HLA ASSOCIATION IN CIDP SPECTRUM NEUROPATHIES

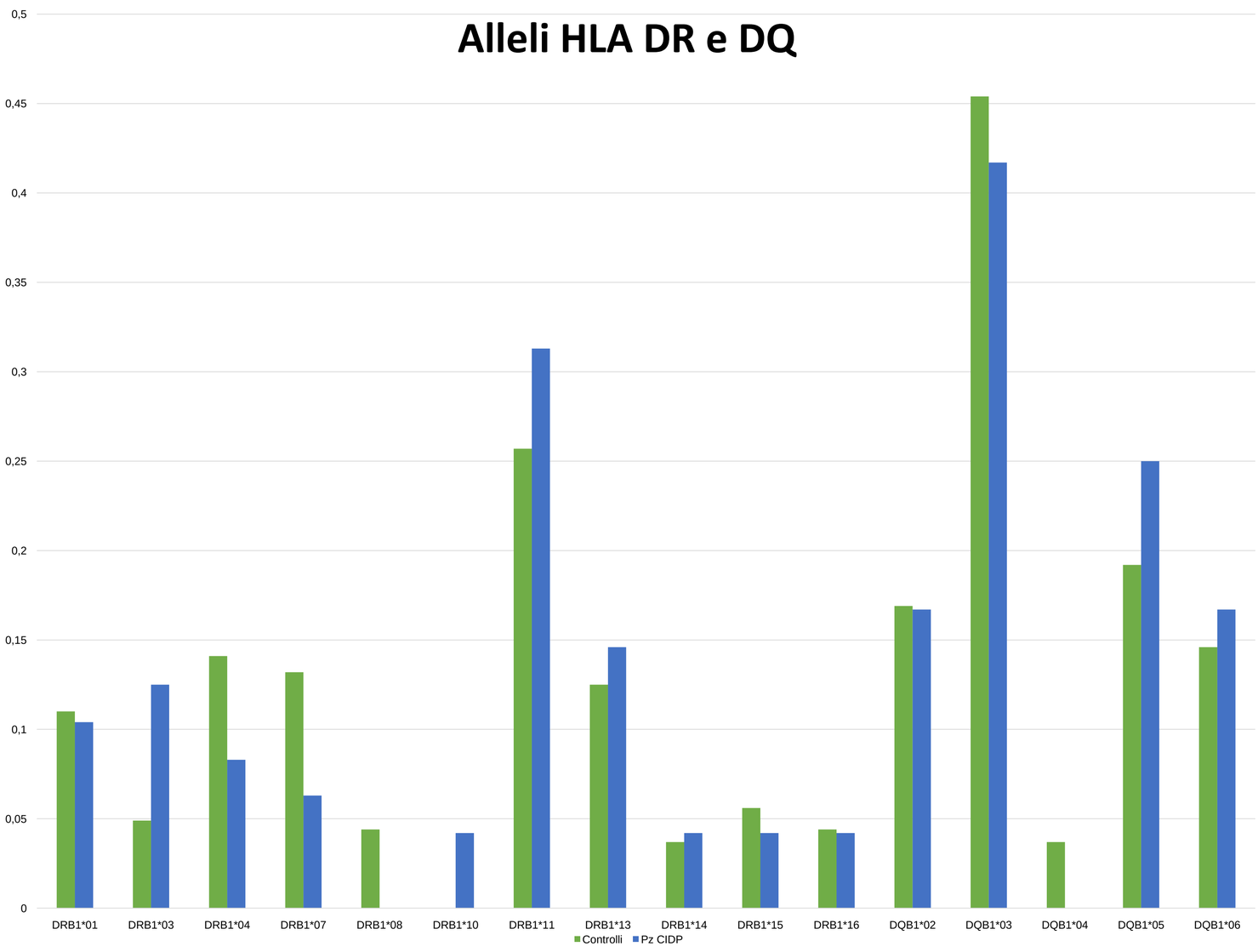
F. Caria[1] , S. Cotti Piccinelli[2], M. Marchesi[2], M. Frassi[3], G. Carella[4], S. Gallo Cassarino[1], S. Rota[1], A. Padovani[1], M. Filosto[1]

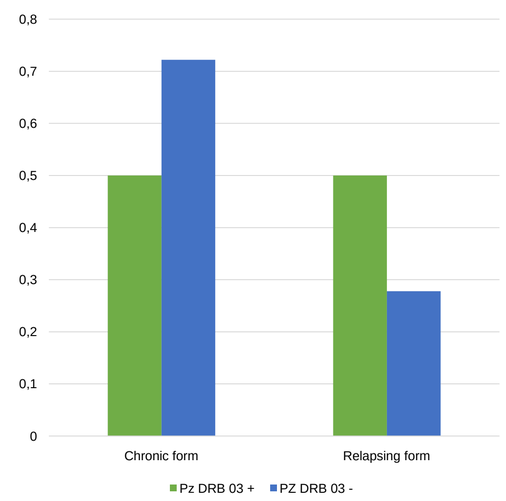
[1] Department of Clinical and Experimental Sciences, Neurology Clinic, University of Brescia (Brescia);
[2] Center for Neuromuscular Diseases, Unit of Neurology, ASST "Spedali Civili" and University of Brescia (Brescia);
[3] Unit of Rheumatology and Clinical Immunology, ASST "Spedali Civili" and University of Brescia (Brescia);
[4] Histocompatibility Laboratory, Immunology and Transfusion Medicine Service SIMT, ASST "Spedali Civili" (Brescia)

