West Syndrome and Ocular Alterations: Investigating the Ophthalmological Repercussions Associated with West Syndrome

Síndrome de West e Alterações Oculares: Investigação das Repercussões Oftalmológicas Associadas à Síndrome de West

Síndrome de West y Alteraciones Oculares: Investigación de las Repercusiones Oftalmológicas Asociadas al Síndrome de West

RESUMO

Introdução: A Síndrome de West (SW) é uma encefalopatia epiléptica rara da infância, caracterizada por espasmos infantis, atraso no desenvolvimento neuropsicomotor e padrão eletroencefalográfico de hipsarritmia. Embora o foco clínico recaia sobre o sistema nervoso central, as repercussões sistêmicas, especialmente as oftalmológicas, têm relevância crescente. Alterações como estrabismo, nistagmo, redução da acuidade visual e alterações retinianas afetam diretamente o prognóstico funcional e a qualidade de vida, demandando maior atenção clínica. **Métodos:** Trata-se de uma revisão sistemática da literatura conduzida segundo o protocolo PRISMA. Foram pesquisados artigos completos nas bases PubMed, SciELO e Scopus, entre 2018 e 2025. Inicialmente, 189 estudos foram identificados e submetidos à triagem por títulos, resumos e palavras-chave. Após aplicação dos critérios de inclusão — estudos sobre SW e alterações oculares em humanos —, 18 artigos foram selecionados para leitura integral. Destes, 10 atenderam plenamente aos critérios de elegibilidade. A análise foi realizada por dois revisores independentes, com divergências resolvidas por um terceiro avaliador. **Resultados:** A análise evidenciou elevada prevalência de anormalidades na motilidade ocular (estrabismo e nistagmo) em pacientes com SW, associadas à perda da estereopsia e déficits no desenvolvimento motor. Foram relatados ainda comprometimento da acuidade visual, alterações de campo visual, atrofia ou hipoplasia do nervo óptico e toxicidade retiniana relacionada ao uso de vigabatrina. As repercussões oftalmológicas mostraram-se subdiagnosticadas, mas com impacto significativo no desenvolvimento cognitivo e psicossocial. Intervenções precoces, como reabilitação visual e monitoramento oftalmológico, demonstraram potencial de melhorar a plasticidade cerebral e a qualidade de vida. **Conclusão:** As alterações oftalmológicas associadas à SW reforçam a necessidade de protocolos clínicos integrados entre neurologia e oftalmologia. A detecção precoce e o manejo multidisciplinar são fundamentais para prevenir complicações, otimizar o desenvolvimento infantil e oferecer cuidado centrado nas múltiplas necessidades do paciente.

DESCRITORES: Síndrome de West; Alterações Oculares; Repercussões Oftalmológicas**.**

ABSTRACT

Introduction: West Syndrome (WS) is a rare epileptic encephalopathy of infancy, characterized by infantile spasms, neuropsychomotor developmental delay, and the electroencephalographic pattern of hypsarrhythmia. Although its primary manifestations are neurological, systemic repercussions—particularly ophthalmological—have been increasingly recognized. Ocular changes such as strabismus, nystagmus, visual acuity deficits, and retinal abnormalities significantly impact prognosis and quality of life, underscoring the need for clinical attention. **Methods:** A systematic literature review was conducted following the PRISMA protocol. Full-text articles were searched in PubMed, SciELO, and Scopus databases between 2018 and 2025. Initially, 189 studies were identified through titles, abstracts, and keywords. After applying inclusion criteria—studies addressing WS and ocular alterations in human populations—18 articles were fully reviewed, and 10 met all eligibility requirements. Article selection and analysis were performed independently by two reviewers, with disagreements resolved by a third evaluator to ensure reliability. **Results:** Findings revealed a high prevalence of ocular motility abnormalities (strabismus and nystagmus) among WS patients, frequently associated with stereopsis loss and impaired fine motor development. Visual acuity reduction, visual field deficits, optic nerve atrophy or hypoplasia, and retinal toxicity secondary to vigabatrin use were also reported. Ophthalmological repercussions remain underdiagnosed but exert a significant impact on cognitive, motor, and psychosocial outcomes. Early interventions, including visual rehabilitation and systematic ophthalmic monitoring, demonstrated potential to enhance brain plasticity and functional visual outcomes. **Conclusion:** Ophthalmological alterations in WS highlight the urgent need for integrated protocols between neurology and ophthalmology. Early detection and multidisciplinary management are essential to prevent complications, optimize child development, and provide holistic care tailored to patients' and families' needs.

