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CARDIO RESPIRATORY PHYSIOTHERAPY MANAGEMENT IN RETT'S SYNDROME-LITERATURE REVIEW

S.HARISH KUMAR[*1], S. KIRUTHIKA[1], R. ARUNACHALAM[1], A. KUMARESAN[1]

[1.] Department of Physiotherapy, Saveetha University, Chennai, India

ABSTRACT

Rett syndrome was identified by Dr.Andreas Rett, an Austrian physician in 1966. It is a neurological disease characterized by arrest of brain development caused by X chromosome mutation that commonly affect girls. The condition is a combination of orthopedics, neurological and cardio respiratory symptoms and most of the people die because respiratory dysfunction. Though medications are used for breathing irregularities, heart abnormalities and seizures, Physiotherapy interventions play an important role in prolonging mobility, preventing cardio respiratory complications and structural abnormalities there by increasing the life span of Rett syndrome survivors. This study intends to review the literature that emphasizes the importance of cardio respiratory physiotherapy for Rett syndrome. Results suggest physiotherapy plays an important role in Rett syndrome especially in cardio respiratory condition. Treadmill and Hydrotherapy were found to be effective for cardio respiratory problem in Rett syndrome.

KEY WORDS: *Rett syndrome, Cardio respiratory physiotherapy management, Hydrotherapy, Treadmill*

S.HARISH KUMAR*
Department of Physiotherapy, Saveetha University, Chennai, India

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1.INTRODUCTION

Rett syndrome, also called as cerebroatrophic hyperammonemia is a rare genetic neurodevelopment disorder with brainstem immaturity. Mostly the grey matter of the brain is affected. Rett syndrome was initially classified under pervasive developmental disorder by the diagnostic and statistical manual of mental disorders, along with the autism spectrum disorders and childhood disintegrative disorder. But later this was classified to non-autistic spectrum disorder with autistic features. The disorder was initially found by Dr.Andreas Rett, an Australian Physician in 1966. Dr.Bengt Hagberg in 1983 explained in detail about the causes, clinical features and thus this disorder became recognizable by the population. In 1999 Huda Zoghbi reported that Rett syndrome is caused by mutations in the gene MECP2(1) [1]. Armstrong reported that there are reduced dendritic territories in the frontal, parietal, and inferior temporal areas, a small area in midbrain, and a large increase in serotonin receptor density in the brain stem, medullary nuclei known to have autonomic and cardio respiratory functions where it is specifically affected. Rett syndrome affects 1 in every 10,000 to 15,000 live female births and in all racial and ethnic groups worldwide.; The risk of a family having a second child with this disorder is less than 1 percent [1.] Females may live upto 40 years.1 per 22800 (0.44/10000) females aged 2 through 18 years of age was reported to have Rett syndrome by Texas Rett syndrome registry[2] . Incidence was 0.96 per 10,000 females to age 12 years in Australian population[1] . The first case in India was reported only in 1994. And few cases were only reported [3].The children's are often misdiagnosed as autism, cerebral palsy or idiopathic mental retardation especially during the early stages. There are 4 stages in Rett syndrome(3). (4)(4)(4)(4)(4)(4)(4)(4)(4)(4)(4)(4)(4)(4)(4)(4)(4)(4)(4)(4)(4)(4)(4)(4)(4)(4)(4)

Stage1: begins between 6- 18 months of age. Symptoms are usually vague. Common features include reduced interest in toys, less eye contact, delay in gross motor skills like sitting, hand wringing and decreased head growth.
Stage 2: begins by 4 weeks to years. Purposeful hand skills, language is lost. Breathing irregularities starts at this phase characterized by episodes of apnea and hyperventilation. These abnormalities are absent during sleep. This breathing dysrhythmia is a striking feature.
Stage 3: begins between 2-10 years. Apraxia and seizures are more common
Stage 4: reduced mobility, scoliosis, rigidity, abnormal posturing of arm leg, walking will be impaired, Heart abnormalities and respiratory problems.

There is no cure for this syndrome often multidisciplinary approach is needed. It consists of Physician, Occupational therapist, Physiotherapist. Most of Retts syndrome patients die because of cardio respiratory problems. Though medications are used for breathing irregularities, heart abnormalities and seizures, physiotherapist interventions play an important role in prolonging mobility, preventing cardio respiratory complications and structural abnormalities thereby increasing the life span of Rett syndrome survivors. Hence this study intends to review the literature that emphasizes the importance of Cardio Respiratory Physiotherapy for Rett syndrome.

2.REVIEW OF LITERATURE

The objective is to review the Cardio-Respiratory signs and symptoms in children's with Rett's syndrome followed by Cardiorespiratory Physiotherapy rehabilitative measures. Some of the inclusion Criteria were selected by common symptoms in Rett's syndrome.. Interventions or exercises that analyzed by the importance of Cardio Respiratory rehabilitation were selected.

