

Prevalence of haemoglobinopathies in young adults from screening camps in Karachi: The importance of using simple thalassemia screen tool for carrier detection in a resource-constrained region

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Abstract

Introduction: Pakistan has a high prevalence of Beta Thalassemia with 5-8% of the population with thalassemia minors, thus there are about 9.8 million carriers. It is estimated that approximately 5000 children are born with thalassemia major, each year. Pakistan's total burden of Thalassemia major affected children may be over 50,000. The expected actual figure is much higher due to unregistered thalasseemics living in rural areas. There is lack of national screening program for thalassemia in Pakistan. Simple tools to identify cases which can later be tested by diagnostic methods is the solution for a developing country with financial limitations. A cross sectional observational study was done in collaboration with Sindlab Diagnostics and JIBA International, Karachi. Total 497 adults were screened using Thalassemia Screening Tool, of which 129 cases were tested for Hb electrophoresis by High Performance Liquid Chromatography (HPLC).

Methods: Complete blood count (CBC) in venous EDTA sample was performed on all subjects. HPLC was later performed on selected subjects using Thalassemia Screening Tool.

Results: A total of 497 subjects were inducted, 21 males and 108 females, mean age 24.5 ± 6.0 years. Normal cases on CBC were 368, and 129 (26.0%) selected for HPLC. Overall prevalence of Haemoglobinopathies was 8.7% (2.2% Beta thalassemia minor, 0.8% Sickle cell trait, 0.2% Hb D Trait, 0.2% Hb E trait and 5.2% Suspected Alpha thalassemia trait). The Positive Predictive Value (PPV) is 81.1% for the CBC tool used to detect Haemoglobin disorders, excluding confirmed cases with iron deficiency anemia as nutritional deficiency states hinders electropho-

retic diagnostic efficacy.

Conclusion: Thalassemia, a major public health concern, is a preventable disease with effective screening programmes. Serious efforts to create preventative measures with Public/Private partnership is the answer. Emphasis on screening with a simple screening tool in schools and colleges may show promising results in a financially restrained country.



Biography

Maliha Sumbul has completed her Membership in Pathology in 2003 and Fellowship in Haematology in 2015 from College of Physicians and Surgeons Pakistan. She did her Post-graduate Diploma in Healthcare Management in 2017 from Institute of Business Administration, Pakistan. She is the Chief Pathologist of Sindlab Diagnostics, Pakistan. She is also a Faculty of Institute of Lab Medicine, Karachi and is actively involved in conducting Technical Workshops and Demo on Learning Lab Software, accredited by AACC. She has extensive teaching experience in the field of Medical Technology and has worked as Senior Lecturer/Assistant Director Medical technology program at Dow and Ziauddin University respectively. She has published articles in Evidentia, UK and Journal of Royal College of Physicians and Surgeons Edinburgh, UK during her work experience in UK, 2005 to 2008. Moreover, she has presented posters in Oxford, London and Brighton Conferences on Malaria audit conducted at Bradford Royal Infirmary, UK

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