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Acute lymphoblastic leukemia in the context of constitutional mismatch repair deficiency syndrome: a case report

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Objective: B-cell precursor acute lymphoblastic leukemia (pre-B ALL) is the most common childhood cancer. Although most childhood B-ALL is sporadic, a subset occurs in children with pre-existing conditions that predispose to leukemogenesis.

Case report: Five and a half year old girl admitted with fever. There were multiple cafe au lait spots without axillary freckling, hepatosplenomegaly and cervical lymphadenopathy on physical examination. Family history elicited that her mother and father were first degree cousins and none of them but her little brother had multiple cafe au lait spots. Laboratory investigations revealed a low hemoglobin level (4.9 g/dL), a low platelet count (13,000 cells/mm^3) and a normal leukocyte count (6610 cells/mm^3). A peripheral blood (PB) examination revealed the presence of leukemic blasts of uncertain origin (30%). A bone marrow (BM) smear showed complete infiltration of L1 blasts. Immunophenotyping was consistent with pre-B ALL. Karyotype of BM blasts could not be analysed because of insufficient number of metaphase cells. FISH analyses showed trisomy 8. Accompanied with ALL IC BFM 2009 chemotherapy protocol, diagnostic work up directed to cancer predisposition syndromes proceeded. Next-generation sequencing (NGS) revealed a mutation in one of the CMMRD genes. The mutation was biallelic in PMS2 gene and according to American College of Medical Genetics and Genomics 2015 guidelines, it was an "uncertain clinical significance" mutation. At the end of induction she was in remission and karyotype and FISH analyses of BM were both normal. The patient experienced hepatosplenic candidiasis and lobar pneumonia during chemotherapy but no dose reduction was made. Follow-up for CMMRD and maintenance treatment of ALL is going on.

Methodology: We present a child with constitutional mismatch repair deficiency syndrome (CMMRD) related pre-B ALL.

Results: Although we do not know the exact karyotype of blasts in our case, at least they have trisomy 8. Trisomy of chromosome 8 is frequently reported in myeloid lineage disorders and also detected in lymphoid neoplasms as well as solid tumors, suggesting its role in neoplastic progression in general. Trisomy 8 is associated with poor prognosis in acute and chronic myeloid leukemias but prognostic significance of extra 8th chromosome in lymphoid malignancies is not reported widely. Trisomy 8 could represent an alternative mechanism for increasing c-myc gene dosage to achieve amplification of c-myc oncogene but mechanisms underlying the events need further study.

Conclusion: CMMRD syndrome is a rare disease and related malignancies need individualization of therapy and novel approaches to optimize care. The child presented herein is a unique case who has CMMRD syndrome phenotype with an "uncertain clinical significance" mutation in PMS2 gene and ALL with trisomy 8. To our knowledge, trisomy 8 has not been reported in ALL in the context of CMMRD syndrome.

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RED BLOOD CELL DISORDERS

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Potential risk of subclinical iron deficiency anemia in misinterpretation of glycosylated hemoglobin a1c (hba1c): a case report

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Objective: Iron deficiency anemia (IDA) which is a global public health problem affecting both developing and developed countries, appears to be more common in diabetic patients compared to non-diabetic population. Gycosylated hemoglobin (HbA1c) which is widely considered as the primary target for glycemic control of diabetic patients, may be altered by certain condition including depletion of iron store with elevation of HbA1C concentrations independent of glycemia. However, reports of the clinical significance of iron deficiency on the glycosylated HbA1c levels have been inconsistent.

Case report: We report a case of 48-year-old diabetic patient with subclinical iron Deficiency, who was in a potential risk of receiving unneeded insulin injection because of false-high values of HbA1c.

Methodology: The false-high values of HbA1c (>7.0%) noticed earlier with the subclinical iron deficiency anemia (hemoglobin 12.7 g/dL, serum iron: 7.94 umol/L, ferritin: 7 ng/mL), then after with frank iron deficiency anemia (hemoglobin 10.1 g/dL, serum iron: 5.68 u, mol/L, ferritin: 5.9 ng/mL).

Results: Interestingly, this high value of HbA1c was subsequently fall (5.8%) simply on correcting the iron deficiency (Ferritin: 10.6 ng/mL) by receiving iron supplementation.

Conclusion: We emphasizes that iron deficiency with or without anemia must be corrected before any diagnostic or therapeutic decision is made based on HbA1c in order to prevent misclassification of diabetes with its hazardous consequences of incorrect treatment.

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