

FEMORAL SHAFT FRACTURE AS A RESULT OF INDIRECT TENSILE FORCES DURING EPILEPTIC SEIZURE IN PATIENT WITH JOUBERT SYNDROME - CASE REPORT

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received: 10.7.2019;

revised: 18.9.2019;

accepted: 1.12.2019

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INTRODUCTION

Musculoskeletal system injuries are rarely the result of convulsive seizures if there is no external trauma during the episode. However, in some cases, convulsive seizures can generate so intense tensile forces that secondary cause musculoskeletal injuries (Finelli & Cardi 1989). In the available literature, the most commonly described are the anterior and posterior shoulder dislocations, as well as various other injuries resulting from convulsions, such as compressive vertebral fracture, humeral fracture, sacral fracture, sternal fracture, femoral neck fracture, etc (Mimata et al. 2016, Sharma et al. 2017, Shah et al. 2014). Femoral shaft fracture has never been described as a complication of a convulsive episode in the literature so far.

The thigh bone or femur is the longest and strongest bone in the human skeleton. Not including femoral neck and pertrochanteric fractures, the femoral (diaphyseal) fractures incidence is 9.5-18.9 per 100,000 people per year. Femoral shaft fractures are most commonly caused by direct force such as in traffic accidents, falling from height, pedestrian accidents or firearms injuries, but can also occur by indirect force like in spiral femoral shaft fracture in skiers. Injury is often associated with predisposing factors such as osteopenia in older people, hyperparathyroidism, hypocalcemia, hypophosphatemia, etc. Femoral shaft fractures may be life-threatening when associated with femoral artery injuries or development of compartment syndrome (Enninghorst et al. 2013, Salminen et al. 2000).

CASE REPORT

Medical history

A 32-year-old male with a previously diagnosed cerebral palsy and symptomatic epilepsy was admitted due to an injury of the left upper leg. By heteroanamnesis from patient's mother, it was acknowledged that

he suffered convulsive seizure, and during that time she heard a breaking noise. There was no external trauma. The last seizure was 10 days prior to this one. He had phenytoin in chronic therapy.

Clinical examination

Neurological examination showed that the patient was conscious, with the production of inadequate speech. His pupils were isochoric, with normal consensual reactions to light. The neck was motile, meningeal syndrome was negative. Patient had severe contractures of the joints. He was in the diaper, with fecal and urine incontinence. The orthopedic examination showed that the patient had an asthenic physique, with deformity in the distal part of the left upper leg. Inspection did not reveal the skin discontinuity or contusions. Interruption of bone continuity was revealed by palpation in area of deformity. No neurovascular deficits were found distally.

Diagnostic work-up

The standard laboratory results revealed sideropenic anemia (hemoglobin 79 g/L) and mineral deficiency. Plain radiograph of the left upper leg in the anteroposterior (AP) projection showed transverse femoral shaft fracture, located in distal third of the bone, with the shift to the longitudinal bone axis (Figure 1A). Additionally, it also showed extensive osteopenic changes, and femoral neck abnormality – possible previous healed femoral neck fracture. Radiograph in laterolateral (LL) projection could not be taken due to patient's condition. Standard multi-slice sections and multiplanar reconstructions through the brain using GE Lightspeed 16 Slice CT Scanner (General Electric, Chicago, IL, USA) showed left cortical atrophic changes, with a thickened cortex. Brain scans using Magnetom Skyra 3T MRI Scanner (Siemens, Erlangen, Germany) revealed following specifics: brainstem, pons and mesencephalon of atypical

