Conference Programme
& Abstract Book

26-27 September 2015
JW Marriott, Hanoi

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Dear Friends,

We are privileged to cordially invite you to attend and participate in the upcoming 2nd Pan-Asian Conference on Haemoglobinopathies, which will take place between 26-27 September 2015 in HaNoi, Vietnam.

Organised by Thalassaemia International Federation in collaboration with the Vietnamese Thalassaemia Association (ViTA) and the National Institute of Hematology & Blood Transfusion (NIHBt), this conference promises to provide an outstanding opportunity for all involved stakeholders to bridge perspectives from various disciplines and to meet the challenges of improving the health and quality of life of those affected by haemoglobinopathies. This conference constitutes a unique forum for sharing knowledge and experiences and for building up new friendships, collaborations, partnerships and networks.

It is comprised of two parallel programmes, one for the health professionals and one for patients and their families and is expected to attract in total around 600 participants. Both the scientific and the patients/parents programme have been developed by national, regional and international experts, both health professionals and patients in a way as to cover in addition to overviews of existing treatment protocols all the new advances relevant to the care and cure of these disorders.

Further to the contents of the 2nd Pan-Asian Conference on Haemoglobinopathies, we trust that your stay in HaNoi will be most enjoyable as this is a very beautiful and exciting city, whose exceptional ambience and charm, is bound to leave an everlasting memory in the minds of all those who will attend the Conference.

We very much look forward to welcoming and seeing you all at this event, aiming to build a brighter future together for patients with haemoglobinopathies in the region and beyond!

Cordially,

Mr Panos Englezos
Chairman of the 2nd Pan Asian Congress on Haemoglobinopathies
President - Thalassaemia International Federation
Wishing to focus attention and encourage collaborations in order to instigate further improvements from the medical/scientific international community, in thalassemia care and management, the Thalassemia International Federation (TIF), cooperated with many countries and international organizations, with a view to propagate and diffuse knowledge and updated information to the medical/scientific communities and patients/parents globally, through the organisations of international conferences, since its establishment in 1987.

The 1st Pan Asian Conference on Haemoglobinopathies was successfully organized in Bangkok, Thailand on February 8 - 10, 2012. This conference was an interactive forum, where medical specialists, scientists, policy makers, and patients and families could update their knowledge on transfusion-dependent thalassemia, as well as other haemoglobinopathies (non-transfusion dependent). We feel privileged and proud that the 2nd Pan-Asian Conference on Haemoglobinopathies will be organized in Hanoi, Vietnam during September 26 - 27, 2015.

Hanoi, Capital of Vietnam, is center of politics, culture, economics and tourism of the country. In 2000, Hanoi was awarded by UNESCO the “City of Peace” Prize. With antique and elegant appearance, the beauty of thousand years of civilization land, our capital is proud to be one of the most attractive cities in Vietnam to both domestic and international guests. With quick social and economic development, modernized hotel and restaurant system, convenient traffic system, high quality security, Hanoi has been relied to be the host of various international and regional conferences as: APEC Summit in 2006; The 9th ASEM Foreign Ministers Meeting in 2009; The 16th ASEAN Summit in 2010; The 63rd session of the WHO Regional Committee for the Western Pacific in 2012; The 63rd session of the WHO Regional Committee for the Western Pacific in 2012; The 12th ASEAN Health Ministers Meeting in 2014.

The scientific and patients/parents programmes, for the 2nd Pan-Asian Conference on haemoglobinopathies in Hanoi, have been developed jointly, between TIF, TIF member associations and the local co-hosting organizations, the Vietnam Thalassemia Association (ViTA) and the National Institute of Haematology and Blood Transfusion of Vietnam (NIHBT).

The 2nd Pan Asian Conference on Haemoglobinopathies includes a spectrum of topics that cover all aspects of thalassaemia control (Prevention and management), ranging from epidemiology, diagnosis and prophylaxis, national control programmes for haemoglobin disorders in countries of the region; standards of safety in blood transfusion therapy, blood safety and adequacy; iron overload; complications: heart, endocrine and liver; stem cell transplantation in thalassemia; advances towards total cure and the vision of thalassemia care in future.

Undeniably, the 2nd Pan Asian Conference on Haemoglobinopathies-2015 promises to be a proliferation in raising awareness on haemoglobin disorders in the region, but more importantly, it promises to be a milestone in the dissemination of information on the new medical and scientific advances in this field for both health professionals and patients/parents.

300 international and 500 local participants, are expected to participate in the 2nd Pan Asian Conference on Haemoglobinopathies, who will have the opportunity to experience typical Vietnamese hospitality, which we hope will have a lasting profound impression of our country.

