CONCOMITANT OCCURRENCE OF BCR-ABL AND JAK2V617F MUTATION


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SUMMARY:

The JAK2 V617F mutation is present in about 95% of patients with polycythemia vera and in 50-60% of patients with essential thrombocythemia and primary myelofibrosis, while the rearrangement BCR / ABL is present in virtually all cases of chronic myeloid leukemia. These two genetic alterations were considered until now mutually exclusive, but are reported in the literature of individual cases of patients with both these mutations. In this paper were analyzed for JAK2 V617F mutation 314 patients with chronic myeloid leukemia, and were thus identified 8 patients with both mutations, representing 2.55% of cases. The predominant phenotype of the disease was that of a typical chronic myeloid leukemia, but this paper suggests that the coexistence of a chronic myeloproliferative disease BCR / ABL negative is possible, and seek it may be indicated especially in those patients who present with atypical features of the disease that could be unmasked by imatinib therapy.

To view the paper: http://www.ncbi.nlm.nih.gov/pubmed/21940831