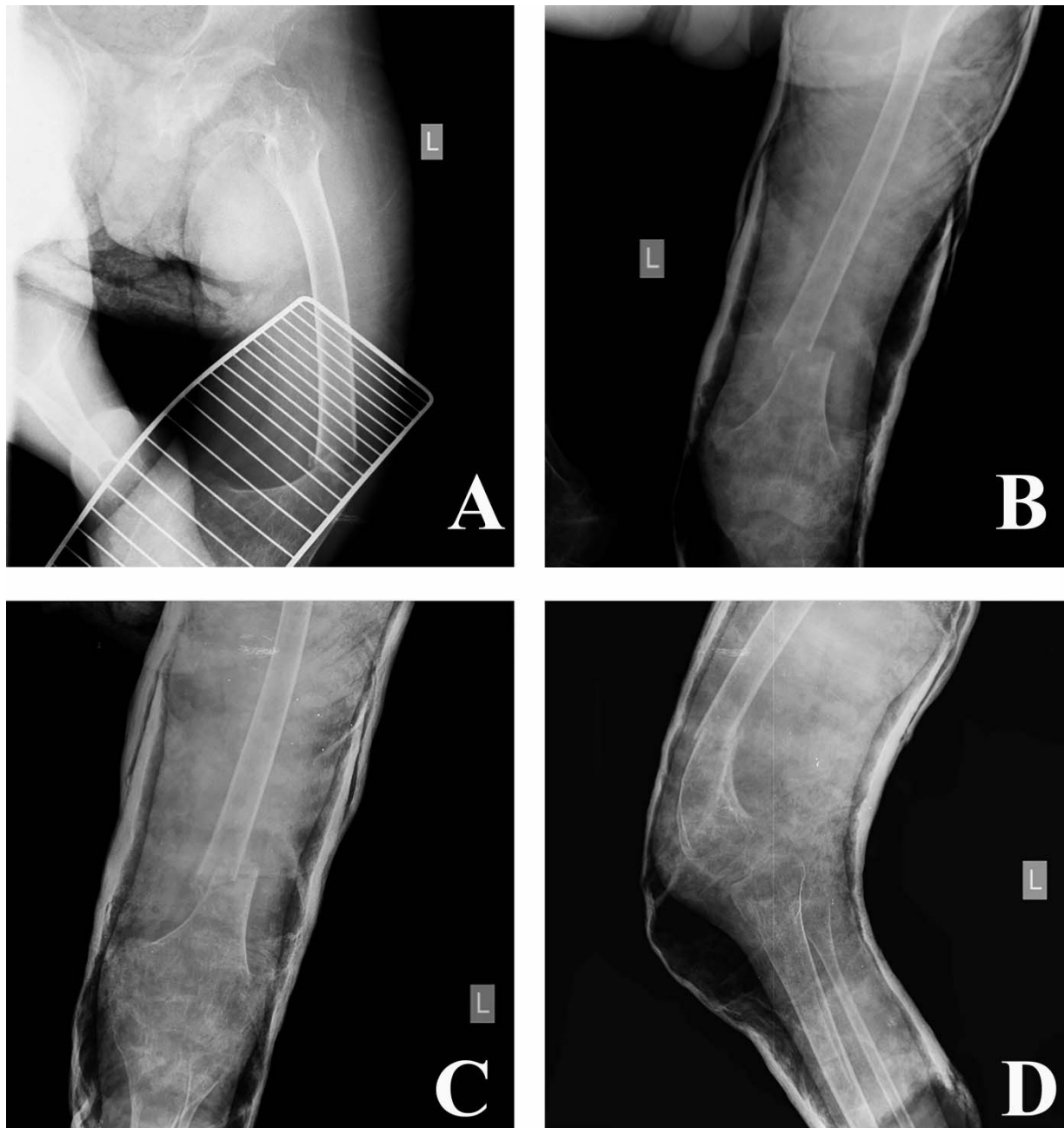


Figure 1. A) X-ray of left upper leg in AP projection upon the arrival – transverse femoral shaft fracture; diffuse osteopenia; B) Control X-ray of left upper leg in AP projection after reduction; C) Control X-ray of left upper leg in AP projection 8 weeks after injury; D) Control X-ray of left upper leg in LL projection 8 weeks after injury

shape in the dorsal area, molar teeth shaped, with a hypoplastic vermis – which is the appearance of Joubert syndrome (Figure 2A, 2B). The electroencephalogram recordings from Neurofax EEG-1200 Diagnostic and Monitoring Platform (Nihon Kohden, Tokyo, Japan) have identified non-specific generalized changes.

