

# Enhancing Walking Ability in Individuals with Rett Syndrome Through the Use of Applied Behavioral Analysis (ABA): Review and a Case Study

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**Abstract:** Rett syndrome (RTT) is a neurodevelopmental syndrome with arrest in brain development. The disorder is caused by a faulty gene on the X chromosome, mainly affecting females. The syndrome usually manifested itself through vast clinical impairments in functional abilities including motor disability. According to different studies only 50-75% of all affected females will achieve walking ability, and some will lose this ability later in life. The benefits of living a physically active life both for handicapped as well as for non-handicapped individuals have been vastly described in the literature. Since life expectancy of individuals with RTT is around 50 years of age, and due to the medical and functional difficulties accompanying this syndrome it is extremely important that the person with RTT will be engaged in an ongoing active life style from a young age. In addition to the achievements of gait there are a few case studies describing the re-gaining of lost walking ability in this population due to an intensive intervention. The present article explores the benefits of walking for this population and describes a case study, where applied behavioral analysis (ABA) techniques were used to extend her daily walking ability. The present case study is unique, as no existing literature supporting the use of ABA techniques for individuals with RTT was found. The findings of the present investigation support existing literature suggesting the importance of an intensive adapted intervention which can improve functional abilities in individuals with RTT.

**Keywords:** Applied Behavioral Analysis (ABA), developmental disability, enhanced walking, intellectual disability, Rett syndrome.

## INTRODUCTION

Rett syndrome (RTT) is a disturbance of cerebral neural development [1] mainly found in females. The syndrome is characterized by normal pregnancy and delivery, later to be followed by apparent normal development for the first 6-12 month of life. After this period the child begins to lose her previously acquired skills, and might show psychomotor dysfunctioning, varied functional communicational and medical symptoms [2].

RTT is due to a faulty gene (MECP-2) located on the X chromosome [3, 4] and the genetic diagnosis in RTT is corroborated by clinical diagnostic criteria. The progression

of the syndrome is described by four stages, e characterized by distinct features. The fourth stage is termed the "late motor deterioration" characterized by the loss of ambulation in individuals possessing the ability to walk during childhood [5]. Clinicians wonder if the fourth stage is indeed part of the syndrome or related to long duration of immobility, lack of the initiative and the sedentary life style that characterizes so many with RTT [6].

The symptomatology of RTT is varied, and intervention usually individually tailored [7]. Present comprehensive global research efforts are in place with intent to find a cure for RTT, which has so far shown impressive improvement of neurological impairment in girls with RTT [8]. These findings suggest a future possibility of obtaining a cure for RTT and indicate that medication might influence young girls and adult women alike. In light of these findings the staff taking care of individuals with RTT should implement intensive therapeutic management, which

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prevent regression and even promote functional abilities with emphasis on walking [9].

### **Why Walking?**

In general, it is important to prevent every person from living a sedentary life style [9]. Walking is a basic physical activity that enables both maintaining physical fitness and acquisition of an advanced functioning level. Walking has been found to prevent osteoporosis in people with disabilities [10], enables guarding of articular ranges of motion in the lower limbs and the back [11], enhances muscle strength in the lower limbs [12], improve functioning of the pulmonary system [12, 14] and improve the cardiovascular system's function [15-17]. Walking is also a social element, and the person that shows pedal ambulation is considered by his human environment as less disabled [18] than non ambulatory individuals. Therefore, it is important to encourage walking in people with disabilities in general, and in individuals with RTT in particular [19].

### **Mobility and Walking in RTT**

Walking is an ability that requires muscles strength, sensory organization and coordination of trunk and limbs in an erect position, while moving. In spite of difficulties in most of these areas, most individuals with RTT achieve pedal ambulation. Yet, some do not learn to walk, while others gain walking ability and lose it later on in life. In most children with RTT motor developmental is delayed and walking is aided, yet as the child grows heavier and older so does her caregivers. Therefore, in adults with RTT walking has an even greater significance, as losing walking abilities makes the person harder to handle. The more the adult woman with RTT is dependent upon external aid for walking, or is dependent upon heavy accessories, her life environments would be restricted and the person with RTT, so keen on social contacts, will be restrained within her growing handicap.

Motor problems and/or sensory problems which are typical for RTT and might influence walking ability are: low muscle tone (hypotonia), which might change in older age to high muscle tone (spasticity), or to the stiff self-induced high muscle tone (rigidity). Additional problems typical to RTT include: Ataxia (impaired muscular coordination), apraxia (difficulty in planning and execution of motor tasks), impairments in sensory spatial body orientation and orthopedic problems such as: Scoliosis, kyphosis, and foot deformities [7].

