International Journal for Multidisciplinary Research (IJFMR)
E-ISSN: 2582-2160 ● Website: www.ijfmr.com ● Email: editor@ijfmr.com

Speech and Language Disorders in Children with Klinefelter Syndrome: A Review Study

Sushant Gupta[1], Mansi Doshi[2]

[1,2]Assistant Professor, Department of Audiology and Speech-Language Pathology, Faculty of Paramedical and Rehabilitation Sciences, Dr. Shakuntala Misra National Rehabilitation University, Lucknow, Uttar Pradesh. Email ID: sgupta_baslp@dsmnru.ac.in, Mob.: 9335927188

Abstract:
Klinefelter syndrome is increasingly diagnosed in childhood, with many boys exhibiting speech and language difficulties throughout development. Studies have shown that most children with this syndrome have some communication impairments. Common findings in speech and language assessments include delayed oromotor milestones, notably poor suckling and babbling, as well as persistent articulation and phonological errors. Receptive language generally surpasses expressive vocabulary and grammar, but deficits in syntactic production, word-finding, and auditory memory are typical. Pragmatic impairments—such as challenges with conversational reciprocity, figurative language, and social language use—are also common in children with this syndrome. These issues tend to worsen as academic and social demands increase. Proposed mechanisms include X-linked gene dosage effects on brain development (e.g., neuroligin-neurexin pathways) and early androgen deficiency. A variety of standardized tools—such as Bayley Scales, vocabulary and grammar assessments, and pragmatic checklists—are used to document these challenges. Early markers like hypotonia, feeding difficulties, and delayed first words should prompt genetic evaluation since early speech-language therapy can improve outcomes if delays are detected promptly. This review covers case reports and cohort studies of KS from infancy through adolescence, summarizing phonological, articulatory, expressive and receptive language, and pragmatic profiles; discussing potential neurodevelopmental mechanisms; and highlighting assessment strategies and early intervention options.

Abbreviation: KS: Klinefelter Syndrome

INTRODUCTION

Klinefelter syndrome is a common condition that results when a person assigned male at birth has an extra copy of the X sex chromosome instead of the typical XY. Klinefelter syndrome is a genetic condition that occurs before birth, but it often isn't diagnosed until adulthood. This syndrome may affect testicular growth. This results in smaller testicles, which can lead to making less of the hormone testosterone. The syndrome also may cause smaller muscle mass, less body and facial hair, and extra breast tissue. The effects of Klinefelter syndrome vary, and not everyone has the same symptoms.

Symptoms of Klinefelter syndrome

Many children with Klinefelter syndrome exhibit few or only mild symptoms. Usually, the condition isn't diagnosed until puberty or adulthood, or it may go undiagnosed altogether. For others, it has a noticeable

impact on growth or appearance. Klinefelter syndrome can influence development, physical appearance, sexual development, and mental health.

Prevalence

According to researchers, it has been estimated that approximately 1 in 500 to 1,000 newborn males has an extra X chromosome, making Klinefelter syndrome (KS) one of the most common chromosomal disorders found in newborns. Having a third or fourth X chromosome in a male is much rarer. Scientists are uncertain about the factors that increase the risk of KS. The error that results in the extra chromosome happens randomly, indicating that it is not hereditary, nor is it passed down from parent to child. Research suggests that women who become pregnant after age 35 may be slightly more likely to have a son with KS. However, about half the time, the extra X chromosome in KS originates from the father.

Klinefelter syndrome affects about 1 in 600–650 boys and is the most common sex chromosome aneuploidy. In addition to characteristic physical findings (tall stature, small testes, hypogonadism), KS is associated with a distinctive neurodevelopmental profile. Nearly all studies agree that boys with KS are at increased risk for language disorders, speech delays, and literacy difficulties. For example, a comprehensive review found that "most studies support that males with KS have an increased risk of language disorders and reading disabilities". School-aged boys often qualify for special education services for language-based learning problems. Boada et al. estimated that approximately 70–80% of children with KS exhibit speech, language, literacy, or social communication challenges. Recognizing these issues is crucial because early intervention can help mitigate later academic and social challenges. This review synthesizes published case reports and studies (observational, clinical, intervention) on speech and language development in KS from infancy through adolescence, with emphasis on phonological/articulatory, expressive/receptive, and pragmatic domains. We also outline possible neurobiological mechanisms and summarize recommended assessment tools and interventions.

