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Sickle Cell Hemoglobin E Disorder – A Case Study in Balasore District of Odisha, India

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ABSTRACT

Hemoglobinopathies are the cause of some major genetic and social health problem in many countries including India and sickle cell disorder is one of the most debilitating genetic disease affecting a large population. Though recognized, in and as various groups of genetic mutations, double heterozygous state of sickle cell and hemoglobin E (HbSE) is uncommon in Odisha state. Due to migration and racial inter-caste marriages between the populations of different neighboring states the possibilities of detecting Hb SE disease in Odisha is rising but this rare case of Hb SE has never been reported in Balasore district so far. In the present instance, a 24 year old male was diagnosed with sickle cell hemoglobin E disorder. The case was confirmed through Hb electrophoresis, slide based sickle test, HPLC and ARMS PCR. The peripheral smear analysis showed the presence of microcytic RBC and Hypochromasia. No splenomegaly and hepatomegaly was observed. The case was mostly asymptomatic and with history of joint and bone pains once or twice per year.

***Keywords:** HbSE, Sickle Cell, Hemoglobin SE, Balasore*

Introduction

Hemoglobinopathies are one of the most commonly encountered monogenic disease of human blood, which constitutes some of the most affected genetic and social health problem in regions of Africa, South America and Southeast Asia, particularly in India (Weatherall and Clegg, 2001; Chhotray *et al.*, 2004).

In the medical history of hemoglobinopathies, sickle cell disease (SCD) is the oldest known molecular disease (Frenette and Atweh, 2007). Among them HbE disease caused by a mutation where in lysine is substituted for glutamic acid in the 26th position of the beta chain of hemoglobin A (HBB Glu26Lys) and it is one of the most widely reported hemoglobin related disorder after the HbS (HBB Glu6Val) disease (Thornburg, 2009; Acipayam *et al.*, 2015). Reports have designated it as the second most common hemoglobinopathy variant globally with observations that this mutation has evolved to provide

PHCOG REV. REVIEW ARTICLE

Search for antisickling agents from plants

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ABSTRACT

The sickle cell disease is fatal in nature. Thousands of children are dying off due to this health problem throughout the globe. Due to the rapid development of diagnosis and clinical managements such patients are living up to a respectable age. But as there is no permanent cure the patients are suffering from bone and joint pain, jaundice, hepato-splenomegaly, chronic infections etc. The main physiological complicacy is due to the polymerization of sickle hemoglobin (HbS), (sickling process) inside the red blood cell (RBC) of these patients during deoxygenating state. The change of RBC from spherical to sickle shape is due to the polymerization of mutant hemoglobin (HbS) inside the RBC and membrane distortion during anoxic condition. The mechanism and the process of sickling are very complex and multifactor in nature. To get rid from such complicacies it is necessary to suitably and accurately stop the sickling of RBC of the patients. The potential anti-sickling agents either from natural sources and/or synthetic molecules may be helpful for reducing the clinical morbidity of the patients. A lot of natural compounds from plant extracts have been tried by several workers in recent past. Most of the studies are based on *in vitro* red cell sickling studies and their mode of action has not been properly understood. Although, few studies have been *in vivo* in nature pertaining to transgenic sickle animal model, there is paucity of data on the human studies. The result of such studies although has shown some degree of success, a promising anti-sickling agent is yet to be established.

