HEMATOLOGY, TRANSFUSION AND CELL THERAPY

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Poster Presentations

Adult Hematology - Categories

Acute Lymphoblastic Leukemia

PP 01_Case Report

A FUSION OF NUP214 TO ABL1 ON AMPLIFIED EPISOMES IN T-CELL ACUTE LYMPHOBLASTIC LEUKEMIA: CASE REPORT AND LITERATURE REVIEW

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Objective: We describe a NUP214::ABL1 fusion identified in a case of T-cell Acute Lymphoblastic Leukemia (T-ALL). **Methodology:** The clinical data of a NUP214::ABL1 fusion gene-positive T-ALL patient were retrospectively analyzed. **Results:** A 13-year-old girl was admitted to our hospital complaining of lower limb edema and leukocytosis. She displayed recurrent edema of both thighs accompanied by cough. A peripheral blood examination showed the following counts: White Blood Cell Count (WBC) 352.5×10^9/L, Neutrophil count 267.91×10^9/L, Lymphocyte count 83.19×10^9/L, Red Blood Cell Count (RBC) 2.2×10^{12}/L, hemoglobin 67g/L, platelet count 79×10^9/L, and C-Reactive Protein (CRP) 12.52 mg/L. Leukemic blasts accounted for 90% of the bone-marrow cells. The patient demonstrated a T-cell phenotype, and showed expression of CD2, CD3(dim), CD4, CD5, CD7 (bri), CD10, CD34, CD38, CD99 and cCD3. A G-band-staining chromosomal analysis revealed normal karyotype. A Fluorescence In Situ Hybridization (FISH) analysis revealed ABL1 amplification (Fig. 1). A ph-like ALL33 fusion gene screening analysis discovered NUP214::ABL1 fusion. In conclusion, the child definitive diagnosed T-ALL with NUP214::ABL1 fusion. Complete remission was achieved after T-ALL induction therapy with vincristine, dexamethasone, PEG-L-asparaginase, daunorubicin, cyclophosphamide, cytarabine, mercaptopurine and dasatinib. To follow-up date, the patient's condition was stable in consolidation therapy phase. **Conclusions:** NUP214::ABL1 fusion is present in 6% of T-ALL cases in both children and adults, it is cryptic by conventional cytogenetics but detected by FISH using a ABL1 probe. FISH analysis reveals multiple extrachromosomal ABL1 sites in metephase cells and amplified ABL1 signals in interphase cells. The amplified signals or episomes are the result of the excision of the 9q34 region between the ABL1 and NUP214 breakpoints followed by circularization of the fragment. NUP214::ABL1 fusion T-ALL represents a distinct form of high-risk leukaemia with early replase and poor prognosis. Because the ABL1 fusions are sensitive to Tyrosine Kinase Inhibitors (TKIs), the strategy of conventional chemotherapy with TKIs can improve outcome in NUP214::ABL1 fusion T-ALL.

Keywords: NUP214::ABL1, ABL1 amplification, T-ALL, FISH.

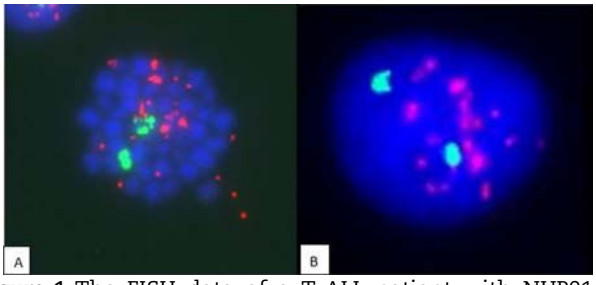

Figure 1 The FISH data of a T-ALL patient with NUP214::ABL1. The red signals indicate copies of ABL1, and the green signals indicate copies of BCR using FISH with a dual-color probe for the detection of the BCR::ABL1 fusion. No fusions are present in these cells. (A) In the metephase cell, multiple red ABL1 sites outside of chromosome. (B) In the interphase cell, the cluster of red signals indicates the amplification of ABL1.

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