SELCUK UNIVERSITY PRESS

GENEL TIP DERGİSİ
The Journal of General Medicine

Received: 16 Oct 2025 | **Accepted:** 04 Mar 2026 | **Published:** 05 Aug 2026 **Genel Tıp Derg. Volume** 36(2026), 1-5

DOI: 10.54005/geneltip.1805086

CASE REPORT

West Syndrome Associated with a *CLTC* Gene Variant: A Case Report on Auditory Perception and Language Development

CLTC Gen Varyantı ile İlişkili West Sendromu: İşitsel Algı ve Konuşma Gelişimi Açısından Bir Olgu Sunumu

[1]Esma ÇAĞAL KARABULUT, [2]Özge ÇAĞLAR ÇİL, [3]Selma YILAR, [4]Turgay ÇOKYAMAN, [1]Beyza AKTİ, [5]Ahmet KABLAN, [6]Kübra Müge ÇELİK, [7]Özge ÖZCAN, [6]Fatma SILAN

[1]Audiology Unit, Department of Otorhinolaryngology, Faculty of Medicine, Çanakkale Onsekiz Mart University, Çanakkale, Türkiye
[2]Department of Otorhinolaryngology, Faculty of Medicine, Çanakkale Onsekiz Mart University, Çanakkale, Türkiye
[3]Department of Audiology, Faculty of Health Sciences, Sakarya Applied Science University, Sakarya, Türkiye
[4]Division of Pediatric Neurology, Department of Child Health and Diseases, Faculty of Medicine, Çanakkale Onsekiz Mart University, Çanakkale, Türkiye
[5]Department of Medical Genetics, Ankara Etlik City Hospital, Ankara, Türkiye
[6]Department of Medical Genetics, Faculty of Medicine, Çanakkale Onsekiz Mart University, Çanakkale, Türkiye
[7]Audiology Unit, Department of Otorhinolaryngology, Çanakkale Mehmet Akif Ersoy State Hospital, Çanakkale, Türkiye

Correspondence
Esma Çağal Karabulut
Prof. Dr. Sevim Buluç Sokak, Barbaros Mahallesi, No:2, 17020, Çanakkale, Türkiye

E-Mail: eesmacagal@gmail.com

How to cite ?
Çağal Karabulut E. Çağlar Çil Ö. Yılar S. Çokyaman T. Akti B. Kablan A. Çelik KM. Özcan Ö. Sılan F. West Syndrome Associated with a CLTC Gene Variant: A Case Report on Auditory Perception and Language Development. Genel Tıp Derg. 36(2026), 1-5

Highlights

- Despite normal hearing levels, patients with West syndrome may exhibit significant delays in auditory perception, as well as receptive and expressive language skills.
- In individuals with genetic mutations, it is clinically critical to evaluate not only hearing thresholds but also auditory perception, as well as receptive and expressive language abilities in detail.
- The rehabilitation process and regular audiological follow-up should be conducted using a multidisciplinary approach to ensure early intervention and the development of individualized treatment plans.

ABSTRACT

Aim: To present a rare case of West syndrome (WS) carrying a heterozygous pathogenic *CLTC* variant and to evaluate the possible relationship between this genetic finding and the patient's neurodevelopmental, auditory, and language profile.
Materials and Methods: The patient, a 5-year-and-1-month-old child, was referred to our clinic for a hearing evaluation and underwent a comprehensive assessment. Brain development was examined using magnetic resonance imaging (MRI). Hearing was evaluated through acoustic immittance audiometry (including tympanometry and acoustic reflex measurements), transient otoacoustic emissions (TEOAE), and auditory brainstem response (ABR) testing. Receptive and expressive language abilities were assessed with the Test of Early Language Development–Third Edition (TELD-3), and auditory perception skills were evaluated using the Children's Auditory Perception Test (CIAT). The Denver II Developmental Screening Test was also administered to assess the child's overall developmental status.
Results: The patient was diagnosed with WS at the age of one year. Clinical findings included cerebellar atrophy, muscle weakness in both upper and lower extremities, epilepsy, facial anomalies, visual impairment, neurodevelopmental delay, and stereotypic movements. Genetic testing identified a heterozygous pathogenic *CLTC* variant (c.2669C>T, p.P890L), which is considered the primary variant associated with the patient's clinical presentation. Additionally, three variants of uncertain significance (VUS) were detected, which may have contributed to the neurodevelopmental phenotype. Audiological evaluation revealed normal bilateral hearing; however, auditory perception as well as receptive and expressive language development were significantly delayed. The patient was referred for rehabilitation to support auditory and language acquisition.
Conclusions: In this case, delayed auditory perception as well as speech and language development were evident despite normal hearing levels. In summary, detailed evaluation of auditory perception, speech, and language skills, alongside hearing thresholds, should be emphasized in patients with WS.

