# Case reports

## Healing fragile bones: a case report on hyperbaric oxygen therapy in pycnodysostosis

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***Corresponding author:*** *Dr Kubra Canarslan-Demir, SBÜ-Gülhane Eğitim ve Araştırma Hastanesi, Sualtı Hekimliği ve Hiperbarik Tıp Kliniği, Etlik/Ankara, Turkey*
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**Abstract**

(Canarslan-Demir K, Yel AK, Aydin G, Zaman T. Healing fragile bones: a case report on hyperbaric oxygen therapy in pycnodysostosis. Diving and Hyperbaric Medicine. 2025 30 June;55(2):191–194. doi: 10.28920/dhm55.2.191-194. PMID: 40544148.) Pycnodysostosis is a rare lysosomal storage disorder characterised by short stature, craniofacial dysmorphisms, dental anomalies, and increased bone fragility due to osteoclast dysfunction caused by cathepsin K gene mutations. This case report describes a 43-year-old female pycnodysostosis patient with recurrent subtrochanteric fractures and delayed bone healing following multiple surgical interventions, including femoral osteotomy and bone grafting. Despite these efforts, bony union was not achieved. The patient underwent 39 sessions of hyperbaric oxygen therapy (HBOT), administered at 243.2 kPa for 120 minutes daily, five days per week. Post-treatment radiographs revealed significant fracture healing, with improvements continuing one month after therapy. Visual analogue pain scores decreased from 4 to 1, and quality of life (SF-36) improved. HBOT enhances tissue oxygenation, stimulating osteogenesis, neovascularization, and immune responses, while optimising osteoclast function, making it a promising treatment for pycnodysostosis-related fracture complications. Although ideal, a controlled trial of HBOT in this rare disorder is probably unachievable. Nevertheless, this report highlights HBOT as a potentially useful adjunctive treatment for enhancing healing of refractory fractures in pycnodysostosis patients.

### Introduction

Pycnodysostosis is a rare lysosomal storage disorder characterised by short stature, acroosteolysis of the distal phalanges, craniofacial dysmorphisms (e.g., midface hypoplasia, convex nasal ridge, prominent forehead, and micrognathia), dental anomalies, osteosclerosis, and increased bone fragility.[1,2] The pathogenesis of pycnodysostosis is associated with a mutation in the gene located on chromosome 1q21 that encodes the enzyme cathepsin K (CTSK). CTSK is an essential lysosomal cysteine protease that plays a key role in bone resorption and remodelling processes[3] by degrading bone matrix proteins such as type I and type II collagen, osteopontin, and osteonectin under low pH conditions.[4] A deficiency of this enzyme leads to osteoclast dysfunction, resulting in inadequate degradation of bone matrix proteins and an abnormally fragile bone structure.[5]

Pycnodysostosis is considered a rare disorder, with its prevalence estimated at approximately 1 to 1.7 per million.[1] Patients are typically diagnosed during childhood due to delayed anterior fontanel closure or short stature. In adulthood, however, they often seek medical attention for recurrent fractures, particularly in long bones, resulting from low-energy trauma.[5] As observed in our case, difficulties in fracture healing pose a significant challenge in the management of pycnodysostosis with no consensus regarding optimal treatment. While surgical methods are predominantly favoured, conservative approaches have been utilised in specific cases.[6]

This case report describes a 43-year-old female patient referred to our clinic by orthopaedic and traumatology specialists due to recurrent subtrochanteric fractures and delayed fracture healing following a right femoral osteotomy. In this case, the impact of pycnodysostosis on the bone healing process was thoroughly evaluated, and the potential role of hyperbaric oxygen therapy (HBOT) in this context was explored.

## Case report

The patient provided written informed consent for the publication of the case details and accompanying images.

A 43-year-old patient, previously diagnosed with pycnodysostosis, underwent surgery for a right femoral shaft fracture. The initial treatment involved open reduction and internal fixation with a plate and screws. Fifteen years later, due to bone deformation, the operation was revised. However, bony union was not achieved after the revision surgery. One-year post-revision, the patient had another fracture proximal to the initial site due to trauma and underwent surgery. The same surgical procedure was employed, yet bony union was again not obtained. Nine months later, a bone grafting operation was performed to address the issue.

