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A Case Presentation of Pemphigus Vulgaris

Gabriel Riguerio[1,2], Folake Ishola[1,*], Cameron Stepler[1], Leah Gehring[1]

[1]Department of Internal Medicine, Ascension St. Vincent Hospital Indianapolis, Indiana, USA
[2]Department of Internal Medicine, Indiana University School of Medicine, Indiana, USA
*Corresponding author: folakeishola26@gmail.com

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Abstract Pemphigus vulgaris is a rare autoimmune disorder affecting the cell to cell adhesion molecules of keratinocytes in the epidermis and mucosa resulting in the production of the classic blister formation. This disease classically presents with intraoral blisters that may rupture and leave painful erosions impairing nutritional intake of patients. We present a case of a male in his early 40s noted to be earlier than the typical age of onset of Pemphigus Vulgaris. The patient had an atypical presentation of his lesions initially starting on his scalp rather than the typical oral involvement with bullous lesions at onset. Our case highlights the increasing incidence of malnutrition in patients with this condition. Our patient was found to have vitamin D insufficiency noting the association of Pemphigus Vulgaris with hypovitaminosis D with the call to action to include vitamin D screening in suspected or confirmed cases of Pemphigus Vulgaris. After discovering this patient's primary diagnosis with punch biopsies, he was immediately started on a treatment regimen including corticosteroids and Vitamin D supplementation. The objective of this case report is to highlight this patient's early onset of this disease as well as his atypical presentation. Additionally, we want to emphasize the association between Pemphigus Vulgaris and hypovitaminosis D to decrease the risk of further complications such as osteoporosis or life threatening injuries.

Keywords: *hypovitaminosis d, vitamin d deficiency, malnutrition, acantholysis, blisters, desmoglein, autoimmune, pemphigus vulgaris, pemphigus*

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1. Introduction

Pemphigus Vulgaris is the most prevalent subtype in a collection of rare, autoimmune diseases known as Pemphigus. The pathogenesis involves autoantibodies against two main desmosomal adhesion proteins, desmoglein (Dsg)1 and desmoglein (Dsg)3. The binding of these pathogenic autoantibodies to these intraepidermal adhesion proteins results in the separation of keratinocytes, known as acantholysis, leading to the characteristic painful, oral erosions, and flaccid blisters on skin [1]. It affects approximately 0.1-0.5 per 100,000 people each year and has about the same prevalence in males and females [2]. The mean age of onset for affected patients is around 45 to 65 years of age [1]. The initial clinical presentation involves intraoral blisters that rupture and leave painful erosions causing impaired nutritional intake in many cases. Patients also have a positive Nikolsky sign on examination which is the easy expansion of bullae when lateral traction is applied [2]. The diagnosis can be reached through direct immunofluorescence of a biopsy involving normal-appearing perilesional skin or mucosa [2]. Histology will show evidence of acantholysis due to autoantibodies binding Dsg1 and Dsg3. Additional workup may include indirect immunofluorescence and enzyme-linked immunosorbent assay (ELISA) to detect circulating autoantibodies [2]. Mortality for patients with Pemphigus Vulgaris has significantly reduced from 75% to around 5% following the emergence of corticosteroids and adjuvant immunosuppressant therapies [3]. As a complication of these treatments, infections such as pneumonia and sepsis, are the leading cause of death in this patient population [3]. We report a case of a male with newly diagnosed Pemphigus Vulgaris.

2. Case Presentation

A 42-year-old Hispanic male with no past medical history presented to the ED with painful lesions on his head, left wrist, axilla, groin, and mouth. Patient reported that the lesions on his head began about one year ago, and the other notable lesions began about six months ago. He traveled to his home country, Mexico, about a year ago, during the onset of symptoms. He reported having fever and chills when the head lesions first appeared a year ago. He admits to having pruritus noting that the lesions were all pruritic except for the lesions in his mouth. Patient reported that his mouth lesions were severely painful when attempting to eat solid or liquid foods. He denied having any dysphagia or globus sensation. He notes that his symptoms have been debilitating as he has not been

working for several weeks. He works as a construction worker where he lays concrete at work. No surgical or family history. No tobacco, alcohol, or illicit drug use. No sexual activity for over a year. No recent HIV or STI testing. He mentioned taking a course of diflucan about two months prior to evaluation at our facility with no improvement in his symptoms.

Patient's physical exam was notable for macerated and erythematous appearing buccal mucosa with lesions on the hard and soft palates along with chelation on his lips as seen in Figure 1. No lymphadenopathy noted in his head and neck. He had several indurated papules with and without eschar, on his left wrist (Figure 2), midline of his back, and flat erythematous lesions on his right axilla. He was found to have right greater than left axillary and inguinal lymphadenopathy. Dry, crusted, psoriatic appearing lesions were noted on his scalp. Dry skin and excoriations were noted on the right groin. No clubbing or pitting were noted on his nails.