Keywords: Auditory perception, genetic, infantile spasms, language development, mutation, West syndrome.

ÖZ

Amaç: Çalışmamızda heterozigot patojenik *CLTC* varyantını taşıyan nadir bir West sendromu (WS) tanılı nadir görülen bir olgu sunmak ve bu genetik bulgu ile hastanın nörogelişimsel, işitsel ve dil profili arasındaki olası ilişkiyi değerlendirmek amaçlandı.
Gereç ve Yöntemler: Hasta 5 yaş 1 aylık erkek hasta olup kliniğimize işitme değerlendirilmesi amacıyla başvurdu. Ayrıntılı bir değerlendirmeye tabi tutuldu. Olgunun beyin gelişimi, Manyetik Rezonans Görüntüleme (MRG) ile değerlendirildi. İşitmesinin değerlendirilmesi için Akustik immitansmetri, Transient Otoakustik Emisyon (TEOAE) ve İşitsel Beyin Sapı Cevapları (ABR) testleri uygulandı. Buna ek olarak Türkçe Erken Dil Gelişim Testi (TEDİL) ile dil becerileri, Çocuklar İçin İşitsel Algı Testi (ÇİAT) ile işitsel algı becerileri değerlendirildi. Genel gelişim düzeyinin değerlendirilmesi için DENVER-II gelişimsel tarama testi uygulandı.
Bulgular: Hasta 5 yaş 1 aylık olup kliniğimize işitme değerlendirilmesi için yönlendirilmiştir. Hastanın 1 yaşından itibaren WS tanısı mevcuttur. Hastada görülen klinik bulgular arasında serebellar yapılarda atrofi, üst ve alt ekstremitelerde kas güçsüzlüğü, epilepsi, yüz anomalileri, görme bozuklukları, nörogelişimsel gelişim geriliği ve stereotipik hareketler yer almaktadır. Genetik incelemede, hastanın klinik tablosuyla ilişkili olduğu düşünülen heterozigot patojenik bir *CLTC* varyantı (c.2669C>T, p.P890L) saptanmıştır. Buna ek olarak, nörogelişimsel fenotipe katkıda bulunabileceği değerlendirilen üç adet klinik anlamı belirsiz varyant (VUS) tespit edilmiştir. Odyolojik değerlendirmede bilateral normal işitme gözlendi, ancak işitsel algı ile alıcı ve ifade edici dil gelişimi belirgin şekilde gecikmiş olarak değerlendirildi. Hasta işitsel algı ve dil becerilerini desteklemek amacıyla rehabilitasyon programına yönlendirildi.
Sonuçlar: Olgumuzda normal işitme düzeyine rağmen işitsel algı, dil ve konuşma becerilerinde gecikme dikkat çekicidir. Özetle, WS'li hastalarda işitmenin yanı sıra işitsel algı, dil ve konuşma açısından da değerlendirilmesinin önemi vurgulanmalıdır.

Anahtar Kelimeler: Dil gelişimi, genetik, infantil spazm, işitsel algı, mutasyon, West sendromu.

Peer-Review: Double anonymized - Two External
Plagiarism Checks: Yes - intihal.net
Complaints: geneltip@selcuk.edu.tr

INTRODUCTION

West syndrome (WS), also known as infantile spasms, is a rare neurodevelopmental disorder characterized by epileptic spasms that typically begin within the first year of life. The condition was first described by William James West in 1841 [1, 2]. The estimated incidence of WS is approximately 2.49 per 10,000 live births [3], and its prevalence among 10-year-old children has been reported as 1 in 10,000 [4]. The hallmark features of WS include clusters of infantile spasms (sudden contractions), neurodevelopmental delay, and a characteristic disorganized electroencephalographic (EEG) pattern known as hypsarrhythmia [5]. These spasms generally present as brief, symmetric contractions involving the neck, trunk, and extremities. It typically occurs in clusters several times per day, most often during transitions between sleep and wakefulness [6].

