



## THE EFFECT OF AN INDIVIDUALLY TAILORED EXERCISE PROTOCOL FOR DEVELOPMENTAL DELAY IN BECKWITH-WIEDEMANN SYNDROME – A CASE REPORT

### Physiotherapy & Rehabilitation

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### ABSTRACT

Beckwith Wiedemann Syndrome is a genetic disorder that presents with various clinical features and one among them is global developmental delay. The lateralized overgrowth and medical complications like umbilical hernia and abdominal wall defects could lead to reduction in time obtained at exploring movements and these children need help to attain the milestones. A tailor-made physiotherapy regimen can be helpful in promoting the child's overall motor skill development.

### KEYWORDS

Beckwith Wiedemann Syndrome, global developmental delay, exercise protocol.

#### INTRODUCTION

Beckwith-Wiedemann Syndrome (BWS) is a human genomic imprinting disorder that presents with a wide spectrum of clinical features including overgrowth, abdominal wall defects, macroglossia, neonatal hypoglycemia and predisposition to embryonal tumors<sup>1</sup>.

Beckwith Wiedemann spectrum is usually caused by dysregulation of the chromosome 11p15 imprinted region and involves overgrowth in multiple tissues, often in a mosaic state. Classical BWS and lateralized overgrowth are considered subsets of the BWS spectrum. BWS is most diagnosed in the neonatal period or in early childhood<sup>2</sup>.

The incidence of BWS is estimated to be 1 out of 13700. The incidence is equal in males and females with notable exception of monozygotic twins that show a dramatic excess of females. BWS usually occurs sporadically but familial transmission occurs in 15% of the cases<sup>3</sup>.

The excess growth in BWS can manifest as whole body or regional overgrowth due to cellular hyperplasia which occurs in 25% of the patients but not apparent at birth<sup>4</sup>. This kind of hypergrowth can also involve one side of the body or be localized. It can be accompanied by hyperplasia of the organs possibly with a component of asymmetry characterized by dissimilar maximum renal diameters between the organs corresponding to the ipsilateral hyperplasia of the body or lower limb<sup>5</sup>. This hyperplasia can result in complications depending on the site of onset. When arising in lower limbs, it can cause varying degrees of asymmetry which can negatively affect walking and can lead to scoliosis and incorrect postural attitudes<sup>5</sup>.

#### CASE PRESENTATION

The study presents the results of a 2-year physiotherapy rehabilitation program in a child with BWS.

Informed consent was obtained from the parent of the minor for the publication of this case report.

The child was born at 32 weeks gestation and was low birth weight. She was born small for her gestational age. Oomphalocele was detected antenatally and correction was done. She developed seizures and apnoea after surgery which was managed with calcium gluconate injections and anticonvulsants. She was also diagnosed with Patent Ductus Arteriosus and Patent Foramen Ovale left to right shunt. She underwent a left palate repair when she was 2 years old. An MRI of the brain and whole spine screening was done when the child was 2 years old. Imaging features were suggestive of periventricular leukomalacia (Grade II). Spinal cord showed normal morphology without myelocoele or myelomeningocoele. Motor and sensory nerve conduction studies were done which did not show any abnormalities.

The diagnosis of BWS was confirmed in the child on 23<sup>rd</sup> December 2019 when the child was 1 year and 4 months old. She was referred for genetic testing because of developmental delay, open mouth appearance, macroglossia, oomphalocele and umbilical hernia.

The child presented to our outpatient rehabilitation unit when she was 2 years old with severe developmental delay. The diagnosis of BWS was already confirmed at that point in time. She had not undergone any rehabilitation prior to this.

On evaluation, the following observations were noted:

- 1. Behavioural Issues:** The child had sensory and motor deficits along with behavioural disturbances. She was startled by any kind of visual or auditory stimuli and would get into a crying fit when exposed to any unfamiliar voice or light. She had severe stranger anxiety. She showed features of tactile defensiveness. She took a very long time to get accustomed to handling by a single therapist.
- 2. Sensory Deficits:** Sensory examination revealed loss of sensation in S1 dermatome (buttocks, anterior aspect of the thigh and leg) bilaterally.
- 3. Motor Deficits:** Motor examination revealed severe tightness in tendoachilles, adductors and hamstrings bilaterally. Her feet were held in plantar flexion. She had collapsed arches in her feet. Genu valgum was also present bilaterally. Strength testing revealed weak scapular muscles, abdominals, gluteal muscles, hamstrings, quadriceps, and dorsiflexors.
- 4. Developmental History:** She had started rolling over to her side in an incorrect pattern by the time she was 2 years old. She was unable to come from side lying to sitting. If she was made to sit, she would assume a sacral sitting posture with maximal support. The child presented with global developmental delay.

Depending on the assessment of the physical impairments, a structured patient-centered exercise program was developed for rehabilitation.



**Fig:1** Child in standing position (Pre-rehab)

**DESCRIPTION OF REHABILITATION**

**PHASE I (2 years - 2 years 8 months)**

**Sensory Approach**

- Desensitization: Exposure to different textures for tactile adaptation in graded progression from soft to rough textures on both palmar and plantar surfaces.
- Exposure to low threshold light and sound stimuli multiple times in between treatment sessions. Lights of different colours were used starting with primary colours of red and blue.
- Exploring different textured toys to improve tolerance of unfamiliar objects.
- Sensory reeducation protocol in the S1 dermatome.

**Motor Approach**

- Neurofacilitatory techniques were used to initiate contraction in the extremely weak group of muscles.
- Mobilization of the joints of the foot, myofascial release of gastrosoleus, hamstring and adductors.
- Structured strengthening protocol for bilateral lower limbs. Trunk flexors, extensors, side flexors and core muscle activation and strengthening.
- NDT intervention designed to target the milestones in a developmental sequence.

