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

Anca Moldovan1, Ioana Felea1, Laura Damian1, Raluca Rancea1, Liliana Bane1, Calin Boloșiu1, Simona Rednic1-3, 1Emergency Clinical County Hospital, Rheumatology, Cluj Napoca, Romania; 2Heart Institute “Nicolae Stanciuc”, 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 Hatleganu, 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 were 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, perungual capillary changes. Her muscular enzymes were elevated (creatine-kinase was 3 times elevated and lactate 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 Sjögren’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.

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


Disclosure of Interests: None declared

AB1096
CLINICAL SPECTRUM AND TREATMENT OF THE RELAPSING POLYCHONDRIITIS: A SERIES OF 12 CASES

Pablo Moreno1, Dolores Martínez-Quintanilla Jiménez1, Noelia García Castañeda1, Jose-Maria Alvaro-Gracia1, Isidoro González-Álvaro1, Alicia Humbria1, Irene Llorente1, Ana Ortiz2, Esther Patiño2, Eva Tomero Muriel1, Esther Vicente1, Rosario García 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 tertiary reference 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, aged 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 Garcia 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-Álvaro: None declared, Alicia Humbria: None declared, Irene Llorente: None declared, Ana Ortiz: None declared, Esther Patiño: None declared, Eva Tomero Muriel: None declared, Esther Vicente: None declared, Rosario García de Vicuna Grant/research support from: Abbvie, BMS, Lilly, MSD, Novartis, Roche, Consultant for: Biogen, BMS, Mylan, Pfizer, Sanofi and Sandоз, Speakers bureau: BMS, Pfizer, Lilly, Sanó, Santos Castañeda Consultant for: Amgen, BMS, Pfizer, Lilly, MSD, Roche, Sanofi, UCB

AB1097
RETROPERITONEAL FIBROSIS- A SINGLE CENTER EXPERIENCE

Mert Oztas1, Emir Cerre1, Izzet Atun2, Cetin Demirdag3, Serdal Ugurlu3, 1Istanbul University Cerahpası, Division of Rheumatology, Istanbul, Turkey; 2Istanbul University Cerahpası, Division of General Internal Medicine, Istanbul, Turkey; 3Mayo Clinic School of Medicine, Arizona, United States of America; 4Istanbul University Cerahpası, 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 encases the ureters or abdominal organs.

REFERENCES

Disclosure of Interests: None declared