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Case Report

Identification of A Novel inv(2)(p13;q14) in Essential Thrombocythemia

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Introduction

A 69-year-old man came at our observation for thrombocytosis. Peripheral blood counts showed: PLT, 805x10^9/L; hemoglobin 14.4 g/dl; WBC, 7x10^9/L (with 58% neutrophils, 27% lymphocytes, 12% monocytes, 2% eosinophils, and 1% basophils). Bone marrow examination at trephine biopsy showed a hypercellular marrow with an obvious megakaryocytopoiesis increase (Figure 1). Some of the megakaryocytes were small, with a reduced ploidy. The karyotype, assessed as previously described [8,9] showed an isolated inv(2)(p13;q14). The patient did not record any previous thrombosis or hemorrhage. Clinically, the disease remained stable, the PLT count being 750 x10^9/L, at the time of writing after 6 years of hydroxyurea treatment, at the daily dosage of 1000 mg p.o. No thrombotic or hemorrhagic event was reported during the follow-up. Some episodes of tinnitus were retailed after a period of 11 months since the date of diagnosis. This is the first time, to the best of our knowledge, that inv(2)(p13;q14) was recorded in ET. In anaplastic large cell lymphomas (ALCL) is rarely observed the inv(2)(p23q35). It involves ALK and ATIC genes, and the fusion protein has a cytoplasmic localization [10]. No other genetic aberrations involving the same chromosomal region are reported in hematological malignancies. Interestingly, however, the loci 2p13 and 2q14 do contain genes known to be involved in the pathogenesis of human cancer, such as MERTK (c-mer proto-oncogene tyrosine kinase), TGFA (transforming growth factor alpha), IL1A (interleukin-1 alpha), and, intriguingly, IL1B (interleukin-1 beta), known to enhance the megaryocyte proliferation in vitro [11]. In conclusion, though we cannot exclude that inv(2) was not strictly related to the neoplastic clone, we hypothesize that the herein described aberration is a newly acquired chromosomal abnormality in ET. Further studies are warranted to better define the exact prevalence and to definitively characterize the phenotypic and molecular features associated with this chromosomal aberration.

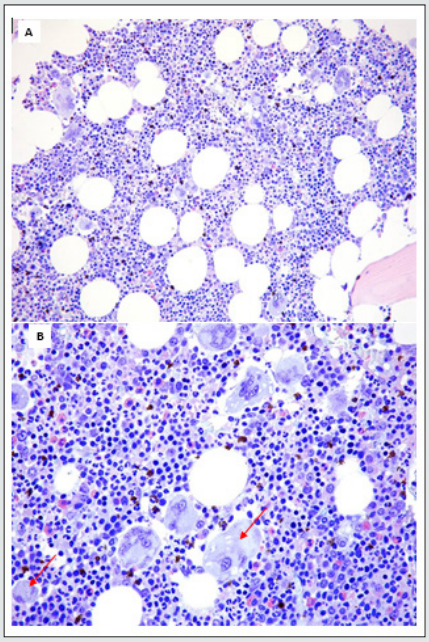

Figure 1: Bone marrow trephine biopsy. A) Hypercellular bone marrow with regular erythropoiesis and granulopoiesis. B) Megakaryocytes are increased in number and mature, with some evidence of dystrophy, and reduced size/ploidy (arrows).

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References

1. Thiele J, Tefferi A, Kvasnicka HM (2017) Essential thrombocythaemia. In WHO Classification of Tumours of Haematopoietic and Lymphoid Tissues IARC, Lyon.
2. Mitelman F, Johansson B, Mertens F (2021) Mitelman database of chromosome aberrations in cancer.
3. Ganvat N, Tefferi A, Thanarajasingam G, Mrinal Patnaik, Susan Schwager, et al. (2009) Cytogenetic abnormalities in essential thrombocythemia:prevalence and prognostic significance. Europ J Haemat 83: 17-21.
4. Sever M, Kantarjian H, Pierce S, Nitin Jain, Zeev Estrov, et al. (2009) Cytogenetic abnormalities in essential thrombocythemia at presentation and transformation. J Hematol 90(4): 522-525.
5. Hsiao HH, Ito Y, Sashida G, Junko H Ohyashiki, Kazuma Ohyashiki, et al. (2005) De novo appearance of der(1;7)(q10;p10) is associated with leukemic transformation and unfavorable prognosis in essential thrombocythemia. Leuk Res 29(11): 1247-1252.
6. Paz DL, Jouanneau-Courville R, Riou J, Jean-Christophe Ianotto, Françoise Boyer, et al. (2020) Leukemic evolution of polycythemia vera and essential thrombocythemia: genomic profiles predict time to transformation. Blood Adv 4(19): 4887-4897.
7. Wong KF (2004) A novel interstitial deletion of 3p in essential thrombocythemia. Cancer Genetics153(1): 84-85.
8. Piccaluga PP, Luatti S, Ascani S, Michele Bianchini, Michele Malagola, et al. (2004) Identification of a novel t(1;9)(q11;q34) in acute myelocytic leukemia. Cancer Genet Cytogenet 151(1): 85-86.
9. Visani G, Bernasconi P, Boni M, G L Castoldi, S Ciolli, et al. (2001) The prognostic value of cytogenetics is reinforced by the kind of induction/consolidation therapy in influencing the outcome of acute myeloid leukemia--analysis of 848 patients. Leukemia 15(6): 903-909.
10. Ma Z, Cools J, Marynen P, X Cui, R Siebert, et al. (2000) Inv(2)(p23q35) in anaplastic large-cell lymphoma induces constitutive anaplastic lymphoma kinase (ALK) tyrosine kinase activation by fusion to ATIC, an enzyme involved in purine nucleotide biosynthesis. Blood 95: 2144-2149.
11. Beaulieu LM, Lin E, Mick E (2014) Interleukin 1 Receptor 1 and Interleukin 1β Regulate Megakaryocyte Maturation, Platelet Activation, and Transcript Profile During Inflammation in Mice and Humans. Arterioscler Thromb Vasc Biol 34(3): 552-564.

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