Among the genes reported in WS cases, *LGI1* (Leucine-Rich Glioma-Inactivated 1) [7], *TSC1* (TSC Complex Subunit 1) and *TSC2* (TSC Complex Subunit 1) [8] have been associated with auditory function [9]. Although a direct association has not been established, studies have shown that the *CDKL5* (Cyclin-Dependent Kinase-Like 5) gene, which is also reported in various WS cases [10], is linked to delays in speech and language development [11]. Other genes frequently implicated in WS, such as *ARX* (Aristaless-Related Homeobox) [12] and *STXBP1* (Syntaxin-Binding Protein 1) [13], play key roles in central nervous system development [14]. These genes are crucial for neurodevelopment, and mutations affecting them have been broadly implicated in pathophysiological alterations of the musculoskeletal system, central nervous system, visual and auditory pathways, as well as language acquisition and overall growth and development [2, 13, 15].

In WS cases, sensorineural hearing loss [16, 17] speech delay despite normal hearing [18], and auditory processing disorders [19] have been reported. However, the relationship between genetic variants associated with WS and auditory perception or language development remains poorly understood. In particular, cases involving concurrent mutations in multiple neurodevelopmentally relevant genes and their potential impact on hearing and language outcomes are exceedingly rare. Therefore, the aim of the current study was to evaluate hearing and language development in a WS patient carrying a heterozygous pathogenic *CLTC* variant and three neurodevelopmentally impactful VUS variants and to discuss the possible contribution of these variants to auditory and linguistic outcomes.

Case Report

This study presents a rare case of a 5-year-and-1-month-old male patient identified as having a heterozygous pathogenic *CLTC* variant. The patient was diagnosed with WS at the age of one year. Prenatal history revealed no parental consanguinity or familial syndromic disorders. During the perinatal period, the patient was delivered via cesarean section at 42 weeks of gestation, with a birth weight of 4020 grams. According to family reports, postnatal development was characterized by marked delays in achieving gross motor milestones relative to chronological age. Developmental milestones were attained as follows: head control at 8 months, independent sitting between 12 and 18 months, crawling at 2 years and 6 months, supported ambulation at 3 years and 6 months, and independent walking at 4 years and 3 months. These findings indicate a significant global motor developmental delay consistent with the underlying neurodevelopmental pathology. Epileptic seizures reportedly began at approximately seven months of age. At the time of clinical evaluation, the family noted that the patient was being received occupational therapy for approximately two years. Clinical examination revealed cerebellar atrophy, muscle weakness in both upper and lower extremities, epilepsy, facial anomalies including brachycephaly, retrognathia, and a drooping columella, dental caries and malformations, visual impairments, neuromotor developmental delay, and stereotypic movements (Figure 1).

Figure 1. Case of West syndrome. Consent for photograph use was obtained. Clinical examination revealed cerebellar atrophy, muscle weakness in both upper and lower extremities, epilepsy, facial anomalies including brachycephaly, retrognathia, and a drooping columella; dental caries and malformations; visual impairments, neuromotor developmental delay, and stereotypic movements.

Test Protocols and Clinical Observations

The patient, a 5-year-and-1-month-old boy, presented to our clinic for a hearing evaluation. Otoscopic examination revealed normal findings in both ears. A detailed medical history was obtained, and a detailed assessment of general development, language skills, and audiological status was performed. Brain development was assessed using magnetic resonance imaging (MRI). Hearing evaluation included acoustic immittance audiometry (tympanometry and acoustic reflex measurements), Transient otoacoustic emissions (TEOAE), and Auditory Brainstem Response (ABR) testing. Receptive and expressive language abilities were assessed with the Test of Early Language Development–Third Edition (TELD-3), and auditory perception skills were evaluated using the Children's Auditory Perception Test (CIAT). The Denver II Developmental Screening Test was also administered to assess the patient's overall developmental status.

Evaluation of muscular development revealed that the patient was able to maintain head control, sit both with and without support, roll to either side in the supine position, and ambulate independently. However, he was unable to ascend or descend stairs or run without assistance, indicating limitations in gross motor coordination and strength. Assessment of the orofacial and articulatory musculature demonstrated normal tone and strength (5/5) in the masseter, temporalis, and sternocleidomastoid muscles, as well as in the tongue, lips, and cervical muscles. In contrast, the buccinator muscle—one of the principal muscles of the cheek—exhibited mildly reduced strength (4/5), suggesting localized weakness that may contribute to articulatory imprecision. Additionally, dental malalignment was observed, potentially serving as a secondary factor negatively influencing articulatory performance and speech intelligibility.

