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Original Research Paper

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DEVELOPMENTAL AND COMMUNICATION PROFILE OF A CHILD WITH WOLF-HIRSCHHORN SYNDROME-A SINGLE CASE STUDY

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ABSTRACT Wolf-Hirschhorn syndrome (WHS) is a rare genetic condition caused by deleted or missing genetic material on the distal portion of the short arm of 4th chromosome (4p16.3), also known as 4P syndrome. WHS is characterized by distinctive craniofacial features, delayed growth and development, intellectual disability and seizures. The distinctive facial features called "Greek helmet facies" includes microcephaly, broad nasal bridge, large and protruding eyes, and a high forehead. The lack of standardized treatment protocols, limited literature and complex needs of these children, it is essential to document and analyse individual cases in depth. Hence a single case study helps speech-language pathologists in understanding the clinical features, communication profile, and management of such children. The findings from this single case study may aid in early identification and intervention planning to the unique needs of individuals with WHS

KEYWORDS : Wolf Hirschhorn syndrome, intellectual disability, seizures, developmental delay

INTRODUCTION

Wolf-Hirschhorn syndrome (WHS) is a rare genetic condition caused by deleted or missing genetic material on the distal portion of the short arm of 4th chromosome (4p16.3), also known as 4P syndrome. WHS is characterized by distinctive craniofacial features, delayed growth and development, intellectual disability and seizures. The distinctive facial features called "Greek helmet facies" includes microcephaly, broad nasal bridge, large and protruding eyes, and a high forehead. Other features include a downturned mouth, small chin and jaw, short philtrum, malformed ears, cleft lip and palate.

Children shows slow growth which begins before birth, and infants struggle with feeding and gaining weight (failure to thrive). Poor nutritional intake leads to generalized weakness, under developed muscles and poor muscle tone. Motor milestones a significantly delayed. Most children and adults with this disorder also have short stature. Wolf-Hirschhorn syndrome can also cause abnormalities of skin, congenital heart defects, vision & hearing impairment, dental anomalies, respiratory tract & ear infections, urinary tract malformations and scoliosis

Intellectual disability ranges from mild to severe in people with Wolf-Hirschhorn syndrome. Compared to people with other forms of intellectual disability, they have better socialization skills with weaker language and verbal communication skills. Most of the affected children also have seizures, which may be resistant to treatment.

NEED OF THE STUDY

As speech language pathologists, we come across many children with neurodevelopmental impairments, many of which significantly impacts speech and language development. Wolf-Hirschhorn Syndrome is a rare multiple congenital anomalies/ intellectual disability syndrome. It presents a broad range of abnormalities including developmental, neurological, craniofacial and feeding related issues.

The lack of standardized treatment protocols, limited literature and complex needs of these children, it is essential to document and analyse individual cases in depth. Hence a single case study helps speech-language pathologists in understanding the clinical features, communication profile, and management of such children. The findings from this study may aid in early identification and intervention planning to the unique needs of individuals with WHS.

AIM & OBJECTIVES:

The aim of the current study is to understand more about wolf-hirschhorn syndrome and describe the developmental and communication profile. It also highlights clinical features, communication challenges, importance of early identification, and need of multidisciplinary management.

METHOD

A case of a 7-year-old boy brought to the department of speech and hearing with the complaint of limited verbal output. Information was collected by directly interviewing the mother. Birth history revealed that the mother had an abortion by 1.5 months of gestation due to bleeding. No significant prenatal history reported. Perinatal history revealed that the child was a full-term baby with a weight of 1.600 kgs and kept in NICU for 16 days. The child was formula fed due to difficulty in sucking. Medical history reveals that the child has been having seizures from the age of 1 year and he is still under medication. MRI report shows bilaterally enlarged lateral ventricle, delayed myelination and thinned out body of corpus callosum which was done at the age of 10 months. Genetic test conducted at 1 year of age and confirmed the diagnosis of Wolf- Hirschhorn syndrome. The child shows very slow weight gaining with a current weight of 11kg. Developmental delays like motor milestones and speech and language milestones are reported. At Six years the child had undergone surgery for ankyloglossia. The Psychological evaluation shows moderate impairment. The child has been subjected to informal and various formal speech and language assessment procedures such as Receptive Expressive Emergent Language scale (REELS), Assessment of Language development (ALD), Speech and language development chart (SLDC), Three Dimensional Language Acquisition test (3D-LAT), Communication DEALL (Developmental Eclectic Approach to Language Learning) Checklist, Oral Peripheral Mechanism examination (OPME), recommended for speech and language intervention.

