thalassemia reports

Indian National Conference on Hemoglobinopathies

17-18 May 2013, Bangalore - India

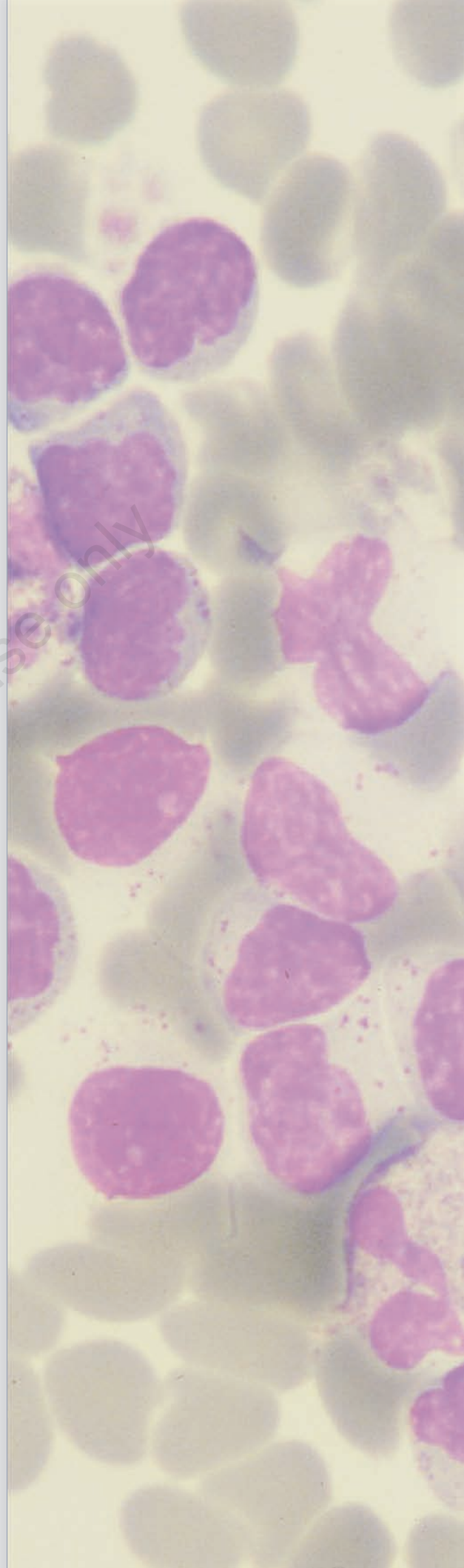
Editors: *Karuna Rameshkumar, Cecil Ross and Anand Prakash*

pagepress

THALASSEMIA REPORTS

ISSN 2039-4357 - eISSN 2039-4365

pagepress



pagepress

THALASSEMIA REPORTS

is published by PAGEPress Publications, Italy.
The journal is completely free online at www.thalassemiareports.org
Publishing costs are offset by a publication fee charged to authors.

For more information
and manuscript submission:
www.thalassemiareports.org

Correspondence

Our publishing offices are located
in via Giuseppe Belli 7, 27100 Pavia, Italy.
Our telephone number is +39.0382.1751762
and our fax number is +39.0382.1750481.
E-mail: info@pagepress.org

INDIAN NATIONAL CONFERENCE ON HEMOGLOBINOPATHIES

17-18 MAY 2013

Bangalore - India

ORGANIZED BY

Departments of Clinical Pathology, Paediatrics & Haematology
St. John's National Academy of Health Sciences
Bangalore - India

ORGANIZING COMMITTEE

ADVISORS

Dr. Roshan Colah
Dr. Cecil Ross
Dr. Karuna Rameshkumar

CHAIR PERSONS

Dr. Sitalakshmi S.
Dr. Sylvan Rego

ORGANIZING SECRETARIES

Dr. Shanthala Devi A.M.
Dr. Fulton D. Souza

TREASURER

Dr. Vanamala A. Alwar

MEMBERS

Dr. Anand Prakash
Dr. Parimala Puttaiah
Dr. Ranjini S.
Dr. Latha Fathima J.
Dr. Saritha Ann Bosco
Dr. Poornima Rao
Dr. Betsy Mathew

Logistics:

Mrs. Pragathi B.
Mrs. Agnes Fabiola
Ms. Veena Maria Saldanha
Mr. Prabir Mukerjee

INDIAN NATIONAL CONFERENCE ON HEMOGLOBINOPATHIES

17-18 MAY 2013

Bangalore - India

ABSTRACT BOOK

TABLE OF CONTENTS

ACKNOWLEDGMENTS

The support of Indian council of medical research (ICMR) for conduct of the program and publication is gratefully acknowledged.

Indian National Conference on Hemoglobinopathies

17-18 May 2013, Bangalore - India

PATHOLOGY POSTERS

PP01

DIAGNOSTIC UTILITY OF RBC INDICES IN DIFFERENTIATING MICROCYTIC HYPOCHROMIC ANEMIAS

*Ramakrishna A., Anuradha A.D., Flynn A.

