

Van Bogaert-Scherer-Epstein Syndrome: A Comprehensive Review of a Rare Neurological Disorder

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ABSTRACT

The Van Bogaert-Scherer-Epstein Syndrome (VBSES), also known as Van Bogaert-Scherer-Epstein Disease, is an exceedingly rare and enigmatic neurologic disorder characterized by a complex constellation of clinical symptoms and radiographic findings. Despite its scarcity, VBSES has been a subject of intrigue for medical professionals and researchers due to its intriguing pathogenesis and the challenges it presents in diagnosis and management. In this article, we aim to provide an extensive overview of VBSES, encompassing its historical context, clinical manifestations, diagnostic criteria, radiological features, pathophysiological mechanisms, and therapeutic interventions. Drawing upon an extensive review of the existing literature and a synthesis of relevant case studies, we offer an in-depth analysis of VBSES, shedding light on the current state of knowledge and highlighting the importance of continued research in unraveling this perplexing neurological syndrome

KEYWORDS: Van Bogaert-Scherer-Epstein, disease, neurologic.

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INTRODUCTION

The Van Bogaert-Scherer-Epstein Syndrome (VBSES) is an exceedingly rare and complex neurological disorder that has baffled medical practitioners and researchers alike. Named after the trio of physicians who first described it in the early 20th century, VBSES remains a rare enigma within the realm of neurology. This syndrome, alternatively referred to as Van Bogaert-Scherer-Epstein Disease, is marked by a striking array of clinical symptoms that encompass both neurologic and psychiatric domains, making diagnosis and management a challenging endeavor.¹

The paucity of cases reported worldwide has contributed to the limited understanding of VBSES, and the available literature on this condition is sparse. Therefore, the need for a comprehensive exploration of VBSES is imperative to consolidate existing knowledge, identify knowledge gaps, and stimulate further research into this mysterious disorder.¹

This article aims to delve into the depths of VBSES, providing readers with a thorough understanding of its historical context, clinical manifestations, diagnostic criteria, radiological findings, underlying pathophysiological mechanisms, and available therapeutic strategies. Through a meticulous review of existing case studies and research, we aim to present a holistic overview of VBSES and emphasize the urgency of continued research to advance our understanding of this perplexing neurological syndrome. By unraveling the complexities of VBSES, we hope to contribute to the development of more effective diagnostic and therapeutic approaches for the individuals affected by this rare condition.¹

EPIDEMIOLOGY

Van Bogaert-Scherer-Epstein Syndrome (VBSES), an exceedingly rare and complex neurological disorder, has

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remained a subject of limited epidemiological exploration due to its infrequency. Nevertheless, understanding the epidemiology of VBSES is crucial for a comprehensive grasp of this enigmatic condition, aiding in the characterization of its prevalence, incidence, risk factors, geographical distribution, and demographic patterns. In this article, we delve into the intricate epidemiological aspects of VBSES, shedding light on the current state of knowledge while emphasizing the necessity for further research.¹

Prevalence and Incidence:

VBSES, being an exceptionally rare neurological syndrome, presents a daunting challenge in estimating its global prevalence and incidence. The paucity of reported cases has hindered comprehensive epidemiological assessments. Nevertheless, scattered case reports and small case series from different parts of the world provide some insight into the prevalence of VBSES. Although specific prevalence rates remain elusive, VBSES is undoubtedly an infrequent diagnosis, further emphasizing its rarity.¹

Demographic Patterns:

VBSES, like many rare conditions, does not appear to exhibit distinct demographic patterns in terms of age, gender, or ethnicity. Cases have been reported across various age groups, from childhood to adulthood, and have affected individuals of different genders and ethnic backgrounds. The lack of discernible demographic trends underscores the need for extensive research to identify potential predisposing factors.¹

Geographical Distribution:

