrome is accompanied by multi-system pathological conditions. These conditions involve delays in basic motor skills, motor impairments and abnormalities in postural and gait control. A large body of literature has documented delays in basic motor skills, such as walking, reaching and grasping, in children with Down syndrome. Also their movements are slower and more variable. There has been debate in the literature over the real cause of atypical motor behaviors observed in individuals with DS. Possible explanations are related to cognitive limitations, biomechanical deficits, neurological disorder, abnormal sensorimotor integration, compromised somatosensory system or adaptive choice. In this chapter, we will first discuss similarities in the control of movement, posture and balance between nonhandicapped individuals and those with DS, and then consider differences. Second, we will review the evidence that relates to whether individuals with DS have specific sensorimotor deficits. Finally we will explore possible explanations for the cause of atypical postural behaviors observed in individuals with DS.

### 2. General characteristics of individuals with Down syndrome

Down syndrome (DS) is a chromosomal anomaly that leaves the individual affected with an additional chromosome (the 21st). The syndrome is associated with approximately 1/800 live births and is one of the leading causes of intellectual disabilities. Intellectual disability of some degree is invariably present in individuals with DS, but, unusual for any major chromosomal disorder, levels of impairment vary greatly across individuals. Most of those with DS fall within the moderate to severe range of disability, but some show levels of cognitive abilities that are borderline normal while others experience profound mental retardation (Roizen, 2002).

In addition to the phenotype characteristics, the Down syndrome (DS) is accompanied by multi-system pathological conditions. The individual with DS faces numerous movement control challenges. These challenges involve delays in basic motor skills, motor impairments and abnormalities in postural and gait control.

The literature has documented delays in basic motor skills, such as walking, reaching and grasping, in children with Down syndrome (Palisano et al., 2001). The complexity of this

developmental pattern is also exhibited in what is described as differences in the structure of cognitive and sensorimotor functioning. That is, children with DS have been found to exhibit particular difficulties in certain areas (e.g., linguistic skills; visual scanning; ability to attend, to discriminate and encode complex stimuli) compared with their overall level of mental function.

One of the most established findings is that children with DS are slower at both initiating and executing goal-directed movements compared to typically developing peers (Savelsbergh, et al., 2000). They also exhibit greater movement time advantages as the accuracy demands of the movement goal are increased (Hodges, et al., 1995). Commonly reported sensorimotor deficits exhibited by children with DS also include perceptual-motor slowness (Elliott & Bunn, 2004), limb control problems and decreased motor proficiency (Wuang, Lin, & Su, 2009). Also the motor items that required aspects of strength and balance (such as standing and walking) developed more slowly than other motor behaviors.

In summary, researchers have found the normal sequencing of motor development in infants with DS, although the pattern of development is slower and the variability higher (Van Dujin et al., 2010). Much of this developmental delay continues to be attributed to isolated factors such as low muscle tone. Ulrich et al., (1997) explain this delay by perception difficulty of postural responses, which undermines the sense of movement and its consequences. Uyanik et al., (2003) suggest the sensory integration dysfunction as a result of limited sensory experience.

Besides the development delay it is generally accepted that motor impairments are inevitably present, to greater or lesser extents, in individuals with DS. Their movements are slower and more variable. Evidences of slowness and lack of smoothness has been observed even in simple elbow-flexion movements (Almeida, et al., 1997), as well as in multijoint pointing tasks (Aruin & Almeida, 1997). As point out by Anson (1992) in a review of DS and reaction time, differences in simple reaction time have varied from 25% to greater than 300%. This slowness in reaction time can have two consequences. First, in all movements made in response to an external stimulus, the initiation of the movement is likely to be delayed and can therefore give the impression of slowness even if the actual movement itself is reasonably quick. Second, when individuals with DS are asked to perform sequences of movements, if each movement in the sequence is treated as a separate movement the sequence will be performed extremely slowly because of the increased reaction time to program each component movement.