Initial lab work included a CBC and BMP that were unremarkable. CT chest/abdomen/pelvis with contrast was negative for any acute process. No acute intrathoracic or intra-abdominal abnormality. No thoracic, abdominal, pelvic, or inguinal lymphadenopathy. CT neck with contrast showed mild diffuse enhancement of the surface of the tongue musculature likely reflecting stomatitis with no deeper extension or abscess seen. Bilateral upper neck reactive lymphadenopathy was noted on imaging.

Infectious disease team was consulted during the patient's hospitalization for further evaluation. CRP, ESR, ANCA screen, ANA, HIV, RPR, Anti Ro/La, Fungitell assay, Urine histoplasma, and Urine blastomyces all came back negative. Vitamin A, B3, C, D, E, and folate levels were within normal limits except for Vitamin D. Patient was started on 50 mcg daily for his Vitamin D insufficiency. He had punch biopsies done on his left wrist and right axilla lesions. Right axillary lesion was remarkable for suprabasilar vesicle formation with an associated mixed inflammatory cell infiltrate within the dermis. Left wrist lesion was remarkable for an extensive suprabasilar vesicle formation with acantholysis of keratinocytes and an associated mixed inflammatory cell infiltrate within the dermis. In summary, both lesions were remarkable for suprabasilar vesicular dermatitis, mixed cell with frequent eosinophils (Figure 3). The histologic features were found to be consistent with pemphigus vulgaris. PAS stains were performed on both biopsy specimens to rule out the possibility of concomitant infection. These stains were negative for fungi. He was started on Philadelphia swish and spit, Prednisone, Protonix, and Bactrim during hospitalization.

The patient was having worsening difficulty eating solid food for six months secondary to pain from the oral lesions. He was found to have severe protein calorie malnutrition evidenced by less than 50% of his estimated energy intake for over a month and 17% weight loss in less than 6 months. Oral Intake was encouraged during hospitalization with foods of pureed consistency. Per registered dietitian recommendations, the patient was also started on protein shakes three times daily with consideration for alternative nutritional supplementation if he was unable to get adequate nutrition from oral intake. Patient was able to eat solid food towards the end of his hospital stay as he had not been able to do so for the last six months.

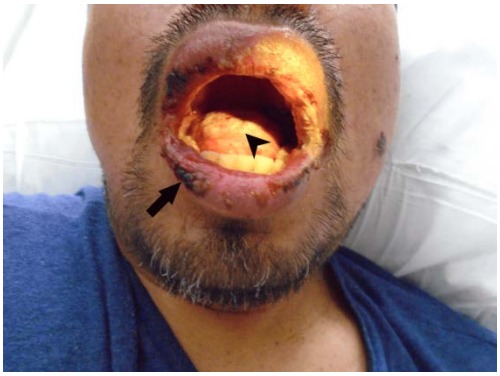

Figure 1. Oral involvement. Tailed arrow indicates healing eschars overlying the upper and lower lips. Tailless arrow shows evidence of mucosal involvement along the tongue

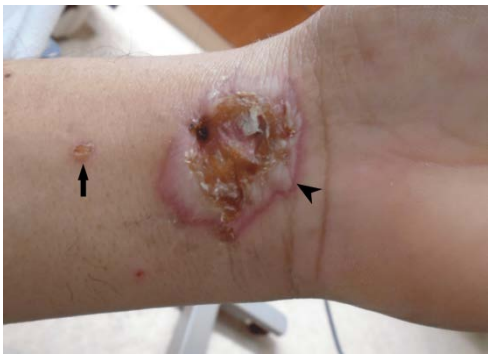

Figure 2. Left wrist involvement. Tailed arrow shows a small, typical deroofed vesicle. Tailless arrow shows a large, ruptured vesicle with central granulation

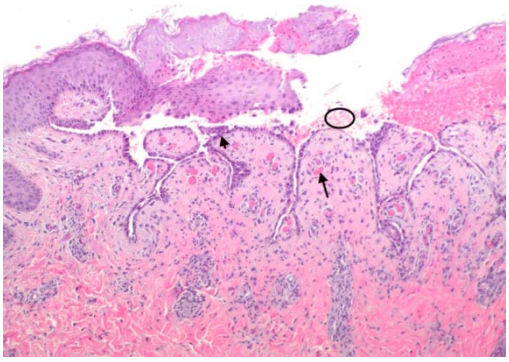

Figure 3. Patient's histology image. Mixed inflammatory cell infiltrates in dermis (circle), frequent eosinophils (tailed arrow), and acantholysis of keratinocytes (tailless arrow)

