



# Speech and Language Delay in Children: Child Neurology Experience

## Çocuklarda Konuşma ve Dil Gecikmesi: Çocuk Nörolojisi Deneyimi

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### ABSTRACT

**Objective:** Speech delay in early childhood can signify various underlying issues, including neurological disorders. The study aims to comprehensively investigate the etiology of speech delay in children aged 2-5 years and to delineate potential neurological disorders associated with this condition.

**Method:** A total of 220 patients presenting with speech delay/disorder between 2018 and 2021 were included in the study. Data obtained detailed analysis of medical history, physical examinations, developmental assessments, brain magnetic resonance imaging scans, electroencephalography recordings, chromosome karyotype analysis and array comparative genomic hybridization, Brainstem Evoked Response Audiometry were retrospectively reviewed, and recorded retrospectively on pre-prepared data forms.

**Results:** Majority (80%) of the study cohort had isolated language delay, while 20% had global developmental delay (GDD). Autism emerged as the most frequently diagnosed condition among children with GDD, followed by other significant diagnoses including neurofibromatosis, cerebral palsy, intellectual disability, hypothyroidism, and Rett syndrome.

**Conclusion:** These results underscore the significance of neurological evaluation in children presenting with speech delay, as it serves as a pivotal step in identifying potential developmental issues early on, such as ASD or GDD. This study contributes valuable insights into understanding the multifaceted nature of speech delay in early childhood and highlights the importance of assuming a holistic approach towards assessment and intervention in affected children.

**Keywords:** Speech delay, autism, global developmental disorders

### ÖZ

**Amaç:** Erken çocukluk döneminde konuşma gecikmesi, nörolojik bozukluklar da dahil olmak üzere altta yatan çeşitli sorunlara işaret edebilir. Çalışma, 2-5 yaş arası çocuklarda konuşma gecikmesinin etiyolojisini kapsamlı bir şekilde araştırmayı ve bu durumla ilişkili potansiyel nörolojik bozuklukları tanımlamayı amaçlamaktadır.

**Yöntem:** Çalışmaya 2018 ve 2021 yılları arasında konuşma geriliği/gecikmesi/bozukluğu ile başvuran toplam 220 hasta dahil edildi. Tıbbi geçmiş incelemeleri, fiziksel muayene bulguları, gelişimsel değerlendirmeler, beyin manyetik rezonans görüntüleme taramaları, elektroensefalografi kayıtları, kromozom karyotip analizi ve array tabanlı karşılaştırmalı genomik hibridizasyon (dizi "comparative genomic hybridization"), İşitsel Beyin Sapı Davranımı Testi verileri önceden hazırlanmış veri formlarına retrospektif olarak kaydedildi.

**Bulgular:** Katılımcı grubunun %80'inde izole dil gecikmesi, %20'sinde ise global gelişimsel gecikme olduğu saptandı. Otizm, global gelişimsel gecikme yaşayan çocuklar arasında en sık teşhis edilen durumdu ve diğer önemli teşhisler arasında nörofibromatoz, serebral palsi, zeka geriliği, hipotiroidizm ve Rett sendromu yer aldı.

**Sonuç:** Bu sonuçlar, konuşma gecikmesi/geriliği ile başvuran çocuklarda nörolojik değerlendirmenin önemini altını çizmektedir; çünkü bu, otizm spektrum bozukluğu veya genel gelişimsel gecikme gibi potansiyel gelişimsel sorunların erken dönemde belirlenmesinde önemli bir adım olarak hizmet etmektedir. Bu çalışma, erken çocukluk döneminde konuşma gecikmesinin çok yönlü doğasının anlaşılmasına yönelik değerli bilgiler sunmakta ve etkilenen çocuklarda değerlendirme ve müdahaleye yönelik bütünsel bir yaklaşımın önemini vurgulamaktadır.

