



Effects of Sensory Integration Therapy on the child with Kabuki Syndrome: A Case Study

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ABSTRACT

Purpose: This case study describes the outcomes of a child with kabuki syndrome using sensory integration therapy.

Method: A 4.6 -year-old boy diagnosed with kabuki syndrome was assessed and given sensory integration activities for 2 months, 5 sessions in a week.

Results: After the Therapy sessions, the child showed improvement in all the areas like cognitive skills, socialization, gross motor & fine motor skills and ADL skills.

Conclusions and Implication: The collaborative efforts of an interdisciplinary team, comprising an occupational therapy, speech pathologist, special educator, parents of the child made it possible to enhance the child's participation and improvement in all the areas.

Key words: Kabuki Syndrome, Sensory Integration Therapy.

INTRODUCTION:

Kabuki syndrome (KS) was first described in the early 1980s in Japan. It was named after the characteristic facial features which resemble the makeup of actors in the Kabuki theatre in Tokyo (Snir Boniel et al, 2021). KS is characterized by typical facial features (long palpebral fissures with eversion of the lateral third of the lower eyelid; arched and broad eyebrows; short columella with depressed nasal tip; large, prominent, or cupped ears), minor skeletal anomalies, and persistence of fetal fingertip pads, mild-to-moderate intellectual disability, and postnatal growth deficiency. Other findings may include: congenital heart defects, genitourinary anomalies, cleft lip and/or palate, gastrointestinal anomalies including anal atresia, ptosis and strabismus, and widely spaced teeth and hypodontia. Functional differences can include: increased susceptibility to infections and autoimmune disorders, seizures, endocrinologic abnormalities (including isolated premature thelarche in females), feeding problems, and hearing loss. (Margaret P Adam et al, 2019).

Loss-of-function variants associated with Kabuki syndrome (KS) lead to the dysregulation of the differentiation of neuronal and myogenic cells. Mechanistically, this may lead to several neurological abnormalities in KS patients. These include infantile hypotonia, developmental delay and intellectual disability, epilepsy, behavioural abnormalities including autistic-like features, and CNS malformations. Neuropsychological symptoms have more commonly been observed in patients with KMT2D variants (Chong-Kun Cheon & Jung Min KO, 2015)

It is important to note that unintelligible or disordered speech originating in oral motor hypotonia has consistently been reported throughout the literature and is commonly observed by the parents and other caretakers (Kristiane M Van Lierde et al, 2000).

Several studies describe neurobehavioral findings in the context of KS. These may include Learning disabilities with reduced IQ, adaptive skill impairment, autistic-like behaviour and psychiatric pathologies, such as anxiety disorder, phobias, bizarre behaviour and impaired emotional control. It has been suggested that developmental delay is more pronounced in patients with KMT2D gene deletions or variants causing a dysfunction of synthesis in the first half of the KMT2D protein. However, no specific genotype-phenotype correlations could be drawn (Francesca Romana Lepri et al, 2018). Varying degrees of developmental delay and speech disability are present in all individuals with KS.

Psychomotor retardation or intellectual disability (mean intelligence quotient of 35–69) occurs in all children with KS. Children with KS tend to have a particularly deficient visual memory and visual-spatial coordination (LCM van Dongen et al, 2019).

Sensory Integration is a theory developed by A. Jean Ayres, an occupational therapist with advanced training in neuroscience and educational psychology. Ayres (1972) defines sensory integration as "the neurological process that organizes sensation from one's own body and from the environment and makes it possible to use the body effectively within the environment". The theory is used to explain the relationship between the brain and behaviour and explains why individuals respond in a certain way to sensory input and how it affects behaviour.

Occupational therapy with a sensory integration approach is designed to guide intervention for children who have significant difficulty processing sensory information, which restricts participation in daily life activities. The theory of sensory integration was developed by A. Jean Ayres [Ayres, 1972, 1979, 1989], an occupational therapist with postdoctoral training in educational psychology and neuroscience. Guided by her roots in the clinical field of occupational therapy (OT), Ayres developed the theory of sensory integration to explicate potential relationships between the neural processes of receiving, modulating, and integrating sensory input and the resulting output: adaptive behaviour. The theory postulates that adequate processing and integration of sensory information is an important substrate for adaptive behaviour. Given its focus on adaptive behaviour and functional skills, this approach is most frequently utilized by occupational therapists as part of a total program of occupational therapy. The goal of intervention is to improve the ability to process and integrate sensory information and to provide a basis for improved independence and participation in daily life activities, play, and school tasks. (Roseann C Schaaf & Lucy Jane Miller, 2005). Yee-Pay Wuang et.al (2020) in their study concluded that school-aged with ASD had impaired sensory and perceptual-motor performances, and these impairments were significantly related to their activity participations. While emphasizing the development of functional skills to facilitate age-appropriate activity participation in multiple scenarios, interventions aiming to improve body functions are indispensable.

