CASE REPORT

Physical therapy rehabilitation of Cohen syndrome in Pakistan

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Abstract

Cohen Syndrome is a rare genetic disorder. It is caused by the mutation of VPS13b gene which is present on chromosome number 8. It was first described in 1973. Here is a case report of a male child who presented to Paediatric Physical Therapy and Neuro Rehabilitation Department of The University of Lahore Teaching Hospital, Lahore, on July 25, 2021, with developmental delay due to hypotonia, typical facial gestalt, neutropenia, intellectual disability and speech delay. His genetic testing confirmed the diagnosis of Cohen syndrome. He received an intensive and holistic physical therapy programme for 3 years (July 2021 till July 2024) which helped him reach his developmental milestones. This study shows that with efficient goal setting, early intervention along with enriched environment and family centred approach can help the child achieve age-appropriate milestones.

Keywords: Cohen syndrome, Physical therapy, Genetic diseases.

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Introduction

Cohen syndrome is a rare autosomal recessive genetic disorder caused by mutation in chromosome 8 at VPS13B gene. The distinct features of this syndrome varies from patient to patient. They may include distinct facial features, truncal obesity, hypotonia, delayed developmental milestones, small head circumference, increased flexibility of joints, abnormal social interactions, intellectual disability, speech delays, cognitive delays, while the child may fall on autism spectrum disorder. This condition can also result in progressive myopia (near-sightedness); 90% of the children develop severe retinal degeneration which starts at an early age.[1,2] Children develop frequent infections due to associated neutropenia. Facial features include open mouth, large ears, thick eyelashes and eyebrows, low hairline, prominent upper central incisors, prominent root of nose, long slender fingers (Figure.1 B,C,D).[3] There is no specific diagnostic criteria of Cohen Syndrome clinically. However, literature review shows that having at least three major criteria (intellectual disability, slender hands and feet, decreased muscle tone, short stature, microcephaly, retinal dystrophy) and one minor criteria (neutropenia, myopia, obesity, dimorphic facial features) can be used to establish a diagnosis.[4]

This study presents a diagnosed case of Cohen syndrome in which early intervention along with family centred approach helped the child to achieve age appropriate milestones.

Case Report

An eight-month-old baby's parents brought him to Paediatric Physical Therapy and Neuro Rehabilitation Department of The University of Lahore Teaching Hospital, Lahore, on July 25, 2021, with the complaint of delayed milestones. The child was unable to hold his neck, perform rolling, or sit even with support. History revealed that the parents were first cousins. After an uneventful pregnancy, the baby was born at full term via C-section due to breech position. His weight at the time of birth was 2.7 kg with occipito-frontal circumference of 37cm. There was no history of delayed cry, seizures, or high grade fever. However, he did have a history of bilateral inguinal herniation at the time of birth which was corrected by B/L herniotomy at two months of age. Systemic reviews revealed low level of vitamin D. His MRI findings were unremarkable; however, EEG revealed focal sharp waves. He suffered from frequent chest infections due to intermittent neutropenia (neutrophils count 0.68×10e 3/µL) while normal values ranges from 2,500 to 7,000/µL. On genetic testing, a homozygous pathogenic variant was identified with VSP13B gene which was consistent with the genetic diagnosis of autosomal recessive Cohen syndrome.

On examination, it was noted that the child had generalised hypotonia with poor neck holding in prone and supine positions and was unable to perform rolling and sitting. His grasp was weak. His attending skills were not age-appropriate (poor focussing on presented objects, poor eye contact, and no recognition of his parents). He had delayed speech—no babbling or any means of non-verbal communication, making it global developmental delay.

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Table-1: Pre and Post Therapy Readings.

Dimensions	Baseline (at 8 months) [%]	After Therapy (at 3 years) [%]
Lying & Rolling	39.21	100
Sitting	10	91.66
Crawling & Kneeling	0	78.57
Standing	0	56.41
Walking and Jumping	0	29.16
Total	9.84	71.16

A holistic physical therapy treatment approach was designed for the child according to the ICF model, including sensorimotor therapy, family-centred approach, targetted and focussed therapeutic playful activities, and hydrotherapy and occupational therapy.[5] His sessions were performed for three hours intermittently (one hour clinic based therapy and two hours home-based therapy) per day, six days a week carried on for three years.

