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, 7.6% of all pts with SLE. The mean age of pts at onset of sJIA was 2.6 y [1.5; 5.75], at onset of SLE – 11.8 y [8.6; 13.95]. 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%), pericarditis (46.2%), hemorrhagic rash (32.7%), neurologic involvement (42.3%), lung involvement (34.6%). We found hyperferritinemia 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+iv). Pts with SLE received: cyclophosphamide iv - 5 (26.3%), cyclosporine per os 1 (5.2%), IVIGs - 6 (31.6%), rituximab - 2 pts (10.5%). Pts with sJIA received: cyclosporine per os - 20 (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. In our observation more pts had MAS during the course of disease, not at onset. Rapid recognition of MAS can improve treatment outcomes and prognosis.

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AB0726

BIOLOGICS IN CHILDREN WITH IDIOPATHIC INFLAMMATORY MYOPATHIES: AN EXPERIENCE OF SINGLE CENTER

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Background: Juvenile dermatomyositis (JDM) is the most common idiopathic inflammatory myopathy (IIM) of childhood, but the involvement of muscle also can be complication of systemic lupus erythematosus (SLE), mixed connective tissue disease (MCTD) and different overlap syndromes. The presence of myopathy can significantly affect the prognosis of the disease and the quality of life. Despite of the absence of official indication for biological therapy for IIM, it's may be needed in severe course of disease.

Objectives: To analyze in retrospective study the efficacy and safety of Biologics (B) in patients (pts) with IIM in our pediatric rheumatology center.

Methods: The study included all pts with rheumatic diseases (RD), who had IIM and received B during past 10 years.

Results: A total of 17 pts (10 females) were identified: 23.5% with JDM, 11.8% - SLE, 35.3% - MCTD, 29.4% - overlap syndromes. The median age at the onset of RD was 9.75 years [interquartile range (IQR) 6.9; 13.5]. The median disease duration at the onset of myopathy was 7.0 months [3.0; 10.0], in all pts with JDM – at onset. All pts had myopathic syndrome. The rash typical for JDM was in 8 pts (47%). The lung involvement observed in 6 pts (35.3%). Levels of serum muscle enzymes (CK, ALT, AST and lactate dehydrogenase [LDH]) were elevated in 88.2% of pts: CK in 52.9% (mean ± std 624.35±1007.2 U/L), ALT – 82.4% (mean ± std 142.18±148.16 U/L), AST – 82.4% (mean ± std 159.6±201.1 U/L), LDH – 88.2% (mean ± std 492.5±229.2 U/L). Abnormal EMG findings were in 10 pts (59.4%). Nailfold capillaroscopic changes typical for IIM had 10 pts (59.4%). Before B 94.1% of pts received corticosteroids (CS), 58.8% - methotrexate, 29.4% - hydroxychloroquine, 17.6% - cyclophosphamide, 11.8% - cyclosporine, 5.9% - mycophenolate mofetil, 5.9% - azathioprine, 47% - IVIG. B therapy was started due to insufficient efficacy of previous therapy. The median age at start of B was 12.3 years [IQR 9.5; 15.0]. The median disease duration prior to treatment with B was 2.25 years [IQR 0.8; 3.6]. 58.8% of pts received rituximab (RTX), 41.2% - abatacept (ABA). The median time between each course of RTX was 182 days [IQR 156–315]. 80% of pts underwent more than one course of RTX therapy, with a maximum of 5 courses. The median duration of ABA therapy was

2.8 years [IQR 1.6; 5.0]. Discontinuation of B due to serious AE was observed in 1 patient with overlap syndrome (5.9%) – macrophage activation syndrome (MAS) 8 day after RTX infusion (5th course, 1st infusion). One patient (female, 12.3 yo) died from MAS (at onset of SLE, developed before B), the others achieved remission. B therapy allowed to reduce the dose of CS to maintenance in all pts, decrease of calcinosis in 2 pts and improve the quality of life.

Conclusion: Our study demonstrated that B is highly effective in children with IIM and have the acceptable safety. Early diagnosis and initiation of intensive therapy are important in reducing symptoms of myopathy in RD. It seems that the development of IIM in other RDs, not JDM, indicates the need for B and its good efficacy. The further extended studies are needed.

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AB0727

STUDY OF MEFV GENE MUTATIONS IN A COHORT OF CHILDREN: A SINGLE CENTER

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Background: Familial Mediterranean fever (FMF) is a monogenic autoinflammatory hereditary disease characterized by recurrent episodes of fever with sterile peritonitis, pleural inflammation, arthritis, and/or erysipelas-like rash. Among all variants of the MEFV gene, according to the literature, five pathogenic ones have been identified, which in 75% of cases lead to the development of a typical clinical presentation: V726A, M694V, M694I, M680I, and E148Q. Among them, the M694V variant is the most common and occurs in patients with FMF in 20-65% of cases. At the same time, approximately 10 to 20% of patients meeting the diagnostic criteria for FMF do not have pathogenic variants in the MEFV gene. Despite the fact that the molecular genetic, pathogenetic and clinical features of the disease have been studied detailed, the diagnosis remains difficult due to the lack of a clear correlation between the patient's clinical and genetic data.

Objectives: To analyze the obtained genetic data of patients with pathogenic variants in the MEFV gene.

Methods: The study included 103 patients who are mainly observed at the rheumatology department of the National Medical Research Center of Children's Health of Ministry of Health of the Russian Federation in Moscow. All patients underwent analysis of the MEFV gene using Sanger sequencing with further statistical processing of the data obtained.

Results: Of 103 patients, the pathogenic variant of the MEFV gene was found in 93 patients (90.3%), in 10 patients (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 homozygous pathogenic variant of MEFV, and 22 (59.5%) children had two mutations in a heterozygous 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 heterozygous 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%);
- Polyarthritis - 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 homozygous pathogenic variant, but also in the combination of two mutations in heterozygous. The presence of one heterozygous mutation, generally, does not lead to the development of FMF.

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AB0728 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, recurrent fractures (75%), 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 anarctic 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 supplementation, 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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AB0729 CLINICO-EPIDEMIOLOGICAL PROFILE OF JUVENILE IDIOPATHIC ARTHRITIS (JIA) PATIENTS IN KENYA; DATA FROM THE KENYA PEDIATRIC RHEUMATOLOGY (KAPRI) REGISTRY

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Background: Pediatric rheumatic diseases are chronic illnesses that impart a significant disease burden upon societies (1-3). Determination of the burden and clinical characteristics of these diseases is a critical first step to improving access to care and optimizing use of existing health systems for the well-being of these patients (4-6). A pediatric rheumatology registry is critical in defining the spectrum, clinical characteristics, outcomes and responses of various interventions for pediatric rheumatic diseases. Given that none exists in Kenya, the Kenya Pediatric Rheumatology Registry (KAPRI) registry offers a platform to generate this much needed data in sub-Saharan Africa.

Objectives: Our objective was to determine the baseline patient characteristics, clinical features and outcomes of the Juvenile Idiopathic Arthritis (JIA) patients assessed at the Aga Khan University Medical College East Africa who were enrolled into the KAPRI registry from inception in March 2019 to December 2020.

Methods: All patients with an ICD 10M 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 database. 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. Majority 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 negative for antinuclear antibody, rheumatoid factor and cyclic citrullinated peptide 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 illustrated in the Table 1 below. Two patients have been lost to follow up.

Conclusion: Seronegative polyarticular JIA was the predominant form of JIA observed 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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AB0730 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 ($p=0.03$).