Tomas Kozak et al., J Blood Disord Transfus 2017, 8:5 (Suppl)
DOI: 10.4172/2155-9864-C1-028

Systemic mastocytosis (SM): Analysis of diagnostic markers and course of the disease prediction

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We analysed data of 40 patients with SM, focusing on markers that could be sensitive to support the diagnosis of SM and that might distinguish indolent and aggressive course of the disease. We looked at C-KIT mutation detection in both bone marrow and peripheral blood and we ańalyzed the tryptase level. Median age of patients at the time of diagnosis was 53 (25-84) years, 45% of patiens were males. Indolent SM (ISM) was diagnosed in majority of patiens: 29 (72%), aggressive SM (ASM) in 9 (23%) patients and in 2 patients was established SM-AHN (ET and AML). All patiens were treated with long term antiallergic profylaxis with both H1 and H2 blockers, 19 patients started specific first line treatment for SM with interferon- alfa (13) or cladribin (5). Three patiens died, 2 for ASM progression, 1 for SM-AHN (AML). Presence of C-KIT D816V mutation by PCR was analysed in 31 patients in bone marrow (BM) and/or peripheral blood (PB). In 27 patients, the mutation was examined in bone marrow, 22 (81%) of them were positive. In 12 patients, the c-kit mutation was examined in peripheral blood, only 4 of them were positive (33%). The c-kit mutation was analysed in 8 patients in both BM and PB, 5 of them (63%) were positive in BM and negative in PB at the same time. We analysed difference of tryptase level in ISM and compared it to the ASM. Median tryptase level in ISM was 37, 1 (6, 03-200) μg/l, in ASM 200 (58-200) μg/l respectively.

Biography

Tomas Kozak has completed his MD from Charles University in Prague and later Post-doctoral studies from Masaryk University in Brno. He is Professor at the Department of Internal Medicine and Haematology at the 3rd Faculty of Medicine, Charles University in Prague, Czech Republic. He has published more than 90 papers in reputed journals as Author, Co-author or Senior Author and has been serving as an Editorial Board Member of repute.

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