Archives of Surgery and Clinical Case Reports
Miura Y, et al. Arch Surg Clin Case Rep 6: 204.
www.doi.org/10.29011/2689-0526.100204
www.gavinpublishers.com

Case Report

Proximal Humerus and Proximal Femur Fractures with Nasu-Hakola Disease Treated with Prosthetic Replacements: A Case Report

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Citation: Miura Y, Kawamura K, Hasegawa H, Kurata S, Uchihara Y, et al (2023) Proximal Humerus and Proximal Femur Fractures with Nasu-Hakola Disease Treated with Prosthetic Replacements: A Case Report. Arch Surg Clin Case Rep 6: 204. DOI: 10.29011/2689-0526.100204

Received Date: 21 August 2023; **Accepted Date:** 24 August 2023; **Published Date:** 28 August 2023

Abstract

Background: Nasu-Hakola disease (NHD) is a rare autosomal recessive disorder characterized by progressive dementia and repeated pathological fractures caused by polycystic lipomembranous osteodysplasia during adolescence. The dementia exhibits a relentless exacerbation, culminating in the eventual transition to a vegetative state within the span of 50 years.

Case: A 41-year-old male afflicted with NHD sustained a fracture of the left proximal humerus, and an ipsilateral femoral neck fracture. Both fractures underwent initial treatment through open reduction internal fixation, which subsequently led to collapses. Subsequently, the patient underwent a salvage procedure involving prosthetic replacements, during which no complications were observed.

Conclusion: The application of prosthetic replacement emerges as a warranted approach for metaphyseal fractures in the context of NHD, driven by the distinctive presence of polycystic lipomembranous osteodysplasia, a hallmark feature of this disorder.

Keywords: Nasu-Hakola Disease; Bone Fractures; Prosthetic Replacement; Bipolar Hip Arthroplasty; Open Reduction Internal Fixation

Introduction

Nasu-Hakola disease (NHD) is a rare autosomal recessive disorder characterized by progressive dementia and repeated pathological fractures during adolescence [1]. NHD was first reported in the early 1970s by Nasu et al. [2] and Hakola [3]. Approximately 200 cases have been reported in Japan and Finland [4-8]. Pathological features of NHD include sclerosing leukoencephalopathic lesions in the cerebrum and membranocystic lipodystrophic lesions in the bone marrow [9]. It is designated polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy. Recent studies have shown that NHD is caused by a mutation in TREM2 or DAP12 [10]. The DAP12 is expressed in NK cells, myeloid cells, and oligodendrocytes, whereas the TREM2 is expressed in myeloid cells. The level of intracellular Ca2+ is elevated to activate microglia and is involved in osteoclast and dendritic cell differentiation and function [11]. Clinically, patients with NHD present with recurrent bone fractures during 30 years of life and frontal lobe syndrome during 40 years of life. The dementia exhibits a relentless exacerbation, culminating in the eventual transition to a vegetative state within 50 years of life [12]. We present a case of non-union with the NHD that was successfully managed through prosthetic replacements subsequent to the lack of efficacy in open reduction and internal fixation procedures for a fracture of the left proximal humerus, and an ipsilateral femoral neck fracture.

Case Presentation

A 41-year-old man with NHD had a fall and sustained proximal humerus and femoral neck fractures. He was referred to the hospital for surgical treatment two weeks after the injury. Eight years before his initial visit, a loss-of-function mutation in the DAP12 gene was identified at another hospital, and a diagnosis of NHD was made. Subsequently, rapid progression to presenile dementia was observed, and the patient was treated at the department of psychiatry. Additionally, he had previously undergone conservative treatment for a fracture in his right ankle around the age of 20. (Figure 1). He had undergone open reduction internal fixation (ORIF) for a left femoral neck fracture (non-displaced type) at another hospital six years before his first visit. At the initial visit, an X-ray photograph (Figure 2, 3A) showed a proximal humerus fracture (Neer classification: 3-part fracture; AO classification: 11-2B) and loosening of the implant in the left hip. Computed tomography showed atrophy of the femoral head and multiple bone cysts in the proximal femur (Figure 2B). Surgery was performed under general anesthesia, and the left femoral implant was removed. An artificial head replacement (DePuy Synthes, Inc. Global Unite®2014) was performed for the left proximal humeral fracture (Figure 4). A postoperative X-ray photograph ten days after the previous surgery showed a collapse of the left femoral head. Subsequently, we diagnosed the left femoral neck fracture and performed Bipolar Hip Arthroplasty (Zimmer Biomet Inc. CMK stem®) (Figure 5). The postoperative course was uneventful. The patient primarily relied on a wheelchair for movement for a period of 2 years after the surgical procedure due to cognitive deterioration.

