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Integrated Multidisciplinary Physiotherapy Intervention in a Child with Baker-Gordan Syndrome, Autism Spectrum Disorder and Global Developmental Delay: A Case Report

Dr. Akshay Singh Parihar[1*]; Dr. Divya Meshram[2]; Dr. Simran Kanojia[2]

[1]1st Year Mpth (Neurophysiotherapy), [2]Bpth

[1,2]Rashtrasant Janardhan Swami College of Physiotherapy (Affiliated to Maharashtra University of Health Sciences, Nashik, Ahilyanagar, Maharashtra, India).

Corresponding Author –Dr. Akshay Singh Parihar[1*]

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Abstract:

- ***Introduction:***

Baker-Gordon Syndrome (BAGOS) is an ultra-rare neurodevelopmental disorder caused by mutations in the *SYT1* gene. It presents with global developmental delay (GDD), hypotonia, motor impairments, intellectual disability, and behavioral problems. Co-occurrence with Autism Spectrum Disorder (ASD) and GDD further worsens functional outcomes, leading to deficits in motor, sensory, social, and cognitive domains. Physiotherapy plays a key role in improving developmental milestones and quality of life through multidisciplinary rehabilitation.

- ***Methodology:***

The patient, a two-year-old female with genetically confirmed Baker-Gordon Syndrome (*SYT1* mutation) and associated hypotonia, gross motor delay, impaired speech, poor eye contact, and delayed social skills, was evaluated using GMFM, SRS-2, ISAA, and BFMF. She underwent a six-week physiotherapy program incorporating neurodevelopmental therapy, sensory integration, oro-motor training, and task-oriented activities, along with parent training for home-based reinforcement.

- ***Results:***

Motor function improved with independent sitting for one-minute, better trunk control, and initiation of crawling. Oro-motor skills advanced with chewing, babbling, and monosyllables, while social responsiveness improved through eye contact, social smile, recognition of mother, and response to name. Outcome measures showed gains: GMFM 9.2%→23.8%, SRS-2 149→102, ISAA 78→70, and BFMF Level V→IV.

- ***Conclusion:***

Multidisciplinary physiotherapy improved motor, sensory, oro-motor, and social functions in a child with Baker-Gordon Syndrome with ASD and GDD. Early diagnosis, family involvement, and individualized rehabilitation are key, and more studies are needed to guide management of SYT1-related disorders.

Keywords: *Baker-Gordon Syndrome, SYT1 Mutation, Autism Spectrum Disorder.*

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I. INTRODUCTION

A hereditary neurodevelopmental disorder (NDD), Baker-Gordon Syndrome (BAGOS) is brought on by pathogenic mutations in the synaptotagmin 1 gene (SYT1) at 12q21. Its symptoms range from moderate to severe intellectual incapacity. One Involuntary movements, behavioral problems, facial dysmorphology, and changes in electroencephalogram (EEG) patterns are the hallmarks of BAGOS. With an estimated prevalence of 1:1,000,000 and an early onset in childhood, it is a rare genetic syndrome. Global developmental delay from birth, including hypotonia, delayed or absent walking, poor or nonexistent speech, and moderate to profoundly impaired intellectual development, are the hallmarks of the condition. 4–6 Additional prevalent traits include self-mutilation, episodes of aggression, sleep disturbances, food issues, acid reflux, and predictable and stereotypical behaviors that may be similar to autism spectrum disorder. [6]

An ultra rare neuro developmental disorder that is Baker Gordan syndrome caused by gene mutation at SYT1 gene shown in Fig.1:

