JAK 2 mutation positive polycythemia vera presenting as internal carotid artery dissection followed by dural sinus thrombosis

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Abstract:

Polycythemia vera is a stem cell disorder, often complicated by thrombotic and hemorrhagic events. We report a case of polycythemia vera in a 60-years-old man which presented with acute ischemic stroke. Computed tomography angiography of head and neck vessels was perfomed which revealed left internal carotid artery dissection. During his hospitalization his neurological status deteriorated and brain MRI, MRA and MRV showed right internal jugular vein, superior sagittal, sigmoid and transverse sinus thrombosis with hemorrhage in left frontal and parietal lobe. Repeated laboratory evaluation showed slightly elevated platelets and hematological consultation was requested. Results of sternal puncture suggested myeloproliferative disorder and JAK2 mutation was positive. Thrombus formation in the dural sinus is extremely rare in PV patients. To best of our knowledge this is the first case describing occurence of acute ischemic stroke, internal carotid artery dissection and dural sinus thrombosis in PV patient. It highlights the need of awareness of the association of PV and cerebrovascular disease and PV should be considered as a part of the differential diagnosis in patient with acute cerebrovascular event of otherwise unknown origin.

Keywords: Polycythemia vera, JAK 2 mutation, dural sinus thrombosis

Sažetak:

JAK2 pozitivna mautacija policitemije vere koja se manifestirala kao disekcija unutarnje karotidne arterija praćena trombozom duralnih sinusa.

Policitemija vera je mijeloproliferativa bolest koju karakterizira povećana sklonost trombozama i krvarenjima. U ovom radu prikazujemo 60- godišnjeg muškarca u kojeg se bolest prvotno prezentirala akutnim ishemijskim moždanim udarom. Učinjena je opširna obrada koja je uključivala kompjutoriziranu tomografsku *angiografiju* mozga i vrata kojom se verificirala disekcija lijeva unutranje karotidne arterije. Tijekom hospitalizacije dolazi do pogoršanja bolesnikova neurološkog statusa te je učinjen MR, MRA i MRV mozga kojima se opisuje tromboza desne unutarnje jugularne vene, superiornog sagitalnog, sigmoidnog i transverzalnog sinusa te krvarenje u lijevom frontalnom i parijetalnom režnju. Laboratorijskom obradom utvrđen je blago povišen broj trombocita te je učinjena daljnja hematološka obrada. Učinjena sternalna punkcija je upućivala na mijeloproliferativnu bolest te je pristigla pozitivna JAK 2 mutacija. Tromboza duralnih sinusa je iznimno rijetka u bolesnika s PV. Prema našim dosadašnjim saznanjima ovo je prvi slučaj u kojem je prisutan akutni ishemijski moždani udar, disekcija

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unutarnje karotidne arterije i tromboza duralnih sinusa u bolesnika s PV. Izradom ovog rada željeli smo naglasiti važnost povezanosti PV-a i cerebrovaskularnih bolesti te potrebu o razmatranju PV-a kao jedne od diferencijalnih dijagnoza u bolesnika s akutnim moždanim događajem nepoznate etiologije.

Ključne riječi: Policitemija vera, JAK 2 mutacija, tromboza duralnih sinusa

Dear Editor,

Polycythemia vera (PV) is a stem cell disorder, often complicated by thrombotic and hemorrhagic events (1). In most patients with PV and other myeloproliferative diseases (MPDs) mutation in *JAK2* is found (1). Thromboembolic events and cardiovascular disease are the major cause of morbidity and mortality in this population (1,2). Vascular complication such as arterial or venous thrombosis often leads to the diagnosis of PV (1,2). Acute stroke is the important cause of death and of long-term disability (3). Patients with PV are a unique subset of stroke patients, both for the pathophysiology and for management. Cerebral infarction and transient ischemic attacks are the most common neurological manifestation of PV, while cerebral hemorrhage, extra/intracranial dissection and dural sinus thrombosis have been rarely reported (4,5).

A 60-year-old right handed male was referred to our Department after being treated in local hospital for acute ischemic stroke presenting with mild right-sided hemiparesis, aphasia and right supranuclear facial palsy. His medical history included atrial fibrillation and was taking warfarin. Diagnostic work up showed acute ischemic lesion in left frontoparietal region as well as diffuse white matter lesions consistent with small vessel disease on brain magnetic resonance imaging (MRI). Computed tomography angiography (CTA) of head and neck vessels revealed left internal carotid artery dissection (ICAD). Thorough immunological work up was done with negative results and patient was switched to rivaroxaban. Suddenly, patient neurological status deteriorated and brain MRI, MRA and MRV were performed revealing right internal jugular vein, superior sagittal, sigmoid and transverse sinus thrombosis with hemorrhage in left frontal and parietal lobe.
Extensive diagnostic work up was performed including screening for occult malignant disease. Repeated laboratory evaluation showed slightly elevated platelets and hematological consultation was requested. Results of sternal puncture suggested MPD and JAK2 mutation was positive.

