Detection of somatic JAK2 (V617F) mutation in patients with chronic myeloproliferative diseases

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**Background:** The mutation JAK2 V617F in Chronic Myeloproliferative Disorders (CMPD) has been described as a frequent genetic event in majority of patients with Polycythemia Vera (PV), Essential Thrombocythemia (ET) and Idiopathic Myelofibrosis (IMF).

**Aim:** To study the mutation JAK2V617F gene in different clinical variants Ph(-) negative CMPD.

**Materials & Methods:** Materials for our study was the DNA samples from 70 patients with Ph(-) CMPD (erythremia, essential thrombocytemia and sub-leukemic myelosis) and 14 patients with Ph(+) chronic myeloid leukemia. The control group consisted of 71 healthy donors. Extraction of DNA was conducted from peripheral blood of patients.

**Results:** In the group of patients with BCR-ABL positive chronic myeloid leukemia mutation V617F in the JAK2 gene was not detected in any of the patients (14 surveyed). In 3 (21.4%) patients of this group were identified various chromosomal changes. Out of 70 examined patients, the V617F mutation in the JAK2 gene was detected in 62.9% (44/70), with erythremia in 80.0% (34/40), in essential thrombocytemia– 46.1% (6/13), in the sub-leukemic myelosis – 23.5% (4/17) of cases. Three patients with erythremia had this mutation in the homozygous form. Among main group, 15 patients was carried out cytogenetic analysis of peripheral blood cells. All patients identified normal karyotype.

**Conclusion:** The presence of JAK2 V617F mutation was associated with a higher hemoglobin level (P<0.05), a higher white blood cell count (P<0.01). Thus, our data confirm that mutation V617F in the JAK2 gene are highly specific diagnostic markers for patients with Ph-negative CMPD.

**Notes:**