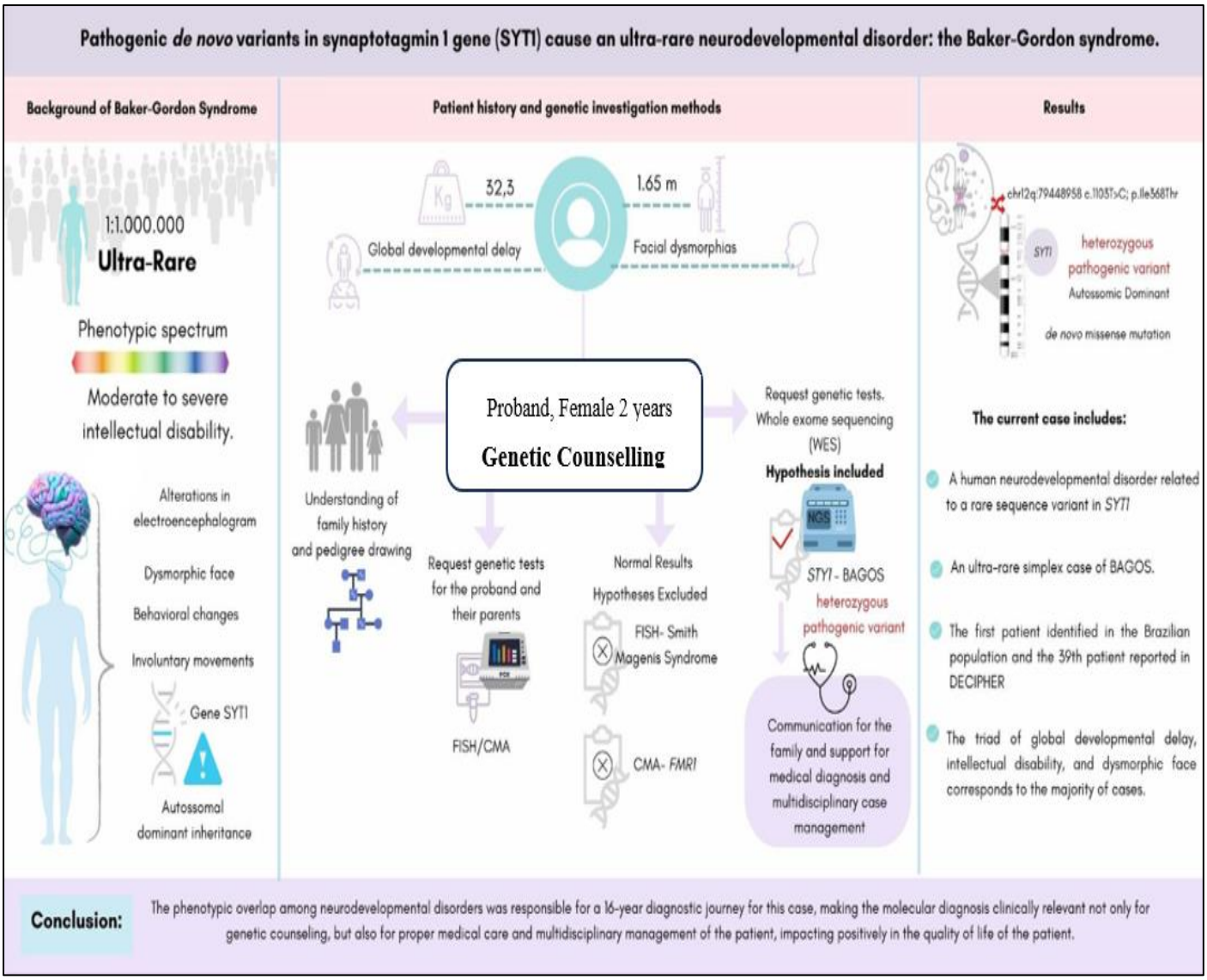

Fig 1 Baker Gordan Syndrome

Autism is a disability characterized by repetitive and stereotypical patterns of behaviour and activities, as well as substantial difficulties with social interaction and communication. Before the age of three, deficiencies in language development and social interaction become noticeable. This disease is known as autism spectrum disorder (ASD) in youngsters. ASD is a condition that impacts a child's whole development, growth, and neurological system [1].

