Screening for Beta Thalassaemia Carriers: A Relook

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Editorial

Thalassaemia is an inherited disorder of haemoglobin synthesis. It is characterised by absence or reduced synthesis of one or more globin chains of human haemoglobin. Public health concern about carrier detection is not only confined to the geographical areas with high disease prevalence but becomes a global issue because of population migration [1]. The majority of carriers are not aware of their status. The homozygous state of β0-thalassaemia results in transfusion dependent thalassaemia.

In the past, screening for thalassaemia involved methods such as osmotic fragility of red blood cells, followed with estimation of Hb level in normal persons is 2-3.4%. In classical beta-thalassaemia trait, hypochromic / microcytic and the HbA2 level is raised.

The grey area in beta- thalassaemia carrier identification are levels 3.5-3.9%. In addition, the carriers with mild beta thalassaemia mutations such as poly A (AATAA to AATAGA) may have normal HbA2 levels. It is important to confirm presence or absence of alpha and beta thalassaemia with DNA studies in those with HbA2 levels between 3.5-3.9%. In a carrier with concurrent alpha and beta thalassaemia, the MCV is 75 fl and above and MCH 25 pg and above respectively [5]. However, the Hb A2 level remains elevated.

Hb electrophoresis is now available by 2 automated techniques that incorporate cellulose acetate and gel in alkaline and acidic media. Hb electrophoresis is not used to quantify Hb subtypes. In beta-thalassaemia carrier identification, it plays a role to alert the presence of a beta-thalassaemia haemoglobinopathy. The H-inclusion test including the heat test may be informative for screening of alpha thalassaemia and presence of abnormal haemoglobins. Functional tests for abnormal haemoglobins is done in research laboratories [6].

Currently automated analysers are available for qualitative measurement of red blood cells, red blood cell indices and haemoglobin subtyping. Measurement of variables are accurate and precise with availability of validation criteria incorporating controls, reference materials and calibrated instruments. Quality control procedures including participation in external quality assurance programs have led to improved screening of beta-thalassaemia carriers.

References

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