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Vacuolization of Myeloid Lineage and Multilineage Dysplasia in a Case of Transcobalamin II Deficiency

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A 6-month-old male child, born of 3rd degree consanguineous marriage, presented with bronchopneumonia and failure to thrive. On examination, there was pallor and mild hepatosplenomegaly, but no icterus or lymph node enlargement.

Hemogram showed pancytopenia with haemoglobin of 4.8 gm/dl, white blood cell count of 4.5×10^9 and platelet count of 45×10^9 per microliters. Reticulocyte count was 1%. LDH level was elevated at 1837 IU/L (Normal range: 225–450 IU/L). Vitamin B12 level was within normal range, however serum homocysteine was high at 47.7 micromole/L. Bone marrow examination showed presence of vacuoles in myeloid precursors (Fig. 1) and dysplasia in erythroid precursors and megakaryocytes (Figs. 1 and 2). The patient was treated with intravenous methylcobalamin daily for seven days with marked clinical improvement. Holotranscobalamin level done at this point was undetectable, confirming diagnosis of transcobalamin (TC) II deficiency. Patient was further put on intramuscular 1 mg methylcobalamin twice per week. At 6 month follow up, patient is thriving well with normal hemogram.

Vacuolization of myeloid precursors is seen in Pearson Syndrome, myelodysplastic syndrome and copper deficiency in children. It can rarely be seen in TC II deficiency, a rare genetic disorder presenting during infancy [1].

Similarly, multilineage dysplasia is also an unusual finding of this disorder [2].

Fig. 1 Bone marrow aspirate showing prominent vacuoles in myeloid precursors and erythroid dysplasia

Fig. 2 Arrow pointing to a micromegakaryocyte

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Compliance with Ethical Standards

Conflict of interest We declare that we have no conflict of interest.

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