Navigating speech and language complexity: A case study of a child with multifaceted health conditions.

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ABSTRACT

Background: Klinefelter Syndrome (KS) is a chromosomal disorder characterized by the presence of one or more extra X chromosomes, affecting males and often associated with language deficits, seizures, and increased risk of stroke. This case report examines a 2.8-year-old male, who presents with multiple health challenges, including KS, post-infectious chickenpox vasculopathy, stroke, and seizures. The child initially displayed inadequate speech and language skills and was diagnosed with right hemiparesis and right upper motor neuron facial palsy, secondary to a stroke caused by post-infectious chickenpox vasculopathy. Subsequent genetic testing confirmed KS, illustrating the combined influence of genetic and acquired factors on his development. **Case procedure**: Linguistic assessments revealed severe regression in both expressive and receptive language abilities following the stroke. Before the stroke, the child had typical early language milestones, but post-stroke, he exhibited significant delays, particularly in expressive language. This regression underscores the effect of stroke on language in children with underlying genetic conditions like KS, necessitating early, multidisciplinary intervention through speech-language therapy and physiotherapy. **Conclusion:** The case emphasizes the need for comprehensive neurorehabilitation such as speech-language therapy, physiotherapy, and continuous neurological monitoring to maximize recovery and support for the family, including counselling, education, and access to resources. Integrated care is essential to manage the psychological and practical challenges in caring for a child with complex health conditions. Further research on speech and language, and early stroke effects in children with KS is recommended to inform interventions that improve developmental outcomes and family well-being.

KEYWORDS- Klinefelter Syndromes, Stroke, Acquired Childhood Aphasia, Acquired Language Disorder.

INTRODUCTION

Klinefelter Syndrome (KS), a condition affecting males, involves the presence of one or more extra X chromosomes and occurs in approximately 1 in 500 to 1,000 males. Most KS cases are identified during puberty or adulthood (1,2). This disorder is often associated with generalized tonic-clonic seizures triggered by low-grade fever. Boys with 47XXY typically exhibit speech and language delays, mild hypotonia, motor delays, academic difficulties, and behavioral issues (3).

Chromosomal abnormalities, such as KS, have been associated with ischemic stroke and seizures (4–6). High fever, a common symptom in patients with chickenpox, can persist for up to 10 days (7). Gray (8) reported that chickenpox (varicella) may also lead to seizures. Additionally, a brain lesion, such as a stroke, can result in childhood aphasia, where a child with acquired childhood aphasia (ACA) may experience a complete or partial loss of language (9,10).

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Received on- 16th October 2024
Published on- 24th February 2025

In this study, we will discuss a child with multifaceted health conditions, including high fever, chickenpox, seizures, stroke, and KS, which have collectively contributed to speech and language problems. Written informed consent for publication was obtained from the parents. The Central Ethics Committee of the academy approved this study.

CASE REPORT

A 2.8-year-old male child, from a Telugu-speaking background, was brought to our Department of Speech Pathology and Audiology by his mother, with concerns about inadequate speech and language skills and inattentiveness for his age. He was born to a non-consanguineous couple and has two clinically normal older sisters. His father had a history of stroke at the age of 4 or 5, although the reports are unavailable. The antenatal history revealed that the child's mother had a miscarriage five years before his birth, with the cause remaining unknown. The child was born via normal vaginal delivery, cried immediately after birth, and had a birth weight of 2.9 kg. He also has a significant medical history, which will be discussed in the following section.

MEDICAL HISTORY

At 1.2 years old, the child initially presented with high fever and skin rashes, which were diagnosed as chickenpox. Four months later, he experienced a sudden onset of weakness in the right upper and lower limbs and was diagnosed with right hemiparesis with right upper motor neuron facial palsy secondary to post-infectious chickenpox vasculopathy (?) inherited vasculopathy. During a subsequent blood investigation, he had two episodes of generalized tonic-clonic seizures and was admitted to intensive childcare for further evaluation.

