Dr Shaikha Salim Al Arrayed, MBChB,DHCG,PHD
Chairperson of Genetic Department, S.M.C, Bahrain.
She had M.B.Ch.B from Cairo university, MSC Human and Clinical Genetics UK 1984, and Ph.D. in Genetic in 1993 from Aberdeen University, She is the Head of National committee for the control of Hereditary diseases since 1993.
She is Member of WHO Expert Advisory Panel on Human Genetics since 2002. Member of American College of Medical Genetics, and Member of Hugo since 1998.
She is founding Member of many societies, and chairperson of Bahrain National Hereditary anemia society.

She organized many National projects such as premarital services, Student Screening for Genetic Blood disease, Newborn screening, and Bahrain Birth Defect Register.
She was awarded the (The Kingdom Competence Order of the first class) in 2004, and was a nominee for the international prize of (1000 women for the Nobel peace Prize)
she has been awarded (the State of Kuwait Prize for Research in Health Promotion 2009), by the WHO.
In 2012 she was Awarded Dr A. T. Shousha Foundation Prize 2012. Following the recommendations of the Dr A.T. Shousha Foundation Committee at the Fiftyeighth Session of the Regional Committee for the Eastern Mediterranean, the Executive Board awarded the Dr A.T. Shousha Foundation Prize for 2012, by decision EB130 (9),in Cairo Egypt
She published more than 60 scientific papers, and attended more than 150 international and national conferences.

Abstract:
Title: Bahrain Success Story in Controlling Sickle Cell Disease
1984-2013
- The goals of this campaign were to reduce the incidence of hereditary diseases in Bahrain, and to improve the standard of management for patients suffering from these diseases.
the campaign to control genetic disease in Bahrain was organized in the period 1984-2013
The prevention strategy depended on health education, screening and counseling. A comprehensive health education program has been launched, to increase public awareness of the diseases and methods to avoid them.
This program used the media, and targeted key opinion leaders in society and the community, in schools and other public places. Screening for haemoglobinopathies included sickle cell disease, thalassaemia, was undertaken on the following categories of the population: antenatal mothers, premarital couples, newborns, and school students, followed by counseling of families. The campaign was supported by both the policy makers and the community.
These efforts continued for more than 25 years. It had tremendous effects in reducing the prevalence of Genetic Blood Diseases (GBD) among the newborns, in 1984 the incidence of SCD among newborn was 21 per thousand, now it is 4 per thousand with more than 75% decline .
-Consanguinity rate also declined gradually due to increase awareness about genetic diseases. The total consanguinity rate in 1990 was 40%, while in 2009 it became 11.3% % with more than 70% decline.
During this campaign the Ethical legal and Social issues were taken care of, such as: equity, informed consent, privacy, confidentiality and prevention of stigmatization and discrimination.

Prenatal diagnosis in Bahrain

Dr Shaikha Al Arrayed ,Dr Amal Hassani

Introduction :

Prenatal diagnosis is testing for conditions in a fetus before it is born. The aim is to detect birth defects such as neural tube defects, Down syndrome, chromosome abnormalities, genetic diseases, such as sickle cell anemia, thalassemia, cystic fibrosis, Muscular dystrophy.

Common testing procedures include chorionic villi biopsy ,amniocentesis, ultra sonography, and serum marker testing, followed by genetic screening.

In KSA, in 1990 ruling (Fatwa) allows termination of pregnancy in the first 120 days after conception if the fetus is shown beyond doubt to be affected with a severe malformation that is not amenable to treatment. The Islamic republic of Iran has same Fatwas for certain condition such as thalassemia .

This service is available in many Islamic and Arab countries such as Turkey, Iran, Pakistan Palestine, Jordan, Egypt, Syria ,United Arab Emirates , Saudi Arabia, Tunisia, Iraq, and Gaza. It is available internationally in countries such as India, Malaysia, south East Asia, Canada, Europe, and USA etc.

In Bahrain the service started in 2002, in a private setting, where prenatal testing only is provided.

Material and method:

Patients were referred to the private clinic from their obstetricians. Indications being at risk for chromosomal abnormalities such as advanced maternal age , or presence of abnormal ultrasound finding . The other Indications being at risk of getting affected babies with SCD or betathalassemis .

Prenatal testing is done early in pregnancy either by CVS at (11 weeks) or amniotic fluid testing at 14 weeks.

Samples were sent to genetic laboratories where cytogenetic or molecular testing was performed. Result was available within 7-10 day.

This service is provided in many other hospitals and clinics in Bahrain now.

Results

Total number of patients who undergo PND during the last 10 years in our clinic was 150 patients. Fifty patients were for chromosomal testing and 100 patients for genetic blood diseases.

Number of affected fetuses was low. In the chromosome cases, it was 4 babies/50 =8%. Among blood disease it was 5 babies/100 = 5%.

Genetic counseling provided prior to testing and after getting the results

Conclusion

PND service aim is to ensure the well being of babies and mothers, it also aim is to give the parents and healthcare staff the chance to prepare medically, psychologically and socially for the delivery of a child with a health problem It allow couple to have further healthy babies. The affected babies' number in our series was low. Further action has to be decided by the couple themselves.

Dr. Michael Angastiniotis, Cyprus
Nationality: Cypriote
Education: Royal College of Physicians of Glasgow,1976, DCH (Diploma in Child Health, University of Aberdeen,1960 – 1966 ,MB, ChB
Training:
Duncan Guthrie Institute, Glasgow, 1986, Medical Genetics
University College Hospital, 1978 – 79, Prenatal Diagnostics
Royal College of Physicians of Glasgow, 1976, Postgraduate Course in Paediatrics

Abstract:

Abstract
Effective Thalassaemia Control: the Cyprus experience
Michael Angastiniotis –Thalassaemia International Federation

In high prevalence areas, haemoglobin disorders are a significant public health issue, which necessitates planning and budgetary support in order to provide optimum patient care and establish preventive services. An example such a comprehensive national programme exists in Cyprus since the 1970s. The disease was recognised on the island in the 1940s but since transfusion started in the 1960s, patient survival improved and so the numbers increased. WHO assistance was requested by the government since the increasing demand for blood, drugs and general costs was projected to rise to unmanageable levels over the next 20 year period. On WHO recommendation in 1972, a 5 year national plan was developed and budgeted, which included a programme to limit new affected births and to improve patient care.

The prevention programme consisted initially on raising awareness, screening to identify carriers, and counselling at-risk couples. Over the years prenatal diagnosis and pre-implantation genetic diagnosis were offered as technology allowed. This programme has led to an over 90% reduction of new affected births.

Concerning patient care, the main issue in the 1970s was to increase the voluntary non-remunerated blood donation. For this purpose a committee was formed responsible to plan a blood donation campaign and also to plan a public education campaign. Members of this committee were doctors and parents of thalassaemia patients as well as other public figures. They have succeeded in making Cyprus self-sufficient in blood (not only for thalassaemia). A group of parents also formed which advocated services for patient care. In 1978 the Council of Ministers approved the free provision of services to all haemogolbinopathy patients as well as free prevention. This relieved families from a heavy burden. Patient care increased over the years to include all new monitoring technologies and all new treatment modalities.
With these comprehensive policies patient survival has been shown, through the national register, to improve with succeeding birth cohorts and life achievements such as education, marriage, parenthood and employment are now a reality for many of the patients.

Abstract 2
Prevention of haemoglobin disorders in the Middle East
Michael Angastiniotis – Thalassaemia International Federation
The need for prevention of haemoglobin disorders has been recognised in most counties of the Middle East where these genetic diseases are highly prevalent. Reduction of new affected births is of benefit to

the whole community but most importantly to the families at-risk and to the patients themselves by saving resources for their needs in terms of proper treatment. The East Mediterranean region expects, overall, around 1800 annual births with sickle cell syndromes, and 9000 thalassaemia births. The numbers of patients are not accurately known since few countries maintain national registries, however the nearest figures that countries can provide sum up to over 42000 SCD patients and over 100000 thalassaemia patients.

A prevention programme includes awareness raising, screening to identify carriers and genetic counselling. A full control programme which includes a comprehensive plan for patient care exists only in about 4 countries (Iran, KSA, UAE and Bahrain), while another 4 countries have national prevention policy. In 7 other countries of the region prevention is possible but not part of a national programme and 5 have no plans at all. 15/20 countries have awareness raising activities but prenatal diagnosis is available in only 6. In most countries in the region the approach is that of mandatory premarital screening with the aim of reducing at-risk marriages. This is favoured by the culture of arranged and consanguineous marriage and it helps in the avoidance of pregnancy termination. Such aprogramme is now showing positive results in the Kingdom of Saudi Arabia (Memish ZA, et al. Ann Saudi Med, 2011)

Prof. Dr. Graham Serjeant, Jamaica

Nationality British

Qualifications: MRCS (Eng), LRCP (London) 1963,
MRCP (London) 1966, FRCP (London) 1977

Abstract:

DEDICATED SICKLE CELL CENTRES; IS THIS A MODEL FOR BAHRAIN?

Sickle cell disease is a lifelong condition of variable severity requiring frequent monitoring and medical support. The current structure for most health care services is that children attend a paediatrician until an arbitrary age varying from 12-25 years, depending on the institution, when they are transferred to adult physicians. Many patients fail to make this 'transition' at adolescence, a critical time requiring care for increasing painful crises, leg ulcers, priapism, problems of pregnancy, enuresis, and retarded growth. Is such a transition necessary? The retarded physical and sexual development of many patients with SS disease makes an arbitrary age of transfer illogical. Why not provide a seamless service regardless of age at a dedicated centre for sickle cell disease? Optimal care for patients with the disease requires regular review even when in good health, in order to establish 'steady state' features clinically and haematologically. Large amounts of data are accumulated and software programmes should make this readily available for patients care. A day care centre allows management of many painful crises and of outpatient transfusion. Special investigations such as transcranial Doppler and assessment by orthopaedic surgeons and other specialists can be performed within such centres accumulating experience in sickle cell disease and avoiding patients having to attend multiple clinics which, in addition to the inconvenience, creates problems of collating information from different sources. In Jamaica, the dedicated centre works well, the patient is reassured by seeing the same nurses, technologists, social workers and psychologists and as they reach the age of 'transition' elsewhere, they simply go to the doctor in Room 2 instead of Room 1. (265 words)

THE PAINFUL CRISIS OF SICKLE CELL DISEASE; JAMAICAN OBSERVATIONS.

Haematological changes in sickle cell disease result in recurrent tissue damage compromising function in the spleen, lungs, brain and kidneys. The bone marrow because of its high metabolic demands, may also develop areas of necrosis. The incidence and significance varies with the patients' age causing dactylitis (hand-foot syndrome) in young children, and a spectrum of conditions in adolescence and young adults including the 'bone pain crisis', and avascular necrosis of the femoral and humeral heads. Bone pain crises typically commence in later childhood, increase in frequency in adolescence and early adult life, especially in males, reduce in frequency and severity at later ages and often cease entirely in patients after the age of 30-40 years. This reflects the distribution of metabolically active bone marrow, which after childhood, occupies the juxta-articular areas of the long bones, ribs, sternum, and vertebral bodies. Enquiries of activities in the 24 hours preceding pains may reveal potential precipitating factors including cold exposure, infections, dehydration and stress which, if avoided, may prevent some painful crises. Longer term risk factors include the last trimester of pregnancy and immediate post partum period and a high steady state haemoglobin, especially in the presence of a low fetal haemoglobin. Once established, the bone pain crisis requires correction of poor hydration and pain relief usually conducted according to the WHO analgesic ladder. Analgesic requirements may be reduced by other

social support mechanisms, an understanding of the origin of bone pain, that it is not life threatening and in most patients becomes less with advancing age. A subgroup of patients, with frequent unexplained pains, demonstrate some features of drug dependence, and often have complex social and psychological problems. (276 words)

Dr. Hanan Hamami, Geneva

Abstract :

<u>Epidemiological profile of sickle cell disease in Arab countries</u>

Sickle cell disease (SCD) is known to be a prevalent inherited disorder in most Arab countries with varying prevalence rates and molecular characterization. The rates of sickle cell trait in Arab countries ranges from 0.3 to 30%, with the Benin, the Arab-Indian and the Bantu haplotypes constituting the bulk of the haplotypes, leading to two major phenotypes; a mild one associated with the Arab-Indian and a severe one with the Benin and Bantu haplotypes. Public health approaches targeting prevention of SCD in Arab countries include newborn screening for sickle cell disease, and premarital screening for carriers of the sickle cell gene. These services are still patchy and inadequate in many Arab countries recommending the upgrade of these services with strengthening of the education and training of health care providers and raising public awareness on the feasibility of prevention and care for sickle cell disease.

