

Osteolysis around the Fassier-Duval nail and spontaneous femur fracture in a young semi-professional swimmer with Osteogenesis Imperfecta type I: case report and review of literature

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Summary

A 12-year-old male patient with OI type I underwent percutaneous osteotomies with the FD telescopic nail (diameter 5.6). The clinical and radiological follow-ups of the implant were satisfactory, with good bone callus formation and correct positioning of the nail. The patient was permitted to resume swimming from the 9th post-operative month, though, unbeknownst to his caregivers, he began a semi-professional freestyle swimming training load. As a result, the patient experienced acute thigh pain with immediate functional impotence 18 months after surgery. The X-rays showed an incomplete transverse fracture in the upper middle third of the femur. The patient was subsequently placed in a long leg cast, with no weight bearing for 3 weeks, and prescribed magnetotherapy until clinical and radiological healing was reached, 2 months later.

We believe this case should give pause for thought as to which indications to give these patients with regard to sports activities, and the need to increase awareness in OI patients and their families. We also believe it is best to discourage, in these particular patients, any form of semi-professional competitive activity, including competitive swimming.

KEY WORDS: Osteogenesis Imperfecta; Fassier Duval nail; femur fracture; sport; activity; bone healing.

Introduction

Osteogenesis Imperfecta (OI) is a rare hereditary multi-systemic pathology that affects the connective tissue (1). The main clinical features of orthopaedic interest are osteoporosis, fragility fractures and the presence of long bone deformities (2). These characteristics result in extreme bone fragility and, consequently, fractures that can occur as a result of low energy traumas (3-5). The associated comorbidities (which can complicate and influence the anesthesiological procedures) include basilar impression, airway deviations and facial mass deformities, respiratory impairment (secondary to the presence of severe scoliotic curves), cardiopathies, capillary fragility and impaired platelet function (6, 7). The current therapy protocol is neridronate 2 mg pro kg in saline solution 100 ml infused intravenously in two hours each 3 months, in order to simultaneously reduce the fracture pain and determine alterations to bone segments by increasing the firmness and thickness of the cortical (5, 8-10).

The objective of the orthopaedic surgeon is to prevent fractures by treating the deformities, in addition to educating and raising awareness, in both the patient and the family, as to the daily living habits and sports activities allowed, or recommended, with regards to this pathology (3, 4, 11-13).

Any sports activity that may expose the patient to trauma and/or fractures is not recommended. Swimming is the best choice for these patients, as it is a low-impact non-weight bearing activity.

We present the case of a 12 year-old male with OI type I who, in 2015, underwent a fracture synthesis of the right femur with a telescopic Fassier-Duval (FD) nail. The clinical and radiological follow-up examinations were deemed satisfactory and he was allowed to pursue swimming as a sport activity. Then, 14 months after the surgery, the patient presented with a spontaneous femoral fracture and the radiological examination revealed a plastic deformity of the telescopic nail and a diaphyseal osteolysis.

Ethical approval: All the procedures performed in the study involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki Declaration and its subsequent amendments or comparable ethical standards. Informed consent was obtained from all the individual participants included in the study.

Clinical case

In January 2015, a 12-year-old male patient with OI type I was admitted with a fracture on the femoral deformity, for which he was treated with percutaneous osteotomies with the FD telescopic nail (diameter 5.6).

In his medical history, the patient described other 5 previous

fractures: the first one happened 45 days after his birth, it was a right femoral shaft fracture, then at the age of 14 months he underwent surgery for a left femoral shaft fracture, which was fractured again 3 months later and treated conservatively. The other two fractures involved both the right femoral shaft. The first one at the age of 5, it was treated surgically with an intramedullary nailing, then one year later the same femoral fracture was treated using just a cast. The patient did not have any other concomitant diseases, nor took any other concomitant medications.

