REFERENCES

- [1] Stone JH, et al. IgG4-related disease. *N Engl J Med*. 2012 Feb 9;366(6):539-51.
- [2] Inoue D, et al. Immunoglobulin G4-related periaortitis and periarteritis: CT findings in 17 patients. *Radiology*. 2011 Nov;261(2):625-33.

Disclosure of Interests: None declared

DOI: 10.1136/annrheumdis-2019-eular.5534

AB1095

THE GENETIC CHAMELEON IN RHEUMATOLOGY'S BACK GARDEN: A 2-CASE PRESENTATION

Anca Moldovan¹, Ioana Felea¹, Laura Damian¹, Raluca Rancea², Liliana Bene³, Calin Boloșiu⁴, Simona Rednic^{1,5}. ¹Emergency Clinical County Hospital, Rheumatology, Cluj Napoca, Romania; ²Heart Institute "Niculae Stancioiu", Cardiology, Cluj Napoca, Romania; ³Emergency Clinical County Hospital, Immunology, Cluj Napoca, Romania; ⁴Emergency Clinical County Hospital, Radiology, Cluj Napoca, Romania; ⁵University of Medicine and Pharmacy Iuliu Hatieganu, Cluj Napoca, Romania

Background: Adult Refsum disease is an autosomal recessive disorder, in which phytanic acid cannot be metabolized and it accumulates in plasma and lipid-containing tissues (1). Although its common clinical features are known, patients are displaying symptoms that can mimic an autoimmune pathology, making this disease a diagnostic dilemma.

Objectives: To highlight the overlap between the clinical and serological features of adult Refsum disease with those of a connective tissue disease, in order to achieve the correct diagnosis and make appropriate therapeutic decisions.

Methods: Clinical and laboratory data were evaluated for 2 patients with adult Refsum disease in a rheumatology tertiary department. The data was retrieved from patients' files.

Results: Both patients were females in their 50s with undiagnosed adult Refsum disease. A medical history revealed that both reported progressive nyctalopia, anosmia, ageusia, peripheral vision loss. In addition to these clinical features one of them reported muscle weakness, weight loss and fatigue, displaying periorbital oedema, heliotrope rash, periungual capillary changes. Her muscular enzymes were elevated (creatin-kinase was 3 times elevated and lactat dehydrogenase was 9 times above normal range), along with C-reactive protein (CRP = 2.4mg/dl). Due to clinical features, increased serum enzymes and a recently diagnosed myocarditis, a high suspicion of dermatomyositis was raised, but was disregarded after extensive investigations (immunology tests, muscular biopsy). The other patient had, in addition, sicca features (dry mouth, eyes, skin) with a Schirmer test of 4mm and an elevated CRP (3.23mg/dl). She was referred to the rheumatology department with a high suspicion of Sjogren's syndrome. Immunology and minor salivary gland biopsy were negative. Interestingly, physical examination showed bilateral brachymetacarpia and brachymetatarsia in both patients, suggesting pseudopseudohypoparathyroidism but, after investigations, the diagnosis was excluded.

Both patients underwent ophthalmological examination to further investigate progressive nyctalopia and peripheral vision loss. Specific changes for pigmentary retinopathy were described, which, together with the above neurological changes and skeletal deformities, were highly suggestive of adult Refsum disease (2). Plasma phytanic acid was 41 and 42 times above normal range respectively, confirming the diagnosis.

After dietary restrictions which excluded phytanic acid containing foods, there was an improvement concerning both clinical and serological manifestations. All connective tissue disease relating symptoms, slowly and gradually started to subside. Muscular enzymes and CRP values returned within normal range (3).

Conclusion: High levels of phytanic acid have triggered a series of uncommon symptoms and serological changes resembling a connective tissue disease, making the diagnosis of adult Refsum disease a very tangled, costly and lengthy process. Although this disease affects many body systems, a case of myositis or sicca syndrome hasn't yet been acknowledged in relation to this condition.

REFERENCES

- [1] Wierzbicki A.S., Lloyd M.D, et al. Refsum's disease: a peroxisomal disorder affecting phytanic acid alpha-oxidation. *Journal of Neurochemistry*, 2002, 80, 727-735.
- [2] Gibberd F.B., et al. Heredopathia atactica polyneuritiformis: Refsum's disease. *Acta Neurol Scand* 1985;72:1-17.