Isaac Odame, **MB ChB, MRCP (UK), FRCPCH, FRCPath, FRCPC, Canada**

Dr. Isaac Odame is the Medical Director of the Global Sickle Cell Disease Network (GSCDN), under the auspices of The Programme for Global Paediatric Research (PGPR) at SickKids. Dr. Odame is an academic clinician in the Division of Haematology/Oncology and co-director of the haemoglobinopathy program at The Hospital for Sick Children (Sick Kids). He is a project investigator with the Research Institute at Sick Kids and Associate Professor of Paediatrics, University of Toronto

Dr. Odame was instrumental in the design and implementation of universal newborn screening for SCD in Ontario. He sits on the Maternal and Child Screening Committee for Ontario as the haemoglobinopathy expert. He chairs the Canadian Haemoglobinopathy Group, a body of all haemoglobinopathy physicians, scientists and nurses in Canada.

He was formerly, Staff Paediatric Haematologist/Oncologist and Associate Professor at McMaster University in Hamilton, where he was also the Director of the Residency Program in Paediatric Haematology/Oncology.

Abstract :

The Global Sickle Cell Disease Network: Progress and Future Plans

Isaac Odame, Hospital for Sick Children, Toronto, University of Toronto, Canada Global SCD Network

The Global Sickle Cell Disease Network (GSCDN) established in 2009 has a mission to foster collaboration between SCD clinicians and scientists to further research and advance clinical care for SCD globally, particularly in low-income countries with the highest disease burden. GSCDN aims to catalyze North-South and South-South partnerships in research, training and education and the enhancement of clinical programs. An International Advisory Council made up of world-renowned SCD leaders provides strategic guidance for GSCDN and supports its partnership and advocacy efforts. Working Groups on newborn screening, infectious diseases in SCD, hydroxyurea therapy in low-income regions, genetic /environmental factors that govern phenotypic diversity and laboratory /data management are at various stages of activity. The Coordinating Office of GSCDN is based at the Hospital for Sick Children, Toronto, Canada.

At the March 2012 GSCDN Conference in Atlanta, USA, the Working Groups held workshops during they discussed tangible areas where collaborative efforts could lead to action. As a result, steps are now advanced to conduct a multicentre pilot study of the safety and efficacy of hydroxyurea in sub-Saharan Africa. Also, a successful survey on leg ulcers proposed at the Atlanta workshop has provided an impetus for conducting a multicentre study of leg ulcers in SCD. A global map of SCD centres, particularly in low-income countries, is providing valuable information about the capacity needs in these regions. This will be valuable information in developing partnerships for capacity building targeted at enhancing clinical care and research, thereby helping to reduce the burden of SCD.

Pain Management in Sickle Cell Disease

Pain is the hallmark clinical manifestation of sickle cell disease (SCD). Severe pain could occur in early infancy and continue through the adult life of the patient. Pain in SCD can be acute, chronic or mixed and may result from tissue injury (nociceptive), nerve injury (neuropathic) or from unknown causes (idiopathic).While improvements in the care of individuals with SCD have resulted in longer life-spans, their lives are impacted even more by the unpredictable intermittent or constant pain that is often poorly manage. To effectively manage pain in SCD barriers to adequate pain assessment and management need to be overcome.

The goals as well as the physiologic, sensory, affective, cognitive, behavioral and sociocultural components of pain assessment process are important considerations in effective pain management. Interventions for pain consist of pharmacologic and non-pharmacologic (psychologic, behavioral and physical) components which should go hand-in-hand. An interdisciplinary approach that brings diverse professionals together for optimal compassionate care enhances the effectiveness of pain management. Many health care institutions now have teams dedicated to acute and chronic pain management. Where available, involvement of these teams at the early stage of pain management leads to better outcomes for patients. Whenever possible, SCD clinicians should play a leading role in the multidisciplinary management of pain.

The Day Hospital and Home Care models of pain management are increasingly being adopted in many SCD centres. These interventions should be patient-focused and family-oriented to ensure appropriate, timely, compassionate care, and thereby prevent and relieve suffering, preserve dignity and quality of life from birth till end of life.

Dr. Rajagopal Krishnamoorthy, France

Name : Rajagopal KRISHNAMOORTHY
Date and place of birth : - INDIA, **Nationality**: French
EDUCATIONAL QUALIFICATIONS : DSc 1979, PARIS VII University, PARIS, FRANCE
PhD 1977, PARIS VII University, PARIS, FRANCE, MSc (Clinical Biochemistry), 1971, University of Madras, Jawaharlal Institute of medicine ,Graduate Medical Education a Research, Tamil Nadu state, INDIA.

PREVIOUS POSITIONS : **INSERM*** 1995 – 2012 Director of Research (1st class), INSERM, Paris, FRANCE
PUBLICATION : More than 250 articles in international peer-reviewed journals and books
FIELDS OF ACTIVITIES - Pharmacogenetics and pharmacogenomics, -Molecular genetics of hemoglobinopathies, -Immunogenetics, -Molecular microbiology, - Other inherited hematological disorders (HLH,CGD...)
COLLABORATIONS:: Extensive experience in mutually beneficial international collaborations in research and molecular genetic technologies, in particular transfer of knowledge and knowhow attested by several co publications with the following collaborating centres; Bahrain, Oman, Qatar, Saudi Arabia, Iran, India, Brazil, Benin, Gabon, Cameroon, Mauritius, Venezuela, UK, USA, Germany etc)

Abstract :

Lessons from the world distribution of Sickle cell haplotypes

Krishnamoorthy R.*[1], Daar S.[2] AlKindi S.[2], Al Zadjali S.[2], Dennison D.[2], Lapoumeroulie C.[1], Pathare A[2].
[1]INSERM U763, Paris, France
[2]Sultan Qaboos University Hospital, Muscat, Oman

Sickle cell mutation arose on five different haplotypes called the “classical β^S haplotypes” and are designated depending upon the geographical location of the original description, into Benin, Bantu, Senegal, Cameroon and Arab-Indian β^S haplotypes. The multicentric origin of sickle cell mutation is believed to reflect a recent mutational event raised in frequency due to selection by malaria at a rate higher than that of meiotic recombination. Two unrelated

subjects sharing a particular haplotype very likely share a common ancestor with respect to this chromosomal block. Recent studies have highlighted that these short range β^S haplotypes (50Kb) indeed represent a high degree of long range haplotype similarity extending over 500 kb region.

Population migration and genetic admixing has introduced the sickle cell gene into many parts of the world and the distribution extends from Indian subcontinent in the east, to Americas in the west with regional-specific patterns. Sickle cell haplotype analysis has been used as a marker to show i) the independent and multiple origin of sickle cell mutation, ii) to identify population relatedness, iii) linked genetic loci in modifying severity at the epidemiological scale (population level) but not as predictors of individual disease severity. Thus knowing the β^S haplotype of a patient per se is not predictive of sickle cell disease severity since several other genetic loci unlinked to the β^S haplotype behave as disease phenotype modifiers. The striking modifier loci include BCL11A on chromosome 2 and HBS1L-MYB on chromosome 6 with respect to HbF expression and alpha globin locus on chromosome 16.

In summary, although the distribution of β^S haplotypes across the world has instructed us on the population affinity / migration and evolutionary adaptation, translation of these data to predict the clinical course at individual patient level is not possible and not recommended.

Genetic Modifiers of Sickle Cell Disease

Krishnamoorthy R.*[1], Lapoumeroulie C.[1], Al Zadjali S.[2], Daar S.[2], Dennison D.[2], AlKindi S.[2], Pathare A[2].
[1]INSERM U763, Paris, France
[2]Sultan Qaboos University Hospital, Muscat, Oman

Autosomal recessive disorders such as sickle cell anemia must, in principle, be clinical rarities but some mutant alleles reach high frequencies due to heterozygote selection with raised homozygous births in inbred/consanguineous communities. When the incidence of such supposedly rare clinical rarities become high and widely distributed, with the improved neonatal care and overall health management, unusual clinical heterogeneity in terms of pattern and severity become manifest. Both environmental and genetic factors do play a role in such diversity of manifestations either by alleviating or aggravating the clinical phenotype to various degrees. Overall, despite being a monogenic disorder, clinical expression of sickle cell anemia is dictated by both genetic and environmental factors (multifactorial). The influence of environmental factors on disease expression is difficult to track down as they vary both in space and time. Whereas the genetic modifiers are much more accessible to research, the study of which has implications in prognosis, prevention, control of complications as well as in treatment response. The proven genetic modifiers of sickle cell disease severity are postnatal expression of HbF and co-inherited alpha thalassaemia both effectively contributing to reduction in the intra-erythrocytic HbS concentration. This results in reduction in HbS

polymerization and hemolysis, which are proximal events of impending distal and long term complications. Genetic loci governing the postnatal HbF expression level have been uncovered in recent years and genome wide or candidate gene association studies have revealed additional loci concerning several sub-phenotypes of sickle cell disease such as stroke, cholelithiasis which will be highlighted. Nevertheless caution is warranted as some of the sub-phenotypes of sickle cell disease may not necessarily have some genetic basis and even if they do, ungovernable environmental influences may outweigh their small contribution.

Dr Salam Alkindi BA, MB, BCh, BAO, DME, MSc, FRCP.
Associate Professor, head- department of haematology
Sultan Qaboos university-Muscat Oman

Following my graduation from Trinity college- Dublin Ireland, in 1993, I have completed my general medicine as well as haematology/ oncology training in Dublin, Ireland as well as the Fred Hutch cancer centre in Seattle USA, where I did my training in Bone marrow transplant. In 1999 I have joined Sultan Qaboos University and in 2005 I was appointed as head of department of haematology. Previously also I held the position of deputy director of Sultan Qaboos university hospital for clinical affairs for about 5 years. Research interests include sickle cell disease, leukaemia and autoimmune disorders with more than 40 articles published in international peer reviewed journals.

Abstract :

<u>Sickle cell disease (SCD) In Oman– from bench to bedside</u>

Dr Salam Alkindi- head- department of haematology, Sultan Qaboos University hospital, Muscat, Oman

Sickle cell disease is a prevalent diseases in Oman with 5.7% of Omani people are carrying the gene and about 0.2% are affected. Although SCD traditionally looked at as predominantly red cell disease, but it's a complex disease demonstrating a model for red cell interactions with white cells and endothelial cell lining. Recent work from our laboratory on acute chest syndrome (which is one of the major causes of death in this disease) and VOC (the most frequent presentation on this disease) just demonstrated this. An alteration in the level of nitric oxide as well as a shift in lymphocytes and monocytes activations plays a role in both conditions. Similarly the altered red cells (sickled cells) leading to perturbed platelets and haemostatic functions plays an important role in stroke development, added to the hereditary component of thrombophilia in this syndrome. These changes are promising an important opening for studies in the various therapeutic interventions that are available for this disease such as hydroxyurea, and more recently low molecular weight heparin, nicosan and other agents that are undergoing testing. Also light is seen at the end of the tunnel with a good progress made in the reduced intensity conditioning (RIC), bone marrow transplant for patients with SCD as seen in the recent experience in our centre, allowing the sickled and normal cells to co-exist together, and the use of stem cells to help patients with avascular necrosis of the hips (AVN), a crippling complication seen in some of our patients.

