INTRODUCTION: Transcobalamin II (TC-II) is a protein for the absorption, transportation, and cellular uptake of cobalamin. Cobalamin plays a critical role in proliferation phase of DNA synthesis. Due to the depletion of intracellular cobalamin, DNA synthesis is effected in TC-II deficiency. Frequent clinical findings are pallor, weakness, failure to thrive, diarrhea, anemia, pancytopenia, agammaglobulinemia, and neurological problems. Symptoms typically develop within the first year of life. Moderately raised plasma homocysteine, urinary and plasma methylmalonic acid levels with normal B12 concentrations are characteristic laboratory findings. Treatment with parenteral cobalamin is highly effective and clinical manifestations are reversible if treatment is initiated early.

CASE REPORT: A 2-month-old infant admitted with the complaints of aphthous ulcers in oral cavity, recurrent systemic infections, and neutropenia. His birth weight was normal. He had been followed-up in a newborn intensive care unit for sepsis and neutropenia. His parents were first degree cousins and a sibling died at the age of 7 months with a suspicion of immune deficiency. Physical examination revealed pallor and mild hypotonia without organomegaly. Laboratory evaluation put forward neutropenia and anemia. Neutropenia continued despite granulocyte colony-stimulating factor treatment with the suspicion of Kostmann syndrome. Due to unexplained multisystemic involvement with neurological, hematological and immunological manifestations, inherited metabolic disease is suspected. Further evaluation revealed elevated levels of serum homocysteine and urinary methylmalonic acid with normal vitamin B12 concentration. With the prediagnosis of a cobalamin metabolism defect, daily intramuscular hydroxycobalamin treatment was initiated. Bicytopenia improved rapidly and homocysteine level was normalized. Molecular analysis revealed a homozygous novel c.33delG mutation in TCN2 gene. On the last visit his neurological and hematological evaluation were completely normal.

CONCLUSION: TC-II deficiency is an ultra-rare multi-systemic inborn error of metabolism. Decreased levels of intracellular cobalamin lead to bone marrow failure, recurrent fatal infections, and irreversible neurological problems. Early treatment with hydroxycobalamin can easily improve all symptoms and is related with better prognosis. Here, we present this case in order to emphasize including TC-II deficiency in differential diagnosis of immunodeficiency syndromes, megaloblastic anemia, and remethylation defects.