

## Case Report

### Toulouse-Lautrec Disease: A Case Report of a Femur Shaft Fracture

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#### Abstract

Toulouse-Lautrec disease is a lysosomal autosomal recessive disease responsible of condensing osteopathy and fragility of the bones. The case report featured a 24-year old woman with a right femoral shaft fracture treated by open reduction and plate fixation. Osteosynthesis by nail was not possible because the femur medullar tunnel was obstructed. Before surgery, the surgeon must be aware of the potential obstruction of the femoral medullar tunnel in this type of patient.

**Keywords:** Condensing Osteopathy; Femoral Shaft Fracture; Pycnodysostosis; Toulouse-Lautrec Disease

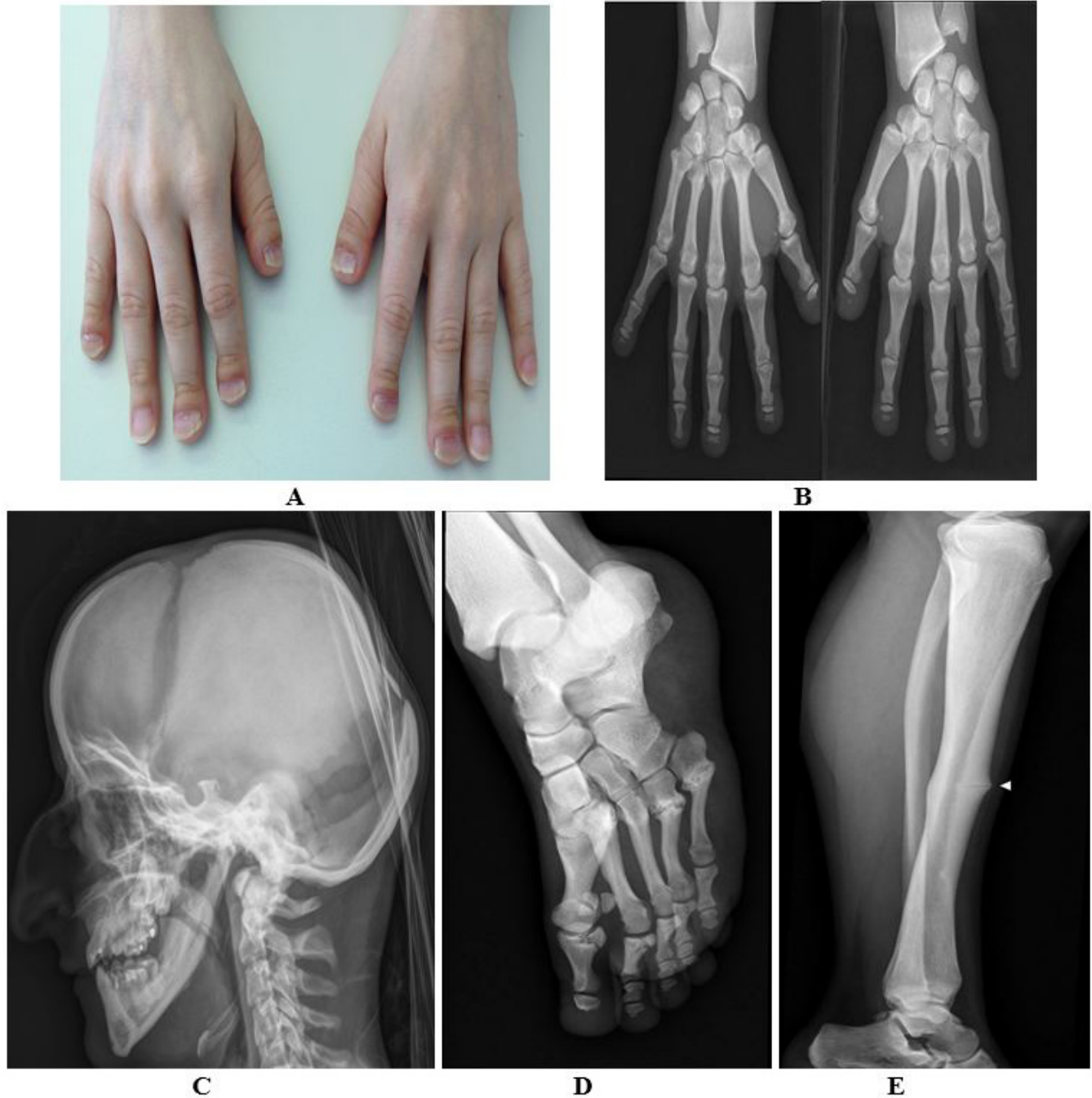
#### Introduction

The first case of pycnodysostosis was described by Montanari in 1962 [1] and then named in 1965 Toulouse-Lautrec disease after it was found out the famous 19th century painter had it [2,3]. The prevalence of pycnodysostosis is unknown in the literature. We report a case of pycnodysostosis with a femur diaphysis shaft treated by open reduction and plate fixation. This method was chosen as a result of the characteristics of the disease forcing to avoid nail fixation.

