IOSR Journal of Nursing and Health Science (IOSR-JNHS)
e-ISSN: 2320–1959.p- ISSN: 2320–1940 Volume 10, Issue 4 Ser. IV (Jul. – Aug. 2021), PP 46-48
www.iosrjournals.org

Physical Therapy for Segawa Syndrome

Dr.Snehal Dharmayat, MPT
Associate Professor, KLEU Institute of Physiotherapy, Belagavi, Karnataka, INDIA

Abstract: *Segawa Syndrome, also known as Dopa Responsive Dystonia, is a rare inherited condition seen in the paediatric age group. This is often misdiagnosed as Cerebral Palsy. Standard protocol of managing these cases is administration of L-Dopa medication which produces a drastic improvement in the symptoms. There is no known evidence of Physical Therapy management of the same.*

Key Words: *Segawa Syndrome, Dopa responsive Dystonia, Physical Therapy*

Date of Submission: 18-07-2021 Date of acceptance: 03-08-2021

I. Case Report:

An 8 year old male child presenting with developmental delay (motor> mental), initially diagnosed as Hypotonic/Dyskinetic Cerebral Palsy with history of 2nd degree consanguinity of the parents. He was born at full term through the vaginal route. He was the 1st of 4 children, other 3 siblings (1 boy & 2 girls) developmentally & neurologically normal. He had a history of delayed birth cry (cried after 5 minutes) & a history of seizures, twice; once at the age of 4 years & second at the age of 6 years was present (after which treatment was started). Examination revealed generalized hypotonia of all 4 limbs (lower limbs> upper limbs) & trunk with wrist dystonia. Knee joint hyperextension was seen bilaterally. Both feet were seen to be in equino-varus at rest changing to valgus on activity such as crawling or walking. Involuntary movements were seen in upper & lower limbs & face, with upper limb movements less severe than lower limb which increased on activity especially walking. Functional evaluation revealed the following:

Gross Motor- Incomplete head control, able to roll prone and back, able to come to sitting from prone but not supine, transit from sitting to crawl position but with a wide base, initiation of crawling but unable to crawl more than few steps due to involuntary movements and fatigue, sitting with wide base of support (preferring a ring sitting position with lower trunk rounding { Flexion}) ,able to come to standing from sitting and squatting but with support, pushing to stand with poor weight shift and locking of knees into hyperextension. He was able to walk with support but with trunk flexion at hip, external rotation of hips and a wide base of support. He did not initiate stepping with hip flexion as he walked with hips in abduction- external rotation. Truncal swaying (involuntary movements) was seen more in standing and walking. Reaching, grasping were fairly good with Athetotic movements seen distally during activity.

Fine Motor- Grasping was done with wrist flexion and metacarpophalangeal joint hyperextension. Release was achieved using tenodesis effect at wrist (wrist extension to neutral). Other activities such as pincer, pad-to-pad and 2-jaw chuck/ 3-jaw chuck were not possible. The child used a palmar grasp for holding a pencil to scribble. Eye- hand coordination was good. Pulling up a zip was not possible

Personal-Social & Adaptive, Speech- Language & Hearing- Vision and hearing were normal. The child had good attention, cognition and comprehension; was friendly & cooperative with good interaction. As per the parents, child had a speech delay but presently had good fluency with extensive vocabulary. Speech was slow and slurred. Feeding difficulties were present during infancy but had recovered with the child being able to self-feed with finger foods but only when sitting with complete back support. Child had difficulty in feeding sitting unsupported, tearing and mixing food, drinking from a cup due to involuntary movements, needing supervision or at times partial assistance. Bowel and bladder control was good. He was able to communicate the need to void but required help to sit on the toilet and for cleaning. He could pull up pants in sitting but with truncal swaying, had difficulty in buttoning and unbuttoning shirt due to involuntary movements. He could assist in wearing a shirt without buttons by shoulder flexion but with abnormal movement in the scapula.