Key words: Antisickling, plants, red blood cell, sickle

INTRODUCTION

Sickle cell disease (SCD) is a genetic blood disorder characterized by red blood cells (RBCs) that assume an abnormal rigid sickle shape. Sickling decreases the RBC's flexibility and results in a risk of various complications. It is the most prevalent human hereditary disorder with prominent morbidity and mortality.[1] It is due to the change of an amino acid in position six within the beta globin chain of hemoglobin molecule whereby glutamic acid, a polar amino acid is replaced by valine, a non-polar amino acid.[2,3] The amino acid change is due to the defective gene (mutation) in chromosome 11. At low oxygen tension, the mutant hemoglobin polymerizes inside the RBCs into a gel or further into fibers leading to a drastic decrease in the red cell deformability. Polymerization and precipitation of sickle hemoglobin (HbS) within the erythrocytes cause the change of shape from the normal spherical form into the one resembling a sickle, hence the name sickle cell. A single nucleotide substitution (Thymine [T] for Adenine [A]) allows HbS to polymerize when deoxygenated, since valine can dock with the complimentary sites on the adjacent globin chains.[4] The presence of sickle shaped RBCs in human blood was first reported by Herrick (1910).[5]

The SCD has been reported to wide spread in Africa, Jamaica, Central India, Saudi Arabia, Greece and Italy and also among the Negroids of America and Britain. SCD affects millions of people throughout the world.[6] The clinical symptoms of patients suffering from the disease vary widely. Some lead a normal life while others suffer from a variety of life threatening complications. The main clinical symptoms are anemia, mild jaundice, repeated vaso-oclusive crises, hepatosplenomegaly, acute chest syndrome, bone and joint pain and growth retardation.[1,7]

SCD widely has no cure. However, treatment can help to relieve symptoms and reduce the complications. Infants who have been diagnosed with SCD through newborn screening are treated with antibiotics to prevent infections. Blood

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CHAPTER

3

Abnormal Haemoglobin Related Health Problem among the Tribals of Mayurbhanj District, Odisha

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Nibarana Satapathy and Nirmal Chandra Dash

3.1 INTRODUCTION

Abnormal haemoglobin related health problems (Haemoglobinopathies) such as Thalassaemia and Sickle cell disorders are recessive inherited diseases, wide spread throughout the World. According to the World Health Organization (WHO) 7% of the global population carries an abnormal haemoglobin gene. Annually 3, 00,000 to 5, 00,000 children are born with clinically significant haemoglobin disorders. About 70% of them are born with sickle cell disease and the rest are with thalassaemia syndromes.

Haemoglobin disorders constitute a major burden of disease, originally believed to be geographically restricted, mainly to malaria endemic or previously endemic regions of the world (Weatherall and Clegg, 2001). However, migration has imported the condition to the low prevalence areas, so that the problem is now global.

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Full length Research Paper

Mutational Spectrum of β-Thalassaemia of Northern part of Odisha, India

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Prevention of β thalassaemia requires knowledge for diagnosing the molecular analysis in the population at risk. This knowledge is particularly necessary when prevention control is applied to a multiethnic population. For this purpose, we are analyzing different populations from northern part of Odisha, India. During the study, we encountered about 98 patients from District Head Quarter Hospital of Mayurbhanj and Balasore district of Odisha. Molecular analysis of β gene mutation were showing that population showing IVS I-5(G-> C), cd 41/42(-CTTT), cd 8/9(+G), IVS I-1(G->T), cd 15(G->A), cd 30(G->C) as well as 619 bp deletion. In most cases, we found the IVS 1-5(G-> C) mutation and cd 41/42(-CTTT) mutation. The novel 619 bp deletion is the first report being analyzed in northern part of Odisha. The patient's age group more prevalence in between 0 - 15 years and their hematological parameters were recorded.

Keywords: β thalassaemia, β gene mutation, IVS 1-5(G-> C), cd 41/42(-CTTT), cd 15(G->A) mutation.