#### Case Report

A 24-year old woman was referred to our hospital with a right femoral shaft fracture which occurred as she got out of her car. She had already been diagnosed with pycnodysostosis as a young child. No history of pycnodysostosis or consanguinity had

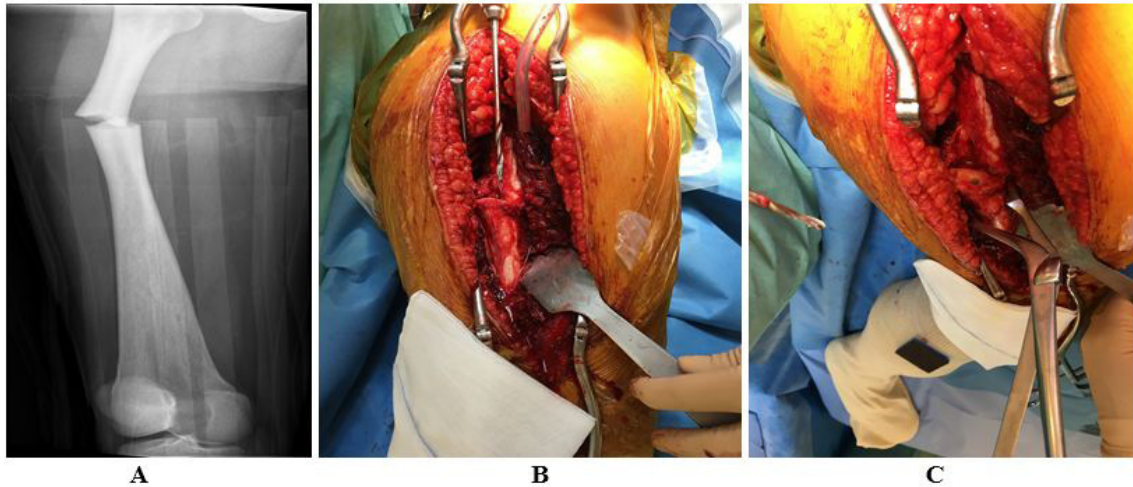
previously been noted in her family. The physical examination highlighted a distinctive dysmorphic syndrome with a stature deficiency (141cm, 43kg), a bradymetacarpia with short and brittle nails (Figure 1A), facial dimorphic disorder showing a small face with prominent frontal lump, a protruding nose, a large forehead and a micrognathia. The mouth analysis displayed dental issues. No abdominal organomegaly was found and the blood tests were standard, especially phospho-calcic balance. Hands and feet X-rays showed an acro-osteolysis and an aplasia of the last phalanges of the fingers and toes (Figure 1B). The cranium radiography emphasized the anterior fontanel and the presence of abnormally large sutures (Figure 1C). The remainder radiographs of the peripheral skeleton underlined asymptomatic fractures at the base of the first phalanges on the first and second toes, at the base of the fourth and fifth left foot metatarsal, and an anterior unicortical fracture of the left tibial diaphysis which had been treated a few months earlier conservatively (Figure 1D & 1E). All these lesions occurred as a result of low energy trauma.