Treatment and management

Closed reduction of bone fragments and immobilization with the coxofemoral spica cast were performed in general anesthesia. Control X-ray of left upper leg showed a satisfactory bone fragments position (Figure

1B). Valproic acid and divalproex sodium were introduced in treatment and phenytoin was gradually excluded. After five days, the patient was discharged home with ordered weekly controls. Eight weeks after injury, clinical examination and X-ray imaging of left upper leg in the AP and LL projection showed a satisfactory femoral axis with healing at the site of fracture (Figure 1C, 1D), and immobilization was removed. In follow up period of two years patient made a remarkable neurological improvement. During this period he did not have any epileptic seizures. He was more active, having better mood, sleep and appetite, and he put on 7 kilograms on weight.

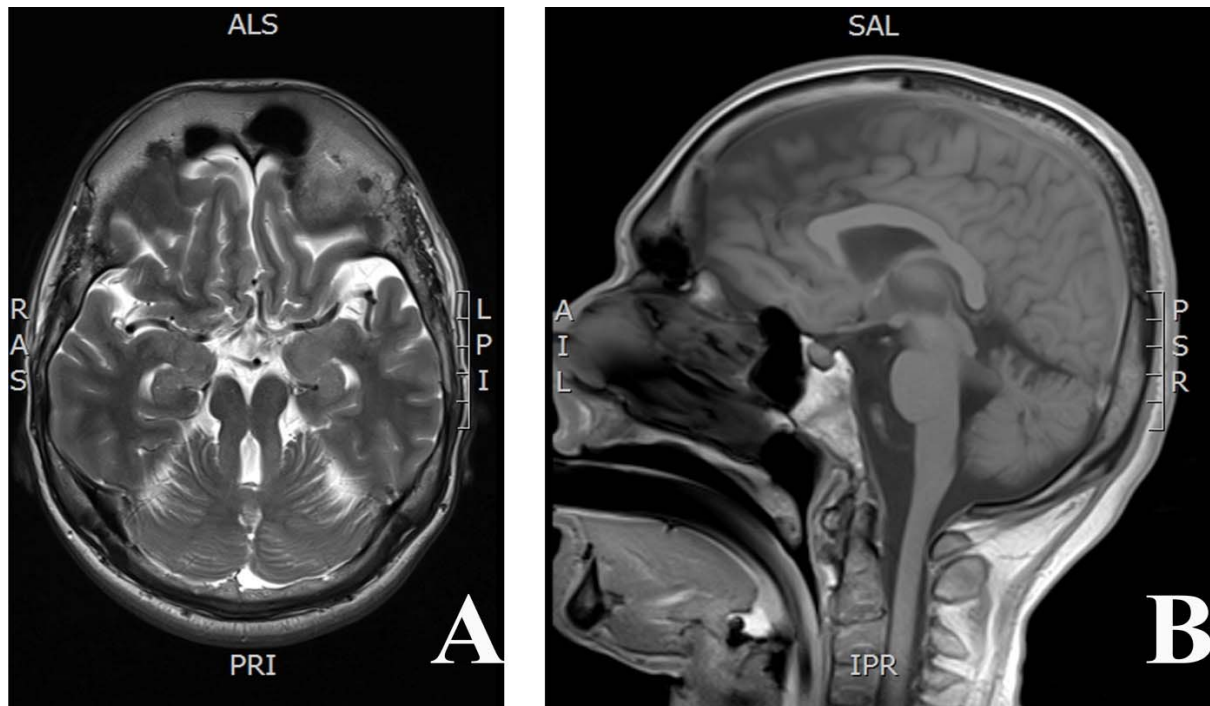


Figure 2. A) MRI T2 weighted image of brain in axial projection – “molar tooth sign” of abnormal midbrain with deep interpeduncular cistern, thickened and elongated superior cerebellar peduncles; abnormal cerebellar folia; B) MRI T1 weighted image of brain in sagittal projection – abnormal 4th ventricle shape; vermian hypoplasia; increased size of pre-pontine cistern

