Background: Osteogenesis imperfecta (OI), is a rare hereditary disease charac-
terized by bone fragility and low bone mass. The clinical presentation is varies
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
among 12 patients with OI, hospitalized in the Rheumatology Department at
Fattouma Bourguiba Hospital Monastir, Tunisia.

Methods: This is a retrospective descriptive study including 12 patients followed
for OI, among the 300 patients enrolled in the KAPRI registry from inception in March
2019 to December 2020. 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 bone deformity (6/12), 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 anarchic deformity of 2 lower limbs (1). Imperfect dentogenesis 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 com-
pression (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, recurrent fractures (75%), blue sclera (16.7), and bone deformity (8.3%).

Conclusion: Despite advances in the OI diagnosis and treatment, more research
is needed. Bisphosphonate treatment decreases long-bone fracture rates, but
is needed. Bisphosphonate treatment decreases long-bone fracture rates, but
did not prevent any infections, allergic reactions or any other untoward events as adverse outcomes following
the use of biological therapies. Five patients have attained remission as illus-
trated in the Table 1 below. Two patients have been lost to follow up.

Methods: All patients with an ICD 10 M code diagnosis of Juvenile Arthritis were
selected from the KAPRI registry database. Age, gender, laboratory and clinical
features at diagnosis and treatment options offered were extracted from the data-
base. A further detailed chart review was undertaken to determine the proportion
of patients who achieved remission or minimally active diseases.

Results: Among the 207 patients enrolled thus far, 16 (7.7%) were diagnosed
to have JIA. Majority of the patients were females (75%; n=12) with a mean age of
7 years and 3 months (Range:1 year – 13 years 7 months).

All patients had joint pain and swelling as the initial presenting complaints. Major-
ity of the patients had polyarticular JIA (75%, n=12). The other 4 patients were
oligoarticular (n=2) and systemic JIA (n=2).Among the polyarticular JIA patients
(n=12), only 3 (25%) were rheumatoid factor (RF) positive and 1 was antinuclear
antibody (ANA) positive. The oligoarticular and systemic JIA patients were all
positive for antinuclear antibody, rheumatoid factor and cyclic citrullinated pep-
tide antibodies (anti-ccp). Seven patients (43.8%) required biological therapies;
tocilizumab (n=2), systemic JIA, adalimumab (n=2), polyarticular JIA, etanercept
(n=2), polyarticular JIA and tofacitinib (n=1); polyarticular JIA. One patient with
systemic JIA on tocilizumab developed herpes simplex which was successfully
managed with oral acyclovir. All the other patients did not develop any infections,
allergic reactions or any other untoward events as adverse outcomes following the
use of biological therapies. Five patients have attained remission as illus-
trated in the Table 1 below. Two patients have been lost to follow up.

Conclusion: The economic burden of juvenile idiopathic arth-
ritis-results from the German paediatric rheumatologic database.

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