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CASE REPORT

An Uncommon Association: HLA-B27 Positivity in Conjunction With Pernicious Anemia - A Case Report and Literature Review

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Abstract

Pernicious anemia, stemming from Vitamin B12 deficiency and autoimmune processes affecting intrinsic factor production, presents challenges in early diagnosis due to vague initial symptoms. This case report introduces a unique occurrence of pernicious anemia-induced peripheral neuropathy in a patient with concurrent HLA-B27 arthropathy, highlighting the complex interplay of autoimmune mechanisms. While HLA-B27 is not typically associated with pernicious anemia, the case underscores the importance of exploring specific HLA haplotypes in understanding the nuanced manifestation of autoimmune disorders. Comprehensive screening for anti-intrinsic factor and anti-parietal cell antibodies is crucial in individuals with signs of pernicious anemia, especially those with a history of HLA-B27 arthropathy, guiding tailored management strategies. This report contributes to the ongoing exploration of the intricate autoimmune landscape in pernicious anemia.

Keywords: Pernicious anemia, HLA-B27, Peripheral neuropathy, Autoimmune disorders, Intrinsic factor antibodies, Vitamin B12 deficiency, Genetic predisposition, Gastroenterology, Macrocytic anemia

1. Introduction

Pernicious anemia, characterized by megaloblastic anemia due to Vitamin B12 deficiency, arises from autoimmune processes targeting intrinsic factor (IF) and parietal cells.[1,2] The disorder poses diagnostic challenges due to its nonspecific initial symptoms, including weakness and difficulty concentrating.[1] Importantly, delayed recognition may lead to irreversible neurological damage.[1]

This case report explores a unique presentation of pernicious anemia causing peripheral neuropathy in a patient with a known HLA-B27 arthropathy, an association not conventionally established.[4] HLA-B27, a key player in autoimmune diseases, prompts an examination of specific HLA haplotypes' roles in pernicious anemia's pathogenesis. Through this case, we aim to contribute to understanding the complex interplay of genetics, autoimmunity, and clinical manifestations in pernicious anemia.

2. Case presentation

We present the case of a 53-year-old male with a medical history notable for osteoarthritis, previously diagnosed HLA-B27 positivity during an arthritis workup, and well-managed hypertension with a calcium channel blocker and ACE inhibitor. The patient sought preoperative clearance for a total knee replacement procedure due to persistent bilateral knee pain.

Noteworthy in the patient's medical history is a previous acknowledgment of low vitamin B12 levels, which, despite being treated with oral B12 supplements, had not undergone further investigation. Upon laboratory examination, vitamin B12 deficiency persisted, with levels measuring <200 at 188, accompanied by elevated methylmalonic acid. Subsequent testing revealed the presence of positive anti-intrinsic factor antibodies.

The definitive diagnosis of pernicious anemia was established based on the laboratory findings.

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Consequently, the patient initiated a therapeutic regimen involving consecutive subcutaneous cyanocobalamin injections. This case underscores the importance of thorough investigation and timely intervention in patients presenting with persistent vitamin B12 deficiency, especially in the context of associated autoimmune factors such as HLA-B27 positivity.

3. Discussion

The etiology of pernicious anemia remains multifaceted, involving a complex interplay of genetic and autoimmune factors. Autoimmune processes targeting intrinsic factor (IF) and parietal cells contribute to the pathogenesis.[1,3] While the mechanism by which these autoimmune processes unfold is not fully elucidated, studies underscore the significance of antibodies directed towards IF and/or parietal cells.[1,5]

In this case, the patient with HLA-B27 arthropathy presented a distinctive clinical manifestation with concurrent pernicious anemia, prompting a closer examination of the potential interconnection between HLA-B27 and pernicious anemia. HLA genes, integral to immune responses, exhibit associations with various autoimmune conditions.[4,6] Contrary to some studies suggesting no overall increased risk for pernicious anemia in HLA-B27 carriers,[7] the patient's unique presentation prompts consideration of heterogeneity in the pathogenesis of pernicious anemia.

Ungar et al. examined HLA patterns in clinically defined subgroups of patients with pernicious anemia, suggesting potential variations in the pathogenesis of the condition.[7] These findings align with the proposal that pernicious anemia results from the interplay of autoimmune and other factors acting on a susceptible gastric mucosa.[7] While HLA-B27 may not confer an increased risk for pernicious anemia on a broad scale, the intricate relationship between specific HLA haplotypes and the pathogenesis of autoimmune conditions warrants further exploration.

The presence of concomitant autoimmune conditions in individuals with HLA-B27 haplotypes is well-documented.[4] This patient's history of HLA-B27 arthropathy underscores the need for comprehensive screening for anti-IF and anti-parietal cell antibodies in those demonstrating signs of pernicious anemia. This approach ensures a nuanced understanding of the autoimmune landscape, aiding in both the diagnosis and tailored management of pernicious anemia in specific patient subsets.

The gaps in our current understanding of pernicious anemia's pathogenesis underscore the necessity for ongoing research. Further investigations should delve into the intricate interactions between HLA haplotypes and autoimmune conditions, shedding light on the complexities of pernicious anemia and its diverse clinical presentations.

4. Conclusion

This case report explores the atypical coexistence of pernicious anemia and HLA-B27 arthropathy, challenging conventional understandings and emphasizing the intricate nature of autoimmune diseases. The complex etiology of pernicious anemia, involving genetic predisposition and autoimmune processes targeting intrinsic factor (IF) and parietal cells, necessitates ongoing exploration.[1,5] Despite conventional wisdom not attributing a broad-scale increased risk of pernicious anemia to HLA-B27 carriers,[7] this case urges a reevaluation of the condition's heterogeneous pathogenesis.

The presented patient, with concurrent HLA-B27 arthropathy and pernicious anemia, prompts reconsideration of the potential interplay between specific HLA haplotypes and autoimmune conditions. Ungar et al.'s findings suggest variations in HLA patterns among pernicious anemia patients, hinting at autoimmune and other factors influencing gastric mucosa susceptibility.[7] While specifics of HLA haplotypes' intricate relationship with autoimmune conditions require further investigation, comprehensive screening for pernicious anemia in patients with signs and HLA-B27 history ensures tailored diagnosis and management.[4] The persistent gaps in understanding pernicious anemia's pathogenesis underscore the necessity for ongoing research, providing valuable insights into its complexities and diverse clinical presentations.

Conflict of interest

No conflicts of interest to declare.

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