

# DIFFICULTIES IN THE TREATMENT OF FEMORAL ITERATIVE FRACTURE ON AN ADULT WITH LOBSTEIN DISEASE

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**Keywords:** Lobstein's disease, iterative fracture, osteosynthesis

**Abstract:** Lobstein's disease is a genetic autosomal dominant disease (rarely as an individual mutation- "de novo") resulting from qualitative defects of type 1 collagen. Most important clinical characteristic of the disease is easily fractures of bone after minor trauma. Treatment is complex and multidisciplinary aimed at: increasing overall bone strength, preventing and treating fractures and maintaining joint mobility. The treatment of femoral fracture in adult with Lobstein's disease is very difficult because of permanent risks of iterative fracture or osteosynthesis deterioration.

**Cuvinte cheie:** maladia Lobstein, fractura iterativă, osteosinteză

**Rezumat:** Maladia Lobstein o afecțiune genetică autosomal dominantă (rareori ca și o mutație individuală - de novo) caracterizată prin deficitul de colagen de tip I. Caracteristica clinică a bolii o constituie fracturile frecvente produse prin traumatisme minore. Tratamentul bolii este complex și multidisciplinar și are ca obiective: creșterea densității osoase, prevenirea și tratamentul fracturilor, menținerea mobilității articulare. Tratamentul fracturii de femur la un adult cu maladie Lobstein este foarte dificil deoarece există permanent riscul de apariție a fracturii iterative sau deteriorare a montajului de osteosinteză. Articolul prezintă particularități clinicoevolutive la un pacient cu maladie Lobstein.

## INTRODUCTION

Lobstein's disease is known as osteogenesis imperfecta. It is one of the most common skeletal dysplasias characterized by generalized disease of connective tissue. Osteogenesis imperfecta is a genetic autosomal dominant disease (rarely as an individual mutation- "de novo") resulting from qualitative defects of type 1 collagen.(1) One of suggested disease mechanism is that the stress state within collagen fibrils is altered at the locations of mutations, where locally larger shear forces lead to rapid failure of fibrils even at moderate loads as the homogeneous stress state found in healthy collagen fibrils is lost.(2) There are eight different types of osteogenesis imperfecta, type I being the most common, though the symptoms vary from ind to other person.

Most common symptoms of disease are: bones fracture easily, slight spinal curvature, loose joints, poor muscle tone, discoloration of the sclera (whites of the eyes), usually giving them a blue-gray color, early loss of hearing in some cases, slight protrusion of the eyes.

At present time there is no cure for Lobstein disease. Treatment is complex and multidisciplinary aimed at: increasing overall bone strength, preventing and treating fractures, maintaining joint mobility. Bisphosphonates, particularly those containing nitrogen, are being increasingly administered to increase bone mass and reduce the incidence of fracture. Physiotherapy is used to strengthen muscles and improve motility in a gentle manner, while minimizing the risk of fracture. Surgery is addressed mostly to fractures or deformities treatment, although improving bone strength and preventing fractures procedures using intramedullary nails or rods inserted in the long bones was developed..

## CASE PRESENTATION

N.C., 48 years old male, known with Lobstein disease, fell down from the same level at home. Following clinical exam and x-ray (fig.1), made in Emergency Room, proximal 1/3 left femoral fracture was diagnosed. Patient has no familial history of Lobstein disease (osteogenesis imperfecta "de novo"). Clinical history reviewed multiple bone fracture on superior and inferior limbs. General clinical exam revealed blue-gray color of sclera, slight protrusion of the eyes, loose joints and 8 cm shortness of right lower leg because of malunited femoral fracture (fig. 2).

**Figure. No. 1** Preoperator (a) and postoperator (b) aspects of fracture



Because of the history of the left femoral fractures the surgical indication was open reduction and plate osteosynthesis (fig.1). Postoperative evolution was good with clinical and radiological consolidation of fracture after 14 weeks. After 18 months patient came on hospital with left thigh abscess and

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