

ROLE OF ADAPTOR PROTEIN SH2D1A IN THE REGULATION OF IMMUNE SYSTEM. DEMONSTRATION ON X LINKED LYMPHOPROLIFERATIVE SYNDROME

E. Mejstříková, D. Sumerauer, T. Freiburger, H. Bučková, J. Starý, Z. Sedláček, O. Hrušák, A. Janda

Thursday, 24 November 2011, 13.45–16.15, Hall A

D1.3 ELECTRONIC TEACHING IN STUDY PROGRAMMES WITH EXTENDED TRAINING IN PEDIATRY

X linked lymphoproliferative disease (XLP) caused by mutation in SH2D1A gene is a rare genetic disorder historically first described in patients with fatal infectious mononucleosis. Later on other disease manifestations were identified in patients with XLP not always associated with EBV infection: Burkitt lymphoma, common variable immunodeficiency, aplastic anemia, pure red cell aplasia, lymphoid vasculitis. Recently in patient with juvenile rheumatoid arthritis SH2D1A mutation was identified. SH2D1A gene is a member of SAP protein family and interferes with the signalling pathways in T lymphocytes, NK and NKT cells. SH2D1A is expressed primarily in lymphocytes, specifically T, NK, and NKT cells, as well as in eosinophils and platelets. Role of SH2D1A family will be described on a recently diagnosed family, where completely different disease manifestations in three affected males were observed (fatal infectious mononucleosis, Burkitt lymphoma followed by aplastic anemia and severe unusual skin affliction so far not described in the literature). With broader availability of molecular genetic testing we expect identification of other less typical disease manifestations. Supported by NS/10480-3, MSM0021620813, MZOFNM2005