RESULTS AND DISCUSSION

Results shows that the child was cooperative during the assessment process. Prelinguistic skills such as eye-to-eye contact, attention span and concentration, sitting tolerance, object permanence etc found to be fair. Receptive language

skills revealed that the child can understand basic lexical items, identify family members, follows simple one-step commands, responds to Yes/No questions through nodding, and understands the emotions of people, Cognitive prerequisites such as the use of objects of few items, object Permanence are present. As reported, she responds to name calls. The child communicates through pointing, pulls the parent to the desired item and has an expressive vocabulary of 5 to 8 words which is consistent and meaningful. Speech and language stimulation at home was found to be good and parental awareness about the child's condition was limited as reported. Delay in Receptive language skills was observed to be 5 years and in Expressive language, the delay was noticed for almost 6 years. The child had the history of repaired ankyloglossia. Poor oral-motor skills (based on OPME) Intelligibility is severely reduced. Fair pragmatic skills, uses gestures, negations, protests, social smiles are present, expresses emotions and initiates some social interactions. Self and parallel play is present, able to scribble. The child still has persistent feeding issues with difficulty in chewing hard solids and drinking water from the cup.

Table:1 Administration Details Of Different Test Materials

REELS	ALD	3DLAT
Receptive Language Age:23-27 months Expressive Language Age:13-22 months	Receptive Language Age:12-18 months Expressive Language Age: 6-12 months	Receptive Language Age:21-23 months Expressive Language Age:12-14 months Cognition:15-17months

SLDC	COM-DEAL
Phonology:1-1.6yrs Semantic:1.6-2yrs Syntax:1-1.6yrs Play:2-2.6yrs Pragmtics:1-1.6yrs	Gross motor:36-40 months Fine motor:24-30months ADL:24-30 months Reception:24-30 months Expression:12-18months Cognition:18-24 months Social skills: 24-30 months Emotional skills:24-30 months

The above findings align with previously reported clinical features of WHS, including low birth weight, seizure disorder, global developmental delay, and craniofacial anomalies (Battaglia et al., 2008). MRI findings of a thinned-out body of corpus callosum, enlarged bilateral lateral ventricle and delayed myelination further support the neurological basis of the observed deficits.

Motor and speech-language milestones were markedly delayed, consistent with literature noting significant global developmental impairments in WHS (Zollino et al., 2015). Feeding difficulties and early failure to thrive are hallmark features of WHS, and in this case, the child was formula-fed due to poor sucking and had persistent chewing and swallowing difficulties. These early feeding issues may have contributed to the overall delay in motor and oral-motor development.

Assessment revealed receptive language delay of approximately five years and expressive language delay of six years. Such disproportion between receptive and expressive skills has been highlighted in WHS, where children often exhibit relatively stronger comprehension compared to verbal expression (Battaglia et al., 2008). The child demonstrated a limited expressive vocabulary (5-8 consistent words) and depended on nonverbal communication such as pointing, pulling caregivers, and gestures.

Socializing and pragmatic skills appeared relatively better preserved. The child was able to request objects, protest, greet, and initiate social interactions through gestures or minimal verbalizations. This observation parallels previous reports indicating that individuals with WHS often demonstrate stronger socialization skills compared to their expressive language abilities (Battaglia et al., 2001).

Play skills were consistent with solitary and parallel play, with emerging abilities in group play. Symbolic play, a key cognitive and linguistic milestone, was limited, reflecting underlying cognitive challenges.

The case highlights the need for early identification and intervention in WHS to optimize developmental and communication outcomes. Speech-language pathologists (SLPs) play a central role, not only in therapy but also in training to use augmentative and alternative communication (AAC) strategies, given the severe expressive delay. Early feeding and oral-motor interventions are also critical due to persistent swallowing and articulation difficulties.

SUMMARY & CONCLUSION

WHS presents complex variable features requiring individualized evaluation. Early identification of speech-language delays is critical. Speech-language pathologists play a key role in improving quality of life and communication outcomes. This case study emphasizes the importance of developmental screening in rare syndromes. As speech-language pathologist we should be aware of the speech and language characteristics of a child with WHS which helps in assessment and intervention and also in interacting with other professionals in a multi-disciplinary team management.

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