LITHUANIAN UNIVERSITY OF HEALTH SCIENCES

International Summer School Practice

Associations of IL11 Gene Polymorphisms with Myeloproliferative Neoplasms

Myeloproliferative neoplasms (MPNs), including polycythemia vera (PV), essential thrombocythemia (ET), and primary myelofibrosis (PMF), are clonal hematopoietic diseases associated with acquired mutations in JAK2, CALR, and MPL. Thrombosis is a major complication of MPNs, arising from interactions between abnormal blood cell production, platelet and leukocyte activation, endothelial dysfunction, coagulation, and chronic inflammation [1,2]. However, interindividual differences in thrombotic outcomes remain incompletely explained, suggesting that additional biological and genetic factors may contribute.

Interleukin 11 (IL-11), encoded by IL11, is involved in hematopoietic regulation and tissue responses. Increased IL-11 levels and production by bone marrow stromal cells have been reported in PV [4]. IL-11, together with hepatocyte growth factor, can promote clonal erythroblast growth independently of the JAK2 p.V617F mutation [5]. These findings support investigating IL11 in MPNs, although previous studies have primarily examined cytokine expression and activity rather than inherited genetic variation. This study will assess whether selected IL11 polymorphisms (rs1042506, rs2298885, and rs4252562) are associated with MPN subtype, laboratory parameters, and a history of thrombotic events.

Aim

To investigate associations between IL11 polymorphisms and the clinical and laboratory characteristics of patients with MPNs.

Objectives

1. To determine the allele and genotype frequencies of the IL11 polymorphisms rs1042506, rs2298885, and rs4252562 in patients with MPNs.
2. To assess associations between the selected IL11 polymorphisms and a history of thrombotic events in patients with MPNs.
3. To investigate associations between the selected IL11 polymorphisms and laboratory parameters in patients with MPNs.

Methods

This retrospective study will include up to 150 patients diagnosed with PV, ET, or PMF according to the 2016 WHO diagnostic criteria at the Department of Hematology, Institute of Oncology, Lithuanian University of Health Sciences, Kaunas, Lithuania. Samples and associated clinical data will be coded, with no access to patient-identifying information during the study. EDTA-anticoagulated blood samples stored at -20 °C will be used for DNA isolation. DNA concentration will be measured and adjusted as required. The selected IL11 polymorphisms will be genotyped using TaqMan allelic discrimination assays with real-time PCR. IBM SPSS Statistics will be used to calculate allele and genotype frequencies and assess associations with clinical and laboratory characteristics using appropriate statistical tests.

Expected results

The study will characterize the selected IL11 polymorphisms in patients with MPNs and analyze their potential associations with disease subtype, laboratory parameters, and thrombotic events. The findings may provide preliminary evidence of associations between inherited IL11 variation and clinical heterogeneity in MPNs. Given the sample size and candidate-gene design, the results will be considered hypothesis-generating and will require validation in larger, independent cohorts.