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MULTIMODAL REHABILITATION PROGRAM IN ATAXIA TELANGIECTASIA - A CASE REPORT

Physiotherapy

Dr Astha Jain Assistant Professor, Amar Jyoti Institute of Physiotherapy, Delhi University.

ABSTRACT

INTRODUCTION: A-T is an autosomal recessive cerebellar ataxia. The prevalence is estimated to be <1–9/ 100,000.

CASE PRESENTATION: 7 years old female child, came to the physiotherapy OPD with complain of difficulty in walking and breathing, and H/O frequent fall, intentional tremors, generalized body weakness and fatigue.

MANAGEMENT AND OUTCOME: Multimodal rehabilitation programme included relaxation, exercises to prevent secondary complication, improve chest expansion, balance, trunk control, gait & Functional independence. Outcome measures were SARA, BBS, TUG, Barthel Index and Trunk Impairment scale.

DISCUSSION: In a total duration of 12 weeks there was a considerable improvement in static and dynamic balance, mat exercises and gym ball training helped to improve trunk control. Added to this patient improved on gait and had a functional independence.

CONCLUSION: Relaxation Exercises, Balance and Mobility Exercises, Gait Training, Co-ordination Exercises and Functional Training had a significant effect on improving the Quality of Life of a 7-year old child with Ataxia Telangiectasia.

KEYWORDS

Ataxia Telangiectasia, SARA, Balance, functional independence.

1] INTRODUCTION

A-T is an autosomal recessive cerebellar ataxia [1]. A-T is often referred to as a genome instability or DNA damage response syndrome and is characterized by progressive cerebellar degeneration, telangiectasia, immunodeficiency, recurrent sino-pulmonary infections, radiation sensitivity, premature aging, and a predisposition to cancer development, especially of lymphoid origin. Other abnormalities include poor growth, gonadal atrophy, delayed pubertal development and insulin resistant diabetes [2].

The prevalence is estimated to be <1–9/ 100,000, although incidences as high as 1 in 40,000 [3,4] and as low as approximately 1 in 300,000 [4].

The mode of inheritance for A-T is autosomal recessive. A-T is caused by mutations in the ATM (ataxia telangiectasia, mutated) gene, which was cloned by Savitsky et al. in 1995 [5]. ATM is located on human chromosome 11q22-q23 [6].

The hallmarks of Neurological involvement include: Progressive cerebellar ataxia (100% of patients) of the trunk and limbs, starting at walking age (is the presenting symptom in 90% of patients) and leading to wheelchair toward the second decade of life. Extra-pyramidal symptoms (both hyperkinetic movement disorders such as choreoathetosis, myoclonus, polymyoclonus, dystonia, kinetic tremor in 60% of patients, and parkinsonism such as bradykinesia, bradylalia, hypomimia, resting tremor in 70% of patients). Eye movement abnormalities (80% of patients) with characteristic oculomotor apraxia, with nystagmus and various defects in saccade and gaze control. Sensorimotor axonal peripheral neuropathy of various degrees, from a mild form characterized only by hypotony and absence of tendon reflexes, to a severe hypotony and weakness with dramatic ankle and foot tendon retractions requiring surgery [7-11].

No established therapy for AT is currently available: treatments are symptomatic and supportive only, and since last few years no controlled study existed regarding the pharmaceutical reduction of ataxia symptoms in AT. The prevention of primary manifestations of AT has been so far unsuccessful. Therapeutic interventions such as early and continued physical therapy minimize contractures, which appear in almost all individuals with tie and lead to other physical problems, whereas IVIG replacement therapy appears to reduce the number and severity of infections in patients presenting with them. Rehabilitation and supportive care includes physical, occupational and speech/swallowing neuro-rehabilitation.

2] CASE PRESENTATION

7 years old female child came to the physiotherapy OPD with complain of difficulty in walking and breathing, and H/O frequent fall, intentional tremors, generalized body weakness and fatigue. She lives with mother, father and two elder siblings (12y/M & 9y/F).

There was a history of Varicella Zoster Virus Infection (VZV) (Chicken pox), for which she was hospitalized for 8 days.

Comprehensive Physiotherapy Assessment was done which included Muscle Performance, Tone, Reflexes, Postural Alignment Control, Balance, Coordination and Functional Assessment.

