presence of sinovitis, power Doppler (PD) signal, bone erosions, and cartilage changes; serology and hand x-ray were requested and evaluated.

Results: We included 5 women and 1 man, mean age was 36.2 ± 10.3 years, and they all had characteristics of acute (4 patients) or chronic (2 patients) VKHD. They received treatment with intravenous methylprednisolone 1 g/day for 3 days combined with cyclophosphamide and thereafter methotrexate (12.5–17.5 mg/week) for maintenance therapy, scheme with which no ocular recurrences have been presented. Two patients had synovitis on physical examination. All patients had PD signal on the ultrasound and they had involvement of both wrists, MCPs and PIPs joints consistent with polyarthritis pattern; additionally bone erosions were detected in 2 patients. Rheumatoid factor and anti citrullinated peptide antibody were negative in all patients

Abstract AB1137 – Figure 1. a) Ocular ultrasonography of patient 1, showing bilateral exudative retinal detachment b) Articular ultrasound of patient 1, showing synovitis with PD signal in both wrists. c) Fundus photographs of the right and left eyes of patient 2 showing exudative retinal detachments d) Articular ultrasound of patient 2, showing synovitis with PD signal in both wrists.

Conclusions: The presence of inflammatory and erosive arthritis in patients with VKHD has only been described in one case recently. We propose that polyarthritis is common manifestation of this rare disease, including erosive arthritis, representing part of the spectrum of the disease, processes that share some features of the genetic susceptibility with rheumatoid arthritis as HLA-DR4, CTLA-4 and STAT4.

REFERENCES:

- [1] Voorduin S, et al. Frecuencia y causa de las uveítis en el Instituto de Oftalmología Conde de Valenciana. *Rev Mex Oftalmol* 2005; 79 (4): 193–196
- [2] Read RW, et al. Revised diagnostic criteria for Vogt- Koyanagi-Harada disease: report of an international committee on nomenclature. *Am J Ophthalmol* 2001; 131:647–652.
- [3] Aydin T, et al. Association of Vogt Koyanagi Harada Syndrome and Sero-negative Rheumatoid Arthritis. *Ethiopian Journal of Health Sciences*. 2016;26(2):193–196
- [4] Baltmr A, et al. Vogt–Koyanagi–Harada syndrome – current perspectives. *Clin Ophthalmol*. 2016; 10: 2345–2361.

Disclosure of Interest: None declared

DOI: 10.1136/annrheumdis-2018-eular.5689

AB1138

HIGH LEVELS OF ANTI-U1RNP AND ANTI-SM IN MIXED CONNECTIVE TISSUE DISEASE PATIENTS

E. Grau García¹, P. Jover Carbonell², I. Martínez Cordellat¹, R. Negueroles Albuixech¹, J.E. Oller Rodríguez¹, F.M. Ortiz Sanjuan¹, E. Vicens Bernabeu¹, C. Alcañiz Escandell¹, K.R. Arevalo Ruales¹, I. Chalmeta Verdejo¹, M. De la Rubia Navarro¹, C. Feded Olmos¹, J.J. Frago Gil¹, R. Gonzalez Mazario¹, L. Gonzalez Puig¹, J. Ivorra Cortés¹, E. Labrador Sánchez¹, C. Najera Herranz¹, I. Canovas Olmos¹, A. Cañada Martínez³, A. Alba Redondo⁴, M.L. Martínez Triguero⁴, J. A. Roman Ivorra¹. ¹Rheumatology Department, HUP La Fe; ²Medical School, UCV; ³Biostatistics Unit, IIS La Fe; ⁴Clinical Analysis Department, HUP La Fe, Valencia, Spain

Background: The mixed connective tissue disease (MCTD) is an autoimmune systemic disease characterised by clinical manifestations that are included in systemic lupus erythematosus (SLE), scleroderma (SSc) or rheumatoid arthritis (RA). Moreover the MCTD exhibits anti-U1RNP high-titer antibodies. However, anti-U1RNP antibodies are not specific or exclusive from MCTD, because of can be detected in other systemic autoimmune diseases as SLE, SSc and RA.

Objectives: To verify the differentiation of MCTD patients from other systemic autoimmune diseases using anti-U1RNP titers. The secondary objective is to characterise anti-U1RNP titer in other systemic autoimmune diseases with clinical manifestation.