Department of Pathology, St. Martha's Hospital, Nrupathunga Road, Bangalore, India

**Contact: saturn424@gmail.com*

INTRODUCTION: Iron deficiency anemia (IDA) and β-thalassemia trait (BTT) are two most common causes of microcytic hypochromic anemia. Several discrimination indices have been formulated to differentiate between these two conditions using parameters obtained from automated cell counters.
AIM: To use Mentzer's index to identify potential patients with β-thalassemia trait and refer them for further testing for confirmation.
MATERIALS AND METHODS: 147 cases of microcytic hypochromic anemia diagnosed in a retrospective period of two months were selected. Certain inclusion criteria of RBC count >4.5 million/cubic mm and MCV <75 fL were used to select the cases. The RBC indices obtained from the automated cell counter (Sysmex XT1800i) were studied and Mentzer's index (MCV/RBC count) was calculated for all cases. The cases were then categorized into potential IDA and BTT according to whether the index was > or <13 respectively.
RESULTS: Out of 147 cases, 82 (56%) were categorised as BTT and 65 (44%) as IDA. Among the BTT cases, maximum number of patients had an MCV value in the range of 60.1-65 fL with RBC count in the range of 5.01-5.5 million/cubic mm. The IDA cases were maximum in MCV range of 70.1-75 fL and RBC count range of 4.51-5 million/cubic mm.
CONCLUSIONS: We thus concluded that Mentzer's index helps in differentiating iron deficiency from thalassemia. It helps the pathologist to guide the physician towards appropriate treatment of the patient, especially those who have been suffering from anemia which does respond to iron therapy.

Key words: Mentzer's index, iron deficiency anemia, β-thalassemia trait.

PP02

NESTROFT

*Dinisha D.P., Anu Priyadharshini S., Parijatham B.O.

Department of Pathology, Sree Balaji Medical College and Hospital, Chromepet, Chennai, India

**Contact: dr.irence@gmail.com*

INTRODUCTION: β-thalassemia is the most prevalent hemoglobinopathyin our country. Due to our limited resources and higher population density, a simple screening test is essential for detecting carriers of β-thalassemia. NESTROFT - naked eye single tube red cell osmotic fragility test - is a screening procedure for the detection of heterozygous β-thalassemia, based on the principle that the red cells of hemoglobinopathies are more resistant to lysis by a hypotonic solution compared to normal red cells. The test is positive in other hemoglobinopathies (HbE, HbS) as well.
AIM: To evaluate the efficacy of NESTROFT as a screening test for the detection of β-thalassemia heterozygotes.
MATERIALS AND METHODS: This study was conducted in the Pathology Department of SreeBalaji Medical College and Hospital, Chennai during August 2012 to February 2013. NESTROFT (using 0.36% buffered saline) was done in blood samples of out-patients suspected with thalassemia. Alkaline hemoglobin electrophoresis was done in positive cases, for confirmation.
RESULTS: NESTROFT done in 26 OPD cases suspected with β-thalassemia showed positivity in 19 cases, out ofwhich, 16 cases had an elevated HbA_2 in alkaline hemoglobin electrophoresis. No cases of HbE or HbS was noted in this study (maybe due to low prevalence in this region).
CONCLUSIONS: NESTROFT is a simple and effective baseline screening tool for β-thalassemia trait, especially in centers with limited resources andmass screening programmes.

Key words: NESTROFT, β-thalassemia trait.

PP03

PERIPHERAL SMEAR EXAMINATION: A CLUE TO THE DIAGNOSIS OF SICKLE CELL ANAEMIA

Jena M., *Lakshmi D.

Department of Pathology, MVJ Medical College and Research Institute, Bangalore, India

**Contact: dldivya87@gmail.com*

INTRODUCTION: Sickle cell anemia is a hereditary haemoglobinopathy with varied clinical and hematological manifestation. The hallmark of sickle cell disease is a group of devastating symptoms known collectively as

a sickle cell crisis which can sometimes be the initial presentation in certain patients when the first diagnosis is made.

CASE REPORT: A 22-year old man presented with high grade fever and vomiting associated with pain abdomen since 1 week. On examination he was pale, icteric with moderate hepatospleenomegaly. His Hb was 4.2; serum bilirubin was 50 mg%. A provisional diagnosis of malaria or leptospirosis was suggested. Peripheral smear examination showed marked dyserythropoeiesis with a very few sickle shaped cells. Reticulocyte count was 4%. On the basis of peripheral smear findings, sickling test was performed which was found to be positive. There after a serum electrophoresis confirmed the diagnosis of sickle cell anemia of heterogeneous type. The imaging studies was later done which showed marked infarction of humerus and haemarthrosis of knee joint.

CONCLUSIONS: A very high level of serum bilirubin with hepatospleenomegaly can misguide the clinician towards a diagnosis of either a liver disease or an infective etiology such as malaria or leptospirosis. In this case a careful examination of the peripheral smear helped in clinching the diagnosis of sickle cell anemia.

Key words: peripheral smear, sickle cell anemia.

PP04

A PROFILE OF SICKLE CELL STATUS IN A HOSPITAL BASED SICKLE CELL SCREENING PROGRAMME OF BHILAI CHHATTISGARH

*Meraj F., Ravindranath M., Singh G., Bhaisare R., Kango M., Bhalla M.

JLNH& RC, Bhilai, Chhattisgarh, India

**Contact: mirazfatima2011@gmail.com*

INTRODUCTION: Bhilai is an Industrial township and has a multispecialty hospital providing health-care to a diverse population to people in and around Bhilai. The Jawaharlal Nehru Hospital and Research Centre Bhilai is a referral centre of Bhilai Steel Plant, which serve the employees of the same and other people also.

