J Neurol Neurophysiol 2018, Volume 9
DOI : 10.4172/2155-9562-C12-095

31ST Clinical Neuroscience and Neurogenetics Conference: Mobilizing Neurons to Rehabilitate

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

Rozikova M A
Tashkent Medical Academy, Uzbekistan

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 thrombocythemia 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 thrombocythemia– 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.

dr.muborak@mail.ru

Notes: