JAK2 46/1 HAPLOTYPE PREDISPOSES TO SPLANCNIC VEIN THROMBOSIS ASSOCIATED BCR-ABL NEGATIVE CLASSIC MYELOPROLIFERATIVE NEOPLASMS


Laura Villani1, Gaetano Bergamaschi2, Massimo Primignani3, Vittorio Rosti1, Adriana Carolei1, Valentina Poletto1, Paolo Catarsi1, Ambra Spolverini4, Alessandro Maria Vannucchi4, Giovanni Barosi1

1Unit of Clinical Epidemiology and Center for the Study of Myelofibrosis, IRCCS Policlinico S. Matteo Foundation, Pavia, Italy
2Department of Internal Medicine, Unit of Clinica Medica 1, IRCCS Policlinico S. Matteo Foundation, Pavia, Italy
3Gastroenterology 3 Unit, IRCCS Ca Granda Ospedale Maggiore Policlinico Foundation, Milano, Italy
4Section of Hematology, Department of Critical Care, University of Florence and Istituto Toscano Tumori, Florence, Italy

SUMMARY:

The germline constitutive JAK2 haplotype, called GGCC or 46/1, is a susceptibility factor for BCR-ABL negative classic myeloproliferative neoplasms (MPNs), although mechanisms linking the haplotype block to the acquired MPN remain unclear. Splanchnic vein thrombosis (SVT) is strongly associated with MPNs, and can occur during the course of an already known MPN or, more often, as the event leading to the diagnosis of MPN. In this study researchers sought to clarify the potential role of 46/1 haplotype in the etiology of SVT. A total of 164 subjects with SVT were enrolled. In 56, a diagnosis of idiopathic SVT was made, while 108 patients were diagnosed as MPN-associated SVT (32 essential thrombocytopenia, 29 fibrotic and 26 pre-fibrotic primary myelofibrosis, and 21 unclassified forms of MPN). Patients with SVT but no MPN had an haplotype frequency similar to control population. Conversely, SVT-associated MPN patients as a whole had an haplotype frequency significantly increased compared to normal population, but this difference was mainly accounted by the presence of the JAK2V617F mutation. In summary, these data suggest that the 46/1 haplotype does not confer susceptibility to SVT, but it is a susceptibility factor for JAK2V617F positive MPNs associated with SVT.

To read the paper: http://www.ncbi.nlm.nih.gov/pubmed/21890200