AIM: i) To screen the patients of JLNH&RC for Haemoglobinopathies; ii) to analyse the trend of Haemoglobinopathies in Bhilai; iii) to educate the carriers and patients of Haemoglobinopathies accordingly.

MATERIALS AND METHODS: The present study was carried out over a period of one year (2011-2012) at Haematology laboratory of Pathology department of JLN Hospital and Research Centre. Twelve hundred ninety patients from paediatric and adult age groups were screened by solubility test. CBC, red blood cell indices, peripheral blood examination was done in all cases using the standard techniques. In abnormal cases family studies were carried out wherever possible. Blood specimens were collected in EDTA vials. All specimens were assessed by Bio-Rad Variant HPLC System, with the use of Variant β-Thalassemia Short Program Recorder Pack (Bio-Rad Laboratories).

RESULTS: Overall 220 families were screened and 1290 samples were tested. Sickle cell trait was detected in 37% of cases, sickle cell disease in 3.8% and combination of sickle disease with thalassemia was observed in 8.7% of case. The trait and disease were equally seen in both the sexes. The average members in the family were found to be 4.5. Nearly 30% of the people screened were unaware that they were carriers of Sickle cell gene and were ignorant about the disease.

CONCLUSIONS: Sickle cell trait is prevalent in and around Bhilai. Significant numbers of people were not aware of their sickle cell status. Efforts have to be initiated to educate the population.

Key words: sickle cell trait, Bhilai, people education.

PP05

INCIDENCE OF ANEMIA IN HOSPITALISED CHILDREN WITH REFERENCE TO HEMOGLOBINOPATHIES

*Saba F.

Pathologist, Bangalore Nursing Home K.G. Halli, Bangalore, India

**Contact: saba.firdos@yahoo.com*

INTRODUCTION: Anemia is a frequent laboratory abnormality encountered in children. The third National Family Health Survey (NFHS) 2005-2006 revealed that at least 80% of Indian children aged 12 to 23 months were anemic.The sparse literature related to anemia in hospitalized children necessitated the present study.

AIM: To estimate the incidence of anemia in children (6 months-12 years), morphological patterns, distribution in different age groups,sex and to assess its severity with special reference to hemoglobinopathies.

MATERIALS AND METHODS: After obtaining Ethical clearance random sample consisting of 882 children between the ages of 6 months to 12 years were included in the study. Consent was taken from their respective parent/guardian. Blood cell counts, peripheral blood smears were studied and statistically analyzed. Investigations like electrophoresis, sickling test, bone marrow study, were done wherever required.

RESULTS: The incidence of anemia was 72.79% (642), non-hemoglobinopathies constituted 98% (629) and hemoglobinopathies constituted to 2% (13). Children 6m0nths-1year were most commonly affected with non-hemoglobinopathies. Among hemoglobinopathies, thalassemia major was most common (69%, 9 out of 13 patients), 7 (54%) children in the age group 6-12 years were most affected followed by 4 (30%) children in the age group 2-3 years and equal incidence of 1 (8%) in the age groups 6 months-1 year and 4-5 years.

CONCLUSIONS: hemoglobinopathies constituted 2% of the anemia in children.

Key words: anemia in children.

PP06

EVALUATING THE ROLE OF HYDROXYUREA AS AN IRON CHELATOR

*Italia K.Y., Colah R., Ghosh K.

National Institute of Immunohaematology, I.C.M.R., KEM Hospital Campus, Parel, Mumabai, India

**Contact: kyitalia4@yahoo.co.in*

INTRODUCTION: Based on our previous study, we hypothesize that hydroxyurea may be playing some role in reduction/removal of labile iron from the body of β-thalassemia patients.
AIM: To evaluating the efficacy of hydroxyurea alone and in combination with most widely used chelators like deferiprone and deferasirox for reducing iron levels of iron overloaded mice.
MATERIALS AND METHODS: CBC and serum-ferritin of 70 Balb/c mice was determined after which they received intraperitonial injections of iron-sucrose. After significant iron overload, the mice were divided into 8 groups and were orally given hydroxyurea, deferiprone, deferasirox alone and its combinations for 4 months, after which, CBC, serum-ferritin and thiobarbituric acid reactive substances-(TBARS) was evaluated. All animals were then sacrificed. Iron staining of the heart and liver tissue was done using Perl's Prussian blue stain. Dry weight of heart and liver iron was done by atomic absorption spectrometry.
RESULTS: Serum-ferritin reduced significantly in groups where iron overloaded mice were administered a single drug with maximum reduction in the group were all three drugs were given together. The increased TBARS levels after iron overload in mice were significantly reduced by combination of the 3 drugs. Dry weight of the liver and heart iron showed maximum reduction in the group treated with a combination of deferiprone and deferasirox and the group treated with deferiprone, deferasirox and hydroxyurea.
CONCLUSIONS: Hydroxyurea proves its role in reducing iron from iron overloaded mice alone and in combination with most widely used iron chelators and also helps in reducing oxidant damage to the tissues caused by iron.

Key words: hydroxy urea, iron chelation.

PP07

PERIPHERAL SMEAR IN THALASSEMIA MAJOR

*Jaisri A.