Dr. Muneer H. Al-Bagshi, KSA.

Nationality : Saudi
M.B.B.S,Arab Board in Pediatrics with its Saudi council for health specialization accredited as PhD Saudi Board,Subspecialty Fellowship of in Pediatric Hematology/Oncology
Fellow of International outreach program at st. jude children cancer research hospital
Current Position : consultant Pediatrician, Hematologist Oncologist and Head of Pediatric Hematology Oncology Unit ,Clinical Assistant Professor of Pediatrics ,Director of Training, Research And Continued Medical Education -Head of Child Protection Team

Abstract :

Hepatic Manifestations of Sickle Cell Disease

Sickle cell disease (SCD) encompasses a group of hemoglobinopathies characterized by a single amino acid substitution in the ß-globin chain. The most frequently occurring form of SCD is sickle cell anemia (HbSS), followed by HbSC and HbSß-thalassemia
Sickling complications occur in all the body organs.
By the age of 2 years over 90% of the organs develop complications, mostly silent and slowly progressive in most cases.
Almost one third of the sicklers have abnormal liver function in their lives and 70% they have high serum bilirubin with no liver disease. The liver is affected by number of complications due to the primary disease or secondary to its complications and therapy
the spectrum of these manifestations are variable from mild form of benign hyperbilirubinemia to a severe life threatening conditions like intrahepatic cholestasis and hepatic sequestration and can leave the patients with morbities. In addition to the vascular complications from the sickling process, patients with sickle cell disease have often received multiple blood transfusions placing them at risk for viral hepatitis, iron overload and(combined with the effects of chronic hemolysis) the development of pigmented stones. The term ''sickle cell hepatopathy'' is used to describe overlaping conditions that have a liver disease in SCD patients. The primary hepatic diseases will be discussed during the presentation.

Sickle Cell Painful Crisis

Sickle cell pain is the most presenting complication in sickle cell disease. The Acute pain syndrome during the course of sickle cell which include acute painful crisis, dactylitis, acute chest syndrome, priapism, avascular necrosis, splenic infarcts, hepatic crisis, and leg ulcers.

Before we start treating the patient for chronic pain or acute on chronic pain, we should understand the origin of the pain and the pathophysiology of pain in sickle cell and the phases of pain during the clinical presentation (four phases in adults and eight phases in children)

The pain is affecting all the systems of the body creating an emergency call for all the hormones in the endocrine system, this tell us to modify our approach toward pain managemet into palliative care policy. The behavior of the patients during and after the acute episode should be reviewed by the treating team in order to distinguish conditions that are high risk to develop addiction like Aberrant drug-taking behaviors, Physical dependence, and the less likely behavior to develop addiction likeTolerance, Drug seeking (Pseudoaddiction)

The understanding of the drugs used in management of pain is vital to improve the pain management and prevent the side effects
All these issues will discussed in details

Dr. Adekunle Adekile, Kuwait

Dr. Adekunle Adekile, Professor and Chairman, Department of Pediatrics, Faculty of Medicine, Kuwait University
Dr. Adekile graduated from University of Ibadan Medical School, Ibadan Nigeria. He had his post-doctoral Fellowship in Pediatric Hematology in Howard University Hospital in 1980-1981 during which time; he worked at the Comprehensive Sickle Cell Center. He established a sickle cell clinic in the Obafemi Awolowo University Teaching Hospital in Nigeria in 1981.
In 1990 worked in the Medical College of Georgia, Augusta, Ga, USA where he worked in Dr. Titus Huisman's laboratory and the Comprehensive Sickle Cell Center. Dr. Adekile moved to Kuwait in 1993 where, he looks after sickle cell patients in the country.
Dr. Adekile also runs a research laboratory .He has a unique perspective on sickle cell disease, having looked after and conducted research on 3 continents.
He has a PhD in Cell Biology from the University of Maastricht, Netherlands based on comparative studies of Nigerian and Kuwaiti SCD patients.

Abstract :

Sickle Cell Disease in Kuwait

The clinical expression of sickle cell disease (SCD) is highly variable and many epistatic factors have been identified. The most studied of these are the βS-globin gene haplotype and Hb F levels. Most patients seen in the Western world carry the African haplotypes, which are usually associated with severe disease and low Hb F levels. Kuwaiti and many other Gulf Arab patients generally have the Arab/Indian haplotype and present a natural model of SCD with elevated Hb F, associated with a mild disease. However, the clinical picture is not homogenous even in this group. We have studied the clinical presentation of our Kuwaiti patients, documenting their Hb F levels at different ages, spleen function, prevalence of avascular necrosis of the femoral head (AVNFH) and silent brain infarcts. Some of our recent studies have focused on neuroimaging using enhanced techniques including SPECT, flair and diffusion-weighted MRI. WE have also been involved with investigating the response to hydroxyurea in selected patients. A summary of our findings will be presented with a focus on the neuroimaging and clinical Hb F response to hydroxyurea.

Kuwait National Sickle Cell Disease Registry

Introduction: Sickle cell disease (SCD) is a chronic inherited condition with different genetic backgrounds, distributed globally. The variant seen among Kuwaitis tends to have a mild phenotype. The Kuwait National Sickle Cell Disease Registry (KNSCDR) was established in 2010 to document the distribution, Hb genotypes, severity, and complications among our patients.
Method: This is a multicenter study involving all the patients attending the clinics in Mubarak, Amiri, Sabah, Adan, and Jahra hospitals in Kuwait. Patients' demographics, investigations, clinical course, complications and management are documented and data entry is done using a purposely designed Microsoft Access program.
Result: So far, 258 patients with (132 males and 126 female), have been registered; 231 were Kuwaitis. Twenty five (9.7%) were <5 years of age, 91 (35.3%) between 5-15 years, 32 (12.4%)between 16-20

years, 64 (24.8%) between 21-40 years and 46 (17.8%) were >40 years. However there is bimodal age distribution, which would suggest that we lose patients in the transition from childhood to adulthood.
Most of the patients had varying frequencies of hospitalization with vaso-occlusive crises; 170 (66.4%) had at 0-1/year (mild), 51 (19.9%) had >3/year(severe) and 35 (13.7%) had 2-3/year (moderate).
The common documented complications included: 76 (30.4%) had cholecystectomy for gallstones, 58 (23.2%) had avascular necrosis of the femoral head. Overt stroke was documented in only 2 (0.8%) patients.
The distribution of Hb F was: 11 - 30% in 163 (71.1%) patients while it was >30% in 34 (15.1%).
Only 68 (26.4%) patients were on hydroxyurea therapy.
Conclusion: SCD is milder in our region hence severe complications are uncommon. There is a loss of patients in the transition period, the reasons for which need further investigation.

Dr. Léon Tshilolo, RD Congo

Dr. Léon Tshilolo is Medical Director of the Centre Hospitalier Monkole in Kinshasa, DRC. He is also the Main Coordinator of the Centre de Formation et d'Appui Sanitaire (a post graduate training centre in Kinshasa).
Dr. Tshilolo received his medical degree and specialization in pediatrics from the University of Padua and the University of Verona, Italy. He also received credits in Tropical Medicine and Mycology from Prince Leopold Institute, Antwerp, Belgium. Dr. Tshilolo's training includes; Ultrlasound (Gecamines, Lubumbashi), Chemical Haematology (Free University of Brussels), and Genetic Haemoglobinpathies investigations (Robert Debré Hospital, Paris and Hammersmith Hospital, London).
He also acts as an expert advisor to several Sickle Cell Disease organizations, examples include; Scientific Advisor in the National Program of Sickle Cell Disease (DRC), Scientific Advisor of Sickle Cell Disease International Organization (OILD), and a WHO expert on Sickle Cell Disease (WHO Afro Region).

Abstract :

Sickle Cell Disease in the Democratic Republic of Congo

L.Tshilolo1, 2,, B. Ngasia1, 2, G. Kazadi1, LM. Aissi2, M Ekwalanga3, S. Wembonyama3,
1. Centre Hospitalier Monkole (CHM), Kinshasa.
2. Centre de Formation et d'Appui sanitaire (CEFA), Kinshasa.
3. Université de Lubumbashi (UNILU), Lubumbashi.

Introduction
SCD SCD is characterized by a variable phenotype expression depending on genetic and environmental factors. The more severe form seems to be related to people bearing the Bantu's haplotype.
The aim of the study : to contribute to the description of the natural history of SCD in DRC by determining the prevalence of sickle cell anaemia; the specific clinical data on SCD in Congolese patients and the genetic and biological parameters.
Material and methods
Early diagnosis of SCA was performed by IEF and /or Capillary electrophoresis. Clinical and biological parameters were regularly determined in course of the follow up of patients in Kinshasa and provinces.

Results and impact of the actions

Early (neonatal) screening of the disease was developed in 18 maternity units and reached 58,000 children aged < 5yrs : 16% were AS and 1.58 %SS. We also determined the ethnic distribution and the impact of malaria on betaS gene distribution.

We determined specific clinical features in Congolese SCA patients: Hand foot syndrome, sepsis and acute anaemia were the early clinical signs; persistent of a large spleen was observed in 40% of patients aged >5yrs. Torrential nose bleeding, tooth decay and hypertophic tonsillitis were frequent in young patients. Osteomyelitis was severe and often with multiple localization.

Most of the Congolese SCA patients (92%) bear the bantu's haplotype ; α-thal deletion was observed in 39.5% of SS and 44.8% of AS. Among the AS, there was a trimodal distribution percentage of HbS corresponding to 2,3 and 4 α genes.

Congolese SCA patients displayed a permanent inflammatory and undernutrition status and developed high titters of auto-antibodies.

Hematologic parameters in SCA patients, in steady state, displayed a mean value of Hb at 7.2 g/dl and reticulocytes at 8.8%; a leucocytosis (14.9 g/L) associated with eosinophilia (7.8%) and monocytosis (14%)

Since specific vaccination (antihaemophylus and antipneumococcus) was administrated in 1229 children aged 0-5yrs, blood transfusion rates was reduced to 40% and the risk of contamination by viral infections (HIV, VHB and VHC) was also reduced from 10 -15% to 5-8%. Globally mortality was also reduced.

Conclusion

Knowledge of the natural history of SCD is one of the main steps for research progress and sustainability of various projects on SCD control in Africa.

Dr. Jameela Mohammed Al-Salman, Bahrain

Dr Jameela al Salman MD:

Chairperson of the medical department In Salmaniya Medical complex,
Chairperson of the infection controls committee in MOH

Dr Jameela graduated with Honor form the medical school at Arabian Gulf University. Then she joined the SMC in 1996. She is triple American Board certifications: American Board in internal Medicine, American Board in geriatric Medicine, American Board in Infectious Diseases.

She has completed her residency and fellow ship programs in the United states where, she finished the internal medicine residency in Eason , Hahnemann University, , Pennsylvania, USA , then she did a fellowship in geriatric medicine in Temple University in Pennsylvania, then did a fellowship in infectious diseases at Yale University IN New haven, Connecticut, USA.

She has many publications in the peer reviewed journals. She is a member in several Professional Society Membership:-American Medical Association, American College of physician, Northampton County Medical Society, Pennsylvania Medical Society, Infectious Diseases Society of America.

Abstract :

Infections in Sickle disease patients

Sickle cell disease is associated with frequent and often severe infections because of immune function impairment and functional asplenia. Infection is the major cause of death in sickle cell disease patients and especially children with Sickle Cell Anemia under the age of five years. The spleen functions as part of the body's defense against infection by serving as a filter to remove bacteria from the blood.

Overwhelming infections caused by encapsulated bacteria are an important cause of morbidity and death in children with sickle cell anemia. The most important contributing factors to this increased susceptibility to infection are an opsonophagocytic defect due to an abnormality of the alternate pathway of complement activation, a state of functional hyposplenia, and a lack of specific circulating antibodies as a developmental phenomenon. Prophylactic therapy with penicillin has been advocated in recognition of the fact that a majority of the causative organisms are sensitive to penicillin. However, no controlled studies have proved the effectiveness of such therapy.