Methods

A narrative review focused on pediatric KS and communication was conducted. Databases from PubMed and Google Scholar were searched for terms like "Klinefelter syndrome language," "47,XXY speech," and "Klinefelter pragmatic language," resulting in case reports, cohort studies, and reviews. Key sources included pediatrics/neurodevelopment journals (e.g., Dev Disabil Res Rev, J Commun Disord, Pediatrics Reports) as well as interdisciplinary literature on sex chromosome aneuploidies. This study highlights findings specific to childhood (from infancy through adolescence). Relevant data on language milestones, standardized assessments, and outcomes of speech-language interventions were extracted.

Results

Early speech and oral-motor characteristics

Infancy and toddlerhood may provide the first hints of KS-related communication risk. Many infants with KS show oromotor hypotonia, which refers to weak muscle tone in the mouth and face. Parents often report difficulties with feeding, such as trouble latching or sucking, which can coincide with delayed motor milestones. The stages of babbling and the emergence of first words are often delayed. For example, Tragantzopoulou and Giannouli note that infants with KS "often exhibit delayed babbling and acquisition of sounds" and difficulties with suck–swallow coordination. These early motor and feeding problems foreshadow later articulation issues.

By preschool age, articulation and phonology deficits are almost universal. In the largest published cohort (n=26, ages 1–17 years), 100% had oromotor impairments, and about half continued to have phonological errors. Specifically, St John et al. reported that 46% of their cases showed phonological delay, and 19% had a phonological disorder; 8.7% exhibited signs of dysarthria, a motor speech disorder. Common patterns include simplification of consonant clusters, sound distortions, and omission of grammatical endings. Hyponasality or nasal speech is also common (around 70% in one series), possibly indicating velopharyngeal dysfunction. Importantly, these phonological and articulatory errors often persist beyond preschool years: Morgan et al. noted that older children had more noticeable articulation and resonance issues, suggesting delays tend to widen into school age.

A Brazilian case report of an adolescent with XXY used a battery of speech tests (e.g., Profile of Speech-Language Abilities, rapid naming, syntactic awareness) and found marked deficits in phonological awareness, simple sentence production (with syntax errors), and access to vocabulary (long naming times). This case also had pragmatic difficulties (see below) and showed that his functional speech-language age was far below chronological (language skills equivalent to ~6 years old). Although singular, such reports echo cohort findings: nearly all children with KS need speech therapy for articulation and phonological delay.

Expressive and Receptive Language

Language development in KS exhibits a characteristic pattern: expressive language is often more impaired than receptive language. Multiple studies of school-aged boys have found that expressive deficits are the most noticeable feature. For example, Graham et al. (1988) compared boys (average age about 9½) with controls and observed significant expressive language impairment—particularly in syntax production and word retrieval—while basic receptive vocabulary and grammar remained relatively intact. In that study, the XXY group's lower verbal IQ was due to poor expressive performance, not receptive understanding. However, both receptive and expressive skills exist on a spectrum; many KS boys do show mild receptive delays, especially with complex syntax. Graham noted that "except for difficulties in understanding complex sentence structures, the receptive language skills of XXY boys did not differ significantly from controls."

St John et al. (2019) similarly observed that about 70% of children with KS had **mild to severe receptive and/or expressive deficits**. When tested, many KS children score 1–2 standard deviations below norms on vocabulary, grammar, and overall language scales. Tragantzopoulou et al. (2024) emphasize deficits in vocabulary breadth, grammar formation, and verbal working memory. In pragmatic language samples, children often produce shorter sentences, make grammatical errors, and have limited vocabulary relative to expectations. Longitudinal cohort data from the extraordinary babies' study (prenatally identified KS) reported that infants who met criteria for early speech therapy ("reactive" group) had significantly lower Bayley-III receptive and expressive scores at 12 months than those who did not. Overall, there is a **downward shift in mean language performance**, though nonverbal IQ is usually in the average range, indicating a specific language-learning weakness.

Pragmatic Language and Social Communication

Pragmatic and social language impairments are very common in KS. By definition, pragmatic language involves using language appropriately in context: taking turns, understanding nonliteral language, recognizing others' intentions, and more. In one group, 83% of KS boys tested had social-pragmatic language challenges. These challenges include difficulties with conversational reciprocity, staying on

topic, making appropriate eye contact, and adapting language for different listeners. Many KS boys also score poorly on standardized pragmatics checklists or social communication scales.

Consistent with this, case studies highlight difficulties with figurative and higher-level language. Melogno et al. (2018) reported on two boys with KS (ages 9 and 13) who had normal verbal IQs but significant pragmatic deficits: they failed to understand metaphors, implied meanings, humor, or appropriate inferences in stories. These children were considered "language-typical" in early childhood and did not receive therapy, yet their pragmatic weaknesses only became evident in later grades. This emphasizes that even when vocabulary appears age-appropriate, subtle deficits in inferencing, perspective-taking, and discourse can develop.