### 3. Postural control

Also individuals with DS are characterized by instable postural control. They are unable to respond rapidly to changes in the environment (Haley 1986). Typically they take longer to initiate and complete a motor task and have difficulty maintaining equilibrium (Galli et al. 2007; Vuillerme et al. 2001). Ulrich et al., (2004) compared levels of stiffness and forcing in preadolescents with and without DS, analyzing gait patterns on a treadmill at different speeds and showing the same adaptation mechanism: all participants increased their stiffness and forcing. The difference between the two groups is explained by the authors in terms of different goals: people with DS actuated the adaptation as a compensatory strategy, in order to maintain stability and to overcome the ligament laxity and hypotonia that characterize DS, while the control group's aim was to optimize metabolic efficiency. Kubo and Ulrich (2006) compared toddlers with DS, to control group and observed that

individuals with DS showed wider step widths but not a larger ML displacement. The authors explained this finding speculating that the increase in step width contributes to ML stability by creating a wider base of support, but toddlers with DS cannot allow their nascent walking system to rock from side to side more than minimally, without losing control.

Although maintaining a bipedal position may appear to be simple, it requires integration of information arriving at the central nervous system (CNS) through the proprioceptive organs and senses, especially vision and the vestibular apparatus of the inner ear. In recent years, study of the behavior of the center of pressure (CoP) has emerged as a way of indirectly understanding the neuromuscular control of equilibrium. The CoP is the point location of the vertical ground reaction force vector. It represents a weighted average of all the pressures over the surface of the area in contact with the ground.

The analysis of the time and frequency domains of CoP data obtained from subjects on a strength platform has been used on several occasions to analyze healthy populations, as well as populations diagnosed with a pathology. Some studies on the equilibrium of individuals with DS performed using this method conclude that this population shows deficient motor control compared to individuals without DS. Adults with DS show significantly higher postural sway velocity than control subjects during a resting stance (Galli et al. 2007; Rigold et al 2011) and adopt different patterns of anticipatory postural adjustments (Aruin & Almeida 1997). Specifically they react using a generalized pattern of co-activation.

### 3.1 Co-contraction

The simultaneous activation of agonist and antagonist (co-contraction) muscles has also been described during quiet conditions (Gomes & Barela 2007), gait (Smith et al. 2007, Rigoldi et al. 2010) and balancing on seesaw (Carvalho & Almeida 2009). The seesaw has been used to study CNS response to external forces because it demands more from the control system and requires an essential change in mode of utilization of incoming proprioceptive information. According to (Ivanenko et al. 1997) healthy subjects primarily use proprioceptive cues for motion perception and postural control when they are supported by a stable surface. For these authors, vestibular information is used to determine the state of the support surface, and, if the support surface is unstable, vestibular information is used to aid balance control. In a previous study we observed that neurologically normal individuals adopted an alternated EMG pattern between agonist and antagonist bursts of ankle muscles and scaled their postural response with the increment of the seesaw's degree of instability (Almeida et al. 2006). Specifically they kept balance by alternating the activation of tibialis anterior and the gastrocnemius medialis muscles. The activation of the gastrocnemius started before the ankle moved from dorsal into plantar flexion and remained until the time the ankle shifted again into dorsal flexion. The activation of the tibialis anterior started before the ankle shifted into dorsal flexion, and remained active until the ankle shifted again into plantar flexion (Figure 1). On the other hand, the individual with DS kept balance by a continuous and simultaneous activation of TA and GM muscles, despite the direction of the ankle movement being into dorsal or plantar flexion (Figure 1). They were able to keep their balance on the seesaw without falling but they did so by using a pattern of muscle activity characterized by a co-activation of the agonist and antagonist muscles while control group did so by using an alternated muscles

pattern. Also, contrary to control group the individuals with DS were not able to graduate the displacement magnitude of ankle joint with seesaw instability. The question is: why did they adopt unusual strategies to keep their balance on the seesaw?