Immunization with broadly polyvalent vaccines against Streptococcus pneumoniae, Haemophilus influenzae type b, and Neisseria meningitidis may ultimately represent the most effective way to reduce the incidence of catastrophic infections.

Bacterial pneumonia is common in patients younger than four years, with most cases being due to S. pneumoniae, Haemophilus influenzae, Mycoplasma pneumoniae, and Chlamydia pneumoniae. Acute chest syndrome is both a difficult differential diagnosis and a common concomitant of bacterial pneumonia. Osteomyelitis is generally due to a salmonella, most often S. enteritidis; multiple foci are common and treatment is difficult, with some patients developing chronic osteomyelitis with sequestration. Parvovirus B 19 infection causes acute bone marrow failure. Malaria does not result in cerebral malaria but can lead to severe anemia or vasoocclusive crisis, and should therefore be effectively prevented. Antimicrobials are generally selected for efficacy against pneumococci (septicemia, meningitis), Salmonella (septicemia, meningitis, osteomyelitis), and mycoplasmas (pneumonia).

Dr. Ahmed Al Arrayed, Bahrain
Dr. Ahmed Al Arrayed Senior Consultant in General Medicine & Renal Transplant – Salmaniya Medical Complex – Ministry of Health. Associate Professor in Arabian Gulf University – Member of many national and international medical and nephrology societies.President of Arab Nephrology & Renal Transplant society. Vice president of Bahrain Kidney Patients Friendship Society since 1978.Editor of (Al Bashayer) Journal in Arabic to educate the public in the field of transplant and organ donation.

Abstract :

The Renal Manifestations of Sickle Cell Disease

Sickle cell disease (SCD) is an autosomal recessive hemoglobinopathy caused by a point mutation in the β-globin chain of haemoglobin, causing the amino acid glutamic acid to be replaced with the hydrophobic amino acid valine at the sixth position.

The renal manifestations of sickle cell disease (SCD) range from various functional abnormalities to gross anatomic alterations of the kidneys

Functional tubule abnormalities such as nephrogenic diabetes insipidus result from marked reduction in vasa recta blood flow, combined with ischemic tubule injury. This concentrating defect leads to dehydration and sickling crises. The concentrating defect also occurs in individuals with sickle trait.

Other tubule defects involve potassium and hydrogen ion excretion, leading to hyperkalemic metabolic acidosis and a defect in uric acid excretion which results in hyperuricemia.

SCD causes cortical infarcts leading to loss of function, persistent hematuria, and perinephric hematomas.

Research evidence suggests that prolonged glomerular hyperfiltration due to any cause, including SCD during childhood, leads to glomerular damage resulting in glomerular sclerosis, proteinuria and progressive renal failure.

The development of chronic renal failure (CRF) is a predictor of poor outcome in patients with Sickle Cell Disease (SCD).

The median survival of patients with SCD becomes only four years from the time a diagnosis of CRF is made.

It occurs in 4-20% of the patients with SCD, the presence of CRF significantly shortens the survival of these patients.

Data suggests that 5 yr survival after renal transplantation is same as that with pt's on hemodialysis , though survival was slightly inferior to the control population.

Prof. Kwaku Ohene-Frempong, USA

Dr. Kwaku Ohene-Frempong is Emeritus Professor CE of Pediatrics, University of Pennsylvania School of Medicine and Director Emeritus – Comprehensive Sickle Cell Center, Children's Hospital of Philadelphia.

Abstract :

Stroke in sickle cell disease

The two basic pathologies of sickle cell disease (SCD) are chronic hemolytic anemia and vasculopathy. The combination of these two pathological effects of red cell sickling accounts for most of the acute and chronic complications of SCD. Many tissues and organs suffer irreparable damage from chronic hypoxia and from ischemic damage caused by both microvascular and macrovascular occlusion. One organ in which the combined effects of anemia and vasoocclusion have been well demonstrated is the brain with stroke (cerebrovascular accident) as the main clinical outcome.

The incidence of stroke varies among the various genotypes of SCD and may be different among beta-globin gene haplotypes; however, in general, it is highest in sickle cell disease SS (SCD-SS). In the US population, prior to the institution of stroke prevention measures in children, the incidence of stroke was higher among children 1-9 years of age with SCD-SS compared to those 10-20 years old. The highest incidence was in those 2-5 years of age. Stroke incidence may be lower in the Arab-Indian sub-haplotype of SCD. Three types of events are included in the definition of stroke in SCD: infarctive, hemorrhagic, and transient ischemic attack (TIA). While these types of events can occur in any age group, in general, hemorrhagic stroke is less common in children.

It has been possible to study the cerebral pathology associated with stroke using various imaging studies including standard angiography using contrast media, computerized tomography (CT), magnetic resonance imaging and angiography (MRI/MRA), positron emission tomography (PET), single photon emission computed tomography (SPECT), and others. These techniques have provided information on both structural and metabolic changes underlying the pathophysiology of stroke in SCD and have expanded understanding of the risk factors for stroke. Neurocognitive testing has added important information on the functional deficits associated with cerebral pathology in SCD. The use of MRI to study neurologically asymptomatic patients with SCD has led to the discovery of "silent infarcts", a previously unknown pathology whose significance in the development of clinical stroke and neurocognitive dysfunction is being assessed in various studies.

While stroke can be one of the most devastating complications of SCD, it is encouraging to note that in recent years, it has been demonstrated clearly that there are screening and intervention strategies that can reduce significantly the incidence of stroke in SCD. The most impressive strategy has been the use of transcranial Doppler ultrasonography (TCD) screening to detect children with elevated cerebral blood flow velocity; chronic transfusion therapy when given to such children reduces significantly the risk of stroke. In recent years and in health systems where MR imaging technology, TCD, and chronic

transfusion therapy are available and applied to children with SCD, the incidence of stroke has been reduced to very low levels.

Dr. Adlette Inati, Lebanon

• Associate Professor in Pediatrics, Lebanese American University and University Medical Center Rizk Hospital, Beirut
• Head, Division of Pediatric Hematology-Oncology, Rafik Hariri University Hospital, Beirut
• Consultant Hematologist, Chronic Care Center, Beirut, Lebanon

EDUCATION:
Dr Adlette Inati received her medical degree from the American University of Beirut Fellowship at Children's Hospital Medical Center and Sidney Farber Cancer Intstitute, Harvard Medical School, Boston. She is American Board certified.

MEMBERSHIPS:
• American Society of Hematology , European Hematology Association , International Society of Thrombosis , World Federation of Hemophilia , Lebanse Society of Hematology , Lebansese Cancer Society , Lebanese Pediatric Society , Georges N Khoriaty Foundation .AWARDS: • Alpha Omega Alpha Honor Medical Society

RESEARCH INTERESTS
Dr Inati has been recognized for her unwavering commitment to the control of inherited haemoglobin disorders worldwide. Dr Inati runs the largest sickle cell disease clinic in Lebanon and has initiated the first Middle East Thought Leaders and Investigators Sickle Cell Disease Scientific Meeting as well as sickle cell disease prevention and early detection campaigns in Lebanon and the region.

Abstract :

Acute Chest Syndrome in Sickle Cell Disease – What Have We Learnt Over Recent Years

Sickle cell disease (SCD), the commonest monogenic disorder worldwide is an inherited hemoglobinopathy characterized by chronic hemolytic anemia, recurrent vaso -occlusion and end organ failure. Despite monumental recent advances in the understanding and management of this disorder, pulmonary complications remain a major cause of morbidity and mortality in affected children and adults.

Acute chest syndrome (ACS), defined as a new pulmonary infiltrate involving at least one complete lung segment and associated with respiratory symptoms, affects up to 50 % of sickle cell anemia (SCA) patients. It is the first cause for early mortality and the second cause of hospitalizations in this patient population. In adults, ACS is associated with higher morbidity and mortality as compared to children. Optimal treatment consists of cautious hydration, parenteral antibiotics, bronchodilators and incentive spiromety. Interventional modalities as hydroxyurea, transfusions and stem cell transplant have been shown to be highly effective in the prevention of ACS.

Optimizing Transfusions and Chelation in Sickle Cell Disease

Patients with sickle cell disease (SCD) are often given chronic or intermittent blood transfusions in order to prevent or treat complications and the use of this treatment modality has been expanding. Based on Stop trials, chronic transfusions represent standard of care for primary and secondary stroke prevention in children with SCD and transcranial doppler (TCD) screening is now recommended for all children with sickle cell anemia (SS) aged 2-16 years. The goal of transfusion therapy in SCD is either to correct severe anemia and increase the oxygen-carrying capacity of the blood or to replace the rigid sickle-shaped red blood cells with deformable red blood cells in order to maintain appropriate blood flow.
Iron overload represents a serious, inevitable and often underestimated consequence of life saving transfusion therapy and is a major determinant of morbidity and mortality in SCD. Unless effectively treated, iron overload can result in severe organ damage and life-threatening complications. For the same degree of iron overload, patients with SCD appear to have less endocrine and cardiac dysfunction compared to those with thalassemia major. A number of studies have shown that iron chelation therapy is effective in reducing iron burden in patients with SCD.

Dr. Zainab Ali Abdulla Aljufairi, Bahrain

Senior Consultant
Acting chairperson of CME council
Assistant Professor at the Arabian Gulf university

Joined Obstetric & Gynaecology Department in Salmaniya Medical Centre January 1989
July 1996- June 1997: postgraduate training in Ireland. Promoted to consultant position in July 1999.Dealing with different obstetrics and gynecology patients with special interest in sickle cell disease
Training Coordinator in Obstetrics& Gynaecology Department from 1999 till December 2006
Representative of the department in licensure examination (1999-2008)
Arab board Representative of Ministry of Health for Obstetrics and Arab board Representative of Ministry of Health for Obstetrics and Gynaecolgy till 2012, Member of examination committee in Arab Board Council and involved in postgraduate examination
Previous Positions:Training Coordinator in Obstetrics& Gynaecology Department from 1999 till December 2006, Representative of the department in licensure examination (1999-2008)
Arab board Representative of Ministry of Health for Obstetrics and Gynaecolgy till 2012

Member of examination committee in Arab Board Council and involved in postgraduate examination

Abstract :

Maternal mortality among women with sickle cell disease in Kingdom of Bahrain between 1977 and 2012

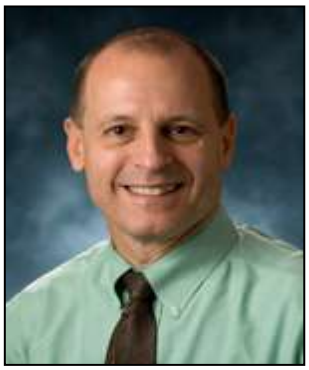
Back ground:
Sickle cell disease (SCD) is a major health problem encountered in the Kingdom of Bahrain. SCD is associated with high maternal morbidity and mortality.

Objectives:
To compare the demographic variables and pregnancy outcome among deceased women with SCD and those with no SCD in the period between 1977 and 2012.
To determine the immediate cause of maternal mortality in women with SCD as compared with no SCD.

Materials and methods
This is a retrospective study which includes all reported maternal deaths in the different maternity hospitals in Bahrain during the period between first of January 1977 up to 31 December 2012. Demographic variables, pregnancy outcome and direct cause of death were compared between women with SCD and the rest of women without SCD.

Results: There were 122 reported maternal deaths in Bahrain in the period between1977 up to 2012. Out of them, thirty seven had SCD accounting for 30% of maternal deaths. The most important direct causes of maternal mortality amongst SCD women include, pulmonary embolism (13), sepsis (9), post partum hemorrhage (6) and acute chest syndrome (5).

Conclusion:
Sickle cell disease is the leading cause of maternal death in Kingdom of Bahrain as it accounts for 30 % of maternal death. All efforts should be focused on managing this high risk group to reduce maternal mortality. Proper thrombo-prophylaxis should be initiated to pregnant women with sickle cell disease. A multidisciplinary medical care should be available as patients with sickle cell disease can deteriorate very fast.