**Figures 1(A-E): Toulouse-Lautrec disease: a case:** **A)** Bradymetacarpia with short and brittle nails; **B)** Hands x-ray showing an acro-osteolysis and an aplasia of the last phalanges of the fingers; **C)** Cranium X-ray showing an osteocondensation of the base of the skull, Hypoplasia of the bones and the lower jaw, with disappearance of the mandibular angle; **D)** Left foot x-ray showing fractures at the base of the first phalanges on the first and second toes, at the base of the fourth and fifth left foot metatarsal. **E)** Left leg x-ray showing an anterior unicortical fracture of the left tibial diaphysis (white arrow).

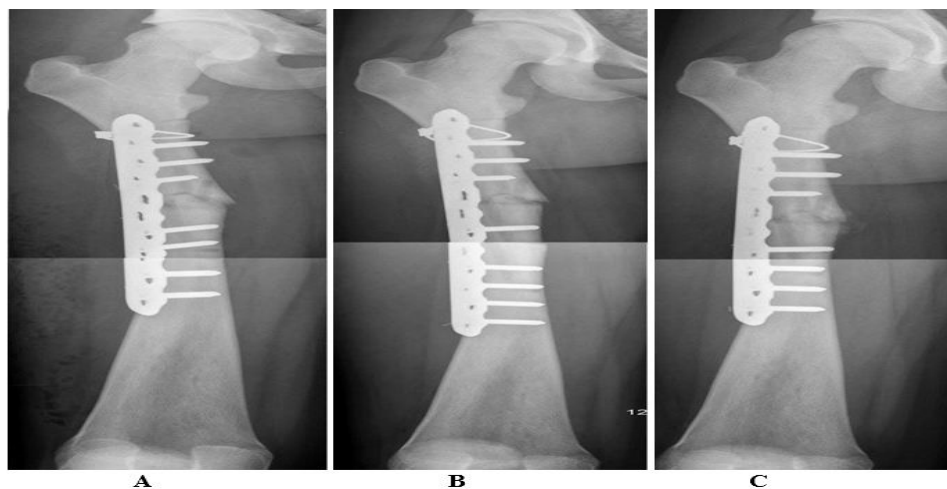
It was decided that the right femur shaft fracture would be treated by surgery (Figure 2A). The patient was laid on the surgery table in a left lateral decubitus position after general anaesthesia associated with a locoregional anaesthesia achieved by femoral nerve block. The lateral approach of the thigh enabled access to the centre of the fracture (Figure 2B). After reduction using Verbrugge pliers, it was

decided to proceed to an osteosynthesis with 10 holes 4,5mm long LCP plate (Depuy Synthes™). Three locked screws were placed above the centre of the fracture and another three were positioned under the centre. During surgery, it was observed that the femur medullar cavities were obstructed which would have prevented an osteosynthesis using an intramedullary nail. The cavities have been made permeable again by drilling through them in order to increase the intake of bone growth factors (Figure 2C). A bone density unusually high made difficult the drilling and screwing of the femoral corticalis.



**Figures 2(A-C): Preoperative view of a right femur fracture in a case of Toulouse-Lautrec disease. A)** X-ray showing a shaft fracture of right femur; **B)** Lateral approach of the thigh enabled access to the center of the fracture. The operator was about to permeabilize the medullary tunnel with the drill; **C)** Appearance of the femoral shaft after drilling.