Genetic Analysis

After DNA isolation from blood, whole-exome sequencing (WES) was performed using the Next Generation Sequencing (NGS) method with the xGen Exome Research Panel v2 kit. The results were analyzed using QIAGEN Clinical Insight Interpret version 7.1.20201218. The evaluation of variations adhered to the recommendations outlined in the 2015 American College of Medical Genetics (ACMG) Standards and Guidelines. Genetic analysis revealed **one pathogenic and three variants of uncertain significance (VUS): a heterozygous pathogenic missense variant** in *CLTC* (**NM_004859.5:** c.2669C>T, p.(Pro890Leu)), and **heterozygous VUS** in *KIF2A* (**NM_001098511.3**): c.1321A>G, p.(Ile441Val)), *LGI1* (**NM_005097.4:** c.749T>C, p.(Val250Ala)), and *TSC2* (**NM_000548.5:** c.3802C>T, p.(Arg1268Cys)).

Magnetic Resonance Imaging (MRI)

Brain MRI revealed bilateral widening of the sylvian fissures and marked atrophy of the bilateral temporal lobes. Additionally, enlargement of the frontal subdural space was observed, accompanied by atrophy in both frontal lobes. Notably, the genu and body of the corpus callosum appeared relatively well preserved, whereas the posterior portion demonstrated significant atrophic changes (Figure 2).

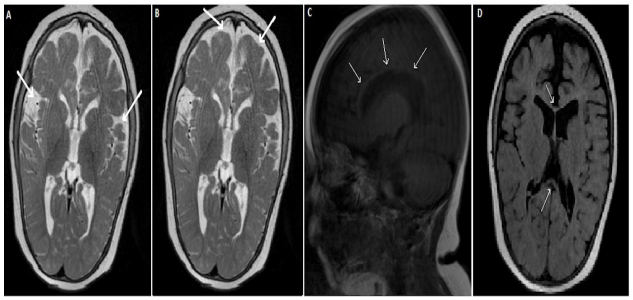

Figure 2. Brain MRI Findings (September 2025). A: Bilateral widening of the Sylvian fissures and marked atrophy of the bilateral temporal lobes are observed. B: Enlargement of the bilateral frontal subdural spaces and atrophy of the bilateral frontal lobes are evident. C: On sagittal T1-weighted images, the genu and body of the corpus callosum appears relatively preserved, whereas the posterior portion demonstrates atrophic changes. D: Axial FLAIR images reveal corpus callosum atrophy.

Acoustic Immittance Audiometry

Bilateral type A tympanograms were obtained, and both ipsilateral and contralateral acoustic reflexes were present, indicating normal middle-ear function.

Otoacoustic Emissions (OAE)

Bilateral transient otoacoustic emissions (TEOAE) were within normal limits, which led to a "pass" result (Figure 3).

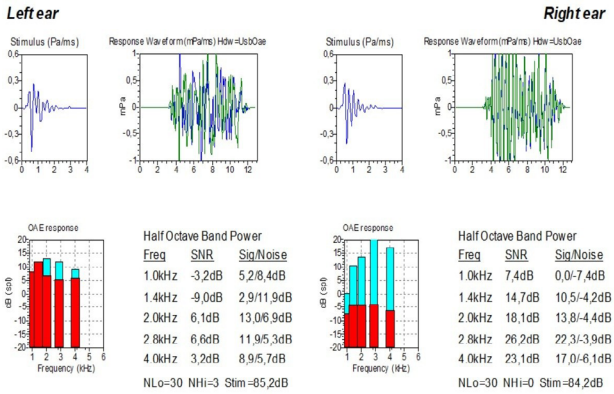

Figure 3. TEOAE results in this case (September 2025).

Auditory Brainstem Response (ABR)

The ABR test, performed using click stimuli, demonstrated bilaterally present wave V responses at 20 dB nHL. Comparison of wave V latencies between the right and left ears at 60 dB nHL and 20 dB nHL revealed an interaural latency difference exceeding 0.4 ms. This degree of asymmetry is considered an electrophysiological indicator suggestive of retrocochlear pathology. The ABR waveforms are presented in Figure 4.

