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*Case Report*

# A Goal Oriented Physical Therapy in Rett Syndrome: A Single Case Study

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## ABSTRACT

**Introduction:** Rett Syndrome is a serious and global developmental disorder that affects the central nervous system. The course of this syndrome is in 4 stages. Till date, there is no cure for Rett Syndrome, so rather than addressing the syndrome as a whole, most treatments try to alleviate specific symptoms.
**Materials and Methods:** For the evaluation of tightness, popliteal angle and adductor angle was measured. For gross motor developmental milestones, Gross Motor Function Measure (GMFM)-88 was used. After the evaluation, goal oriented Physical Therapy was given for 45 minutes per day for 5 days per week for 6 weeks. After 6 weeks, post-intervention assessment was done using same measures.
**Results:** After intervention, both popliteal angle on right & left side and adductor angle was increased. The improvement was seen in scores of Gross Motor Function Measure (GMFM)-88 Dimension B, C, D and E those are Sitting, Crawling, Kneeling, Standing, Walking, Running and Jumping Component. Also the total score of Gross Motor Function Measure (GMFM)-88 was improved.
**Conclusion:** Physical Therapy helps the children with Rett Syndrome in reducing the tightness and achievement in gross motor developmental milestones.

***Key words:*** Rett Syndrome, neurodevelopmental disorder, Physical Therapy.

## INTRODUCTION

Rett Syndrome is a serious and global developmental disorder affecting the central nervous system. [1] In 1960's Andreas Rett, described Rett Syndrome as an x-linked neurodevelopmental condition characterized by loss of spoken language and hand use with the development of distinctive hand stereotypies. Later in 1999, Amir et al discovered that mutation in gene encoding Methyl-CpG-binding protein 2 ($MECP_2$) is associated with both rare familial cases as well as with more common sporadic occurrence of typical Rett Syndrome. [2]

Rett Syndrome can be Typical (Classical) or Atypical (Variant). Some individuals present with many of the clinical features of Rett Syndrome, such as regression, but do not necessarily have all of the features of the disorder and are called as Atypical Rett Syndrome. [2] Prevalence for Typical Rett Syndrome is 1 in 10,000 to 22,000 females[1] and possibly 1 in 100 000 males [3] and for Atypical Rett Syndrome is 1 in 45,000.

The course of Rett Syndrome includes Stage One, Early-Onset (six-18 months of age). The early symptoms are not always noticeable in Stage 1. The infant may not make eye contact with family

members and may not show much interest in toys, delays in gross motor skills such as sitting or crawling, noticeable hand-wringing and slowing of head growth. This stage usually lasts for a few months but can persist for more than a year.

Stage Two, Rapid Deterioration (one–four years). This stage may be either rapid or gradual in onset. Child loses her ability to speak and to make purposeful hand movements, hand-to-mouth movements may appear, as well as hand-wringing or hand-clapping gestures. These movements may be nearly constant while the child is awake but disappear during sleep. Episodes of breath holding and hyperventilating. The child may have trouble sleeping and may become irritable. If the child is able to walk, she will start to look unsteady on her feet and may have periods of trembling or shaking. Slowed growth of the head is usually most noticeable during this stage.

Stage Three, Plateau (two-10 years). Motor problems (i.e. Apraxia) and seizures often appear during this stage. The child's behavior, however, often shows some improvement, with less irritability and crying. She may show greater interest in her surroundings, her attention span and communication skills often improve. Many patients with RS remain in stage 3 for most of their lives.

Stage Four, Late Deterioration of Motor Skills (usually after 10 years of age). Patients gradually lose their mobility; some stop walking while others have never learned to walk. There is, however, no loss of cognitive or communication skills. The repetitive hand movements may decrease, the spine begins to develop an abnormal sideways curvature (scoliosis), the patient may develop muscle rigidity, puberty begins at the same age as in most girls. [4]

Molecular Genetic Test helps in the diagnosis of Rett Syndrome. There are various neuroimaging studies used for diagnosis of Rett Syndrome, which are Diffusion Tensor Imaging Studies, Magnetic Resonance Spectroscopic Imaging, and Cerebral Blood Flow Measurement with MRI. [5] Clinical diagnosis is made by DSM IV – Diagnosis of Rett's Disorder; 299.80; Rett's Disorder [4] and Rett Diagnostic Criteria 2010 by J L Neul et al. [2]

Till date, there is no cure for Rett Syndrome. But there are a variety of ways to minimize its effects. Rather than addressing the syndrome as a whole, most treatments try to alleviate specific symptoms. [1] Current recommendations regarding on-going treatment for individuals with Rett Syndrome include aggressive physical, occupational and speech therapy. Appropriate interventions required for nutrition and optimal growth, orthopedic concerns, proper assessment and management of epilepsy and GI dysfunction, and other issues as they become relevant. [6]

Physical therapy intervention focus on normalization muscle tone, improvement in range of motion, maintain and improve mobility, develop and maintain transitional skills, improve body awareness, stimulating hand use, increase and improve cardiovascular fitness, reduce apraxia and ataxia, promote balance, reverse progression of scoliosis. [7] The current case report is an effort not only to define and describe the developmental and behavioral features of Rett Syndrome, but to discuss the changes that occurs secondary to physical therapy intervention in Rett Syndrome.

## CASE DESCRIPTION

A 9-year old girl brought to the Physiotherapy Outpatient Department by her mother, with the complaint of gradual loss of speech, social interaction, hand skills and developmental milestones with stereotyped movements of the hand and body since last 6 years.

**Birth and developmental history:** The child's mother gave history of non-consanguineous marriage, an uncomplicated pregnancy with full term normal delivery at home. Height and weight at birth were normal.