A youngster with ASD may struggle to develop social skills and frequently struggles to communicate. According to the Diagnostic and Statistical Manual of Mental Illnesses-5 (DSM-5), ASD is a neurodevelopmental disease marked by limited, repetitive patterns of behaviour, interests, or activities, as well as ongoing deficiencies in social communication and social interaction in a variety of contexts [4].

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Children with GDD had a 62.3% prevalence of ASD.[7] GDD refers to a child's inability to follow age-appropriate developments.

Children with GDD often experience challenges in their gross motor skills, which can in turn affect other areas of development such as: Fine motor skills, language and speech, cognition, social and behavioural problem, poor intellectual skill and problem in self-care. [1]

A gross development delay is when two or more developmental stages are missed or stopped, whereas a specific developmental delay is when there is deficit in one area. The causes of neurodevelopmental delay are numerous. These consist of some neurological or non-neurological impairments, medications, infections, prenatal difficulties and genetic anomalies.[2]

The International Clinical Epidemiology Network-India (INCLEN) carried out a multi-centric study to assess the prevalence of neurodevelopmental abnormalities in children aged 2 to 9 from both rural and urban backgrounds. Using appropriate interviewing techniques, validated instruments and consensus criteria, this study found that the prevalence was 3.1% in children aged 2 to 6 and 5.2% in children aged 6 to 9.[3]

GDD may result from birth trauma, maternal alcohol use or exposure to chemicals, including lead, during pregnancy, hypoxia at delivery, neonatal cerebral infections, traumatic brain insults, and perinatal infections, including Rubella, HIV, CMV, and others. Poor nutrition, lack of environmental stimulation, poverty, and inadequate child care or supervision are less common causes [1].

The development of a child's gross motor skills is essential. If gross motor skills do not develop according to the child's age, it can lead to delays in other movements. Children with head control balance issues, for example, may experience difficulty swallowing food, breathing effectively, sleep disturbances, and functional hand disorders. Additionally, gross motor skills are closely linked to cognitive development and understanding. Recognizing the role of gross motor skills in development underscores the importance of mastering treatment with a gross motor functional improvement approach. According to normal movement development, head control is the first milestone, followed by other body developments. One aspect of head control is the midline position, which refers to the head being in the middle of the body.[8]

The gross motor function is measure by a “gold standard” scale known as Gross motor function measure (GMFM). GMFM obtained an ICC of 0.99 (95% Confidence Interval = 0.972–0.997), indicating good levels of validity. [9]

The Social Responsiveness Scale-2 (SRS-2) is a widely used screening tool for statistically evaluating symptoms of autism spectrum disorders. SRS can assist in identifying and measuring the spectrum of autism symptoms in the general child population. Parents can seek early intervention if the at-risk population is identified. It obtained an ICC of 0.72-0.97.[10]

The Ministry of Social Justice and Empowerment, the Ministry of Health and Family Welfare, and the National Trust collaborated to create the Indian Scale for Assessment of Autism (ISAA). Its intended goal was to diagnose and rate severity, which was then translated into the degree of impairment, in order to facilitate certification and the use of the Welfare of Persons with Autism, Cerebral Palsy, Mental Retardation and Multiple Disabilities Act" benefits. Inter-rater and test-retest reliability were 0.99 and 0.93-0.99, respectively. [11]

Bimanual Fine Motor Function (BFMF) is a five-level measure that classifies children's hand functions, with level I being the best function and level V the most restricted. The ICC was 0.86 overall. High intra-rater (κw: >0.90) and inter-rater (κw: >0.85) reliability were shown by Cohen's weighted kappa.[12]