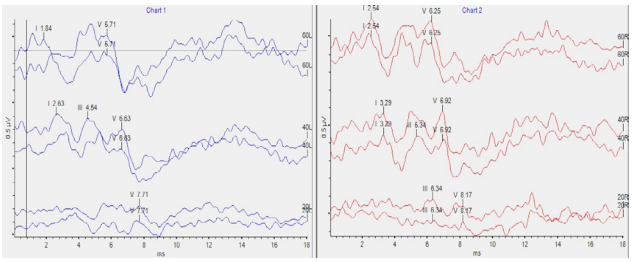

Figure 4. ABR results in this case (September 2025).

Test of Early Language Development (TELD-3)

The patient's language skills were evaluated using the Test of Early Language Development–Third Edition (TELD-3). The assessment revealed a receptive language age equivalent to 2 years and 2 months and an expressive language age of 1 year and 4 months. Considering the chronological age of 5 years and 1 month, these findings indicate a pronounced delay in both receptive and expressive language domains, with expressive abilities being more severely affected.

Children's Auditory Perception Test (CIAT)

To assess auditory perception skills, subtests from the Children's Auditory Perception Test (CIAT) were administered, including pattern perception, word recognition, and sentence recognition. In the pattern perception subtest, the patient achieved a score of

87.5% (21/24), demonstrating the ability to perceive auditory patterns and partially recognize individual words. In the word recognition subtest, accuracy was 70.8% (17/24) for three-syllable words and 58.3% (14/24) for single-syllable words. Sentence recognition performance was 44.4% (8/18). These results indicate a significant delay in auditory perception skills across all subtests compared with typically developing peers. This suggests deficits in higher-order auditory processing and linguistic integration.

Denver II Developmental Screening Test

The patient's overall developmental profile was evaluated using the Denver II Developmental Screening Test. Results revealed that personal-social development corresponded to an age range of 2 years to 2 years and 2 months, fine motor skills were consistent with 1 year and 4 months to 1 year and 7 months, language development corresponded to 1 year and 11 months to 2 years and 2 months, and gross motor skills were equivalent to 1 year and 2 months to 1 year and 6 months. These findings indicate global developmental delay across all domains of the Denver II assessment.

Although the patient demonstrated normal hearing thresholds, a significant delay in auditory perception and language development was evident. Based on these findings, intensive auditory rehabilitation was recommended under the supervision of an audiologist to improve auditory perceptual abilities. In addition, comprehensive speech and language therapy was advised to enhance articulatory muscle coordination and overall linguistic competence. A follow-up evaluation was scheduled for six months later to monitor progress in auditory perception and language development.

Discussion

WS is a rare neurodevelopmental disorder associated with genetic mutations that often result in multiple systems involvement and epileptic encephalopathy [1]. In the present case, mutations were identified in the *CLTC, KIF2A, LGI1,* and *TSC2* genes. Among these, mutations in *KIF2A* [20], *LGI1* [7], *TSC1*, and *TSC2* [8] have been previously associated with WS and related epileptic and neurodevelopmental phenotypes. Although CLTC has not been directly associated with WS, variants in this gene have been reported in connection with epilepsy and broader neurodevelopmental disorders [21]. In this case, the existing clinical findings are primarily attributed to the pathogenic *CLTC* variant, which is considered the main target for therapeutic management. However, the potential modulatory effects of the other variants on the clinical phenotype are being monitored over time.

In early-onset epileptic syndromes such as WS, chronic or recurrent epileptic discharges can negatively influence the functional integrity of language-related neural networks, particularly within the frontal and temporal lobes [19, 22-25]. Functional neuroimaging and electrophysiological studies have demonstrated that epileptic activity in WS inhibits cortical plasticity and impedes synaptic reorganization [19, 22, 24, 26]. This disruption compromises the connectivity of brain networks involved in language processing, particularly those responsible for auditory perception and verbal communication. Consequently, individuals with WS often exhibit significant impairments in auditory processing, auditory perception, verbal memory, and both receptive and expressive language abilities [26, 27].

