

Physical therapy for Rett syndrome

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The challenge

Rett syndrome (RTT) causes a neurological and developmental arrest that manifests itself in a variety of difficulties, such as loss of functional hand use, loss of acquired speech, apraxia, ataxia, autonomic system dysfunction, epilepsy, breathing abnormalities, failure to thrive and muscle tone irregularities [1-3]. In order for the children and adults with RTT and their families to live life to the fullest, proper intervention should be applied.

The present chapter addresses some of the main physical difficulties typically exhibited by individuals with RTT and suggests possible intervention techniques according to the scarcely existing literature and mainly based on the experience of the author for the past 25 years, by evaluating over 500 individuals with RTT across the globe, as part of the Israeli RTT national evaluation team, and also as a therapist managing about 13 individuals with RTT on a weekly basis.

Due to the longevity of individuals with Rett syndrome (RTT) [4] and the complex nature of the RTT disability, physical therapy is an important part of the management of the disorder. Individuals with RTT show a considerable functional diversity. Some young people might not achieve independent sitting or standing, while others may gain high functional abilities such as running, skiing and trampoline jumping [5]. Due to such versatility, a thorough evaluation should be performed of each child entering an intervention program. After such an evaluation, preferably by a multi-disciplinary team [6,7], an intervention program should be drawn up, specifically tailored for each client.

In many cases, the child with RTT is treated by a team of therapists, each of the disciplines involved in the therapeutic program uses a combination of different techniques intended to maintain and maximize function of the individual with RTT. While these therapies do not cure RTT, they can help the person with RTT by lessening the challenges experienced by her, thus helping her and her family cope with some of the functional limitations typical of RTT [8].

Team work

It is vital for the success of the intervention program that different members of the team coordinate their therapeutic efforts into a combined management approach, in accordance with all team members' agreement (including that of the parents and the child with RTT).

A well-planned intervention is of great importance to individuals with RTT [9, 10]. Such a program may maintain or improve function, prevent deformities and provide positioning and mobility [10], thereby contributing to the social accessibility of the client.

Goals

The basic goal of the Physical Therapy intervention program is to enhance the quality of life and functional abilities of the client with RTT. These goals might be achieved if the intervention is aimed at:

- Managing muscle tone and preventing/reducing abnormal movement patterns
- Maintaining articular range of motion and prevent contractures from a young age
- Reducing Dyspraxia by repetitive functional experiences
- Increasing and improving cardio-vascular fitness
- Stimulating hand use
- Achieving dynamic postural control
- Promoting better co-ordination and balance
- Preventing progression of scoliosis from a young age

The importance of early intervention

Early intervention in RTT is intended to facilitate acquisition of developmental skills, to inhibit or minimize the long-term effects of specific risk factors on development [11], and reduce the “setting in” of pathological movement patterns which become difficult or impossible to change.

Supplementary program

The term “supplementary program” implies a 'round the clock' care for the individual, intensity of appropriate handling, continuous positioning and suitable assistive devices. A supplementary management program is both in addition to, and supportive of the client's therapeutic regime by modifying her activities so that each daily task can be used to reinforce the improvement of learned skills within meaningful contexts [12].

Scoliosis

The most common orthopaedic problem seen in RTT involves the spine. The phenomenon can be found in 80%-85% of adults with RTT [1,13]. Due to its frequency, all young people with RTT should be checked for spinal position, asymmetry and curvature during their yearly physical examination.

Another problem involving scoliosis in RTT is its rapid progression, averaging at 14-degree annual rate [1,13]. Moreover, due to the high incidence of scoliosis it is recommended that each child with RTT be involved in a scoliosis prevention program from a young age.

The prognosis of scoliosis development in RTT is better when scoliosis is observed at an older age, and when walking and stair climbing can be maintained by the child [1]. The complexity of actively and dynamically controlling the human movement is due to proper sensory input, proper evaluation of that sensory information, and the large number of musculoskeletal elements involved in every movement [14].

Scoliosis and kyphosis in RTT begin with lack of such coordinated ability, leading the child with Rett to compensate with individualized fixations of hands (stereotypical movements) and body rigid fixations which eventually lead to the development of spinal deformities in RTT.

Active exercise and passive range of movement routines are helpful. Maintaining spinal alignment is important and can be facilitated by activities designed to provide proprioceptive (alignment of the body), kinaesthetic (sensations received from joints and muscles) input, thereby enhancing the individual's awareness [15].

Inserting dynamic core control techniques from an early age are strongly suggested. The common intervention approach for treating scoliosis is usually a combination of the following techniques:

- Intensive physical and hydro-therapy treatments yield maximal benefits [13, 16].
- Intensive preventive measures before the appearance of the first spinal asymmetry.
- Intensive treatment starting as soon as the first spinal asymmetry is noticed [16].
- Intensive walking (or standing if the individual is non-mobile) [16].
- “Over-correction” treatment, suggested by Hanks [13,18], is applied to re-adjust the skewed mid-line perception of females manifesting scoliosis dominated by their skewed sensory system. There are reports of 4-5 cases where the progression of scoliosis was arrested using this method [19].
- Active anti-scoliosis regime – this intervention has managed to reverse the scoliosis from 30 to 20 degrees in a child with RTT. [18].
- Dynamic core activities adapted to each child’s level.
- Follow ups - visits to an orthopaedic surgeon (at least twice a year; more if the child is showing initial signs of scoliosis and is younger than five years of age) [2, 20]

Walking

Ambulation, or walking, is a skill which requires a complex level of coordination. 50%-85% of all children with RTT achieve the ability to walk [1]; some lose this ability later in life. Walking is extremely important in RTT as it prevents osteoporosis, strengthens lower extremity muscles and activates the respiratory-vascular system.

It is good to encourage walking in those who can do so [16], but it is sometimes impossible to teach a child to walk if the required co-ordination is not present [21]. On the other hand, some individuals with RTT have gained walking ability at the age of six, sixteen, and even 21. And there are reports of individuals with RTT who have lost walking ability, yet regained it, after being wheelchair bound for 5 [22], 12[10] and even 20 years [23] and our findings suggest that 80% of individuals with RTT presenting independent walking may lose and regain this ability twice on average.

Due to the importance of ambulation on health and quality of life issues, it is believed that the pursuit of walking should be constantly maintained for persons with RTT including individuals with RTT who were able to walk and lost their ability to ambulate [2].

Generally, it is a good idea to establish routine daily walks as a preventative measure against inactivity and a sedentary life-style. Correlation between ambulation ability (specifically stair-climbing) and milder cases of scoliosis was found [24]. Walking in RTT was found to correlate with improved physical fitness [17]

Summation

Rett syndrome is a developmental disability presenting a complex clinical picture. The versatility of those diagnosed with RTT requires intensive, individual, and knowledgeable care from childhood, including acquisition of postural control, prevention of spinal asymmetries, and maintenance of functional and physical fitness.

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