The pharmacological therapy consisted in neridronate 2 mg pro kg in saline solution 100 ml, infused intravenously in two hours each 3 months. It was suspended post-surgically for 4 months in order to do not interfere with the healing process of the fracture and the osteotomies (7, 9, 11). The vitamin D intake administered to the patient was 1000 International Units (IU) per day. The calcium intake with the diet was 1 g per day. The last bone metabolism exams performed by the patient before the fracture showed the following values:

- serum calcium: 9.6 mg/dL (range: 8.4-10.8 mg/dL)
- inorganic phosphorus: 5 mg/dL (range: 3.4-6.2 mg/dL)
- alkaline phosphatase: 264 U/L (range: 42-362 mg/dL)
- osteocalcin: 59.94 ng/mL (range: 24-70 ng/mL)
- beta-Cross Laps (CTX): 0.64 ng/mL (range: 0.1-0.584 ng/mL)
- Parathormon (PTH): 21.9 pg/mL (range: 15-65 pg/mL)
- Vitamin D: 23 ng/mL (range: 4.92-42.70 ng/mL).

The clinical and radiological follow-ups of the implant were satisfactory, with good bone callus formation and correct positioning of the nail (Figure 1). The patient did not report any pain and had a complete articular range of motion.

Per request of the parents, swimming was permitted from the 9th post-operative month. Infusion therapy with bisphosphonates was suspended on September 2016 to normalise the DEXA values (Z score = -2.0).

Unbeknownst to his caregivers, the patient, in light of his optimum clinical and psychical condition, began a semi-professional freestyle swimming training load with 4 workouts per week, consisting of 40 laps per training session.

18 months after surgery, during one of his training session, the patient experienced acute thigh pain with immediate functional impotence.

The radiological examination showed an incomplete transverse fracture in the upper middle third of the femur, with a diaphyseal osteolysis around the entire FD nail (Figure 2). Although the device was positioned correctly, with good proximal/distal anchorage and the expected slide-extension, it appeared bent, permanently deformed (plastic deformation) along its entire length.

Targeted and repeated laboratory tests were performed 3 times per week, in order to rule out possible infectious complication. The absence of any acute signs and symptoms, as well as the negative results of the anamnesis and laboratory tests, excluded such complication.

The patient was subsequently placed in a long leg cast, with no weight bearing for 3 weeks, and prescribed magnetotherapy until clinical and radiological healing was reached after 2 months (Figure 3).

Discussion and conclusions

Considering their high bone fragility, the choice of sport activities for patients with OI is influenced by the risk of trauma



Figure 1 - X-ray in frontal view, showing the inferior limb and the right femur 1 year after the surgical treatment for correction of deformities and osteosynthesis with Fassier Duval telescopic nail.

and fracture.

It is however described by Van Brussel et al. (14) that a supervised training program can improve aerobic capacity and muscle strength, reducing levels of subjective fatigue in children with OI type I and IV in a safe and effective manner. Other Authors recently asserted that physical activity, incor-



Figure 2 - X-ray showing the fracture in the upper middle third of the femur, occurred to the patient during a swimming training session.



Figure 3 - X-ray showing the fracture healing 2 months after the injury.

porating strengthening exercises, could be an interesting approach for improving muscle function in children and adolescents with OI type I, so an higher volume of physical activity could prevent adverse effect of aging on muscle function observed in OI patients type I (15).

Swimming is, therefore, considered safe due to the low impact and non-weight bearing nature of this activity. Literature analysis proves that non-weight bearing activities, such as swimming, do not stimulate the development of bone mineral density (16, 17).

Additionally, repetitive technical gestures can place long bones under constant stress, influenced by the motion levers.

When free style swimming is correctly executed, the athlete coordinates his/her breathing with the upper and lower limb movements, averaging 6 leg kicks per 2 arm cycle rotations (18-20).

Our patient, therefore, performed an average of 8 arm cycles and 48 leg kicks per lap (25 metres). The training included 40 laps per training session, 4 times a week, which equals to 7680 legs kicks.

Given the correct positioning of the implant (with the expected slide-extension of the nail), as well as a positive clinical and radiological fracture healing assessment, we reached the conclusion that the intramedullary synthesis medium plastically deformed during the repeated hip and knee flexion against the water's resistance, resulting in an osteolysis around the implant and a subsequent spontaneous femoral fracture.

We believe this case should give pause for thought, as to which indications to give these patients with regard to sports activities and the need to increase awareness in OI patients and their families.

It has already been established that traumatic or contact sports are not recommended as they put patients at risk of fractures. However, we also believe it is best to discourage any form of semi-professional competitive activity, including competitive swimming, which, despite being low-impact and/or conventionally harmless, involves cyclic gestures and repeated stresses, particularly in patients fitted with intramedullary nails.

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