At her initial evaluation at our clinic, conducted two years after the revision surgery, she reported experiencing hip pain during movement, with a visual analogue scale (VAS) score of four out of 10, as well as reduced hip mobility. Computerised tomography (CT) and X-ray imaging revealed one subtrochanteric fracture line with bone graft, one fracture line on the femoral shaft, one plate and 16 screws.

We performed 39 HBOT sessions (243 kPa for 120 minutes), once daily, five days a week. No complications or adverse effects related to HBOT were observed. A radiograph taken after the 39 HBOT sessions revealed callus formation at the distal fracture site. Additionally, a radiograph taken one month after the HBOT sessions showed healing at the proximal fracture line (Figure 1). The patient's hip pain, as measured by the VAS score, decreased from 4 to 1.

The Short Form-36 (SF-36) and Beck Depression Inventory (BDI) was administered at the first and last of the treatments (Table 1). Four months after the treatment, the patient reported no recurrence of fractures or pain.

## Discussion

This case report highlights the challenges associated with long bone fracture healing in pycnodysostosis patients and examines the potential benefits of HBOT in overcoming these difficulties. Pycnodysostosis is characterised by defective bone resorption due to osteoclast dysfunction, leading to increased bone fragility.[5] This condition results in frequent recurrent fractures, either spontaneous or caused by low-energy trauma, as well as delayed healing processes in pycnodysostosis affected patients.

In our case, multiple surgical interventions were performed to address osteotomy and non-union of recurrent subtrochanteric fractures in the right femur. However, bony union could not be achieved following any of these procedures. This outcome is consistent with the osteoclast dysfunction that underlies the fundamental pathophysiology of pycnodysostosis. The inability of osteoclasts to degrade bone matrix proteins disrupts the bone remodelling process, significantly increasing the risk of nonunion.[5]

Following 39 sessions of HBOT, radiographic evaluation demonstrated healing at the distal femoral fracture site, with additional healing observed at the proximal femoral fracture site one month later. Furthermore, HBOT was associated with a reduction in the patient's pain VAS score, an improved quality of life as measured by the SF-36 scale, and an enhanced mood state as indicated by the Beck Depression Inventory.

HBOT involves the administering 100% oxygen to patients in a hyperbaric chamber, where the pressure exceeds normal atmospheric levels (101.3 kPa). This process

**Figure 1**

Radiographic images taken before HBOT (left), at the end of HBOT (middle), and one month after the completion of HBOT (right); green arrows indicate areas of fracture healing; blue arrows indicate callus formation at the fracture site

