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Prenatal thalassemia screening in Viet Nam: a prospective cross-sectional comparative study

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Objective

Vietnam is a Southeast Asian country with high burden of Thalassemia. Mean Corpuscular Volume (MCV) or Mean Corpuscular Hemoglobin (MCH) is the main hematological algorithm commonly used for primary prenatal Thalassemia screening in Vietnam, followed by hemoglobin electrophoresis and genotyping of both parents. The aim of this study was to investigate the utility of genetic profiling compared to current hematological method as a first-line Thalassemia screening tool.

Methods

Blood samples from 547 pregnant women attending routine pregnancy care at the Ha Noi Obstetrics and Gynecology Hospital were collected. Screening was performed in parallel using MCV/MCH method and Thalassemia mutation profiling by Mass Array (Agena Bioscience, United States of America).

Results

Among 131/547 (23.95%) pregnant women screened positive by MCV/MCH, only 64/131 (48.85%) carried Thalassemia mutation(s), resulting in a Positive Predictive Value (PPV) of 49.89% for the MCV/MCH method. PPV for detection of α-thalassemia and β-thalassemia by MCV/MCH was 36.36% and 30.78%, respectively. MCV/MCH-based screening sensitivity for β-thalassemia was 96.00%, higher than that of α-thalassemia (87.23%). Small deletions (α3.7 and α4.2) and single nucleotide polymorphism mutations (Constant Spring) are α-globin mutations often missed by MCV/MCH. Only 49/75 (65%) husbands whose wives carried a thalassemia mutation attended further screening.

Conclusion

Current pregnancy thalassemia screening strategy in Vietnam employs a multi-stage hematological methodology which is affordable yet complicated and inaccurate. We demonstrate the potential utility of primary Thalassemia genotyping for pregnant women compared to current method.