Conclusion: Our study has shown that rheumatology fascinates Moroccan medical students by its clinical abundance and the diversity of musculoskeletal pathology and also by the quality of life it can offer. According to the students, the main barriers to choose rheumatology are the interest in a surgical specialty, and the limited therapeutic aspects of the specialty.

Disclosure of Interests: None declared


AB1506

RHEUMATOLOGY TRAINING EXPERIENCE: ARE RHEUMATOLOGY RESIDENTS WELL-PREPARED NOWADAYS? A SELF-APPRaisal

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Background: Rheumatology residence in Europe is a 4-year training program where Medical Doctors (MD) purchase all capacities for turning into Rheumatologists at each Hospital. European guidelines concerning which contents should be acquired and recommendations for its obtaining are available (UEMS, Union Europeenne des Medecins Spezialistes). Several researches have proven high self-reported abilities, but some deficiencies are displayed in deep investigations: musculoskeletal ultrasonography, poorly crystal identification by optic microscopy and injection skills. As well, low clinical experience remains as one of the most self-reported deficiencies.

Objectives: To assess the quality of one-year rheumatology trainee consultation at the outpatient clinic in a Spanish hospital.

Methods: The last year of outpatient consultation of a resident training in Rheumatology in one Spanish hospital was assessed, from May 2021 to May 2022. All patients visited by the trainee were included, absent patients in the consultation date were excluded from the study. Main variables were collected, such as gender, main diagnosis, biological or targeted synthetic treatments and supplementary tests. The last resident’s opinion about his training was also checked.

Results: During one-year period of the Resident’s consultation, 1419 visits were performed. From those, 470 were the first visit of the patient and 119 of them were solved in the first visit and no follow-up was needed. 60.7% of patients were women. Most frequent diagnosis were arthropathies: 21.8% psoriatic arthropathies, 16.9% rheumatoid arthritis and 8.4% axial spondyloarthritis. Regarding collagenopathies, 3.4% of patients were diagnosed of systemic erythematosus lupus, 1.3% antiphospholipid syndrome, 2.3% systemic sclerosis and 1.3% myopathies. Bone-related diseases as Paget’s disease and osteoporosis were also frequent (21 and 132 patients). The most prevalent vasculitis was giant cell arteritis with 51 patients, although patients with Behcet’s disease, Takayasu arteritis and polyarteritis nodosa were also followed. A total of 207 musculoskeletal ultrasonography were made during daily clinical practice. As well, 67 articular injections were performed. Capillaroscopy and salivary gland biopsy were the least frequent procedures (21 and 18, respectively). Biological therapies most supervised by the resident were anti-TNF (76.9%), followed by anti-IL-6 (12.8%) and anti-IL-17 (10.3%).

Conclusion: Most of rheumatic diseases can be diagnosed and followed during one-year period in the resident’s training program. Also, ultrasonography and injection skills are granted during this time. The most important self-reported deficiency is the low number of patients under biological/targeted synthetic treatments.

REFERENCES:

Disclose of Interests: None declared


Educational cases

AB1508

A NOVEL PATHOGENIC VARIANT IN ZNF462 GENE ASSOCIATED WITH WEISS-KRUSZKA SYNDROME AND SLE

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Background: Weiss-Kruszka syndrome (WSKA) is an autosomal dominant congenital anomaly syndrome due to mutations in the ZNF462 gene and manifests with developmental delay and multiple craniofacial abnormalities with variable expressivity. It is also characterized by cognitive impairment, whilst about a third of the affected individuals belong to the autism spectrum. Although the disease is inherited with the autosomal dominant manner (most of the described subjects 95%) had de novo variants with no affected family members1. WSKA has been recently described and only 26 (including our patient) affected individuals have been classified to date2. Systemic lupus erythematosus (SLE) is a systemic autoimmune disease characterized by the presence of autoantibodies and multi-organ inflammation. Genetic factors might play an important role in disease pathogenesis in patients with childhood-onset SLE.

None declared

Disclosure of Interests: None declared

Objectives: To describe the case of a SLE patient who was found to have WSKA related to a novel pathogenic autosomal dominant (AD) variant in the ZNF462 gene, and inform clinicians of a possible association between the 2 conditions.

