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Incidence of inherited thrombophilia in Greek patients with cerebral venous thrombosis

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Background

Hereditary thrombophilia has been reported to be present in approximately 30% of all patients with cerebral venous thrombosis (CVT). However, data on the incidence of inherited thrombophilia in Greek CVT patients are scarce.

Materials and methods

We report the results of the diagnostic work-up including a full thrombophilia screening in a consecutive case series of 27 patients (7 males, 20 females, age range 17 - 59 years) with CVT from a Greek tertiary healthcare facility.

Results

Cephalalgia was the leading symptom in 85% of the patients (n=23), focal neurological signs were present in 48% (n=13), and epileptic seizures in 22% (n=6). Multiple thrombosis of cerebral sinus was a common finding in MRI and MRV: Thrombosis of the superior sagittal sinus was found in 78% (n=21), of the transverse sinus in 41% (n=11), the sigmoid sinus in 7% (n=2), of the sinus rectus in 18% (n=5) and of the cavernous sinus in one patients only. Elevated D-dimers were found in 48% (n=13), hyperhomocysteinaemia in 30% (n=8), heterozygous mutation of the MTHFR gene in 44% (n=12) and homozygous MTHFR mutation in 18% (n=5). Other hereditary thrombophilias (e.g. FV-Leiden mutation, n=1, or the prothrombine G20210A mutation, n=2) were found in single cases only.

Conclusions

In this consecutive open case series of Greek patients with CVT, the incidence of inherited thrombophilia was considerably higher than reported from other comparable study populations.