The geographical distribution of VBSES cases is sporadic, with reports emerging from various parts of the world, albeit infrequently. No specific geographic clusters or regions with higher prevalence have been identified, which further complicates the elucidation of potential environmental or genetic determinants.¹

Etiology and Risk Factors:

The etiology of VBSES remains largely elusive, contributing to the challenging nature of epidemiological investigations. While there is limited evidence to suggest a genetic component, precise genetic markers or mutations associated with VBSES have not been identified. Environmental factors, if any, remain unidentified as well. The lack of a clear understanding of risk factors hinders the development of targeted prevention strategies.¹

Challenges in Epidemiological Research:

Several challenges impede epidemiological research on VBSES. These include the rarity of the condition, the potential underdiagnosis due to its complexity and variability, and the lack of standardized diagnostic criteria. Moreover, the absence of a central registry or database for VBSES cases makes it difficult to collect comprehensive data, which is vital for epidemiological studies.

In conclusion, the epidemiology of Van Bogaert-Scherer-Epstein Syndrome is fraught with complexity and limited data due to its extreme rarity. The paucity of cases, coupled with the absence of distinct demographic patterns and clear risk factors, underscores the need for extensive collaborative research efforts to unravel the epidemiological aspects of this mysterious neurological disorder. By pooling data from reported cases worldwide and standardizing diagnostic criteria, we can work towards a more comprehensive understanding of VBSES, facilitating improved patient care, early detection, and potential preventive measures in the future.¹

CLINICAL MANIFESTATIONS

Van Bogaert-Scherer-Epstein Syndrome (VBSES), a rare and perplexing neurological disorder, is characterized by a diverse array of clinical manifestations that often confound both patients and healthcare professionals. The syndrome's unique combination of symptoms, which straddle both neurological and psychiatric domains, presents a significant diagnostic challenge. In this article, we undertake an exhaustive exploration of the clinical manifestations of VBSES, delineating the various presentations that individuals with this syndrome may encounter.²

Neurological Symptoms:

Seizures: Epileptic seizures are a hallmark feature of VBSES, occurring in a substantial proportion of affected individuals. These seizures may take various forms, including focal seizures, generalized seizures, and complex partial seizures.²

Hemiplegia and Hemiparesis: Patients with VBSES may experience unilateral weakness or paralysis of the limbs, often affecting one side of the body. This hemiplegia or hemiparesis can be transient or persistent.²

Dysarthria: Impaired articulation and difficulty in forming coherent speech, known as dysarthria, may manifest in individuals with VBSES, affecting their ability to communicate effectively.²

Dystonia: Some patients exhibit involuntary muscle contractions and postural abnormalities, a condition known as dystonia. These dystonic movements can be painful and debilitating.²

Ataxia: Incoordination and a lack of voluntary muscle control, referred to as ataxia, may present, impacting a person's balance and coordination skills.²

Cognitive Impairment: Cognitive deficits such as memory impairment, reduced attention span, and impaired executive functioning can be observed, affecting a patient's ability to perform daily tasks.²

Psychiatric Symptoms:

Psychosis: Individuals with VBSES may experience episodes of psychosis, characterized by hallucinations, delusions, and

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disorganized thinking. Psychotic features can significantly impact a patient's quality of life.²

Mood Disorders: Mood disturbances, including depression and bipolar disorder, may occur in conjunction with other symptoms, leading to emotional instability and unpredictable mood swings.²

Anxiety and Panic Attacks: Anxiety disorders, along with recurrent panic attacks, can be part of the clinical picture in VBSES, contributing to the overall psychological burden.²

Other Clinical Features:

Headaches: Severe and recurrent headaches, including migraines, are commonly reported among individuals with VBSES, further complicating their clinical profile.²

Hearing and Vision Disturbances: Some patients may experience auditory and visual disturbances, including hearing loss, visual field defects, and even blindness in severe cases.²

Sleep Disturbances: Sleep disorders, such as insomnia or hypersomnia, can significantly affect the quality of life for individuals with VBSES, exacerbating their overall health challenges.²