Right now, please plan and prepare for your trip to Vietnam to attend the conference in Hanoi. We are welcoming you to be here and desire to bring you hospitality and friendship from Vietnam citizen.

Prof. Nguyen Anh TRI, MD, PhD.
Co-Chairman of the 2nd Pan Asian Congress on Haemoglobinopathies
President - Vietnam Thalassemia Association
Director - National Institute of Hematology and Blood Transfusion, Vietnam
Hanoi - Our Host City

About Hanoi

With an area of 3,329 km² and population of 6.7 million, Hanoi is a political, cultural, economic and tourist centre of Vietnam. In 2000, Hanoi was recognized by UNESCO as the “City of Peace”. With an antique and elegant appearance, the beauty of thousands of years of civilization, our capital is proud to be one of the most attractive cities in Vietnam to both domestic and international guests. In 2013, Hanoi was ranked among the 25 most attractive destinations of Asia.

Climate

The end of the long rainy season in Hanoi is also the beginning of its beautiful autumn - the season of flower blossoming and love. The beauty of Hanoi’s autumn is a popular topic of uncountable songs and poems, which describe how wonderful it is.

September is the transforming time between summer and autumn, so the weather is cooler and more comfortable. Although the temperature is still at a high level, the temperatures during this month are not as extreme as in June and July. Although the temperature range remains between 26°C to 31°C, the sunlight is not too strong and the highest temperature remains below 35°C. The low humidity levels in combination with Hanoi’s cool breeze, soft sunlight and clear blue sky make this city such a romantic place. Besides T-shirts and shorts, a good camera is recommended to capture the charming tree-lined boulevards of milkwood pines or streets piled high with yellow leaves.

Cuisine

Being the cultural centre of the North of Vietnam for centuries, people can find and taste dishes from various lands in Hanoi.

Pho is a popular dish in Vietnam, but Pho in Hanoi has a typical way of cooking. Hanoi Pho is flavoured by cow bone, medium and not tough beef which has a soft taste, pure stock, and slim and soft noodle. After being passed though boiled water, the noodle is placed in bowl, with the beef on top with spring onion and herbs. Through time, many types of pho have developed with different way of cooking, such as stir-fried pho and fried pho.

Green sticky rice from Vong Village is prepared by the villagers of Vong village with typical taste and colour. Green sticky rice is made from young sticky rice covered by lotus leaves and is brought through the city by crowds of salesmen and women.

Another dish of Hanoi, which has recently appeared but is very famous, is La Vong grilled chopped fish. During French Colony, The Doan Family at Hang Son Street, which is now at 14 Cha Ca Street, created a dish of which fame has changed the name of the street. The dish is made from hemibagrus (snake-headed fish) or mudfish - although not as good as hemibagrus - which is cut into thin slices, mixed with galingale liquid, saffron, pepper, fish sauce; then put into bamboo stick to grill on coal right on the dining table. La Vong grilled chopped fish is best tasted while hot and eaten with dry pancake or Vietnamese noodle, peanut, herbs, and, sliced spring onion, and shrimp paste.

Other typical dishes in Hanoi are rolled pho, Vermicelli and chicken soup, Vermicelli and grilled chopped meat, vermicelli and spring roll, etc.
**Places of interest in Hanoi**

- Ho Chi Minh Mausoleum and Ho Chi Minh Museum.
- One Pillar Pagoda located in the same complex (built in 11th century).
- West lake and Tran Quoc Pagoda (built in 544th - an island pagoda in the west lake).
- Quan Thanh Temple (where Taoism is worshipped), located in Thanh Nien road.
- And Old Quarter in the centre of Hanoi, Hoan Kiem Lake, Ngoc Son Temple.
- Catholic Churches, Old Citadel.
- Temple of Literature (the first university of Vietnam which was also built to venerate Confucius, built in 11th century) and many other pagodas and temples.
- Bat Trang Ceramic Village.
- Van Phuc Silk Village.

**Suggested Day trips**

- Perfume Pagoda (half an hour by boat to the foot of the mountain and then climbing up).
- King’s Island in Ba Vi Mountain with Golf course of 18 holes.
- Tam Coc, the Ha Long Bay in land (boating trip through three natural mountain tunnels).
- Hoa Lu - the Old Citadel in the 10th Century.
- Phat Diem - the stone Cathedral Church,
- Bai Dinh pagoda.
- Ha Long Bay (World Natural Heritage, with more than two thousand islands and beautiful caves).
Hanoi - Our Host City

Useful Information

- **Local time**
  The official time in Vietnam throughout the congress is seven hours ahead of Greenwich Mean Time (GMT+7). No daylight savings time applies.