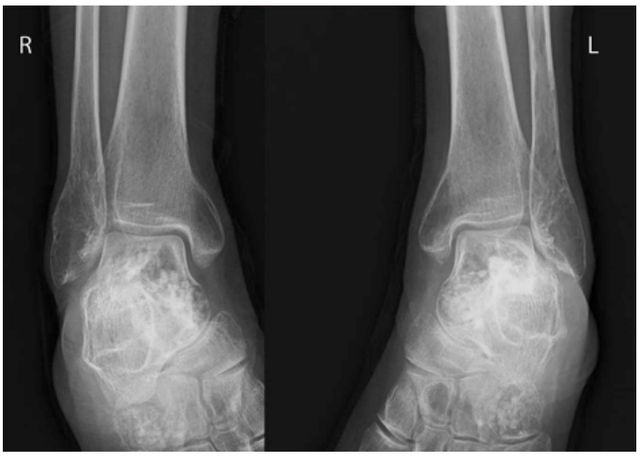

Figure 1: Radiographic image of the bilateral ankles at the time of the initial visit. Multiple bone cysts are observed in both the ankles showing characteristics of NHD.

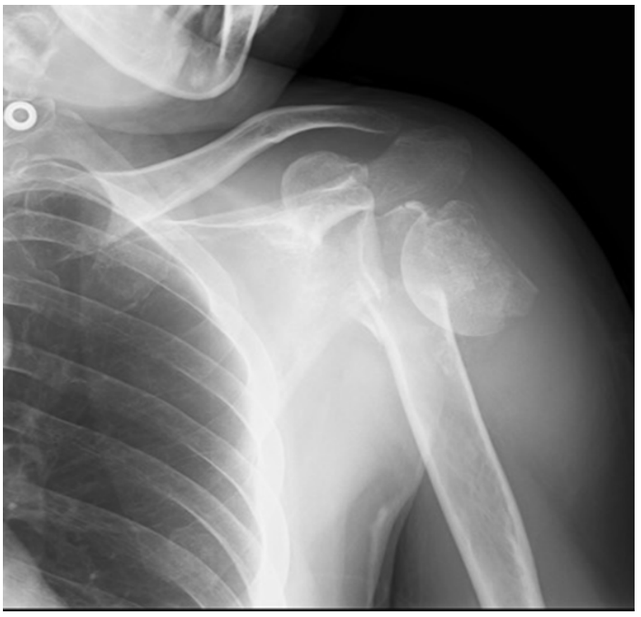

Figure 2: Radiographic image of the left shoulder at the time of the initial visit.

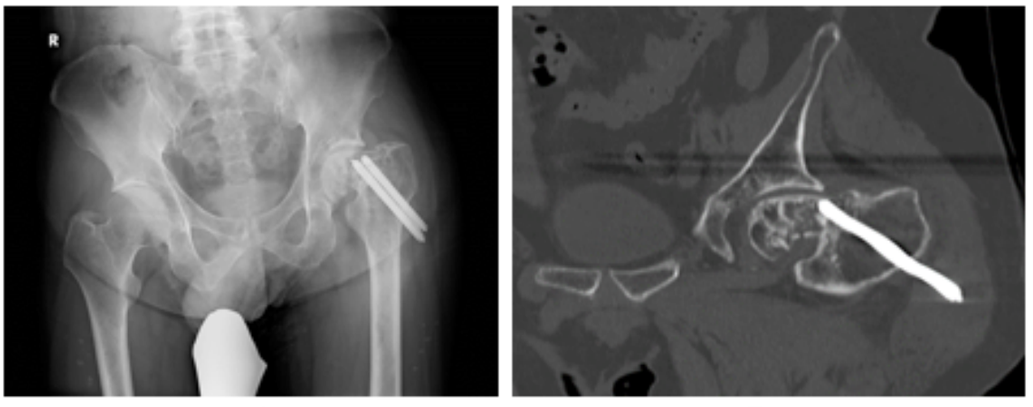

Figure 3: Radiographic images of the left hip joint at the time of the initial visit. A: X-ray photograph. B: Computed tomography image.

