Case Report

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Effectiveness of Multidisciplinary Rehabilitation Program of a Child with Joubert Syndrome at Inclusive School in Bangladesh

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ABSTRACT

Background: Joubert syndrome (JS) is an extremely rare autosomal dominant gene disorder that causes developmental delay, hypotonia, ataxia, oculomotor apraxia, and brain malformation. The aim of this study was to assess the effectiveness of multidisciplinary rehabilitation programs for a child with JS at inclusive school settings.

Case Description: The reported case for the study was 9 years old boy recruited from an inclusive school of CRP diagnosed with pure JS. His gross motor function was at Level II according to the GMFCS.

Method and Materials: Purposive sampling technique was applied considering the inclusion and exclusion criteria. The Child and his mother gave consent and agreed to participate in this observational case report study for 2 years.

Intervention: The research was carried out between January 2021 and December 2022. The subject has participated in 2 years multidisciplinary rehabilitation program of physical therapy, speech therapy, occupational therapy, orthotics services, and special education. This study employed a repeated-measures design conducted using the Gross Motor Function Measure (GMFM), Pediatric Functional Independence Measure (WeeFIM), and Developmental Quotient (DQ). Result: The results after two years of evaluation demonstrated significant improvements in all the dimensions of the GMFM score. After 2 years evaluation WeeFIM scores have found 64 where baseline assessment scores were 29. All traits of DQ have observed favorable outcome after completion of 2 years rehabilitation program.

Conclusion: The study highlights the benefit of multidisciplinary rehabilitation team approach for better outcomes of JS in coping with developmental delay at inclusive school setting.

Keywords: Joubert Syndrome; Molar Tooth Sign; Rehabilitation Program; Inclusive Education

Opinion

Joubert syndrome (JS) is a rare autosomal dominant gene disorder that is heterogeneously inherited and manifests itself as hypotonia, ataxia, breathing abnormalities, developmental delay, intellectual disabilities, oculomotor apraxia, and brain malformation [1-3]. It is a multisystem syndrome that mostly impacts the neurological, urogenital, ophthalmic, and gastrointestinal systems in varied degrees. The disease first manifests during the course of pregnancy when the cerebral cortex starts to grow and develop [4]. Jobourt syndrome was originally recognized in 1961 by a French Neurologist named Dr. Marie Joubert [4-6]. The instances of JS around 1:80,000 and 1:100,000 according to reports [7-9]. There have been 200 instances of JS recorded globally [10]. The Joubert syndrome may be inherited or sporadic [11]. The majority of children with this syndrome survive to adulthood [12]. Common clinical manifestations of JS are lower muscle tone, ataxia, eye-related issues, facial dysmorphism, unusual respiratory issues, growth retardation, and a specific mid-hindbrain brain malformation [5,13-15]. The molar tooth sign (MTS) on brain axial magnetic resonance imaging (MRI) can be used to identify JS in addition to the clinical presentation [1,16]. The molar tooth sign is a radiographic hallmark which reveals unusually deep interpeduncular fossa, thick and horizontally arranged superior cerebellar peduncles, and cerebellar vermis hypoplasia or dysplasia [17-19]. The main purpose of this case report are to highlight the benefits of multidisciplinary rehabilitation team approach at inclusive education program.

Case Presentation

The 9 years old term boy delivered via cesarean section, weighed 2.8 kg and measured 48 cm tall. It was the mother's first pregnancy as well as delivery. The mother didn't have any significant illnesses or challenges during her pregnancy. The parents are not related in any kinship relation. Nobody else in the family bears this peculiarity. At the age of 8 months, the case was identified as having JS. The MRI impressions of the baby reveal enlarge superior cerebral peduncles showing molar tooth sign. Initially baseline assessment was done by multidisciplinary rehabilitation team when the case was admitted at inclusive school of CRP. The case was provided therapeutic rehabilitation program and inclusive education prior to Individual Education Plan (IEP) for achieving specific academic skill and knowledge. The main complaints were the inability to stand independently, walk freely, as well as weakness in both bilateral upper and lower limbs, with the lower limbs being more evident. At the age of 18 months, he mastered neck control; at the age of two years, he mastered rolling under supervision, and when he was three years old, he learned to sit with assistance. His delayed gross motor skill caused challenges with daily tasks and social interaction. At the age of 5 years, he was admitted to an inclusive school for improving interaction, communication, academic knowledge, and therapy interventions as inclusive ambient provides all supportive services for children with disabilities. After schooling here, his academic skills, communication, and physical profile especially gait pattern has improved through multidisciplinary rehabilitation approaches including special education, physical therapy, speech therapy, and occupational therapy services.

Investigation

The key results of JS from cranial MRI investigations are deepened interpeduncular fossa, hypoplasia of cerebellar vermis, thick and horizontal superior cerebellar peduncles become enlarged [19]. It is linked to faulty brain stems, which create neural pathways between the spinal cord and brain, as well as a lack of inadequate development of the cerebellar vermis, which regulates coordination and balance [4,20]. The MRI impression of this case reveals prominent thick elongated superior cerebellar peduncles (molar tooth sign) with prominent 4th ventricle, forming bats wing configuration. Bilateral 7th and 8th cranial nerve complexes are unremarkable.