II. CASE REPORT

A female infant, who is currently 2 years old and the first born child, was born on May 02, 2023.Consent was obtained from her mother. At 8 months old, she present with a complaint of not being able to hold her neck and being unresponsive to visual or aural stimuli. For that her parents went to a pediatric neurology and cardiology center, and she was referred for genetic counselling and medical support. According to the results, she has Baker-Gordan syndrome because of a mutation in the SYT1 gene. During history taking, the mother told the baby weighed 3.4 kg and was full term and delivered via caesarean section because of malpresentation in the womb. The cry was audible as soon as the baby was born. Due to the mother's inadequate supply of breast milk, the baby was unable to breastfeed after birth for 3 days. The child was referred for paediatric physiotherapy treatment in January 2024 at the age of 8 months for delayed milestone treatment and autism spectrum disorder. The treatment focused on neck control, rolling, sitting, fine motor skills, language, social skills. However, due to family issues, the patient was unable to receive regular physiotherapy treatment for 6 months. She is currently receiving routine physiotherapy treatment at the age of two years and two months, with crawling, independent sitting, and walking being the primary areas of concern.

➢ *Examination*

Accordingly, the child exhibited delayed gross and fine motor domains when compared with the typical developmental age detailed in Table 1.

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Table 1 Gross Motor Development Assessment

Gross Motor	Typical Developmental Age	Chronological Age of the Patient
Head Control	3 months	Achieved at 9 months
Rolling	4-6 months	Achieved at 12 months
Sitting- with support	6 months	Not Achieved
Sitting without support	8 months	Not Achieved
Crawling	9-11months	Not Achieved
Standing- with support	9-12 months	Not Achieved

Table 2 Fine Motor Development Assessment

Fine Motor	Typical Developmental Age	Chronological age of the patient
Hand regard	12-20 weeks	Not Achieved
Hand opening	3-4 months	Achieved at 4 Months
Bidexterous reach	4 months	Not Achieved
Unidexterous reach	6 months	Achieved at 6 Month
Objects to midline	3-4 months	Not Achieved
Transfer object to one hand to other	6-7 months	Not Achieved
Pincer grasp	9-10 months	Not Achieved
Hold out objects but does not release	8-9 months	Not Achieved

Regarding social development of the child exhibits delay when compared to typical developmental age Table 3:

Table 3 Social Developmental Skill Assessment

Social Skills	Typical Developmental Age	Chronological age of the patient
Gaze contact	2 Months	Not Achieved
Smile	3 Months	Achieved at 2.5 Months
Social Smile	2 Months	Not Achieved
Recognises mother	3 Months	Not Achieved
Recognises strangers	6 Months	Not Achieved
Waves bye	9 Months	Not Achieved
Comes when called	12 Months	Not Achieved
Play in group	4 Year	Not Achieved

Regarding language development of the child exhibits delay when compared to typical developmental age Table 4:

Table 4 Language Developmental

Language development	Typical developmental age	Chronological age of the patient
Babbling	3 Months	Not Achieved
Monosyllables	6 Months	Not Achieved
Bi syllables	9 Months	Not Achieved
One word with meaning	12 Months	Not Achieved
5 word with meaning	18 Months	Not Achieved
Simple command	2 Years	Not Achieved
Identifies parts of the body	4 Year	Not Achieved
Speech intelligent to strangers	5 Year	Not Achieved

- *Reflex Examination:*

Table 5 Reflex Examination

Primitive reflexes	Typical age	Integrated at age
Sucking	Birth to	Integrated
Rooting	Birth to 3-4 Months	Integrated
Moro's reflex	Birth to 4-6 Months	Integrated
Palmer grasp	Birth to 3-4 Months	Present
Plantar grasp	Birth to 6-8 Months	Integrated
ATNR	Birth to 2 Months	Integrated
STNR	6 Months to 9-11 Months	Integrated
TLR	Birth to 3 Years	Integrated
Parachute reflex	8 Months and not disappear	Absent

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Landau's reflex	Birth to 12-24 Months	Present
Galant reflex	Birth to 2 Months	Integrated
Flexion Withdrawal	Birth to 2 Months	Integrated
Extension Withdrawal	Birth to 4 Months	Integrated
Neck on body	Birth to 4-6 Months	Integrated
Body on body	Birth to 4-6 Months	Absent

➢ *Clinical Diagnosis*

Genetic testing revealed, A heterozygous Missense variant c.909G>A In Exon 9 of the SYT1 Gene that results in the amino acid substitution p. Met303Ile was identified. The observed variant is a novel variant in gnom AD exomes and genomes, respectively. The severity of the impact of this variant on the protein is high, based on the effect of the protein and REVEL score. Rare Exome Variant Ensemble Learner (REVEL) is an ensemble method for predicting the pathogenicity of missense variants based on a combination of scores from 13 individual tools: Mut Pred, FATHMMv2.3, VEST 3.0, PolyPhen-2, SIFT, PROVEAN, Mutation Assessor, Mutation Tester, LRT, GERP++, SiPhy, PhyloP, and phast Cons. The REVEL Score for an individual missense variant can range from 0 to 1, with higher score reflecting a greater likelihood that the variant is disease causing. Based on the above evidence, the variant has been classified as Uncertain Significance according to the ACMG guidelines.

III. PHYSIOTHERAPY INTERVENTION

Table 6 Physiotherapy Intervention as Mentioned in Detail.

Problem List	Physiotherapy Goals	Physiotherapy Interventions
Poor response to name	To improve response to name and visual tracking	• Pairing names and call with favorite object. • Eye contact games. • Light and colour tracing • Parent training for social interaction.
Impaired speech and oro-motor skills	To enhance speech and oro-motor skills	• Oro-motor stimulation using senso brush. • Chewing tubes • Blowing through straw and candle • Blowing balloon • Soap bubble blowing game • C-icing 15 secs with 5 reps/session • Practice one syllable to two syllable word
Impaired Sensations	Sensory integration	• Brushing protocol- Fast brushing • Play with different texture for eg. Sand, slime, beans etc. • Swinging and rocking on vestibular ball • Visual tracking games • Light and colour tracing objects • Sound discrimination tasks • Deep pressure eg. Joint compression, tight hugs.
Delayed developmental Milestone	To improve Gross motor function – Fine motor functions-	• Neural developmental technique- Rhythmic initiation, Gentle weight shift and in sitting and rocking in quadruped • Vojta therapy for crawling 10 reps /session • Tunnel crawling • Kneeling and half kneeling 10 sec hold with 10 reps • Sit to stand with support on different surfaces • Fine motor bi manual therapy to improve eye and hand coordination • Putty clay activities • Scribbling using colored chalks • Sensory bin play using rice, sand, grains • Peg board activity • Reaching and grasping activities using toys on different shape and size • Building block activities • Squeezing ball
Hypotonia	Tone management	• Rood's approach- Slow stroking, fast brushing, icing 3-5 strokes with movement (Hamstring, calf), quick stretching, heavy joint compression 10-15 reps/session

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Weak core, upper and lower limb muscles	To improve the strength of core, upper and lower limb muscles	• Vestibular stimulation • Gentle weight shifts in sitting and quadruped then progress to standing • Pelvic bridging- 10 reps x 3 sets • Abdominal curls – 10 reps x 3 sets • Back extensor exercise (On wedge and swiss ball) • Task oriented activities- Throwing and catching the ball, Kicking the ball, Reach outs. (On mat and swiss ball)
Impaired balance	To improve balance	• Static and dynamic balance training with play therapy • Weight shifts and pertubations • Balance board activity • Rocking activity
Difficulty in ADL's	To improve ADL's	• Practice brushing, bathing, grooming activities, donning on and off t-shirt. • Practice holding spoon.
Impaired cognition	Cognition and skill development	• Activity with flash cards • Puzzle activity • Showing family member's photos • Identifying 5 common objects/body parts • Following 2 steps command (for eg: take the ball and put it in the box)

