Background: In the course of familial Mediterranean fever (FMF), the frequency of other inflammatory diseases increases compared to the general population. Multiple sclerosis (MS) or demyelinating diseases (DD) of central nervous system (CNS) are also more common in FMF patients than in the general population.

Objectives: In this study, we would like to report 5 cases with MS/DD accompanied by FMF or MEFV mutations in two families.

Methods: 4 patients with FMF and 1 patient with MEFV mutation were included in this study. The patients with FMF were diagnosed according to Tell-Hashomer clinical criteria for FMF. The diagnosis of MS was made according to McDonald criteria.

Results: The clinical features of the patients were shown in Table 1.

Conclusion: FMF and MS/DD are characterized by repetitive attacks. Familial association can be seen in 12% of patients with MS (1). This is related to both HLA and non-HLA-related genetic tendency. The probability of developing MS increased 4 times in FMF patients (2). This seems to be related to the presence of MEFV gene creating a pro-inflammatory background. In such family samples, combining HLA and non-HLA gene related studies with MEFV gene analysis will be useful in common genetic factors investigation.

References:

Acknowledgments: None

Disclosure of Interests: None declared. DOI: 10.1136/annrheumdis-2020-eular.6273

AB1041
BIOLIGICS IN ADULT’S ONSET STILL’S DISEASE: TREATMENT STRATEGIES AND SAFETY IN SINGLE CENTER COHORT WITH LONG-TERM FOLLOW-UP
N. Koukouas1, N. Avgoustidis1, S. Pitsigavdaki1, K. Pateromichelaki1, A. Repa1, A. Molla Ismail Sali1, A. Eskinizis1, G. Bertisis1, University Hospital of Heraklion, Rheumatology, Clinical Immunology and Allergy, Heraklion, Greece

Background: Adult-onset Still’s disease (AOSD) is a rare systemic inflammatory disorder. In recent years biological disease modifying antirheumatic drugs (bDMARDs) are becoming increasingly important for its treatment.

Objectives: To evaluate disease outcomes, treatment strategies and their long-term safety in a cohort of AOSD patients treated with bDMARDs.

Methods: A single-center retrospective study of patients diagnosed with AOSD until 2019 was conducted. Patients were included if they: a) were 16 years old or older, b) met the Yamaguchi criteria and c) had received a bDMARD. Demographics, clinical and laboratory parameters were collected at the time of diagnosis. Data regarding treatment lines included: the previous and concomitant conven-
tional disease modifying antirheumatic drugs (cDMARDs), the type of initial bDMARD, switches, and corticosteroids discontinuation. Adverse events related to treatment and disease outcomes including death and amyloidosis were also recorded.

Results: Sixteen patients with AOSD (Table 1) refractory to cDMARDs were administered biologics. The median duration of follow-up was 14 years (range 1-24). Consistent with recent literature, two distinct disease patterns were recognized: the systemic form (SF) and the chronic articular form (CAF). In the SF the leading clinical symptoms were fever, pericarditis and pleuritis. In CAF the leading clinical symptom was persistent RA-like arthritis.

Table 1. Summary of patient characteristics at the time of diagnosis

<table>
<thead>
<tr>
<th>Characteristics</th>
<th>Results</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age at the time of diagnosis median, (range) years</td>
<td>32.5 (18-64)</td>
</tr>
<tr>
<td>Sex (N)</td>
<td>11 female, 5 male</td>
</tr>
<tr>
<td>Fever</td>
<td>14 (87%)</td>
</tr>
<tr>
<td>Rash</td>
<td>8 (50%)</td>
</tr>
<tr>
<td>Lymphadenopathy</td>
<td>2 (12.5%)</td>
</tr>
<tr>
<td>Arthritis</td>
<td>15 (93.75%)</td>
</tr>
<tr>
<td>Pleuritis</td>
<td>7 (43.7%)</td>
</tr>
<tr>
<td>Pericarditis</td>
<td>9 (56.25%)</td>
</tr>
<tr>
<td>Hepatopeliosismegaly</td>
<td>2 (12.5%)</td>
</tr>
<tr>
<td>Elevated liver enzymes</td>
<td>13 (75%)</td>
</tr>
<tr>
<td>Hyperferritinaemia</td>
<td>4 (25%)</td>
</tr>
</tbody>
</table>

