

Potential Correlation between Kienböck's Disease and Hereditary Hemochromatosis: A Case Report

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Learning Point of the Article:

Hereditary hemochromatosis may be a predisposing factor for Kienböck's disease, which in turn may aid in the early detection and treatment of these conditions.

Abstract

Introduction: Avascular necrosis of the lunate bone has been extensively researched, although the etiology of the condition remains controversial. Even though many treatments for the disease exist, a better understanding of the pathophysiology can improve our decision-making between preventive and therapeutic measures. Various hematological disorders have been found to predispose for Kienböck's disease. On the other hand, there has not yet been any reference in literature to a relationship between this condition and hereditary hemochromatosis (HH).

Case Report: We present two cases of Kienböck's disease in two patients who are third-degree relatives and diagnosed with HH. A 61-year-old Caucasian female patient with type 1 HH presented with symptomatic Kienböck's disease on the left side. The patient is a third-degree relative of a 51-year-old male Caucasian patient with Kienböck's disease on the right side, known as having the same hereditary hematological condition.

Conclusion: Our findings suggest a potential correlation between the aforementioned conditions. The prevalence of these coexisting pathologies should be studied further.

Keywords: Kienböck, lunate, osteonecrosis, hemochromatosis.

Introduction

Kienböck's disease is a condition characterized by avascular necrosis (AVN) of the lunate bone. It was first described in 1910 by the Austrian radiologist Robert Kienböck [1]. Symptoms can include wrist pain, decreased grip strength, and range of motion of the affected joint. However, incidental determination of the condition is common [2]. Other than the patient's perceived symptoms, there are little other findings that guide the choice of therapy. Although distinct degrees of lunar collapse could evoke outspoken symptoms in some, numerous studies found no clear link between these radiological findings and the symptomatology [2-4]. Treatment is therefore predominantly

based on patient complaints. Just as the factors predisposing to symptoms are not clearly defined, the etiology of the condition remains dubious. In general, it affects 20–40-year-old men regularly performing physical labor or having a history of wrist trauma. More recent literature describes equal involvement in terms of gender with the onset of the disease usually later in women [3]. Other low evidence predisposing factors include smoking, excessive alcohol consumption, ulnar variance, and specific comorbidities such as diabetes, peripheral vascular, and certain genetic conditions [4]. Factors largely associated with AVN of the hip as well. Despite inconclusive data, one can assume that the vascularization of the lunate plays an important

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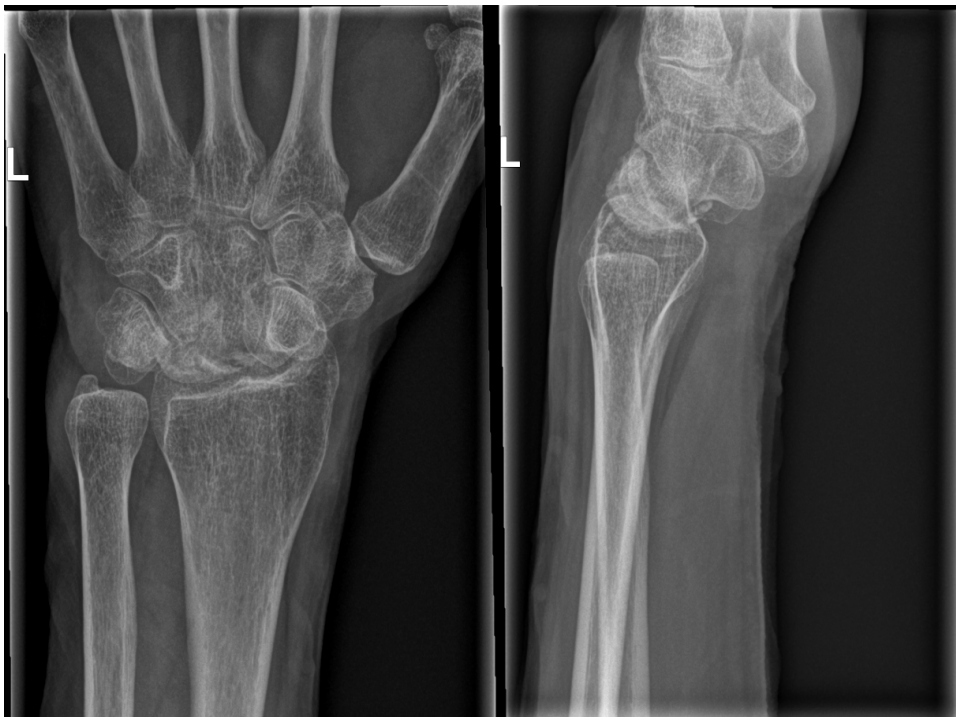


