Polycythemia vera (PCV) is the most common myeloproliferative neoplasm in the United States. It is characterized by erythrocytosis and the presence of a mutation in JAK2, a molecule that binds to erythropoietin, myeloid precursors and results in constitutive activation of the JAK-STAT pathway. PCV increases the risk of vascular events, venous and arterial thrombosis, splenomegaly, myelofibrosis, acute leukemia.

We present a new case of PCV:

- 68 year old male with history of massive pulmonary embolism 6 years prior on long-term anticoagulation with warfarin therapeutic INR 2-3.
- He presented with left leg swelling and was found to have a recurrent left deep venous thrombosis in the left peroneal vein.
- His hematocrit (HCT) was abnormally elevated 59.7% and hemoglobin (HGB) 18.7 g/dL. ULN 16.0g/dL. White blood cells and platelet levels were normal. Prior labs showed a normal HGB and HCT over the past nine years.
- Secondary etiology of erythrocytosis was evaluated by testing the erythropoietin (EPO) level which was appropriately suppressed at 1.6 mIU/mL; normal carboxyhemoglobin 1.7%. He had no history or symptoms of obstructive sleep apnea, heart failure or lung disease, and was a non-smoker. A congenital high oxygen affinity hemoglobin was not suspected. Quantitative JAK2 V617F mutation by polymerase chain reaction peripheral blood testing showed abnormal JAK2 V617F mutant DNA detected at 52%.

Anticoagulation was continued with warfarin and weekly therapeutic phlebotomy (removal of 500mL whole blood) for a goal HCT of 45%. Bone marrow biopsy was performed for evaluation for myelofibrosis and showed normocellular marrow with progressive trilineage hematopoiesis, no fibrosis.

Due to his age over 60, history of thrombosis he was deemed high risk PCV and was initiated on 1000mg hydroxyurea a day for cytoreduction. After 1 month of therapy and 4 phlebotomies performed the HCT had improved to 52%.

- Polycythemia vera is a cause of recurrent thrombosis and should be considered on the differential diagnosis.
- Evaluation for secondary erythrocytosis by testing EPO to rule out paraneoplastic production, chronic carbon monoxide exposure, sleep apnea and high oxygen affinity HGB, cardiopulmonary disease is important as management is directed to treating the underlying disorder.
- Bone marrow evaluation for progression to myelofibrosis or blast phase should be performed.
- Cytoreduction with hydroxyurea and therapeutic phlebotomy to reduce risk of thrombosis, vascular events should be initiated for those over age 60 with thrombosis as this is a leading cause of mortality.

Angela Fleischman, Kristen Pettit, 2022, Myeloproliferative neoplasms, American Society of Hematology Self-Assessment Program figure 17-2