Demographic, clinical, neurological features and genetic mutations did not differ in surviving and deceased DADA2 patients. Deceased and surviving subjects differed in terms of medication usage after the diagnosis of DADA2. Anti-TNF alpha treatment has been initiated in 5 surviving patients soon after the diagnosis of DADA2. However, unfortunately three patients who have died, were able to use either a few doses of anti-TNF alpha treatment or none; one patient due to reluctance of patient and two patients due to establishment of a definite diagnosis with genetic analysis after the patients had died.

Conclusions: Although this study includes limited number of patient, to our knowledge this study for the first time compares the phenotypic, genotypic and medication differences between surviving and deceased DADA2 patients. Anti-TNF alpha treatment seems to be very efficient and lifesaving in DADA2 patients.

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AB0995 CLINICAL, THERAPEUTIC, AND GENETIC ANALYSES IN A PATIENT WITH PAPA SYNDROME COMPLICATED WITH INFLAMMATORY BOWEL DISEASE

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Background: PAPA syndrome is an autoinflammatory disease linked to mutations in the *PSTPIP1* gene [1]. These mutations produce a hyper-phosphorylated PSTPIP1 protein and alter its participation in the activation of the "inflammasome" [2]. PAPA syndrome is characterized by pyogenic arthritis with pyoderma gangrenosum and acne, and ususally treated with corticosteroids. Reports from the literature suggest that patients with poorly controlled PAPA syndrome may benefit from IL-1 blockade [2]; however, we cannot use these biologics for PAPA syndrome in our country.

Objectives: To elucidate the pathogenesis of PAPA syndrome, we examined the clinical status and complications before and after treatment and also analyzed the *PSTPIP1* gene.

Methods: We herein report a 25-year-old Japanese male who suffered from recurrent arthritis in his knee and ankle joints, pyoderma gangrenosum, and acne. He had experienced melena and multiple colonic ulcers had been detected by colonfiberscopy. His ulcerations resembled ulcers associated with Crohn's disease. A histological examination was then performed for the synovium of this knee joints, skin lesions of pyoderma gangrenosum, and the colon. The genomic DNA of *PSTPIP1* were analysed in both the patient and his family. We also examined the serum level of IL-1, IL-6, and TNF-α before and after treatment of biologics (adalimumab and infliximab) in this patient.

Results: 1) A histological analysis revealed that a large number of neutrophils had accumulated in the skin lesions; however, very few neutrophils were detected in the pathological lesions of the knee joints and colon. 2) According to a gene analysis, we detected a novel heterozygous mutation (E101G) in the *PSTPIP1* gene; however, his healthy father also had the same mutation, thus suggesting that this mutation of *PSTPIP1* might not be related to his phenotype. We are searching for other affected genes besides the *PSTPIP1* gene for PAPA syndrome in this case. 3) After treatment of biologics (infliximab), the clinical symptoms, such as arthritis and multiple colonic ulcers were considerably improved and the serum level of IL-6 and TNF-α were decreased in this patient.

Conclusions: We herein reported a Japanese PAPA syndrome patient who was complicated with inflammatory bowel disease and had a good response to biologics. A genetic analysis suggested that this particular phenotype might not have been affected by a mutation of the *PSTPIP1* gene.

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AB0996 FREQUENCY OF HLA-B*51 AND THE ASSOCIATION WITH CLINICAL MANIFESTATIONS IN ARMENIAN PATIENTS WITH BEHÇET DISEASE

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Background: Behçet disease (BD) is a system inflammatory disease mostly characterized by oral and genital ulcers and variable manifestations affecting other organs, mainly skin and eye. The HLA class I molecule HLA-B*51 allele is strongly associated with BD in many different ethnic groups and appears to be a significant risk factor for BD in the ancient Silk Route areas in contrast with Western and Northern Europe and U.S.[1–4]

Objectives: Our purpose was to show the frequency of presence of HLA-B*51 allele in Armenian patients with BD and estimate any correlation with common symptoms of disease depended on presence of HLA-B*51.

Methods: Forty-seven patients of Armenian origin (28 males 59.6% and 19 females 40.4%) fulfilling the International Criteria for BD (ICBD) with mean age 29.7±11.3 years; disease duration – 5.3±12.4 years were enrolled. We observed the clinical manifestations of BD of both HLA-B*51 carriers and non-carriers.

Results: HLA-B*51 was detected in 38 (80.8%) patients, of whom 25 male and 13 female (65.8% and 34.2%, respectively). Arthritis, erythema nodosum and genital ulcers were significantly more common in HLA-B*51-positive patients (83.3%, 83.3% and 80.6% respectively) than in HLA-B*51-negative ones (16.7%, p=0.032, 16.7% p=0.032 and 19.4% p=0.023 respectively).

Conclusions: In this ever first estimation of HLA-B*51 gene frequency in patients with BD in Armenia, our results indicated that the frequency of HLA-B*51 allele in BD is 80.8% which is higher than elsewhere (in Japan 58.9%, Iran 61.9%, Turkey 75%, Saudi Arabia 76.9% Greece 78.9%)[5]. Furthermore, this investigation revealed HLA-B*51 carriage much more often in male than in female BD patients. In addition HLA-B*51 carriers are in significant higher risk of development of arthritis, erythema nodosum and genital ulcers.

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Diagnostics and imaging procedures

AB0997 ULTRASONOGRAPHIC EVALUATION OF ANTERIOR KNEE PAIN IN YOUNG ADULTS

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Objectives: The Primary outcome of the present work is to detect type and frequency of Ultrasonographic changes in patients with anterior knee pain. Secondary outcome is to determine the role of MSUS in early detection of causes of anterior knee pain (AKP).

Methods: Sixty patients with AKP and 20 sex and age matched healthy control were included in this study obtained from the outpatient of Physical Medicine and Rheumatology Department of El Hussein and Bab El-Sharia university hospitals. Inclusion criteria: age between 20–40. Exclusion criteria: Trauma, Crystal induced arthritis, connective tissue diseases (RA, SLE, SpA), chronic liver or kidney disease and previous surgery. Full medical history and clinical examination as well as WOMAC, knee Conventional Radiography and Musculoskeletal Ultrasonography (MSUS) were done to all patients. Bilateral MSUS examination of the following common sites: Insertion of quadriceps tendon, Ligamentum patellae and Anterior aspect of the knee from medial to lateral. According to EULAR guide lines for musculoskeletal ultrasonography.

Results: The mean duration of knee pain in patients group was 11.42±9.134 months. ESR, CRP and WOMAC were higher in patients group ($p<0.01$). MSUS showed significantly more frequent Supra-patellar and Infra-patellar bursitis in patients than control group (p=0.000). patients had higher frequency of cartilage clarity loss (p=0.039) as well as lower cartilage thickness than control, 2.28±0.258 vs 2.73±0.261 mm respectively (P=0.000, 95% CI: 0.31 to 0.58)

Conclusions: The degree of Knee pain, measured by WOMAC, is positively correlated with inflammatory biomarkers (ESR and CRP) in patients with Anterior knee pain. MSUS revealed a positive correlation between Femoral Articular Cartilage thickness and the periarticular tendon- thickness, namely Patellar origin, patellar insertion and Quadriceps insertion. Interestingly, our data present an evidence of the negative impact of the duration of knee pain over Femoral Articular Cartilage thickness as well as Patellar origin thickness. (FAC thickness get worse with longer duration of pain)