P17. JAK2V617F MUTATION IN ESSENTIAL THROMBOCYTHEMIA.

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Essential thrombocythemia (ET) is a clonal myeloproliferative disease involving a hematopoietic stem cell and, manifesting predominantly as thrombocythosis, and it is associated with thrombohemorrhagic complications and myeloid transformation to diseases such as myelofibrosis and acute myeloid leukemia. In 2005 a unique acquired clonal mutation in JAK2 was found. This mutation was observed in the majority of polycythemia vera patients and in about half of ET or primary myelofibrosis patients. We analyzed 104 cases of ET, from a single institution to determine the prevalence of JAK2V617F mutation and the clinical correlations. Mutation screening was performed on genomic DNA from peripheral blood from all 104 patients. The JAK2V617F mutation was found in 52,4% of cases. 7,2% were homozygous for the mutant allele (>75%). Patients with JAK2V617F positive had higher haematocrit, leucocytes levels and advanced age.