Other notable features were:

Sleep benefit - Severe generalized hypotonia improving in the morning after sleep and rest periods & Occulogyric crisis episodes occurring once in 3-4 days lasting 15 minutes which improve with sleep

Medical Management:

Neurologist's impression- Features highly suggestive of Dopa Responsive Dystonia (DRD) [presenting as global hypotonia]. Although no familial history exists, he has to be considered for DRD.

Global hypotonia and oculogyric crisis suggest possibility of Sepiapterase Reductase deficiency-SEGAWA SYNDROME.
Medications- Tab Syndopa (1 mg/kg/d) in divided doses. Drug induced involuntary movements with syndopa noted, managed by tapering the dose to complete cessation which greatly helped in controlling the movements.

Physical Therapy: Assessment was done for muscle strength & functional performance using Manual Muscle Testing (MRC Grading), GMFM 88 and Wee FIM before and after intervention (Tables 1, 2 and 3). Intervention was provided as follows:

- Active exercises in the gravity eliminated plane were started for muscles of grade 1 &2 along with electrical stimulation using Faradic type of current with trapezoidal wave form to prevent accommodation. Minimal resistance was used when the power increased to 2. Child was able to tolerate the resistance. Care was taken to control hypermobility.
- Functional training was started on mat beginning with activities in lying progressing to various transitions up to standing gradually.
- Trunk muscles were strengthened in the lying position initially progressing to working in kneeling & on the therapy ball.
- Balance training was given in sitting, kneeling & half kneeling. Progression to standing balance could not be started as child got discharged from the hospital & failed to follow up.
- Weight bearing on upper limbs in different positions such as sitting, kneeling were started to facilitate co-contractions & achieve girdle stability with care taken to prevent hypermobility.
- Hand muscle strengthening was started by using the soft ball & putty clay to increase the intrinsic muscle strength. Also resistance was provided for finger abduction- adduction using rubber bands across the fingers .Hand function training was started with the use of ADL boards where activities such as turning a large screw, opening door knobs of different types, turning on/off light switches, etc. were taught. Radial grasp with a pen was initiated by increasing the diameter of the pen using sticking tape near the writing end. Later progression was made to scribbling & drawing lines. Modification of other daily use items such as spoon, cup, etc were told to the parents.
- Static Ankle foot orthosis (AFO) was prescribed for aligning the ankle during activities.

Table 1- MUSCLE STRENGTH (MRC Grading)

	Pre	Post
Upper limb: Shoulder flexors		
Extensors	1+	2+
Abductors	1	3
Internal rotators	1+	2
External rotators	1+	3
Elbow Flexors	2	4
Extensors	1+	3
Wrist Flexors	1+	2
Extensors	1	3
Radial Deviators	1	2+
Ulnar Deviators	1	2+
Lower limb: Hip Flexors	1+	3
Extensors	1	3
Abductors	1	3
Adductors	1+	3
Internal Rotators	1	2
External Rotators	1	2
Knee Flexors	1+	3+
Extensors	1	3
Ankle Plantar flexors	1+	2+
Dorsiflexors	1	3
Invertors	1	2
Evertors	1	2

Table 2- GMFM 88 SCORE

Dimension	Pre	Post
A	**43**	**49**
B	**39**	**54**
C	**12**	**28**
D	**05**	**18**
E	**05**	**10**