Ethical Approval

A subject for this case study was recruited from an inclusive school. Approval was obtained from the selected school and from the parents' willingness to participate in this study. The Child and his mother agreed to participate in this observational case report study for 2 years.

Rehabilitation Strategies

There is no specific treatment for joubert syndrome. Therapy alternatives encompass physical therapy, occupational therapy, and speech therapy [4]. Treatment for JS consists primarily of supportive and symptomatic measures [21]. Special treatments are being used for the pulmonary and nutritional issues that arise in connection with breathing problems and hypotonia in the initial stages [22]. Early diagnosis of this problem is crucial in order to begin multidisciplinary action, which includes medical care and rehabilitation, as soon as feasible [2]. If addressed and treated early through a multidisciplinary intervention program that includes physical therapy, special education, occupational therapy, and speech therapy, then the prognosis of JS cases may be favorable. For numerous patients with JS, the early intervention has demonstrated notable advantages in promoting the achievement of developmental stages [23]. One of the key roles of occupational Therapist is children performing play skills, academic part, activities of daily living skills, balance control, developmental coordination, sensory integration that influence participation and engagement in the functional activities [24]. Rehabilitation approaches are also impacted by occupational therapist in behavioral and cognitive challenges additionally to visual impairment [22].

Physical rehabilitation through physiotherapy appears to be quite crucial in controlling symptoms of JS children. This method would help the child to perform the general daily tasks and enhance their quality of life. Brain experts, physical therapy, occupational therapy, speech therapy, nephrologists, neurologists, opticians, and stimulation therapy can all be used to treat developmental delays for newborn JS. The process of rehabilitation entails a number of therapies that follow the Bobath neuro developmental approach, motor training, postural approaches, different forms of balance, range of motion, various rehabilitation session, symptomatic treatment is given, and all of these techniques aid the patient in recovering and making easier life [10]. Physical therapy strategy comprised positioning, sensory integration therapies, and functional exercises to support the necessary stages of development. The incorporation of orthotic and assistive devices, like special chair, ankle foot orthosis (AFO), standing frame, and balancing board, as well as weight-bearing exercise and joint compression maneuvers, motor skill techniques, and stretching are also included in the list of exercises [2]. Speech and language therapist used oral-motor exercise and articulation therapy for improving oral movement and speech sound as well speech intelligibility [25]. Receptive language skill is achieved through following instruction of object identification and complex direction, where expressive language skill achieved through naming the daily object, body part, action verb define [26].

Discussion

The purpose of the study was to see the development of a child with JS over 2 years after receiving multidisciplinary therapeutic rehabilitation. The Prime goals were to identify the gross motor expertise, functional abilities, and developmental profile of a kid attending an inclusive school. The Gross Motor Function Measurement (GMFM) test was utilized to assess both the level of expertise and motor level of function of the case. The examination comprises 88 total items and 5 functional dimensions, which are arranged sequentially in the order of neurological development which include lying and rolling (17 items), sitting (20 items), crawling and kneeling (14 items), standing (13 items), and walking, running, and jumping (24 items). The overall score for each section is derived by multiplying the sum of the individual section's scores by 100, then dividing the result by the sum of those scores. The sum of all these 5 sections is divided by 5 to determine the final score [27,28]. The case has seen improvement after two years intervention in GMFM-88. The total baseline assessment score was 15.90%, after 1 year evaluation score was 36.36%, and after 2 years evaluation score was 65.16%. The improvement was seen in all dimensions of GMFM scores. In dimension Eat walking, running and jumping has observed massive improvement after 2 years evaluation compared to baseline assessment. In dimension B, differences were observed in sitting after 1- and 2-years interventions. In dimension D there was noticed marked changed in standing after 2 years evaluation score. In dimension C, crawling and kneeling has observed 52.38% improvements after 2 years evaluation where baseline assessment score was only 2.38%.

In dimension A, the lowest difference was recorded internally at lying and rolling as 39.21%, 54.90%, and 70.58% at baseline, after one-, and two-years intervention respectively (Table 1). The Pediatric Functional Independence Measure (WeeFIM) was used to assess the level of independent functioning of the case. The Functional Independence Measure (FIM), which was utilized as the assessment method for senior rehabilitation, served as the basis for the creation of the WeeFIM evaluation scale. The functional independent levels of children with cerebral disability along with various developmental delays are assessed using the WeeFIM scale, as well as alterations in time-related functions [29]. In 2001, Erkin and Aybay carried out the WeeFIM validity and reliability investigation in Turkey. Self-care, sphincter control, mobility-transfer, locomotion, communication, and cognitive function are the six components and 18 elements that make up the WeeFIM evaluation method. When the prescribed task is completed entirely with assistance, it obtains a score of 1, however when it is completed entirely autonomously, on schedule, and safely, it obtains a score of 7 [30]. Our case got 29 scores of Functional Independence Measure (WeeFIM) at baseline assessment. We have observed improvement respectfully 49 and 64 after one- and two-year's evaluation. All dimensions of WeeFIM have found significant outcome after 2 years evaluation. In dimension self-care got scores 8 at initial assessment were got scores 18 after 2 years evaluation. In dimension mobility-transfer got scores 3 at initial assessment were got scores 15 after 2 years evaluation. In dimension locomotion scores were 2 at initial assessment where after 2 years evaluation the scores were 10. The dimension of self-care, mobility-transfer, and locomotion has observed greater improvement than sphincter control, communication and cognitive function after 2 years evaluation (Table 2).