**Anahtar kelimeler:** Konuşma gecikmesi, otizm, global gelişimsel bozukluklar

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## INTRODUCTION

Speech and language skills play a crucial role in child development, and deficiencies in these areas are referred to as communication disorders. These disorders are common neurodevelopmental conditions of childhood, classified by the "Diagnostic and Statistical Manual of Mental Disorders-V" (DSM-V) into categories including language disorder, speech sound disorder, childhood-onset fluency disorder (stuttering), social (pragmatic) communication disorder, and unspecified communication disorder<sup>(1)</sup>.

According to DSM-V criteria, to establish the diagnosis of language or speech sound disorder, the condition must not be attributable to general health status, neurological disorders, or general developmental delay<sup>(2)</sup>. Thus, a thorough neurological evaluation in patients with speech and language disorders is essential to identify any potential underlying or concomitant neurological conditions, such as epilepsy or cerebral palsy (CP).

Speech delay refers to a delay in achieving language development milestones and it is one of the most common complaints among individuals with speech and language disorders. While the prevalence of speech delay is around 10-15% among 2-year-old and drops to approximately 4-5% after the age of three<sup>(3,4)</sup>.

The aim of this study is to investigate the etiology of speech delay in the 2-5 year age group and to shed light on possible underlying neurological disorders.

## MATERIALS and METHODS

Children aged 2-5 years who presented to the pediatric neurology outpatient clinic between 2018 and 2021 with complaints of speech delay, or disorder were enrolled in the study. Detailed medical histories, physical examination findings, developmental assessments, brain magnetic resonance imaging (MRI) scan, electroencephalography (EEG), chromosome karyotype analysis, vitamin B12 and hemoglobin test results of the patients were retrospectively reviewed.

The patients were evaluated to determine whether they had isolated language impairment or global developmental delay (GDD) using the Denver Developmental Screening Test (DDST). DDST is a tool used to screen developmental delays in children aged 0-6 years. It evaluates children in four domains: motor skills, language development, social skills, and personal-social development. Patients who are delayed only in the

language domain compared to their peers are considered to have an isolated language delay, while those who are delayed in multiple developmental domains (motor, language, social, cognitive) compared to their peers are considered to have GDD. In our study, isolated language delay was defined as a delay only in the language domain of the DDST. Since patients with a delay in the language domain were included in the study, GDD was defined as a delay in at least one additional domain (motor, social, or cognitive) along with the language domain.

In case of need auditory evaluation was conducted using the Brainstem Evoked Response Audiometry (BERA) test.

For each patient presenting with speech delay, screen time was inquired and appropriate recommendations were provided.

The diagnosis of Attention-Deficit/Hyperactivity Disorder (ADHD) was taken into consideration for patients monitored by child psychiatry under this diagnosis.

Children who exhibited normal receptive language skills and had undergone a normal diagnostic work-up but displayed clear expressive language delay were categorized as "late talkers".

## Statistical Analysis

Statistical analysis of data was performed using the Statistical Package for the Social Sciences version 25 (SPSS 25.0) software, and results were presented as numbers and percentages. The descriptive statistics of the quantitative variables that conformed to the normal distribution were shown as mean  $\pm$  standard deviation, and the descriptive statistics of the quantitative variables that were not normally distributed were shown as median (minimum-maximum) or mean  $\pm$  standard deviation.

Ethical approval was obtained from the Clinical Research Ethics Committee of the University of Health Sciences Turkey, Kanuni Sultan Süleyman Training and Research Hospital (approval number: 2022/05, date: 01.10.2022).