Dr. Russel E. Ware, USA

Dr. Russell Ware joined Baylor College of Medicine and Texas Children's Hospital in March 2011. At Baylor College of Medicine, Dr. Ware serves as Professor of Pediatrics and Vice-Chairman of Global Health for the Department of Pediatrics, as well as Director of the Baylor International Hematology Center of Excellence. Dr. Ware is also the Director of the Texas Children's Center for Global Health and Director of the Texas Children's Hematology Center. He holds an Endowed Chair in Sickle Cell Disease.

Dr. Ware received an undergraduate degree in Chemistry from Furman University in Greenville, S.C., (1979) and both his medical degree (1983) and Ph.D. degree in Immunology (1991) from the Duke University School of Medicine.

He was a faculty member at Duke from 1990-2004, after which he served as the Lemuel Diggs Endowed Chair of Sickle Cell Disease and Chair of the Department of Hematology at St. Jude Children's Research Hospital in Memphis until early 2011.

Clinically, Ware focuses on sickle cell disease, as well as anemia, thrombocytopenia, and autoimmune blood disorders. In his new role as Director of the Center for Global Health, Ware will lead an international sickle cell initiative to begin newborn screening in developing countries, to establish care and treatment plans for these patients, and also test the safety, feasibility, and efficacy of introducing hydroxyurea in this setting.

Abstract :

Hydroxyurea Therapy in Sickle Cell Disease.

Based on potent laboratory and clinical effects, coupled with convenient oral dosing and a mild toxicity profile, hydroxyurea has emerged as the primary disease-modifying treatment modality for infants, children, teens, and adults with sickle cell anemia (SCA). Hydroxyurea increases the hemoglobin (Hb) concentration and induces fetal hemoglobin (HbF), while lowering the white blood cell count, neutrophils, reticulocytes, platelets, and measures of hemolysis. Most young patients with SCA will reach a maximum tolerated dose of hydroxyurea at 20-30 mg/kg/day, and will achieve key laboratory thresholds (Hb ≥9 g/dL and HbF ≥20%) with mild myelosuppression. Clinically, hydroxyurea significantly reduces the frequency and severity of painful vaso-occlusive events, and lowers the number of acute chest syndrome, transfusions, and hospitalizations. Data now also document some protection against chronic organ damage and prolonged survival for patients taking hydroxyurea. Potential long-term toxicities of hydroxyurea therapy including effects on carcinogenesis, teratogenicity, and fertility have not yet been fully defined, yet accumulating data suggest that these risks are low for patients with SCA. No increased risk for stroke, myelodysplasia, or carcinogenicity have been reported for SCA patient cohorts, with drug exposure now reaching 15-20 years for some treated adults and children. In the North America and Europe, available evidence suggests that hydroxyurea represents an inexpensive and effective treatment option that should be offered to most, if not

all, patients with SCA. As countries around the world develop newborn screening programs to identify SCA, hydroxyurea may prove to be a useful treatment for affected children. Particularly in resource-limited settings, hydroxyurea could become a safe and effective treatment for patients with SCA.

Dr. Asaah Nkohkwo BScFrench, FRSPH, UK

A Clinical Scientist & Public Health Consultant
SUMMARY EXPERIENCE: Nationwide Adviser, Comprehensive Care for Sickle Cell Disease, 2009 to-date ,CLAHRC Experience-based Public Health Improvement Methodology, 2010- NIHR-CLAHRC NWL GP SCD Education Project Board, 2010.
NHS Panellist (various), 2004- to-date ,UK NICE Medical Guidelines Development 2010-
WHO Expert Panellist on Haemoglobinopathies, 2006-
Dr Nkohkwo is a chartered and state-registered clinical scientist in Chemical Pathology, who qualified (with an award-winning PhD Medicine) at Hope Hospital, Manchester, in 1993. He then took up day-to-day charge of the Nuclear Medicine laboratory at the Royal Marsden Hospital in London for almost 10years to 2002, gaining in-service a specialist manager diploma from the South Bank Business School, London. For 7yrs to 2009, he took a career break from routine NHS to serve as chief executive of the Sickle Cell Society, (sicklecellsociety.org). In 2009, he was appointed as a DH-funded Nationwide Adviser to champion the marketing of comprehensive care for sickle cell disease across the NHS in England.

ABSTRACT:

PROTOCOLS FOR PAIN MANAGEMENT: the experience of surrogate advocacy in ensuring a patient-centred pain management guideline development.

Under mandate from the UK Department of Health, the National Institute for Health & Clinical Excellence (DH, NICE) published the "Guidelines for Managing Acute Painful Sickle Episodes in Hospital Setting". It took 9 months to deliver the project steered very strictly according to NICE's novel GRADE system of appraising clinical evidence. An important aspect of the process was the encouraging empowerment of the patient's voice as a key stakeholder throughout the development. The final report was fully endorsed by the DH and published as national guidelines in June 2012. It would be interesting to share the experience from the standpoint of surrogate advocacy and its potential impact on user-focused public healthcare development.

ABSTRACT:

AVOIDANCE OF THE CAUSES OF DEATH: impact of a UK mortality audit of sickle-cell disease.

The UK National Confidential Enquiry into Patients Outcomes & Death (NCEPOD) over 2yrs undertook a rigorous mortality audit for sickle-cell disease and published the NCEPOD Report ("a sickle crisis?") in 2008. That landmark report has since instigated several key changes in improving sickle cell care in the UK NHS and abroad, including
a national standard of care for adults living with sickle-cell disease (2008)
an All-Party Parliamentary Group for Sickle-cell & Thalassaemia (2009)
a Peer Review Inspection Programme for NHS Treatment Centres (2010-)
a DH Commissioning Framework for Sickle-cell & Thalassaemia (2011)
a Community-centred Hub concept for integrated care commissioning (2012)

The presentation would appraise the impact of the above outcomes. Of particular contribution on the above would be the salient thread that the developments are user-focused or user-responsive. The contemporary global thought is that patients/ carers are seriously untapped resources in responsive healthcare designs. Hence the momentum is building towards co-creating health or shared-decision making in healthcare (Salzburg

Dr. David Dennison, Oman

Dr. David Dennison completed his undergraduate medical education at the Christian medical college hospital in Vellore, India in 1980. He then did his post-graduation in internal medicine at the same institution in 1986. He subsequently went to the United States of America where he did his fellowship in hematology and oncology between 1988 and 1991 at the University of Nebraska Medical center. At this time he was trained in bone marrow transplantation at the Fred Hutchinson Cancer Research Center at Seattle, USA and the University of Minneapolis Hospital and Clinic in Minnesota, USA. Dr. Dennison returned to Vellore, India in 1991 where he resumed his work as a faculty member of the hematology department at the CMC hospital. He was one of the team of transplanters at this institution which pioneered bone marrow transplantation especially for thalassemia in India. In 1998 he moved to Muscat, Oman where he is senior consultant hematologist and director of BMT at the Sultan Qaboos University Hospital. For the past 14 years Dr. Dennison has developed the BMT program in Muscat to make it one of the leading transplant centers in the Gulf region. His areas of interest are BMT

for sickle cell disease, thalassemia and primary immunodeficiency. He has also set up a haploidentical transplant program at the Sultan Qaboos University. Dr. Dennison has more than publications in peer reviewed journals and is on the editorial board of the bone marrow transplant journal. He is principal investigator on a major project in Oman funded by His Majesty Sultan Qaboos Bin Said's strategic initiative, where his team is investigating the role of mesenchymal stem cells in sickle cell disease related osteonecrosis.

Abstract :

HEMATOPOIETIC STEM CELL TRANSPLANTATION FOR SICKLE CELL DISEASE

Hematopoietic stem cell transplantation (HSCT) for sickle cell disease (SSD) is rapidly gaining popularity worldwide. Many patients with SSD are now free from the devastating multisystem consequences of this disease and can enjoy a pain free existence. Two areas of rapid research in this field include the investigation of alternate donor sources to increase the donor pool for SCD transplants and newer strategies in conditioning. Four large studies from 1986-2002 using myeloblative conditioning regimens have shown event free survivals in the range of 82-85% and transplant-related mortality of less than 10%. While such conditioning regimens work well to cure the young sickle cell anemia patient, modification in conditioning strategies are clearly needed so as to be able to include older patients and to prevent permanent gonadal toxicity. A recent study on ten patients using a reduced intensity conditioning regimen showed promising results with all ten surviving the transplant and only one rejection. In the Sultan Qaboos University Hospital, in Oman, between 1995 and 2012, a total of 16 patients with SCD underwent matched sibling-donor HSCT using a reduced intensity conditioning regimen which consisted of low-dose busulfan, fludarabine and anti-thymocyte globulin. The median age was 16 yrs ranging from 7 years to 40 years. 88% (14/16) of these patients are cured from their disease with a median follow up of 6 months (range 2 yrs to 4 yrs) while two patients (12%) rejected their grafts and are alive with SCD. The forty year old patient tolerated the conditioning regimen remarkably well and is now completely off all medications. 83% (5/6) of the post pubertal female patients however have primary ovarian failure despite the low dose of busulfan given in this regimen. Our data confirms the safety of reduced intensity conditioning which has the potential to be utilized for the older patient with SCD. Better regimens which preserve fertility also need to be investigated to provide the cured patient with an even much better quality of life.

Dr. Abdalla A. Malki, Bahrain

Abdalla A Malki, FRCS Ed, M.D.Ortho, Professor & Orthopedics Consultant

Qualified from Cairo University in Egypt 1968, Obtained higher training and qualifications from UK (FRCS Ed 1980), Obtained M.D. (Medical Doctorate in Orthopaedics) 1986, Egypt, In 1981 joined the Orthopaedic Department and Medical School in Kuwait.
In Kuwait: Established a unit for joint replacement arthroplastries.
Consultant at the Orthopaedic Hospital & Assistant Professor at Kuwait University
Published 30 papers (search pub med for: Malki A "orthopedics") and made more than 40 presentations to International and Regional Journals and Conferences.
Member of: SICOT, Education for Health Network & British Orthopedic association (BOA) and Egyptian Orthopedic Association (EOA),One of the founders of Bahrain Orthopedic Society, GCC & Pan-Arab Orthopaedic Associations.
Currently: Consultant Orthopaedic Surgeon at Salmanyia Hospital since 1994, Professor at Arabian Gulf University in Bahrain & Deputy Chief of Medical Staff SMC.

Abstract :

<u>Management of Femoral Head Osteonecrosis in Sickle Cell Disease</u>

Due to improvement of health care, the life span of Sickle Cell Disease (SCD) patients has increased and many of them live to middle age and beyond. Osteonecrosis of the femoral head is one of the common musculo-skeletal problems in SCD patients. Once osteonecrosis starts in the femoral head, it can progress in many patients from early to late stage in few years.
In the early stages of osteonecrosis, it is advisable to treat by femoral head preserving procedures such as core decompression, bone graft, osteotomy and other preserving procedures. In advanced painful stages, hip replacement arthroplasty is indicated.
In a report from Bahrain on 35 replaced hips for 28 SCD patients, Harris hip score improved from a mean 36 pre-operative to 86 post-operative. However, many early complications were documented such as excessive bleeding, acetabular perforations, sickle cell crisis, and deep venous thrombosis. At a mean follow up of 9.5 (5-15) years, six hips failed due to symptomatic aseptic loosening and one due to late deep infection.
It is concluded that SCD patients for major surgery need appropriate preparation and care for surgery to minimize the incidence of serious complications. It is a must that hematologists,

anesthetists and orthopedic surgeons should work together from the pre-operative preparation until the patient is discharged from the hospital

Dr. S. A. Azeez Pasha, Bahrain

Abstract :

Perioperative Management of Sickle Cell Disease patients

Sickle Cell Disease patients pose immense challenges during the perioperative management of their surgical conditions. As this group of patients are at a higher risk of perioperative complications including death, adequate attention has to be given to the diagnosis of their medical and surgical conditions, assessment of severity of sickle cell disease, preoperative preparation, advanced intraoperative monitoring and anesthetic techniques and postoperative care of cardiovascular system, respiratory system and their intractable acute pain.