Southern Railway Headquarters hospital, Perambur, Chennai, India

**Contact: jaisri12@yahoo.com*

INTRODUCTION: The peripheral smear of a 29-year old known thalassemic patient was studied and the microscopic findings were analysed. .
AIM: The objective is to highlight the variation in the peripheral smear findings and blood counts in a thalassemic patient and to correlate it with the clinical findings.
MATERIALS AND METHODS: The leishman stained peripheral smears and blood counts from the automated haematology analyzer were studied.
RESULTS: Hemoglobin 8.1g/dL mcv 79 fL, total WBC count: 490,000/cmm. The peripheral smear showed marked leukocytosis with a shift to the left comprising numerous stab forms, monocytes, basophils, normoblasts and thrombocytosis. The corrected WBC count was 26,000/cmm. RBCS showed hypochromia microcytosis and marked anisopoikilocytosis. On reviewing the patient with the clinical history, it was found that she was splenectomised in 1995.
CONCLUSIONS: The immature cells are due to splenectomy. Thrombocytosis is due to reactive marrow because of anemia. The query is about the numerous basophils. Are they due to an active marrow or due to a rare juvenile coexisting chronic myelomonocytic leukemia as reported in the literature.
LAP score and genetic translocation studies were not done. There were two other patients diagnosed with thalassemis major by peripheral smear and clinical history during infancy. One child was lost to follow-up at 4 months of age. The other child died at 8 months and was born to parents of consanguineous marriage. This 29-year old patient is on treatment in our hospital with blood transfusions and iron chelators since childhood.

Key words: peripheral smear, thalassemia.

PP08

PROFILE OF HAEMOGLOBINOPATHIES IN A TERTIARY CARE HOSPITAL IN SOUTH INDIA

*Karuna R.K., Ross C., Parimala P., Shanthala Devi A.M., Latha F.S., Purnima R., Vanamala A.A., Sitalakshmi S.

Department of Clinical Pathology, St. John's Medical College Hospital, Bangalore, India

**Contact: karunark@yahoo.com*

INTRODUCTION: The incidence of hemoglobinopathies is high in northern and north eastern part of India.
AIM: To estimate the incidence of various haemoglobin disorders and to identify mutations whenever possible.
MATERIALS AND METHODS: From January 2009 to December 2012, patients with anaemia who were clinically suspected and/or families of known patients were screened. HPLC (high performance liquid chromatography) was done using Bio-Rad variant. Reverse dot blot hybridization (RDB) was used for identifying mutations.
RESULTS: A total of 2137 patients were screened, among which 842 patients (39.4%) were diagnosed with hemoglobinopathies. Thalassemia group (major, trait, coexistent variants and intermedia) was the highest (n. 612 - 28.6%) followed by sicklecell group (n.135 - 6.3%). 533 patients were diagnosed with Thalassemia trait. The rest comprised of 95 cases which were haemoglobin C, D and E including both homozygous and heterozygous cases. The number of patients with haemoglobin E was more compared to haemoglobin C and D. For 90 patients mutation study was done. The common mutations observed were IVS 1-5G-C and Codon 15. In 3 patients with thalassemia trait, RDB done showed no known common mutations.

CONCLUSIONS: The incidence in the current study is 4% which is slightly higher compared to the previous reports (1-3%). The pattern of mutation indicates a strong Dravidian influence. Socio economic mobility had a strong influence in the incidence indicated by presence of increase in Hb E and D cases. A slow and steady rise indicates efforts by individual institutions are not adequate to address the issue. In India which is ethnically and socially diverse, for the optimum delivery of screening and prevention programmes, a structured intervention at state level is required.

Key words: hemoglobinopathies, HPLC, screening programs.

PP09

PREVALENCE OF HAEMOGLOBINOPATHIES IN THE NEWBORN: A STUDY ON CORD BLOOD BY HPLC

*Khonglah Y.,[1] Raphael V.,[1] Das N.,[1] Singh A.S.[2]

Departments of [1]Pathology and [2]Obstetrics & Gynaecology, North Eastern Indira Gandhi Regional Institute of Health and Medical Sciences (NEIGRIHMS), Shillong, India

Contact: yookarink@gmail.com

INTRODUCTION: Thalassaemia and haemoglobinopathies are major public health problems worldwide. The prevalence of HbE in South East Asia reaches up to 30-40%. There is also a high prevalence of HbE disease in North East India. However, no study has been done in this region to know the prevalence of HbE status in newborn so that an effective screening tool may be established.
AIM: To know the prevalence of haemoglobinopathies in newborns, using high performance liquid chromatography (HPLC) on cord blood samples.
MATERIALS AND METHODS: Two mL of cord blood was collected in EDTA vial immediately after delivery and subjected to complete haemogram, peripheral blood smear and HPLC using β thalassaemia short programme.Following cut-off values were used for the diagnosis of common haemoglobinopathies in cord blood:

Type of haemoglobinopathy	HbA2	HbF	HbA
HbE heterozygous	0.5-4.1%	>80%	>2.9%
HbE homozygous	>4.2%	>80%	>2.9%
β-thalassaemia major	-	>80%	0-0.8%
β-thalassaemia minor	-	>80%	0.8-2.8%
β-thalassaemia/HbE	>0.5%	>80%	0-2.8%

RESULTS: 504 cord blood samples were evaluated. The prevalence of haemoglobinopathy was found to be 23.21%. HbE heterozygous was the most common haemoglobinopathy followed by HbE homozygous. Cases of β-thalassaemia minor, β-thalassaemia major and β-thalassaemia/HbE were also noted.
CONCLUSIONS: This study showed a high prevalence of haemoglobinopathy in neonates in this region necessitating an appropriate screening strategy for newborns. We also conclude that HPLC is a sensitive technique for studying haemoglobinopathies in newborn and may be utilised for screening.