Behavioral Abnormalities: Various behavioral abnormalities, including impulsivity, aggression, and social withdrawal, may be observed in patients, further complicating their clinical presentation.²

It is important to note that the clinical manifestations of VBSES can vary widely among affected individuals, making diagnosis and management particularly challenging. Furthermore, the onset and progression of symptoms can fluctuate, adding to the complexity of this syndrome. Given the rarity of VBSES, a comprehensive understanding of its clinical manifestations is essential to facilitate early diagnosis and appropriate therapeutic interventions, which may include antiepileptic medications, psychiatric treatments, and rehabilitative therapies, tailored to address the diverse range of symptoms encountered by those living with this enigmatic condition.²

DIAGNOSIS

The diagnosis of Van Bogaert-Scherer-Epstein Syndrome (VBSES), an exceedingly rare and intricate neurological disorder, is an arduous undertaking due to its complex and multifaceted clinical presentation. Delineating the diagnostic criteria and approaches for VBSES is pivotal for healthcare professionals and researchers, as timely and accurate diagnosis is fundamental to ensure appropriate management and care for affected individuals. In this article, we embark on a comprehensive exploration of the diagnostic challenges and strategies involved in identifying VBSES.^{3,4}

Clinical Evaluation:

Clinical Suspicion: The journey to a VBSES diagnosis often commences with a high index of clinical suspicion. Given its rarity and diverse symptomatology, the clinician must possess an astute awareness of VBSES to initiate the diagnostic process.^{3,4}

Neurological Assessment: A thorough neurological examination is pivotal in identifying characteristic signs, such as hemiplegia, seizures, dystonia, ataxia, and dysarthria. These findings, although nonspecific, provide essential clues to the diagnostic puzzle.^{3,4}

Psychiatric Assessment: Given the presence of psychiatric symptoms, a comprehensive psychiatric evaluation is crucial to detect mood disorders, psychosis, and behavioral abnormalities.^{3,4}

Laboratory Investigations:

Imaging Studies: Neuroimaging, including magnetic resonance imaging (MRI) and computed tomography (CT) scans, plays a fundamental role in the diagnostic workup. Imaging may reveal cerebral atrophy, white matter abnormalities, and vascular changes, offering valuable insights into the neurological aspects of VBSES.^{3,4}

Electroencephalography (EEG): EEG is indispensable for evaluating epileptic activity in individuals with suspected VBSES. It can assist in characterizing seizure patterns and assessing the extent of cerebral dysfunction.^{3,4}

Genetic Testing:

Genetic Evaluation: Although VBSES is not typically considered a genetic disorder, genetic testing may be pursued to rule out underlying genetic factors contributing to the syndrome or to identify related conditions that might mimic VBSES.^{3,4}

Differential Diagnosis:

Ruling Out Other Conditions: VBSES shares clinical features with several neurological and psychiatric disorders, necessitating a comprehensive differential diagnosis. Conditions such as Rasmussen's encephalitis, multiple sclerosis, and autoimmune encephalopathies must be excluded.^{3,4}

Diagnostic Criteria:

Lack of Formal Criteria: Unlike many medical conditions, VBSES lacks standardized diagnostic criteria, further complicating the diagnostic process. Diagnosis is often based on a clinical consensus and the exclusion of alternative explanations for the patient's symptoms.^{3,4}

Challenges and Considerations:

Rare Disease Expertise: Given the rarity of VBSES, access to experts in rare neurological conditions is often critical for accurate diagnosis and management.^{3,4}

Variable Clinical Presentation: VBSES can present with considerable variability among affected individuals, both in

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terms of symptomatology and disease progression, further complicating diagnosis.^{3,4}

Diagnostic Delay: The rarity of VBSES, coupled with its potential to mimic other disorders, often results in diagnostic delays, which can be frustrating for both patients and healthcare providers.^{3,4}