Post-surgery, weight-bearing was prohibited for a three months period. The day following surgery, rehabilitation, by strengthening of the quadriceps muscle, begun along with support of the articular range on the limb which underwent surgery, and the learning of how to walk using crutches. The immediate post-surgery consequences did not show any complications such as cutaneous healing or hematoma issues. The progression was effective and allowed the patient to stand at three months with no associated pain or physical aid. The femur X-rays taken during the follow-up eventually showed a bone callus on the third month (Figure 3). This finding confirmed the existence of a slow bone healing with this type of patient.



**Figure 3: Radiographic postoperative result of a right femur fracture treated by plate fixation. A)** after 1 week; **B)** after 1 month; **C)** after 3 months.

## Discussion

The pycnodysostosis (from Greek, pycnos = dense, dys = faulty, osteon = bone) is a seldom bone dysplasia at the root of the increase in density and fragility of the bones leading to repetitive shock less fractures impacting mainly the lower limbs and especially the femur. It is a lysosomal autosomal recessive disease responsible of condensing osteopathy. The anomaly is located on the 1-chromosome in 1q21 which codes for cathepsin K [4, 5]. It is a lysosomal protease secreted by the osteoclast enabling the proteins breakdown from the bone matrix such as Type I collagen, osteonectin, or osteopontin. The pycnodysostosis results from mutation of this gene and is characterised by bones that are highly dense but also fragile due to bone restructuring related problems [6-9].

Generally, this disease is diagnosed during childhood where a small body height clashes with a relatively large skull, a lack of closure on the anterior fontanel and a facial dysmorphism. The illness can sometime be spotted during the adult age either after a fracture or during a punctual check-up. From a clinical perspective, the patients display distinctive cranial malformations with a large skull associated with the presence of wormian bones, a persistence of the anterior fontanel and a mandibular hypoplasia. The thorax is narrow with a collarbone dysplasia. Small and square hands and feet feature short fingers and toes resulting from an acro-osteolysis and an aplasia of the last phalanxes. Dental and ungual issues can also be observed as well as a bone fragility, which causes fractures. These fractures occur after impacts happening at low-energy, in a repetitive manner, and essentially affecting the lower limbs [10,11]. The exact pycnodysostosis prevalence remains unknown but is estimated to range within 1 and 1.7 over a million childbirths with an even distribution across both sexes [12]. As per any recessive autosomic transmissible diseases, consanguinity increases the risk of appearance. It arises in 30% of the cases.

No specific treatment against pycnodysostosis is currently available [13]. The use of growth hormones produces encouraging results on the development [14]. The genetic therapy is still at the experimental stage similarly to the substitutive enzymotherapy and the cathepsin K inhibitors used in the treatment against osteoporosis [15-17]. Most of the treatment primarily bears a preventive aim with an early screening and a regular monitoring of the fractures, which strengthening is often slowed down. The screening test in the pre-birth diagnosis is not done systematically, but is considered for the couples that have already had a child affected by the disease [18-23]. In our cas, the obstruction of the medullar tunnel has been noticed while surgery was in progress. This physical anomaly has a crucial impact upon the choice of the fixing system that will be used during the osteosynthesis. Several authors have mentioned about the technical difficulties related to the osteosynthesis via intramedullary nail with locking pin. Some even prefer using a plate in the event of a contralateral femur osteosynthesis [19].

Regardless of the risk of surgical site infection and the loss of the fracture hematoma, a plate fixation yet appeared as an astute choice given the anatomical particularity. This therapeutic alternative is also proposed by Azagui, et al. [20].

Overall, the pycnodysostosis is a seldom cause of condensing osteopathy often diagnosed during childhood, responsible of a typical dysmorphic syndrome associated with an increased bone fragility. It belongs to the genotypic osteopathy group. No curative treatment for this affection currently exists. The approach therefore lies on the complications presented by the patient and particularly any fractures of the peripheral bones such as the case of this report. While proceeding to the pre-surgery exercise, the surgeon must be aware of the potential obstruction of the femoral medullar tunnel.

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