## RESULTS

A total of 220 patients who presented to the pediatric neurology outpatient clinic with complaints of speech delay or disorder were included in the study. Study population consisted of 46 (20.9%) female and 174 (79.1%) male patients. Family history of speech delay

was revealed in 34.1% of the patients, and 1.4% of them was bilingual. The characteristics of the patients are detailed in Table 1. EEGs were performed for 86 (39%) patients. EEG results were abnormal in 5.1% of the

Personal and family history	n (%)
Prematurity	33 (15)
Neonatal intensive care hospitalization	46 (20.9)
Phototherapy	18 (8.2)
Febrile seizure	8 (3.1%)
Afebrile seizure	3 (1.4%)
Speech delay in family	75 (34.1%)
Consanguinity	21 (9.5%)
Bilingualism	3 (1.4%)

EEG findings (n=86)	n (%)
Normal	77 (35)
Background activity disorganisation	1 (0.5)
Interictal epileptiform activity	8 (4.6)

EEG: Electroencephalography

Etiologic factors and accompanying diagnoses		n	% (-)
Isolated language disorder (n=176/220)	Late talkers	62	28.1
	Fluency disorder-stuttering	19	8.6
	Other isolated language disorder	95	43.1
	ADHD*	4*	
	Epilepsy*	1*	
GDD (n=44/220)	Autism	19	8.6
	Mental retardation	2	1
	NF-1	2	1
	Cerebral palsy	1	0.5
	Hypothyroidism	1	0.5
	Rett syndrome	1	0.5
	Undiagnosed patients	18	8.1
	Epilepsy*	1*	
	ADHD*	2*	
Cerebellar mass*	1*		

\*Accompanying diagnosis, ADHD: Attention-deficit/hyperactivity disorder, NF-1: Neurofibromatosis Type 1, GDD: Global developmental delay

patients (Table 2). Two patients were diagnosed with concomitant epilepsy. Cranial imaging was performed for 31 patients, revealing periventricular leukomalacia, hydrocephalus, a cerebellar mass, and an arachnoid cyst in one patient each. BERA test performed on 29 patients could not detect hearing loss in patients. In 5 patients, chromosome karyotype analysis and array comparative genomic hybridization were performed; and, single-gene analysis carried out in one patient established the diagnosis of Rett syndrome.

Mean hemoglobin value was  $12.4 \pm 0.88$  g/dL and median B12 value was 368 pg/mL (104-2000). Three patients had iron deficiency anemia, and four patients had vitamin B12 deficiency.

Isolated language disorder was diagnosed in 176 (80%) patients, while GDD was identified in 44 (20%) patients. In patients with GDD, autism was the most frequently identified etiology (8.6%). The etiology GDD could not be determined in 8.1% of the patients. The etiologic factors and accompanying diagnoses are summarized in Table 3. The patients with isolated language delay, had also asthma (n=3), had atrial septal defect (n=1) and ventricular septal defect (n=1). Incidentally, a cerebellar mass was detected in one patient with GDD.

## DISCUSSION

In our study, we have demonstrated that 20% of patients presenting solely with complaints related to speech delay were diagnosed with GDD. Therefore, patients with speech delay should be thoroughly examined. A multitude of etiologies can contribute to childhood speech and language delay. In children presenting with speech and language retardation, a comprehensive developmental assessment is essential, as atypical language development may serve as the first indicator of other physical and developmental issues<sup>(5)</sup>.

Many studies have reported that the ratio of affected males to females in specific language disorder is approximately 2:1 to 3:1<sup>(6,7)</sup>. Consistent with the literature, in our study there was a male gender predominance with a ratio of 3.8:1 (79.1%)<sup>(5,8)</sup>. Additionally, conditions associated with language delay, such as GDD/intellectual disability and autism spectrum disorder (ASD), are more common in males rather than females<sup>(9-11)</sup>.

Some authors have reported an increased incidence of language-related disorders among family members of children with language disorders, with a range of 28% to 50%<sup>(12-14)</sup>. In the present study, a positive family history was revealed in our 75 (34.1%) patients.

### Isolated Language Delay

For early recognition of speech delay, it is crucial to have a good understanding of the developmental process. Red flags for speech delay include not using the words mama/papa/dada to call parents at 12 months, not having learnt at least 5 words at 18 months, and using less than 50 words and not being able to connect two words at 24 months of age. In our study, the final diagnoses of patients who presented with complaints of speech delay and were diagnosed with isolated language delay are outlined in Table 3. Among patients with isolated speech delay, 62 out of 176 (28.1%) were identified as late talkers. Late talkers typically have a significantly lower word count compared to their peers at the age of 24 months. However, half of these children catch up with normal speech milestones by the age of 3 years<sup>(15,16)</sup>. Since we lack data on the final language development status of all patients, we cannot make a comment on this issue.

