Familial Occurrence of Kienbock’s Disease

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Abstract

We present the first reported case of a mother and daughter with unilateral Kienbock’s disease, along with a radiographic and genetic study of the case. The radiographic study demonstrated ulnar minus and flattened radius in both patients. The karyotype of both affected individuals (mother and daughter) was normal 46XX (peripheral lymphocytes). Since the chromosomal analysis was normal, we could not speculate with regard to genetic etiology, or suggest further molecular investigation. If more familial cases become available, it would be possible to investigate this in the future.

Introduction

In 1910 Robert Kienbock for the first time described a series of 16 cases of lunate “traumatic malacia” [1]. This was the first osteonecrotic clinical report of the ulnar and has since been termed as Kienbock’s disease. Kienbock’s disease tends to occur in active adults aged between 20 and 40 years [2]. It is usually a unilateral condition but several cases of bilateral disease have been described [3]. The etiology of the disease is unknown and many causes have been proposed. Gelberman et al. investigated the vascular anatomy of the lunate and suggested a theory of “repeated trauma mechanism” as the most likely cause of Kienbock’s disease [4]. The vascular vulnerability of the lunate was also demonstrated by Panagis et al. [5] Another hypothesis suggested that venous congestion plays a role in this disease [6] The anatomy of the lunate and the distal radius and ulna may be of importance to the appearance of the disease, also an association between ulnar negative variance and Kienbock’s disease has been described in the literature [7-10]. Some authors found flattened radial inclination [10-11] and smaller lunate [11] predisposing to the disease. Systemic etiology has not been established but reports of the disease associated with other conditions such as septic emboli, sickle cell disease, gout, carpal coalition, cerebral palsy and corticosteroid use have been described [12]. Familial cases of kienbock’s disease have not been reported [13] apart from one incident of two brothers with bilateral disease [14]. In this article we present the first reported case of a mother and daughter with unilateral Kienbock’s disease, along with a radiographic and genetic study of the case.

Case Report

The mother was diagnosed as suffering from left Kienbock’s disease when she was 29 years old. She is right-handed and complained of left wrist pain for several years; the diagnosis was made by plain x-ray and a TC99-bone scan (Figure 1). She underwent distal radial shortening and is asymptomatic at present.

The daughter was diagnosed with left Kienbock’s disease when she was 16 years old. She is right-handed and complained of left wrist pain for two years; the diagnosis was made by plain x-ray (Figure 2), TC99-bone scan and MRI (Figure 3). She refused any surgical treatment.

We performed a radiographic comparison between the patients (Table 1). The ulnar variance was measured according to the recommendation of Palmer and Epner [15,16], They both had a negative ulnar variance (Mother-3 mm, Daughter-1 mm). Radial inclination angle was measured and found to be flattened as compared to Tsuge and Nakamura’s report of normal individuals (23.9 ± 2.8 Vs. 25.2 ± 2.8) [10] (Mother-20, Daughter-21). As part of the investigation, a family tree was made (Figure 4) and we performed chromosomes analysis to rule out familial structural variant. The family tree was characterized by multiple consanguineous marriages, as often seen in Arab Moslem families in our region. The karyotype of both affected individuals (mother and daughter) was normal 46XX (peripheral lymphocytes).

Discussion

We described a case of familial kienbock’s disease, with the two patients demonstrating similar clinical and anatomical characteristics. The radiographic study demonstrated ulnar minus and flattened radius in both patients. These findings match the anatomical variances that were suggested to predispose the disease. Possible environmental (non-genetic) causes for the condition in these two women were revoked.

It is generally accepted that Kienbock’s is a sporadic disease; therefore environmental factors could be considered as prime

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etologic factors. But the recurrence of a similar phenotype in the two individuals we present, a mother and her daughter, raises the premise genetic factor involvement and predispose for the disease. The pedigree complies both monogenic and multifactorial inheritance. For example, vertical transmission is consistent with autosomal dominant inheritance. Scarcity of affected individuals in such a large family can be explained either by de-novo mutation of the mother, or reduced penetrance or variable expression of the clinical features in other carriers, consequently other affected individuals were overlooked. Since the family is characterized by multiple consanguineous marriages, autosomal recessive inheritance, manifesting as pseudo-dominant transmission, should also be considered.

