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Case Report

Renal Failure, Spina Bifida, and Porphyria: A Case Report and Review of the Literature

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ABSTRACT

This case report details the complex clinical presentation of a 33-year-old female with a history of meningomyelocele spina bifida, who developed early-onset renal failure and was later diagnosed with porphyria. The patient presented with bullous lesions on sun-exposed areas, which led to the investigation of potential underlying metabolic disorders. Despite surgical interventions for spina bifida, the patient's urinary function remained compromised, eventually progressing to stage 5 vesicourethral reflux and requiring long-term dialysis. The porphyria diagnosis was confirmed through a spot urine porphobilinogen test followed by a 24-hour urine analysis, and the specific subtype was found to be VP (Variegate Porphyria) confirmed by a genetic analysis. This case emphasizes the importance of considering porphyria in patients with unexplained dermatological and neurological symptoms, particularly when coexisting with other congenital or chronic conditions. The overlap of renal failure, spina bifida, and porphyria in this patient underscores the challenges in diagnosis and management, highlighting the need for multidisciplinary care. Early diagnosis and intervention are crucial to prevent complications and improve quality of life, particularly in rare and complex presentations such as this one. This report aims to raise awareness among clinicians about the potential for delayed diagnosis of porphyria, which can lead to severe and irreversible complications.

Keywords: Porphyria; Dialysis; Renal failure; Spina bifida; Bullous lesions; Metabolic disorders

Introduction

Porphyrias are a group of metabolic disorders caused by enzyme deficiencies in the heme biosynthetic pathway, often leading to the accumulation of porphyrins and their precursors. Clinical manifestations vary widely, ranging from cutaneous symptoms to potentially life-threatening acute neurovisceral attacks. Early recognition is crucial for preventing complications, yet diagnosis can be challenging due to the rarity and diverse clinical presentations of these disorders.

Case Presentation

A 33-year-old female was referred to our clinic with recurrent bullous lesions on her face, arms, and hands following sunlight exposure. These lesions, which initially left black scars and progressed to permanent white scars, prompted investigation into underlying metabolic disorders. Her medical history was significant for meningomyelocele spina bifida at birth, complicating urinary function despite surgical intervention.

Renal complications emerged in her early twenties,

progressing to stage 5 vesicourethral reflux requiring weekly dialysis. Over eight years, dialysis frequency increased to three times weekly, managed at home since the age of 28. Family history was notable for congenital adrenal hyperplasia in her mother and diabetes mellitus in her father, although no direct familial link to her condition was identified.

1. Symptomatic management of porphyria exacerbations: Initiating heme therapy, such as intravenous hemarginate, to alleviate acute symptoms and prevent porphyria attacks.

2. Renal supportive care: Continuation of thrice-weekly dialysis, which the patient performs independently at home, to manage uremic symptoms and maintain fluid and electrolyte balance.

3. Skin protection strategies: Implementing stringent sun protection measures, including the use of broad- spectrum sunscreen, protective clothing, and avoidance of direct sunlight exposure during peak hours, to mitigate cutaneous symptoms associated with porphyria.

In the treatment regimen for this patient, we administered intravenous hemarginate at a dose of 3 mg/kg during acute porphyria attacks, which were confirmed by the Ehrlich aldehyde test (**Figure 1**). This approach was effective in managing the acute symptoms and preventing further exacerbations. Additionally, we prescribed Plaquenil® (hydroxychloroquine) at a dosage of 100 mg per week to address the chronic cutaneous manifestations. After a 10-month course of Plaquenil®, the patient's bullous lesions have completely resolved, indicating a successful outcome in the management of her porphyria-related skin symptoms.

Figure 1: Positive Ehrlich Aldehyde Urine Test.

Methods

Diagnostic evaluation included spot urine porphobilinogen testing with Ehrlich's reagent, confirming elevated levels suggestive of porphyria. Subsequent 24-hour urine testing determined the specific subtype. Clinical correlation with cutaneous manifestations and neurological history supported the diagnosis of porphyria.

Discussion

Porphyrias are a group of inherited or acquired disorders characterized by a defect in one of the eight enzymes involved in the heme biosynthetic pathway, leading to the accumulation of porphyrins or their precursors. These accumulations can result in a wide range of clinical manifestations, from acute neurovisceral symptoms to chronic cutaneous lesions, depending on the specific type of porphyria and the affected enzym[1,2]. The complexity of porphyria's clinical presentation often leads to misdiagnosis or delayed diagnosis, as seen in this patient, where the diagnosis was only made after significant disease progression.

