## AB1089 CLINICAL FEATURES OF FAMILIAL MEDITERRANEAN FEVER PATIENTS IN NORTH-WESTERN PART OF TURKEY: ANALYSIS OF 139 PATIENTS

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**Background:** Familial Mediterranean fever (FMF) is known the most common monogenic autoinflammatory disease. Its prevalence is reported high from the eastern Mediterranean areas (1) The disease is characterized by episodes of fever, serositis, arthritis,renal complications and other different clinical manifestations (2)
**Objectives:** Here, we aimed to present our data of our 139 FMF patients for demonstrating the demographic and clinical features of the study group,from North-western part of Turkey.
**Methods:** A total of 139 FMF patients who were diagnosed and treated in the department of Internal medicine/Rheumatology, Sakarya University (North-western area of Turkey) were included in the study and the demographic and clinical characteristics of the patients were examined.
**Results:** The mean age of the patients was 39.02 ± 11.3. Male gender was 42 (30.2%) and female gender was 97 (69.8%).107 (77%) of patients had fever and 32 (23%) had no history of fever. 127 (91.4%) patients complained about peritonitis, 27 (%19.4) patients had pleuritic pain, 19 (13.7%) patients had erysipelas like erythema and 53 (38.1%) patients had arthritis attack. 34 (24.5%) patients also had sacroiliitis. The ratio of resistance of treatment response to colchicine drugs that can be available in Turkey (Colchicum dispert®), 6 (4.3%) was determined. Interestingly these patients responded to the colchicine drugs available from some other countries from Europe (as Colchicine-opocalcium® and Colchicina-lirca®) None of our patients needed anti-IL1 therapies. The rate of amyloidosis was 5 (3.6%).
**Conclusion:** FMF is a disease with high morbidity and mortality, 95.7% of the patients in our region have response to colchicine drugs which is available in our country. The remaining patients have also response to colchicine available from some other countries. None of our patients had anti-IL1 therapies.

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## AB1090 CECR1/ADA2 MUTATION IN A BRAZILIAN FAMILY

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**Background:** Deficiency of adenosine deaminase 2 (DADA2) is a recently identified disease caused by mutations in the CECR1/ADA2 gene, encoding for adenosine deaminase 2 protein. Clinical presentation is variable and includes early-onset polyarteritis nodosa, hemorrhagic and ischemic strokes, hypogammaglobulinemia and cytopenia
**Objectives:** To present the clinical cases of two Brazilian siblings with early stroke episodes carrying a homozygous CECR1/ADA2 mutation.
**Methods:** Chart review of clinical data, laboratory tests and mutation analysis
**Results:** The index case is a 7-year-old boy who presented to the emergency unit with right lower limb weakness, rhyme deviation and palpebral ptosis, associated with recurrent and intermittent fever, mood change and hypertension. The physical exam revealed drowsiness, lateral and vertical ocular paresis, diplopia, facial palsy, and bilateral ataxia. The brain MRI showed acute left mesencephalic small vessel lacunar stroke, previous right mesencephalic subacute stroke, and cerebellar cavity related with anterior cerebellar artery segmental narrowing. Laboratory tests revealed increased inflammatory markers and anemia. Autoantibodies and viral screening were negative. Renal ultrasound showed a pattern of low resistance in the intrarenal arteries bilaterally. At this time, he was diagnosed as polyarteritis nodosa (PAN). Despite of adequate treatment (cyclosphosphamide and corticosteroids), two new stroke episode occurred at left head of caudate and right thalamus. His brother, a 9-year-old healthy boy at that time, had a previous history of ischemic stroke when he was 4 years old, after receiving a vaccine, with complete recover. Considering this family history and the fact that parents are consanguineous, mutation analysis of CECR1/ADA2 gene was performed and showed homozygosity for the p.Y453C mutation in exon 9 in both siblings and heterozygosity for the same mutation in both parents. Although the eldest boy remained asymptomatic for 5 years without any specific treatment, shortly after the identification of the gene mutation, he presented recurrent fever episodes, myalgia, livedo reticularis and increased inflammatory markers. Anti-TNF treatment was initiated for both patients with good disease control.
**Conclusion:** DADA2 should be suspected in patients with PAN-like phenotype and history suggestive of an inherited disease (eg.: affected siblings and consanguineous parents) or resistance to conventional treatment.

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## AB1091 THE RELATIONSHIP BETWEEN NAILFOLD CAPILLAROSCOPY FINDINGS IN BEHÇET'S PATIENTS AND COURSE OF THE DISEASE

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**Background:** Behçet's disease is a chronic, recurrent and systemic vasculitis that may affect veins and arteries at all diameters. Small vessel involvement is responsible for most of its pathological signs.
**Objectives:** We aimed to compare the nailfold capillaroscopy findings of patients with Behçet's disease to a healthy control group and examine the relationships, as well as revealing the relationships with the sub-type, activity and other characteristics of Behçet's disease.
**Methods:** We conducted a cross-sectional analysis of 153 patients with Behçet's disease and 165 healthy volunteers in a single center. The capillaroscopic findings of the 2nd-5th fingers of both hands of the participants in the Behçet's patients and control groups were included in the analysis. Capillaroscopic findings were evaluated by two different experts who were experienced in this field by using the scoring at Atlas of Capillaroscopy in Rheumatic diseases by Maurizio Cutolo (1).
**Results:** There was no statistically significant difference between the two groups in terms of age or sex (respectively p=0.189 and p=0.585). There was no difference between the Behçet's patients and healthy volunteers in the qualitative analysis on capillary density, capillary visibility, aneurism, capillary tortuosity, capillary enlargement and presence of avascular areas (p values respectively: 0.610, 0.147, 0.481, 0.057, 0.514 and 0.110). In the Behçet's patients, bushy capillaries (24.2%, 37/153), capillary dilatation (32%, 49/153) and microhemorrhage (39.2%, (60/153) rates were significantly higher than those in the healthy control group (p<0.001). In the quantitative analysis, total capillaroscopy score was significantly higher in the Behçet's patients than those in the healthy control group (p<0.001) (Table 1). No statistically significant relationship was found between the presence of clinical signs and capillaroscopy scores, except for erythema nodosum.
Conclusion: the Behçet's patients had significantly higher total capillaroscopy scores in comparison to those in the healthy control group. Based on these data, we believe that the capillaroscopic changes found in Behçet's patients, though unspecific, may support clinical diagnosis in uncertain cases where Behçet's disease is considered as a probability. There is a need for well-planned prospective studies to support this our thought.