A series of radiological investigations were conducted. A Computerized tomographic scan on November 11, 2022, indicated an acute infarct in the left fronto-temporo-parietal regions and basal ganglia within the middle cerebral artery territory, with a midline shift to the right side of approximately 6 mm and no signs of haemorrhagic transformation. A magnetic resonance imaging (MRI) performed on the same day revealed a chronic global left middle cerebral artery territorial infarct, accompanied by gliosis and encephalomalacia, involving the left frontal, parietal, and temporal lobes. The left basal ganglia and thalamus appeared atrophic, likely due to vascular affliction and secondary neuronal degeneration, respectively. Additionally, the MRI showed evidence of Wallerian degeneration along the left corticospinal tract with atrophy of the left hemi-brainstem.

On November 12, 2022, a time-of-flight magnetic resonance angiography was conducted, revealing focal high-grade narrowing in the M1 segment of the left middle cerebral artery. An electroencephalogram (EEG) performed on November 21, 2022, showed abnormal activity in the left cerebral hemisphere with low voltage signals. Steroids were prescribed, and the child was referred for physiotherapy. During a follow-up visit on April 25, 2023, he was diagnosed with global developmental delay (motor > cognition) with right hemiparesis and right upper motor neuron facial palsy, secondary to post-infectious chickenpox vasculopathy. Given the family history, the child was referred for genetic evaluation, which revealed the presence of an XXY sex chromosome, confirming Klinefelter syndrome and an inherited vasculopathy. To assess his linguistic abilities, the Receptive and Expressive Emergent Language Scale (11) was administered on November 11, 2022, revealing a receptive language age (RLA) of 12-14 months and an expressive language age (ELA) of 10-11 months.

The lab investigations and other tests mentioned in the medical history were conducted across various hospitals. The linguistic profiling of the child was conducted in our department on March 13, 2024. He was able to comprehend simple instructions, identify common objects and animals, respond to basic 'what' and 'where' questions, and select items from a group of five or more varied items. He expressed his needs through vocalization accompanied by pointing to the desired item, used jargon during play, and would say /amma/ (mother) when needing attention or assistance. The Assessment Checklist for Speech and Language Skills (12) was administered, revealing a receptive language age (RLA) of 1.7-1.9 years and an expressive language age (ELA) of 0.10-1.0 years. The child did not exhibit any comorbid conditions such as apraxia, dysarthria or dysphagia. Based on these results, the initial clinical impression was Spoken Language Disorder. However, there was uncertainty about whether the condition was secondary to stroke due to limited information from the initial interview. To clarify, a follow-up telephonic interview was conducted, during which the parents disclosed that the child had typical premorbid developmental

milestones and had begun using two-word utterances before the stroke. This led to the clinical impression of regression in speech and language skills secondary to stroke. The child was receiving physiotherapy (PT) for his hemiparesis and speech and language therapy (SLT) to address target behaviors such as improving attention span, expressing kinship terms, and articulating specific words in Telugu, including /dʒija/ (water) and /bua/ (food). After 10 sessions, the therapies were discontinued due to difficulties with commuting for the parents. Despite this, significant progress was made: his attention span increased from 5 seconds to 20 seconds; he achieved 60% consistency in saying /ka/ for /akka/ (sister) and /pa/ for /appa/ (father); he expressed /am am/ for /bua/ with 65% consistency; and he was able to point to his mouth to request water with 80% consistency.

DISCUSSION

In this case study, we present a male child with multiple health conditions, including KS, high fever, chickenpox, stroke, and seizures. Klinefelter syndrome is often diagnosed during puberty or adulthood due to its distinctive characteristics, such as tall stature, gynecomastia, hypogonadism, small and firm testes, elevated follicle-stimulating hormone levels, decreased facial or pubic hair, and azoospermia (13)

Individuals with KS are at increased risk for stroke and seizures (4–6,14). However, in this case, medical reports indicate that his hemiparesis is secondary to post-infectious chickenpox vasculopathy, a condition also described by Gray (8).