DISCUSSION

Femoral shaft fracture is a very rare repercussion of indirect force acting. Although other authors reported the fractures of the neck and the pertrochanteric area of the femur caused by convulsive seizures, by reviewing available literature, we could not find any case of femoral shaft fracture (Grimaldi et al. 2009, McMillan et al. 2016). In our case, one of the factors that could contribute to the most effective action of the tensile forces in the area of diaphysis is contracture of the adjacent joints (hips, knees). Also, important factors in the substrate were the osteopenia, hypocalcaemia and hypomagnesaemia, which can be found in most fractures of long bones occurred from indirect force acting (Sharma et al. 2015). Osteopenia in our case developed due to disuse of locomotor system as patient was immobile, and secondary to anticonvulsant therapy.

Furthermore, in our case there is interesting approach to injury by the non-operative treatment, which is used when there are strict contraindications for operative treatment (internal/external fixation) (Neumann et al. 2015). Given the general condition of our patient, immobility, bedridden and the presence of moderate sideropenic anemia, we have opted for a conservative treatment. Cast is a very safe method of treatment, with minimal possibility of infection. We estimated that the increased risk of its complications (malunion, malalignment, and stiffness of the adjacent joints) was acceptable for set goals, which are determined for each patient

individually. Because of the limited use of the locomotor system in biomechanics, for disabled patients it is not imperative to achieve the perfect axis of the femur. Given the condition of our patient, the main goals were bone healing and pain management. Generally, when there is no complications, we are practicing immobilization of minimally 6 weeks, up to 12 weeks, depending on the clinical course. The ultimate outcome of our patient is radiologically and functionally very satisfying.

From the neurological side, the aim of treatment was to achieve control of epileptic seizures. Apart from the neurological improvement, this was indicated due to possibility of femoral refracture in the case of new convulsions. We performed extensive neurological diagnostics in exploration for potential pathological substrate associated with the described clinical findings. Based on the MRI of brain and its comparison with the phenotypic and clinical characteristics of the disease we came to the conclusion that the primary diagnosis of cerebral palsy was wrong, and that patient had a rare autosomal recessive disease – Joubert syndrome (prevalence 1: 100,000 - 258,000 of live births), which is in clinical practice often mistaken for cerebral palsy. Establishment of diagnosis is regularly based on clinical features and MRI. Joubert syndrome has a very heterogeneous symptomatology, but is always characterized by three primary findings: a distinctive cerebellar and brain stem malformation called the molar tooth sign, hypotonia and developmental delays (Parisi et al. 2007). Seizures are unusual in Joubert's syndrome and the description of the

MRI report suggests an additional neuronal migration disorder. Our patient had a form of illness with the expression of severe clinical features of syndrome – cranio-facial dysmorphism, cerebellar vermis hypoplasia, severe mental retardation, muscular hypotonia, immobility, bladder and bowel incontinence, scoliosis, oculomotor apraxia, strabismus. The patient had good response on the modified therapy, and no epileptic seizures were observed in the follow up period.

CONCLUSION

The presented case report is interesting for a series of specifics – unreported traumatic manifestation of convulsive seizures, conservative approach to treatment of femoral shaft fracture and detection of rare autosomal recessive disease. This paper once again demonstrated that a high-quality interdisciplinary approach is a pledge for the best outcome of cases with comorbidities from different human body systems. Patients welfare and maximum life quality depend on the complementary work of physicians from various medical disciplines, like in our case it was from - emergency medicine, orthopedics, traumatology and neurology.

Contribution of individual authors:

Jure Pupić-Bakrač contributed to the design of the study, manuscript writing, literature searches and analyses.

Martin Kajić contributed to the manuscript writing.

Marko Ostojić contributed to the manuscript writing, analyses and interpretation of data, stylistic and grammatical revisions to manuscript.

Anita Ivanković contributed to the literature searches, analyses and interpretation of data.

Nataša Pejanović Škobić contributed to the literature searches, analyses and interpretation of data.

Ana Pupić-Bakrač contributed to the manuscript writing, graphic design, stylistic and grammatical revisions to manuscript.

Acknowledgements: None.

Conflict of interest : None to declare.

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