Current practice in the management of sickle cell disease patients will be presented and suggestions to improve the quality of care during their perioperative stay will be made. Further prospective research to define the various factors of importance in predicting complications is stressed.

Dissemination of such vital information will help in having better control and more effective management of perioperative complications in these patients.

Dr. Pradeep Kumar Patra, India

Dr. Pradeep Kumar Patra
Current Position : Professor & Head, Department of Biochemistry, J.N.M.Medical College, Raipur (C.G.)
Qualification :M.B.B.S-(1990) from VSS Medical College, {Sambalpur University} Burla, Sambalpur (Orissa).M.D. in Biochemistry (1994) from Banaras Hindu University, Varanasi
Responsibilities:
Incharge Centre for Genetic Disease & Molecular Biology, Raipur..
Principal Investigator of Sickle Cell Screening Project, Chhattisgarh,Deputy Director, Medical Education, Raipur (C.G.) from Jan 2000 to Feb. 2003. Deputy Secretary, Govt. of Chhattisgarh (Biotechnology & Medical Education) and Joint CEO- Chhattisgarh Infotech and Biotech Promotion Society, Raipur (C.G.) from Jan 2003 till date.

Abstract :

SICKLE CELL IN INDIA WITH SPECIAL REFERENCE TO SCREENING PROGRAMME FOR SCHOOL GOING CHILDREN IN CHHATTISGARH

Sickle cell anaemia is a genetic disorder quite prevalent in India specially in few states like Chhattisgarh, Gujarat, Madhya Pradesh, Maharashtra, Odisha, Jharkhand, Kerala etc. Globally this disorder is prevalent in African countries, USA and few countries of Europe and few Asian countries. Unfortunately this disorder is prevalent in poor socio-economic people. Non-availability of well trained health person, less public awareness, no economical investigation facilities, no cheap screening technique, no effective & economical treatment, low cost vaccine, no dedicated sickle cell institute, and no sincere political will are limiting to handle this genetic disorder.

In our centre at Medical College, Raipur, Chhattisgarh, India, sickle cell screening project is going on in which little modification in Hb solubility test, substantial cost reduction of the project has been achieved without compromise in the quality of the test.

Till date 9,38,874 no. of samples have been screened out (children aged 3-15 years) in state of Chhattisgarh, in which 92,762 (9.88%) population [AS-90,058, SS-2,704] are suffering from sickle cell disorder. In our state fetal hemoglobin concentration is relatively high in comparison to the American and African sickle cell disorder patients. The gene frequency data were not in Hardy-Weinberg equilibrium most readily accounted for by a deficiency of the SS phenotype who failed to enter the samples population, through either sickness or early death. All subjects with abnormal haemoglobin received permanent cards bearing personal information and haemoglobin genotype along with personal counselling and educational materials written in Hindi. It is hoped that those with abnormal haemoglobin genotype may factor this information in to decisions regarding marriage and avoid the risks of having children with sickle cell disease.

HERBAL TREATMENT FOR SICKLE CELL PATIENTS

Sickle cell anaemia is a genetic disorder where defective synthesis of globin chain of hemoglobin molecule occur. In result of this average life span of RBC reduced to 10-15 days and create so many clinical presentation mainly anaemia, pain related issues and organ failure. Though few variations in clinical presentation are noticed in different parts of the world but basic clinical features are almost same all over the world.

In different parts of the world, so many herbal medicines/ formulations are available / practised by different local communities and almost none of the formulations are standardized, but they are used for

symptomatic relief. In our place so many herbal medicines are used by local healers. These are as follows-

1. Bilwa (Aegle marmelos) :- Fruits and leaves are used
2. S.Compound:- mixture of some herbs 500mg BD
3. Karwa- Indarjau extract
4. Used by Kamar, Gond, Halba tribes, underground rhizome of Curcuma angustifolia (Tikhur), tuberous root of Abelmoschus crinitus (Datokand), Flowers of Indigofera Cassoides (ghirgholi), boiled seeds of Dolichos biflorus (kulthi), Unriped fruits of Carica Papaya (Papita) and Musa Paradisica (Kela), Whole plants - Andrographis Paniculata (Bhuineem), Dried roots of Scoparia Dulcies, Tubers of Dioscorea species, Dried powder of Chlorophytum Tuberosum (Safed Musli).
5. Plant extract of - Punarnava (Boerhamia diffusa), Rakt chandan (P. Santalinum), Palaash (Butea monosperma), Horsetail (Equisetum arvense), Asparagus root (Asparagus officinalis), Aloevera etc.

In the other part the world, herbs like Spirulina, Stinging nettle, Roots of dandelion, burdock, yellowdock are used for Sickle cell treatment.

Extract of Piper guineensis, Pterocarpa Osun, Eugenia Caryophyllata, Sorghum bicolor, Pterocarpus santolinoides, Aloe vera, Terminalia catappa, thirteen congolese plants (Alchornea cordifolia, Afromomum alno vilaceum, Annona senegalensis, Cymbopogon densiflorus, Bridelia ferruginea, Ceiba pentandra, Morinda lucida, Hymenocardia acida, Coleus kilimandcharis, Dacryodes edulis. Caloncoba welwithsii, and Vigna unguiculata) are used in treatment of SCD.

Crude extracts of Zanthoxylum macrophylla roots, Garcinia Kola-seeds consumed by Nigerians for SCD.

Extract of four plants [NIPRISAN, (1) Piper guineenser seeds, (2) Pterocarpus Osum stems, (3) Eugenia Caryophyllum fruit, (4)Sorghum Bicolor leaves] are used for anti sickling properties.

Some medicinal mushrooms are also used for the treatment of SCD in few region

These plant product/extract relief SCD patients clinically specially pain, anaemia etc. The basic mechanism of action is through RBC membrane stabilization, alter morphology of RBC, providing micronutrients, antioxidants etc.

Dr. Abdulnabi Derbas, Bahrain

Consultant Psychiatrist, Psychiatric Hospital, Bahrain. Ass.Prof.CMMS, AGU

Abstract :

Management of substance abuse among patients with sickle cell disease

Sickle cell disease (SCD) is a complex condition that affects the patient, the family, and the patient's and family's relationship with health care providers and the community. The fear of addiction is one of the greatest obstacles to adequate pain control in sickle cell disease, a fear that might be due to inadequate knowledge of the clinical pharmacology of opioids. This lack of understanding causes confusion between physical dependence and addiction. Addiction is a socio-psychological state that is characterized by abnormal behavior pattern of drug abuse, by the craving of a drug for other than pain relief, by becoming overwhelmingly involved in the use of the drug, and by the tendency to relapse after withdrawal. Drug tolerance is not addiction; it indicates that large doses of a narcotic are needed for an analgesic effect, often without expected adverse effects.Physical dependence is a physiological response to the pharmacologic effects of opioids characterized by the development of withdrawal symptoms when an opioid is

abruptly discontinued or if an opioid antagonistis administered. Drug addiction therefore should not be the primary concern of a physician treating patients with sickle cell disease for pain. The physician should focus on providing patients with adequate relief by understanding drug tolerance, physical dependence, and the clinical pharmacology of the drugs. This knowledge should translate into a practice that includes tapering to prevent withdrawal, thereby eliminating physical dependence after treatment for acute pain. Only occasionally true drug addiction develops in patients with sickle cell disease. Psychological, social, and economic factors are major forces in the patient's addiction rather than the use of prescribed drugs. Addicted patients should be referred to the addiction unit. It should be remembered that drug-dependent patients can have painful episodes; management of such situations requires kindness, firmness, and most important, knowledge of opiate pharmacology. It is very important that teaching the skills necessary for coping with this illness begin at the time of diagnosis and continue throughout the life of the patient, and those providers recognize that including the extended family and the community in the education process will ensure the most positive outcome.

Dr. Najat Mahdi, Bahrain

Abstract :

Sickle cell program in pediatric department

Overview

Sickle Cell Disease is the most common genetic disease in Bahrain affecting part of population and it is an unpredictable, chronic disease requiring a multidisciplinary approach by specialized health care professionals. Optimal management of sickle cell disease and its complications may result in improved outcome for those affected by this potentially devastating disease. Some of the complications of sickle cells disease include:

Episodic pain crises.
Frequent hospitalizations, emergency room/clinic visits.
Increased susceptibility to bacterial infections.
Slowed growth.
Blindness / Visual impairment.
Anemia.
Gallstone disease.
Overt strokes and sub clinical strokes.
Ischemia with tissue damage to heart, lungs, bone marrow, bone, kidneys, spleen etc.
Painful erections in men (Priapism)
Jaundice (hemolytic, obstructive or hepatic).
Psychological stress due to the unpredictable nature of the illness and/or attendance at school or work. This can also affect one's interpersonal relationships in a negative manner.
The potential for lowered self-esteem. Lowered self-esteem can have a direct impact upon the patient's compliance with home management, the general treatment plan, and keeping clinic appointments.
The program was established in 1997.comprehasive program serving all sickle cell disease patients cared for in pediatric department.
The aims: Health education., Vaccination. Prophylaxis. Screening. Treatment.

Health education: many workshops were conducted for patients and their parents, focusing on the nature of the disease and how to deal with it.
Vaccination: Many new vaccines were introduced.
Prevenar (conjugated pneumococcal vaccine) introduced 2002, since then we had no cases of pneumococcal meningitis or septicemia in sickle cell disease patients, with noticeable reduction in the admission rate for pneumonia in infants and toddlers.
Hepatitis A vaccine is advancement in the management of sickle cell disease.

Dr. Erol Baysal, PhD, FRCP, UAE

Dr. Erol Baysal graduated from University College London (UCL) with honours and completed his doctorate training at the Royal Free Hospital School of Medicine, University of London, in 1987
he moved to the USA where he completed a 3-year postdoctoral fellowship at New York University Medical Center (NYUMC).
Dr. Baysal has been working as Consultant Molecular Geneticist at the Dubai Genetic & Thalassemia Center, since 1995. He is currently the Head of the Molecular Genetics and DNA Diagnostic Unit. He was the first person in the UAE to establish the Prenatal Diagnosis program for thalassemias,..
Dr. Baysal's experience in the field of hemoglobinopathies and genetic diseases spans all continents. He has served as consultant to the UN, WHO, NIH, TIF (Thal. Int. Fed.) and serves on the review board of several scientific and medical journals.
. In 1995, he was appointed as senior lecturer in the clinical faculty at Dubai Medical College. In 2002, he was awarded the Sheikh Hamdan Award for Medical Sciences.
He has published 100 peer-reviewed articles in reputable journals. He has authored and co-authored 10 book chapters. In 1997, he co-authored "The Syllabus of Thalassemia Mutations", one of the most cited reference books in the literature.
In 2006, the Royal College of Physicians, London, bestowed upon him an Honorary Fellowship for his distinguished contributions to science and medicine.
Healthcare.

Abstract:

ADVANCES IN THE PRENATAL DIAGNOSIS OF HEMOGLOBINOPATHIES IN THE UAE

The hemoglobinopathies were the first genetic diseases to be characterized at the molecular level. For over 30 years hemoglobinopathies have been used as a prototype for the development of new technique of mutation detection.
In the UAE, the need for prenatal diagnosis of hemoglobin (Hb) disorders has provided impetus for the development of DNA-based diagnoses. Although direct detection of mutant genes is applicable to other diagnostic tasks, prenatal diagnosis (PND) as a principal application will be the focus of this paper. Advances in chorionic villus sampling (CVS) have rendered the first trimester PND a standard practice. The ability to detect mutant globin genes has provided a rapid, safe, accurate and reliable methodology for CVS. In the UAE, PND is now available for pregnancies at risk for virtually all inherited hemoglobin disorders. In the last decade, the use of preimplantation genetic diagnosis (PGD) has become an alternative and increasingly reliable diagnostic approach. Recently, non-invasive PND (NIPND), whereby

fetal cells from the maternal circulation are tested, may soon become a practical reality and be widely available in many diagnostic laboratories.
DNA-based detection of globin gene mutations has been facilitated greatly by the polymerase chain reaction (PCR) revolution, and many reliable diagnostic methods are now available in our DNA diagnostic laboratories. PCR-based methods rely on restriction fragment length polymorphism (RFLP) analysis, allele-specific oligonucleotide (ASO) hybridization, amplification refractory mutational system (ARMS), DNA sequence analysis and several other sophisticated approaches. These methods are available for detecting all types of hemoglobinopathies that affect α or β-globin loci.
This paper will discuss advances made in the UAE in the prenatal diagnosis of hemoglobinopathies among the UAE national families as well as expatriate communities from the UAE and other Gulf States. A number of pertinent issues such as ethical, legal and social concerns regarding its use, identifying families for testing, appropriate conditions for testing will also be discussed. The goal of more widespread utilization of PND will be addressed in the context of prevention programs for the future generations.