- [3] Baldwin E., Gibberd FB, et al. The effectiveness of long term dietary therapy in the treatment of adult Refsum disease. *J Neurol Neurosurg Psychiatry* 2010;81:954-957.

Disclosure of Interests: None declared

DOI: 10.1136/annrheumdis-2019-eular.4797

AB1096

CLINICAL SPECTRUM AND TREATMENT OF THE RELAPSING POLYCHONDritis: A SERIES OF 12 CASES

Pablo Moreno¹, Dolores Martinez-Quintanilla Jimenez¹, Noelia García Castañeda¹, Jose-Maria Alvaro-Gracia¹, Isidoro González-Álvarez¹, Alicia Humberia¹, Irene Llorente¹, Ana Ortiz¹, Esther Patiño¹, Eva Tomero Muriel¹, Esther Vicente¹, Rosario Garcia de Vicuna¹, Santos Castañeda¹. ¹HU La Princesa, Reumatology, Madrid, Spain

Background: Relapsing polychondritis (RP) is a systemic autoimmune disease characterized by inflammation of the cartilaginous tissues associated with the appearance of symptoms caused by the involvement of other non-cartilaginous tissues and the presence of systemic symptoms.

Objectives: To describe the clinical characteristics and treatment of patients diagnosed with RP in a third level university hospital.

Methods: Data from RP patients were obtained from the computerized medical history of our center, including demographics, disease phenotype, treatment, laboratory and imaging studies.

Results: A total of 12 patients were included; 66.7% were women, age at diagnosis was 36 [34-52.5] years (median [p25-p75]). The main manifestation observed was auricular involvement (75%) followed by nasal (66.7%), joint (66.7%), and laryngo-tracheo-bronchial disease (50%). All patients were on DMARD's treatment and two thirds of the patients were receiving biological therapy at the time of analysis. Infliximab was the first biological line in all the cases (100%). Other biological therapies used, in order of frequency, were: adalimumab (25%), rituximab (25%) and tocilizumab (12.5%). 50% of the patients associated another autoimmune disease diagnosed before or during RP follow-up: Crohn's disease, epidermolysis bullosa, primary biliary cholangitis, autoimmune hypothyroidism, systemic lupus erythematosus, granulomatosis with polyangiitis and ulcerative colitis. Only one patient (8.33%) associated hematologic disease (myelodysplastic syndrome) and another one died during follow-up.

Conclusion: RP is a rare disease with a broad spectrum of clinical expression, which often requires intensive management to control its manifestations, potentially serious when there is involvement of the tracheo-bronchial tree or is associated with other autoimmune diseases. A large part of our patients require biological therapy at some point in their evolution.

REFERENCES

Disclosure of Interests: Pablo Moreno: None declared, DOLORES MARTINEZ-QUINTANILLA JIMENEZ: None declared, Noelia García Castañeda: None declared, Jose-Maria Alvaro-Gracia Consultant for: AbbVie, Bristol-Myers Squibb, Eli Lilly, MSD, Novartis, Pfizer Inc, Roche, Sanofi, and UCB, Speakers bureau: AbbVie, Bristol-Myers Squibb, Eli Lilly, MSD, Novartis, Pfizer Inc, Roche, Sanofi, and UCB, Isidoro González-Álvarez: None declared, Alicia Humberia: None declared, Irene Llorente : None declared, Ana Ortiz: None declared, Esther Patiño: None declared, Eva Tomero Muriel: None declared, Esther Vicente: None declared, Rosario Garcia de Vicuna Grant/research support from: Abbvie, BMS, Lilly, MSD, Novartis, Roche, Consultant for: Biogen, BMS, Mylan, Pfizer, Sanofi and Sandoz, Speakers bureau: BMS, Pfizer, Lilly, Sandoz, Santos Castañeda Consultant for: Amgen, BMS, Pfizer, Lilly, MSD, Roche, Sanofi, UCB

DOI: 10.1136/annrheumdis-2019-eular.7518

AB1097

RETROPERITONEAL FIBROSIS- A SINGLE CENTER EXPERIENCE

Mert Oztas¹, Emir Cemre², Izzet Atun³, Cetin Demirdag⁴, Serdal Ugurlu¹. ¹Istanbul University Cerrahpasa, Division of Rheumatology, Istanbul, Turkey; ²Istanbul University Cerrahpasa, Division of General Internal Medicine, Istanbul, Turkey; ³Mayo Clinic School of Medicine, Arizona, United States of America; ⁴Istanbul University Cerrahpasa, Department of Urology, Istanbul, Turkey

Background: Idiopathic retroperitoneal fibrosis (iRPF) is a rare, chronic, progressive disorder of unknown etiology and characterized by the presence of inflammatory and fibrous retroperitoneal tissue that often encompasses the ureters or abdominal organs.

