Abstract AB1128 – Figure 1. Serum Versican levels between FMF and Control groups

Conclusions: Serum versican level was statistically significant lower in FMF group than in control. On the other hand, there was no statistically significant correlation between versican levels and other parameters in FMF patients with attack-free period. Clinical trials including FMF patients with attack-period, attack-free period and amyloidosis would be designed in order to investigate the possible role of versican.

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AB1129 EPIDEMIOLOGICAL AND CLINICAL PROFILE OF PATIENTS WITH PARANEOPlastic RHEUMATOLOGIC SYNDROMES IN A SPANISH TERTIARY HOSPITAL

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Background: Paraneoplastic rheumatic syndromes (PRS) are defined as rheumatic symptoms caused by a malignancy, but which are not caused directly by the tumour or metastasis. PRS are characterised by a close temporal relationship with an underlying malignancy and fast regression of the symptoms after the treatment of the primary tumour.

Objectives: To describe the epidemiological and clinical profile of patients diagnosed with PRS in a Spanish tertiary hospital.

Methods: A retrospective observational study was performed, which included all patients who were diagnosed of PRS in our centre during the period 1985–2017. The demographic, clinical and outcome data were obtained from their medical records.

Results: During the study period 48 patients with suspected paraneoplastic syndrome were identified, but diagnosis was confirmed in 14 of them, 7 women (50%), with a mean age at diagnosis of 69.8±14.5 years. 42.9% of patients were smokers, 14.3% reported alcohol consumption, 35.7% and 7.1% had personal and family history of neoplasia, respectively; no patient reported personal or family history of autoimmune disease. The comparison between patients with and without PRS is shown in the table, but no significant differences were identified. The most common neoplasm was breast cancer (28.6%). 21.4% of the cases of PRS appeared as a consequence of a tumour recurrence. In 42.9% of patients the diagnostic of the tumour was concomitant to PRS, while the remaining 57.1% SPR was previous. The most frequent paraneoplastic syndrome was polymyalgia rheumatica (42.9%), followed by dermatomyositis (28.6%) and paraneoplastic polyarthritis (28.6%). The most common musculoskeletal manifestation was shoulder girdle pain and stiffness (35.7%) followed by polyarthritis and proximal

muscular weakness (28.5%). 85.7% of the patients had high erythrocyte sedimentation rate and C-reactive protein levels with a mean value of 60.2±39.4 mm/h and 77.6±9.5 mg/L, respectively. Antinuclear antibodies were positive in 7 patients (50%), while rheumatoid factor, anti-CCP and anti-RNP were positive in 1 patient each (7.1%). The response of PRS to the treatment of the tumour was available in 9 patients, of which 6 (66.7%) presented remission of the PRS. Regarding mortality, 5 patients (35.7%) died due to complications associated with the primary tumour.

Abstract AB1129 – Table 1

	PRS (n=14)	NO PRS (n=34)	P-value
Age (years)	69.78±14.57	73.23±10.81	0534
Sex (Male)	7 (50%)	14 (41,17%)	0575
Personal history of malignant neoplasm	5 (35,71%)	16 (47,05%)	0471
Personal history of autoimmune disease	0 (0%)	4 (11,76%)	0180
Family history of malignant neoplasm	1 (7,14%)	0 (0%)	0115
Family history of autoimmune disease	0 (0%)	1 (2,94%)	0115
Smoking	6 (42,86%)	13 (38,24%)	0766
Alcoholism	2 (14,29%)	4 (11,76%)	0810

Comparison of the demographic characteristics of patients with and without a diagnosis of paraneoplastic rheumatic syndrome.

Conclusions: Polymyalgia rheumatica was the most frequent PRS in our study, in contrast to several published series where other PRS are more common such as hypertrophic osteoarthropathy, polyarthritis and dermatomyositis/polymyositis. The laboratory data and response to treatment of the tumour were similar to those reported in the literature. The mortality in our series was high, but in none of the cases was related directly to PRS.

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AB1130 LATE ONSET TUMOUR NECROSIS FACTOR RECEPTOR (TNFR)–ASSOCIATED PERIODIC SYNDROME (TRAPS) CAUSED BY SOMATIC TNFRSF1 MOSAICISM

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Background: Tumour necrosis factor receptor (TNFR)–associated periodic syndrome (TRAPS) is an autosomal-dominant disease caused by gain-of-function mutations in the TNFRSF1A gene, which encodes the 55-kd TNFR type I (TNFRI) protein. Mosaicism has been identified in a single patient. A 60 year old male presented with a 6 year history of intermittent fever as high as 103.5, lasting 3–4 weeks with associated peritoneal symptoms, arthralgias, myalgias, lymphadenopathy, bilateral episcleritis, erythematous rash in his torso. Prednisone up to 60 mg daily and colchicine was ineffective where he responded fully to canakinumab at a dose of 150 mg every 4 weeks.

Objectives: To identify the extent of mosaicism in a patient with adult onset TRAPS phenotype.

Methods: DNA was extracted from the patient's whole blood, saliva and hair root. The TNFRSF1A gene was analysed by Sanger sequencing in all tissues, whole blood and fractionated cell subsets. *In silico* molecular modelling was performed to predict the structural and functional consequences of the tumour necrosis factor receptor (TNFR) type I protein mutation.

Results: Sanger sequencing revealed differential tissue and hematopoietic cell presence of a missense mutation at c.265 T>C p.Phe89Leu (F89L) Chr12 (GRCh37):g.6442960A>G ex3 rs104895245. The mutant allele was present in whole blood and buccal mucosa and absent in hair root, supporting the presence of somatic TNFRSF1A mosaicism. Additionally, quantitative analysis revealed allele frequency of 50% in B cells, 30% in NK cells and 30% in neutrophils. The mutation was absent in T cells and monocytes. *In silico* prediction modelling with SIFT and PolyPhen2 suggested that this mutation led to numerous structural rearrangements which resulted in changes in the protein surface profile.