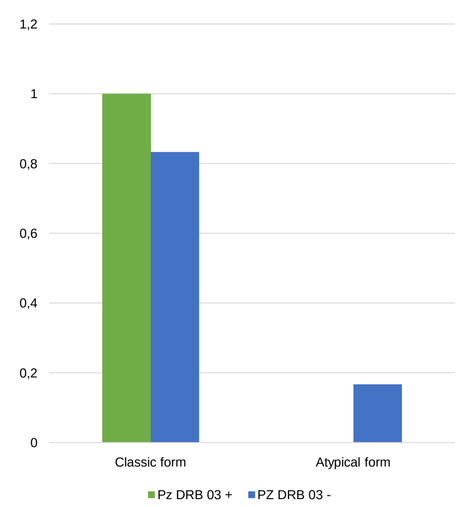
European Reference Network for rare or low prevalence complex diseases
Network Neuromuscular Diseases (ERN EURO-NMD)
Member Spedali Civili di Brescia — Italia

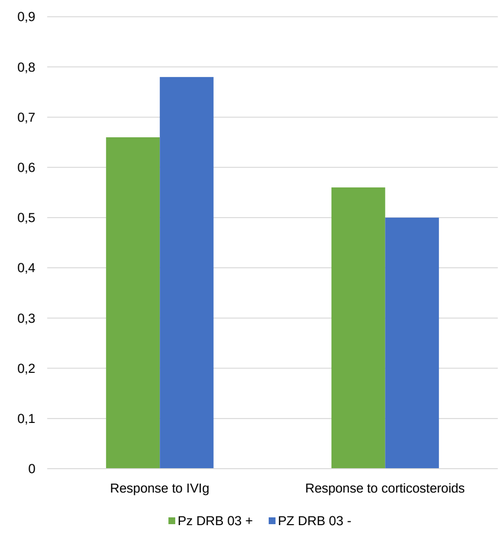
OBJECTIVES: To describe human leukocyte antigen (HLA) haplotype association in an italian cohort of chronic inflammatory demyelinating polyradiculoneuropathy (CIDP) patients and to analyze relation with clinical presentations and response to therapy.

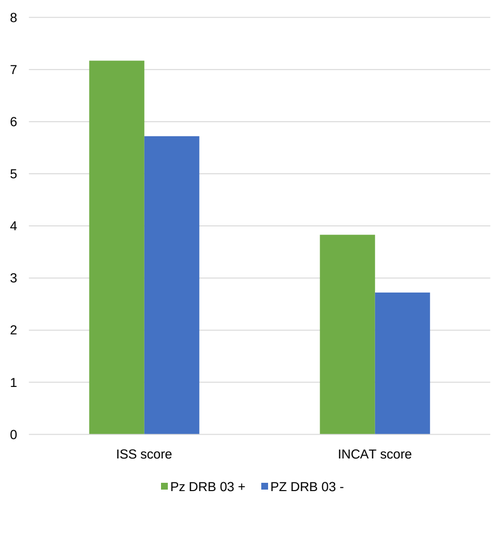
PATIENTS AND METHODS: Lymphocitic expression of HLA-DR and DQ haplotypes of 24 CIDP patients was analyzed by using PCR-SSP. Twenty-one patients had classical form, 2 had pure sensory CIDP, 1 had DADS. Sixteen patients had a chronic form, while 8 presented a relapsing course. Two hundred sixteen healthy samples from Brescia Bone Marrow Donor Registry were random selected as the control group. The results were related to the clinical presentation, course and response to therapy. For each patient INCAT score, ISS score, MRC sum score and Quality of Life score were calculated.











RESULTS: HLA-DR3 and DR3/DQ2 are more significantly frequent in the cohort of patients with CIDP (13% and 10% respectively) than control group (4% and 3% respectively), $p<0,05$. Specifically, six classic CIDP patients were DR3 positive. Out of these, five were DR3/DQ2 positive. The other 15 classic patients and the three non classic patients were DR3 and DR3/DQ2 negative. The DR3 and DR3/DQ2 positive patients present with a more frequent relapsing course, a worse response to I.V. Ig, a more frequent upper limb distal sensory and cranial nerve involvement and higher ISS and INCAT scores than DR3 negative patients.

DISCUSSION: CIDP clinical spectrum encompasses a classical CIDP form and several CIDP variants. The causes of different clinical expression and variable response to treatments are scarcely known. Specific HLA associations are common in dysimmune conditions and the presence of an HLA association is a criterium for diagnosing some autoimmune diseases. In CIDP, few studies reported no associations or HLA-DR13/DQ6 association in some populations. To date, a clear confirmed HLA association for CIDP is lacking.
In this study, we found an association between HLA-DR3 and DR3/DQ2 and a subset of CIDP patients which seems to present a more frequent relapsing course and more severe disease.

CONCLUSIONS: Although our sample is too small for a definitive evaluation, our results may support a possible role for HLA haplotypes in modulating CIDP clinical features and deserve further studies on a greater number of patients.