However, other types of inheritance cannot be fully excluded based on the available data. Presently no genetic cause is known to associate Kienbock’s disease. Therefore, we performed chromosomal analysis, assuming that any chromosomal rearrangement, in particular if segregates with the clinical phenotype in the family - might lead to suspected genomic regions. Since the chromosomal analysis was normal, we could not speculate with regard to genetic etiology, or suggest further molecular investigation. If more familial cases become available, it would be possible to investigate this in the future.

### References

1. Kienbock R, Peltier L (1980) Concerning traumatic malacia of the lunate and its consequences: Degeneration and compression fractures. Clin Orthop149: 4-8.
2. Almquist EE (1986) Kienbock’s disease. Clin Orthop Relat Res 202: 68-78.
3. Yazaki N, Nakamura R, Nakao E, Iwata Y, Tatebe M, Hattori T (2005) Bilateral Kienbock’s disease. J Hand Surg 30: 133-136.
4. Gelberman RH, Bauman TD, Menon J, Akeson WH (1980) The vascularity of the lunate bone and Kienböck.s disease. J Hand Surg Am 5: 272-278.
5. Panagis JS, Gelberman RH, Taleisnik J, Baumgaertner M (1983) The arterial anatomy of the human carpus: Part II The intraosseous vascularity. J Hand Surg Am 8:375-382.
6. Schiltenwolf M, Martini AK, Mau HC, Eversheim S, Brocai DR et al. (1996) Further investigations of the intraosseous pressure characteristics in necrotic lunates (Kienböck’s disease). J Hand Surg Am 21: 754-758.
7. Hultén O (1928) Über anatomische Variationen der Handgelenkknochen. Acta Radiol Scand 9:155-168.
8. Goeminne S, Degreef I, De Smet L (1994) Negative ulnar variance is not a risk factor for Kienböck’s disease. Acta Orthop Belg 76: 38-41
9. Nakamura R, Imaeda T, Miura T (1990) Radial shortening for Kienböck’s disease: factors affecting the operative result. J Hand Surg Br 15: 40-45.
10. Tsuge S, Nakamura R (1993) Anatomical risk factors for Kienböck’s disease. J Hand Surg Br 18:70-75.
11. Watanabe K, Nakamura R, Horii E, Miura T (1993) Biomechanical analysis of radial wedge osteotomy for the treatment of Kienböck’s disease. J Hand Surg Am 18: 686-690.
12. Allan CH, Joshi A, Lichtman DM (2001) Kienbock’s disease: diagnosis and treatment. J Am Acad Orthop Surg 9: 128-136.
13. Iirisarri C (2010) [Aetiology of kienbock’s disease] Handchir Mikrochir Plast Chir. 42: 157-161.
14. Ringsted A (1932) Doppelseitiger Mb. Kienboeck bei 2 Br.udern. Acta Chirurgica Scandinavica, LXIX: 185-196.
15. Epler RA, Bowers WH, Guilford WB (1982) Ulnar variance—the effect of wrist positioning and roentgen filming technique. J Hand Surg Am 7: 298-305.
16. Palmer AK, Glisson RR, Werner FW (1982) Ulnar variance determination. J Hand Surg Am 7: 376-379.

### Table 1: Anatomical comparison between the mother and the daughter.

|                     | Mother | Daughter |
|---------------------|--------|---------|
| Age of onset        | 29     | 16      |
| Side affected       | left   | left    |
| Dominant hand       | right  | right   |
| Ulnar variance      | -3mm   | -1mm    |
| Radial inclination  | 20°    | 21°     |

**Figure 2:** AP X-ray of the daughter’s left hand demonstrating increased density changes in the lunate and ulnar minus variant.

**Figure 3:** T2-weighted image – coronal view of the daughter’s left hand demonstrating increased signal in the lunate.

**Figure 4:** Family tree. The affected individuals demonstrating multiple consanguineous marriages.