A prothrombin mutation and hormone replacement therapy: a dangerous combination

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Abstract

Introduction: The prothrombin G20210A mutation is the second most common inherited thrombophilia after the factor V Leiden mutation. Transmission is considered to be autosomal dominant. The combination of the mutation and other acquired risk factors such as hormonal contraceptive use increases the risk of cerebral vein thrombosis (CVT).

Case Report: A 41-year-old woman, with a personal history of obesity, hormone replacement therapy for early menopause and depressive syndrome, presented to the emergency department with a new onset of right unilateral headache, intensity of 10/10, associated with prostration, vomiting and increased blood pressure (175/115 mmHg). Brain computed tomography showed "...right temporal-parietal heterogeneous lobar haemorrhagic lesion with surrounding vasogenic oedema...". She also performed an angio-CT and angio-MR which confirmed a diagnostic hypothesis of haemorrhagic vascular lesion secondary to venous sinus thrombosis. She was admitted with diagnosis of haemorrhagic stroke secondary to thrombosis of sigmoid and transverse right sinus. The aetiological investigation identified the existence of heterozygosity for prothrombin mutation (G20210A variant) and hyperfibrinogenaemia (545 mg/dL). The patient was informed about the treatment options and decided to use long-term oral anticoagulation with rivaroxaban.

Conclusion: This case deals with an intracerebral haemorrhage in a young woman. The aetiology of the event was multifactorial, related with the use of hormone therapy, hyperfibrinogenaemia, heterozygosity for prothrombin mutation (G20210A variant) and obesity. The clinical presentation of venous sinus thrombosis is highly variable. Headache is the most frequent symptom, occurring in almost 90% of patients, with or without vomiting, papilledema, and visual complaints.