

Cerebral venous thrombosis risk factors and JAK2 mutation

Cerebral venous thrombosis is a rare type of cerebrovascular disease. The widespread use of neuroimaging has aided early diagnosis of CVT and its parenchymal complications. Etiology and natural history of CVT differs from DVT mainly due to different pathophysiologic conditions of sinuses.

The main objective of this study was to find out prevalence of genetic and acquired thrombophilic factors and prevalence of JAK2 V617F mutation (typical for myeloproliferative diseases) in CVT patients and discover their relation to prognosis and treatment. Next target of our survey was to identify typical symptoms of CVT and to verify significance of clinical indicators (time to diagnosis, type of therapy, recurrence, recanalization, etc.) and laboratory tests (D-dimers, blood count).

Minority of the data were collected as retrospective (5 patients from a hospital in Pardubice, 18 patients from the University hospital in Hradec Králové). Majority of study was prospective; all patients with CVT prior to the starting point of survey were re-evaluated in outpatient department during the study. Healthy controls were recruited from Transfusion department of the University hospital in Hradec Králové

We enrolled 64 patients and 70 healthy controls. JAK2 mutation was more frequent in patients group than in controls ($p=0.015$). In majority of cases (3/5 at the end of the study) positivity of JAK2 mutation led to development of myeloproliferative disease. Genetic thrombophilic factors were found in 25 % of the cases. Most common acquired thrombophilic risk factor was use of hormonal contraception (59% of women). We found 19% of idiopathic CVTs.

Cephalaea was most common leading symptom (67% of cases). Patients in critical state had shorter time to diagnosis, higher D-dimers (on the edge of statistical significance) and they were more frequently treated with thrombolysis. We identified recanalisation, contraception use and low D-dimers as predictors of good prognosis. Calculated D-dimers sensitivity was 80.6%. D-dimer level was higher at the time of diagnosis and decreases in time. Complete or partial recanalisation was found in 83% of cases and was more frequent in younger patients and in women with hormonal contraception.

Within cooperation in CEVETIS multicentric study (data from 45 patients were sent) we identified personal history of venous thromboembolism, malignity and recent head injury as risk factors of recurrence.

By monitoring all relevant parameters of CVT in time of diagnosis and during follow-up we found several interesting facts for diagnosis, treatment and prognosis of patients.