CASE REPORT

Improvement in Neurocognitive Manifestations with Short-term Multidisciplinary Intervention in DiGeorge Syndrome

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Background: DiGeorge syndrome involves deletion of chromosomal region 22q11.2. **Case characteristics**: 3-year-old girl presenting with speech delay showed defiant behaviour and sensory concerns. **Outcome**: Multidisciplinary intervention with parental counselling improved communication and social skills. **Message**: Cognitive and behavioral issues in DiGeorge syndrome should be addressed through timely, multidisciplinary intervention.

Keywords: *DiGeorge Syndrome, Management, Neurocognitive dysfunction*.

DiGeorge syndrome includes developmental defects in the III and IV pharyngeal pouches, aplasia or hypoplasia of thymus and parathyroid, and cardiac outflow tract defects [1]. Scarce research exists on neurocognitive manifestations in children having DiGeorge syndrome.

This report focuses on neuro-cognitive manifestations in a child with DiGeorge syndrome. Parents reported with delayed speech, at three years and three months of age *i.e.* after the critical period of language development. This reflects the general pattern of management in such patients where acute medical problems (*e.g.*, heart disease, immune disorders, feeding problems) are focused in infancy and preschool years; while cognitive, behavioral and learning disorders are focused during school years [2].

Case Report

A 3-year 3-month-old girl, diagnosed with DiGeorge Syndrome in early infancy, reported to a multidisciplinary child development center.

The child was born of a non-consanguineous marriage. The congenital cardiac defect and palate-pharyngeal defect were identified in early infancy and Fluorescent in-situ Hybridisation (FISH) showed chromosomal deletion 22q.11.2 at 1.5 months of age. She presented with delayed speech and language. Mother reported frequent behavioural concerns including hyper-activity, restlessness, stubbornness and aggression (*e.g.* screaming in anger, throwing objects or hitting others) and sensory concerns (*e.g.* discomfort during head bath, seeking movement all the time).

Audiogram and ophthalmic evaluation results were normal. She underwent a comprehensive assessment by a developmental pediatrician, psychologist, occupational therapist and speech therapist. On Vineland Social Maturity Scale [3], she obtained a social quotient of 74, indicating borderline social functioning. Areas of concern included self-help eating and self-help dressing. According to fifth edition of the Diagnostic and Statistical Manual of Mental Disorders [4], she met the diagnostic criteria for Attention Deficit/Hyperactivity Disorder of moderate severity with speech delay. On Receptive Expressive Emergent Language Scale [5], she obtained a score of 20-22 months (receptive language) and 11-12 months (expressive language). In terms of clinical impression, comprehension and pragmatic skills were fairly developed (*i.e.* facial expressions, eye contact, social smile and play skills). On occupational therapy assessment, sitting tolerance, attention span and frustration tolerance was low. Visual-perceptual skills (*e.g.* color and shape recognition) and cognitive skills were underdeveloped. Vestibular seeking behaviour was observed. She needed assistance in performing activities of daily living.

Based on the assessments, a multidisciplinary, individualized, goal-oriented intervention program was implemented for a period of six months. Occupational therapy (including sensory integration) and speech therapy were provided along with monthly parental counselling sessions. After undergoing 16 weeks of intervention and consistent follow-up of therapy at home, she showed marked improvement in behavior and communication skills (*e.g.*, she was able to say 15-20

meaningful words) and behavioral concerns such as defiance, temper tantrums and aggression markedly decreased.

DISCUSSION

The child was referred at the age of 3.25 years, reflecting loss of critical period for language development. She inadequately compensated with non-verbal communication. Deficits in verbal communication could have exacerbated the existing behavioral difficulties.

Despite the magnitude of developmental delay and behavioral problems, we demonstrated positive outcomes after a short duration of intervention. The intervention was developed for a period of six months and within first three months, the child showed improvement across domains such as communication, personal-social skills and behaviour.

Pre-school children with DiGeorge sydrome commonly have developmental delays, mild hypotonia, and language and speech delays [6]. However, given the limited awareness in the Indian context, it is important to inform caregivers on neurocognitive manifestations and risk of developmental delay in children having this syndrome. This will improve care-seeking and ensure early intervention and mitigation of difficulties due to developmental delay.

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