- **Arrival & Transportation**
  Noi Bai International Airport: the biggest airport of the North of Vietnam serves Hanoi and nearby provinces. The airport is about 45 km from the city centre.

  Outside Noi Bai Airport, you can see taxis with quoted price of about $18-20 USD for travelling to the centre of Hanoi. Several taxi corporations provide services for this route. We recommend Mai Linh Taxi (green signboard), Noi Bai Taxi (white signboard with yellow border), or Taxi Group (white signboard with red border). To avoid the danger of overcharging, we recommend that you look at the indicator on the taxi meter or even bargain for the charge before hiring the taxi.

- **Banking Service / Currency**
  - VND (Vietnam Dong) is the currency of Vietnam. Notes are in denominations of VND 1,000; 2,000; 5,000; 10,000; 20,000; 50,000; 100,000; 200,000 and 500,000.
  - The official rate of exchange is approximately VND 23,000 to one USD and VND 29,000 to one EUR.
  - Foreign currencies are easily converted at banks, jewellery shops, hotels, or at authorised exchange counters.
  - Visitors are advised to carry USD cash for easy acceptance nationwide. Tourists may encounter difficulties in exchanging other currencies than the USD. However, at Hanoi - Noi Bai and HCM - Tan Son Nhat airports, upon their arrival or departure, visitors can exchange EUR, GBP, CAD, USD, AUD, SGD, HKG, YPY, CHF, and THB to USD, Vietnamese Dong, or to their own currency.
  - Travellers’ cheques, VISA and MASTERCARD are accepted in most tourist destinations with 5% commission usually being charged when paying.
  - ATMs (Automatic Teller Machines) are located at the Congress Venue and are available at all banks and bank branches, but also at many popular places in Hanoi.
  - ANZ bank’s services are available 24/24.
  - Do not exchange money in the street.
  - Secure all valuables into hotel safes.
  - Upon leaving Saigon or Hanoi, foreigners should arrange to arrive at the airport about 3 hours before taking an international flight and 2 hours before taking a domestic flight.

- **Shopping & Business hours**
  - Shops are open Monday to Sunday: 8.00 - 20.00, although many shops in tourist resorts are open until 21.00 or even 22.00.
  - Favourite shopping places for overseas visitors are Dong Xuan Market, Old Quarter and Hang Gai Street.
  - Banks are open from 7.30 am or 8.00 am until 11.30 am and from 1:00 pm to 4pm. -Saturday afternoon and Sunday they are closed.
  - Governmental agencies work 8 hours per day from 7.30 am to 5:00 pm (excluding a 1 hour lunch break). Closed on Saturday and Sunday.
• **Phone Calls & Mobile Phone Services**
  For international calls, dial the international dialing code (00), the country code, the area code, and the individual number. Domestic phone cards are available for sale at most convenient stores, hotels, and airports.

• **Electricity**
  The standard household and hotel electrical supply in Vietnam is 220 Volts/50 Hertz, but you may find that 110 Volt/50 Hertz outlets are still in use at some places.

  In Vietnam, the standard socket accepts a two round pins’ plug without a ground pin, but non-standard two flat blade or two rectangular blade sockets and plugs are still in use. Some modern hotels and office blocks have sockets for three round pin plugs or the UK three pin square sockets.

  Before leaving for Vietnam, please ensure that the various items that you will be bringing, which will require electricity to operate, have the proper adapters, converters, or transformers.

• **Traffic**
  Public traffic in Hanoi is very comfortable. You can travel cheaply to various destinations. All the bus routes from the centre of Hanoi are charged with cash. The bus cards are used by regular passengers only. The easiest way for tourists to travel around Hanoi is by taxi. Ciclo is a favorite means of transportation for tourists to discover the peaceful old city (36-Street Quarter). Besides, the local authorities are improving disability access to public facilities.

• **Tax & Tipping**
  Value Added Tax (V.A.T.) is levied on most goods and services at a standard rate of 10% and is included in the retail price. Tipping is not customary in Vietnam. In tourist hotels, a 5% service charge is added to the bill, and 5-10% at some large restaurants.
Conference Committees

ORGANISING COMMITTEE:

Panos Englezos - President Thalassaemia International Federation (TIF), President Cyprus Alliance for Rare Disorders and Honorary President Pancyprian Thalassaemia Association

Androulla Eleftheriou - Executive Director Thalassaemia International Federation (TIF), Executive Director Cyprus Alliance for Rare Disorders, Head of the World Health Organisation (WHO) Collaborating Centre for Thalassaemia Control in Cyprus.

