Example of Abstract

Interaction of hemoglobin QH disease with β-thalassemia: hematological, molecular and diagnostic aspects

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Background and rationale: In an area with high prevalence of thalassemia and hemoglobinopathies, several complex thalassemia syndromes can be resulted from genetic interaction of several defects, leading to a difficulty in laboratory diagnosis of the cases. We report the hematological and molecular features associated with a thalassemia syndrome caused by interaction of several defects in both α- and β-globin genes, found in an adult Thai individual.

Materials & Methods: Study was done in a 29-year-old Thai male, encountered with a mild hypochromic microcytic anemia. Rbc parameters were recorded using standard blood cell counter and Hb analysis was done using the MINICAP FP capillary electrophoresis (Sebia, France). α- and β-globin gene defects were examined using PCR and related methods.

Results: The proband had mild anemia with Hb 12.6 g/dL, Hct 37.8 % MCV 45.5 fL, MCH 15.2 pg and RDW 21.5 %. Hb analysis revealed no normal Hbs but two abnormal peaks. Analysis of the patient's blood mixed with normal specimen identified an abnormal peak of Hb A (95.2%) and an abnormal Hb A_2 (4.8%). No other normal Hb detected. Globin gene analysis identified α^0-thalassemia (SEA), α^+-thalassemia (4.2 kb deletion) linked to the Hb Q-Thailand mutation [α74(EF3)Asp-His (α1), GAC>CAC] and heterozygosity for a β^0-thalassemia (codons 41/42) mutation.

Discussion: The subject was affected by an unusual form of αβ-thalassemia syndrome, the Hb QH disease with β-thalassemia trait, a hitherto undescribed condition in Thailand. Based on the molecular analysis, it could be concluded that the two abnormal peaks observed are the Hb Q-Thailand ($\alpha^Q{}_2\beta_2$) co-separated with Hb F in zone 7 (95.2%) and the Hb QA_2 ($\alpha^Q{}_2\delta_2$) at zone 1 (4.8%). Because the subject also carried α^0-thalassemia, no normal Hb A ($\alpha_2\beta_2$) and Hb A_2 ($\alpha_2\delta_2$) was detected. Nonetheless, an elevation of Hb QA_2 of 4.8% could still point to a co-inheritance of β-thalassemia trait. Identification of this case with the unusual form of thalassemia confirms that in an area where both thalassemia and hemoglobinopathies are prevalent and heterogeneous, complex syndromes may result from interaction of several defects with a spectrum of hematological and clinical manifestations.

Conclusion: Accurate diagnosis of such cases using combined hematological testing and molecular analysis is required to understand the gene-gene interaction and improve genetic counselling.

Keywords: α-thalassemia, Hb Q-Thailand, β-thalassemia, Hb analysis, Molecular analysis