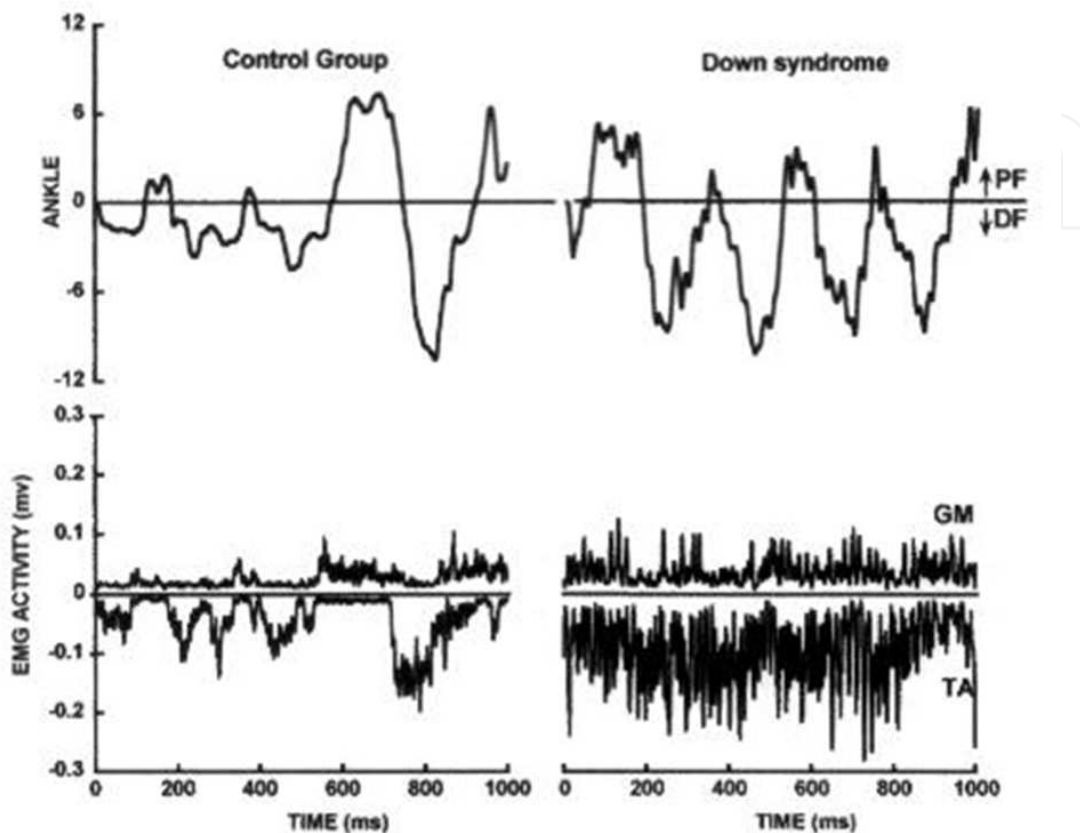


Fig. 1. Balance on a seesaw. Left panel are for control group subject and right for subject with DS. Positive values are for plantar flexion (PF) and gastrocnemius medialis (GM) activity and negative values for dorsal flexion (DF) and tibialis anterior activity.(Carvalho & Almeida 2009)

#### 4. Explanations of atypical postural behaviors

There has been debate in the literature over the real cause of atypical postural behaviors observed in individuals with DS. Possible explanations are related to cognitive limitations (Latash & Anson 1996), neurological disorder (Moldrich et al. 2007), abnormal sensorimotor integration (Vuillermé et al. 2001), compromised somatosensory system (Brandt 1995) biomechanical deficits (Cioni et al. 1994) such difference in bone density, hypoplasia of cartilage, ligaments properties changes. These changes may affect the ability to generate joint torque and strength in isokinetic contractions. For Shields & Dodd (2004) the muscle weakness can also influence the ability to perform daily tasks such as equilibrium.

##### 4.1 Neurological disorders

The brains of individuals with DS are smaller and lighter than those of normal individuals and exhibit a lower neuronal

density; they also show synaptic irregularities due to the reduction of neurotransmitters and anomalies in myelination processes.

Favour to idea of neurological disorders are the studies investigating cerebral development have indicated that although persons with Down syndrome depend on their right hemisphere for speech perception, their left hemisphere appears to play the executive role in speech production (Maraj et al., 2002). Of relevance to the motor behaviour domain, left hemisphere specialisation for speech production is associated with a general lateralised proficiency for specifying the magnitude and timing of muscular force. That is, persons with Down syndrome appear to perceive speech with their right cerebral hemisphere, but depend on their left cerebral hemisphere for the organisation and control of movement thus, exhibiting atypical patterns of brain organisation.

In the motor domain, relating to visual and verbal-motor development, persons with Down syndrome have demonstrated relative proficiency on skills involving the visual demonstration of movement (Maraj et al., 2002). Several studies have shown that adults with Down syndrome exhibit more errors performing single manual oral gestures to a verbal command (e.g., "place your finger on your nose") than following the visual demonstration of a task. Elliott, Gray and Weeks (1991) proposed that the functional isolation of the speech perception (right hemisphere) and movement production (left hemisphere) systems has led to a breakdown in communication between these systems, adversely affecting tasks that require verbal-motor behaviour. This proposal had been previously formalised into a model of cerebral specialisation.