Studies also highlight an overlap between KS and autism-like profiles. Bishop and Scerif found that a small percentage of KS boys meet criteria for autism spectrum disorder (ASD) features. In their analysis, the overall KS communication profile was "very similar to that seen in Specific Language Impairment (SLI)," but some individuals showed ASD-like pragmatic deficits. Clinicians should therefore be attentive to social communication issues—such as difficulty understanding jokes, sarcasm, or making inferences—in children with KS, even if they do not meet formal ASD criteria. In practice, parents often report that their son with KS "talks too much about one topic," "doesn't ask follow-up questions," or "takes things very literally," all of which suggest pragmatic challenges.

Literacy and Learning

Language impairments in KS often extend into literacy. Many boys with KS struggle with reading and writing. Graham et al. found that expressive language deficits are linked to poor reading and spelling achievements. As the authors state, difficulties in phonological processing and syntax likely cause later dyslexia-like problems: "difficulty learning how to read and spell may be due to a preexisting language disability." In Tragantzopoulou's review, persistent phonological errors and limited vocabulary predicted issues with reading comprehension and written expression. Therefore, even if a child with KS seems to have adequate language skills, early screening for emerging literacy skills is important.

Neurobiological and Developmental Mechanisms

Several lines of evidence explain why 47,XXY results in this language profile. One hypothesis focuses on X-linked gene dosage effects. The extra X chromosome in KS causes overexpression of genes that escape X-inactivation. Bishop and Scerif proposed the neuroligin–neurexin hypothesis: neuroligins (synaptic proteins encoded on X and Y chromosomes) are involved in language and social communication networks. Having three neuroligin gene copies (due to the extra X) along with variants in neurexin may disrupt synaptic functioning, similar to what is observed in SLI and autism. This genetic model matches the observation that the KS language phenotype resembles SLI/dyslexia.

Neuroimaging research supports a brain-based difference in KS. Functional MRI in KS men and boys shows reduced left-hemisphere dominance for language tasks. For example, KS individuals display less lateralized activation in Broca's and Wernicke's areas during verb generation and semantic tasks. This unusual lateralization may contribute to slower processing and more diffuse networks for language, aligning with subtle processing inefficiencies. Structural MRI studies indicate decreased gray and white matter volumes in regions associated with language (such as the superior temporal gyrus) in KS compared to XY controls, though findings vary with age and mosaicism. Notably, many MRI analyses reveal a general reduction in total brain volume, enlarged ventricles, and thinning in left frontal motor and language

regions in KS. These anatomical differences are linked to neuropsychological test scores in some studies (e.g., frontal lobe volume correlating with language scores).

Hormonal factors might also influence development. Testosterone is essential for early brain growth; boys with KS experience prenatal and early postnatal androgen deficiency. Some researchers have noted that early testosterone supplementation could slightly enhance language skills, although solid evidence from large trials is lacking. StatPearls states that hypogonadism starts in utero for KS, which may contribute to underdeveloped neurocognitive functions. In practice, it remains unclear how much androgen therapy directly impacts language, but it remains an active area of research.

Finally, cognitive factors such as auditory memory and processing speed are consistently weak in KS and likely limit language development. For example, both Boada et al. and Tragantzopoulou's review highlight deficits in short-term auditory memory and the processing of rapid speech. These deficits would make learning new words and following complex verbal instructions more difficult, adding to expressive delays.

In summary, the KS language phenotype seems to result from a combination of X-chromosome effects, neuroanatomical differences (especially in left-hemisphere language areas), and general processing and memory limitations.

Assessment Tools

Studies involving children with KS use a wide range of tools for language and speech assessment. For infants and toddlers, researchers often used the Bayley Scales of Infant and Toddler Development (3rd Ed.) to measure receptive and expressive language quotients. In longitudinal studies of extraordinary babies, cohort, receptive and expressive Bayley scaled scores, and growth metrics were analyzed to track development. Standardized assessments for preschool and school-age children include the Clinical Evaluation of Language Fundamentals (CELF) and the Preschool Language Scales, along with the Peabody Picture Vocabulary Test (PPVT) for receptive vocabulary, and the Expressive Vocabulary Test (EVT) or other naming tests. Tests like the Goldman-Fristoe Test of Articulation and the DEAP (Diagnostic Evaluation of Articulation and Phonology) are commonly used to evaluate articulation and phonology. One case study used the Brazilian Profile of Speech-Language Abilities, the Token Test (for auditory comprehension), and Rapid Automatized Naming to document deficits.

