

Abstract

Background/Aims: Thrombophilia, ie an increased predisposition to venous and arterial thrombosis, is a complex disease caused by disorders of platelets and endothelial, among others. Circulating endothelial cells (CEC) and endothelial progenitor cells (EPC) have been described as markers of endothelial damage and dysfunction, respectively as markers of its recovery in many diseases, including thrombotic complications. However, their significance in patients with known thrombophilia has not yet been investigated. Both CEC and EPC represent extremely rare cell populations found in peripheral blood. Therefore, it is essential to use exclusively standardized and sensitive methods for their identification and quantification. The aim of the study was to identify and quantify CEC and EPC in the peripheral blood of patients with congenital thrombophilia and to evaluate their importance as markers of endothelial and platelet activity in context with the risk of thrombosis occurring and recurrence. **Methods:** Analysis of the number of CEC and EPC in the peripheral blood of patients with thrombophilia with or without a history of thrombosis and patients with acute thrombosis was performed by multicolor flow cytometry. The CEC and EPC reference values were determined on a group of healthy controls. Patients with hematological malignancies after high-dose chemotherapy and patients with acute myocardial infarction were used as positive controls. **Results:** CEC and EPC immunophenotypes were determined as CD45 negative to weakly positive, CD34 strongly positive, CD146 positive, CD31 positive and CD133 negative, respectively CD133 positive. Elevated levels of endothelial cell subpopulations were confirmed in a positive patient group. No significant changes in CEC or EPC counts were detected in patients with thrombophilia or in patients with acute thrombosis compared to healthy controls. **Conclusion:** In this work, the optimized method of multicolor flow cytometry allows unambiguous identification and quantification of endothelial cells in the peripheral blood. The results support previous studies showing that elevated CEC levels could serve as an indicator of endothelial damage and dysfunction. Normal CEC and EPC levels were determined in patients with congenital thrombophilia.