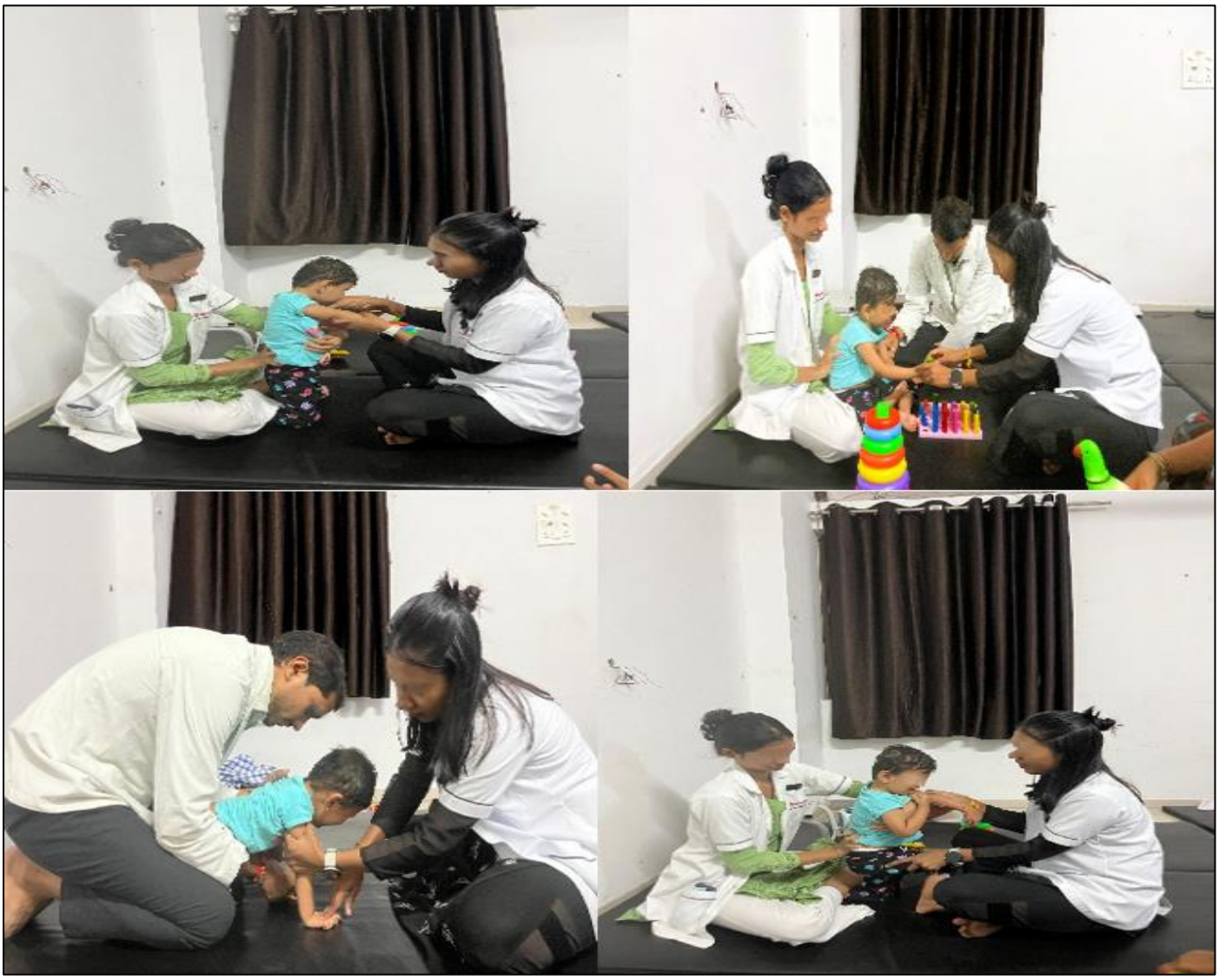

Fig 2 Physiotherapy Intervention

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IV. POST PHYSIOTHERAPY ASSESSMENT

A six-week assessment of the participants' muscular tone, motor control, primitive reflexes, and functional improvement was conducted following the prescribed protocol. The child's oromotor performance and visual tracking both improved following the intervention. She chewed her food and started talking incoherently, using terms like "ma." 30 minutes of sitting without aid was attained, and sitting with support got better. She can use her fine motor abilities to align the object with the midline. She could also make eye contact, smile socially, and recognize her mother. Her trunk control has improved, and she can now carry objects on the midline.