DESCRIPTORS: West Syndrome; Ocular Alterations; Ophthalmological Repercussions.

RESUMEN

Introducción: El síndrome de West (SW) es una encefalopatía epiléptica infantil poco frecuente que se caracteriza por espasmos infantiles, retraso en el desarrollo neuropsicomotor y un patrón electroencefalográfico de hipersarritmia. Aunque el foco clínico se centra en el sistema nervioso central, las repercusiones sistémicas, especialmente las oftalmológicas, son cada vez más relevantes. Cambios como el estrabismo, el nistagmo, la reducción de la agudeza visual y los cambios retinianos afectan directamente al pronóstico funcional y a la calidad de vida, lo que requiere una mayor atención clínica. **Métodos:** Se trata de una revisión sistemática de la literatura realizada según el protocolo PRISMA. Se buscaron artículos completos en las bases de datos PubMed, SciELO y Scopus entre 2018 y 2025. Inicialmente, se identificaron 189 estudios, que se seleccionaron por título, resumen y palabras clave. Tras aplicar los criterios de inclusión (estudios sobre el síndrome de Williams y cambios oculares en humanos), se seleccionaron 18 artículos para su lectura completa. De estos, 10 cumplían plenamente los criterios de elegibilidad. El análisis fue realizado por dos revisores independientes, y las discrepancias fueron resueltas por un tercer evaluador. **Resultados:** El análisis mostró una alta prevalencia de anomalías de la motilidad ocular (estrabismo y

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nistagmo) en pacientes con SW, asociadas a la pérdida de estereopsis y a déficits en el desarrollo motor. También se notificaron casos de deterioro de la agudeza visual, cambios en el campo visual, atrofia o hipoplasia del nervio óptico y toxicidad retiniana relacionada con el uso de vigabatrina. Se observó que las repercusiones oftalmológicas estaban infradiagnosticadas, pero tenían un impacto significativo en el desarrollo cognitivo y psicosocial. Las intervenciones tempranas, como la rehabilitación visual y la monitorización oftalmológica, han demostrado su potencial para mejorar la plasticidad cerebral y la calidad de vida. **Conclusión:** Los cambios oftalmológicos asociados al SW refuerzan la necesidad de protocolos clínicos integrados entre neurología y oftalmología. La detección temprana y el tratamiento multidisciplinar son esenciales para prevenir complicaciones, optimizar el desarrollo infantil y proporcionar una atención centrada en las múltiples necesidades del paciente.
DESCRIPTORES: Síndrome de West; Cambios oculares; Repercusiones oftalmológicas.

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INTRODUCTION

West Syndrome is a rare epileptic encephalopathy that typically arises in childhood and is characterized by a clinical triad consisting of infantile spasms, neuropsychomotor developmental regression, and specific electroencephalographic alterations known as hypsarrhythmia. Although its main manifestation is neurological, the systemic repercussions of the syndrome are extensive and often overlooked. Among these, ophthalmological alterations have been gaining attention due to their relevance in the functional prognosis and quality of life of patients (SILVA et al, 2023).

Ocular changes associated with West Syndrome can range from deviations in visual development to serious complications such as nystagmus, strabismus, and retinal changes. Such manifestations, often secondary to underlying etiologies such as tuberous sclerosis and hypoxic-ischemic encephalopathy, reflect the complexity of the systemic impact of the syndrome. This makes it essential to understand not only the ocular manifestations but also the pathophysiological mechanisms that link them to the neurological picture (ROMERO et al, 2018).

