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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

Maliha S

Sindlab Diagnostics, Pakistan

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Introduction:

Pakistan has a high prevalence of Beta Thalassemia with 5-8% 1 of the population with thalassemia minors, thus there are about 9.8 million carriers 1. 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 1. The expected actual figure is much higher due to unregistered thalassemics living in rural areas.

This genetic condition requires frequent blood transfusions throughout the life span requiring regular iron chelation therapy. The huge financial burden on family, society and blood banks is inevitable. The only curative approach is Bone Marrow Transplant which is not affordable in a developing country resulting in sub-optimal treatment in most of the cases.

Ironically there is lack of national screening program for thalassemia in Pakistan. Simple screening tools 2 to identify problem cases which can later be tested by diagnostic methods is the solution for a developing country with financial limitations.

It is important to exclude coexisting iron deficiency anemia by serum iron profile determination as nutritional deficiency states suppresses Hb A2 levels 3. Moreover, presence of silent mutations in our population, though rare, requires PCR testing in all cases with suspicion of thalassemia trait if Hb electrophoresis does not reveal raised HbA2 levels, a prerequisite for diagnosis of thalassemia trait.

Objective:

Prevalence of Haemoglobinopathies in young adults from screening camps in Karachi.

Study Design:

A cross sectional observational study

Setting:

Three Thalassemia Camps in collaboration with Sindlab Clinical Laboratory and JIBA International, for the Students, Trainees and Factory workers of a School, Technical Institute and Undergarment Factory, Karachi.

Sample size:

Total 497 children and adults were screened by Complete Blood Count. Of which 129 cases were tested for Hb electrophoresis by High Performance Liquid Chromatography (HPLC) selected by Sindlab Thalassemia Screening Tool.

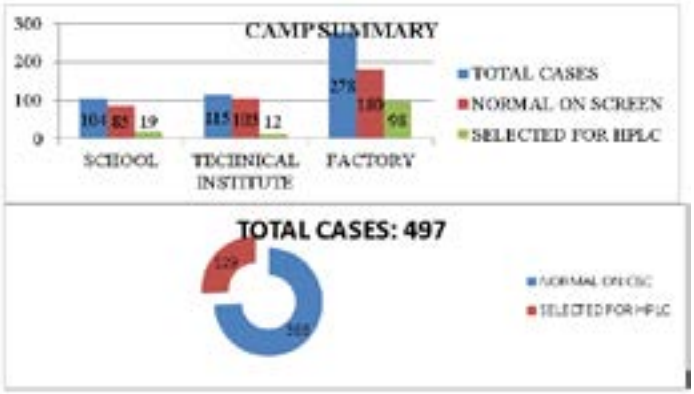
Methods:

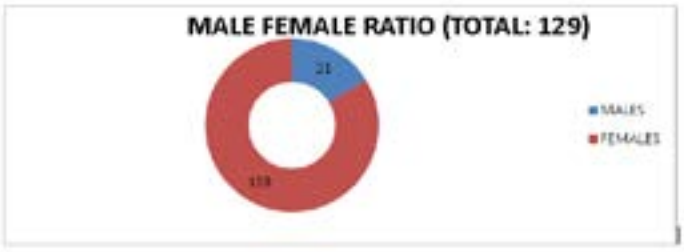
Informed consent was obtained. Complete blood count (CBC) in venous sample collected in EDTA top was performed on all subjects. Hb electrophoresis by HPLC on the same tube was performed on selected subjects (microcytic hypochromic anemia with raised red cell count) using Sindlab Thalassemia Screening Tool.

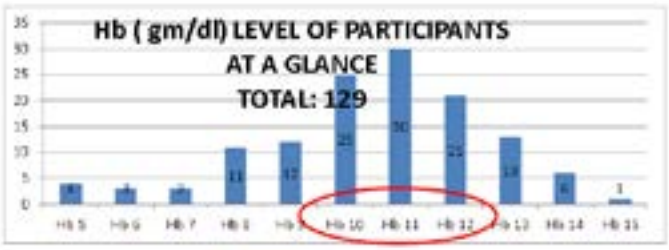
Results:

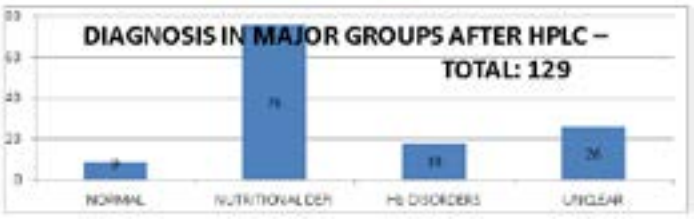
A total of 497 subjects were inducted, 21 males and 108 females, mean age 24.5 ± 6.0 years. Normal cases on CBC were 368, and 129 (26.0%) selected for HPLC. Overall prevalence of Haemoglobinopathies was 8.7% (2.2% Beta thalassemia minor, 0.8% Sickle cell trait, 0.2% Hb D Trait, 0.2% Hb E trait and 5.2% Suspected Alpha thalassemia trait).

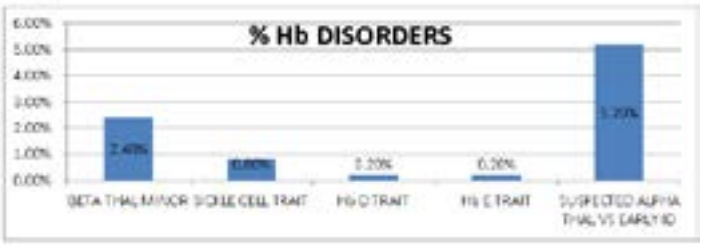
The Positive Predictive Value (PPV) is 81.1% for the CBC tool used to detect Haemoglobin disorders, excluding confirmed cases with iron deficiency anemia as nutritional deficiency states hinders electrophoretic diagnostic efficacy.

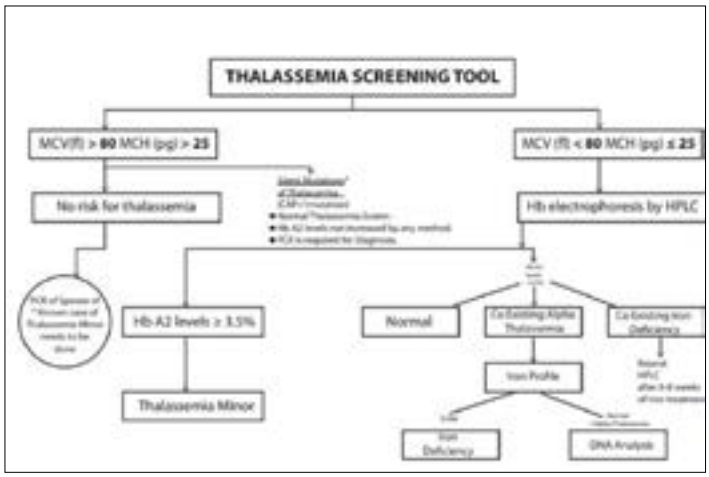












Conclusion:

Thalassemia, a major public health concern, is a preventable disease with effective screening programmes. Serious efforts to create Thalassemia preventative measures with Public / Private partnership is the ultimate answer. Strategies should include awareness programmes, screening and counselling of target families, mass screening of general population, premarital and prenatal screening. Emphasis on screening with a simple screening tool in schools and colleges may show promising results in a financially restrained country.

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