Results: We observed 52 pts with RD and MAS: 31 (59.6%) with sJIA, 19 (36.5%) – SLE, 1 (1.9%) – juvenile dermatomyositis (JDM), 1 (1.9%) – overlap syndrome (JIA-JDM). Pts with MAS accounted of 26% of all pts with sJIA, 76% of all pts with SLE and 1 (1.9%) of pts at onset of SLE was 2.6 y [15.5; 7.7], at onset of SLE – 11.8 y [8.6; 13.9]. The patient with JDM was 6.5 years old, pts with overlap syndrome – 3.5 years old. Male to female ratio was 1:1.7. A total of 63 episodes of MAS was recorded (41 – in sJIA, 20 – SLE, 1 – JDM, 1 - overlap syndrome). 23 pts (44.2%) had MAS at onset, 26 pts (50%) – “classic” MAS (during the course of disease), 3 pts (5.8%) – recurrent MAS. The clinical manifestations of MAS included fever (90.4%), hepatomegaly (50%), polyarthritis (46.2%), hemorragic rash (32.7%), neurologic involvement (42.3%), lung involvement (34.6%). We found hyperferrinemia in 98%, thrombocytopenia in 78.8%, increased transaminases in 88.5%, hypofibrinogenemia in 40.4%, hypertriglyceridemia in 42.3 % of pts. Most severe course of MAS was established in pts with ferritin levels >1500 ng/ml and with hypertriglyceridemia more than 6.0 mmol/l at an early stage. Bone marrow investigation was performed in 21 pts, and the evidence of haemophagocytosis was confirmed in 8 pts (38%). First features of MAS were fever, sleepiness, lower platelet counts, high TA. Lesions of the skin and mucous were mainly represented by point hemorrhages at an early stage in pts with SLE and MAS, the development of a bright rash with itching was typical for pts with sJIA and MAS. There are no principal differences between course of MAS in sJIA and other RD in children, but mild “subclinical” cases of MAS were observed only in pts with sJIA on biologics. For treatment of MAS all pts were administered high dose of glucocorticoids (per os). Pts with SLE received: cyclophosphamide iv - 4 (26.3%), cyclosporine per os 1 (5.2%), IVIGs - 6 (31.6%), rituximab - 2 pts (10.5%). Pts with sJIA received: cyclosporine per os - 2 (64.5%), IVIGs - 25 (80.6%), 1 etoposide - (3.2%). Patient with overlap syndrome received cyclosporine per os. 8 pts (15.4%) died from MAS (3 with sJIA, 5 – with SLE).

Conclusion: MAS can develop in various children's RD, but more often in pts with sJIA and MAS. There are no principal differences between course of MAS in sJIA and other RD in children, but mild “subclinical” cases of MAS were observed only in pts with sJIA on biologics. For treatment of MAS all pts were administered high dose of glucocorticoids (per os). Pts with SLE received: cyclophosphamide iv - 4 (26.3%), cyclosporine per os 1 (5.2%), IVIGs - 6 (31.6%), rituximab - 2 pts (10.5%). Pts with sJIA received: cyclosporine per os - 2 (64.5%), IVIGs - 25 (80.6%), 1 etoposide - (3.2%). Patient with overlap syndrome received cyclosporine per os. 8 pts (15.4%) died from MAS (3 with sJIA, 5 – with SLE).

Results: Of 103 patients, the pathogenic variant of the MEFV gene was found in 93 pts (90.3%), in 10 pts (9.7%) - the pathogenicity of the revealed variant was contradictory. Of 93 patients with the pathogenic variant of MEFV, the clinical presentation of the disease fits to FMF in 37 patients (39.6%), 11 (29.7%) of them had a mutation in M694V. Out of 37 children who met the criteria for FMF diagnosis, 15 (40.5%) children had a homozgous pathogenic variant of MEFV, and 22 (59.5%) children had two mutations in a heterozgous state. 57 patients who do not have a typical clinical presentation, which is specific for FMF are observed at the departments of rheumatology, cardiology and nephrology. 13 patients are on an outpatient observation, and 6 patients at the time of the study are over 18 years old. 8 (14%) of them had a mutation in M694V. Among 57 patients with pathogenic heterozgous variants in a, 22 patients (38.6%) are observed in the rheumatology department, among them:

- Enthesitis-related arthritis - 2 patients (9%);
- Systemic juvenile arthritis - 13 patients (59%);
- Oligoarthritis - 5 patients (23%);
- Polyarthritids- 2 patients (9%).

Conclusion: Analysis of the obtained data showed that FMF is characterized by a combination of the clinical presentation and the pathogenic variant in the MEFV gene. However, the disease manifests itself not only in the homozgous pathogenic variant, but also in the combination of two mutations in heterozogous. The presence of one heterozygous mutation, generally, does not lead to the development of disease.

REFERENCES:

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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 I 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 pain and osteoporosis (12%), blue sclera (6.7%), hearing loss (1.7%), 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 vertebra (7/12), femur (6/12), tibia (3/12), femurs (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 anacardic 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 10±3.8 ng/mL. The average Lequesne index was 14. Joint space narrowing and erosions were noted with a predilection to affect more girls and boys. Over a period of 2 years, remission has been attained among 31.25% of the patients (5 of 16) with use of synthetic disease modifying anti-rheumatic drugs and biological therapies.

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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%), enthesis-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).