Patients with the SF were treated with anakinra (n=4), tocilizumab (TCZ; n=3), canakinumab (n=1) and anti-TNFα (1 adalimumab, 1 etanercept) (n=2). Patients with the CAF received anti-TNFα (3 infliximab, 1 etanercept) (n=4) and TCZ (n=2). The median time from biologic initiation to corticosteroids discontinuation was 6.5 months, (range 2-32), (Table 2), 9 patients (56.25%) remained on treatment with the initial bDMARD, 4 patients (25%) received treatment with two and 3 patients (17.5%) with ≥ 3 bDMARDs. All patients with the CAF were on bDMARD at the end of follow-up, while 4/10 patients (40%) with the SF discontinued it. During follow-up only one serious adverse event was attributed to bDMARD (allergic reaction to infliximab infusion). There were no cases of amyloidosis or deaths.

Conclusion: Dichotomous phenotype in AOSD can determine treatment strategy for initial biologic treatment. Inhibition of IL-1 and IL-6 was the preferred therapeutic option for systemic form while inhibition of TNF and IL-6 was the preferred option for the chronic articular form. All of the above bDMARDs have favorable long-term safety profile in patients with AOSD.


Disclosure of Interests: Nikolaos Koukouas: None declared, Nestor Avgoustidis: None declared, Sofia Pitsigavdaki: None declared, Katerina Pateromichelaki: None declared, ARGYRO REPA: None declared, Ainour Molla Ismail Sali: None declared, Anastasios Eskinizis: None declared, George Bertisis Grant/research support from: GSK, Consultant of: Novartis.

DOI: 10.1136/annrheumdis-2020-eular.4120

AB1042
ASSESSMENT OF BONE MINERAL DENSITY AND FREQUENCY FRACTURES PERIPHERAL SKELETON BONES IN PATIENTS WITH ALKAPTONURIA
A. Kuzin1,2, A. Smirnov2, E. Zaytseva1, K. Kudinskiy2, L. Blank2, A. Ginzburg1, Y. Anisimov2, A. Shchukin2, L. Ryzhkov2, E. Dolzhnenkova3, N. Poltiova2, 1Russian Medical Academy of Continuous Professional Education, Moscow, Russian Federation; 2V.A. Nasonova Research Institute of Rheumatology, Moscow, Russian Federation; 3Ryazan Regional Clinical Hospital, Ryazan, Russian Federation; 4Kirov State Medical University, Kirov, Russian Federation

Background: To assess the bone mineral density (BMD) of the skeleton using the Hologic Discovery A DXA, determine the frequency of low-energy skeletal bone fractures among adult patients with alkaptonuria (AKU), and identibobjective fac-
tors that affect the occurrence of fractures.

Methods: AKU is a rare genetic disease (1 case per 250,000) which occur to severe damage to the spine and large joints. Serious problem in this category of patients is a decrease in BMD. The study included 40 patients with a reliable diagnosis of AKU (23 men and 17 women) aged from 33 to 78 years (mean 60.32±9.1). Densitometry of the lumbar spine was performed in 40 patients; of the forearm bones in 34 patients; of the proximal femur in 32 patients (of the proximal femur in 32 patients (8 patients were not examined due to bilateral hip joint replacement).

Results: Normal values of spinal BMD were found in 26 patients (65%), osteopoenia – in 12 (30%) and osteoporosis – in 2 (5%) patients. In the proximal parts of the femur, osteoporosis was detected in 12 patients (30%), osteopenia in 13 (32.5%), and normal in 7 (17.5%) patients. In the bones of the forearm,