Table 7 Post Treatment Assessment

Sitting with support	**Achieved**
Sitting without support	Achieved (For 30 Min)
Gaze Contact	Achieved
Social Smile	Achieved
Recognizes Mother	Achieved
Babbling	Achieved
Monosyllables	Achieved
Parachute Reflex	Present

Table 8 Outcome Measures

Outcome Measure	**Pre**	**Post**
GMFMS	9.2%	23.8%
SRS-2	149	102
ISAA	78-Mild Autism	70-Mild Autism
BFMF	Level V	Level IV a

- *Outcome Measure:*

(GMFM-Gross Motor Function Measure, SRS-2-Social Responsiveness Scale 2, ISAA-Indian Scale for Assessment of Autism and BFMF-Bimanual Fine Motor Function).

V. DISCUSSION AND CONCLUSION

➢ *Discussion*

- *Clinical Significance of Baker-Gordon Syndrome*

With varied manifestation, Baker-Gordon Syndrome (BAGOS) is an extremely rare neurodevelopmental disease linked to pathogenic mutations in the SYT1 gene. Among the characteristics of the illness shown by the child in this study were hypotonia, delayed gross and fine motor development, damaged speech, and social interaction impairment.

These results support already reported phenotypes of SYT1-related illnesses, which usually present as general developmental delay and behavioral dysregulation.

- *Co-occurrence with Autism Spectrum Disorder and Global Developmental Delay*

BAGOS's coexistence with autism spectrum disorder (ASD) and Global Developmental Delay (GDD) greatly worsened developmental impairments. Particularly challenges in communication, repetitive behaviors, and social interaction problems are among the high incidence of ASD traits shown in children with GDD. Here, lack of eye contact, babbling, and response to basic instructions point up the synergistic influence of ASD traits on development.

- *Rehabilitation Approach*

An interdisciplinary physical therapy program was started in light of the several impairments. Included were neurodevelopmental therapy techniques (e.g., rhythmic initiation, Vojta therapy), sensory integration (vestibular stimulation, brushing, texture play), oro-motor therapies (straw blowing, chewing tubes, bubble play), and task-oriented functional training (crawling, sit-to-stand, balance board). These methods complement data showing activity-based, family-centered rehabilitation in children with intricate neurodevelopmental problems.

Gross motor skills (partial head and trunk control, crawling attempts, better sitting balance), oro-motor activities, and social responsiveness (greater response to name, eye contact start) all showed improvement. These increases support the part of physiotherapy in using neuroplasticity and enabling developmental milestones, even in children with major genetic syndromes.

- *Limitations and Future Directions:*

Although results are good, the rarity of BAGOS restricts availability of standard clinical guidelines, so necessitating reliance on protocols created for ASD, GDD, and cerebral palsy groups. Irregular treatment in early months owing to family conditions most probably slowed growth as well.

Long-term prognosis is still unknown, therefore ongoing treatment and careful follow-up are emphasized. Future research should emphasize multicenter partnerships to record clinical trajectories and create consistent rehabilitation plans for SYT1-related illnesses.

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➢ *Conclusion:*

This case emphasizes how a well-organized, individualized physiotherapy program could help a kid with Baker-Gordon Syndrome complicated by autism spectrum disorder and Global Developmental Delay to improve motor, sensory, and functional areas.

- *Important Consequences are:*

✓ Early genetic diagnosis and counseling help patients to start therapy promptly.
✓ Integrated, multidiscip line rehabilitation enables functional independence despite serious genetic defects.
✓ Optimizing results depends on family involvement and continuity of care.

While long-term prognosis in BAGOS is still unknown, this example shows how goal-oriented and intense physiotherapy can produce clinically significant improvements. Standardized treatment routes for rare neurodevelopmental syndromes have to be developed via more longitudinal studies.

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