Key words: HPLC, screening, hemoglobin E.

PP10

THALASSEMIA INTERMEDIA - AN ENIGMA TO BE SOLVED

*Latha F.J., Karuna R.K., Parimala P., Shanthala A.M., Sitalakshmi S.

Department of Clinical Pathology, St. John's Medical College Hospital, Bangalore, India

Contact: fathimashaju@rediffmail.com

INTRODUCTION: Thalassemia intermedia is characterised by haemoglobin levels around 7-10 gm/dL with/without the need of regular transfusions, more severe RBC abnormalities than thalassemia minor, varying degrees of splenomegaly and increased susceptibility to infections reflecting a spectrum between major and minor.
AIM: To study the incidence and the clinico pathological profile of thalassemia intermedia.
MATERIALS AND METHODS: All cases which came for evaluation of anemia from January 2009 to December 2012 were reviewed. The peripheral smears, high performance liquid chromatography (HPLC) patterns along with red cell indices were analyzed.
RESULTS: Among 2157 cases screened, 24 cases were diagnosed as TI (1.1%). The age of presentation ranged from 3 to 50 years (M:F-11:13). The main clinical presentation was fatigue followed by repeated infections. Splenomegaly was found in all cases. Haemoglobin values ranged from 3.8 gm% to 10.6%. Fetal haemoglobin values ranged from 7.9 to 98.2%. 6 of them (25%) of them needed repeated transfusion once in 6 months. Others needed transfusion rarely (pregnancy, prior to surgical procedures) Among 9 cases where mutational analysis was done, IVS 1-5 GC (N-5) and Codon 15 (N-4) were observed.
CONCLUSIONS: The incidence of TI was 1.1%. among patients who were screened, while among patients with hemoglobinopathies it was 2.8%. The age at onset is higher compared to other Indian studies, while range of hemoglobin and fetal hemoglobin values and transfusion history are similar to other studies. With a wide spectrum of clinical presentation and severity, clear guidelines for optimal management plan are yet to evolve.

Key words: thalassemia intermedia, HPLC, fetal haemoglobin, transfusion history.

PP11

TITLE: δ-β-THALASSEMIA CASE REPORT OF TWO FAMILIES FROM SOUTHERN INDIA

*Shanthala Devi A.M., Parimala P., Karuna R.K., Sitalakshmi S.

Department of Clinical Pathology, St. John's Medical College Hospital, Bangalore, India

**Contact: shanthalaam@gmail.com*

INTRODUCTION: δ-β-thalassemia is characterized by elevated levels of fetal hemoglobin (HbF). Homozygous δ-β-thalassemia manifest clinically as thalassemia intermedia but not as severe as β-thalassemia. Only few cases of δ-β-thalassemia are reported from India. We report for the first time two families with this disorder from Karnataka.
CASE REPORT: Among the two families that were studied all the children were affected (two siblings in both the families). Age at presentation ranged from 7 to 22 yrs. Clinically all presented with anemia, jaundice and splenomegaly. Hemoglobin ranged from 9.0-12.9 g/dL. All cases had microcytic hypochromic anemia. Siblings of one faily were on regular blood transfusions, among whom one sister had splenectomy done. Hemoglobin analysis on high performance liquid chromatography (HPLC) showed HbF ranging from 88.6%-95.5%, HbA2 0%-4.5%. Investigations of the parents showed thalassemic red cell indices with increased levels of HbF ranging from 18.7%-22% and HbA2 2.4%-2.9%, features consistent with δ-β-thalassemia trait. Mother of one of the families had features of β-thalassemia trait with HbA2 4.6%.
CONCLUSIONS: δ-β-thalassemia is a rare disorder resulting in an increased percentage of HbF in adulthood. Clinical and hematological parameters should be evaluated for accurate diagnosis and to exclude hereditary persistence of fetal hemoglobin which also shows increased HbF levels.

Key words: δ-β-thalassemia, fetal hemoglobin.

PP12

CO-EXISTENT HBQ INDIA - β-THALASSEMIA TRAIT: CASE REPORT OF AN UNCOMMON VARIANT

*Parimala P., Shanthala Devi A.M., Karuna R.K., Sitalakshmi S.

Department of Clinical Pathology, St. John's Medical College Hospital, Bangalore, India

**Contact: puttaiahparimala@yahoo.com*

INTRODUCTION: HbQ India is a rare α chain variant and usually presents in the heterozygote state. Normally it is clinically silent. It becomes symptomatic when associated with other conditions like β-thalassemia, α thalassemia, Hb E or Hb H. We report one such case in a migrant Punjabi Khatri family.
CASE REPORT: A thirty three year old male presented with fatigue, weakness and body ache for two months. He was found to have pallor and mild jaundice (unconjugated bilirubinemia). He was diagnosed outside as double heterozygous for HbD-β-thalassemia on hemoglobin electrophoresis. Hematological investigations showed Hb 13.9 g/dL, MCV 56.1 fL, RBC count 7.15 m/μL, RDW 19% and microcytic hypochromic blood picture. Hemoglobin analysis on high performance liquid chromatography (HPLC) showed a unknown peak with a retention time of 4.76 min (9.6%) along with HbA2 of 5.7%. Investigations of father showed normal hematological parameters and a similar peak on HPLC (18.3%). Mother was not available. A final diagnosis of double heterozygous for HbQ India - β-thalassemia with parent being carrier was done.
CONCLUSIONS: HbQ India with co-existent β-thalassemia trait is a very rare variant of thalassemia. Such a rare entity can be confused with HbS and HbD. A careful screening with HPLC and molecular techniques should be performed in cases with band in S,D, G region on hemoglobin electrophoresis and negative sickling test to exclude Hb Q India.

