

## C8. THE RELATIONSHIP BETWEEN FACTOR V LEIDEN, PROTHROMBIN G20210A AND *MTHFR* MUTATIONS AND THE FIRST MAJOR THROMBOTIC EPISODE IN POLYCYTHEMIA VERA AND ESSENTIAL THROMBOCYTHEMIA.

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### **Background/aim**

Arterial and venous thrombosis are the most frequent complications in patients with polycythemia vera and essential thrombocythemia. We sought to demonstrate a possible contribution of the Factor V Leiden, Prothrombin G20210A, and *MTHFR* 677 C>T and 1298 A>C mutations to the thrombotic risk in patients with polycythemia vera and essential thrombocythemia, along with other biological features of these patients.

### **Material and methods**

We included 86 patients with polycythemia vera, of which 34 (39.5%) had major thrombosis, and 95 patients with essential thrombocythemia, of which 22 (23.1%) had major thrombosis. Factor V Leiden, Prothrombin G20210A, and *MTHFR* 677 C>T and 1298 A>C mutations were genotyped by PCR-RFLP and AS-PCR techniques.

### **Results**

In the whole cohort of patients, only the Factor V Leiden mutation was significantly associated with both arterial and venous thrombosis, in univariate and multivariate analysis (OR = 4.3; 95% CI = 1.5 – 12.5; p=0.008 and OR = 4.3; 95% CI = 1.2 – 15.9; p = 0.02, respectively). Other factors significantly associated with thrombosis in both univariate and multivariate analysis were: male sex (OR = 2.8; 95% CI = 1.4 – 5.4; p = 0.002 and OR = 3.5; 95% CI = 1.6 – 7.6; p = 0.002, respectively) and the *JAK2* V617F mutation (OR = 5.5; 95% CI = 2.1 – 15; p = 0.0001 and OR = 6.9; 95% CI = 2.2 – 21.2; p = 0.001, respectively).

### **Conclusions**

In conclusion, among the four mutations analyzed (Factor V Leiden, Prothrombin G20210A, and *MTHFR* 677 C>T and 1298 A>C), only Factor V Leiden is a major contributor to thrombosis in polycythemia vera and essential thrombocythemia.