Figure 1: Anteriorposterior and lateral left wrist radiographs of case 1 demonstrating Kienböck's disease. Radiographs of the left wrist of case 1 at the first presentation showing Lichtman stage IIIb Kienböck's disease with notable scaphoid flexion and ring sign. Limited degeneration of the neighboring intercarpal joints.

role in the onset of the disease.

Lamas et al. mapped the arterial supply of this carpal bone by studying 27 wrists using latex injections and the Spalteholz technique, finding palmar nutrient vessels entering the proximal end of the os lunatum through the radioscapocapitate ligament [4]. Degeneration of this ligament through blunt force and repetitive microtrauma could therefore be a cause of the vascularization of the bone. Vessels entering the dorsal aspect of the lunate could anastomose with the volar ones, preventing ischemia. Considering this delicate vascularization, not only the blood supply but also the vital blood-carried minerals and nutrients could impact the health of this bone.

We refer in this context to one specific condition. Hereditary hemochromatosis (HH), a common autosomal recessive disorder in Caucasians, is characterized by excessive iron absorption. A C282Y mutation of the HFE gene is found in most cases [5,6]. Men and women are affected equally in regard to gene inheritance. In vitro studies have proven the excess of iron to impact different cascades, altering osteoclastogenesis, resulting in bone loss in HH-impaired patients [7]. These studies help us better understand the impact on bone integrity but currently fail to adequately emulate these mostly animal-based and vitreous findings in clinical cases. Little evidence can be found on the correlation between HH and lunar

osteomalacia.

We present two affiliated patients with unilateral Kienböck's disease and HH who received treatment in our center.

Case Report

Case 1

A 61-year-old female patient, housewife, right hand dominant with type 1 HH and homozygous for the Cys282Tyr mutation of the HFE gene presented with increasing mechanical left wrist tenderness and concomitant functional deficit without any history of wrist-involved injuries, professional or recreational activities associated with excessive stress on the wrist joint. The patient is an active smoker. Physical examination showed 45° palmar flexion (contralateral 80°) and dorsiflexion (contralateral 70°). Prosupination was full and

symmetric. Posteroanterior and lateral radiographs displayed lunate collapse with a revised carpal height ratio of 1.26 (measured using the Nattrass Method), differing significantly from the mean value of the revised carpal height ratio in the general population of 1.57 ± 0.05 [8] (Fig. 1). Magnetic resonance imaging of the left wrist confirmed the diagnosis of stage IIIb Kienböck's according to the Lichtman classification [9] (Fig. 2). Conservative treatment consisting of 8 months of intermittent bracing yielded limited results in terms of subjective or functional improvement. Consequently, a proximal row carpectomy was performed. Six months post-operatively, clinical examination showed complete symmetrical prosupination, 30° palmar flexion, 45° dorsiflexion and a grip strength of 36 kg on the right, and 22 kg on the left, measured with the Jamar Dynamometer.