Loss of the independent walking and transfer abilities are the first abilities lost by individuals with RTT [20, 21] thereby leading to severe functional lost.

All individuals with disabilities gain from being able to walk yet this ability is even more important when relating to individuals with RTT [22]. One example for the advantages of walking for individuals with RTT is the adverse correlation found between mobility in general, and ascending and descending stairs specifically, and the prognosis of scoliosis.

In this group of clients scoliosis appears in 80-100% [5, 22, 23], and therefore general physical activity is warranted with emphasis on stairs descending and ascending. Another

reason for maintaining walking ability in individuals with RTT is the fact that most with this disorder suffer from constipation. Since physical activity was found to improve blood flow to internal organs it could be found beneficial in relieving constipation [24].

Moreover, the autonomous system in RTT was found impaired, including the reduction of blood flow to the limbs [25]. Since walking enhances blood flow to the lower limbs, walking is a recommended activity for individuals with RTT.

Osteoporosis, another known aspect of RTT [26, 27] has been improved in individuals with RTT showing the ability to walk as compared to non-ambulant individuals [10].

Due to the multiple challenges faced by the person with RTT and her family it is recommended that a multi disciplinary intervention be applied to meet the vast symptomatology presented by persons diagnosed with this disorder [28]. A joint collaboration by therapists from various disciplines, where each brings knowledge and acquired clinical experience, can advance the client with RTT to achieve her best abilities [29, 30]. It might be that the severity of the phenotypic expression can not be changed, but the quality of life could be improved if the therapeutic intervention is intense enough and individually adapted to the needs of each client. As suggested by a daily walking program implemented for the duration of two months with five individuals with RTT resulting in improvements in functional as well as cardio-vascular outcomes [31].

### **APPLIED BEHAVIOR ANALYSIS (ABA)**

Applied behavior analysis (ABA), is an application to modify human behaviors, especially as part of a learning or treatment process [32]. The method is applied by a person qualified in behavior analysis (The second author; E.S.) who assesses the observable relationship between a targeted behavior of a specific person to his physical and human environments. Research in the field of ABA has supported the effect of this method [33-35]. The main characteristics of this method are 1) the application of operant learning principles, 2) the precise measurement of observable behavior, and 3) the use of experimental analysis methodologies to identify behavior-environment relations. Behavioral analysis of a specific problem is designed to determine what factors support or maintain the observed behaviors. The identification of these variables is possible due to careful interview with peers or caregivers and observation of the individual in his natural environment [36].

### **THE STORY OF MS**

MS was born a full term baby in a normal delivery, after an uneventful pregnancy. During her first post gestation year she presented a normal yet somewhat slow development of motor milestones. During her second year her development arrested and stereotypical movements appeared. MS began pedal ambulation at the age of 18 month. She was diagnosed with RTT at the age of two years and placed in a residential care center at the age of 15 years due to familial circumstances. She was able to ambulate independently, yet actively chooses to stay inactive by hiding in other clients' beds, so she cannot be detected, during activity hours.

At the beginning of the intervention MS was residing in a residential setting at the center of Israel, her morning hours spent in a day care center with little to no activity and spend her time in a chair or sitting on a beanbag. She was showing normal tone with spinal deformity suggesting extreme (untreated) C-shaped left scoliosis. It was decided to initiate an intervention program, which included three steps:

- An ABA pre-intervention evaluation
- A pre-intervention baseline evaluation
- The intervention itself with an ongoing outcome measure assessment.

The pre-intervention evaluation included an interview with caregivers familiar with MS and an observation of her behavior. The findings suggest that MS spends most of her time sitting on a sofa (a tangible negative reinforcement). Her daily walking experiences mostly included walking to the dining room for breakfast, from the dining room to the car driving her to the day care center, from the car to the room within the day care center, where she spends her day sitting, from the day care center to the car driving her back to the residential setting, from the car to the residential setting, and later during evening times from the sofa, where she spends the afternoon sitting to the dining room for dinner. The overall daily steps count was collected over a one month period and averaged at 800 steps per day.

MS was able to sit down on a chair or on a sofa at will, as well as to get up from a sitting position. She was found to get up from the sofa and independently move to sit by other residents, who would play with her hair. MS understands simple instructions and responds favorably to verbal praise. The support providers were found to give MS higher levels of assistance than she really needed.

The pre-intervention evaluation found the following goals for intervention: to enhance number of steps taken by MS daily, to use verbal praise and tactile stimuli for reinforcement and outcome measures were defined as number of steps taken. The results of the pre-intervention evaluation were presented to M.S's caregivers and they were asked to comply with the program.