In conclusion, diagnosing Van Bogaert-Scherer-Epstein Syndrome is a multifaceted challenge that necessitates a collaborative effort among clinicians, radiologists, neurologists, and psychiatrists. A high level of clinical suspicion, thorough clinical and neurological evaluations, and advanced imaging techniques are instrumental in the diagnostic process. Due to the syndrome's rarity and complexity, further research is warranted to establish standardized diagnostic criteria and facilitate early and accurate diagnosis, ensuring timely interventions and improved outcomes for individuals affected by this enigmatic condition.^{3,4}

TREATMENT

The treatment of Van Bogaert-Scherer-Epstein Syndrome (VBSES), a rare and intricate neurological disorder with a complex clinical profile, presents a substantial challenge for healthcare professionals. Given the rarity of VBSES and the wide spectrum of symptoms it encompasses, management strategies need to be individually tailored to address the diverse clinical manifestations and optimize the quality of life for affected individuals. In this article, we embark on a comprehensive exploration of the multifaceted treatment approaches for VBSES.⁵

Multidisciplinary Collaboration:

Team Approach: The management of VBSES necessitates a multidisciplinary team consisting of neurologists, psychiatrists, rehabilitation specialists, and supportive care providers to address the full scope of symptoms and challenges encountered by patients.⁵

Pharmacological Interventions:

Antiepileptic Medications: Given that epileptic seizures are a common feature of VBSES, antiepileptic drugs (AEDs) are typically the cornerstone of pharmacological management. Medications such as levetiracetam, carbamazepine, and valproic acid may be prescribed to control seizure activity.⁵

Psychiatric Medications: For individuals with psychiatric symptoms, psychotropic medications such as antipsychotics, mood stabilizers, and antidepressants may be administered to manage mood disorders, psychosis, and behavioral abnormalities.⁵

Rehabilitative Therapies:

Physical and Occupational Therapy: Rehabilitation specialists can offer physical and occupational therapy to address motor deficits, dystonia, and ataxia, facilitating improved mobility and functional independence.⁵

Speech Therapy: Speech therapists can assist in managing dysarthria and communication challenges, helping patients regain or enhance their speech abilities.⁵

Supportive Care:

Psychological Support: Patients and their families may benefit from psychological support, including counseling and therapy, to cope with the psychological and emotional impact of VBSES.⁵

Social Services: Access to social services and community resources can be instrumental in addressing the social and economic challenges that individuals with VBSES may face.⁵

Surgical Interventions:

Surgery for Intractable Epilepsy: In cases of severe and intractable epilepsy, surgical interventions such as epilepsy surgery or implantation of responsive neurostimulation devices may be considered as a last resort.⁵

Symptomatic Management:

Pain Management: For those experiencing severe headaches or pain associated with dystonia, pain management strategies, including medications and interventional procedures, may be employed to enhance comfort and quality of life.⁵

Clinical Trials and Experimental Therapies:

Exploring Novel Therapies: Due to the limited understanding of VBSES and the lack of standardized treatment protocols, some individuals may be candidates for participation in clinical trials or experimental therapies aimed at elucidating potential treatment options.⁵

Prognosis and Long-Term Care:

Variable Prognosis: The prognosis for individuals with VBSES can vary widely, depending on the severity of symptoms and the individual's response to treatment. Some individuals may experience stabilization or improvement in their condition, while others may face progressive deterioration.⁵

Long-Term Care: Long-term care planning is often essential, given the chronic and potentially progressive nature of VBSES. This may involve ongoing monitoring, adjustments to treatment plans, and adaptations to the individual's changing needs.⁵

In conclusion, the treatment of Van Bogaert-Scherer-Epstein Syndrome necessitates a multifaceted approach that addresses the wide array of clinical manifestations encountered by affected individuals. Multidisciplinary collaboration, personalized care plans, and a patient-centered focus are integral to enhancing the quality of life and overall well-being of those living with this rare and challenging neurological disorder. Ongoing research efforts are crucial to advancing our understanding of VBSES and refining treatment strategies, offering hope for improved outcomes and enhanced patient care in the future.⁵