Table 1: Evaluation scores of Gross Motor Function Measure (GMFM).

Dimension	Initial Assessment	After 1 year	After 2 years
Lying and rolling	20	28	36
Sitting	16	32	49
Crawling and kneeling	1	8	22
Standing	2	9	24
Walking, running and jumping	3	19	41
Total GMFM	42 (15.90%)	96 (36.36%)	172 (65.16%)

Table 2: Evaluation scores of Functional Independence Measure (WeeFIM).

Dimension	Initial Assessment	After 1 year	After 2 years
Self-care	8	13	18
Sphincter control	3	7	9
Mobility-transfer	3	12	15
Locomotion	2	8	10
Communication	6	8	9
Cognitive function	7	9	12
WeeFIM Total	29	49	64

Development quotient (DQ) evaluates to determine whether the development velocity has been attained or not. The evaluation was measured based on the DQ, which was divided into four categories: gross motor, fine motor, language, and social DQ. Calculating an overall general DQ is as simple as dividing the participant's overall developmental age (DA) throughout domains by their chronological age (CA), then multiplying that result by 100 (DQ = DA/CA × 100). This study looked into how well DQ could be used to evaluate cognitive level of kids with JS [4]. In our case baseline assessment score was 13.88 at observations scores of DQ. After 1 year evaluation we found changed in observation score 22.21 of total DQ. It was seen significant improvement after 2 years observations scores of DQ. All traits of DQ have observed favorable outcome after completion of 2 years rehabilitation program. After 2 years evaluation total DQ scores has found 34.71 where baseline assessment scores was 13.88 (Table 3).

Table 3: Observations scores of Developmental Quotient (DQ).

DQ	Baseline assessment	After 1 year	After 2 years
Gross Motor	11.11	16.66	22.22
Fine Motor	11.11	22.22	38.88
Language	16.66	27.77	33.33
Social	16.66	22.22	44.44
Total DQ	13.88	22.21	34.71

It was determined by Mouna Saghir, et al. assumed that there is no curative treatment for this disease. Rehabilitation plans for joubert syndrome assist the individual in overcoming significant delays in comprehension. A few research points out that supportive and therapeutic interventions, such as physical therapy, speech therapy, genetic counseling, and specialized education, helpful for the patient's improvement [10]. In accordance to the neuro developmental principle, ooze epic, et al. (2017) had a physiotherapist deliver a physical therapy regimen for an hour, 5 days a week, for a total of 13 months. The finding showed a positive outcome where the individual was successfully able to execute some tasks independently [3]. Six days a week, the kid attended a 90-minute session of treatment that included physical therapy and speech therapy in addition. Genetic consulting has been suggested as one of the key steps to avoid JS. JS can be diagnosed prenatally at around 11 gestational weeks by doing sampling of the chorionic villus [1,31]. Anita Kumara et al. (2013) demonstrated that a 5 years old joubert syndrome child's ataxia decreased as a result of rehabilitation therapy that also assisted the child in regaining a normal walking pattern [10]. Shema Vipin Sharma et al. (2017) also discussed the importance of early diagnostic and genetic counseling for therapeutic goals as well as the rehabilitative program [32]. Mounisghir, et al. (2016) commenced a rehabilitation program combining muscle strengthening, stimulation therapy, occupational therapy, and speech therapy [10].

Conclusion

The rare autosomal recessive disorders cannot currently be treated, while prenatal screening and genetic counseling are readily accessible and available. To overcome dependency and milestones, early recognition, genetic consultation, different physiotherapy approaches, speech therapy, occupational therapy, and symptomatic treatment can be taken into consideration. We believe that multidisciplinary rehabilitation strategies will enhance typical motor function and growth children with JS. The study emphasizes the importance of multidisciplinary rehabilitation teams for more effective outcomes. This instance shows the significance of raising awareness of JS to enhance stages of development, balance, academic ability, and mobility with comprehensive care. The research indicates that multidisciplinary rehabilitation programs at inclusive school settings can be beneficial for dealing with the symptoms which causes delays in development for children with JS.

Consent for Publication

The patient's mother provided written, fully informed consent for the publication. The chief editor of this journal has a copy of consent for review.

Conflict of Interest

Authors disclose no conflict of interest relevant to this publication.

Fund

The manuscript was developed with researcher's own resource.

Availability of Data Support

The assessment and evaluation form are available of this case.

Author Contribution

All authors contributed to preparing write up, revising the article, and giving final approval of the version to be published. Authors consent to accept responsibility for all contents of the tasks.

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