Irreversible chorea as the initial presentation of polycythemia rubra vera in an elderly woman

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Janus Kinase 2 (JAK2) protein controls the signal transduction of the erythropoietin, thrombopoietin, and related receptors that control erythrocyte and megakaryocyte expansion. A somatic mutation of JAK2-V617F in the exon 14 of the JAK2 gene (9p24) is the most common cause of myeloproliferative disorder called polycythemia rubra vera (PRV).1-3

Polycythemia rubra vera is characterized by elevated hemoglobin, red cells mass, platelets and white cell counts. The incidence of PRV is 0.4-2.8/100,000 persons/year in Europe and the prevalence is 44-57/100,000 persons.3 The PRV is most common in those aged between 50-70 years and the risk of death with PRV is 1.6 times higher than the general population. We present a patient whose first clinical manifestation of the PRV was chorea though chorea as the first presentation of the PRV is rare (less than 5%). However, other neurological manifestations like headache, visual symptoms or cognitive impairment are common in PRV.1-3

Polycythemia rubra vera is a chronic disease and the treatment protocol should be individualised. Phlebotomy is performed to reduce hematocrit below 45% to improve blood circulation. Since this condition leads to hyperviscosity, low dose aspirin is recommended to reduce the risk of thrombosis.3 However, in people above 60 years of age with previous hypertension or previous thrombosis, drugs like hydroxyurea can be tried.2,4

An 80-year-old woman presented with abnormal limb and facial movements for 6 months. She was slightly fidgety and unsteady but reported no change in mood, memory or cognition. She reported headache after 6 months of the abnormal limb movements. She was taking a regular antihypertensive and related receptors that control erythrocyte and megakaryocyte expansion. A somatic mutation of JAK2-V617F in the exon 14 of the JAK2 gene (9p24) is the most common cause of myeloproliferative disorder called polycythemia rubra vera (PRV).1-3

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The hyperviscosity due to the raised red cell mass in the neostriatal area probably led to the venous stasis, low blood flow, impaired oxygen and glucose metabolism, causing movement disorder.2,4,5 However, the persistence of chorea in our patient despite the improvement of blood viscosity raises 2 possibilities (namely, either hyperviscosity resulted in a lacunar infarct with irreversible injury to the striatum or the mechanism of chorea in PRV was not hyperviscosity alone).3,4 Lew J et al3 reported chorea in a patient with normal hematocrit but with positive JAK 2 V617F mutation. The other possible mechanisms of chorea are altered dopamine metabolism in platelets or receptor

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hypersensitivity secondary to estrogen deficit in postmenopausal women. However, magnetic resonance imaging is often inconclusive in polycythemic chorea like our patient.

Polycythemic chorea is more prominent in the orofacial and lingual muscles though it can be generalized also. Polycythemia rubra vera should be considered as one of the causes of adult onset treatable chorea. The other, more common, causes of chorea in the adult are genetic, drug-induced, infective, paraneoplastic or vascular.

In conclusion, an 80-years-old woman presented with predominantly orofacial and lingual chorea. She tested positive for JAK 2 V617F mutation and was diagnosed as PRV. The improvement of hematocrit after phlebotomy did not lead to the resolution of her chorea.

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