Results: The frequency of heterozygote rs33996649GA genotype was higher in pSS patients than HS [OR=3.143 (1–10.234), p=0.046], and also, rs33996649GA genotype was associated with high SSDAI score (p=0.01). The pSS patients showed 44-fold more mRNA expression in comparison with HS (p=0.002), and mRNA expression correlates with SSDAI (r^2=0.512, p=0.006).

Conclusion: The rs33996649G>A genetic variant of the *PTPN22* gene is associated with increased development risk of pSS in the western Mexican population. The expression mRNA correlates with disease activity in pSS.

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Disclosure of Interests: None declared

DOI: 10.1136/annrheumdis-2020-eular.2173

AB0017 CONSTANT GENETIC MARKER IL1B T-31C IS ASSOCIATED WITH ANAMNESIS OF BIOLOGICAL DRUGS TREATMENT IN RHEUMATOID ARTHRITIS

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Background: Rheumatoid arthritis (RA) is chronic progressive joint disease with erosions formation. Timely and effectiveness treatment is important due to quickly structural damage and progressive losing of active motion. Synthetic DMARDs didn't have a sufficient effect. Using biological drugs seemed like a panacea, but according to investigations at least 30-40% RA-patients lost treatment efficiency. Biological drugs act through immune cascade, that's why mutation in regulatory region of cytokine genes may partly determine treatment failure.

Objectives: The objective of our study was to analyze the frequency of *IL1 T-31C* single nucleotide polymorphism in patient with rheumatoid arthritis and its association with biological drugs prescribing.

Methods: One hundred two Caucasian RA-patients (age – 56 yrs [45; 61]; DAS28 4.7 [3.8; 5.9]) were enrolled in our study. All of them had American College of Rheumatology (ACR)-defined RA (1987 classification criteria) and gave written inform consent. Single nucleotide polymorphisms *IL1B T-31C* (rs1143627), *IL4 C-590T* (rs2243250), IL10 C-592A (rs1800872), *IL10 A-1082G* (rs1800896) were determined by restriction fragment length polymorphism. Descriptive statistics, Chi-squared test were used for data analysis. Results are presented as median and 25th/75th percentiles (Me [25th percentile; 75th percentile]).

Results: The most of SNPs analyzed had corresponded to the Hardy Weinberg equilibrium (HWE). The only exception was IL1B T-31C – the frequencies were differed statistically significant from HWE (p=0,03). Forty seven (46.1%) patients were treatment with biological drugs. Homozygotes IL1b -31CC were founded more frequently beside patients with biological treatment compare with other group (13 from 47 (27,7%) vs. 6 from 52 (11.5%), p=0,042). Other SNPs didn't demonstrate any associations.

Conclusion: Single nucleotide polymorphism *IL1B T-31C* (rs1143627) may be used for prognosis of basic anti-inflammatory therapy inefficiency and the needing for prescribing biological therapy.

Disclosure of Interests: None declared

DOI: 10.1136/annrheumdis-2020-eular.2404

AB0018 TNFA RS1800629 POLYMORPHISM: WHAT ABOUT ITS ASSOCIATION WITH CLINICAL MANIFESTATIONS AND ANTI-TNFA THERAPY? DATA FROM A SERIES OF ITALIAN PATIENTS WITH BEHÇET SYNDROME

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Background: Tumor Necrosis Factor-alpha (TNF-α) is a pleiotropic cytokine with a critical role in the pathogenesis of Behçet syndrome (BS). Anti-TNF-α therapy is useful for patients with refractory, severe BS, in particular for ocular, central nervous system, and gastrointestinal manifestations. However, although biological treatment with anti-TNF-α agents are effective in BS, not all patients are definite responders. Non-responders patterns could be due to: alternative non-TNFα related pathway of inflammation; anti-drug antibodies presence or development; polymorphic alleles of *TNFα* gene. *TNFα* rs1800629 (-308G>A) is a drug-response single nucleotide polymorphism (SNP) located within the gene promoter. Poor and conflicting data are currently available about the association of this polymorphism and clinical manifestations of BS, as well as about the responsivness to the TNFα blockers in BS patients [1-3].

Objectives: Aims of this study were to investigate in a cohort of Italian patients with BS the frequency of rs1800629 genotypes and its association with clinical features and anti-TNFα therapy response.

Methods: Consecutive patients with BS were recruited. Patients demographic and clinical data were collected by medical records and analyzed. Home-made specific primer pairs were used for rs1800629 coverage. gDNA was isolated and amplified using PCR. Good-quality amplicons were sequenced (Sanger method). *In silico* analysis was downstream performed using specific software for query-subject similarity analysis.

Results: 130 BS patients (64M:66F; mean age: 45.8±12.3 years) were included in the study. Patients predominant lesions were oral aphtosis (100%), eye involvement (86.2%), skin lesions (72.3%) and genital ulcers (57.7%). *TNFα* rs1800629 wild-type GG genotype was found in 106/130 BS patients (81.5%); the heterozygous genotype (GA) was identified in 24/130 patients (18.5%). No statistically significant differences were found in genotypes frequencies when the patients were stratified for presence and absence of each clinical manifestation (p>0.05), while statistical significant differences were found when the patients were compared for therapy (anti-TNFα drugs) response. In detail, 73/130 patients (56.2%) were treated with anti-TNFα agents. We found 16/73 (21.9%) non-responders patients (NRP). In NRP group, we identified 9/16 patients (56.3%) with GG genotype and 7/16 (43.7%) with GA genotype, while 8/57 (14.0%) responder patients showed GA genotype and 49/57 responder patients (86.0%) showed GG genotype (p=0.0093; OR: 0.21, CI: 0.06-0.729).

Conclusion: Here we described a low frequency of *TNFα* rs1800629 SNP-containing allele and the lack of association between SNP and BS clinical hallmark, as previously reported in literature [1-4]. We also found higher percentage of GG genotype in case of therapy response than GA genotype. The SNP is a promoter polymorphism that could affect the auto-inflammatory response and the therapy responsivness, as suggested by our preliminary data of pharmacogenomics. Analyses of a larger cohort of patients are need to confirm the study findings and to explain the SNP role as outcome predictor.

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Disclosure of Interests: Maria Carmela Padula: None declared, Pietro Leccese: None declared, Nancy Lascaro: None declared, Giusi Gaia Sorrento: None declared, Rosa Paola Radice: None declared, Antonina Rita Limongi: None declared, Teresa Carbone: None declared, Angela Padula: None declared, Giuseppe Martelli: None declared, Salvatore D'Angelo Consultant of: AbbVie, Biogen, BMS, Celgene, Eli Lilly, MSD, Novartis, and UCB, Speakers bureau: AbbVie, BMS, Celgene, Eli Lilly, Novartis, Pfizer, and Sanofi

DOI: 10.1136/annrheumdis-2020-eular.2295

AB0019 GENETIC POLYMORPHISM OF THE INFLAMMATORY MARKER SAA1 RS12218 (-13 T/C) IS ASSOCIATED WITH AN APTITUDE TO CLINICAL PHENOTYPES OF JUVENILE IDIOPATHIC ARTHRITIS

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Background: Juvenile idiopathic arthritis (JIA) is one of the most widely-spread immuno-inflammatory diseases of an unknown etiology, the leading manifestation of which is chronic joint inflammation, occuring in children under the age of 16. The disease is a complex of chronic arthropathies with various phenotypic manifestations.