Methods: An observational retrospective study of patients with inflammatory autoimmune disease evaluated in the Rheumatology Department from 2012 since 2016 was performed. In all cases a blood-test with anti-U1RNP, anti-Sm, anti-Ro, and anti-La analysis was conducted. Clinical data was registered according to the patients' medical history, with special emphasis being placed on renal affection, vascular affection, pulmonary hypertension, arthritis-synovitis, tendonitis-tenosynovitis, dry eye syndrome and Raynaud's phenomenon. Biostatistical analysis was performed using R.

Results: We collected data from 355 patients with a mean age of 50.84 (15.49) years, 98.55% of them were female. 13.8% of patients showed anti-U1RNP high titers (up to 20 pg/mL), and a significant increase of anti-U1-RNP in MCTD patients in contrast to other connective pathologies ($p < 0.0001$) was observed. Anti-Sm antibody also exhibit significantly higher values in MCTD patients than in RA ($p = 0.025$) or scleroderma ($p = 0.003$). No differences in anti-Ro and anti-La levels among all diagnosis were observed. Patients with the high anti-U1-RNP levels, regardless of the diagnosis, showed more Raynaud's phenomenon and vascular affection, ($p < 0.001$ y $p = 0.008$). Related to Anti-Ro and anti-La, high titer of these antibodies in patients with Dry eye syndrome was observed ($p < 0.001$). Specifically in SLE patients, those with the highest levels of anti-U1-RNP exhibit Raynaud's phenomenon ($p < 0.001$), highest levels of anti-La was shown in those patients with renal affection ($p = 0.02$) and the highest levels of anti-Ro and anti-La was shown in those patients with Dry eye syndrome ($p = 0.002$ and $p = 0.006$).

Conclusions: In our patient series anti-U1RNP were significantly elevated in MCTD diagnosis, and in lesser extent anti-Sm antibodies. Anti-Ro and anti-La antibodies are increased in dry eye syndrome patients. In SLE patients, anti-La increased levels were associated to renal affection.

Disclosure of Interest: None declared

DOI: 10.1136/annrheumdis-2018-eular.6402

AB1139

RITUXIMAB FOR SCLERITIS AND PERIPHERAL ULCERATIVE KERATITIS ASSOCIATED WITH RHEUMATOLOGIC DISEASE

E. Valls Pascual¹, I. Benítez Hurtado², L. Martínez-Costa Pérez², C. Vergara Dangond³, M. Aguilar Zamora¹, L. Montolio Chiva¹, A.V. Orenes Vera¹, A. Ybáñez García¹, Á. Martínez. Ferrer¹, J.J. Alegre Sancho¹. ¹Rheumatology; ²Ophthalmology, HOSPITAL DOCTOR PESET, València; ³Rheumatology, Hospitales Madrid, Madrid, Spain

Background: Some types of ocular surface inflammatory diseases are often related to rheumatic conditions: 37% of scleritis (especially diffuse and necrotizing forms) and 60% of peripheral ulcerative keratitis (PUK). Rheumatoid arthritis (RA) and ANCA-associated vasculitis are the most frequently related conditions. Significant loss of visual acuity can be observed if these ocular diseases are not properly treated. To date, no approved therapies are available. Consequently, the management of these ocular diseases are based on published evidence coming from clinical trials (scarce and often with a low sample size), observational studies and case reports. There are positive efficacy data of rituximab (RTX) for ocular surface inflammatory disease.^{1–4}

Objectives: To describe our experience with RTX as a therapy for severe ocular surface inflammatory diseases associated to rheumatic conditions.

Methods: This is a retrospective observational study. It includes patients with severe scleritis or PUK associated to rheumatic diseases diagnosed and managed at our Multidisciplinary Uveitis Clinic between January 2006 and November 2017. We recorded demographic and clinical variables. As outcome variables we used the