In pragmatics, tools like the Test of Pragmatic Language (TOPL) or the Children's Communication Checklist (CCC-2) are useful, although few published studies on KS directly reference a specific pragmatics test. Instead, pragmatic impairments are often described qualitatively or reported by caregivers. Neuropsychological assessments (e.g., WISC, Raven matrices) are used to place language scores within the broader context of cognition. In various studies, parental concerns or referrals for speech therapy frequently act as triggers, with many papers relying on the Clinical Evaluation of Language conducted by speech-language pathologists. The key point is that no single test is designed specifically for KS; however, clinicians should use age-normed tools to thoroughly evaluate each child's speech articulation, syntax, semantics, and pragmatics, while noting any discrepancies (e.g., normal social understanding but poor articulation, or the opposite).

Interventions and Early Detection

Since speech and language delays are common in KS, early detection is crucial. Signs that should prompt evaluation in infancy include persistent hypotonia, feeding issues, and little or no babbling by 9–12 months. If KS is diagnosed prenatally or through newborn screening, proactive developmental monitoring is recommended. The StatPearls review states that "any male receiving speech and language therapy

should be evaluated for Klinefelter syndrome," emphasizing the importance of awareness on both ends. Once identified, speech-language therapy should begin immediately when delays are observed. Morgan et al. noted that language and literacy problems tend to worsen as demands increase with age and argued that "earlier detection and intervention of phonological errors may reduce the risk for later language and literacy challenges." Graham et al. also highlighted that early attention to expressive language issues could help prevent future learning disabilities and behavioral problems.

About half or more of boys with KS receive speech-language intervention. In large samples, 50–75% have observable delays necessitating therapy. In one prospective cohort, ~61% of prenatally identified KS infants had begun speech therapy by age 3. Speech therapy is rarely contraindicated, and many therapists recommend intensive, frequent sessions. There are no KS-specific intervention protocols, so clinicians typically adapt evidence-based strategies from SLI or phonological delay programs. Interventions often include:

- **Phonological remediation:** drills to correct specific sound errors, minimal pair practice, and auditory discrimination exercises. Visual supports (e.g. pictures, cues) and structured vocabulary lists are used to reinforce target sounds.
- **Language enrichment:** expanded modeling of grammar, interactive book-reading, and vocabulary building. Multisensory approaches (visual gestures, signing) can assist word learning.
- **Oromotor exercises:** feeding therapy and oral-motor play may help hypotonic children develop better speech coordination.
- **Pragmatic training:** role-playing social scenarios, using social stories or comic-strip conversations to teach turn-taking, metaphor understanding, and perspective-taking. Group therapy with peers can also be useful.

Graham et al. (1988) specifically recommended simplified instructions, visual cues, and structured vocabulary programs to help these children learn effectively. Parents play a vital role: studies of other conditions with pragmatic deficits show that coaching families to "narrate" actions, repeat expansions of child utterances, and model social language can enhance formal therapy (adapted from parent accounts). Consistency and repetition are essential, as KS boys often need extra practice to generalize skills.

Some evidence suggests that early testosterone treatment may offer cognitive benefits. Los et al. note that some providers administer "mini-puberty" testosterone in infancy to support development. One study (SciDirect Annals of Pediatrics, 2013) found that neonatal testosterone improved language outcomes, although this remains an emerging area. Endocrine consultation is recommended as part of the KS care team, but any hormonal therapy must be balanced with goals and risks.

In summary, early detection and intervention are crucial. Pediatricians and therapists should closely observe milestone checklists in boys with any developmental delays. Due to the high prevalence of speech delay, a diagnosis of KS should be considered when language lag occurs, and similarly, a KS diagnosis should lead to early referral to speech therapy. Even in children who "catch up" by preschool, ongoing monitoring is essential because higher-level language and pragmatic deficits may not appear until later.

Discussion

The evidence reviewed shows that children with Klinefelter syndrome are highly vulnerable to speech and language disorders, including articulation, phonology, syntax, semantics, and pragmatics. In practical terms, clinicians should expect that any male with KS will likely need individualized speech-language assessment and often therapy. The phenotype varies, but the following broad points emerge:

- **Articulation and Phonology:** Virtually all KS children struggle with clear speech sounds. This may reflect underlying motor planning or hypotonia. Therapy should include direct phoneme work and, if needed, referral to feeding/oral-motor specialists for severe hypotonia.
- **Receptive vs. Expressive**: While comprehension often remains relatively strong, expressive skills tend to lag, especially in forming complex sentences. This "discordance" means a child might seem to understand but speak in short, simple phrases. Receptive language can also decrease under cognitive load, so testing should include complex syntax.
- **Pragmatics:** High rates of social-communication problems mean KS overlaps with pragmatic language impairment (PLI). Routine screening for ASD is sensible, but even without ASD, pragmatic intervention (e.g., social-pragmatic groups) can be helpful.
- **Academic Impact:** Given the linkage to reading and writing difficulties, KS children benefit from literacy support (e.g. phonics-based reading programs) in parallel with speech therapy. Early intervention programs (e.g. IDEA Early Start) should be offered based on language delays alone.
- **Neurobiology:** The neurogenetic model indicates that therapies targeting auditory memory—such as memory games and repeating multisyllabic words—could be beneficial. There is also speculative interest in early hormone therapy to support brain development. More research is needed to determine whether androgen supplementation during infancy or childhood can improve language delays, as current evidence is anecdotal and retrospective.
- **Gaps and Future Directions:** Many published KS studies are cross-sectional; more longitudinal research is necessary to monitor language development and therapy outcomes. There is a shortage of KS-specific intervention trials.

Conclusion

In conclusion, speech and language disorders are key features of Klinefelter syndrome in childhood. Most affected boys exhibit expressive language and pragmatic difficulties, with relative strengths in basic comprehension. Oromotor hypotonia and phonological issues often appear early and tend to persist without intervention. Clinicians should screen for KS in any boy with unexplained speech delay, and conversely, a KS diagnosis should prompt a formal communication evaluation. Early, intensive speech-language therapy—customized to the child's profile (phonological, grammatical, or social goals)—is recommended, along with educational support for literacy. Understanding the neurodevelopmental causes (such as neural asymmetry or gene dosage) of these issues may help guide future treatments. Ultimately, proactive management of speech-language problems can significantly enhance academic success and overall quality of life for boys with KS.

Future Scope of Study

Future research should test whether adapted SLI therapies (e.g., Fast ForWord, narrative intervention) work equally well in KS. Genetic studies might identify which extra-X genes most impact language, possibly pointing to novel targets.

References

1. Graham JM Jr, Bashir AS, Stark RE, Silbert A, Walzer S. Oral and written language abilities of XXY boys: implications for anticipatory guidance. Pediatrics. 1988 Jun;81(6):795-806.
2. Bishop DVM, Scerif G. Klinefelter syndrome as a window on the aetiology of language and

communication impairments in children: the neuroligin–neurexin hypothesis. Acta Paediatr. 2011 Jun;100(6):903-7. doi:10.1111/j.1651-2227.2011.02150.x

3. Boada R, Janusz J, Hutaff-Lee C, Tartaglia N. The cognitive phenotype in Klinefelter syndrome: a review of the literature including genetic and hormonal factors. Dev Disabil Res Rev. 2009;15(4):284-94. doi:10.1002/ddrr.83.
4. Tragantzopoulou P, Giannouli V. Understanding the neuropsychological implications of Klinefelter syndrome in pediatric populations: current perspectives. Pediatr Rep. 2024;16(2):36. doi:10.3390/pediatric16020036.
5. St John M, Ponchard C, van Reyk O, Mei C, Pigdon L, Amor DJ, Morgan AT. Speech and language in children with Klinefelter syndrome. J Commun Disord. 2019 Mar-Apr;78:84-96. doi:10.1016/j.jcomdis.2019.02.003.
6. Melogno S, Pinto MA, Orsolini M, Tarani L. Beyond the literal meaning of words in children with Klinefelter syndrome: two case studies. Brain Sci. 2018 Sep;9(9):171. doi:10.3390/brainsci8090171.
7. Los E, Leslie SW, Kadam SJ, Ford GA. Klinefelter syndrome. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2025.
8. Laing BA, Letourneau KN, Bird LM, Mazzocco MMM. Prevalence and application of speech-language therapy in individuals with Klinefelter syndrome: a scoping review. Cureus. 2025 Jun;17(6):e85523. doi:10.7759/cureus.85523.
9. Nielsen, J., & Wohlert, M. (1991). Chromosome abnormalities found among 34,910 newborn children: Results from a 13-year incidence study in Arhus, Denmark. Human Genetics, 87(1), 81–83.
10. Klinefelter, H.F., Reifenstein, E.C., & Albright, F. (1942). Syndrome characterized by gynecomastia aspermatogenesis without A-Leydigism and increased excretion of follicle-stimulating hormone. Journal of Clinical Endocrinology & Metabolism, 2, 615–627.
11. National Human Genome Research Institute. Learning about Klinefelter syndrome. Retrieved June 5, 2012, from https://www.genome.gov/Genetic-Disorders/Klinefelter-Syndrome