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JAK2-mutated essential thrombocythemia-associated chorea: a case report

Alessandra Calculli[1], S. Arceri[2], M. Porcino[1], E. Rigoni[2], A. Pisani[1,2]

[1]Department of Brain and Behavioral Sciences, University of Pavia, Pavia, Italy
[2]IRCCS Mondino Foundation, Pavia, Italy

Introduction: The variant $JAK2^{V617F}$ is the most common somatic mutation associated with myeloproliferative disorders, such as polycythemia vera, essential thrombocythemia and primary myelofibrosis. While chorea is a well-known neurological complication of polycythemia vera, to date, only two cases of chorea associated with $JAK2^{V617F}$-positive essential thrombocythemia have been described. In addition, its pathophysiology remains unclear.

Objective: To report a case of $JAK2^{V617F}$-positive essential thrombocythemia-related chorea.

Methods: A 61-year-old woman was admitted to our department with a three-months history of slowly progressive chorea involving oromandibular district, left upper limb and ipsilateral foot and toes. Five years before, the patient was diagnosed with $JAK2^{V617F}$-mutation essential thrombocytemia. She was treated with hydroxyurea 500mg OD. Of interest, this treatment was interrupted right before the neurological symptoms onset. At the admission, blood cell count showed platelet 816×10^3/mL, brain MRI showed chronic vascular infarction in the right caudate nucleus, with no evidence of acute lesions. Hydrossyurea was reintroduced increasing the dosage up to 1000mg OD, together with tetrabenazine, 12.5mg twice daily, with clinical improvement. At follow-up, nine months later, her neurological examination was substantially improved, and platelet count normalized.

Results: Autoantibodies panel including ENA, ANA, ANCA, anti-gliadin, anti-cardiolipin antibodies, anti-beta-2-glycoprotein and lupus anticoagulant resulted negative. EEG detected slow-wave activity of non-specific meaning. Brain MRI showed right caudate nucleus ischemic chronic lesion.

Conclusions: Current literature suggests that hyperviscosity and venous stasis may alter metabolic turnover of neurotransmitters in the basal ganglia, (i.e. dopamine and serotonin) thereby causing adaptive changes in local receptor expression. In our case, we hypothesize that withdrawal of hydroxyurea determined the increase in platelet count, blood viscosity and the consequent caudate infarction. However, the subacute presentation of chorea, its gradual progression, together with the impossibility to establish a temporal correlation with the ischemic lesion, rather suggest a re-arrangement at circuit level, causing the symptom onset.