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Fund “Nauka” Project № 11001 Resume

“Chronic PH‘-negative myeloproliferative neoplasms: correlation between JAK2V617F pathological expression, clinical and biological prognostic markers and angiogenesis in the bone marrow”

Project leader: Prof. Liana Gercheva, MD, PhD

JAK2V617F mutation is the most common genetic event established among BCR-ABL negative chronic myeloproliferative neoplasms (CMNs). Many investigations aimed to discover some correlations between mutational burden and clinical manifestations within every disease entity have been accumulated but with very contradictory results. The aim of this work is to analyze correlations between JAK2V617F mutational burden on one side and clinical symptoms, risk score and laboratory deviations on the other. 131 newly diagnosed patients have been prospectively investigated at the hematology clinic in “St. Marina” University hospital, Varna. Correlations between mutational burden and clinical parameters have been followed out separately in each nosological unit. We found out more frequent JAK2V617F mutational burden in patients with primary myelofibrosis (PMF) (70%) compared to literature data. Strong correlation between JAK2V617F mutational burden and the risk score has been found in all patients. The level of JAK2V617F mutational burden correlates firmly with constitutional symptoms in all diagnostic groups with the strongest relation in homozygote patients with polycythemia vera (PV) and primary myelofibrosis (PMF). Marked leukocytosis has been determined in patients with PMF and essential thrombocythemia (ET) who are JAK2V617F mutation carriers. The marked symptoms in all patients with JAK2V617F mutation in all diagnostic groups rise the question for expansion of the indications for JAK2 inhibitors therapy.

Scientific publications:

Clinical Hematology, L 2014, № 1-2
Journal of the Bulgarian Medical Society of Hematology L 2014, № 1-2
Journal of IMAB - Annual Proceeding (Scientific Papers) 2014, vol. 20, issue 4
http://dx.doi.org/10.5272/jimab.2014204.526