INTRODUCTION

The term 'Haemoglobinopathies' is given to the inherited disorders of structure and synthesis of globin part of haemoglobin molecule and falls into several overlapping groups. It has been estimated that approximately 7% of the world population are carriers of such disorders and that 3, 00,000–4, 00, 000 babies with severe forms of these diseases are born each year.In developed countries it has been estimated that genetic disease constitutes upto 40% of the requirements for chronic care in pediatric practice. Inherited haemoglobin disorder falls into two main groups i.e. the structural haemoglobin variants (Sickle cell disease) and the thalassaemia. From a public health view point α and β thalassaemia are sufficiently common to be of importance. According to World Health Organization (WHO) estimate about 7% of World population carries an abnormal haemoglobingene [1]. The average incidence of the β thalassaemia trait in India is 4% with one or two couples per 1000 being at risk of having affected offspring each year and annually at least 8000 – 10000 children with β thalassaemia major are born, constituting 10% of the total number born in the World every year[2].

By convention, haemoglobinopathies are classified according to the qualitative nature of the resultant haemoglobin (i.e., sickle cell disease) and the quantitative amount of haemoglobin produced (i.e., thalassemia). Several studies have reported occurrence of haemoglobinopathies at variable frequency in different states and caste population of India [3-10]. There is a paucity of information regarding the occurrence thalassaemia and abnormal haemoglobins other than sickle cell haemoglobin (HbS) in the state of Odisha which is inhabited by 36.7 million people comprising of 22.4% Scheduled Tribes and 16.2% Scheduled Caste population. The earlier studies from Odisha were from

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Research Article

Hypocholesterolaemia in Beta-Thalassaemia

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Abstract

Background: To assess the plasma total cholesterol of male and female β-thalassaemia patients. The plasma cholesterol of normal individual compared with the β-thalassaemia cases.

Methods: From October 2010 to March 2011, we enrolled 36 consecutive patients with β-thalassaemia that visited District Head Hospital, Balasore and Odisha every month for routine examinations. The sample was collected in the EDTA vial (5 ml), immediately centrifuged at 5000 rpm for 5 min and supernatant (plasma) collected in to the microcentrifuge tube and stored in -20°C freezer for other biochemical experiment. We have used Infinite cholesterol kit. We also found 5 sickle patients and included them in our analysis.

Results: Of the 36 thalassaemic patients, 25 were males (12 ± 5.91 years) and 11 were females (9.08 ± 4.32 years) with comparison to 30 normal cases (15-35 years). Data analysis revealed that total cholesterol level, in case of normal (178.8.5 ± 31.1mg/dl), in thalassaemic cases (135.8.6 ± 29.3mg/dl) and in case of 5 sickle patients (143.3 ± 15.7mg/dl). The total cholesterol levels in case of normal is high than that of thalassaemic and sickle patients. In case of thalassaemia cases the low total cholesterol is 47.4 mg/dl. The expected values of total cholesterol is <200 mg/dl. In case of thalassaemia the values are too low than expected values. The 't' test data revealed that there is comparison of two mean of the sample (normal and thalassaemic) is negligible small.

Conclusion: The present study revealed that the majority of males and females with beta-thalassaemia have their blood lipid levels within the normal range, and lower than the healthy individuals of the same age and population. Such low level of cholesterol in thalassaemic patients seems to reflect the inability of the organism to balance the increased cholesterol requirement for red cell membrane formation. It is conceivable that the availability of cholesterol, ordinarily used in steroid hormone synthesis to control hypercoagulability.

Keywords: Beta-thalassaemia; Mean; Standard deviation; 't' test; Total cholesterol (TC)

Introduction

Thalassemia is among the most common genetic disorders worldwide; 4.83 percent of the world's population carries globin variants, including 1.67 percent of the population who are heterozygous for α-thalassemia and β-thalassemia. In addition, 1.92 percent carries sickle hemoglobin, 0.95 percent carry hemoglobin E, and 0.29 percent carry hemoglobin C. Thus, the worldwide birth rate of people who are homozygous or compound heterozygous for symptomatic globin disorders, including α-thalassemia and β-thalassemia, is no less than 2.4 per 1000 births, of which 1.96 have sickle cell disease and 0.44 have thalassemias [1].