Because epileptic activity in WS typically follows a diffuse pattern, its effects are not confined to a single cortical area but extend across multiple regions, including the frontal, temporal, and parietal lobes [24, 27]. This widespread cortical involvement disrupts neural synchronization- especially within the superior temporal gyrus and associated auditory processing regions- leading to deficits in auditory perception, attention, and verbal information processing. These findings suggest that the language and speech delays observed in WS are primarily attributable to central auditory processing dysfunction arising from diffuse cortical impact, rather than to peripheral hearing loss [27].

In line with previous literature, a prospective study of patients with WS reported that although hearing thresholds were within normal limits in most cases, auditory orienting responses were significantly impaired, and this impairment was correlated with both visual function and neurodevelopmental level [16]. Moreover, while some case reports of WS have indicated normal hearing function [18, 28], auditory perception and language development have not been comprehensively evaluated in those studies.

The present case is distinguished by a comprehensive evaluation encompassing hearing thresholds, language development, overall developmental level, cerebral structural findings, and auditory perception skills. In disorders such as WS, it is essential to conduct not only detailed audiological testing to identify potential hearing loss but also in-depth assessments of auditory processing, language, and speech abilities. In this case, despite normal hearing thresholds, the patient demonstrated marked delays in auditory perception, language, and speech development. This underscores the importance of early monitoring and intervention strategies targeting auditory processing, auditory perception, and language–speech development in children with WS.

Conclusion

Rehabilitation plays a critical role in supporting the development of language and speech skills in individuals with WS. Regular audiological follow-ups are also vital for early detection and intervention, as hearing loss may emerge during the disease course. Moreover, the evaluation and rehabilitation of children with WS should be guided by a multidisciplinary team approach. Collaborative efforts among specialists in pediatric neurology, audiology, speech and language therapy, occupational therapy, physiotherapy, and child psychology are essential to design individualized treatment and rehabilitation programs tailored to each patient's specific needs.

Conflict of interest statement

The authors declare no conflicts of interest.

Financial support

This research received no specific grant from any funding agency in the public, commercial, or not-for-profit sectors.

Acknowledgments

We would like to express our sincere gratitude to our participant and parents for their valuable contributions to this study.

References

1. Mytinger JR. Definitions and Diagnostic Criteria for Infantile Spasms and West Syndrome - Historical Perspectives and Practical Considerations. Semin Pediatr Neurol. 2021;38:100893.

2. Pavone P, Polizzi A, Marino SD, Corsello G, Falsaperla R, Marino S, et al. West syndrome: a comprehensive review. Neurol Sci. 2020;41(12):3547-3562.

3. Jia JL, Chen S, Sivarajah V, Stephens D, Cortez MA. Latitudinal differences on the global epidemiology of infantile spasms: systematic review and meta-analysis. Orphanet J Rare Dis. 2018;13(1):216.

4. Lux AL. Latest American and European updates on infantile spasms. Curr Neurol Neurosci Rep. 2013;13(3):334.

5. D'Alonzo R, Rigante D, Mencaroni E, Esposito S. West Syndrome: A Review and Guide for Paediatricians. Clin Drug Investig. 2018;38(2):113-24.

6. Mackay MT, Weiss SK, Adams-Webber T, Ashwal S, Stephens D, Ballaban-Gill K, et al. Practice parameter: medical treatment of infantile spasms: report of the American Academy of Neurology and the Child Neurology Society. Neurology. 2004;62(10):1668-81.

7. van der Knoop MM, Maroofian R, Fukata Y, van Ierland Y, Karimiani EG, Lehesjoki AE, et al. Biallelic ADAM22 pathogenic variants cause progressive encephalopathy and infantile-onset refractory epilepsy. Brain. 2022;145(7):2301-12.

8. Chu-Shore CJ, Major P, Camposano S, Muzykewicz D, Thiele EA. The natural history of epilepsy in tuberous sclerosis complex. Epilepsia. 2010;51(7):1236-41.

9. Fu X, Sun X, Zhang L, Jin Y, Chai R, Yang L, et al. Tuberous sclerosis complex–mediated mTORC1 overactivation promotes age-related hearing loss. J Clin Invest. 2018;128(11):4938-4955.

10. Kluckova D, Kolnikova M, Medova V, Bognar C, Foltan T, Svecova L, et al. Clinical manifestation of CDKL5 deficiency disorder and identified mutations in a cohort of Slovak patients. Epilepsy Res. 2021;176:106699.