Methods: A 25-year-old Caucasian female with a history of SLE, diagnosed at the age of 14, manifested with malar rash, Raynaud’s phenomenon, arthritis, thrombocytopenia, positive ANA, ds-DNA antibody and hypocomplementemia. At the age of 17, she developed renal dysfunction due to lupus nephritis class IV, and she was treated with glucocorticoids (GCs), cyclophosphamide and hydroxychloroquine. In Oct 2020, she was admitted to the hospital for thrombotic thrombocytopenic purpura due to lupus exacerbation. She was successfully treated with GCs, plasma exchange and rituximab. During her hospitalization, the presence of various clinical features raised the suspicion of a genetic syndrome. First, the patient exhibited dysmorphic features such as hypertelorism, prototithism, arched eyebrows, flattened nasal bridge, small upper lip, mild intellectual disability, and a history of childhood-onset SLE. Whole exome sequencing (WES) by NGS was used for the genetic investigation of the patient.

Results: Figure 1.

Methods:

Results: The patient underwent genetic evaluation with WES and the novel heterozygous AD pathogenic variant c.4142delT (p lle1381ThrfsTer16) in ZNF462 gene was identified. Confirmation of the identified variant was also verified by Sanger sequencing. Pathogenic variants in the ZNF462 gene were previously described in patients with the recently reported WSKA of which several characteristics are compatible with our patient’s features. The ZFN462 encodes a zinc-finger transcription factor that plays a role in embryonic development, transcriptional regulation, and chromatin remodelling. Given that chromatin remodelling has been implicated in the pathogenesis of SLE, the association of this novel ZNF462 variant in the development of SLE, needs to be determined.

Conclusion: This is the first report of a patient with coexisting SLE and WSKA due to a novel variant. It illustrates the need for further research in order to elucidate any possible pathophysiologic link among the 2 conditions.

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OVARIAN CARCINOMA MIMICKING SYSTEMIC SMALL VESSEL VASCULITIS: CASE REPORT

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Background: Few cases of digital ischemia and gangrene associated with primary solid tumors have been described in literature[3]. The exact mechanism of severe occurrence has not been completely understood and the available treatment options have an extremely limited utility [1,2]. In the most cases the patients were elderly women with adenocarcinomas of digestive or gynaecologic apparatuses [4].

Objectives: We describe a new case of digital gangrene as unusual presentation of ovarian cancer in a 36 years old woman.

Methods: A 36 years old female was admitted to our Reumautology department with blackish Blackish discoloration of the toes of one week duration. She had history of COVID -19 infection 8 months prior to the presentation then developed hemoptysis, picture suggestive of ILD, generalized anasarca and skin rash; accordingly an initial diagnosis of post COVID-19 vasculitis was made by dermatologist. The blood tests were ESR:21 mm/hr, CRP:25.7, D.Dimer:8.8, Ferritin:575 ng/ml, lymphopenia:0.9, S.Creatinine:2, 24h urinary protein: 325 mg/24h and all autoimmune markers were negative except anti nuclear antibody (ANA) with titer:1/160. Further assessment revealed that she had multiple site coagulopathy; internal jugular vein thrombosis, bilateral lower limbs Deep Venous Thrombosis (DVT). Neck ultrasound surprisingly showed bilateral enlarged suspicious looking supraclavicular lymph node with lost hilum which was Biopsied for histopathological correlation which revealed focally necrotizing adenocarcinoma with significant signet ring differentiation. Searching for the primary malignancy, tumor markers were sent CA125: 584 u/ml (up to 35), Pelvi-abdominal Magnetic resonance imaging (MRI) revealed Left ovarian mass measuring 3.6 x 3.4 x 4.4 cm highly suspicious of malignant neoplastic growth for histopathological correlation, Suspicious looking pelvi-abdominal lymph nodes mostly representing malignant lymphadenopathy, Scattered peritoneal nodules suspicious of metastatic deposits.

Results: During admission the patient received full dose anticoagulation (Low Molecular Weight Heparin: 60 iu /12h). Upon diagnosis we arrange the transferal to the oncology department to continue her management plan. Unfortunately; the case was terminal for palliative therapy and she died after 2 weeks.

Conclusion: Bluish discoloration of digits and toes may be a clue for diagnosis of many diseases not only vasculitis. Malignancy can disturb the immune system in a way that mimic any systemic connective tissue disease. Acute insult aggressive multiple site deep venous thrombosis (DVT) necessitate thinking outside the box and consider other causes of coagulopathy like visceral malignancy.

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