Table 3- **FUNCTIONAL ASSESSMENT**

	Wee FIM Score
Pre	64
Post	70

II. Discussion:

Dopa responsive Dystonia (DRD)/ Segawa Syndrome also known as 'hereditary progressive primary dystonia' characterized by onset during childhood, circadian fluctuation of symptoms & a dramatic & sustained response to low doses of oral administration of levodopa, was first described by Segawa et al in 1976.[1] Age of onset is usually between 4-8years (age range 9 months to 16 years). The disorder is more common in females inherited in the autosomal dominant fashion [2, 3, 4] but has also been reported in males [5, 6, 7,8]. Dystonia is progressive & more pronounced in the leg than in the arms, as was seen in the present child; but can also affect the neck & axial musculature.[2] The dystonia characteristically worsens throughout the day & there is marked benefit from sleep which was seen in the present child.[3] The condition is often misdiagnosed as Cerebral Palsy, or other forms of neurodegenerative disorder [9, 10,11]There are 2 forms: mild & severe. In the mild form, symptoms typically begin in childhood. Children develop jerky movements that quickly progress to physical rigidity. These children show spastic movements, and make very little voluntary movement. If untreated, children with Segawa syndrome may have expressionless faces, drooping eyelids, tongue tremors, and drooling problems. They will show both intellectual and physical developmental delays. The severe form of the disease will appear in infancy, usually before six months of age. Affected infants have delayed motor skills, weakness in the chest and abdomen, rigidity in the arms and legs, and problems with movement. These children will eventually have learning disabilities, problems with speech, and behavioral/psychological problems. [12] Segawa Syndrome/ DRD has also been reported in Indian literature but less frequently [5,13,14]. The role of Physical therapy in the management of DRD has not been documented till date. This present report, therefore, is an effort to bridge this gap in knowledge. Physical therapy will help the children in improving their motor control, strength & function, thus improving quality of life.

References:

[1]. Segawa M, Hosaka A, Miyagawa F, Nomura Y, Imai H. Hereditary progressive dystonia with marked diurnal fluctuations. Advances in Neurology 1976; 14:215-33

[2]. Livanainen M, Kaakkola S. Dopa responsive dystonia of childhood. Developmental Medicine & Child Neurology 1993; 35 (4): 362-67

[3]. Nygaard TG, Marsden D,Fahn S. Dopa responsive dystonia : Long term treatment, response & prognosis: Neurology1991;41:174-81

[4]. Gordon N. Dopa responsive dystonia: A widening spectrum. Developmental Medicine & Child Neurology 1996;38:554-59

[5]. R. Verma,S. Misra,N.N Singh, D. Kishore. Patient with limb girdle dystrophy presenting with dopa- responsive dystonia- A case report. Neurology India, April-June 2003; 51(2): 252-253

[6]. Neel Kamal, Deepti Pagare Bhat, Emma Carrick. Dopa- Responsive Dystonia (Segawa Syndrome). Indian Pediatrics July 17, 2006; 43: 635-38

[7]. Mohammad Ghofrani, Hassan Tonekaboni. Dopa Responsive Dystonia: A case report. Medical Journal of the Islamic Republic of Iran, Fall, 1999 November; 13(3):235-236

[8]. Yosunkaya E, Karaca E, Basaran S, Seven M, Yuksel A. Marked improvement in Segawa Syndrome after L-dopa & selegiline treatment. Pediatric Neurology, 2010 May;42(5): 348-50

[9]. Jae-Hyeok Lee, Chang-Seok Ki, Dae- Seong Kim, Jae-Wook Cho, Kyung-Phil Park & Seonhye Kim. Dopa- responsive Dystonia with a Novel Initiation Codon Mutation in the *GCH1* Gene Misdiagnosed as Cerebral Palsy. Journal of Korean Medical Sciences. 2011 September; 26(9): 1244–1246.

[10]. Boyd K, Patterson V. Dopa responsive dystonia: A treatable condition misdiagnosed as Cerebral Palsy. British Medical Journal1989; 298: 1019-1020

[11]. Mohammed M. S Jan. Misdiagnoses in Children with Dopa-responsive Dystonia. Pediatric Neurology 2004;31(4):298-303

[12]. https://www.counsyl.com/services/family-prep-screen/diseases/segawa-syndrome/ accessed on 13th September 2013

[13]. Rajiv Mittal, Jatinder S. Goraya, Srikanta Basu. Dopa-Responsive Dystonia. Indian Pediatrics 2001;38:1056-58

[14]. Kuljeet Singh Anand, Jyoti Garg, Yogesh Valecha, Ranjit Singh Makar. Dopa-Responsive Dystonia: An Unappreciated Movement Disorder. Journal, Indian Academy of Clinical Medicine October-December 2007;8(4):339-41

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