Key words: hemoglobin Q, α chain.

PP13

HEREDITARY PERSISTENCE OF FETAL HEMOGLOBIN. A CASE REPORT

*Sitalakshmi S., Parimala P., Latha F., Shanthala Devi A.M., Karuna R.K.

Department of Clinical Pathology, St. John's Medical College Hospital, Bangalore, India

**Contact: slvbs@yahoo.co.in*

INTRODUCTION: Hereditary persistence of fetal hemoglobin (HPFH) is characterized by persistence of elevated levels of hemoglobin F in adult life in the absence of any anemia. Only a few cases are reported. During a routine antenatal screening for pregnancy this rare disorder was detected.
CASE REPORT: A 25-year old female on a routine antenatal checkup was found to have mild jaundice and splenomegaly. Hematological investigations showed Hb 12 g/dL, MCV 75.3 fL, Retic 9.5%. Peripheral smear showed normocytic normochromic blood picture with polychromasia. Patient had a past history of recurrent attacks of jaundice. Similar complaints were present on the paternal side. HPLC showed HbF 100%. In view of high levels of HbF in the mother, son was investigated at 2 years of age. He was asymptomatic and hematological findings were Hb 13.2 g/dL, MCV 63 fL with microcytic hypochromic blood picture. HPLC showed HbF 23.6% and HbA2 2.2%. As the patient was clinically silent a final diagnosis of homozygous HPFH and heterozygous HPFH on the child was made.
CONCLUSIONS: Hereditary persistence of fetal hemoglobin (HPFH) is a rare disorder with no apparent clinical abnormality. Therefore a careful screening by HPLC and molecular techniques are required to detect and confirm these disorders.

Key words: hereditary persistence of fetal hemoglobin, HPFH.

PP14

SICKLE HEPATOPATHY. UNCOMMON PRESENTATION OF INFREQUENT COMPLICATION

*Poornima D.R., Karuna R.K., Parimala P., Prakash A.

Departments of Clinical Pathology and Pediatrics St. John's Medical College Hospital, Bangalore, India

**Contact: drpoornima.ashok@gmail.com*

INTRODUCTION: Intrahepatic sequestration is an uncommon complication of sickle cell disease and occurs in homozygous sickle cell anemia.

CASE REPORTS: Case #1: A 7-year old boy (non consanguineous marriage, sibling not affected) presented with fever of 5 days duration. On examination, he was very pale, icteric, had hepato splenomegaly (7 cm below costal margin) and lymphadenpathy. 8 months back had a similar episode with history of blood transfusion. 10-year old sibling is asymptomatic. Significant laboratory results: haemoglobin: 3 gm%. Corrected total count 22,800/μL corrected reticulocyte count: 7.75% high conjugated hyper bilirubinemia; enzymes not elevated, smear for malaria was negative.

Case #2: A 5-year old boy (consanguineous marriage, sibling with H/o blood transfusion) presented with fever of 5 days duration. H/o transfusion of 3 years back. On examination, pale, icteric and had hepatosplenomegaly (4 cm below the costal margin). Significant laboratory results: haemoglobin: 8.9 g/dL; conjugated hyperbilirubinemia (12.8/10.8); enzymes elevated (608/255); reticulocyte count: 1.75%. HPLC done on both patients showed features of sickle cell-β thalasemia. Both patients did not have gallstones on ultrasound evaluation.

CONCLUSIONS: Sickle cell anemia usually presents with mild splenomegaly and unconjugated hyperbilirubinemia. Acute sickle hepatic crisis occurs in approximately 10% of patients with sickle cell anemia and relate predominantly to vascular occlusion with acute ischemia and sequestration. These two cases with hepatosplenomegaly and conjugated bilirubinemia highlight the varied presentation of sickle hepatopathy in these patients with sickle-β-thalassemia.

Key words: sickle cell anemia, co-existent β-thalassemia, hepatopathy.

Indian National Conference on Hemoglobinopathies

17-18 May 2013, Bangalore - India

CLINICAL POSTERS

CP01

SPECTRUM OF SICKLE CELL DISEASE IN PATIENTS DIAGNOSED AT INDIRA GANDHI INSTITUTE OF CHILD HEALTH ALONG WITH THEIR FAMILY SCREENING AND SPECIAL EMPHASIS ON CLINICO-HEMATOLOGICAL PROFILE OF PATIENTS

Hemalata L., *Pradeep R., Premalatha R., Viswanath V., Rajashekar Murthy G.R.

