IDENTIFICATION OF MPL-W515L MUTATION IN THROMBOPOIETIN RECEPTOR -*COULD BE MPL-W515L MUTATION AN ADDITIONAL VASCULAR „RISK FACTOR" IN WOMAN DIAGNOSED WITH ESSENTIAL THROMBOCYTHEMIA?*-

Éva Pósfai[1], Imelda Marton[1], Balázs Kotosz[3], Márta Széll,[2] Zsuzsanna László[2] , Zita Borbényi[1]

[1]*2nd Department of Medicine and Cardiology Center ,Albert Szent-Györgyi Clinical Center - University of Szeged, Korányi fasor 6. , Szeged H-6720, Hungary evaposfay@gmail.com*
[2]*Institute of Medical Genetics, Albert Szent-Györgyi Clinical Center - University of Szeged, Somogyi B. u. 4 Szeged H-6720*
[3]*Institute of Economics and Rural Development, University of Szeged, Mars tér 7. Szeged H-6724*

Background: Essential thrombocythemia (ET) is a clonal BCR-ABL1-negative myeloproliferative neoplasm (MPN). Life expectancy of ET patients is strongly affected by thrombotic events. Investigation of risk factors of thrombotic events in ET women should be important, since changes in their lifetime conditions such as pregnancy or climacterium could have an additional effect on the relatively frequent occurrence of vascular complications. 30–40% of ET patients are JAK2 V617F mutation negative, thus, further mutation analysis could be important. [1] Our aim was to evaluate the frequency of acquired MPL W515L mutation, in JAK2 V617F-negative ET woman patients and to answer the question whether the MPL-W515L mutation has an additional role as a vascular *„risk factor"* in ET women?

Patients and methods: Between 1999 and 2011, 96 patients with essential thrombocythemia were selected randomly. Among them 27 JAK2 V617F-negative female ET patients could be found with the mean age of 55.5 years [range: 14–95]. DNA was isolated from EDTA stabilized peripheral blood samples, and screened for the mutation by allele-specific PCR reactions and subsequent agarose gel electrophoresis. The method has 1% to 5% sensitivity in terms of allele frequency.

Results: The MPL W515L mutation could be detected in 16 patients. Mann-Whitney tests, and multivariate binary logistic regression was run to estimate the probability of thrombotic events in combination with MPL mutation status, and with other main cardiovascular risk factors. The MPL mutations - although not significantly due to the small sample - showed a correlation with the clinical appearance of the disease, and its possible prognostic value could be detected in our group of patients.

Conclusion: Based on our findings we suppose that ET female patients with cardiovascular risk factors (especially high blood pressure, hyperlipidemia, smoking) may have a higher risk for thrombotic events, and the MPL-W515L mutation could have an additional role in this special group of patients.

References: 1. Tefferi A. Polycythemia vera and essential thrombocythemia: *American Journal of Hematology* 2012;87(3):284-93.