Abstract AB1139 – Table 1. RTX for scleritis and PUK

	Age (ys)	Gender	Ocular disease	Rheumatic disease	Comorbidities	Previous treatment	RTX cycles	Δ VA	Inflammatory activity	AE	Follow up (ms)
1	55	F	PUK	RA	Amiloidosis	MTX, CS-A, IFX, MPLN ev	1	+0.1	No	No	132
2	58	F	Scleritis	GPA	HBV chronic carrier, LTI	MPLN ev, PDN	1	+0.1	No	No	19
3	59	F	Scleritis	RP	DM	MTX, CS-A, PDN, DXM io	1	0	Diffuse nodular scleritis	No	17
4	75	M	Scleritis	RP	DM, Sweet's syndrome, myelo displasic syndrome	MTX, CS-A, ETN, PDN	1	+0.5	No	No	11
5	75	F	PUK	RA	Previous thyroid CA, asthma	MTX, PDN	1	+0.5/ +0.1	No	No	25
6	60	F	PUK	RA	No	MTX, AKR, IFX, PDN	2	+0.1	No	No	128
7	70	M	PUK	RA	LTI	MTX, CS-A, TCZ, PDN	1	+0.1	No	No	10
8	53	F	PUK	RA	CSC	MTX, LFN, PDN	1	+0.1/0	No	No	7

change in visual acuity and the presence of inflammatory activity by biomicroscopy.

Results: VA: visual acuity, AE: adverse events, RA: rheumatoid arthritis, GPA: granulomatosis with polyangiitis, RP: relapsing polychondritis, LTI: latent tuberculous infection, DM: diabetes mellitus, CSC: central serous chorioidopathy, MPLN: metilprednisolone, PDN: prednisone, DXM: dexametasone, MTX: methotrexate, CS-A: cyclosporine, LFN: leflunomide, IFX: infliximab, ETN: etanercept, AKR: anakinra, TCZ: tocilizumab.

Conclusions: As previously described we consider rituximab as an effective therapy for severe ocular surface inflammatory diseases related to rheumatic conditions when other immunosuppressant drugs fail or are contraindicated.

REFERENCES:

- [1] - Albert M, Beltrán E, Martínez-Costa L. Rituximab in rheumatoid arthritis-associated peripheral ulcerative keratitis. *Arch Soc Esp Oftalmol*. 2011; 86 (4):118–20.
- [2] - Sims J. Scleritis: presentations, disease associations and management. *Postgrad Med J*. 2012;88(1046):713–8.
- [3] - Jabs DA, Mudun A, Dunn JP, Mardsh MJ. Episcleritis and scleritis: clinical features and treatment results. *Am J Ophthalmol* 2000;130:469.
- [4] - Suhler EB, Lim LL, Beardsley RM, Giles TR, Pasadhika S, Lee ST, et al. Rituximab therapy for refractory scleritis: results of a phase I/II dose-ranging, randomized, clinical trial. *Ophthalmology*. 2014;121(10):1885–91.

Disclosure of Interest: None declared

DOI: 10.1136/annrheumdis-2018-eular.1419

AB1140

COMPARISON OF CUT-OFF VALUES FOR FERRITIN TO DISCRIMINATE ADULT ONSET STILL'S DISEASE FROM OTHER CAUSES OF FEVER OF UNKNOWN ORIGIN: RESULTS OF A PROSPECTIVE STUDY

E. Bilgin¹, A. Erden², B. Armagan², L. Kılıç², A. Sarı², G. Yardımcı², E. Bilgin³, A. Akdoğan¹, Ö. Karadağ², S. Apraş Bilgen⁴, I. Ertenli², S. Kiraz², U. Kalyoncu².

¹Internal Medicine; ²Internal Medicine, Division of Rheumatology; ³Physical Medicine and Rehabilitation, Hacettepe University Faculty of Medicine; ⁴Internal Medicine, Division of Rheumatology, HACETTEPE UNIVERSITY, Ankara, Turkey

Background: Adult onset Still's disease (AOSD) is a rare, auto-inflammatory disease that commonly presents as fever of unknown origin (FUO). As AOSD can cause complications as reactive lymphohistiocytosis that can be mortal, discriminating AOSD from FUO is important. Ferritin, an acute phase reactant, is commonly high in patients with AOSD as a part of systemic inflammation.

Objectives: To determine a cut-off value of ferritin that favours AOSD over FUO.

Methods: Data from patients who admitted to Hacettepe University Hospitals, inpatients sections of department of internal medicine with the complaint of FUO collected prospectively during 30 months. Patients with uncertain diagnosis after all diagnostic procedures excluded. AOSD patients followed at Hacettepe University department of rheumatology were included. Of AOSD patients whose initial test ferritin levels were not available also excluded. For determination of cut-off values, receiver operator curve (ROC) analysis were done for each predetermined value. Upper normal level in our laboratory for ferritin is 336 ng/ml.