β-thalassaemia can be broadly defined as a syndrome of inherited hemoglobin disorders characterized by a quantitative deficiency of functional β globin chains. Although it is defined as a reduction in the synthesis of β- globin, some forms result from structural hemoglobin variants that are ineffectively synthesized or are so unstable that they result in a functional deficiency of the β chains and a thalassaemia phenotype [2]. Beta thalassaemia is a very serious blood disorder since individuals with it are unable to make enough healthy red blood cells and depend on blood transfusion throughout their life. However, quality and duration of life of transfusion-dependent thalassaemic patients has been transformed over the last few years, with their life expectancy increasing well into the third decade and beyond with a good quality of life [3].

During the past years many scientific evidences have raised the adverse effect of abnormal blood lipid levels, like total-cholesterol and other lipids and lipoproteins on atherosclerotic disease [4-6]. At this point it should be mentioned that the relationships between blood lipids and atherosclerosis might be influenced by several other lifestyle-related factors, like glucose intolerance; blood pressure levels, dietary and smoking habits [7]. In recent years, several authors reported a high incidence of endocrine abnormalities in children, adolescents and young adults suffering from thalassaemia.

To the best of our knowledge, data regarding the distribution of blood lipid levels among patients with beta thalassaemia are lacking. Therefore, we investigated the distribution of glucose, protein, bilirubin, urea and total cholesterol in plasma samples of patients with beta-thalassaemia in district of Balasore, Odisha, India. Among these the total cholesterol profile gives the good result with comparison to the normal cases.

Total cholesterol (TC) consists largely of the cholesterol in LDL particles (LDL cholesterol) plus the cholesterol in high-density lipoprotein particle (HDL cholesterol). Cholesterol is not only a fundamental element of cell membranes but also the principal precursor for steroid and sexual hormone biosynthesis. Furthermore, cholesterol, through its intermediary products such as farnesyl diphosphate and geranyl diphosphate, is involved in the regulation of ras-protein intracellular signal transduction.

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REVIEW ARTICLE

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Beta Thalassaemias in Odisha: Current Status and Future Need

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ABSTRACT

Thalassaemias are a group of hereditary disorders of haemoglobin. The carrier frequency of these disorders varies considerably in different caste and population groups of India. As per ICMR report, about 3.9% in an average of the Indian populations are carrier for β thalassaemia. There is no population based survey data on prevalence of β- Thalassaemia in Odisha. Sporadic study data on the occurrence of β thalassaemia and ? thalassaemia have been reported among few caste and tribe population from Odisha. The percentage of the beta thalassaemia trait varies from 0-8.5% among different studied tribal groups. Beta thalassaemia gene in the form of trait was found in Paraja (8.5%), Santal (8.0%), Lodha (6.7%), Kolha (2.0%), Bhumiz (1.7%) of Mayurbhaj, Bhatra (6.6%) of Nawarangpur , Kondha (6.3%) of Phulabani , Saora (6.2%) of Ganja and Gajapati , Munda (5.2%), Oraon (1.9%), Kharia (1.9%), Kissan (1.5%) of Sundargargh and Gond (0.5%) of Kalahandi districts tribal school students of the state. Few cases of homozygous beta thalassaemia major have been reported by several authors from referred hospital samples. The double heterozygous state such as Haemoglobin E ? thalassaemia (14 cases), Sickle cell β^+ thalassaemia (17 cases) and few Sickle cell ?0 thalassaemia cases have also been documented from Odisha. Out of the thirty beta thalassaemia mutations reported from Indian population, only one i.e. IVS 1-5 (G->C) mutation was found from few studied thalassaemia patients of Odisha. Keeping in view of the present information a detail comprehensive strategy has to be adopted to know the exact magnitude of genetic health load and to develop a control measure for thalassaemia in the state.

Key words:

Beta thalassaemias, Odisha, Tribes.

INTRODUCTION

Beta thalassaemia is the commonest monogenic hereditary disorder affecting the population world-wide. It is widespread throughout the Mediterranean region, Africa, the