Objectives: We evaluated iRPF cases of a single institution cohort to describe the features of patients (pts) with iRPF.

Methods: Twenty-eight iRPF patients who were being followed in our outpatient clinic between 2009 and 2018 were reviewed retrospectively. The demographic characteristics, clinical and radiologic findings, medical and interventional therapies were retrospectively collected and reported.

Results: We evaluated the data of 28 patients (17M/11F). The mean age at the time of diagnosis was 53±10.57, median follow up was 46 (IQR:23-67) months. Fifteen (50%) patients were presented with abdominal-pain which was the most common initial complaint; 9 (32%) flank pain, 1 anuria, 1 nocturia, and 2 had nausea. Twenty-one patients had hydronephrosis (11 bilateral hydronephrosis, 5 right and 5 left hydronephrosis) at initial visit and one patient diagnosed while on temporary hemodialysis therapy. Laboratory tests showed that ESR was elevated in 15 of 23 cases in which it was available (65.2%), with a median value of 39 mm/h (IQR:17-88); CRP was elevated in 19 of 28 patients (68%), with median value 11 (IQR:5.5-46.5). Mean serum creatinine level was 1.14 mg/dl IQR (0.83-1.9). Tissue involvement was detected with computed tomography in 25 (89%) patients. The remaining 3 patients' involvement was located with ¹⁸F-fluorodeoxyglucose positron emission tomography (¹⁸F-FDG-PET). ¹⁸F-FDG-PET has been performed in 19 patients and 16 of them (84%) revealed tissue involvement of iRPF. Histopathological samples were available for further analysis in 8 cases and 5pts' findings were consistent with iRPF (storiform fibrosis and dense lymphoplasmacytic infiltration were observed) and of these 5 pts, 2 of them were classified as IgG4-related subtype. The remaining 3 pts were diagnosed with positive radiologic findings. Medical and interventional therapies are presented in Table 1. The mean initial oral prednisone dose was 45.5±13.8 mg/day. Prednisone was discontinued in 4 patients after remission and for the remaining 23 pts; 12 pts' prednisone was tapered <5 mg/day at mean 9 (IQR:6.7-22.5) months. One patient diagnosed AML-M2 during the follow-up and had allogeneic stem cell transplantation. Final median values of ESR and CRP were 10 mm/h (IQR:4.7-17.7) and 4.4 mg/l (1.9-7.7), respectively (p= 0.001 and p=0.002). Final median serum creatinine level was 1.03 (IQR:0.78-1.28) (p=0.2). Thirteen pts (46) were treated successfully with Rituximab who were not responded initial prednisone therapy. Twenty-one pts had ureteral stents and 11 of them are stent free at the final visit. Two pts had ureterolysis but the procedure was failed in one patient. None of the pts was deceased on the follow-up.

Conclusion: Steroids are the mainstay treatment for iRPF and steroid resistant cases could be treated with rituximab rescue therapy. Prompt interventional approaches are valuable bridging therapies for pts who presented with hydronephrosis. Imaging modalities add great value on the diagnose of iRPF.

. Types of medical and interventional therapy

Types of medical therapy	n, (%)
Glucocorticoid	27(96.5)
Glucocorticoid only	3 (10.5)
Tamoxifen	8 (28)
Azathioprine	8 (28)
Cyclophosphamide	1 (3.5)
Methotrexate	10(35)
Rituximab	13(46)
Mycophenolate Mofetil	1 (3.5)
Types of interventional therapy	n, (%)
Left ureteral stents	15(53.5)
Right ureteral stents	16(57)
Right nephrostomy	4 (14)
Left nephrostomy	2 (7)
Ureterolysis	2 (7)
Temporary Hemodialysis	1 (3.5)