Results: Total 150 patients (n=63, for AOSD; n=87, for FUO) were included. Median ferritin level was significantly higher in AOSD group [1705 (657–6417) ng/ml vs. 424 (141–1188) ng/ml, p<0.001]. FUO group were also divided into three subgroups: rheumatologic (n=31, 35.6%), infectious (n=28, 32.2%) and malignant (n=28, 32.2%) causes. Median ferritin levels for these subgroups were 222 (104–1020), 527 (145–1057), 599 (166–2766) ng/ml, respectively. Area under curve

(AUC) 95% confidence intervals, sensitivity and specificity for predetermined cut-off values are given in Table-1. ROC curve for all ferritin levels are given in Figure 1.

Abstract AB1140 – Table 1

Cut/off value (ng/ml)	Area under curve (AUC)	Sensitivity, %	Specificity, %	p value	95% Confidence Interval (CI)
>UNL	0,73	98	49	<0001	0,65–0,81
>3xUNL	0,66	64	68	0001	0,56–0,74
>5xUNL	0,67	52	82	<0001	0,58–0,74
>750	0,69	75	63	<0001	0,60–0,77
>1250	0,66	59	76	0001	0,57–0,75
>2500	0,66	43	85	0001	0,57–0,75

*UNL: Upper normal limit (for ferritin: 336 ng/ml)

Figure-1: ROC analysis for each ferritin level (AUC 0,77%95 CI: 0.70–0.84, p<0.001)

Conclusions: For each cut-off values for ferritin, test performances were quite well to differentiate patients with AOSD from FUO. While ferritin levels higher than upper normal level have 98 percent sensitivity, ferritin levels higher than 5 times of upper normal level have 82 percent specificity. Large number of patients in each FUO subgroup is needed to determine ferritin test performance for each particular group of patients to differentiate from AOSD.

Disclosure of Interest: None declared

DOI: 10.1136/annrheumdis-2018-eular.3473

AB1141 UTILITY OF MUSCLE BIOPSY WITH NEEDLE IN A RHEUMATOLOGY SERVICE. A 12 YEARS EXPERIENCE

E. Casado¹, S. García-Cirera¹, M.R. Bella², M. Arévalo¹, M. Larrosa¹, J. C. Ferreres², J. Gratacós¹. ¹Rheumatology; ²Pathology, University Hospital Parc Taulí. I3PT. UAB. Sabadell, Sabadell, Spain

Background: Muscle biopsy with needle is presented as a faster and less invasive alternative than conventional open biopsy for the diagnosis of some myopathies. However, its use as a diagnostic technique is still very limited in the Rheumatology services.

Objectives: To describe the experience of 12 years and the diagnostic usefulness of needle biopsy in a Rheumatology service.

Methods: Descriptive study including all patients who, since 2005, had undergone a needle biopsy in the Rheumatology service of the Parc Taulí University Hospital in Sabadell, as a diagnostic technique for suspected myopathy.

The technique was performed in all cases on the lateral aspect of the thigh, about 10 cm above the knee. After disinfection and local anaesthesia, an incision of 1 cm longitudinal to the thigh was made until reaching a depth of about 3–4 cm, then introducing the Bergstrom needle for the muscle biopsy (about 4–5 muscle fragments of 2–3 mm taken in different directions) from the vast lateral. Finally, the incision was sutured with a single stitch.

In obese patients, a previous ultrasound was performed to exactly knowing the depth at which the muscle sample should be taken.

The collected samples were sent fresh to the Pathology service, wrapped in a gauze moistened with 0.9% physiological saline solution.

Results: In these 12 years we have performed a needle biopsy on 49 patients (29 women). Age 52±10 years (range 25–70). The reason for performing the biopsy was always the increase of muscle enzymes, mainly creatine kinase (CK), which in 9 of the cases was isolated, without any underlying disease, myotoxic drugs or other symptoms. Eight patients presented myalgia or weakness as the only symptomatology. Twelve patients had a rheumatic or autoimmune disease, and in 7 of these 12 cases there was a suspicion of antimalarials myopathy. In 6 cases the suspicion was dermatomyositis and in 5 cases of vasculitis. The biopsy was performed in 4 patients with fibromyalgia and in a patient with diabetes. In 4 of the cases, the suspicion was a lipid-lowering drug myopathy.

