A three-year-old boy was referred to our hospital with the complaint of anemia resistant to iron treatment. Laboratory examination revealed hemoglobin (Hb): 8.7 g/dl, hematocrit (Hct): 30%, red blood cell (RBC): 5.5x10⁹/L, mean corpuscular volume (MCV): 54 fl, red cell distribution width (RDW): 21.6%, and platelet: 448x10⁹/L. Peripheral blood smear examination revealed hypochromia, microcytosis, poikilocytosis and anisocytosis. Reticulocyte count was 1%. The personal history was unremarkable and there was no consanguinity between parents. He had received no transfusions. Physical examination was unremarkable except for mild pallor and mild splenomegaly. Serum iron concentration and serum total iron binding capacity were 77 μmol/L and 362 μmol/L, respectively, and the transferrin saturation was 21.2%. Hemoglobin electrophoresis revealed HbA₂: 1.5%, HbF: 1.9%, and HbH: 3.6%. We observed numerous RBCs containing hemoglobin H (HbH) inclusions in the peripheral blood smear stained with brilliant cresyl blue (Figure 1). Beta Globin Strip Assay (Vienna Lab; Vienna, Austria) was based on reverse hybridization. The mutational analyses using alpha Globin Strip Assay (Vienna Lab; Vienna, Austria) of the family revealed that the patient had -MED II(-26.5)/-α³.7 deletions; the mother and father were found to have -MED II(-26.5) and -α³.7 deletions, respectively.

Although HbH is a common disease, it is rarely reported because of low suspicion [1,2]. In patients with hypochromic microcytic anemia without β-thalassemia trait and iron deficiency, a simple reticulocyte stain can serve as a useful early rapid screening tool for detecting inclusions in patients with HbH disease. This staining can be used as a screening procedure where the molecular diagnosis of HbH disease is not possible.

References