Extensive family counselling was done regarding the importance of early intervention and enriched environment. They were instructed to carry out a home programme which included full body massage and texture exposure on body through brushes, textured balls, playing on grass, in his oral cavity via finger brush, and different textures of food like crunchy food. GMFM-88 was used to record the readings at baseline and after two and a half years of therapy (Table-1).[6] Significant improvement was seen in gross motor function of the child. He achieved his independent sitting with good manipulation and exploration of toys at 15 months of age. By three years of age, he was able to walk independently.

Discussion

Cohen syndrome is a rare autosomal recessive disorder. It is challenging to diagnose clinically at early ages due to its diverse and overlapping clinical manifestations.[7] As a result of its variable presentation, limited literature is available on the management of Cohen syndrome through physiotherapy. Given the unique manifestations in each child, individualised treatment programmes are crucial for achieving optimal developmental outcomes.

The heightened plasticity of the neonatal brain, as emphasised in neurodevelopmental research, allows for significant improvement in motor, cognitive, and behavioural functions when exposed to targetted sensory and motor inputs.[8] In this case of Cohen syndrome, early physical therapy facilitated remarkable developmental progress, with the child achieving independent sitting by 15 months and walking by three years, aligned with findings that highlight the critical role of early intervention (Figure.1 A).

Research consistently emphasises that an enriched

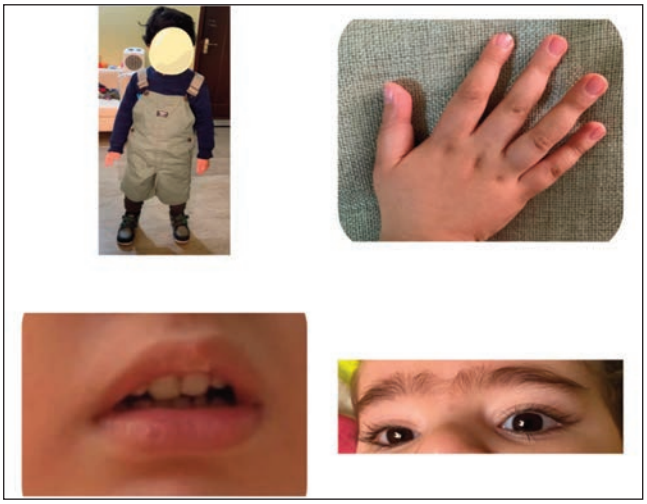

Figure-1: A: Independent standing achieved after intervention. B: Long slender fingers. C: Open mouth appearance D: Thick eyebrows and eyelashes.

environment, combined with family-centred care, plays a vital role in improving developmental outcomes. Enriched environments, such as those involving sensory integration therapy (SIT), provide varied sensory stimuli—tactile, visual, and auditory—that stimulate neuroplasticity and promote cognitive and motor improvements, particularly effective when applied early in children with neurodevelopmental disorders (NDDs).[9] This study integrated such principles, using varied sensory stimuli within a family-centred care model. Daily parental feedback through logbooks and videos played a crucial role in creating a consistent and stimulating therapy programme.

The holistic approach, involving SIT, which emphasises both sensory and motor development, reinforces the importance of tailoring interventions to meet individual needs.[10] Early and intensive therapy, as highlighted in the literature, is essential for promoting long-term developmental progress in children with Cohen syndrome, enabling them to reach key milestones, such as walking independently, often by the age of five.[4] The present case also focussed on this holistic approach. The child received an intensive physical therapy programme designed to address delayed milestones and prevent secondary complications in clinic and home environment.

This discussion highlights the need to capitalise on the critical early years of neuroplasticity through individualised, enriched, and family-centred interventions. These strategies hold promise for improving developmental trajectories and overall quality of life for children with Cohen syndrome, contributing valuable insights to the limited body of research on this rare genetic disorder.

Conclusion

It is concluded that early intervention, holistic approach,

enriched environment, and targetted and focussed therapy of genetic syndromes result in early improvement.

Consent: Parents' consent was obtained prior to publishing the case report.

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Conflict of Interest: None.

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