In 48/49 cases (98%) sufficient muscle sample was obtained. The technique had only to be repeated in one patient due to insufficient or inadequate tissue. Only in 2/49 cases (4%) the histological diagnosis was not concordant with the definitive clinical diagnosis (dermatomyositis), which was confirmed in one patient with open muscle biopsy.

In 31 cases (63%) the biopsy was normal. Among the pathological biopsies, the most frequent histological diagnosis was polymyositis, in 12 cases. In 3 cases a dermatomyositis was confirmed, in 3 cases a vacuolar myopathy by antimalarials and in 1 case a necrotizing myopathy.

As complications, it should be noted that only 2/49 (4%) patients presented moderate pain, which subsided in less than a week with analgesia, and one patient presented a hematoma in the area. No case of wound infection was observed.

Conclusions: Muscle biopsy with a needle is a quick, simple, low invasive and safe technique that can be very useful in a Rheumatology department. The incorporation of this technique as a diagnostic tool should be extended to the majority of Rheumatology departments.

Disclosure of Interest: None declared

DOI: 10.1136/annrheumdis-2018-eular.4066

AB1142 REDUCTION OF ABSOLUTE NUMBER OF CD4+CD25+FOXP3+ TREG CELLS IS ASSOCIATED WITH PATHOLOGICAL FEATURES OF PATIENTS WITH IDIOPATHIC INFLAMMATORY MYOPATHY

H. Zhao, H.Y. Wen, X.F. Li, Y. Liu. *Rheumatology, the Second Hospital of Shanxi Medical University, Taiyuan, China*

Objectives: To explore the alterations and their significance of peripheral CD4⁺CD25⁺ FOXP3⁺ regulatory T cells (Tregs) and Th17 cells in Idiopathic inflammatory myopathy.

Methods: Clinical indicators of IIM cases (n=85) and healthy controls (n=60) were enrolled. The absolute number of peripheral Tregs and Th17 cells were analysed by flow cytometry. The clinical features were collected retrospectively. Since the data was disregarded from the normal distribution, the median four quantile method was used for statistical description. Two samples were compared with Mann-Whitney U test, and the correlation between variables was Spearman rank correlation analysis.

Results: (1) The absolute number of Treg cells in the patients was significantly lower than that in the control group ($P<0.05$); the ratio of Th17/Treg was also significantly higher than that in the control group ($P<0.05$); (2) Peripheral Treg cells levels were negatively correlated with CRP ($r=0.279$, $p<0.05$); (3) According to the involvement of important organs was classified into two groups: organ group and non-organ group. The absolute number of Treg cell in organ group is fewer than

that in non-organ group ($p<0.05$), and the ratio of Th17/Treg in organ group was significantly higher than that in non-organ group.⁴ The peripheral Th17 cell absolute number in patients with skeletal muscle inflammatory oedema was significantly higher than that of non-inflammatory oedema patients ($p<0.05$);⁶ The levels of Th17, Tregs and ratio of Th17/Treg did not correlated with pathological features of inflammatory infiltration ($p>0.05$).

Conclusions: The absolute number of peripheral Treg cells decreased significantly in IIM, and correlated with CRP. Patients with organ involvement had fewer Treg cells, and imbalance between Th17 and Treg. When muscle MRI appeared inflammatory oedema, it has a higher level of Th17 cells. Our results suggest that Treg cells plays an important role in the pathogenesis of IIM and increasing the number of Treg cells and maintaining Th17/Treg immune balance will become a new therapeutic strategy for IIM.

REFERENCES:

- [1] Noack M, Miossec P. Th17 and regulatory T cell balance in autoimmune and inflammatory diseases.[J]. *Autoimmunity reviews*,2014,13(6):668–677.
- [2] Bluestone J A, Trotta E, Xu D. The therapeutic potential of regulatory T cells for the treatment of autoimmune disease.[J]. *Expert opinion on therapeutic targets*,2015,19(8):1091–1103.
- [3] Vercoulen Y, Bellutti Enders F, Meerding J, et al. Increased presence of FOXP3+ regulatory T cells in inflamed muscle of patients with active juvenile dermatomyositis compared to peripheral blood.[J]. *PloS one*,2014,9(8): e105353.
- [4] Young N A, Sharma R, Friedman A K, et al. Aberrant muscle antigen exposure in mice is sufficient to cause myositis in a Treg cell-deficient milieu.[J]. *Arthritis and rheumatism*,2013,65(12):3259–3270.
- [5] Milward K F, Wood K J, Hester J. Enhancing human regulatory T cells in vitro for cell therapy applications.[J]. *Immunology letters*,2017,190:139–147.