Indira Gandhi Institute of Child Health, Bangalore, India

**Contact: docpradi@gmail.com*

INTRODUCTION: Sickle cell anemia is the most common heritable hematological disease affecting humans. Because of their prevalence and worldwide distribution, disorders resulting from sickle haemoglobin are of enormous clinical importance.
AIM: The aim of this study was to identify the spectrum of all sickle cell diseases diagnosed at Indira Gandhi Institute of Child Health, Bangalore and to screen parents and siblings of the patients for their carrier status.
MATERIALS AND METHODS: 26 children diagnosed to have sickle cell disease over a period of five years (2008 to 2012) from inpatient and outpatient departments of IGICH, Bangalore were studied. 38 Parents and 10 siblings of these children were studied for their carrier status. Standard methods were used for routine hematological and biochemical investigations. Hemoglobin electrophoresis was done by alkaline gel method.
RESULTS: Age of the children ranged from 1 ½ years to 14 years. 12 out of 26 children presented in hemolytic crisis, one child presented in aplastic crisis and the others presented with constitutional symptoms. 11 out of 26 children were diagnosed with sickle cell anemia, 8 were diagnosed with sickle thalassemia and 7 were diagnosed with sickle cell trait. Among the parents and siblings, 40 were found to have sickle cell trait, 7 were found to have thalassemia trait and one was electrophoretically normal.

Age group (years)	Number of children
0-5	08
5-10	12
10-15	06

Gender ratio (male/female): 1.9.

CONCLUSIONS: Sickle cell anemia was the most common sickle cell disease in the patients followed by sickle thalassemia and then sickle cell trait. Sickle cell trait was the most common entity among the parents and siblings, followed by thalassemia trait.

Key words: sickle cell anemia, thalassemia.

CP02

MORBIDITY PROFILE OF SICKLE CELL DISEASE IN CENTRAL INDIA

Yadav R., Singh M.P.S.S., Vishwakarma C.P., Lal Neelkar R., Gupta R.B., *Rajasubramaniam S.

Regional Medical Research Center for Tribals, Jabalpur, India

**Contact: raja.rmrct@gmail.com*

INTRODUCTION: Sickle haemoglobin (HbS) in Madhya Pradesh is mainly confined to the tribal predominant areas. Out of fifty, thirty (32) two districts of MP (comprising of 240 blocks/ taluk) have sickle hemoglobin. The frequency of sickle cell trait varies from 3 to 35%.
MATERIALS AND METHODS: Till date no facilities for diagnosis exist at the district or PHCs. The patients remain undiagnosed. The Regional Medical Research Center for Tribals established the first clinic to identify and provide necessary aid to sickle cell disease patients at the NSCB Medical College, Jabalpur. The sickle clinic will identify the clinical profile and natural history of sickle cell disease in Central India and develop a simple protocol for its management and prevention.
RESULTS: A total of 691 sickle cell disease patients were registered in sickle cell clinic between 2002 and 2012. Majority of the registered patients were males (68.7%). About 46.7 % belonged to Scheduled caste category, Backward Class (29.1%) and Scheduled tribe (14.2%). About 73.1% of patients were demonstrated Splenomegaly. Splenectomy was observed in about 1.2 % of the patients. The most frequent clinical symptoms observed these patients was pallor (95.1%), Icterus (85.1%), joint pains (80.8%), fever (76.0%), bony pains (61.2%), abdominal pain (38.8%), joint swelling and chest pain (21.0%). General weakness was observed among (81%) patients. Nearly 50% of the patients reported at least one blood transfusion during their follow-up period. History of multiple blood transfusions was observed in about 16.5% cases. Majority of the patients (69.0%) reported onset of the disease prior to attaining the age of 6 yrs. Seventeen % patients showed first manifestation of the disease on or after 9 years of age.
CONCLUSIONS: A reduction in severity of the disease was observed in the majority of cases post intervention. Percentage of severe cases were reduced and shifted to mild category consequent to intervention.

Key words: sickle cell anemia, tribal population.

pagepress

CP03

SICKLE CELL ANAEMIA PRESENTING AS ACUTE RENAL FAILURE AND ACUTE AXONAL NEUROPATHY

Murthy M., Lakshmi D., Kavitha S., *Thivya R.

Department of Pathology, Coimbatore Medical College Hospital, Coimbatore, India

Contact: drrthivya@gmail.com

INTRODUCTION: Sickle cell disease is an inherited chronic haemolytic anaemia whose clinical manifestations arise from the tendency of the haemoglobin to polymerize and deform red blood cells into the characteristic sickle shape. In our country it is more common in tribals of Central India. Sickle cell anaemia in homozygous state (HbS is more than 70%) usually present in early childhood.
AIM: To evaluate a case of 42-year old male presented with complaints of diarrhea for past 2 days and sudden onset of weakness in both lower limbs and reduced urine output for past one day. He was apparently normal before.
MATERIALS AND METHODS: After the clinical examination, routine blood investigations including complete Hemogram and peripheral smear were done in addition to electrophoresis. Ultrasonogram, MRI Spine and Nerve conduction study were carried out in view of his weakness of lower limbs.
RESULTS: On examination the patient was anaemic, Icteric and dehydrated. Hepatomegaly was mild. Tone and power were reduced in lower limbs, deep tendon reflex and plantar reflex were absent in both lower limbs. Ultrasonogram showed hepatomegaly with medical renal disease. MRI Spine was normal. Nerve conduction study done showed severe Axonopathy affecting motor nerves with demyelination. Biochemical investigaions including blood urea, serum creatinine, serum bilirubin, serum LDH were elevated. Complete hemogram showed presence of anaemia, neutrophilic leucocytosis with increased reticulocyte count Peripheral smear picture clinched the diagnosis as sickle cell anaemia with leukemoid reaction. Electrophoresis confirmed the diagnosis with HbS (79.7%), HbA (2.1%), HbF (14.2%), HbA2 (4%).
CONCLUSIONS: Sickle cell anaemia in hemolytic crisis with acute renal failure and acute axonal neuropathy.