Subsequent research based on this model has indicated that individuals with Down syndrome experience difficulties in performing motor tasks based on verbal instruction. The model has been used in accounting for the information processing difficulties on the basis of verbal instruction. Further, there is some evidence to suggest that persons with Down syndrome may consolidate visual information such that positive transfer is seen when they are switched from a visual to verbal mode of learning. Although much work has been done on simple upper limb movements, real progress toward influencing broader health and education practices demands that we assess gross motor skills. Gross motor skills are an important component of many physical activities. Moreover, the acquisition of these types of motor skills can facilitate many other activities of daily living.

#### **4.2 Adaptive choice**

Other explanations are adaptive choice used in unexpected situations to enhance security and stability (Latash & Anson 1996). For these authors, while the movements produced by those with DS appear clumsy, they can be viewed as adaptive reactions due to changed priorities within the central nervous system. The central nervous system is able to generate solutions to provide movement outcomes accept to itself such as a wrong co-contraction pattern of pre-programed response to increase the stability. On the other hand, wrong reciprocal contraction pattern would increase the effects of perturbation. Interesting fact is that with the extensive practice of simple movements, these individuals are able to adopt a tri-phasic pattern of contraction, favoring the idea that the co-contraction is a choice made by the nervous system in view of its flexibility and adaptability. Although it is a mechanically sub-optimal choice, co-contraction offers more security and reflects insecurity of postural system to generate universal postural reactions. On the other hand, the high levels of co-contraction described above does not match with the characterization of lower

tone and low ability to generate force of teenagers with SD. Exists therefore an inconsistency between the clinical evaluation of tone in this population and the abovementioned findings remain the relationship between hypotonia and co-contraction little understood. For Webber et al., (2004) and Vuillerme et al., (2001) evaluation of tone about passive conditions can provide limited information about the strategies used by SNC.

Among the explanations for the postural deficits found in individuals with DS, the favored explanatory hypothesis is that of a compromised sensorimotor system (Carvalho & Almeida 2009). Moreover, children with DS often exhibit significant perceptual problems. Auditory problems, often in association with ophthalmologic disorders such as cataracts, strabismus, nystagmus, visual and tactual impairments have been reported. However sometimes the apparent visual-perceptual problems in children with DS are actually due to deficits in the ability to physically perform the required task. Only a small proportion of children with DS were able to perform successfully on tactual and kinesthetic discrimination tasks (e.g., to discriminate among objects by texture, size, and weight while blindfolded), although the inclusion of visual input improved tactual performance in these children. In sum, children with DS show both motor and perceptual impairments that may influence the development and learning of various fundamental and complex actions. These influences have been widely reported over the years, but unfortunately not many findings have addressed the functional coupling of information and movement such as coupling of information and postural control.

#### **4.3 Sensory contribution to postural control**

One of the most widely used experimental approaches for understanding the sensory contribution to postural control is the manipulation of sensory information during postural disturbance. Galvanic vestibular stimulation (GVS) can induce postural reactions that are useful in determining the influence of vestibular function on balance (Fitzpatrick & Day, 2004). The vibration of muscle tendons is commonly used to determine the relative role of muscle proprioception in human posture control (Ruget, Blouin, Teasdale & Mouchnino, 2008). A number of studies have demonstrated that tendon vibrations, which almost selectively activate the primary endings of muscle spindles and elicits a discharge in the fast-conducting large-diameter Ia afferent fibers, can induce postural and orientation imbalance (Kavounoudias, Gilhodes & Roll, 1999). In order to better understand the sensory contribution to postural adjustments, we analyzed the effect of bipolar galvanic stimulation (GVS) and the vibration of Aquilles tendon on the pattern of muscle activity and joint displacements of individuals with DS.

