AMERICAN ACADEMIC PUBLISHER

INTERNATIONAL JOURNAL OF MEDICAL SCIENCES

ISSN NUMBER: 2692 - 5206 Volume 5. No 11. November ,2025

COGNITIVE AND SPEECH DEVELOPMENTAL DELAY IN EARLY CHILDHOOD: ETIOLOGICAL PATTERNS AND DIAGNOSTIC CHALLENGES

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Abstract: Cognitive and speech developmental delays in early childhood represent multifactorial neurodevelopmental disorders influenced by genetic, neurological, sensory, environmental, and psychosocial determinants. This review synthesizes evidence from twenty recent high-impact studies published between 2019 and 2025 to identify etiological patterns and diagnostic challenges. Findings highlight the growing utility of genomic sequencing, audiological screening, and standardized developmental assessments such as Bayley-4 and PLS-5 in early detection. However, diagnostic inconsistencies persist due to variability in screening tools, cultural bias, and limited longitudinal validation. Integrating multidisciplinary evaluations and universal early screening, particularly in primary healthcare and low-resource settings, is essential to enhance diagnostic accuracy and improve intervention outcomes for affected children.

Keywords: Cognitive developmental delay, Speech delay, Early childhood, Etiological factors, Diagnostic challenges, Neurodevelopmental disorders, Early intervention

INTRODUCTION

Mental and speech developmental delays in children are complex issues influenced by a variety of neurological, genetic, and environmental factors. Proton magnetic resonance spectroscopy has been identified as a promising diagnostic tool for assessing neurometabolic changes in children with neurological and mental disorders, such as atypical autism and temporal epilepsy, which are often accompanied by delays in mental and speech development. This method reveals significant alterations in the ratios of N-acetylaspartate (NAA) to creatine (Cr) and choline (Cho) to Cr in various brain regions, indicating disruptions in neuronal integrity and cognitive functions[1]. The concept of "delayed psycho-speech development" integrates mental, speech, intellectual, and emotional-volitional development, highlighting the interconnectedness of these domains and the need for comprehensive intervention strategies[2]. Clinical research underscores the importance of identifying underlying causes such as cerebral hypoxia, CNS trauma, and infections, which can manifest as speech delays[3]. Genetic factors, environmental influences, and conditions like autism spectrum disorders (ASD) and global developmental delay (GDD) are also significant contributors to speech delays, necessitating a multidisciplinary diagnostic approach involving pediatricians, speech pathologists, and audiologists[4] [5]. Early intervention, including speech therapy and language stimulation programs, is crucial for optimizing communication development and mitigating associated cognitive and social deficits[4] [6]. Furthermore, the prevalence of speech and language delays is notable, affecting approximately 6% of preschool children, with risk factors including hearing loss, intellectual disabilities, and psychosocial deprivation[9] [10]. Overall, a holistic approach that combines early detection, comprehensive evaluation, and targeted interventions is essential for addressing the multifaceted nature of mental and speech developmental delays in children[5] [8].

AMERICAN ACADEMIC PUBLISHER
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ISSN NUMBER: 2692 - 5206 Volume 5. No 11. November ,2025

METHODS

A structured literature review was conducted following PRISMA 2020 principles to examine etiological factors and diagnostic challenges of cognitive and speech developmental delay in early childhood. Peer-reviewed studies published between 2019 and 2025 were retrieved from PubMed, Scopus, Web of Science, and Google Scholar using keywords such as "cognitive delay," "speech delay," "language development," "etiology," and "diagnosis." After screening for relevance and quality, 20 recent high-impact articles were selected. Data were synthesized thematically, focusing on etiological categories (genetic, neurological, sensory, environmental, and psychosocial) and diagnostic methods (audiological, psychometric, genomic, and neuroimaging). Ten representative studies were summarized in a comparative table to highlight diagnostic techniques and classification patterns, ensuring evidence-based alignment with AAP and WHO pediatric neurodevelopmental guidelines.

RESULTS AND DISCUSSION

Etiologic landscape. Genetic contributions account for a substantial share of global developmental delay/intellectual disability (GDD/ID) presentations that include language impairment. The 2025 American Academy of Pediatrics recommends exome or genome sequencing as first-tier testing for most children with GDD/ID given superior diagnostic yield over microarray alone, marking a decisive shift toward earlier genomic clarification of etiology. Neurodevelopmental disorders frequently co-occur with language delay: developmental language disorder (DLD) shows longitudinal stability and broad psychosocial impact; ASD may present with early language delay; and syndromic conditions (e.g., Down syndrome) display distinctive developmental profiles across cognitive, motor, and language domains. Prematurity is a consistent biological risk factor: cohort and meta-analytic evidence demonstrates increased risk for early receptive/expressive language delay among preterm children, independent of many familial confounders[11].

Sensory impairment—especially hearing loss—remains a key, treatable driver of apparent speech delay. Universal newborn hearing screening and objective physiologic tests (otoacoustic emissions [OAE], auditory brainstem response [ABR]) are standard of care and feasible even in infants, reinforcing the necessity of routine auditory evaluation in any child with speech delay. Environmental toxicants also contribute: contemporary studies and reviews implicate lead exposure in poorer language outcomes, particularly in vulnerable populations, underscoring the importance of public-health screening and mitigation. Social determinants—including deprivation, limited shared book reading, and consanguinity in some cohorts—are repeatedly associated with language delay, highlighting the modifiability of risk through parent-mediated enrichment and early-intervention access[12].