One of the important diagnoses that should not be overlooked in patients presenting with speech delay is hearing loss. In our study, although hearing loss was not detected in any patient with were identified, we observed that hearing test could be applied at a low rate which may be attributed to patients' poor compliance with the tests and the families' adherence to the diagnostic procedures.

Stuttering can be classified as developmental, neurogenic, or psychogenic. Neurological stuttering is rare and typically follows a neurological event such as acquired brain damage, brain injury, or stroke<sup>(17,18)</sup>. In the present study, 19 children were diagnosed with stuttering, and none of them had neurogenic stuttering.

Early detection of patients with isolated language delay and initiation of appropriate therapy are crucial for prognosis. Furthermore, delays in language development may serve as early signs of more common problems such as ASD, GDD, and intellectual disability<sup>(19,20)</sup>.

### Global Developmental Delay

GDD is defined as a delay in two or more developmental domains, including gross/fine motor skills, speech/language, and personal/social communication skills, affecting children under the age of 5 years<sup>(21)</sup>. Common etiologies of GDD include genetic syndromes/chromosomal abnormalities, intrapartum asphyxia, cerebral dysgenesis, psychosocial deprivation, metabolic diseases, neurocutaneous diseases, intrauterine infections, hypothyroidism, and toxin exposure<sup>(22-24)</sup>. Additionally, GDD can occur in

combination with behavioral problems such as ASD and attention-deficit hyperactivity disorder<sup>(25)</sup>.

In our study, GDD was observed in 44 out of 220 (20%) patients, and etiologic factors were identified in twenty-six of them. Etiologic factors could not be determined in 18 (40.9%) cases. The most prevalent diagnosis was autism, identified in 19 patients. Other identified diagnoses included mental retardation, Neurofibromatosis Type 1, CP, hypothyroidism, and Rett syndrome (Table 3). In Ozmen et al.<sup>(26)</sup>, investigated 247 patients with GDD, and reported perinatal complications in 21%, chromosomal abnormalities in 9%, genetics/dysmorphic syndromes in 3%, metabolic disorders in 4%, hypothyroidism in 4%, neurocutaneous syndromes in 3%, intrauterine infection in 2%, and disorders of unknown etiology in 36% of their cases. GDD has a wide variety of etiologies. In our study, the low proportion of patients with identified etiology and limited diversity is due to our inclusion criteria, which only encompassed patients with complaints of speech delay. The presentation of patients with conditions related to asphyxia/perinatal complications, chromosomal abnormalities, genetics/dysmorphic syndromes, and metabolic disorders would have different, irrelevant complaints. Zengin-Akkuş et al.<sup>(20)</sup> found non-autistic developmental delay in 15%, autism in 16%, isolated speech delay in 50%, and normal language development in 19% of patients presenting to the developmental pediatric outpatient clinic with speech delay<sup>(20)</sup>. Unlike our study, the rate of isolated speech delay was lower in their study<sup>(20)</sup>. This difference could be attributed to the inclusion of patients with normal language development in their study.

The overall etiologic yield in an unselected series of children under 5 years of age with GDD is close to 40% and 55% in the absence of any concomitant autistic features<sup>(23)</sup>. These rates may vary according to the study method and design, as well as the severity of GDD. Consistent with the literature, the etiologic factors could be determined in 59% (26/44) of patients with GDD<sup>(24)</sup>.

Shan et al.<sup>(27)</sup> analyzed clinical data of 521 children with GDD aged between 24 and 60 months. The prevalence of ASD in children with GDD was 62.3%<sup>(27)</sup>, whereas this rate was 43.2% (19/44) in our study. The high rates of autism highlight the importance of autism screening in patients presenting with language disorder and GDD. Children with autism often experience delays in speech/language and personal/social developmental domains. Additionally, some patients may also exhibit delays in gross/fine motor domains.

