remission after 12 months of treatment. Interestingly, about half of patients did not need to be treated with a biologic to reach predefined T2T (2). Thus, the early treatment escalation seems advantageous indicating a window of opportunity to successfully treat polyarticular JIA.

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COMPARISON OF THE CLINICAL DIAGNOSTIC CRITERIA AND RESULTS OF THE NEXT GENERATION SEQUENCE GENE PANEL IN PATIENTS WITH PERIODIC FEVER

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Background: Autoinflammatory diseases (AID) are a group of hereditary diseases characterised by inflammation periods accompanied with clinical findings such as fever, skin rash, lymphadenopathy, abdominal pain, musculoskeletal symptoms, and with sign of inflammation in the blood. Each disease has own typical clinical findings and they are associated with mutations in specific genes such as in MEFV gene in familial Mediterranean fever (FMF), MVK gene in mevalonate kinase deficiency (MKD), TNFRSF1A gene in TRAPS and NLRP3 gene in cryopyrin associated periodic fever syndrome (CAPS). There are also patients exhibit the incomplete phenotype of a disease or overlap signs of more than one AID. The diagnosis of these patients have difficult and may not be possible by a single target gene analysis. Screening of the periodic fever syndrome (PFS) genetic panel including various AID genes may be beneficial to define the atypical cases.

Objectives: The aim of this study was to compare the phenotypic diagnoses to the genotypic results obtained from the PFS genetic panel, which studied in patients with phenotypic findings of AIDs except the FMF, such as MKD, TRAPS and CAPS.

Methods: This is a prospective study and conducted between June 2016 and December 2018. Patients who met "The Eurofever clinical diagnostic/classification criteria" for MKD, TRAPS and CAPS were included in the study. Next-generation sequencing (NGS) analysis was performed including 8 genes (MEFV, MVK, NLRP3, NLRP12, TNFRSF1A, TNFRSF11A, LPIN2 and PSTPIP1) in 37 patients with phenotypic findings of MKD, TRAPS or CAPS. The patients phenotypic preliminary diagnoses and genotypic results were compared.

Results: Thirty seven patient included in the study who met the clinical diagnostic criteria for MKD, TRAPS or CAPS. As a result of clinical signs; 19 patients were diagnosed with MKD, 10 were TRAPS and 8 were CAPS.

In the PFS genetic screening panel of 19 patients with phenotypic diagnosis of MKD; disease causing mutation was found in 8 patients in MVK gene, in 3 patients in MEFV gene, in 2 patients in NLRP12 gene and in 1 patient in TNFRSF1A gene. No pathogenic mutation was shown in 3 patients and genetic variants of uncertain significance (VUS) in different genes were shown in two patient.

In the result of PFS panel of 10 patients with phenotypic diagnosis of TRAPS; disease causing mutation was found in 5 patients in MEFV gene and in 2 patients in TRAPS gene. No pathogenic mutation was identified in 3 patients.

In 8 patients who were diagnosed phenotypically as CAPS; disease causing mutation was found in 3 patients in NLRP3 gene, in 2 patients in NLRP12 and in one patient in MEFV. No mutation was detected in remaining two patient.

The final diagnosis was made by both phenotypic and genotypic data. In 37 patients (phenotypically 19 MKD, 10 TRAPS, 8 CAPS), respectively; 8 had MKD, 9 had FMF, 3 had CAPS, 3 had TRAPS, 4 had FCAS2 and 10 had undifferentiated PFS.

Conclusion: In our study, we have shown that clinical diagnostic criteria may not always be sufficient to establish the correct diagnosis. In case of clinical suspicion or in the presence of more than one autoinflammatory disease findings, NGS analysis may help to determined the diagnosis.

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QUALITY OF LIFE IN JUVENILE IDIOPATHIC ARTHRITIS-ASSOCIATED UVEITIS: DO WE NEED A NEW ASSESSMENT QUESTIONNAIRE?

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Background: Uveitis, the most common extra-articular manifestation in juvenile idiopathic arthritis (JIA), occurs in 10-20% of patients. Although prognosis of uveitis within JIA (JIA-U) improved, complications still cause severe impairment of visual function in 25-33% of children that affects their psychophysical and psychosocial development and quality of life (QoL).

Objectives: To study the QoL and it's dimensions in children suffering from JIA-U as well as to investigate is there any difference in childhood's and parent's perception of disease between the group of children with JIA-U and children with JIA without uveitis.

Methods: The study included 42 children with JIA and their parents. Patients were divided into two groups. The first consisted of 21 children with JIA-U and the second of 21 children with JIA and no uveitis. Both groups of patients and their parents filled the Juvenile Arthritis Multidimensional Assessment Report questionnaire (JAMAR) for monitoring and assessing the health status of children with JIA. The variables used to test differences were: QoL, functional ability, pain level, disease activity estimation, and current emotional state of the child. The significance of differences between groups of children and parents was verified by the independent-samples t-test. The Pearson correlation coefficient was used for measurement of the strength of the linear relationship between variables.

Results: There were no statistically significant differences in the JIA-U group and the control group in either of the examined variables. Although there is a tendency of higher scores in children with JIA-U, which indicates their worse functioning, higher pain intensity and worse current emotional state, these differences were not statistically significant. Two groups did not differ significantly in the assessment of their own overall functional ability, which was associated with experienced pain intensity. Stronger pain intensity was associated with dysfunction ($r = 0.642$, p