Prevalence of α-thalassemia-thal-1 in Hb E Trait (Hb E 25-45%) and Mean Corpuscular Volume (MCV) Less Than 80 Femtolitre (fL)

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Background: Detection of α-thalassemia-thal-1 co-in Hb E trait (Hb E trait) is important to identify risk couples for Bart’s hydrops fetalis. In previous study, Hb E level less than 26% and MCV < 75 fL are associated with high prevalence of co-inherited α-thalassemia thal-1. However, there is no study on prevalence of α-thalassemia-thal-1 in Hb E trait (Hb E level 25-45%) and mean corpuscular volume (MCV) less than 80 fL.

Objective: To identified prevalence of α-thalassemia-thal-1 co-occurrence in Hb E trait (Hb E level 25-45%).

Methods: We collected 390 blood specimens from patients who were diagnosed as Hb E trait in our study. Then, we selected 242 specimens with Hb E level 25%-45%, MCV less than 80 fL, and Red blood cell distribution width (RDW) less than 14.5%. We determined Hb E level by Isoelectric focusing (IEF) and/or High-performance liquid chromatography (HPLC) technique and detected α-thalassemia-thal-1 (--SEA, --Thai, --Philippine, --Mediterranean, --20.5Kb deletion), α-thalassemia-thal-2 (3.7,4.2 Kb deletion) by polymerase chain reaction. Furthermore, we analyzed cut-off value of Hb E level, MCV, and MCH to detect co-inherited α-thalassemia-thal-1 in Hb E trait.

Results: From 390 blood specimens, we found co-exist α-thalassemia-thal-1in Hb E trait (12.3%), but we could not detect co inheritance of α-thalassemia-thal-1 in Hb E (25-45%). A suitable cut-off value for detection of co inherited of α-thalassemia-thal-1 in Hb E trait was Hb E < 21% by HPLC technique with 100% sensitivity, 91.2% specificity, 70.6% positive predictive value (PPV), and 100% negative predictive value (NPV). In IEF technique, Hb E < 33% achieved 95.8% sensitivity, 89.5% specificity, 56.1% PPV, and 99.4% NPV. Whereas, the cut-off point of MCV < 72 fL yielded 100% sensitivity, 70.2% specificity, 32% PPV, and 99.6% NPV. In MCH < 22.5 pg/cell, the sensitivity and NPV were both 100%. When we combined Hb E level with MCV and MCH cut-off points, the specificity and PPV were increased to 95.6% and 81.8%, respectively by HPLC technique.

Conclusion: In this study, we can exclude α-thalassemia-thal-1 co-exist in Hb E trait (Hb E>25%) with mean MCV 73.78 fL (66-80). Hb E level<21% by HPLC technique and <33% by IEF technique, MCV <72 fL, and MCH <22.5 pg/cell could be applied for screening of α-thalassemia-thal-1 in Hb E trait, with 95.8% sensitivity and 96% specificity.

Keywords: α-thalassemia-thal-1, Hb E trait, MCV, RDW