Past literature indicates that individuals with KS typically experience more impairment in expressive language compared to receptive language (15). However, St John et al. (3) found impairments in both receptive and expressive language domains. Reports on infants with KS describe delays in babbling, sound acquisition, and reduced semantic and syntactic abilities in 18-month-old males (16,17). In this study, the child did not exhibit impairments in either receptive or expressive language prior to the stroke. Post-stroke, there was noticeable regression in his linguistic skills. St John et al. (3) also reported that two participants aged 1.1 and 1.11 years showed no impairments in receptive or expressive language.

Stroke is a primary cause of childhood aphasia (9), raising concerns about diagnosing the child with Acquired Childhood Aphasia (ACA). According to Van Hout (18), two criteria are essential for diagnosing ACA:

1. Age: The child must be at least 2 years old, as there needs to be some prior language development for deterioration to be evident.

2. Language Disruption: There must be a noticeable disruption in language components such as syntax, vocabulary, and phonetic relations.

Van Hout also notes that brain lesions acquired before age 2 lead to abnormalities in lexical acquisition, which may be termed "developmental language breakdown" or "disruption of language milestones." Although the "Children's Acquired Aphasia Screening Test (CAAST)" (19) is available, no specific diagnostic tool for ACA exists. An adapted version of CAAST in Kannada (20) is available, but it is for ages 3 to 7 years, and the child's native language is Telugu. Despite the stroke occurring at 1.6 years of age, and his development of 2-word utterances before the stroke, the significant deterioration in language skills assures considering the diagnosis of ACA.

In this scenario, the child, who has Klinefelter syndrome and is therefore prone to language deficits (3), experienced a stroke that led to loss of previously acquired language skills and developed inattentiveness. This inattentiveness can be attributed to the infarct in the basal ganglia, which plays a critical role in attention regulation (21). Significant improvements in speech and language skills have been observed, despite the limited time. Use of augmentative and alternative communication may also support the growth in speech and language skills (22).

CONCLUSION

This case report illustrates the complex challenges of managing multifaceted health conditions in a pediatric patient, specially focusing on the interaction of Klinefelter Syndrome, post-infectious chickenpox vasculopathy, stroke, and seizures. The significant regression in speech and language skills following stroke in an otherwise typically developing child highlights the need for prompt and integrated intervention strategies.

The healthcare providers should prioritize early and comprehensive evaluations, particularly in children

with known genetic vulnerabilities like Klinefelter Syndrome. Multidisciplinary teams, including neurologists, geneticists, speech-language pathologists, and physiotherapists, should collaborate closely to design individualized care plans that address both the medical and developmental needs of these patients. Further assessments of cognition and sensory-motor development may yield added information for management.

Additionally, providing ongoing support to the family is crucial, as managing complex medical conditions can be overwhelming. Regular counselling, education on the child's condition, and connecting families with support networks can improve overall well-being and adherence to therapeutic regimens.

Further research into the long-term impacts of early stroke in children with genetic syndromes like Klinefelter Syndrome is essential to inform evidence-based practices and optimize interventions that support both the child's immediate needs and long-term developmental trajectory.

ACKNOWLEDGEMENT

We sincerely acknowledge the parents of the child for their cooperation and invaluable support throughout the assessment and intervention process, which significantly contributed to the understanding and documentation of this case. We also extend our heartfelt gratitude to Ms. Anu Lokheshwar for proofreading and constructive feedback, which greatly enhanced the clarity and presentation of this article.

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CITE THIS ARTICLE:
M.I. Maseera, B. M. Mohan, C. S. Sebastian, S Lokheshwar, Navigating speech and language complexity: A case study of a child with multifaceted health conditions. J Ind Fed NR, 2025, March 2025; 1 (2): 42-46.