Experiencing GVS, individuals with DS lacked the ability to maintain balance. The lack of balance under the effect of GVS cannot be explained by a change in muscle strategy as the pattern of co-activation was not changed by GVS. The DS individuals were more sensitive than control subjects to GVS (Carvalho & Almeida 2011). If somatosensory loss due to chronic neuropathy (Brandt 1995) or vibration (Carvalho & Almeida 2009) increases the reliance on vestibular information for control of postural orientation and individuals with DS also increases the reliance on vestibular information (shown by increased responses to GVS), we would suggest that individuals with DS have somatosensory deficits, and because of this, they were not able to compensate for a deficit of vestibular information with somatosensory feedback. Consistent with these findings are the results with vibration. The vibration was more detrimental to the balance performance of control group compared with

group with DS. One possible reason is that vibration disrupted the somatosensory information of control group but not in individuals with DS already disrupted by some deficit. It is possible that their proprioceptive deficit may prevent them from detecting the vibration effects. Previous studies showed that postural sway in subjects with somatosensory loss was significantly larger than normal on a firm surface but not on the sway-referenced surfaced, suggesting that sway-referencing disrupts somatosensory information for postural control already disrupted by neuropathy (Horak et al 2002).

Our findings support the hypothesis of somatosensory deficits defended by such authors as Cole, Abbs & Turner (1988) showing that individuals with DS failed to modulate the grip force when were asked to lift one object with different surfaces and Brandt & Rosen (1995) showing low amplitudes for sensory nerve action potential following stimulation of the thumbs suggesting impaired peripheral somatosensory functions. Other possible explanation is a delay in central processing the afferent and efferent information at the cerebellum level because the cerebellum weight has been reported to be lower (Bellugi et al., 1990).

Despite the importance of knowledge of sensory changes, biomechanical and neurobiology for understanding motor deficits, characteristics such as environmental context, experience and practice have great influence over these deficits. The positive effects of the practice have been demonstrated. Repeated room mobile exposure of babies (illusory) led to a more coherent and stable coupling between visual information and the body oscillation (Polastri e Barela, 2005). Reduced stiffness over the trials during maintenance of static posture signaled the ability of adults to vary its stiffness with practice (Webber et al., 2004). Similarly Smith et al., (2007) observed the reduction of muscle stiffness values in tweens with SD after treadmill training, although the kinematic patterns adopted before and training have deferred of the control group patterns. According Tudela et al., (2011) the intervention should be started up to the 3rd month so that the infant can have adequate stimulus in different postures. If stimulation is started earlier, it can be a way of minimizing long periods necessary to improve a skill in the motor development required by the infants with Down syndrome, and thus facilitate motor acquisitions, mainly antigravitational postures.

Overall, the motor control studies in individuals with DS indicate deficits in postural control mechanisms. The acquisition of this control is delayed and postural mechanism seems to be organized in order to maximize stability, adapting them to the slowness and poverty of the responses to environment changes. The functional consequence of this principle is the reduction of speed and coordination of movements that become clumsy. Although restricted in laboratory conditions to practice has influenced positively the postural control. We believe that this practice should be focus in the function and not in the correction of compensatory adjustments since the SNC can adopt numerous motor patterns to realize motor tasks successfully (normal variability).

## 5. Conclusions

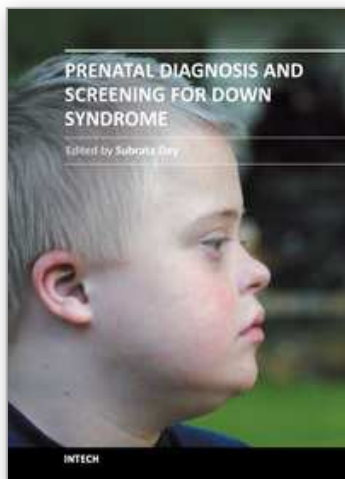
The Down syndrome is a multimodal disability affecting several systems therefore it is very difficult to pinpoint specific organic dysfunction for motor problems in these individuals. However, taking into consideration the lack of balance under the effect of GVS together with the fact that somatosensory loss increases the reliance on vestibular information, we could suggest that the balance difficulties observed in DS individuals during GVS can reflect deficits in the proprioceptive system.

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## **Prenatal Diagnosis and Screening for Down Syndrome**

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This book provides a concise yet comprehensive source of current information on Down syndrome. Research workers, scientists, medical graduates and paediatricians will find it an excellent source for reference and review. This book focuses on exciting areas of research on prenatal diagnosis - Down syndrome screening after assisted reproduction techniques, noninvasive techniques, genetic counselling and ethical issues. Whilst aimed primarily at research worker on Down syndrome, we hope that the appeal of this book will extend beyond the narrow confines of academic interest and be of interest to a wider audience, especially parents and relatives of Down syndrome patients.

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