The evaluation focused on eliminating negative reinforces such as preventing the availability of chairs and bin bags in her vicinity, and enhancing positive reinforces when MS was ambulating, which she was found to like such as verbal praise and gentle hair stroking.

The results of the intervention program were measured through the use of accelerometers, as these appliances have been found to be a valid and reliable objective assessment of physical activity [37] and as means to document the frequency and distribution of physical activity of a person or within a population [38].

Within three month of the initiation of the program a learning curve developed showing a constant and steady enhancement of number of daily steps from an average of 800 to more than ten times of the initial value (see Fig. 1).

The comparison of average daily steps taken by MS pre-post intervention present a significant change ( $P < 0.00$ ) (see Fig. 2).

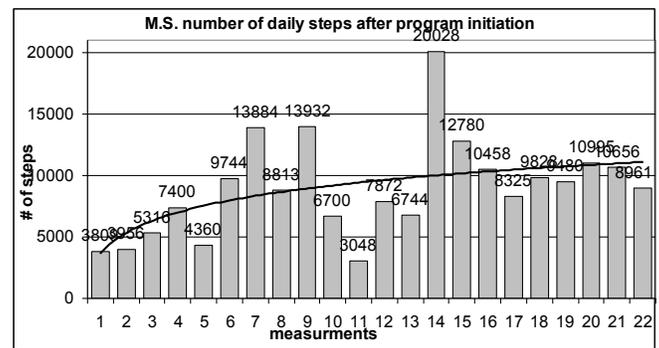


Fig. (1). Number of daily steps taken after program initiation.

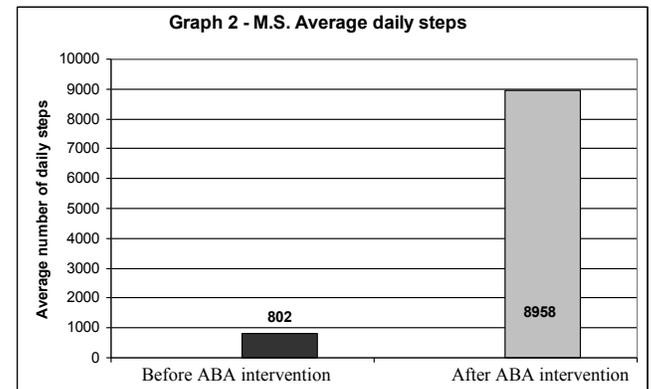


Fig. (2). Average daily steps taken.

**DISCUSSION**

We have described a case study of a woman with Rett syndrome (RTT) enhancing her daily walking capacity due to an ABA (Applied behavior analysis) program. The change from 800 daily steps to over 8,000 steps per day accrued slowly within a period of three month. At the end of the program MS was walking around the residential care center almost constantly looking for staff members, who would give her verbal praise and gentle hair stroking. The results suggested that an intensive program comprising of positive reinforcements was a good motivation for MS to become a more active person with all the health benefits associated with increased activity.

It is obvious now that the reduction in her ambulation abilities in her late teens was caused by changes in her surrounding with reduced demand to stay an active person as well as the tendency of individuals with RTT to refrain from physical effort.

Due to known negative consequences of passivity and immobility [9] and the positive influence of activity programs on morbidity [10-18] of individuals with disabilities it is essential that individuals with RTT will be kept active as long as possible throughout their life. This case study, as well as other findings suggesting improvements in functional abilities in individuals with RTT, when an intense program is implemented [31, 39, 40], should encourage clinicians working with this population to invest their professional skills in preventing this group of clients from losing their pedal ambulation and implement further attempts at enhancing previous functional abilities. It is emphasized that RTT is defined today as a "developmental

disorder" (in contrast to its previous definition as a "degenerative disorder") and therefore, the future of the person with RTT is not pre-determined. Furthermore, since RTT was found almost completely reversible in genetically engineered RTT mice [8], it is possible that such a change will be achieved in humans with RTT in the future. It is therefore our moral duty as therapists and health related professions, to invest our efforts in maintaining the person with RTT at her full functional potential, ready to gain recovery at a time when gene therapy will be able to reverse RTT.

## CONCLUSION

Walking is a primary health promoter for all people but especially for those with motion and postural difficulties such as individuals with RTT. Walking is achieved in many with RTT and might be lost at a later age in this group of clients.

The present article presented a case study of an adult woman with RTT where enhancement of her walking ability was improved 10 folds, through the use of Applied Behavior Analysis. This case study shows that with proper intervention some of the disabling features of this syndrome are reversible and are prone to rehabilitation.

## CONFLICT OF INTEREST

The authors confirm that this article content has no conflict of interest

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