Correlation of BACH1 and hemoglobin E/Beta-thalassemia globin expression

Abstract

Objective: The diverse clinical phenotype of hemoglobin E (HbE)/β-thalassemia has not only confounded clinicians in matters of patient management but has also led scientists to investigate the complex mechanisms involved in maintaining the delicate red cell environment where, even with apparent similarities of α- and β-globin genotypes, the phenotype tells a different story. The BTB and CNC homology 1 (BACH1) protein is known to regulate α- and β-globin gene transcriptions during the terminal differentiation of erythroid cells. With the mutations involved in HbE/β-thalassemia disorder, we studied the role of BACH1 in compensating for the globin chain imbalance, albeit for fine-tuning purposes.

Materials and Methods: A total of 47 HbE/β-thalassemia samples were analyzed using real-time quantitative polymerase chain reaction and correlated with age, sex, red blood cell parameters, globin gene expressions, and some clinical data.

Results: The BACH1 expression among the β-thalassemia intermedia patients varied by up to 2-log differences and was positively correlated to age; α-, β-, and γ-globin gene expression level; and heme oxygenase 1 protein. BACH1 was also negatively correlated to reticulocyte level and had a significant correlation with splenectomy.

Conclusion: This study indicates that the expression of BACH1 could be elevated as a compensatory mechanism to decrease the globin chain imbalance as well as to reduce the oxidative stress found in HbE/β-thalassemia.

Keyword: BACH1, Gene expression, Hemoglobin E/β-thalassemia, oxidative stress, Red blood cell parameters