Disclosure of Interests: None declared

DOI: 10.1136/annrheumdis-2019-eular.7266

AB1098

COMPARISON OF THE PERFORMANCE OF CLASSIFICATION/DIAGNOSTIC CRITERIA FOR POLYMYALGIA RHEUMATICA. SINGLE CENTERSTUDY OF 100 PATIENTS

D. Prieto-Peña, Monica Calderón-Goercke, Miguel A. González-Gay, Ricardo Blanco. *Marqués de Valdecilla University Hospital, Santander, Spain*

Background: Polymyalgia rheumatica (PMR) diagnosis is based on the combination of shoulder and pelvic girdle pain associated with elevated

acute phase reactants. Traditionally, Bird et al.¹ and Chuang et al.² criteria have been used for establishing PMR diagnosis. In 2012 an international working group developed new European League Against Rheumatism (EULAR)/American College of Rheumatology (ACR) classification criteria for PMR.³

Objectives: a) To compare the performance of Bird et al., Chuang et al. and 2012 EULAR/ACR classification criteria for PMR in a single-center study. b) To describe the characteristics of the patients excluded by these criteria.

Methods: We included 100 patients with new-onset PMR who were consecutively diagnosed over a 7-year period by experienced rheumatologists from a single center. PMR diagnosis was confirmed during a prospective 24-months follow-up after excluding other mimicking conditions. Rheumatoid factor (RF) and anticitrullinated protein antibodies (ACPA) were determined in all patients. Subjects were classified by each of the three different criteria.

TABLE 1. 2012 ACR/EULAR CLASSIFICATION CRITERIA

Age at onset ≥ 50 years (required)	94/100	94%
Bilateral shoulder aching (required)	89/100	89%
Abnormal CRP and/or ESR (required)	96/100	96%
Morning stiffness duration > 45 min (2 points)	54/100	54%
Hip pain or limited range of motion (1 point)	90/100	90%
Absence of RF or ACPA (2 points)	100/100	100%
Absence of other joint involvement (1 point)	95/100	95%
Required for classification: score of 4 or more	84/100	84%

TABLE 2. CHUANG ET AL. CRITERIA (1982)

Age at onset ≥ 50 years (required)	94/100	94%
ESR ≥ 40 mm/hr	96/100	96%
Bilateral aching for ≥ 1 month involving two of the following areas:		
- Neck	47/100	47%
- Shoulders or proximal regions of the arms	89/100	89%
- Hips or proximal aspects of the thighs	90/100	90%
Exclusion of all other diagnosis of mimicking entities	100/100	100%
All criteria are required for classification	94/100	94%

TABLE 3. BIRD/WOOD CRITERIA (1979)

Age at onset > 65 years	59/100	59%
ESR > 40 mm/hr	96/100	96%
Bilateral shoulder pain/stiffness	89/100	89%
Duration onset < 2 weeks	77/100	77%
Stiffness > 1 h	54/100	54%
Depression and/or weight loss	16/100	16%
Bilateral upper arm tenderness	89/100	89%
Probable: 3 or more	96/100	96%

Results: We studied 100 patients (61 women/39 men); mean age 67.1 ± 10.2 years. 96% of the patients fulfilled Bird et al. criteria, 94% fulfilled Chuang et al. criteria and 84% of the patients fulfilled 2012 EULAR/ACR criteria (TABLE 1, 2 and 3). We assessed the characteristics of the 16 patients who did not fulfilled 2012 EULAR/ACR criteria: 6 of them were under 50 years of age (range age 43-48 years). Another 10 patients did not complain of pain/stiffness in the shoulder girdle at any time. However, they had typical inflammatory pain involving the pelvic girdle and elevated acute phase reactants.

Conclusion: In our single-center experience we found that 2012 EULAR/ACR criteria classified most of our patients. However, patients who presented predominant pelvic girdle affection and those who were under 50 years old despite showing all the rest of clinical and analytical required criteria were excluded. Some of these patients did fulfill Bird and/or Chuang criteria.

REFERENCES

- [1] Bird HA, et al. *Ann Rheum Dis.* 1979;38:434-9
- [2] Chuang T, et al. *Ann Intern Med.* 1982; 97:672-80
- [3] Dasgupta B, et al. *Ann Rheum Dis.* 2012; 71:484-92

Disclosure of Interests: D. Prieto-Peña: None declared, Monica Calderón-Goercke: None declared, Miguel A González-Gay Grant/research support