Acknowledgements: Thanks for my teachers, classmates and my family.

Disclosure of Interest: None declared

DOI: 10.1136/annrheumdis-2018-eular.4116

AB1143 AUTOIMMUNE CHARACTERISTICS IN A COHORT OF 89 PATIENTS WITH PRIMARY BILIARY CHOLANGITIS

I. Carballo Fernández¹, B. Sopena¹, E. Otero², S. Tomé², R. Alende², E. Páez¹, A. García-Villafranca¹, A. González-Quintela¹. ¹Immunopathology Unit, Internal Medicine; ²Hepatology and Liver transplant Unit, Complejo Hospitalario Universitario de Santiago de Compostela. Universidad de Santiago de Compostela., Santiago de Compostela, Spain

Background: Primary biliary cholangitis (PBC) is associated to other autoimmune diseases with an unknown prevalence. Their treatment can prevent progression to liver cirrhosis and other systemic complications

Objectives: To describe the clinical and analytical autoimmune characteristics in a cohort of patients with PBC diagnosed and followed in a specific unit of Hepatic Diseases. All patients were also studied in the Autoimmune and Systemic Diseases Unit of our hospital to check for the presence of extrahepatic autoimmune diseases.

Methods: We have studied patients with PBC diagnosed in our Service since 1994 who are currently under follow-up. The diagnosis of PBC was made taking into account: the presence of colostasis enzymes with positive anti-mitochondrial antibodies (AMA) and/or compatible liver biopsy

Results: We studied 89 patients with PBC. 81 of them were women (female/male ratio 10/1) with a mean age at diagnosis of 56 years (range 23–84 years). The mean follow-up was 106 months (range 9–286). IgM was elevated in 70% of the patients in whom it was found (56/80). The ANA were positive in 71% (61/86) and the AMA in 75% (67/89). 43% of patients AMA-negative PBC had other suggestive antibodies: 7 anti-centromere, 2 AMA-2 and 1 anti-sp-100. A liver biopsy was performed on 75 patients (87%), resulting in a diagnosis of 58% and useful to exclude other pathologies in the rest. In 18 patients (20%) an overlapping condition was diagnosed: PBC +Autoimmune hepatitis. In 11 patients (12%) a Reynolds syndrome was diagnosed: PBC +Scleroderma, in all of them Raynaud phenomenon was present. On another 11 of 41 (27%) Raynaud phenomenon was also present. In 17 patients (19%) there was a history or new diagnosis of autoimmune thyroiditis and in 13 patients (15%) of Sjögren syndrome. None of them was diagnosed of IgG4-related disease. Serum IgG4 was measured in 56 patients (63%) with a mean value of 36.6 mg/dL.^{2,8–125}

The patients with pure PBC were treated only with ursodeoxycholic acid, with a complete response of 41%, a partial response of 51% and an absence of response of only 6 (7%). There was no difference regarding liver response to treatment among patients with pure PBC and patients with overlapping autoimmune hepatitis, Reynolds syndrome, Raynaud phenomenon, Sjögren syndrome nor autoimmune thyroiditis. Only 3% of patients with complete response and 12% of

those who responded partially evolved to liver cirrhosis, with a similar follow-up time in both groups (105 vs 113 months). 5 of the 6 patients who did not have a biochemical response developed liver cirrhosis. 7 of the 13 patients with cirrhosis (54%) already presented clinical or histological data of cirrhosis in the initial evaluation

Conclusions: PBC patients have frequently other autoimmune diseases such as Autoimmune Hepatitis, Sjögren syndrome or Scleroderma so we must actively seek the presence of these pathologies. The treatment with ursodeoxycholic seems to be useful in all patients but it is important to make an early diagnosis

Disclosure of Interest: None declared

DOI: 10.1136/annrheumdis-2018-eular.7546

AB1144

HISTOLOGY OF ROSAI-DORFMAN DISEASE IN A SUBSET OF PATIENTS WITH ERDHEIM-CHESTER DISEASE: A DISTINCT ENTITY MAINLY DRIVEN BY MAP2K1