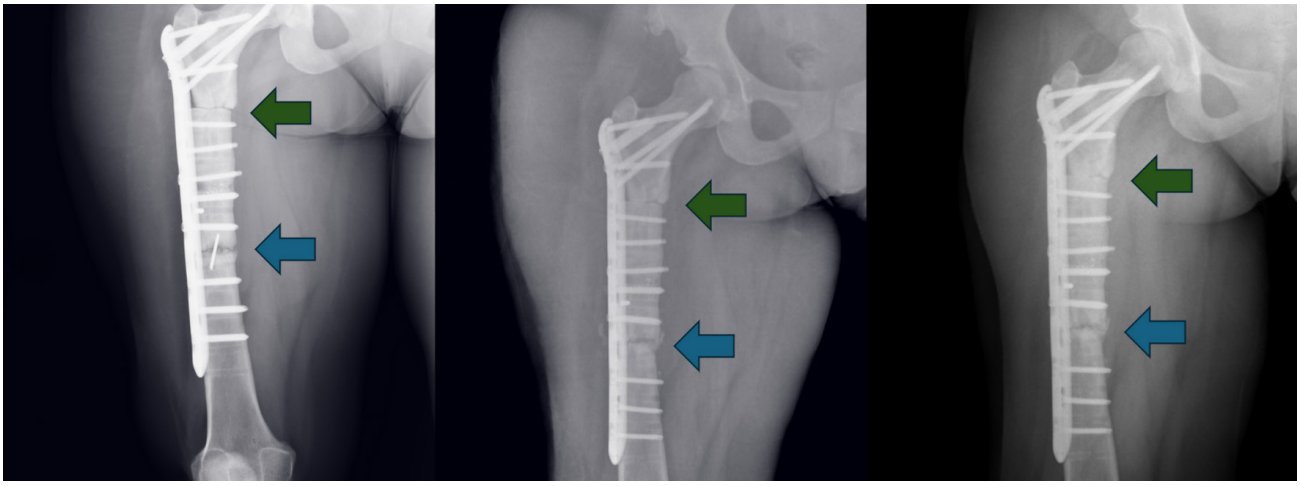

**Table 1**
Short Form-36 and Beck Depression Inventory scores before and after treatment

| Short Form-36 scores | | |
|---|---|---|
| **SF-36 Domains** | **Pre** | **Post** |
| Physical functioning | 5 | 40 |
| Role Limitations – Physical | 0 | 0 |
| Role Limitations – Emotional | 0 | 0 |
| Energy/fatigue | 40 | 55 |
| Emotional well-being | 56 | 52 |
| Social Functioning | 50 | 75 |
| Pain | 25 | 75 |
| General Health | 25 | 55 |
| Health Change | 50 | 75 |
| **Beck Depression Inventory scores** | | |
| **Pre** | **Post** | |
| 12 | 3 | |

increases oxygen delivery to partially ischaemic or hypoxic tissues, enhancing oxygen-dependent leukocyte activity by stimulating the production of reactive oxygen species such as hydrogen peroxide and superoxide.[7] Additionally, hyperoxia promotes bone osteogenesis and supports the development of neovascularization, aiding in the replacement of damaged tissue with healthy bone. The improved vascular network enables better infiltration of immune cells, antibodies, and antibiotics into affected areas. Furthermore, HBOT aiding the clearance of bone debris by optimising osteoclast function.[7]

HBOT appeared to make a significant contribution to the treatment of this patient. Numerous studies have demonstrated that HBOT supports fracture healing both histologically and radiologically, accelerates callus formation, increases bone mineral density, and enhances osteoblastic activity and neovascularization.[8–10] Furthermore, a recent study suggests that hyperbaric oxygen therapy (HBOT) may support bone healing in refractory orthopaedic conditions such as avascular necrosis by preventing disease progression and reducing the need for surgical intervention.[11] Given their high metabolic activity, osteoblasts and osteoclasts are highly dependent on oxygen. HBOT enhances tissue oxygenation, providing the oxygen required by both osteoblasts and osteoclasts, potentially boosting their activity. This process supports new bone formation and accelerates the fracture healing process. Therefore, HBOT contributes to the early union of fractures and positively influences the balance between bone formation and resorption processes.[12,13] These mechanisms suggest that HBOT may serve as a potential treatment option for addressing complications such as non-union in pycnodysostosis patients.

Importantly, throughout the treatment sessions, no complications or adverse effects related to HBOT were observed. HBOT is a safe treatment when administered under proper protocols and monitoring. While there are potential side effects of HBOT, such as barotrauma or oxygen toxicity, the absence of such complications in this case highlights its tolerability, even in a complex condition like pycnodysostosis.

The 39-session HBOT protocol applied in this case was associated with a significant reduction in the patient's pain scores and a noticeable improvement in quality of life (SF-36). Significant improvements observed in the 'Physical Function' and 'Pain' subscales underscore the potential benefit of HBOT not only in pain alleviation but also in facilitating the patient's engagement in daily activities and enhancing overall quality of life. These findings suggest potential value of HBOT as an adjunctive treatment option in the challenging fracture healing processes associated with conditions such as pycnodysostosis.

Although ideal, a controlled clinical trial of HBOT in this rare indication is probably unachievable. Nevertheless, this report identifies HBOT as a potentially useful adjunctive treatment for fracture healing in pycnodysostosis patients. Additional factors influencing bone healing in pycnodysostosis patients, such as nutritional status, comorbidities, and genetic variations, must also be considered. The optimal HBOT protocol, including parameters such as the number of sessions, pressure, duration, and combination with other therapeutic modalities is unknown, but as indicated above, would be hard to study.

In conclusion, this case report highlights the potential efficacy of HBOT in addressing fracture healing impairments in pycnodysostosis patients and emphasises the need for exploring novel therapeutic strategies for this rare genetic disorder.

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