A STUDY OF PREVALENCE OF THALASSEMIA TRAIT AMONG YOUNG ADULT PATIENTS WITH MICROCYTIC HYPOCHROMIC ANAEMIA

By

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ABSTRACT

INTRODUCTION

Among microcytic hypochromic anemias, the most common disorders are iron deficiency anemia and co-pathological conditions such as α- or β-thalassemia (α- or β-thal) traits.

Thalassemia is a alpha/globin gene disorder that results in a diminished rate of synthesis of one or more of the alpha/globin chains. Thalassemia minor is often an asymptomatic carrier state (a mild clinical phenotype when one normal copy of the alpha/beta globulin gene is present) but exhibit marked microcytosis that can be mistaken for iron deficiency.

About 1.5% of the global population (80 to 90 million people) are carriers of Thalassemia and more than 200 mutations are described in thalassemia, the only effective way to reduce burden of thalassemia is to prevent birth of homozygotes.

OBJECTIVE

To estimate the prevalence of beta thalassemia trait in adult individuals with microcytic hypochromic anaemia and also among antenatal mothers visiting the obstetric department of SDMCMSH.

METHODS

Adult population between 18-40 years who were IPD (department of general medicine and OBG) or attending OPD (department of general medicine and hematology) were included and their blood sample was sent for CBC , iron studies and hemoglobin electrophoresis.

RESULTS

Out of the 100 microcytic hypochromic anemia patients analyzed, 7 patients have beta thalassemia trait . 4 of the 7 are female. Their hemoglobin electrophoresis report showed high

HBA2 value of >3.8. There were no carriers among the pregnant individuals tested. A regression model was fitted for predicting HBA2 based on values of serum iron, serum ferritin, and MCV.

CONCLUSION

In conclusion, identification of thalassemia gene mutations in our population is necessary because of the country's multiracial population. This will prevent unnecessary treatment with iron supplements in thalassemic patients and also helps in pregnant women to prevent thalassemia major children.

Molecular genotyping provides a rapid and reliable method for identification of common, and unknown α- and β-gene mutations, which help to diagnose unexplained microcytosis and thus prevent unnecessary iron supplementation.

We conclude that microcytosis due to thalassemia is common in north Karnataka. This illustrates the importance of adequate prenatal and laboratory investigation for these abnormalities.

KEY WORDS

Beta thalassemia trait; Microcytosis; Haemoglobin electrophoresis