2.1CARDIO RESPIRATORY SYMPTOMS

Hagebeuk EE, 2012 studied respiratory disturbances in Rett's syndrome where patients are evaluated with respiratory disturbance overnight and symptoms were clinically relevant apnea in 10 subjects. In 8 children central apneas were present during the day often with obstructive apneas at night. In 3 children severe respiratory problem, with frequent oxygen desaturations were also found.[3] Acampa & Guideri, 2006 summarized the anatomical findings, ECG, cardiac autonomic nervous system findings. This Study also explained about the pathogenesis and their mechanism for sudden death. There are no cardiomyopathies or valve alterations but prolongation of corrected QT interval and hyperventilation were the common features.[5] Julu PO1, Kerr AM, 2001 studied breathing and associated central autonomic dysfunction in the Rett's disorder. 56 female patients with Rett's disorder aged 2-35 years were recorded for breathing movement, BP, ECG R-R interval, HR, transcutaneous blood gases, cardiac vagal tone, and cardiac sensitivity to baroreflex measured on-line with synchronous EEG and video. Respiratory rhythm was normal during sleep and abnormal in the waking, forced and apneustic breathing were prominent. Resting cardiac vagal tone and cardiac sensitivity to baroreflex were reduced.(6)[8] Ellawaya & Shollerb, 1999 did study with 34 girls with Rett's syndrome, a cohort study. Electrocardiography and 24 hour Holter monitoring were the outcomes. Compared to healthy subjects the RS subjects showed longer corrected QT values.[7] Southall DP1, Kerr AM, Tirosh E, 1988 assessed respiratory function in 18 RS and 23 healthy people. These subjects were assessed for ventilation, apnoea, and arterialpH. 56% subjects had hyperventilation when awake. Accompanied with prolonged periods of apnoea, hypocapnia and pH was 7.47-7.60.[8] Pertile N , Vergerio A , Galliani E , Turrin A , Caddia V, analyzed the SAO2 and pH during sleep and wakefulness in a girl with RS. Hyperventilation- SAO2 98%, alkalosis and apnea. These patterns seen during wakeful period.[9]

From these review it is clear that the subjects often complaints of signs and symptoms of Cardio-Respiratory abnormalities. Common symptoms found

were hyperventilation, altered ECG pattern, Prolonged periods of apnea.

2.2CARDIO RESPIRATORY REHABILITATION

Lotan, Shavit, & Merrick, 2015 did study on 15 year old subject. Her history was that she spends most time in sitting with less walking experience. Applied behavioral analysis intervention was given for 3 months and the outcome was number of steps measured using accelerometers. The results showed improvement from 800 to 8000 steps following intervention. And the study concluded walking is a basic physical activity that enables both maintaining physical fitness and acquisition of an advanced functioning level.[10] Harris, summarized the development, gait abnormalities, complications, assessment and common treatment strategies used for Rett's syndrome. Maintaining fitness is more important. Fitness can be improved by treadmill and activities like walking, stair climbing, swimming and riding. Hydrotherapy is also used for both fitness maintenance and enhancing gait.[11] Meir Lotan, 2004 did a study that investigated the feasibility of treadmill training on fitness and physical activity among Rett's syndrome. 4 females were given training on a 1400 model treadmill (Trim line, capable of very low speeds < 0.5 k/h), for 2 months. The outcome measures were pulse and activity levels. Tests showed that general functional abilities had improved considerably. Pearson correlation showed high linkage between functional improvement and change in physical fitness following treadmill training.[12] Bumin, Uyanık, & Yılmaz, 2003 did study on the hydrotherapy. Treatment was given in swimming pool twice a week for 8 weeks. The girl's physical abilities were assessed 3 times: before and 5 minutes after a single hydrotherapy session and after 8weeks of hydrotherapy. After hydrotherapy stereotypical hand movements decreased and purposeful hand functions and feeding skills increased.[13] Reeve, did study on Hippoteraphy for the Rett's Syndrome , 15 year old female child was given horse therapy for 7 months duration. Subject suffered from hyperventilation, anxiety, scoliosis, lack of balance, gait disturbances, no social communication. The child showed improvement in most aspects like gait, balance, posture etc. Hippoteraphy was also proven to be effective for reducing hyperventilation. 3 types of respiratory phenotype were identified: forceful, feeble and Apneustic phenotypes each require unique treatment. Physiotherapy plays an important role for these respiratory problems. For forceful breathers rebreathing mask and breath control technique is needed because they develop respiratory alkalosis, for feeble breather's external respiratory support, for apneustic and feeble breathers artificial ventilation is needed because they fail to restart breathing.[14]

3.CONCLUSION

The key words used in the literature search were Rett's syndrome, Physiotherapy management, Cardio Respiratory symptoms and exercises for Rett's syndrome. The electronic databases for this review included were from science direct, Pub Med, The lancet, NCBI, Europe PMC plus, NIH, Journal of Intellectual Disability Research. Literature search was done for past 27 years i.e. from 1988 to 2015. 30 articles are collected regarding Rett's syndrome**.** Only case studies were obtained. There are no Randomized Controlled trails, Systemic Review, Meta-Analysis. Among the studies reviewed, A case study provided evidence for the effectiveness of Hippotherapy and Applied Behavioral Analysis. 2 studies on Treadmill was proven to be effective. A observational study identified 3 types of respiratory pattern and the management to prevent respiratory complications. An article summarized the importance of Hydrotherapy and Treadmill training.. If QT interval is prolonged strenuous activities should be avoided.Rett's syndrome commonly affects females and the subject's survival currently is not possible estimate but the average life expectancy is beyond age 40. Often there is combination of musculoskeletal, neurological & cardio-respiratory signs and symptoms. So often multi-disciplinary approach is needed. Compromise in cardio respiratory system can occur either because of the condition severity or postural compromise such as scoliosis and reduced ambulatory status. People with Rett's' syndrome often die due to Cardio-Respiratory complications or sudden death without causes. Hence the team should work to increase the survival rates.. Though only less studies were available about Retts syndrome, the aim of this literature review is to analyze the importance of Cardio Respiratory physiotherapy. Maintaining fitness is an important criteria to prolong the mobility and prevent structural complications. Interventions like Hydrotherapy, Treadmill training and Hippo therapy can be encouraged and the intensity of exercises should be moderate to prevent respiratory distress.

CONFLICT OF INTEREST

Conflict of interest declared none.

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