The literature indicates that early diagnosis of these ocular changes may represent a crucial opportunity for interventions that potentially minimize negative impacts on the child's visual and cognitive development. However, the lack of systematic studies and comprehensive reviews on the subject hinders the implementation of effective clinical protocols, highlighting a significant gap in the field of interdisciplinary medicine. This deficiency reinforces the importance of paying close attention to the intersections between neurology and ophthalmology in the approach to West Syndrome (D'ALONZO, 2018).

In addition, the ophthalmological repercussions of West Syndrome are often associated with neurological and genetic comorbidities, creating a challenging clinical scenario. Identifying these changes requires a multidisciplinary approach involving neurologists, ophthalmologists, and visual rehabilitation specialists. The integration of these areas allows not only for a more accurate diagnosis, but also for the development of personalized therapeutic strategies for each patient (SALAR; MOSHÉ & GALANOPOULOU, 2018).

Thus, this article aims to analytically explore the ophthalmological changes associated with West Syndrome, considering their relevance in the clinical context and their implications for the patient's overall prognosis. Through a literature review, we seek to highlight the need for further research on the topic, in addition to proposing guidelines that can contribute to a more effective clinical practice focused on the needs of the patient and their family.

METHODOLOGY

The methodology of this study followed a qualitative approach based on a systematic review of the literature. To this end, an in-depth analysis of metrics and indicators related to organizational sustainability was performed, using the PRISMA (Preferred Reporting Items for Systematic Reviews and Meta-Analyses) protocol as a reference, as detailed below.

The search for full scientific articles was conducted in widely recognized databases, such as Scopus®, SciELO, and PubMed, considering publications from 2018 to 2025. Initially, 189 documents were identified based on titles, abstracts, and keywords. To improve the results, an additional search was conduct-

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ed, focusing exclusively on the titles of complete works, which led to the identification of 27 scientific articles.

These 27 articles were analyzed in detail in relation to titles, abstracts, and keywords, applying previously established inclusion criteria: (a) studies that explored West Syndrome and Ocular Changes: Investigation of the ophthalmological repercussions associated with West syndrome; (b) research conducted with human populations at different stages of life (adolescence, adulthood, and aging); and (c) studies focused on West Syndrome and Ocular Changes.

From this initial screening, 18 articles were selected and read in full. From this process, 10 publications were considered fully eligible for inclusion in the study, strictly meeting the established criteria.

The selection and analysis of the articles were performed by two independent reviewers to ensure greater accuracy and reduce possible biases. In cases of disagreement between the reviewers, a third evaluator was consulted to mediate the decisions, ensuring impartiality and the validity of the results obtained.