The following improvements were noted in the child:

1. Improvements in sensory and behavioural responses: The tactile defensiveness and stranger anxiety had reduced significantly after 6 months of rehabilitation. Different therapists were slowly introduced for her sessions to improve tolerance to handling and to promote social bonding. Her stranger anxiety had reduced as demonstrated by her reduced startle response to unfamiliar voices and reduced crying episodes to the presence of new faces. The physical environments during therapeutic sessions were kept the same to enhance cooperation. Once she adjusted to the surroundings, her behavioural responses became stable and calmer.

2. Milestone history: By the end of phase I, the child started to roll into prone and back into supine. She could come into side sitting from supine in a correct pattern with upper limb support. She was able to maintain kneeling and quadruped with mild support. She was able to initiate crawling with commands and tactile prompting.

3. Improvements in strength: Hamstring and adductor tightness reduced and night splinting was started for TA tightness. Strength improved in gluteal muscles with a score of 2 on MMT. Hamstring was at score 2 as well and dorsiflexors started to show a flicker of contraction bilaterally. Quadriceps strength was at 3 on MMT. Scapular muscle strength and control improved significantly. Trunk and core musculature still showed weakness and pelvis was in anterior tilt with exaggerated lordosis.

After significant improvements in muscle strength were noted, weightbearing through feet had to be introduced and for that an ankle foot orthosis was used and we progressed to the next phase of the rehabilitation plan.



**Fig:2:** Child standing with mild support after Phase-I rehab.

**Manual Muscle Testing Scores At The End Of Phase I Were As Follows:**

S. No	Muscle	SCORE (Right)	SCORE (Left)
1.	Hip flexors	2+	2+
2.	Hip abductors	2	2-
3.	Hip extensors	3	3
4.	Quadriceps	3	3
5.	Hamstrings	1+	1+
6.	Dorsiflexors	1	1
7.	Trunk flexors	2	
8.	Trunk extensors	2	
9.	Sideflexors	2	2

**PHASE II**

(2 years 8 months to 3 years 6 months)

Weightbearing activities were started with an AFO with arch correction for adequate weight distribution.

Weight bearing activities included the following:

S. No	Type of exercise	Frequency	Intensity	Time
1.	Sit to stand	6 times a week	10 reps*1 set	
2.	Half kneeling	6 times a week		≈10 minutes
3.	Standing with support	3 times a week		≈30 minutes
4.	Standing weight shifts	3 times a week		≈10 minutes
5.	Balance reaction training	3 times a week		≈10 minutes
6.	Squats	3 times a week	10 reps*2 sets	
7.	Lunges	3 times a week	10 reps*2 sets	

In addition to the above, potty sitting was maintained for 10 minutes every alternate day during sessions and was also continued at home. A 45-minutes session was dedicated for lower limb, trunk, and scapular muscle strengthening. Lower limb strengthening and trunk strengthening were carried out in a standing position. Adductor stretches were done using a bolster and sit to stand from the stretched sitting position was added to the protocol. Standing stretch for tendo-achilles was started as well and maintained for 5 minutes before weightbearing activities were started. Hippotherapy was also introduced during this time for vestibular stimulation. 15 minutes of hippotherapy was added to the protocol 3 times a week.

Once the child was able to stand independently with considerable control over weight shifts during stepping, gait training was initiated with the ankle foot orthosis. The child attained gait with mild support by the end of phase two. Once the child was able to ambulate with support for her needs, she started school. After the initiation of schooling, the child was found to be able to participate in the new social environment under supervision and mild assistance from her teachers. With this level of functioning achieved, we tapered her physiotherapy rehabilitation to once a week for maintenance and continuous assessment, thus entering phase III of our rehab.

**PHASE III**

In this phase, the focus was to maintain the strength gains achieved during the first two phases. All the strengthening protocols are being carried out at home by her devoted mother. The child actively engages in group play and participates in all activities at her school. She comes in to the rehabilitation center once a week for monitoring. Weekly assessments of strength, flexibility and mobility are taken to make sure there are no further additions needed in the protocol followed at home.

**Present Status Concern**

Her postural control and balance reactions are the area of concern at present. She has an anterior pelvic tilt and weakness in her trunk musculature which is affecting her posture and weight distribution during standing and gait at normal pace.

**DISCUSSION**

Beckwith-Wiedemann syndrome is the most common congenital syndrome associated with overgrowth. In this case study, the relevant presentation from a rehabilitation point of view was global developmental delay. She was born small for her gestational age. She did not show features of any disproportionate overgrowth and hence there were no postural concerns. The features that led to the diagnosis

was macroglossia, open mouth appearance, oomphalocoele in addition to her global developmental delay.

Through our structured rehabilitation program, we planned to address the issue of developmental delay. When she came to us for rehabilitation, she was medically stable and was not undergoing any treatments.

The rehabilitation program was planned at a pace that could be tolerated by the child as she had severe sensory and behavioural issues. Once that hurdle was crossed, the protocol was made more vigorous to attain maximal functional independence.

A rehabilitative program that was consistent over a period of 1 year and 6 months along with reliable support from the family in the form of adherence to a structured home exercise program is what helped the child in attaining her developmental milestones. She is now able to perform age-appropriate activities under supervision. She has difficulty standing for prolonged duration without support during school assembly. She can walk along the hallways by touching the walls. She is apprehensive of falling and prefers to have someone by her side as she walks.



**Fig:3** Current status of the child

## CONCLUSION

A well-structured rehabilitation exercise plan as an early intervention when developed as per the child's needs and when continued with utmost consistency at home, can give better results in the achievement of functional independence of the child in future. Individualized tailored interventions need to be developed by the physical therapist to improve the outcome measures in children with gross developmental delay of genetic origin.

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