Dr. Amani Ali Isa Al Hajeri, Bahrain

Dr. Amani Ali Isa Al Hajeri MD, MSc Medical Genetics (Scotland- University of Glasgow), B.Sc Basic Medical Sciences, Joint Board Certificate in Family Practice (ICGP) & (RCSI), Arab Board of Family & Community Medicine. She is a Bahraini clinical geneticist who graduated from the Arabian Gulf University.
Currently is employed as a Consultant Medical Geneticist at the Genetic Dept. and a Consultant Family Physician at the Ministry of health in Bahrain & Co-Director of The Bahrain Branch of the UK Cochrane Centre/The Cochrane Collaboration.
She is actively involved in producing systematic reviews in collaboration with the Cystic Fibrosis & Genetics Diseases & Heart Cochrane groups. She has a number of publications in several peer-reviewed journals. She is also a member of the editorial board of Bahrain Medical Bulletin. Her principal research interests are in the common genetic disorders in the region such as hemoglobinopathies, the methodology of systematic reviews, and in family medicine.

Abstract :

Inhaled Nitric Oxide for Acute Chest Syndrome: A Systematic Review and Future Genetic Implications

Objective
To perform a systematic review of the literature on the effectiveness of inhaled nitric oxide (NO) in the treatment of acute chest syndrome in patients with sickle cell disease and to explore the genetic implications of this review in the form of gene therapy.
Methods
Electronic databases were searched from their initiation up to date. Additional searches were carried out of abstracts of conference proceedings. These comprehensive searches yielded 9 citations all of them are randomized controlled trials. None of the trials examined the use of inhaled nitric oxide in acute chest syndrome.
Literature review of studies in which investigators have managed to increase the production of NO in different types of tissues has been conducted.

Results
No relevant trials matching the inclusion criteria of this systematic review were identified. This reflects the lack of clinical trials on patients with acute chest syndrome despite the high prevalence of the disease.
Many gene therapy studies were able to restore the bioactivity of NO in different types of tissues. A descriptive summary was presented and a new therapeutic modality in the form of NO gene therapy is proposed.
Conclusion
Although no trials were identified to answer the research question in this review, there is evidence that inhaled nitric oxide may be effective in the treatment of acute chest syndrome, but to support this, well-designed randomised controlled trials are needed.
Nitric oxide synthase gene transfer may hold promise as a future therapeutic strategy for acute chest syndrome.

Public Awareness about Sickle cell Disease in Bahrain

Amani Al Hajeri, MD, CABFM, IBFM, MSc MG, Shaikha Al Arrayed, MBCHB. DHCG. PhD.DHCM
Objectives: To measure the public awareness level about SCD in Bahrain.
Methods: A questionnaire was distributed among 2000 persons from the general public**.** It was conducted from December 2006 to February 2007. The participants got personally interviewed face to face either by a health professional or a trained interviewer.
Results: A majority of 93% has heard of SCD and 89% knew that it can be diagnosed by a blood test. However 51% didn't know about the prevalence of SCD in Bahrain. 84% recognized it as a hereditary disorder and 72% said it can skip generations. Females showed better knowledge than males and married persons seems to know more than singles.
Conclusion: The study reflects a good status of knowledge among the tested sample. Though some of the respondents were confused about the difference between the carrier state of a disease and the affected state. There is a wide acceptance and appreciation of the preventive campaigns being conducted in Bahrain such as the premarital service and the student screening program.

Hussain Jaffar Al Mukharraq, Bahrain.

Gallstones in Sickle Cell Disease patients under 15 years old in Bahrain between 2007-2011.

A retrospective descriptive study.

Fatima Al-Mousawi [1], Maryam Jadeed [1] and Zainab Taraif[1]

Supervised by: Hussain Al-Mukharraq[2]

[1]Royal College of Surgeons in Ireland, Medical University of Bahrain, Kingdom of Bahrain.
[2]Consultant Physician in Pediatric Hematology, Salmaniya Medical Complex, Kingdom of Bahrain.

Word count: 2871

Abstract:

Background: Sickle cell disease is one of the very common chronic hematological diseases in Bahrain specifically and in certain geographic regions internationally. It has major devastating complications namely abdominal crisis and cholelithiasis, owing to the recurrent hemolysis. Surgery is the main interventional approach for almost all the SCD patients suffering from cholelithiasis.

Aims: The study aimed at observing the course, consequences and management of cholelithiasis in pediatric SCD patients in SMC, the major tertiary hospital of Bahrain.

Methods: A retrospective study based on chart reviewing of all SCD patients who were under 15 years old and were diagnosed of having gallstones within a five-year period. (2007-2011).

Results: There were more occurrences in males (53%) than females (47%). The mean age at diagnosis was 10 years for females and around 9 years for males. Cholelithiasis found to be more associative with homozygous SCD (HBSS 54.15%), than heterozygous SCD (HBSA) and other hematological disorders. Of the diagnosed patients, the majority was symptomatic with predominantly abdominal pain. However, symptomatic as well as asymptomatic individuals underwent laparoscopic/ open cholecystectomy as a curative management. In a few exceptional cases conservative management was assumed, but an eventual surgical intervention was considered lately.

Conclusion: large population of SCD patients is at a considerable high risk for developing cholelithiasis. Cholelithiasis appeared early in the pediatric patients with clear and indicative symptoms, most commonly around the age of 10. Almost the vast majority of the observed patients were symptomatic, with abdominal pain being the most commonly encountered symptom. Cholecystectomy has to be considered in symptomatic and asymptomatic patients, as those will frequently develop symptoms in the long run and emergent surgery may be needed.

Headings: Cholelithiasis, Sickle Cell Disease, pediatrics, Hemolysis, Gallstones, Lap Cholecystectomy.

Dr. Abdulrahman Alsultan, KSA

ABDULRAHMAN ALSULTAN, M.D. FAAP
Assistant Professor and Attending Physician, Hematology & Oncology section, Department of Pediatrics, College of medicine and King Khalid University Hospital, Riyadh, Saudi Arabia. November 2008-current
Clinical Fellowship, Hematopoietic Stem Cell Transplantation, Fred Hutchinson Cancer Research Center, Seattle, Washington. July 2008-Oct 2008.
Postdoctoral Research Fellowship, Integrated Department of Immunology, University of Colorado Denver and National Jewish Research & Medical Ctr. Denver, CO. July 2006-June 2008
Clinical Fellowship, Pediatric Hematology/Oncology/Bone Marrow Transplant, The Children's Hospital, Denver, CO. July 2005- June 2008
Residency, Department of Pediatrics, University of Iowa Hospitals and Clinics, Iowa City, IA. June 2001-June 2004
Residency, Department of Pediatrics, King Khalid University Hospital, Riyadh, Saudi Arabia. September 2000-April 2001

Abstract :

Genetic modifiers of the severity of sickle cell disease

Sickle cell disease (SCD) is a common inherited blood disorder in Saudi Arabia. SCD phenotype in Saudi Arabia is widely variable similar to other ethnicities. This indicates the involvement of other genes in determining SCD phenotype. Identification of genes that modify the phenotypic severity of monogenic diseases, SCD included, represents a paradigm shift in our view of these disorders and an important step toward a more meaningful phenotype/genotype correlation. Genetic modifiers of SCD will be discussed focusing on data available from Saudi Arabia.

The molecular basis of high HbF in the Arab Indian haplotype.

Sickle cell anemia is prevalent in the eastern province of Saudi Arabia where the HbS gene is on the Arab-Indian (AI) HBB gene cluster haplotype. Polymorphisms in genes trans to the HBB loci, like BCL11A and HBSL1-MYB and also in regions linked to HBB have been associated with HbF levels in many different populations. The mean fetal hemoglobin (HbF) concentration in AI haplotype sickle cell anemia is higher than that among patients with HbS haplotypes of African origin. Using autozygosity mapping, Sanger and deep DNA sequencing and focused polymorphism analysis we searched for cis- and trans-acting elements that might account for higher HbF. An autozygous region of 101 kb on chr11p containing the entire HBB gene cluster and locus control region was shared by 79 AI haplotype

patients. Other runs of SNP homozygosity were not found. Focused genotyping confirmed homozygosity for AI haplotype-specific elements cis to HBB. SNPs in BCL11A explained less than 10% of the variation of HbF in Saudi patients. KLF1 mutations associated with high HbF were not present. High HbF in most AI haplotype HbS homozygotes is likely to be a result of the interaction of shared cis-acting elements that have a dominant role in determining HbF level, with trans-acting modulators that differ amongst patients. Understanding the mechanisms involved could provide insights into the normal biology of HbF expression.

Dr. Sana Al Khawaga, Bahrain

Abstract :

Predictors of risk of death in adult sickle cell patients admitted to intensive care unit in SMC

Dr. Sana Al Khawaja, Zainab Mahdi, Ridha Al Hammam

Purpose: This study aims at identifying the predictors of mortality in adult sickle cell disease patients admitted to ICU in SMC.

Background: Sickle cell disease (SCD) is one of the commonest reasons for hospital admission in Bahrain and has the highest rate for multiple readmissions for individual patients. Sickle patients even those who appear fit, are susceptible to cardiovascular collapse and sudden death, this finding illustrate the need for thorough study of actually ill patients to identify those at high risk, so they can be treated aggressively before fatal events develop.

Literature: only two studies found reported the SCD's ICU mortality rate and the associated predictors. A study done in UK in 2006 revealed 16% mortality rate among SCD patients in ICU and identified: length of stay (LOS) in ICU, need of blood transfusion, need of inotropes, and cardiovascular comorbidity as mortality predictors. A recent study (2012) in Oman showed 19% mortality rate and found: LOS, need of inotropes, and need of mechanical ventilations as major predictors to SCD mortality in ICU. None of the mentioned studies described the collective prediction power of these predictors but rather studied individually.

Method: retrospective, descriptive, and correlational design. Records of 206 SCD patients who were admitted to the ICU in SMC during the years 2011 and 2012 were reviewed.

Results: The mortality rate among SCD in ICU in SMC is 12.7%. Four significant predictors of SCD mortality in ICU were identified: patient's age, number of hospital admissions, length of stay in ICU, and patient's need of mechanical ventilation. Non-survivors were elder than survivors (M=37.6 vs. M=30.3 years, U=1685, z=-2.2, P=0.025), less frequently admitted to hospital (M=19 vs. M=46 times, U=1274, z=-3.7, P=0.000), have shorter length of stay in ICU (M=3.1 vs. M=5.5 days, U=1145, z=-4, P=0.000), and usually come to ICU on mechanical ventilation (54% vs. 9%, $X^2(1, n=204)=34.5$, p=0.000). When these predictors regressed on mortality, a statistically significant logistic model yielded ($X^2(4, n=197)=62$, P=0.000). This model is highly accurate (85%), sensitive (88%), and specific (85%) in predicting SCD mortality in ICU.

Conclusion: accurate prediction and characterization of SCD patients in term of their survival outcome is a crucial tool to guide timely intervention aims at reducing mortality among them.

