EVALUATION OF RED CELL AND RETICULOCYTE PARAMETERS FOR SCREENING BETA THALASSEMIA CARRIERS

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INTRODUCTION

Beta-thalassemia is the most prevalent monogenic and inherited blood disorder worldwide with approximately 317,000 affected individuals each year. In Egypt, β-thalassemia is the most common genetically-determined, chronic, hemolytic anemia. It had been calculated in Egypt that one thousand children out of 1.5 million live births are born annually with thalassemia major and its carrier rate has been estimated at 9–10.2%. Thalassemia exhibit a wide spectrum of phenotypes. Depending on the clinical severity. Thalassemia is generally divided into three groups: carriers of Thalassemia minor/trait are often asymptomatic and do not need any treatment, those with Thalassemia intermedia have moderate anemia, and occasionally require red blood cell transfusion and Thalassemia major. β-Thalassemia is caused by reduced (β+) or absent (β0) synthesis of the β-globin chains of hemoglobin. Thalassemia exhibit a wide spectrum of phenotypes. Depending on the clinical severity. It is generally divided into three groups: carriers of Thalassemia minor/trait are often asymptomatic and do not need any treatment, those with Thalassemia intermedia have moderate anemia, and occasionally require red blood cell transfusion and Thalassemia major. Thalassemia major patients need a regular blood transfusion and iron chelation therapy which leads to poor life quality for them and a massive financial burden on the clinical care system. Therefore prevention policies are greatly needed, they include carrier screening, gene counselling and prenatal diagnosis. For reliable screening test, different blood count parameters are investigated and several mathematical formulae are developed.

AIM OF THE WORK

Our study aimed to determine an economic and accurate test for screening beta-thalassemia carriers using some reticulocytes parameters and the application of 2 mathematical formulae.

SUBJECTS AND METHODS

Subjects:

A study was carried out on 130 relatives of β-thalassemia patients up to 4th-degree relatives) from Alexandria, attending Hematology /Oncology clinic at Alexandria University Children's Hospital at El Shatby. While, the exclusion criteria included Other hemolytic diseases and Relatives of known α- thalassemia patients.

Methods:

This research is a cross-sectional study. All participants were subjected to full history taking and physical examination. Venous blood samples were collected from all participants for CBC (complete blood count) for measuring RBC and Reticulocytes indices and apply to the formulae, Haemoglobin electrophoresis, Serum iron for all participants, and serum ferritin for participants who had low serum iron.

Formula (1) $$\frac{1.449994 \times (MCV - 82.80762)}{10.28125} + \frac{0.662983 \times (MCH - 27.02007)}{3.904478} + 0.981605$$

Formula (2) $$\frac{(RBCs \times MCHC \times 50)}{MCV} / CHr$$

RESULTS

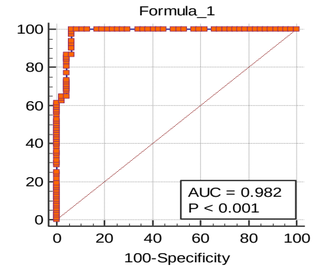
Table 1: ROC curve analysis between Formula1 and carrier state

AUC	95% CI	Cut off point	Sensitivity (95% CI)	Specificity (95% CI)	+PV (95% CI)	-PV (95% CI)	p
0.982	0.941 – 0.997	≤ -0.62	100.0 (95.7 – 100.0)	93.62 (82.5 – 98.7)	96.5 (90.3 – 98.8)	100.0 (--)	**<0.001***

AUC: Area under the curve, +PV: positive predictive value,-PV: negative predictive value

Table 2: ROC curve analysis between Formula2 and carrier state

AUC	95% CI	Cut off point	Sensitivity (95% CI)	Specificity (95% CI)	+PV (95% CI)	-PV (95% CI)	p
0.998	0.968 – 1.000	>5.25	95.18 (88.1 – 98.7)	100.0 (92.5 – 100.0)	100.0 (--)	92.2 (81.9 – 96.8)	**<0.001***



Figur1: roc curve analysis between Formula1 and carrier state

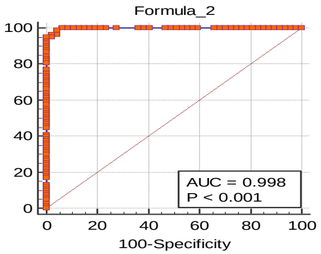


Figure 2: roc curve analysis between Formula2 and carrier state

CONCLUSION

Mathematical formulae using MCV and MCH had high sensitivity and formula used a combination of reticulocyte parameters and RBCs parameters are considered as a reliable and inexpensive method for mass screening of the population to enable the selection of β-thalassemia carriers for further electrophoresis analysis to confirm the diagnosis.