Background Patients with demyelination may manifest features of autoimmune rheumatic diseases. We evaluated patients presenting with a demyelinating syndrome and clinical or autoantibody evidence for an underlying connective tissue disease (CTD).

Methods Patients with clinical and/or imaging evidence of demyelination referred to a rheumatology unit. Diagnoses of multiple sclerosis (MS) and systemic lupus erythematosus (SLE) were made according to McDonald and SLICC classification criteria, respectively. Patients with features of CTD not fulfilling criteria for a specific disease were labelled as demyelination with autoimmune features (DAF). Demographics, clinical/serological imaging/data and treatments were recorded at every visit, following multidisciplinary evaluation (neurology, rheumatology, neuroimaging).

Results Sixty-five patients (n=65) were included in the study [93.8% female, mean (SD) age at first demyelinating episode 37.3 (11.8) and median (IQR) duration to last follow-up 4 (7) years]. Rheumatologic clinical manifestations and autoantibodies of all patients are summarized in table 1. Fifty-two patients had lesions in the brain (80%), 32 in the spinal cord (49.2%) and 5 in the brainstem (7.7%), while 17 developed optic neuritis (26.2%). Among the 65 patients, at last follow-up, 32 patients (49.2%) had fulfilled diagnostic criteria for MS with 11 patients (34.4) diagnosed as overlap between SLE and MS and 16 patients (50%) had CTD features not fulfilling criteria for any known CTD. Of patients with demyelinating syndrome not fulfilling criteria for MS (n=33), 7 patients (21.2%) had SLE while 22 patients (66.7%) were classified as DAF. Most common features of DAF patients were ANA, arthritis and mucocutaneous features of SLE.

Conclusions Among patients with demyelination and features of CTD a significant number of patients do not fulfill criteria for either MS or SLE. These patients exhibit lupus-like autoimmune features and may represent a distinct group of patients.