More factors need to be studied to uncover their predictive potential of SCD patients' mortality.

Dr. Ashraf Wazeer Zakariya Refaie, Bahrain

Abstract :

Pain management in SCD patients and the role of pain clinic.

Sickle Cell Disease affects around 300,000 people around the world. The hallmark symptom of SCD is pain. The advances in SCD management has resulted that SCD patients are living longer but their life is affected by the negative impact of pain. Pain management in SCD patients forms a big challenge for doctors. Understanding the nature of pain in SCD patients is a major step for the proper approach in SCD pain management. They suffer acute pain attacks during Vaso Occlusive Crisis (VOC) as well as chronic pain due to the effects of repeated VOC. Also SCD patients are not immune from other chronic pain conditions like osteo-arthritis or neuropathic pain. The role of pain clinic to improve quality of life of SCD patients should be emphasized.

Ms. Shafeeqa H. Y. Yaqoob, Bahrain
Ward Nurse Supervisor
Master of Science in Nursing (MSc), [2009-2011], Royal College of Surgeons in Ireland-Bahrain, Bachelor of Science in Nursing (BSc) Degree of Honor from the Collage of Health Sciences, and an Associate Degree in General Nursing, Degree of Honor from the Collage of Health Sciences , Manama, Bahrain
WORKING EXPERIENCE: [Oct 2009-Now] Nurse Supervisor-Ward 61(Haematology/Medical)

Abstract :
Knowledge and Attitudes of Nurses in Bahrain Regarding Pain Management in Adult Patients with SCD
Background: Unmanaged pain is a widespread problem that many SCD patients face on a daily basis. SCD pain can be either acute or chronic. Unmanaged pain increased the morbidity and mortality rate in SCD patients; moreover, it has devastating effects on the quality of life.
Aim and objective: to assess the level of knowledge and attitudes of nursing staff working in sickle cell disease units in Kingdom of Bahrain regarding pain management for adult SCD patients.
Methodology/ Design: A positivist, quantitative study approach was used.
Method: The sample in this study consisted of 30 staff nurses working in adult SCD wards. Staff nurses asked to complete the demographic data section and the modified Nurses' Knowledge and Attitude Survey Regarding Pain management tool.
Results: The results of the study showed that a mean score of 15.8 ± 3 (47.8%) out of 33 was achieved by staff nurses regarding SCD pain management. The data showed that staff nurses demonstrated significant knowledge deficit and negative attitudes regarding pain management. In addition, no significant relationship was found between the demographic data (age,

nationality, level of education, year of working experience, attendance of pain management course), of staff nurses and their knowledge and attitudes of SCD pain management.
Conclusion: The results were seen to be consistent with previous studies that show lack of knowledge and negative attitudes regarding assessment and management of pain.
Implications for practice: The findings of this study suggest the need for the development of specific strategies to effectively educate the staff nurses about pain assessment and management, as well as integrate pain management as a major component of the in-service programs and undergraduate-nursing curriculum to improve patient outcomes.

Prof. Dr. Nafea MH, Bahrain

Prof Dr Nafea Mohamed Hasan ; professor of Hematology Alexandria University and Hematology consultant Salmaniya Hospital(Ibrahim khalil centre).

Abstract :

Protocols for pain management in Sickle cell disease

Pain is the most annoying event for patients with SCD. Pain caused by SCD can be acute, chronic or a mixture of the two. The mechanism of pain is vascular occlusion resulting in ischemia and accumulation of inflammatory products .This results in pain of variable degree in many organs in the body especially in skeletal system.
The protocol of management includes management at home, ED treatment, a day hospital, and hospital admission treatment.
The principles of managing pain for adults with SCD are similar to WHO guidelines for the treatment of cancer-related pain, using a stepwise approach.
Starting with regular analgesia (RA) as paractamol or diclofenac, passing through combination of RA and non-opioid therapy,to stronger pain medication including controlled release opioids, such as controlled release (CR) oxycodone and CR) morphine, should be considered. A reasonable starting dose of the opioid based on knowledge of doses required during the individual's previous pain episodes and the intensity of the current pain episode.
For morphine , an intravenous dose of 0.1 to 0.15 mg/kg (maximum 10 mg) or for hydromorphone, an intravenous dose of 0.02 to 0.05 mg/kg (maximum 1.5 mg), with a reassessment in approximately 15 to 30 minutes after the completion of the infusion, is typically appropriate . This is followed by smaller dose every 3-4 hours until pain is ameliorated.
When the acute pain begins to resolve, the dose is tailed off gradually rather than stopped abruptly, so as to avoid withdrawal symptoms, which can mimic those of sickle-cell crisis.
NSAIDS or Slow-release oral morphine or nonopoid analgesia is used for long-term analgesia.
Of course parallel treatment with liberal fluids, oxygen, warmth, physiotherapy and hydroxyurea which can decrease the frequency and severity of pain.

Mr. Ali Abdulnabi Mohamed, Bahrain
A student at the Royal College of Surgeons in Ireland – Medical University of Bahrain (RCSI Bahrain), Busaiteen, Bahrain. Candidate for Bachelor of Medicine and Surgery, MB BCh BAO. Expected graduation in June 2013.

Abstract :

Does Sickle Cell Disease protect against Diabetes Mellitus? : An exploratory cross-sectional study

Ali Abdulnabi Mohamed[1], David L Whitford[2], Fathia Al-Qurashi[3]
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[2]Department of Family and Community Medicine, Royal College of Surgeons in Ireland - Medical University of Bahrain, PO Box 15503, Adliya, Kingdom of Bahrain
[3]Section of Haematology/Oncology, Salmaniya Medical Complex, PO Box 12, Manama, Kingdom of Bahrain

Background
The co-existence of diabetes mellitus and sickle cell disease has been shown to be rare. However, this has not been established in a population where both conditions are highly prevalent. We carried out this preliminary study in order to explore whether patients with sickle cell disease had a similar prevalence of diabetes mellitus as the general population in Bahrain.
Methods
Cross sectional study in Bahrain. A random sample of 520 patients aged over 18 years with sickle cell disease was taken. Patients' files and laboratory records were examined for the presence of diabetes. Data were analysed using descriptive statistics and chi-squared tests of association.
Results
376 patients were included (mean age 33.5 years). 24/376 (6.4%) patients with sickle cell disease were determined to have diabetes. 32/376 (8.5%) patients had impaired glucose tolerance. The age/sex standardized prevalence of diabetes was 8.25%.
Conclusions
The prevalence of diabetes in patients with sickle cell disease in Bahrain is high at 8.25% but lower than expected in this population (15.8%). There may be a protective effect of sickle cell disease towards diabetes. Alternatively, a lower level of obesity in the sickle cell disease population may be responsible for the low observed prevalence of diabetes. Nevertheless, the impact of these two conditions on vascular disease suggests a need for screening and aggressive treatment of vascular risk factors in this population.

Mr. Ali Darwish, Bahrain
Sickle Cell Care Coordinator, REGISTERED NURSE, BSC NURSING, SPECIAL NURSING SPECIALIST
Mr. Darwish earned his degree in nursing at the College of Health Sciences 2006. He has participated in a study on triage system of accident and emergency S.M.C (2010) Mr. Ali has worked in accident and emergency department (5 years) and has been a clinical nurse instructor for Royal College of Surgeons Irish and College of Health Sciences since 2009-2010-2011. Since 2011, he has been delegated to work as the Sickle Cell Disease Care Coordinator in Salmaniya Medical Complex and he acts as a deputy chairman of nursing media coordinators committee. He is a member in Ministry of Health, sickle cell care improving team since 2012.

Abstract:
Psycho-Social Problems Vs. Narcotic Drug Dependency in Sickle Cell pts. with Frequent Visits to Accidents & Emergency

Background: Group of SCD pts. attend hospital facilities daily either twice a day or more along the year, manifesting significant drug seeking behavior on narcotic medications such as morphine and pethadine. This issue influenced their quality of interpersonal relationships within families & community, besides the risk of drug over dose had increased the morbidity and mortality ratio among this group of SCD pts.
Aim and objective: Based on hypothesis that Narcotic Drug Dependency is related to psycho-social distress, this study aimed to compare the psycho-social problems between the SCD pts. with frequent visits to A&E who exhibited drug seeking behaviors with infrequent SCD visitors.
Settings: A&E Department, Hematology follow-up Clinic, Salmaniya Medical Complex, Kingdom of Bahrain.
Design: Quantitative Study.
Method: The study sample involved 60 adults Bahraini SCD pts., equal in gender, aged 20-35 years, 30 pts. of frequents daily visitors and other 30 pts. of infrequent visitors Psycho-Social Analysis Interview and Questioner been used for data collection.
Results: Data analysis showed(n=35/60,p=58%)±2 reported social and psychological distresses, as it was higher among frequents visitors(n=24/30,p
=80%) which remarkably 75% validate that daily narcotic medication therapy reduce their psycho-social anxiety and stress. However infrequent patients reported less psycho-social problems(n=11/30,p=36%). Moreover In aspect of demographic data's, unemployed, age group of 20-25 years, male exhibited higher level of distress than others.
Conclusion: Its notable that the frequent pts. demonstrated significant reduce in daily life functions with negative cognitive behaviors toward community along with deficit in knowledge and psycho-social adaptation techniques, which may double the risk of Psycho-Depression, Aggression and Narcotic Drug Dependency.
Implications: The recommendations suggest developing preventive narcotic drug dependency strategies and psychological interventions such as Detoxification, Congestive Behavior Therapy, Anger Management along with providing comprehensive community awareness programs and patient-family counseling through forming multi-disciplinary team with contributions of governmental and non-governmental associations.

Dr. Deogratias Munube

Dr. Deogratias Munube, Kampala, Uganda
Profession Medical Doctor/Paediatrician
Educational Background>
2008 to 2012. Makerere University – Masters of Medicine in Paediatrics and Child Health MMed Paediatrics & CH) , Makerere University -(MBChB).1995-1997 Caltec Academy, Kampala - (UACE) – A Level, King's College, Lagos - OLevel (UCE),1982-1986 Corona Primary School, Victoria Island, Lagos,Nigeria –(PLE),rofessional Experience: Aug 12 to date Paediatrician Neurology/Haematology and Oncology Ward

Abstract :
Stroke in Children with Sickle Cell Anaemia in Mulago Hospital

Background: Stroke is a catastrophic complication of sickle cell anaemia (SCA): 10% of patients with SCA will have had a stroke by age of 20.
Objective: To describe the clinical presentation of children with SCA stroke and hematological indices in those with and without stroke. The secondary objective was to describe interventions in both groups.
Methods: 184 SCA children aged 2-18 years attending Mulago Hospital were enrolled in this cross sectional study from February to June 2011. 46 children with stroke were compared with 138 without stroke. Data on the history, physical examination and blood tests was collected. Data was analyzed using Stata Version 10 and was summarized in tables and charts. The Fisher's exact test with odds ratios and 95% confidence intervals (CI) was used.
Results: 35% of the participants were <5 years. The M: F ratio was 1:1.The median age for SCA stroke was 7 years (IQR 5-11) and 8 years (IQR 5-11) for those without stroke. Symptoms of SCA stroke included behavior changes, headache and seizures. Physical examination findings included hemiplegia, aphasia, and limb ataxia. Overall hematological indices, other than HbS level, were not significantly different between the two groups. 4/46 with SCA stroke had an HbS<30%, versus 0/138 without stroke (p=0.004). SCA strokes (44/46) were more likely to have
received a blood transfusion than those without stroke (99/138): OR 8.6 (95%CI 2.0, 37.5). Children with SCA stroke (33/46) were more likely to have had multiple transfusions than those without stroke (62/138): OR 3.1 (95% CI 1.5, 6.6).
Conclusion:
Symptoms in SCA stroke include behavior change, headache and seizures; clinical signs include hemiplegia, aphasia and limb ataxia. Few children with SCA regardless of their stroke status, had HbS<30%. SCA stroke were more likely to have received a blood transfusion than those without stroke probably as a consequence of stroke management.