Essential thrombocythemia: Rare cause of chorea

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Abstract

Essential thrombocythemia (ET) is a clonal myeloproliferative disorder (MPD), characterized predominantly by a markedly elevated platelet count without known cause. It is rare hematological disorder. In ET clinical picture is dominated by a predisposition to vascular occlusive events and hemorrhages. Headache, transient ischemic attack, stroke, visual disturbances and light headedness are some of the neurological manifestations of ET. Here, we describe a 55 year-old female who presented to us with generalized chorea. On evaluation, she was found to have thrombocytosis. After ruling out the secondary causes of thrombocytosis and other MPD we confirmed diagnosis of ET in her by bone marrow studies. Polycythemia vera (PV) another MPD closely related to ET may be present with generalized chorea. There are few case reports of PV presenting as chorea in the literature, but none with ET. We report the first case of ET presenting as generalized chorea.

Key Words

Chorea, essential thrombocythemia, polycythemia vera

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Introduction

Essential thrombocythemia (ET) is a clonal myeloproliferative disorder (MPD), characterized predominantly by a markedly elevated platelet count without known cause. It is a rare hematological disorder. Here, we report a case of 55-year-old female who presented to us with chorea. On evaluation, she was found to have ET. ET is a rare treatable cause of chorea and we report the first case of ET presenting as generalized chorea.

Case Report

A 55-year-old female patient presented to us with an acute onset of involuntary movements involving limbs, face and tongue. These movements were random and fleeting from one part of the body to another causing dysarthria and social embarrassment. There was no history of headache, visual disturbances or transient ischemic attack (TIA). There was no history of recent exposure to anti psychotic's medication. There was no family history of chorea or dementia and no history of rheumatic fever. On examination, patient had generalized chorieform movements involving limbs face and tongue. There was no extra pyramidal or pyramidal sign. Higher mental functions were normal with mini mental score of 28. There was no facial plethora or splenomegaly.

On investigation, she had total white blood cell count 14.2 × 10^3/uL, total red blood cell count 5.07 × 10^6/uL, platelet count 1092 × 10^3/uL, hemoglobin 15.1 g/dL, hematocrit 46.3%, mean corpuscular volume 91.3 fL, mean corpuscular hemoglobin 29.8 PG, mean corpuscular hemoglobin concentration 32.7 gm/dL and erythrocyte sedimentation rate 01 mm. The peripheral blood smear revealed no abnormal cell or acanthocytes. Neutrophil alkaline phosphatase was 240, ceruloplasmin was normal and bcr-abl gene expression was negative. Janus kinase 2(JAK2) V617F mutation was positive. Bone marrow showed hyper cellular marrow with a marked increase in megakaryocytes without any atypical blast cells. Magnetic resonance imaging (MRI) brain was normal and the ultrasound sonography abdomen did not reveal organomegaly. Renal, hepatic and other metabolic profiles were normal. Anti-nuclear antibody and anti-double-stranded deoxyribonucleic acid were negative. The chest X-ray and echocardiogram were normal. Hence, a diagnosis of ET was made and she was started on haloperidol initially to control chorea symptomatically. She showed only some improvement, but her platelet count was rising further and was started on hydroxyurea. On 3rd month of follow-up her chorea completely subsided, haloperidol was slowly withdrawn and her platelet count was 562 × 10^3/uL.

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Discussion

ET is an acquired MPD characterized by a sustained elevation of platelet number with a tendency for thrombosis and hemorrhage. The prevalence in the general population is approximately 30/100,000.[1] The median age for diagnosis is 65-70 years, but the disease may occur at any age. The female to male ratio is about 2:1. The clinical picture is dominated by a predisposition to vascular occlusive events and hemorrhages. Arterial and venous thrombosis, as well as platelet-mediated transient occlusions of the microcirculation and bleeding, represent the main risks for ET patients. Headache, transient ischemic attack, stroke, visual disturbances and light headedness are some of the neurological manifestations of ET.[2] Chorea has been rarely reported as the presenting feature in patients with polycythemia vera (PV) in the literature. PV is one of the treatable causes of chorea and must be considered as a differential for any chorea. Most of the cases of polycythemia chorea have occurred in elderly women with an acute onset. The chorea is usually generalized with predominant involvement of the orofacial lingual muscles, but it might be unilateral rarely.[3] There are few reports of PV presenting as generalized chorea in literature, but none with ET. Hiroyuki Ito *et al*., published one case of ET presenting as sudden onset hemichorea with MRI showing T1 hyperintense pallidum and putamen similar to diabetic hemichorea.[4] An acquired V617F mutation in JAK2 occurs in 90% of patients with PV, but is seen in only in half of those with ET and idiopathic myelofibrosis. Mutation-positive ET patients had features resembling PV like more venous thromboses and a higher rate of polycythemia transformation than those without the mutation. V617F-positive individuals were more sensitive to therapy with hydroxyurea, but not anagrelide, than those without the JAK2 mutation. JAK2 V617F-positive ET and PV form a biological continuum.[5] The exact cause or pathogenesis of chorea in MPD is unknown. Since JAK2 positive ET closely resembles PV we suggest that same mechanism may be responsible for causing chorea in both. Blood hyper viscosity in PV or ET reduces and impairs oxygen transport, particularly in the basal ganglia, probably plays an important part in the pathogenesis.[6] The neostriatal hyper viscosity syndrome produces venous stasis, reduced brain blood flow and impaired tissue oxygen/glucose metabolism. A state of dopaminergic hyperactivity is presumably enhanced by relatively increased neostriatal catecholestrogens.[7]

Conclusion

To conclude, ET is a rare MPD, which may present with neurological manifestations. We report the first case of generalized chorea as a presenting feature of ET. Most of the cases of chorea in adults are due to neurodegenerative diseases and hence diagnosing ET/PV is essential as they are treatable. JAK2 positive ET may be a biological continuum of PV. Early diagnosis and prompt treatment will avoid serious complications of MPD.

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