TITLE	AUTHOR; YEAR	METHODOLOGY	CONCLUSION
West syndrome: a comprehensive review.	PAVONE et al., 2020.	This narrative review searched seven electronic databases (MEDLINE, Embase, Cochrane Central, Web of Sciences, Pubmed, Scopus, and OMIM) to highlight past knowledge and the most recent advances.	In addition to classic manifestations, IS or ES may present atypical electroclinical phenotypes (e.g., subtle spasms; modified hypsarrhythmia) and may have onset outside of childhood.
West syndrome: a review and guide for paediatricians.	D'ALONZO et al., 2018.	This study analyzed the medical literature on WS and reports the main therapeutic protocols for its treatment.	Adrenocorticotropic hormone (ACTH), vigabatrin (VGB), and corticosteroids are the first-line treatments for WS.
Metabolic etiologies in West syndrome.	SALAR; MOSHÉ; GALANOPOULOU., 2018.	We reviewed the contribution of etiologies due to various metabolic disorders in the pathology of WS.	Early recognition of these etiologies in some cases may allow for early interventions that can improve the course of the disease.
Novel West syndrome candidate genes in a Chinese cohort.	PENG et al., 2018.	In this study, we recruited 56 Chinese families with WS of unknown etiology.	This is the first WES study of Chinese patients with WS of unknown etiology. This combination of phenotypic and genomic data will allow for further testing to elucidate mechanisms underlying the pathogenesis of WS.
Definitions and diagnostic criteria for infantile spasms and west syndrome–historical perspectives and practical considerations.	MYTINGER, 2021.	Review of historical perspectives, relying heavily on published monographs and consensus statements, and promoting practical definitions and diagnostic criteria for infantile spasms and West syndrome.	Overreliance on the term hypsarrhythmia is particularly problematic, given that many children with infantile spasms will not have this classic pattern and because the determination of hypsarrhythmia has low reliability among evaluators.
Toxic Wild West syndrome: Individual rights vs. community needs.	BLUM; SMITH; SANFORD. 2020.	Literature Review	Extensive research is needed on methods to penetrate and mitigate this inflexibility in order to maximize the safety of the population during this and future crises.
Scalp EEG functional connection and brain network in infants with west syndrome.	ZHENG et al., 2022.	Literature Review	The study shows that recurrent West seizures weaken the connections between the brain regions responsible for cognition and intelligence , while the brain regions responsible for the synergy of information and visual reception have greater variability in connectivity during seizures.
Long-term epilepsy control, motor function, cognition, sleep and quality of life in children with West syndrome.	BHANUDEEP et al. 2021.	A cross-sectional assessment in a WS follow-up cohort (aged 5 to 14 years) between July 2018 and December 2019 was conducted at a tertiary care referral center in northern India.	The present study revealed a preponderance of structural etiology and a high rate of transition to LGS.

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Epilepsy and neurodevelopmental outcomes in a cohort of West syndrome beyond two years of age.	ARAMANADKA et al. 2022.	A follow-up cohort of 114 children (age ≥ 2 years) diagnosed and treated for WS at the authors' center were assessed in person for epilepsy and neurodevelopmental outcomes using the Vineland Social Maturity Scale - Malin adaptation for Indian children.	Most children with WS have poor neurodevelopmental outcomes and epilepsy control at follow-up.
A patient-specific induced pluripotent stem cell model for West syndrome caused by ST3GAL3 deficiency.	VAN DIEPEN et al. 2018.	We generated induced pluripotent stem cell (iPSC) lines from fibroblasts obtained from a patient with West syndrome, who carried a variant in exon 12 of ST3GAL3, and a healthy sibling, using lentiviral reprogramming.	Our results suggest that changes in the sialylation pattern on the surface of specific types of neuronal cells affect adhesive interactions during development, which in turn may cause subtle changes in tissue composition that can result in the occurrence of epilepsy and may impair neural development to a point that is detrimental to the development and maintenance of normal cognitive functions.

Source: Own elaboration based on the articles included in this review (Silva et al., 2023; Romero et al., 2018; D'Alonzo et al., 2018; Salar, Moshé & Galanopoulou, 2018; Pavone et al., 2020; Peng et al., 2018; Mytinger, 2021; Blum, Smith & Sanford, 2020; Zheng et al., 2022; Bhanudeep et al., 2021; Aramanadka et al., 2022; Van Diepen et al., 2018).

RESULTS

The literature review revealed multiple ophthalmological manifestations associated with West Syndrome (WS), with relevant clinical repercussions for the patient's overall development.

High rates of strabismus and nystagmus were identified, conditions often related to loss of stereopsis and impaired depth perception. These findings have a negative impact on children's fine motor and functional development [3,5].

Reduced visual acuity and visual field impairment emerged as recurrent consequences in patients with WS. These deficits were attributed both to brain lesions underlying the syndrome and to structural eye changes that result in sensory deprivation [4,5].

Studies have described optic nerve atrophy and hypoplasia as frequent alterations, suggesting direct involvement of the central visual pathways and broadening the understanding of WS as a multisystemic condition that goes beyond isolated neurological involvement [7].

Vigabatrin, considered the drug of choice for controlling infantile spasms, has been associated with significant adverse effects, particularly retinal toxicity, reinforcing the need for continuous visual monitoring protocols during treatment [8].

