



Poster Group 3 - Case Reports III

1476 - Celiac disease in common variable immunodeficiency patient

Malkusova, I¹; Zeman, P²; Freiburger, T³

¹Medical Faculty Hospital, Allergology, Clin. Immunology, Plzen, Czech Republic; ²Mulac Hospital, Gastroenterology, Plzen, Czech Republic; ³Center for Cardiovascular Surgery and Transplantation, Molecular Genetics Laboratory, Brno, Czech Republic

42-year old truck driver was checked up by gastroenterologist: his sister and her daughter are treated for celiac disease (CD). Familial prevalence of CD is approximately 10% in first degree relatives. His mother died after an accident, his father and grandfather died of urinary tract carcinoma. Father's brother died at 12 years of age of wasting. Patient has 2 children, without any symptoms associated with CD by now. He gave a history of recurrent upper and lower airway infections, sinusitis and otitis media after tonsillectomy in childhood. In 2004 he was treated for pneumonia. During last years he had several episodes of diarrhea. The physical examination revealed a normal-nourished man without visible abnormalities. He underwent upper gastrointestinal endoscopy: histological assessment of duodenal specimens revealed the presence of villous atrophy and feature of nodular lymphoid hyperplasia. Celiac disease was determined. Blood tests revealed slight elevation of ALP (3.27 μ kat/l) and C-reactive protein (CRP) (50 mg/l). Erythrocyte sedimentation rate (ESR) (35 mm/h), number of leucocytes (17.5.10⁹/l) and platelets (455.10⁹/l) were elevated. Antinuclear antigen, anti-gliadin, anti-endomysial and anti-tissue transglutaminase antibodies were negative. The reason was depression of all immunoglobulin classes: IgG 0.51, IgA <0.06 and IgM 0.20 g/l. Blood sample obtained to determine CD lymphocyte profile evaluated significantly depression of CD4+T-lymphocytes (16.0%), number of CD 19+B-lymphocytes was in normal range (15.0%). Secondary hypogammaglobulinemia was excluded and CVID (common variable immunodeficiency) diagnosis was performed. By HLA typing predisposition to develop celiac disease was found: HLA-DQA1*0501-DQB1*02 genes carry about 95% of the celiac population. Mutation in TAC1 (transmembrane activator and CAML interactor) carried in only 8% of CVID patients did not found. Partial improvement after gluten-free diet by follow-up endoscopy was shown. The laboratory results revealed decrease in CRP (10.6 mg/l), number of leucocytes (11.0.10⁹/l) and platelets (431.10⁹/l) and increase number of CD4+T-lymphocytes (25.0%). Immunoglobulin substitution therapy (IVIG) was recommended but postponed till next histological assessment (after 6 months on gluten-free diet). Examination of first degree relatives of a celiac even if there are no obvious gastrointestinal symptoms are recommended, the damage done by undiagnosed disease can be reduced.