J. Razanamahery¹, E. Diamond², F. Cohen-Aubart¹, P. Karl-Heinz³, F. Charlotte⁴, Z. Helias-Rodziewicz⁵, A. Dogan⁶, O. Abdel-Wahab⁶, B. Durham⁶, N. Ozkaya⁶, Z. Amoura¹, J.-F. Emile⁵, J. Haroche¹. ¹Internal medicine, Pitié-Salpêtrière hospital, Paris, France; ²Neurology, Memorial Sloan Kettering Cancer Center, New York, USA; ³Neurology, Frankfurt-Cancer-Institute, Frankfurt, Germany; ⁴Pathology, Pitié-Salpêtrière hospital; ⁵Pathology service, Ambroise Paré hospital, Paris, France; ⁶Pathology and Laboratory, Memorial Sloan Kettering Cancer Center, New York, USA

Background: Diagnosis of Erdheim-Chester disease (ECD) is based on characteristic imaging of bone, retroperitoneal and/or cardiovascular involvement.¹ Biopsy is mandatory to exclude other diagnoses and confirm infiltration of histiocytes, but histology is not specific.² By contrast diagnosis of Rosai-Dorfman disease (RDD), a rare histiocytosis, is based on histology, which is characterised by infiltration by CD68 +CD1 a- S100+ histiocytes with large nuclei and abundant lesions of emperipolesis.² Up to 70% of ECD have *BRAF* or *MAP2K1* mutations,³ which are rare in RDD.

Objectives: We investigated patients harbouring an ECD phenotype but RDD histology.

Methods: We reviewed records of ECD patients followed in Pitié-Salpêtrière hospital (Paris, France) and Memorial Sloan Kettering Cancer Centre (New-York, NY, USA) between 2007 and 2018. Biopsy samples of the patients were systematically investigated for mutations of genes of MAP kinase pathway.

Results: Among 209 patients with ECD, we found 10 (4.7%) patients who had RDD histology. These 10 patients had typical ECD clinical and radiological presentation, in particular bones (n=7), vascular (n=5) and peritoneal (n=6) involvements. Patients also had typical neurological involvement of ECD (n=6). All patients except one had at least one biopsy with a compatible histology of ECD at diagnosis. ECD biopsies showed non-specific fibrosis (n=5), foamy CD 68 +CD1 a- histiocytes (n=3) and/or Touton cells (n=1). Biopsies disclosing RDD histology were performed during the course of the disease involving testes (n=5), stomach (n=1), tibia (n=2), cheek (n=1) and omentum (n=1). All tissues showed lympho-plasmocytic infiltrate with large histiocytes infiltration. Histiocytes were CD68 +CD1 a- S100 +with large nuclei and abundant lesions of emperipolesis. Five patient harboured *MAP2K1* mutation and one patient had *PIK3CA* mutation. None of the patients had *BRAF* mutation.

Conclusions: Some patients with ECD may also present the iconic histological lesions described by Rosai and Dorfman. Overlap forms of such distinct histiocytoses between ECD and RDD is mainly driven by *MAP2K1* but not by *BRAF*.

REFERENCES:

- [1] Diamond EL, Dagna L, Hyman DM, Cavalli G, Janku F, Estrada-Veras J, et al. Consensus guidelines for the diagnosis and clinical management of Erdheim-Chester disease. *Blood*. 2014;124:483–92.
- [2] Emile J-F, Abia O, Fraïtag S, Horne A, Haroche J, Donadieu J, et al. Revised classification of histiocytoses and neoplasms of the macrophage-dendritic cell lineages. *Blood*. 2016;127:2672–81.
- [3] Haroche J, Charlotte F, Arnaud L, von Deimling A, Hélias-Rodziewicz Z, Hervier B, et al. High prevalence of *BRAF* V600E mutations in Erdheim-Chester disease but not in other non-Langerhans cell histiocytoses. *Blood*. 2012;120:2700–3.

Disclosure of Interest: None declared

DOI: 10.1136/annrheumdis-2018-eular.4917

AB1145

ACUTE POSTERIOR MULTIFOCAL PLACOID PIGMENT EPITHELIOPATHY: CLINICAL PATTERN AND MANAGEMENT OF 79 PATIENTS

J. Álvarez Troncoso¹, A. Schlinker Giraud², F. Arnalich Fernández¹, Á. Robles. Marhuenda¹. ¹Internal Medicine; ²Ophthalmology, Hospital Universitario La Paz, Madrid, Spain

Background: Acute Posterior Multifocal Placoid Pigment Epitheliopathy (APMPPE) is an uncommon inflammatory disease causing acute-onset chorioretinal bilateral disease. It typically affects the posterior pole of both eyes leading to visual blurring or scotomas. Although it is thought to be benign, APMPPE has been associated with central nervous system (CNS) manifestations: cerebral vasculitis, meningoencephalitis and cerebral vascular disease.

