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Screening of Thalassaemia Carriers and Its Limitations

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The α and β-thalassaemias (thal) are common genetic disorders of globin chain synthesis where the carriers have deficiency of α or β globin chain respectively. In resource limited settings, the naked eye single tube red cell osmotic fragility (NESTROFT) has been used for screening of thalassaemia. It has low specificity compared with its sensitivity. Screening of carriers encompasses automated/semi-automated techniques and manual methods for presumptive diagnosis of thalassaemia. The current strategy in screening involves a step by step process simplified into the following acronym, BHES. B, refers to blood counts and a film; H, haemoglobin (Hb) subtyping; E, Hb electrophoresis; and S, stability (H-inclusion and Sickle cell test). Blood counts and red blood cell indices are now available by automated instrumentation where a cut-off value of mean corpuscular Hb (MCH)<27 pg necessitates further investigation for thalassaemia or iron deficiency. The distribution of normal Hb subtypes (HbA, HbA2 and HbF) is done commonly by high performance liquid chromatography (HPLC) or capillary zone electrophoresis (CZE). The thalassaemia trait phenotype is indicated by a near normal Hb, microcytic and hypochromic red cell indices. In classical β-thalassaemia trait, the cut-off value of HbA2 value is 4% and above. However, milder mutations can result in HbA2 values between 3.2 to 3.9%. In alpha thalassaemia trait the HbA2 falls within the normal range. In a carrier of α0-thalassaemia, occasional or few H-inclusions suggests its presence. However, absence of H-inclusions does not rule out its presence. Recently, it has been shown that the current screening method of thalassaemia carriers has its limitations. Mild mutations may show normal or near normal red cell indices and be missed in the screening process. Carriers with these mutations can cause thalassaemia intermedia. In addition there are mutations that have more profound effects resulting in more severe outcome but have carriers with normal red cell indices. In α-thalassaemia intermedia, HbH disease, those with nondeletional mutations are clinically more severe. These non-deletion mutations include Hb Constant Spring, Hb Pakse and Hb Adana. Carriers with these non-deletional α-thal mutations have normal red cell indices. Cases of hydrops foetalis have been reported with a homozygous state of Hb Adana and also in α0 with Hb Adana. Coinheritance of α or β-thal also effects the red cell indices making them near normal. Thalassaemia intermedia and major have serious implications and prevention requires strategies to detect couples at risk. Mutational spectrum of the globin genes is updated continuously with improved diagnostic technology. Clinical programs preventing thalassaemia are presently based upon prospective screening and prenatal diagnosis. In order for prevention to be effective, there is a need to be aware that mutations can exist in thalassaemia carriers where the red cell indices are normal.