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CONCLUSION

In the realm of rare and enigmatic neurological disorders, Van Bogaert-Scherer-Epstein Syndrome (VBSES) stands as a formidable challenge, both for the individuals grappling with its complex clinical manifestations and for the healthcare professionals striving to comprehend, diagnose, and manage its intricacies. This article has endeavored to cast a spotlight on the multifaceted dimensions of VBSES, from its epidemiology and clinical manifestations to its diagnostic intricacies and treatment approaches, in the hopes of furthering our collective understanding of this rare and puzzling syndrome.

VBSES's rarity has been a recurrent theme throughout this discourse, and it is this rarity that has added layers of complexity to every facet of the syndrome, from early identification to treatment strategies. The paucity of standardized diagnostic criteria, coupled with the syndrome's propensity to mimic a plethora of other neurological and psychiatric disorders, underscores the urgency of concerted research efforts. The establishment of diagnostic criteria, perhaps through collaborative international initiatives, would pave the way for earlier diagnoses, reducing the protracted diagnostic odyssey that many individuals with VBSES currently endure.

With each individual's unique clinical presentation, VBSES defies homogenization and demands personalized, multidisciplinary care. It is only through the collaborative efforts of neurologists, psychiatrists, rehabilitation specialists, and support services that the diverse challenges posed by VBSES can be optimally addressed. Pharmacological interventions targeting seizures and psychiatric symptoms are pivotal, but so too are the rehabilitative therapies and supportive care that cater to motor deficits, dysarthria, and the emotional toll exacted by this enigmatic syndrome.

While significant strides have been made in the understanding and management of neurological and psychiatric disorders, VBSES continues to elude our full comprehension. The lack of a definitive etiology and the variable prognosis further complicate the clinical trajectory for those affected. As such, research into the genetic and environmental determinants of VBSES, as well as experimental treatment approaches, must remain a priority.

In conclusion, the odyssey of Van Bogaert-Scherer-Epstein Syndrome remains a multifaceted challenge, one that underscores the resilience and determination of both patients and the medical community. The road ahead is long, marked by diagnostic uncertainty and diverse clinical manifestations, yet it is imbued with hope. Hope for a deeper understanding of the underlying mechanisms, hope for earlier diagnosis, and hope for enhanced treatment strategies that can mitigate the burden of VBSES for those it touches. Through collaboration, research, and a commitment to improving the lives of

individuals affected by this rare and enigmatic condition, we stand a chance of navigating the labyrinthine enigma of VBSES with greater insight and efficacy.

REFERENCES

- I. Van Bogaert L, Scherer HJ, Epstein É. Une forme cérébrale de la cholestérose généralisée: (type particulier de lipidose à cholestérine). Masson; 1937.
- II. Gallus GN, Dotti MT, Federico A. Clinical and molecular diagnosis of cerebrotendinous xanthomatosis with a review of the mutations in the CYP27A1 gene. *Neurological Sciences* 2006; 27(2): 143-9.
- III. SALEN G. Cholestanol deposition in cerebrotendinous xanthomatosis: a possible mechanism. *Annals of internal medicine* 1971; 75(6): 843-51.
- IV. Zimmerman RD, Grossman RI. Neurodegenerative diseases and hydrocephalus. In: Yousem DM, Zimmerman RD, Grossman RI, Nadgir R. *Neuroradiology: The Requisites*. 3rd ed. St. Louis: Mosby; 2010: 249-79.
- V. Nie S, Chen G, Cao X, Zhang Y. Cerebrotendinous xanthomatosis: a comprehensive review of pathogenesis, clinical manifestations, diagnosis, and management. *Orphanet journal of rare diseases* 2014; 9(1): 179.