中文題目:兩種 BCR-ABL 融合基因同時出現在急性淋巴性白血病一個案報告

英文題目: Dual BCR-ABL fusion transcript, P190 and P210, in Acute lymphoblastic leukemia—Case Report

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Background：The positive Philadelphia chromosome: t(9;22)(a34;q11) and BCR/ABL is one of the most frequent rearrangement in adult acute lymphoblastic leukemia (ALL). The incidence of t(9;22) increases with age and is present in 40 to 50 percent of patients older than 60 years. There are three principal forms of BCR-ABL (p190, p210, and p230 BCR-ABL), and BCR-ABL P190 fusion transcript is more common in acute lymphoblastic leukemia. We report one case with ALL who expressed dual BCR-ABL P190 and P210 fusion transcript expression.

Case Presentation：A 38-year-old healthy woman came to OPD with a sudden onset of blurred vision of the retinal hemorrhage related IN Jan. 2015. Marked leukocytosis, anemia and thrombocytopenia were also noted. Bone marrow biopsy showed acute B lymphoblastic leukemia with positive CD79a, TdT, CD34 and negative CD3 and MPO in specified stain. The bone marrow cells contained Philadelphia chromosome, and the molecular analysis revealed BCR-ABL P210 fusion transcript. The patient received the target therapy with Imatinib and induction chemotherapy with MRC-ECOG protocol. Then the patient received the intensification of chemotherapy with MTX + L-asparaginase for 3 courses, and remission of the disease was achieved. In the follow-up regular bone marrow aspiration, previous bcr-abl P210 fusion transcript disappeared after Imatinib use. But, a new fusion transcript, bcr-abl p190, developed after using Imatinib for 6 months. Later, we shifted the TKI from Imatinib to Dasatinib. The bcr-abl p190 fusion transcript was disappear after 6 months of Dasatinib. The karyotype was also showed no Philadelphia chromosome. Then she attained a complete remission, and the allogeneic hematopoietic cell transplantation was performed on Dec. 2015 smoothly. The disease status is in CR to now.

Discussion：In this case, we showed dual BCR-ABL fusion transcript in one patient. We try to propose several mechanisms of alternating expression of the BCR-ABL P190 and P210 fusion transcript. First, initially, the level of BCR-ABL P190 fusion transcript is too low to detect. The second mechanism is alternative spicing during transcription. Further detailed investigation should be performed to confirm the mechanism.

Reference:

British Journal of Haematology, 1998, 103, 711-715