Patient was reffered to outpatient neurological and hematological follow up and was treated with venepunction, hydroxycarbamide and low molecular weight heparin which was eventually switched to dabigatran. His neurological symptoms improved with only discrete right hemiparesis on last outpatient visit.

We argue that PV is associated with endothelial dysfunction that may predispose to arterial disease and ICAD by promoting thrombosis, leucocyte adhesion, inflammation and proliferation of smooth muscle cells in the arterial wall (6). Thrombus formation in the dural sinus is extremely rare in PV patients. To best of our knowledge this is the first case describing co-occurence of both in PV patient. It highlights the need of awareness of the association of PV and cerebrovascular disease and should be considered as a part of the differential diagnosis in patient with acute cerebrovascular event of otherwise unknown origin.

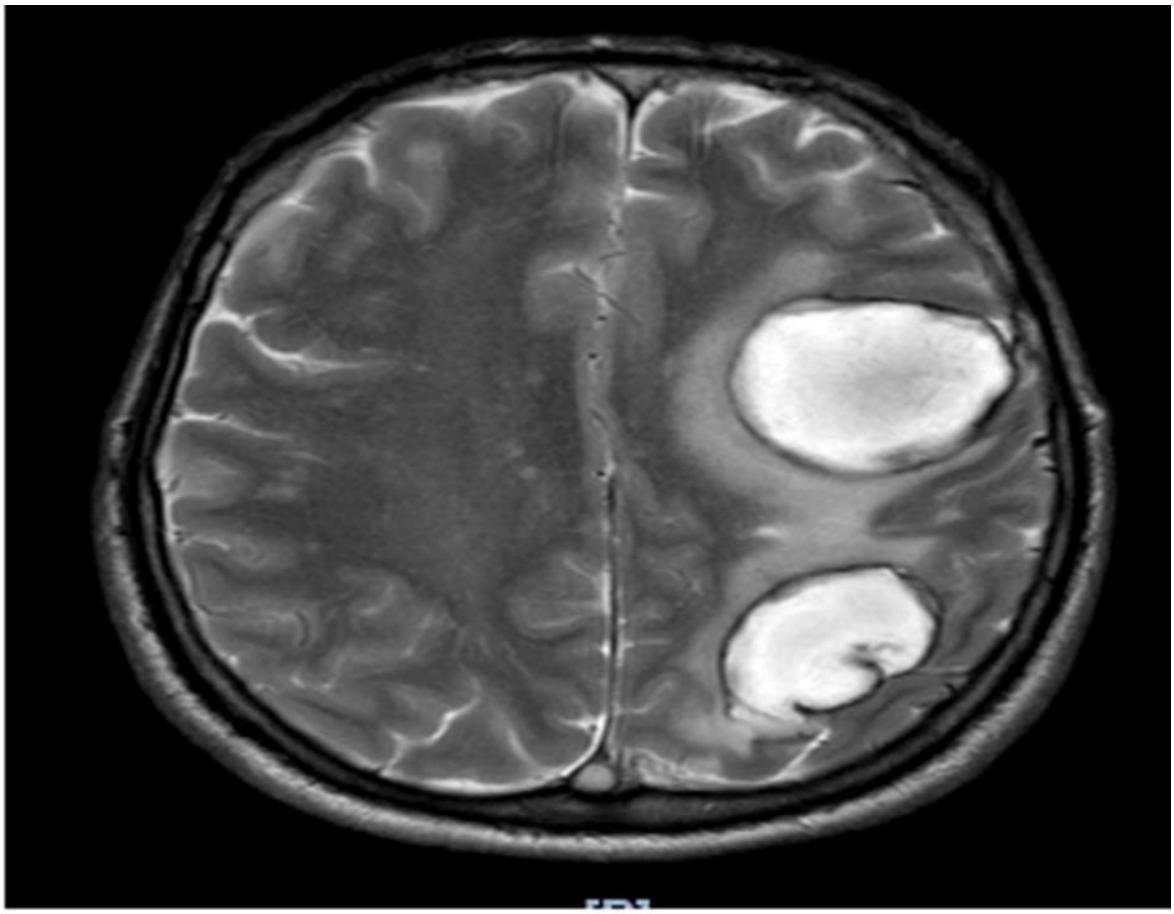

Figure 1: MRI showing two atypical subacute intracerebral hematoma in the left frontal and in the left parietal lobe

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