This patient's history of meningomyelocele spina bifida is a significant factor in her overall clinical presentation. Spina bifida, particularly the meningomyelocele subtype, is associated with a range of complications, including neurological deficits, chronic urinary retention, and renal dysfunction due to vesicourethral reflux[3]. In this case, the patient's renal failure began in her early twenties, requiring escalating dialysis interventions. It is well-documented that chronic renal failure can exacerbate the clinical manifestations of porphyria due to the impaired clearance of porphyrins and their precursors[4,5]. This interplay between renal impairment and porphyria likely contributed to the severity of the patient's symptoms and the difficulty in managing her condition.

The cutaneous symptoms experienced by the patient, particularly the formation of bullous lesions upon sun exposure (**Figures 2,3**), are characteristic of cutaneous porphyrias, such as porphyria cutanea tarda (PCT) or variegate porphyria (VP)[6]. These lesions occur due to the accumulation of porphyrins in the skin, which become photoactivated by ultraviolet light, leading to oxidative damage and blister formation[7]. The chronicity and progression of these lesions, resulting in permanent scarring, further underscore the impact of delayed diagnosis and inadequate management.

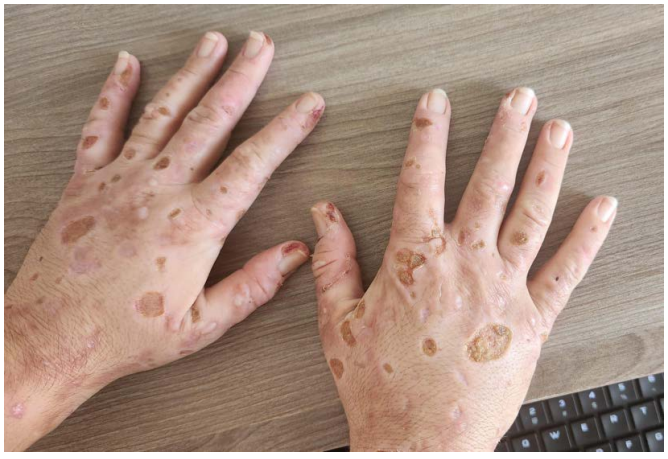

Figure 2: Lesions on hands, parts exposed to sunlight.

Interestingly, the patient's family history, while not directly indicative of porphyria, includes congenital adrenal hyperplasia in her mother and diabetes mellitus in her father. Although these conditions are not directly linked to porphyria, they may suggest a genetic predisposition to metabolic disorders, warranting further investigation into potential familial links or genetic mutations contributing to the patient's condition[8,9]. The absence of similar symptoms in immediate family members does not preclude a hereditary basis, particularly in cases where porphyria may manifest with varying severity or may remain asymptomatic in other family members.

Figure 3: Lesions on sun exposed areas, both hands and face.

The diagnostic process for porphyria in this patient involved a spot urine porphobilinogen test, which is a critical initial step in identifying acute porphyrias[10]. Elevated levels of porphobilinogen, along with clinical symptoms, strongly suggest a diagnosis of porphyria. The subsequent 24-hour urine analysis is essential for measuring the excretion of porphyrins and their precursors, helping to differentiate between the various types of porphyria[11]. In this case, the specific subtype is found to be VP, highlighting the need for genetic testing to confirm the diagnosis and guide management.

Management of porphyria, particularly in the context of concurrent renal failure, requires a multidisciplinary approach. Dermatological care is necessary to manage the cutaneous symptoms and prevent further skin damage, while nephrological support is critical in managing the patient's renal failure and ensuring the safe administration of treatments[12]. Given the complexity of this case, involving multiple systems and rare conditions, a coordinated effort among specialists in neurology, nephrology, dermatology, and genetics is essential to optimize patient outcomes.

Conclusion

In conclusion, this case report highlights the challenges in diagnosing and managing porphyria, particularly when it coexists with other congenital or chronic conditions like spina bifida and renal failure. The delay in diagnosis and the subsequent complications underscore the importance of early recognition and intervention in patients with complex medical histories. Clinicians should maintain a high index of suspicion for porphyria in patients with unexplained dermatological and neurological symptoms, particularly when these symptoms are accompanied by renal dysfunction or a history of congenital anomalies. Early diagnosis and a multidisciplinary approach to care are essential to prevent the progression of symptoms and improve the patient's quality of life.

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