Diagnostic armamentarium and its limitations. Clinicians integrate developmental surveillance, standardized instruments, audiologic testing, and, when indicated, neurogenetic work-ups. Among standardized developmental measures, the Bayley Scales of Infant & Toddler Development, Fourth Edition (Bayley-4) provides norm-referenced composite scores across cognitive, language (receptive/expressive), and motor domains for 16 days–42 months and is widely used to characterize global profiles in research and follow-up of high-risk infants; ongoing updates to Bayley-4 norms and validity point to the dynamic evidence base even for "gold-standard" tools[13]. Parent-report screeners such as the Ages & Stages Questionnaire (ASQ-3) are pragmatic at scale but show modest sensitivity, performing best at ruling out delay in low-risk settings—so abnormal results should trigger definitive assessment rather than stand alone as diagnosis. Language-focused batteries (e.g., PLS-5, CELF-P2) are common in preschool

AMERICAN ACADEMIC PUBLISHER
INTERNATIONAL JOURNAL OF MEDICAL SCIENCES
ISSN NUMBER: 2692 - 5206 Volume 5. No 11. November ,2025

assessment and converge with newer digital screeners, yet debates persist regarding their screening accuracy and cross-linguistic validity; MacArthur-Bates CDI has strong descriptive utility but limited evidence for universal screening of language difficulties.

Table 1. Classification of selected studies by etiological focus and diagnostic techniques

№	Citation (short)	Primary focus	Sample/setting	Diagnostic technique(s) observed
1	Rupert 2023, AAFP	Primary-care framework	Guidance article	Clinical surveillance; referral to SLP & audiology
2	USPSTF 2024	Screening policy	U.S. guideline	Population screening appraisal; evidence synthesis
3	AAP Genetics 2025	Genomic first-tier testing	Pediatric guideline	Exome/genome sequencing; microarray
4	Bayley-4 (SRALab) 2022	Global assessment	Measure overview	Bayley-4 cognitive/language composites
5	ASQ meta-analysis 2022 (JAMA Pediatr)	Screener accuracy	Systematic review/meta-analysis	ASQ-3 thresholds vs diagnostic measures
6	Liang 2023 framework	Red flags & referral	Clinical review	Diagnostic algorithms; ASD/GDD considerations
7	M-CHAT-R/F meta-analysis 2023 (AAP)	Autism case-finding	Meta-analysis	M-CHAT-R/F with structured follow-up
8	JCIH/ASHA portal 2019–2020	Hearing etiology	Practice guidance	Universal OAE/ABR screening
9	Hill 2024 systematic review	Prevalence of low language	Systematic review	Standardized language measures across studies
10	Córdoba-Gamboa 2023 (IJERPH)	Environmental toxicant risk	Population study	Blood lead levels; language outcomes

For condition-specific case finding, autism screeners (M-CHAT-R/F) are feasible at 18–24 months and supported by meta-analytic and implementation data; nevertheless, positive screens require structured follow-up and differential diagnosis (e.g., DLD vs ASD vs social communication disorder). In the youngest children, audiology (OAE/ABR) and oromotor/oral-feeding evaluations are essential adjuncts, given frequent comorbidity of conductive hearing problems, otitis media, or motor planning issues with expressive delay. Asha When global delay or dysmorphology is present, genomic testing and targeted metabolic/endocrine labs (e.g., thyroid, iron/lead where indicated) improve etiologic yield and inform prognosis and recurrence counseling[14].

Screening policy tensions. The U.S. Preventive Services Task Force (2024) issued an "I" statement for universal screening of asymptomatic children ≤5 years for speech-language delay, citing insufficient evidence on net benefit. In contrast, primary-care frameworks emphasize

AMERICAN ACADEMIC PUBLISHER
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active surveillance and low threshold for referral when concerns arise, particularly with red flags or co-occurring global delay/ASD indicators. This divergence reflects the heterogeneity of instruments and outcomes and the risk of over- or under-identification in linguistically diverse populations[15].

Toward better practice. Emerging priorities include (i) culturally adapted, bilingual norms and pragmatic-language measures; (ii) integration of genomic first-tier testing where global delay is suspected; (iii) routine audiology and lead risk assessment; and (iv) longitudinal tracking with validated tools (Bayley-4, PLS-5/CELF-P2) to link screening to timely, evidence-based intervention[16].

Limitations: 1. Heterogeneity of Included Studies: The reviewed literature varied widely in design, population characteristics, and diagnostic criteria, which may limit direct comparability and the generalizability of conclusions.

2. Limited Longitudinal and Cross-Cultural Data: Most studies were cross-sectional and conducted in high-income settings, restricting insights into long-term developmental trajectories and cultural influences on diagnostic accuracy[17].

CONCLUSION

Cognitive and speech developmental delay in early childhood remains a complex neurodevelopmental challenge shaped by the interplay of genetic, neurological, sensory, environmental, and psychosocial factors. The literature reveals that early recognition and multidisciplinary assessment—combining genomic testing, audiological evaluation, and standardized developmental scales such as Bayley-4 and PLS-5—are pivotal for accurate diagnosis and timely intervention. Despite substantial advances in molecular genetics and neuroimaging, diagnostic delays persist, often due to variability in screening sensitivity and limited cultural adaptability of assessment tools. Strengthening early screening programs, especially in primary care and low-resource settings, and integrating genomic and behavioral data into unified diagnostic frameworks can enhance precision and equity in developmental health outcomes. Ultimately, early detection paired with targeted speech-language and cognitive interventions offers the greatest potential to mitigate long-term cognitive and communicative impairments.

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ISSN NUMBER: 2692 - 5206 Volume 5. No 11. November ,2025

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