Prior to discharge, the patient reported having significant improvement in his pain and pruritus. He was given instructions to follow up with a primary care provider and dermatologist for further management. The patient was discharged with prescriptions for 70 mg Prednisone daily, 40 mg Protonix daily for GI prophylaxis, Bactrim for Pneumocystis jirovecii pneumonia prophylaxis, Philadelphia swish and spit for oral symptoms, and Vitamin D for his vitamin D insufficiency. Following discharge, the patient was followed by the dermatology team at our primary care clinic. He was found to have elevated desmoglein 1 antibody at 58 and elevated desmoglein 3 antibody at 160. He had positive

circulating IgG anti-epithelial cell surface antibodies with a titer of 1:80. Patient is currently on Mycophenolate Mofetil (Cellcept) with the plan to transition him to Rituximab and taper prednisone.

3. Discussion

Pemphigus Vulgaris classically affects elderly individuals within the age of 45 to 65 years old [1]. Our patient presented at our facility for evaluation a year after onset of his symptoms at age 42. In about 80% of patients affected with Pemphigus Vulgaris, the first sign of the condition usually starts with oral involvement in the form of blisters that eventually rupture resulting in excruciating erosions [2,4,5,6]. The typical pattern of Pemphigus Vulgaris is the development of cutaneous lesions after the development of oral blisters [2]. Our patient had a different presentation as his symptoms initially began on his scalp six months prior to the development of lesions in his oral mucosa, and cutaneous lesions to his left wrist, axilla, back, and groin.

The characteristic skin finding of this condition is noted to be a blister formation in the form of a "thin walled bulla" [4]. This is due to the fluid buildup from the destruction of the cell to cell adhesion of keratinocytes in the epidermis and mucosa [4,7]. This destruction is done by autoantibodies targeting desmoglein, a known cell adhesion molecule. Our patient did not present with the typical blister formation on his left wrist as evidenced in *Figure 2*. During the patient interview on presentation, he denied having any known blisters prior to evaluation at our facility. However, the lesions found may represent the remains of bullous lesions that had ruptured.

Due to the excruciating pain of oral lesions in affected patients, there is usually a degree of malnutrition noted as patients have decreased oral intake [2]. Our patient was no different in this aspect as he had worsening oral intake since the presence of oral lesions six months prior to evaluation at our facility. He mentioned that he began having blended foods to get some nutrition in for the day. This was evidenced by his severe protein calorie malnutrition with less than 50% of his estimated energy intake for over a month and 17% weight loss in less than 6 months.

Our patient was found to have vitamin D deficiency with low vitamin D noted on workup. A meta-analysis done by Yang et al showed a remarkably lower vitamin D levels in patients with autoimmune bullous dermatoses such as Pemphigus Vulgaris than in the control population [8]. Affected patients with Pemphigus Vulgaris have an increased risk of hypovitaminosis D and vertebral fractures [9]. One of the mainstay treatments for pemphigus vulgaris is glucocorticoid therapy. Steroids are known to cause hypovitaminosis D and osteoporosis. The meta-analysis done by Yang et al noted that patients with no steroid treatment had vitamin D insufficiency as well suggesting that the hypovitaminosis D is not just an adverse effect of steroid therapy but a result of the condition [8]. Hypovitaminosis D puts affected patients at risk for osteopenia, osteoporosis, fractures from falls with debilitating injuries that can affect quality of life. This can be avoided if they are routinely monitored and repleted with supplementation. Future research work on Pemphigus Vulgaris should be targeted at pathophysiology behind pemphigus vulgaris and hypovitaminosis D.

4. Conclusion

Pemphigus Vulgaris is a rare autoimmune disease usually seen with equal prevalence between males and females. Our case involves a male who is younger than the mean age of affected patients with this condition. Initial imaging and lab work were helpful in ruling out potential diagnoses. Ultimately, the primary diagnosis was confirmed with the punch biopsies of lesions in the right axilla and left wrist. This resulted in starting the regimen of 70 mg Prednisone daily, 40 mg Protonix daily for GI prophylaxis, and Bactrim for Pneumocystis jirovecii pneumonia prophylaxis. Our patient had rapid improvement in symptoms and was able to eat solid food prior to discharge. The aim of this case report was to highlight this individual's early presentation of Pemphigus Vulgaris based on his age at symptom onset, and his atypical presentation of initial lesions originating in the scalp prior to oral involvement. Additionally, we wanted to highlight the association of hypovitaminosis D and pemphigus vulgaris with a call to action on including vitamin D labs in suspected or confirmed cases. Future research work on Pemphigus Vulgaris should be targeted at pathophysiology behind pemphigus vulgaris and hypovitaminosis D.

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