KS is a recently identified genetic syndrome that is increasingly becoming recognised in primary care settings. Clinicians should be prepared to provide interventions informed by research that have addressed these types of symptoms for other childhood disorders. Effective therapy is likely to include finding appropriate resources and support for the parents, where little resources are available. Both the child and family also benefit from the development of a collaborative treatment team of Doctors, Teachers, Physical,

Occupational and Speech Therapists and working collectively for the child and family. Most importantly, practitioners who encounter this diagnosis should expect to be flexible and creative in finding effective interventions where none currently exists (Bethany D Kasdon & Judith E Fox, 2012)

This case study describes the outcomes of a child that utilised sensory integration as a means of enhancing functional outcome in a young boy with Kabuki syndrome (KS).

METHOD:

Case summary:

A 4.6 years old boy diagnosed with Kabuki syndrome (KS) was referred to CRC (COMPOSITE REGIONAL CENTER FOR SKILL DEVELOPMENT, REHABILITATION AND EMPOWERMENT OF PERSON WITH DISABILITIES) NELLORE. The child was brought by his parents who reported that he had inadequate speech output, drooling, weakness, behavioural issues, not following commands, not sitting in one place. Prenatal history reveals mother went regular check-ups. Rh incompatibility present. History of hypertension present at 7th month of gestation. Taken medications as per doctor advice. Perinatal history revealed the child was born with c section delivery. Birth weight was 3.5 kg. Hyperbilirubimia present and the child was under observation for 24 hours. Post natal history revealed that the child had seizures at seventh month & the child was in incubator for 2 days.

Pre therapy evaluations:

To evaluate the current level the detailed occupational therapy assessment was carried out. The occupational therapy assessment reveals that the child has generalised weakness, poor eye contact & name call response , poor gross motor skills(not able to jump in the trampoline), poor dynamic balance, sensory issues ,poor fine motor skills, poor cognitive perceptual skills, poor socialization skills (no interaction with his brother & peer groups), not playing with the peer group, no toilet indication , Dependent on his mother for all ADL skills & not cooperative for the ADL skills.

Therapy Details:

The child attended the regular occupational therapy, speech therapy for 5 sessions per week, OT sessions for 45 minutes duration & speech therapy for 30 minutes duration. Counselling was done for the parents. Mother was inside the therapy room. Occupational

Therapy sessions started with free play for identifying the child's likes and dislikes. Initially the child was screaming and not cooperative for any activity. Gradually with the help of mother sensory stimulation activities was started (tactile stimulation, visual stimulation, auditory stimulation vestibular & proprioceptive activities like jumping in the trampoline, playing in the swing). After some days slowly some more sensory integration activities were started, peg board activities, balance board activities & cognitive skills training also given. After every session, home-training activities were explained to the mother.

Post therapy assessment:

Occupational Therapy assessment done for the child after 2 months of therapy.

RESULTS AND DISCUSSION

After 2 months of Occupational Therapy sessions the following changes was observed in the child. The child starts showing interest to the therapy sessions. He maintained eye contact, developed social smile, trying to communicate sometimes with sound & sometimes non verbally by pointing out. Able to jump in the trampoline without support, dynamic balance was improved. He started playing with his brother alone. Following simple command, his sensory issues was reduced, sitting tolerance improved (Able to complete the task) & cooperative for the ADL skills.

The Sensory Integration Therapy (SIT) approach attempts to facilitate normal development and improve the child's ability to process and integrate sensory information (visual, perceptual, proprioceptive, and auditory, etc.). One important aspect of choosing the SIT approach is that the motivation of the child plays a crucial role in the selection of the activities. Most children tend to seek out activities that provide sensory experiences which will be beneficial to them at that point in their development. It is this active involvement and exploration that enables the child to become a more mature, efficient organizer of sensory information (Linda & JoAnn 1996).

Robert M. Kantner et al (1976) have shown that effectiveness of the vestibular stimulation training has a positive effect on motor skills in children with Down syndrome. Kelly G (1989) described the effects of rotational vestibular stimulation to increase gross motor co-ordination in children and adults. He showed that rotational vestibular stimulation was effective in increasing reflex integration, balance, intellectual functions, perception– motor skills, and hearing–language and socio emotional development(Mine uyangk et al, 2003).

AR Shamsoddini & MT Hollisaz (2009) in their study concluded that Sensory Integration therapy intervention had a significantly positive effect on gross motor function in the children with Diplegic spastic CP. Sajad Haghshenas et al (2014) in their studies with ADHD children concluded that the vestibular stimulation can increase auditory comprehension. Vestibular stimulation gradually calms children by provoking uniformity in the autonomic nervous system, leading to a significant impact on children's hyperactivity in therapy sessions (Kantner R & Tocco AM, 1980).

CONCLUSION

This case study shows that the child exhibited improvement in all the areas. Hence sensory integration therapy can be used. Evidence based practice in terms of documenting is essential as this paves the way to successful counselling and greater acceptability among families. Also an interdisciplinary team comprising occupational therapist, speech therapist and the parents' efforts should collaborate to achieve progress. Future research into intervention and treatment of K S is crucial in understanding how we can be most beneficial to this growing population. Most important, however, is the need for other clinicians to share their experiences in treating children with this diagnosis.

Limitations:

The study was limited to only one child. It is further limited that the basic assessment only done and therapy continued based on the assessment. Not used standardized scales to monitor the child progress. The progress is subjective post therapy assessment & parental feedback.

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