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An evolving case of a myeloproliferative disorder in a youngster

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Essential Thrombocytosis (ET) is a myeloproliferative disorder (MPD) in which an increased megakaryocytic proliferation leads to abundant platelet production. It has a prevalence of 1-2 cases/100,000, 50% of which express JAK2 mutation. ET can evolve to another MPD such as Policitemia Vera (PV). We report this case to raise awareness for MPD in youngsters. A 28 year old woman with a history of asthma, obesity and smoking habits who was diagnosed with an asymptomatic thrombocytosis (548 × 109/L → 820 × 109/L), leucocytosis (14,2 × 109/L) and neutrophilia (9,24/µL) in routine blood tests, with the haemoglobin concentration in the upper limit. Further exams revealed normal renal, liver and thyroid function with no increase in inflammatory parameters, excluding reactive thrombocytosis. The abdominal ultrasound (US) showed a slightly enlarged spleen. Considering a MPD we referred the patient to a haematologist who confirmed the results, prescribed acetylsalicylic acid and further investigated. He found normal serum levels of iron, ferritin, vitamin B12 and folate, decreased erythropoietin and JAK2 V617F heterozygous mutation. The bone marrow biopsy (BMB) showed hypercellular bone marrow (haematopoiesis 80%), megakaryocytic hyperplasia with increase in size and number and enlarged dysmorphic cells, no abnormalities in other lineages or evidence of blast cells and was negative for bcr-abl mutation, which allows the diagnosis of ET. The patient maintained acetylsalicylic acid and was encouraged to lose weight and quit smoking to reduce the risk of thrombotic events. Nonetheless, due to borderline haemoglobin and to low erythropoietin levels, we consider the possibility of a pre-policitemic stage of PV. The workup enabled the exclusion of reactive thrombocytosis and myelodysplastic syndrome. Considering the presence of JAK2 mutation, borderline haemoglobin and low erythropoietin levels, there's a high probability of this TE evolving to PV, which forces us to be alert to raising haemoglobin, thrombotic events and blood dyscrasia.