Case 2

A 51-year-old male patient, driver and third-degree relative of our female study participant, also known with type 1 HH and homozygous for the Cys282Tyr mutation in the HFE gene is presented. He quit smoking at age 35 and is known for psoriasis. He is right-hand dominant and presented with 14-year history of atraumatic right wrist complaints. The physical examination showed 30° palmar flexion (contralateral 75°), 80° dorsiflexion

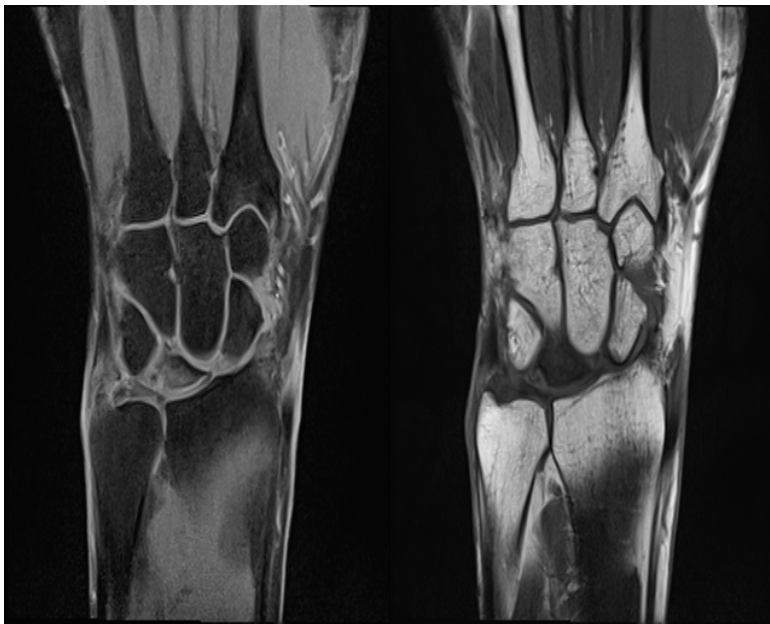


Figure 2: Magnetic resonance imaging-images. T2- and T1-weighted anteriorposterior left wrist images of case 1. T2- and T1-weighted anteriorposterior magnetic resonance images of the left wrist of case 1 at first presentation showing Lichtman stage IIIb Kienböck's disease. These images depict no significant effusion or degeneration of neighboring joints.

(contralateral 80°) and Grip strength of 30 kg on the right, and 50 kg on the left, measured with the Jamar dynamometer. X-rays were obtained showing lunar collapse with a revised carpal height ratio of 1.31 (Fig. 3) and stage IIIb Kienböck's disease in accordance with the Lichtman classification. Additional therapy was consensually waived given the current unstated



Figure 3: Anteriorposterior and lateral right wrist radiographs of case 2 demonstrating Kienböck's disease. Radiographs of the right wrist of case 2 at first presentation showing Lichtman stage IIIb Kienböck's disease with a near total collapse of the lunate bone. Scaphoid flexion and ring signs are seen as well. There's little degeneration of the neighboring intercarpal joints.

clinical implications.

Discussion

Since its discovery in 1910, several pathogenetic pathways have been proposed for Kienböck's disease with many well-defined risk factors playing a key role. These can be further categorized into different groups, including traumatic events ranging from repetitive microtrauma to acute wrist injuries, deprived vascularization, rheumatological conditions, and certain medical conditions, associated with impaired osseous blood flow [10].

Concerning Kienböck's disease, HH is not commonly mentioned in the literature and its role is probably underestimated. After all, most current research largely focuses on morphological predisposing factors, rather than hematological ones. This iron overload disorder can lead to osteoporosis, chondrocalcinosis, and arthropathy. A few studies linked the condition to AVN of the femur [11, 12].

Both patients had successful monthly phlebotomy treatments for their iron excess. Both relatives presented with clear lunate collapse. One chose skillful neglect, and the other one underwent an operative treatment with good relief of pain and fine recovery of function.

Conclusion

We suggest a close pathological relationship between HH and AVN of the lunate (Kienböck's disease) based on these two case reports in a male and female relative, which have never been published in the literature. Awareness of this possible cause of wrist pain with this hereditary condition is mandatory.

Clinical Message

In patients with Kienböck's disease, possible predisposing conditions such as HH should not be overlooked. Identification of both pathologies may help us better understand the pathophysiology and possibly future treatment of lunatomalacia.

Declaration of patient consent: The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given the consent for his/ her images and other clinical information to be reported in the journal. The patient understands that his/ her names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Conflict of interest: Nil **Source of support:** None

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Consent: The authors confirm that informed consent was obtained from the patient for publication of this case report

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