Case Reports

A boy with probable essential thrombocythaemia

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Introduction

Essential thrombocythaemia (ET) is a disorder characterized by sustained proliferation of megakaryocytes in the bone marrow which results in persistent thrombocytosis (platelet count 450,000 per cu mm or more) with a tendency for complications such as thrombosis or haemorrhage[1]. ET is extremely rare in childhood[1]. A mutation which results in a valine to phenylalanine substitution at position 617 in the Janus Kinase 2 gene is identified in 40 to 60% of patients with ET[2]. We report a 10 year-old boy with persistent thrombocytosis who probably has ET.

Case report

A 10 year old boy presented with a grossly elevated platelet count of 2,062,000/cu mm during the course of investigating an upper respiratory tract infection. He was the second child of non-consanguineous parents. His past medical history was unremarkable except for an afebrile convulsion at the age of 7 years. A high platelet count of 1,054, 000/cu mm was noticed at that time, too, but was not investigated. He had no family history of similar illnesses. Physical examination was unremarkable.

Investigations showed normal white blood cell counts and haemoglobin. There were no abnormal cells or large platelets in the blood picture. Inflammatory markers, liver function tests and renal function tests were normal. Serum ferritin and alpha fetoprotein levels were normal. His parental platelet counts were normal. Ultrasound scan of the abdomen showed no hepatosplenomegaly. Contrast enhanced computed tomography (CECT) scan of brain was normal. Bone marrow examination revealed a normocellular marrow with normal erythroid and myeloid lineages but clusters of large megakaryocytes. Myeloblasts were about 3% of all nucleated cells. There was no evidence of myelofibrosis or blast proliferation. The overall histological features suggested a chronic myeloproliferative disorder without myelofibrosis suggestive of ET.

No BCR/ABL chimeric transcript was demonstrated by reverse transcription polymerase chain reaction. JAK2- V617F mutation was negative. Evaluation of the MPL W515L/K and CAL-R genes tests were not done because of local unavailability of the tests. He was referred to a haematologist and started on low dose aspirin 75mg daily and hydroxyurea 500mg every other day. The platelet count came down over a few months and the latest platelet count was 604, 000/cu mm. He remains asymptomatic.

Discussion

Thrombocytosis is relatively common in young children though extreme thrombocytosis (platelet count more than 1,000,000/cu mm) is rare, occurring in less than 2% of children[1]. The most common aetiologies of thrombocytosis are reactive or secondary and once these are excluded, primary or essential thrombocythaemia should be considered.

According to the 2008 World Health Organization (WHO) recommendation, 4 criteria are essential to diagnose ET[3]:

1. A minimal platelet count of 450,000 per cu mm.
2. Bone marrow showing proliferation of megakaryocytes with enlarged mature morphology and no significant increase of granulopoiesis or erythropoiesis.
3. Identification of JAK2V617F mutation or other clonal marker or no evidence of reactive thrombocytosis.
4. Not meeting WHO criteria for chronic myelogenous leukaemia, polycythaemia

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vera, myelodysplastic syndrome, primary myelofibrosis, or other myeloid neoplasm.

Diagnosis of ET in children may also require exclusion of manifestations due to hereditary transmission involving genetic defects of the thrombopoietin receptor, thrombopoietin and myeloproliferative leukaemia mutations expressed in both somatic and germ line cells[4].

In our patient, elevated platelet count of more than 450,000 per cu mm, bone marrow changes and not meeting the criteria for myeloid neoplasms were suggestive of ET. Furthermore, causes for reactive thrombocytosis were excluded. The negative JAK2 mutation in our patient cannot exclude ET as it is positive only in 40 to 60% of patients[2]. However, other clonal markers were not tested because of the non-availability of tests in Sri Lanka.

Treatment consists of antiplatelet and cytoreductive therapy due to the possibility of thrombotic complications[5,6]. Our patient was initially started on aspirin and as there was no improvement hydroxyurea was added. The recommended starting dose of hydroxyurea is 15 mg/ kg/day and this can be increased up to 30 mg/kg/day. Alternative drugs include interferon-alpha and anagrelide[7]. Allogeneic bone marrow transplantation is regarded as an experimental therapy in patients whose condition has transformed into acute leukaemia or myelodysplasia or progressed into myelofibrosis[2].

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