On Observation the child had neck and Truncal tremors which were intentional in nature and decreasing with rest. Gait was with wide base, irregular step length, exaggerated arm swing, reduced speed and cadence, difficulty in turning and frequent loss of balance. Eyes were having telangiectasia.

On examination, ROM was passively full in all joints, Generalized Hypotonia, Weakness of all muscles, Sluggish reflexes, intact Cranial Nerves and reduced chest expansion at all three levels. Also, the patient had Ataxia specific features like Trunkal Ataxia, Staccato scanning speech, handwriting larger than the normal, positive Riddoch's Test, Rebound Phenomenon present and poor Co-ordination.

3] ASSESSMENT AND MANAGEMENT

Pre intervention and post interventional assessment of several outcome measures were taken like Ataxia specific scale, SARA (Scale for Assessment and Rating of Ataxia); Balance assessment measures like BBS (Berg Balance Scale) and TUG (Timed up and Go Test); TIS (Trunk Impairment Scale) and Barthel Index.

An extensive rehabilitation was given to the child during this phase including relaxation exercises, Gait and Balance Training, Co-ordination Exercises like Frenkel's Exercise, Functional Training.

Table-1 Multimodal Exercise Program

AIM	OBJECTIVE
To promote Relaxation	Relaxed posture Breathing exercises Frequent rest periods in between exercises
To prevent Secondary Complications	Tightness- Stretching Correcting faulty postures Positional stretch posture Respiratory Complications- Deep Breathing Exercises Segmental Breathing Exercises Prevention of Falls- Progressive balance Exercises Postural Control Training Weight Shifts and Reach-outs Postural Control Training Weight Shifts and Reach-outs

To improve Chest Expansion	Breathing Exercises with spirometer or play therapy like blowing balloon and bits of paper
To improve Balance	Various balance demands in sitting and standing One leg standing Marching Standing on Foam Multidirectional Reach-outs while standing on foam
To improve Trunk Control and Core Stability	Gym Ball exercises Mat Activities – Bridging, Abdominal Curl-ups, Kneeling, Half-Kneeling, Prone Kneeling
To improve Co-ordination	Frenkel's Exercises
To Improve Gait	Gait Training using Mirror, cues, Markers, Obstacle Training
To Improve Functional Independence	Functional training

Table 2: Sara Scores

	0 WEEKS	6 WEEKS	12 WEEKS
Gait	6	4	3
Stance	5	4	2
Sitting	3	2	1
Speech Disturbance	4	4	4
Finger Chase	3.5	3	2
Nose-finger Test	3	2	2
Fast Alternating Hand Movements	2.5	2	1.5
Heel-shin Slide	3.5	2.5	2

Table 3: Outcome Measures

	0 weeks	**6 weeks**	**12 weeks**
BERG BALANCE SCALE	33/56	42/56	48/56
TIMED UP AND GO TEST	26 Sec.	22 Sec.	19 Sec.
BARTHEL INDEX	80/100	90/100	90/100
TRUNK IMPAIRMENT SCALE	20/23	21/23	21/23

4] RESULT

The results of this comprehensive rehabilitation show a significant improvement in Static and Dynamic Balance, Trunk Control, Gait, Co-ordination and Functional Independence.

5] DISCUSSION

As the Genetically inherited condition of Ataxia Telangiectasia is having delayed diagnosis and no standard protocol of Medical or Rehabilitative Treatment for its improvement available; the physiotherapy services are mandatory for the symptomatic improvement and betterment of the quality of life. Whatever the lifespan of a patient with AT should be active and self-sufficient to lead an independent life with no secondary complications.

Based on the therapy provided to child for a duration of 12 weeks it has been found that Balance of the child improved significantly on static as well as dynamic scales where there was a greater improvement in first 6 weeks as compared to the next 6 weeks, with various exercises like one leg standing, Marching, Standing on Foam and balance demands placed in siting and standing position while various routine activities. Trunk control of the child improved with various Mat Exercises and Gym ball Exercises. Co-ordination improved a bit very gradually as observed in the scores of SARA. Gait also showed a significant improvement in the same duration.

There is lack of evidence about the standard protocol for the rehabilitation of patients with AT, so there is a need to perform various such case studies on patients and develop the guidelines for the AT patients.

6] CONCLUSION

Multimodal Rehabilitation Programme including Relaxation Exercises, Balance and Mobility Exercises, Gait Training, Co-ordination Exercises and Functional Training had a significant effect on improving the Quality of Life of a 7-year old child with Ataxia Telangiectasia.

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