Key words: sickle cell anemia, axonal neuropathy, haemolytic crises.

CP04

ENCOURAGING RESULT OF HYDROXYUREA IN Eβ AND THALASSEMIA INTERMEDIA: EXPERIENCE FROM A SINGLE CENTRE

*Chopra Y., Ramzan M., Prakash Yadav S.

Pediatric Haematology and Bone Marrow Transplant Unit, Fortis Memorial Research Institute, Gurgaon, Haryana, India

Contact: yogiraj.chopra@gmail.com

INTRODUCTION: Hydoxyurea is a promising drug in thalassemia intermedia and Eβ-thalassemia to increase transfusion free interval.

MATERIALS AND METHODS: We retrospectively analysed data regarding clinical efficacy and long term toxicity of hydroxyureain children with thalassemia intermedia. Complete blood count was monitored monthly.
RESULTS: We had 3 patients of thalassemia intermedia and 3 of Eβ-thalassemia (4 male and 2 female). Mean age at diagnosis was 3.5 years (range: 1.0-7.5 years). Mean age for first transfusion was 2.59 years (1 to 5.75 years) except one child who received no transfusion before starting hydroxyurea. Mean haemoglobin (Hb) level at the time of starting. Hydoxyurea was 5.8 g/dL (5 to 7 g/dl). Mean dose of hydroxyurea used was 18.33 mg/kg (15-20 mg/kg). One patient of Eβ-thalassemia was started on hydroxyurea after rejection of bone marrow transplant, who also did not respond to hydroxyurea for 12 months and was restarted on regular transfusions. Another patient of thalassemia intermedia responded to hydroxyurea therapy which was started at 1.5 years of age. He achieved mean Hb of 8 g/dL and remained transfusion independent till the age of 4.5 years when he dropped his Hb to <7 g/dL and had growth faltering so was restarted on transfusions. Rest four patients responded to hydroxyurea. Their mean Hb level were 8.16 mg/dL (7-10 g/dL). Mean transfusion free interval on hydroxyurea therapy was 7.5 months (6-21 months). No adverse effects were noted in these patients due to hydroxyurea.
CONCLUSIONS: Results are encouraging and hydroxyurea is a valuable and promising drug even in Eβ-thalassemia.

Key words: hemoglobin Eβ-thalassemia, hydroxyurea.

CP05

STUDY OF PULMONARY FUNCTION TESTS IN MULTITRANSFUSED CHILDREN WITH THALASSEMIA

Bhagyalakshmi S., Mallya P., Jyotirmanju C.S., Somshekhar A.R., *Rau A.T.K.

Department of Pediatrics and Physiology, M.S. Ramaiah Medical College, Bangalore, India

Contact: atkrau@gmail.com

INTRODUCTION: Thalassemia is a quantitative disorder of haemoglobin production. It is a multisystem disorder which mainly affects the heart, liver, endocrine and bone and is treated with regular blood transfusions and chelation therapy. A number of recent studies have also reported alteration in pulmonary functions with this disorder which have been ascribed either to the disease or its therapy. Affected children frequently develop pulmonary complications and subsequently growth retardation. This study was undertaken to assess pulmonary function in thalassemic children who had received regular transfusions and chelation therapy.
AIM: To study pulmonary function tests (PFT) in multitransfused children with thalassemia and compare them with normal children.
MATERIALS AND METHODS: Pulmonary function tests were done on 35 children with β-thalassemia major attending the thalassemia clinic of a teaching hospital

over a period of 1 year. Serum ferritin and haemoglobin levels were also estimated in these children. The PFT was compared with 35 age and sex matched controls. Student's *t* test was used to compare the PFT parameters between these groups and the Pearson's correlation analysis was used to correlate various PFT parameters with serum ferritin, number of blood transfusion received and duration of chelation therapy.

RESULTS: Mid peak expiratory flow (PEF) in thalassemic children was significantly lower than the control group (P=0.023). Among the thalassemic children, 27.71%, 14.28% and 2.85% had restrictive, obstructive and a mixed pattern of abnormalities respectively. In the study subjects, the assessed lung volumes (forced expiratory volume in 1 second to forced vital capacity (FEV1/FVC) and PEF, PEFR inversely correlated with serum Ferritin levels (r=0.23, 0.12, 0.09, respectively). Similarly, all measured lung volume parameters (FEV1, FVC, PEFR) also inversely correlated with cumulative blood transfusions given (r=0.20, 0.27, 0.276, respectively). Among the study group, children who were chelated early were found to have better lung functions (FEV1, FVC, PEFR) than those chelated late or not at all (r=0.27, 0.48, 0.25, respectively). This correlation was particularly significant between FVC and duration of chelation therapy (P=0.014)

CONCLUSIONS: Our study found a significant reduction in lung volume parameters in chronically transfused thalassemia major patients. Iron overload and number of blood transfusions given correlated with deterioration in lung functions but early initiation of chelation therapy correlated well with better lung functions when compared to those in whom chelation was started late or not at all.

Key words: thalassemia, transfusions, lung function, iron chelation.

Index of authors

PAGEPress Publications, Pavia, Italy I www.pagepress.org I