OSTEOGENESIS IMPERFECTA: ABOUT 12 CASES

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Background: Osteogenesis imperfecta (OI), is a rare hereditary disease characterized by bone fragility and low bone mass. The clinical presentation is various with varying severity skeletal signs and inconstant extra-skeletal signs. Type 1 is the most common form (60% of cases).

Objectives: Our objective is to describe the various clinical features observed over a period of 15 years.

Methods: This is a retrospective descriptive study including 12 patients followed for OI, hospitalized in the Rheumatology Department at Fattouma Bourguiba Hospital Monastir TUNISIA between 2006 and 2019. Files were collected and analyzed.

Results: They are 9 boys and 3 girls with an average age of 14.9 ± 8.6 years. Consanguinity was reported in 25% of cases. The reason leading to consultation was a frequent fracture (5/12), blue sclera (16.7%), and bone deformity (8.3%). The number of previous fractures was on average of 5, all of which were caused by a low energy trauma. Similar family cases were noted in 41.6%. The mean age of the first fracture was 4.41 ± 3.2 years. The most frequent fracture sites were respectively: femur (7/12), leg (6/12), tibia (3/12), humerus (4/12), ankle (2/12), and forearm (2/12). A deformity was noted in 58.3% of the cases: lumbar kyphosis (2), exaggerated dorsal kyphosis (2), femurs in parenthesis (2), and an arachnoid deformity of 2 lower limbs (1). Imperfect dentinogenesis was found in 8.3% of cases, while ENT examination revealed conductive and sensorineural hearing loss in 2 patients each. The main radiological abnormalities were diffuse bone demineralization (9 patients), cortical thinning (5 patients), vertebral compression (3 patients), and fracture (2 patients). The bone densitometry showed a mean Z score of 3.49±1.4 in the lumbar spine. The average serum calcium level was 2.38±1.15, alkaline phosphatases were elevated in all cases with an average of 756±624.9. The vitamin D level was deficient in all cases with an average of 22.75±5.3. All patients received in addition to the vitamin-calcium supplementa- tion, pamidronate intravenously at a dose of 9mg/kg/year with a mean number of 6 cures. The main side effects observed during the infusion were abdominal pain, polyarthralgia and asthenia (1 patient), chest pain (1 patient) and fever and chills (1 patient). The control bone densitometry showed a mean Z score of 1.81±1.2 in the lumbar spine.

Conclusion: Despite advances in the OI diagnosis and treatment, more research is needed. Bisphosphonate treatment decreases long-bone fracture rates, but such fractures are still frequent. New antiresorptive and anabolic agents are being investigated but efficacy and safety of these drugs, especially in children, need to be better established before they can be used in clinical practice.

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HIP INVOLVEMENT IN JUVENILE IDIOPATHIC ARTHRITIS

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Background: Juvenile idiopathic arthritis (JIA) is a heterogeneous group of diseases that have in common an onset before the age of 16 years, more than 6-month duration, and the presence of arthritis for at least 6 weeks with no identifiable cause. Hips are the commonly affected joints in severe destructive JIA. Hip disease develops in 20% to 50% of children with JIA.

Objectives: We aimed to analyze the epidemiological, clinical, and radiological aspects of hip involvement in Tunisian JIA.

Methods: A retrospective study including 35 patients was conducted between 2010 and 2019. The patients enrolled met the ILAR criteria for the diagnosis of JIA. Clinical, biological and radiological parameters relating to the hip involvement were collected.

Results: Thirty-five patients were enrolled. The mean age of the disease onset was 9 years [3-15]. The mean age of the patients at the time of the study was 37.8 years [17-69]. The mean duration of the disease was 27 years [2-56]. These patients were assigned to discrete JIA categories: rheumatoid factor positive polyarthritis (43.5%), rheumatoid factor negative polyarthritis (21.7%), enthesitis-related arthritis (17.4%), oligoarthritis (13%) and psoriatic arthritis (4.4%). A biological inflammatory syndrome was noted in 52% of patients. Hip involvement was noted in 43.5% of patients. Coxitis occurred on average 15 years after the JIA onset [4-37]. Thirty four percent of the patients had bilateral hip involvement. The mean Lequesne index was 14. Joint space narrowing and erosions were noted in respectively 68% and 34% of cases. The majority of patients (82.6%) received medical treatment combining nonsteroidal anti-inflammatory drugs (NSAID) and rehabilitation. In the other cases, a total hip replacement was necessary. Coxitis was significantly correlated with rheumatoid factor positive polyarthritis subtype (p=0.02) and with the presence of biological inflammatory syndrome (p=0.03).
CERVICAL SPINE INVOLVEMENT IN PATIENTS WITH JUVENILE IDIOPATHIC ARTHRITIS

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Background: Juvenile idiopathic arthritis (JIA) is the most common chronic rheumatic disease in childhood. Approximately 40-60% of patients with juvenile idiopathic arthritis (JIA) have continuous or recurrent disease activity extending into adulthood.

Methods: A retrospective study including 35 patients was conducted between 2010 and 2019. The patients enrolled met the ILAR criteria for the diagnosis of JIA. Clinical, biological, and radiological data were collected. Patients had a radiological evaluation that included cervical spine x-rays in antero-posterior, lateral and lateral views with flexion.

Results: Thirty-five patients were enrolled. The mean age of the disease onset was 9 years [3-15]. The mean age of the patients at the time of the study was 37.8 years [17-60]. The mean duration of the disease was 27 years [2-56]. These patients were assigned to discrete JIA categories: rheumatoid factor positive polyarthritis (43.5%), rheumatoid factor negative polyarthritis (21.7%), enthesitis-related arthritis (17.4%), oligoarthritis (13%) and psoriatic arthritis (4.4%). Sixteen patients (45%) reported neck pain. Cervical spine involvement occurred on average 7 years [0-13] after the JIA onset. Cervical spine radiographs showed anterior atlantoaxial subluxation (> 5mm) in 8 patients. Magnetic-resonance imaging was performed in 9 patients that had abnormal neurological examination showing a pannus formation of C1-C2 junction (3 cases), a basilar invagination (4 cases) and erosions of the odontoid process (2 cases). A cervical collar has been used for immobilization in patients with significant cervical spine damage. A C1-C2 arthrodesis was proposed to 4 patients.

Conclusion: These findings suggest that the presence of cervical involvement in JIA patients is frequent. Radiologic assessment of cervical spine should be systematically performed for early detection and to prevent its complications.

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Figure 1. Gangrene of left 1st-5th digits and right 4th-5th digit resorption

Figure 2. T2W FLAIR image showing bilateral fronto parietal, occipital, putamen, left insula and subcortical and cortical T2W FLAIR hyperintensity without diffusion restriction suggestive of PRES.