Disclosure of Interests: None declared


AB1103  NEUROLOGICAL IMPAIRMENT DURING SARCOIDOSIS

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Background:
Objectives: To describe neurological impairment characteristics in sarcoidosis.

Methods: This was a descriptive and retrospective study including 65 patients with sarcoidosis, followed in the departments of internal medicine and neurology at the Military Hospital of Tunis over a period of 20 years from 1997 to 2017.

Results: A total of 65 patient files have been selected, of which 38 have neurological involvement. Thirty-eight patients met the inclusion criteria for Neurosarcoïdosis. According to Zajicek’s criteria, the diagnosis of Neurosarcoidosis was certain in 2 cases, probable in 18 cases and possible in 18 cases. Neurological disorders were symptomatic in 58.5% of the studied population. Neurological signs were inaugural in 9 patients (14% of cases). A central neurological involvement was demonstrated in 33 patients (86.8%), the peripheral nervous system was affected in 5 patients (13.1%), and cranial nerve involvement was found in 10 patients (26.3%). Ten patients have had both central and peripheral impairment. Neurological involvement was significantly associated with cardiac, renal extra-thoracic, ophthalmologic, articular and cutaneous involvement (p <0.05).

Conclusion: Neurological impairment was frequently observed in our series. It was also significantly associated with multisvircular involvement without particular genetic pattern.

Disclosure of Interests: None declared


AB1105  A NOVEL AUTOINFLAMMATORY AND LYMPHOPROLIFERATIVE SYNDROME ASSOCIATED WITH PIM1 MUTATIONS

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Background: Whole exome sequencing can allow genetic diagnosis in subjects with long lasting clinical stories not supporting any well-defined disorder. A 35-year-old man was referred to ophthalmologist’s evaluation for blurry vision in his left eye. The fundus examination showed choroidal lesions in both eyes. His past medical history was relevant for celiac disease, recurrent episodes of fever and skin rashes with leukocytoclastic vasculitis, inflammatory lesions of the osteoarticular and muscular system, one episode of aseptic meningitis, an intracranial granuloma and two episodes of anterior uveitis. He had also splenomegaly with non-caseating granulomas. Brain TC found multiple lytic and sclerotic skull lesions. He was diagnosed with atypical sarcoidosis and treated with oral steroid and methotrexate.

Objectives: To describe functional and genetic data supporting the role of a PIM1 mutation in the multisystemic inflammation and lymphoproliferation of the patient.

Methods: Whole exome sequencing (WES) analysis. Flow-cytometry to evaluate Pim1 expression. Bad phosphorylation (target of Pim1 kinase) and the effect of Pim1 inhibitor on peripheral blood mononuclear cell (PBMC) viability. RNAseq was on primary fibroblasts from the patient and