Objectives: The aim of this study was to define clinical features, systemic manifestations, treatment and outcomes of a review of 79 patients with APMPPE.

Methods: We retrospectively analysed the epidemiology, potential triggers, prodromes, clinical data, ophthalmological study, extraocular manifestations, treatment and outcomes of 79 patients collected through an extensive review of the literature from the first description of Gass JD up to the present time.

Results: A total of 79 patients were reviewed (47 male). Mean age at diagnosis was 30 years (with a range of 8 to 58 years old). 27 patients (34.2%) presented with a previous triggering being flu-like illness the most frequent.¹⁵ However, the complete serological study was only requested in 28 patients (and the immunological study just in 22) Median time from trigger to overt APMPPE was 9 days. Main clinical symptoms were: decreased visual acuity/blurred vision (100%), headache (51.8%) and photophobia (12.2%). Average decreased visual acuity was 13/20. APMPPE is defined by the presence of multiple white-yellowish plaques in funduscopy, and early hypofluorescent areas with late hyperfluorescence in fluorescein angiography. The fundus was pathological and compatible with APMPPE in all cases (100%), as was fluorescein angiography in those that had been performed (59). CNS involvement appeared in up to 50.6% (40 patients). The CNS manifestations were divided into language disorders (11 patients), motor deficit,²⁰ sensory deficit²⁰ and other CNS manifestations.²³ The mean time from visual deficit to neurological manifestations was approximately 2 weeks. Cerebrospinal fluid was studied in 37 patients, with a predominance of lymphocytic pleocytosis (mean of 46 cells/mm3) and elevated proteins (mean of 111 mg/dl). Within the neuroimaging studies carried out (58) up to 69.7% were pathological. 67 patients (84.8%) received treatment with corticosteroids. 14 patients (17.7%) also received other immunosuppressants (mainly azathioprine and cyclophosphamide), especially if CNS involvement. Regarding the evolution, 55 patients (69.6%) presented improvement, 12 (15.2%) relapsed and 6 (7.5%) died due to APMPPE.

Conclusions: APMPPE is a rare inflammatory disease which primarily affects the retina. However, the CNS involvement could be more frequent than what is classically described. Also, it seems that there might be a trigger effect either inflammatory or infectious. Steroids and immunosuppressants should be considered in patients with CNS involvement from the beginning.

REFERENCE:

- [1] Case D, Seinfeld J, Kumpe D, Folzenlogen Z, Jones W, Simpson J, et al. Acute posterior multifocal placoid pigment epitheliopathy associated with stroke: a case report and review of the literature. *J Stroke Cerebrovasc Dis* 2015;24:e295-e302.

Disclosure of Interest: None declared

DOI: 10.1136/annrheumdis-2018-eular.3777

AB1146

THE ASSOCIATION OF COMMON MEFV GENE MUTATIONS WITH AXIAL SPONDYLARTHROSITIS IN FMF PATIENTS: A RETROSPECTIVE STUDY

K. Ayar¹, E.K. Ozturk², O. Yesiloz². ¹Rheumatology; ²Physical therapy and Rehabilitation, University of Health Sciences, Bursa Yuksek Ihtisas Training and Research Hospital, BURSA, Turkey

Background: Familial Mediterranean fever (FMF) is an autoinflammatory disease seen with autosomal recessive inheritance and is characterised by recurrent and self-limiting attacks with peritonitis, pleuritis, arthritis or fever alone. The association of spondylarthrosis and FMF is reported in some studies. There are few studies evaluating the association of MEFV gene mutations with axial spondylarthrosis in FMF patients.

Objectives: The aim of this study is to identify patients with FMF associated spondylarthrosis retrospectively and to compare the frequency of common MEFV gene mutations in FMF patients with and without axial spondylarthrosis.

Methods: We have reviewed 138 charts of FMF patients. The data of 116 patients (70 female, 46 male) who met the diagnosis of FMF with Tel Hashomer classification criteria and have the results of MEFV gene mutation were examined. Patients' age, sex, MEFV gene mutations were recorded. The presence of