**Milestones:** All the developmental milestones, that are gross motor, fine motor, social, emotional, speech and language were achieved normally till first three years of life.

**History of presenting illness:** After three years of normal development, she started showing signs of developmental regression. By this age of 9 years, she was not able to crawl and walk, but able to stand for 5 minutes when passively made stand, where as she used to run previously. She lost her acquired and purposeful hand skills and was unable to hold, pick up and grasp things in her hands. She was showing stereotyped hand movements and clapping. Teeth grinding was also reported. She was not able to speak any meaningful words, where as she used to narrate small sentences earlier. Her mother noticed her lack of attachment with family members and would not interact or make eye contact with them, even when family member would try to do so. She was unable to indicate her need for daily activities such as toilet, passing stools or for food.

**Family history:** There was no significant family history present.

**Examination:** On Physical Therapy evaluation, child showed delayed gross motor developmental and tightness on bilateral hamstring muscles and adductor muscle. For assessment of gross motor development, Gross Motor Function Measure (GMFM)-88 was used. For tightness of hamstring and adductor muscle, popliteal angle and adductor angle measurement respectively was used.

**Pre Therapy Score:** On Gross Motor Function Measure (GMFM)-88 she scored 30.44%. She was having popliteal angle 100° on right and 105° on left. Adductor angle was 60°.

**Consent:** After examination, the therapist explained the findings and the importance of daily Physiotherapy exercises to the mother and took her consent in written.

**Management:**

For tightness, initially hot moist pack was given for hamstring and adductor muscles bilaterally, followed by passive stretching of both the muscles.

For developmental gross motor milestones, mat activities were given.

For the first two weeks, active rolling from supine to prone. From supine position, supine to sit facilitation. In prone position maintaining weight on bilateral forearms followed by weight bearing on palm was initiated.

Then on third and fourth weeks when she was maintaining the weight bearing on palm, quadruped positioning started, by passively flexing both the knees and coming to quadruped position and maintaining that position. In quadruped position, passively doing flexion and extension of hip alternatively and simultaneously moving both the hands forward for crawling facilitation. Then from quadruped position coming to kneeling position passively and maintain that position. From kneeling to kneel standing to half kneeling position then started.

Subsequently on fifth and sixth weeks, once she started maintaining the half kneeling position, half kneeling to standing was facilitated by child holding therapist in front and coming to standing position (pull to stand). From sitting on the chair, sit to stand while therapist holds hands from front. Followed by standing in standing frame. Walking facilitation was done by mother holding both hands from front and therapist doing alternate hip flexion and extension.

The same protocol was repeated for 45 minutes per day for 5 days per week for the duration of 6 weeks. After 6 weeks, a complete reassessment was done.

**Post Therapy Scores:** Score of Gross Motor Function Measure (GMFM)-88 was 41.66%. Popliteal angle was 110° on right and 117° on left. Adductor angle was 68°.

**Table 1.1: Pre-Intervention and Post-Intervention measurement of Tightness**

| | Popliteal Angle | | Adductor Angle |
|---|---|---|---|
| | Right | Left | |
| Pre-Intervention | 100° | 105° | 60° |
| Post-Intervention | 110° | 117° | 68° |

**Table 1.2: Pre-Intervention and Post-Intervention scores of Gross Motor Function Measure (GMFM)-88**

| | Dimension A (Lying & Rolling) | Dimension B (Sitting) | Dimension C (Crawling & Kneeling) | Dimension D (Standing) | Dimension E (Walking, Running & Jumping) | Total Score |
|---|---|---|---|---|---|---|
| Pre-Intervention | 64.70% | 48.33% | 23.80% | 15.38% | 0% | 30.44% |
| Post-Intervention | 64.70% | 61.66% | 45.23% | 25.64% | 11.11% | 41.66% |

## DISCUSSION

The purpose of the study was to describe the results of Physical Therapy Rehabilitation Program for a child with Rett Syndrome. The therapy was aimed primarily at reducing the muscle tightness and achievement of gross motor developmental milestones. After the intervention, the tightness was reduced on both the hamstring muscle and hip adductor muscles as shown by change in measurement of both popliteal and adductor angles.

Improvement was seen in the Gross Motor Function Measure (GMFM)-88 scores also. She showed improvement in Dimensions B, C, D and E those are Sitting, Crawling, Kneeling, Standing, Walking, Running and Jumping Component. In sitting component, she started initiating rolling in supine to right & left, attaining sitting and sitting on mat and lowering to prone with control. In Crawling and Kneeling component, she started crawling reciprocally forward 1.8 meters and half kneeling using arms and maintaining arms free for 10 seconds. In standing component, she started coming to standing from large bench and from half kneeling position. In Walking, Running and jumping component, She started walking 10 steps forward initially with two hand support and later by one hand support.

Sayan Chattopadhyay et al said that, according to the International Rett Syndrome Association, Physical Therapy should be one of the priorities in patients with Rett Syndrome. When patient is in stage 1, we should focus on independent sitting, standing and walking; stage 2, focus on range of motion and ambulation,; stage 3, focus should emphasize on sitting and stage 4, requires focus on all the aspects from previous stages. [8]

Meir Lotan et al said that, the individual with Rett Syndrome present with vast array of orthopedic and neurological difficulties. To cope with such difficulties and overcoming the associated limitations, an informed and intensely applied Physical Therapy regime can help the child and family both. [7]

Therefore, the present article was written with the intention to introduce Rett Syndrome to the Physical Therapist, familiarize them with this population, and to offer some of the authors' clinical experience in applying physical therapy intervention programs for these individuals.

## CONCLUSION

From this case study, we concluded that, Physical Therapy helps the children with Rett Syndrome in improvement of tightness and achievement in gross motor developmental milestones.

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