Perinatal asphyxia is commonly associated with developmental delay<sup>(23,26)</sup>. In our study, we observed only one patient diagnosed with CP. We attributed detection of scarce number of CP cases to the presentation of these patients to the outpatient clinic predominantly with motor retardation before language delay becomes apparent.

The history of agnosia should be investigated in patients presenting with a language disorder because language comprehension (auditory verbal agnosia) and verbal expression loss (aphasia) are characteristic diagnostic features of Landau-Kleffner syndrome (LKS). LKS is a rare childhood disorder, and most of these patients exhibit severe EEG abnormalities during sleep. However, there were no patients diagnosed with Landau-Kleffner syndrome in the present study.

EEG recordings provide valuable information within specific clinical contexts such as Angelman syndrome, Wolf-Hirschhorn syndrome, as well as in cases of agnosia/aphasia or a clinical history of seizure/epilepsy<sup>(28)</sup>. In our study, EEGs were performed in 86 out of 220 patients, and two of these patients with a history of seizures were diagnosed with epilepsy. However, we did not identify any specific EEG patterns for particular clinical conditions. In larger series, specific EEG findings may be detected.

In acquired language disorders, patients should be evaluated for potential neurodevelopmental and neurodegenerative diseases such as Leigh encephalopathy, Rett syndrome, metachromatic leukodystrophy, and mucopolysaccharidoses. Other considerations include middle ear infections, neglect/abuse, and head trauma. In the current study, one patient with acquired language disorder was diagnosed with Rett syndrome. Rett syndrome is characterized by stereotypical hand movements, gait abnormalities, loss of speech and purposeful hand use, and normal early development. It is well known that Rett syndrome occurs in females, and acquired microcephaly is its significant clinical characteristics. Mutation analysis of the *MECP2* gene should be ordered in patients exhibiting characteristic symptomatology.

Mental retardation, learning disability, ADHD, and pervasive developmental disorder (autism spectrum) are conditions commonly associated with language disorders in children and adolescents<sup>(29)</sup>. Research has shown that 30.4% of children in the speech-language impaired (SD/LI) group had ADHD based on the DSM-III (1980) criteria, compared to 4.5% of controls<sup>(30)</sup>. In

our study, six patients had coexisting ADHD. The lower rate observed in our study may be attributed to the retrospective nature of the study and the relatively young age range of our patient group.

Language disorders commonly occur as a symptom of ASD and GDD. Therefore, developmental assessment tools such as the DDST, along with diagnostic tools like MRI, EEG, genetic, and metabolic screening tests, should be considered for patients whose language disorders cannot be diagnosed through detailed history and physical examination<sup>(24,27,31,32)</sup>.

## CONCLUSION

In conclusion, it's important to emphasize the possibility of secondary neurological and behavioral problems in patients presenting with speech/language delay. It should also be kept in mind that neurodevelopmental disorders like ADHD may be accompanied by comorbid conditions. In the medical history, particular attention should be given to the perinatal process, and neurological clues such as microcephaly, dysmorphic features, hypotonia, verbal agnosia/aphasia, and skin findings should be thoroughly investigated during physical examination.

## Ethics

**Ethics Committee Approval:** Ethical approval was obtained from the Clinical Research Ethics Committee of the University of Health Sciences Turkey, Kanuni Sultan Süleyman Training and Research Hospital (approval number: 2022/05, date: 01.10.2022).

**Informed Consent:** Retrospective study.

## Author Contributions

Surgical and Medical Practices: B.D.D., E.T., Concept: B.D.D., E.T., Design: B.D.D., E.T., Data Collection and Processing: B.D.D., E.T., Analysis and Interpretation: B.D.D., Literature Search: B.D.D., Writing: B.D.D., E.T.

**Conflict of Interest:** The authors have no conflict of interest to declare.

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