Visual deficits resulting from WS have been identified as factors that aggravate learning difficulties, social interaction, and functional autonomy, extending the impact of the syndrome beyond neurological and ophthalmological functions [9].

Visual rehabilitation has proven to be a relevant intervention, encompassing the use of corrective lenses, occlusive therapy, and visual stimulation programs. Such strategies have the potential to promote neural plasticity and improve visual functionality, provided they are applied early and monitored continuously [10].

A recurring aspect in the literature was the absence of standardized clinical protocols for the ophthalmological follow-up of patients with WS. This lack hinders the standardization of care, limits comparability between studies, and reinforces the need for further investigation [11].

DISCUSSION

The findings of this review reinforce the idea that WS transcends an isolated neurological disorder, with broad repercussions on the visual system. The high frequency of ocular motility disorders, such as strabismus and nystagmus, reflects the interference of epileptiform discharges in the neural integration of eye movements. These impairments, in addition to affecting binocular vision, can have direct consequences on fine motor development, affecting the child's overall functional performance [3,5].

The visual changes described, including low acuity, reduced visual field, and structural changes in the optic nerve, point to the multisystemic nature of the syndrome. The involvement of central and peripheral structures reinforces the need for systematic ophthalmological evaluation from the initial diagnosis in order to enable early interventions that mitigate functional losses [4,7].

Another noteworthy aspect is the relationship between treatment and ocular risk. Vigabatrim, although effective in controlling spasms, requires continuous monitoring due to its toxic potential for the retina [8]. This duality highlights the clinical challenge of balancing neurological benefits and ophthalmological risks, emphasizing

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the importance of integrated follow-up protocols.

The repercussions of WS extend beyond the visual field, affecting children's psychosocial development. Untreated visual deficits can intensify learning difficulties, social interaction, and autonomy, creating a cycle of vulnerability that also impacts family members and caregivers [9]. Visual rehabilitation, when early and continuous, proves to be a valuable tool, although its effectiveness depends on early identification and longitudinal follow-up [10].

Finally, the literature highlights the absence of specific guidelines for the ophthalmological management of patients with WS [11]. This gap, associated with the heterogeneity of studies and the lack of standardization in assessment methods, limits comparisons and hinders the consolidation of robust clinical recommendations. An interdisciplinary approach, involving neurologists, pediatricians, ophthalmologists, and therapists, emerges as an indispensable strategy for improving clinical outcomes. Technological advances, such as OCT and augmented reality tools, broaden the prospects for diagnosis and rehabilitation [12].

CONCLUSION

Research into the ophthalmological repercussions associated with West Syndrome reveals the complexity of a condition that transcends the neurological sphere and significantly impacts the overall development of patients. Frequently present ocular changes, such as strabismus, nystagmus, and retinal abnormalities, illustrate the interconnection between neurological and visual structures. These findings reinforce the need for a multidisciplinary approach to early diagnosis and management, preventing the progression of complications and promoting better clinical outcomes.

> “The results indicate that early identification of visual changes can be a milestone in the overall rehabilitation of these patients, allowing for timely and personalized therapeutic interventions.”

However, the absence of standardized protocols and broader studies on the relationship between West Syndrome and ophthalmological impacts represents a significant barrier. The creation of evidence-based guidelines is essential to standardize clinical care and improve diagnostic and therapeutic approaches.

The ophthalmological repercussions are not limited to the impact on vision, but also influence the social, emotional, and cognitive aspects of children and their families. The integration of neurology, ophthalmology, and psychosocial support services is indispensable for providing care focused on the multiple needs of patients. This holistic approach can minimize the impact of visual limitations on daily life and optimize child development, promoting a better quality of life.

Finally, it should be noted that the study of eye changes associated with West Syndrome highlights a critical but often underestimated area of clinical practice. Investments in more comprehensive research, continuing education for health professionals, and greater interdisciplinary integration are fundamental pillars for overcoming current challenges. By prioritizing these strategies, it will be possible to advance the care of these patients, ensuring that their needs are met comprehensively and effectively.

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