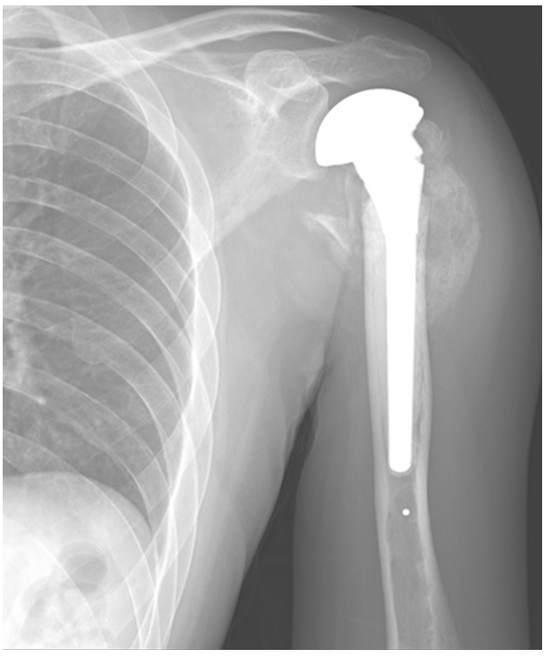

Figure 4: Radiographic image after artificial humeral head replacement.

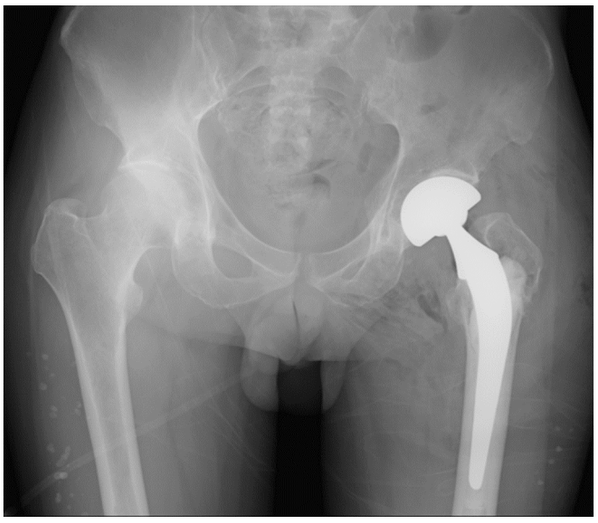

Figure 5: Radiographic image after BHA.

Discussion

The clinical manifestations of NHD often begin with multiple fractures in young patients due to multiple cystic lesions. Thus, orthopedic surgeons are likely to treat NHD first. However, orthopedic surgeons are not familiar with NHD because they are extremely rare. When multiple fractures with bone cysts are observed in young patients, we should be suspicious of NHD and cognitive decline should be detected. Generally, Garden stages I and II are classified as non-dislocation types, and stages III and IV are classified as dislocation types. ORIF is indicated for non-dislocation type fractures [13,14]. ORIF and late segmental collapse were more frequent in displaced fractures than in non-displaced fractures. Thus, prosthetic replacement is recommended for displaced fractures14. In this case, ORIF was selected for a non-dislocated type femoral neck fracture at the time of the initial surgery at a previous hospital. However, bone cysts occur frequently in the epiphyseal region of patients with NHD, which indicates a risk of implant failure and pseudoarthrosis. Prosthetic replacement should be recommended even in younger patients with NHD. In addition, the short average life expectancy of patients with NHD suggests a more positive selection for prosthetic replacement [15]. Although total hip replacement is recommended for young and active patients with femoral neck fractures, [16] BHA may be a better choice for femoral neck fractures with NHD because multiple bone cysts are located in the epiphyseal region and not in the acetabulum. ADL impairment due to cognitive decline will provide us with a more positive selection of BHA. Haruta et al. demonstrated that patients with NHD did not manifest any typical neuropsychiatric symptoms during the 16-year follow-up period [17]. The patient then underwent curettage and bone grafting. No recurrence of lesions or occurrence of new cystic lesions was noted. Curettage and bone grafting may be indicated in young patients with NHD.

Conclusion

Prosthetic replacement should be indicated for metaphyseal fractures with NHD because of polycystic lipomembranous osteodysplasia, which is a specific feature of NHD.

Ethical Considerations: The patient was informed that their data and images would be submitted for publication and gave their consent.

Conflict of Interest: None.

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