11. Fehr S, Leonard H, Ho G, Williams S, de Klerk N, Forbes D, et al. There is variability in the attainment of developmental milestones in the CDKL5 disorder. J Neurodev Disord. 2015;7(1):2.

12. Kato M, Saitoh S, Kamei A, Shiraishi H, Ueda Y, Akasaka M, et al. A longer polyalanine expansion mutation in the ARX gene causes early infantile epileptic encephalopathy with suppression-burst pattern (Ohtahara syndrome). Am J Hum Genet. 2007;81(2):361-6.

13. Takeda K, Miyamoto Y, Yamamoto H, Iwasaki T, Sumitomo N, Takeshita E, et al. Mutation in the STXBP1 Gene Associated with Early Onset West Syndrome: A Case Report and Literature Review. Pediatr Rep. 2022;14(4):386-95.

14. Jaafar A, Alsiddiq F, Ling KH. Pathogenic mutations in ARX, CDKL5 and STXBP1 genes are not associated with the early-onset epileptic encephalopathy in Malaysian pediatric patients: A pilot study. Neurosci Res Notes. 2018;1(3):5-17.

15. Caglayan AO, Gumus H, Kato M. Two female siblings with West syndrome: Familial idiopathic West syndrome with genetic susceptibility and variable phenotypic expression. J Pediatr Neurosci. 2010;5(2):147-9.

16. Baranello G, Randò T, Bancale A, D'Acunto MG, Epifanio R, Frisone MF, et al. Auditory attention at the onset of West syndrome: correlation with EEG patterns and visual function. Brain Dev. 2006;28(5):293-9.

17. Kaushik JS, Patra B, Sharma S, Yadav D, Aneja S. Clinical spectrum and treatment outcome of West Syndrome in children from Northern India. Seizure. 2013;22(8):617-21.

18. Sharma M, Parmar M, Kumar S. Speech Language and Hearing Aspects in West Syndrome: A Single-Case Report. Journal of Integrated Health Sciences. 2023;11(1):63-5.

19. Werner K, Fosi T, Boyd SG, Baldeweg T, Scott RC, Neville BG. Temporal lobe impairment in West syndrome: event-related potential evidence. Ann Neurol. 2015;77(1):47-57.

20. Tian G, Cristancho AG, Dubbs HA, Liu GT, Cowan NJ, Goldberg EM. A patient with lissencephaly, developmental delay, and infantile spasms, due to de novo heterozygous mutation of KIF2A. Mol Genet Genomic Med. 2016;4(6):599-603.

21. Helbig KL, Farwell Hagman KD, Shinde DN, Mroske C, Powis Z, Li S, et al. Diagnostic exome sequencing provides a molecular diagnosis for a significant proportion of patients with epilepsy. Genet Med. 2016;18(9):898-905.

22. Aydinli N, Calişkan M, Ozmen M, Tonguç E. Neuroradiologic aspects of West syndrome. Pediatr Neurol. 1998;19(3):211-6.

23. Çetin ÖE, Yalçınkaya C, Karaman B, Demirbilek V, Tüysüz B. Chromosome 14q11. 2-q21. 1 duplication: a rare cause of West syndrome. Epileptic Disord. 2018;20(3):219-24.

24. Hattori H, Yamano T, Tsutada T, Tsuyuguchi N, Kawawaki H, Shimogawara M. Magnetoencephalography in the detection of focal lesions in West syndrome. Brain Dev. 2001;23(7):528-32.

25. Hughes JR, Rechitsky I, Daaboul Y. Long term changes in patients with hypsarrhythmia-infantile spasms: 505 patients, up to 43 years follow-up. Clin Electroencephalogr. 1997;28(1):1-15.

26. Enatsu R, Kubota Y, Kakisaka Y, Bulacio J, Piao Z, O'Connor T, et al. Reorganization of posterior language area in temporal lobe epilepsy: a cortico-cortical evoked potential study. Epilepsy Res. 2013;103(1):73-82.

27. Fosi T, Chu C, Chong WK, Clark C, Scott RC, Boyd S, et al. Quantitative magnetic resonance imaging evidence for altered structural remodeling of the temporal lobe in W est syndrome. Epilepsia. 2015;56(4):608-16.

28. Mohan V. West Syndrome: A Case Study Through Audiological Perspective. Int J Health Sci Res. 2022;12(7):115-117.