Nguyen Anh Tri - President Vietnam Thalassemia Association (ViTA), Director National Institute of Hematology and Blood Transfusion (NIHBT), Vietnam

LOCAL ORGANIZING COMMITTEE:

Prof. Nguyen Anh TRI, MD., PhD. - President Vietnam Thalassemia Association, Director National Institute of Hematology and Blood Transfusion, Vietnam

Prof. Pham Quang VINH, MD., PhD. - Vice President, Vietnam Thalassemia Association, Deputy Director National Institute of Hematology and Blood Transfusion, Vietnam

Dr. Bach Quoc KHANH, MD., PhD. - General Secretary, Vietnam Thalassemia Association, Deputy Director National Institute of Hematology and Blood Transfusion, Vietnam

Pharm. Le LAM, MSc. - Deputy Director National Institute of Hematology and Blood Transfusion, Vietnam

Dr. Pham Tuan DUONG, MD. - Deputy Director National Institute of Hematology and Blood Transfusion, Vietnam

Dr. Nguyen Trieu VAN, MD., PhD. - National Institute of Hematology and Blood Transfusion, Vietnam

Dr. Ngo Huy Minh, MD., MSc. - National Institute of Hematology and Blood Transfusion, Vietnam

Dr. Nguyen Thi Thu HA, MD., PhD. - National Institute of Hematology and Blood Transfusion, Vietnam

Dr. Tran Thanh TUNG, MD. - Cho Ray Hospital, Vietnam
Assoc., Prof. Lam Thi MY, MD., PhD. - Ho Chi Minh City Medicine and Pharmacy University, Vietnam

Dr. Phu Chi DUNG, MD. - Ho Chi Minh Hospital of Blood Transfusion and Hematology, Vietnam

Dr. Nguyen Van TRANH, MD. - Hue General Hospital, Vietnam

Dr. Nguyen Thi Minh THY, MD., MSc - CanTho Central General Hospital, Vietnam
Assoc., Prof. Bui Van VIEN, MD., PhD. - Hanoi Medical University, Vietnam

Dr. Duong Ba TRUC, MD., PhD. - National Pediatric Hospital, Vietnam

INTERNATIONAL SCIENTIFIC ADVISORY COMMITTEE:

Honorary Chairpersons:

Sir David Weatherall - Emeritus Professor of Haematology, University of Oxford, Founder of the Weatherall Institute of Molecular Genetics, United Kingdom
Prawase Wasi - Professor of Medicine, Division of Haematology, Department of Medicine, Faculty of Medicine, Siriraj Hospital, Thailand

Chairperson:

Suthat Fucharoen - Professor of Haematology, Head of Thalassaemia Research Centre, at Institute of Science & Technology for Research of Mahidol University, Salaya Campus, Thailand

Members:

Michael Angastiniotis - TIF Medical Advisor, Ex-Director of the Paediatric Clinic and WHO Thalassaemia Collaborating Centre, Ministry of Health, Nicosia, Cyprus

Nguyen Anh Tri - Director of the National Institute of Haematology and Blood Transfusion and President of the Vietnamese Thalassaemia Association, Vietnam

Ahmed Waqar Khan - Professor of Pathology at Bangladesh Institute of Child Health and Senior Consultant, Department of Pathology, Dhaka Shishu (Children’s) Hospital, Dhaka, Bangladesh

Ching Tien Peng - Professor of Paediatric Haematology at the China Medical University in Taichung, Taiwan, China

Vivian Chan - emeritus Professor of Molecular Medicine, university Department of Medicine, University of Hong Kong, Hong Kong-China

Elisabeth George - Professor and Senior Consultant Haematologist at the Department of Pathology, Faculty of Medicine and Health Sciences, University of Putra Malaysia, Malaysia

Rai Mra - Professor of Haematology and President of the Myanmar Medical Association, Myanmar

Ivy Ng - Professor of Paediatrics, KK Women’s and Children’s Hospital, National Thalassaemia Registry, Singapore

Iskandar Wahidiyat - Professor of Paediatrics, Department of Child and Health, Medical School, University of Indonesia, Indonesia

Roshan Colah - Scientist G & Director -in- Charge, National Institute of Immunohaematology, Mumbai, India

INTERNATIONAL PATIENTS ADVISORY COMMITTEE:

Shobha Tuli - Founder Member and Secretary of Thalassemics India, Vice-President of Thalassaemia International Federation (TIF), India

Mr Loizos Pericleous - Member TIF’s Expert Patient Panel, Thalassaemia International Federation (TIF) Board Secretary, past President of the Cyprus Ant-Anaemia Association, Cyprus

Mr George Constantinou - Chairperson TIF’s Expert Patient Panel, Board Member Thalassaemia International Federation (TIF), United Kingdom

Ramli Mohammed Yunus - Federation of Malaysian Thalassaemia Societies and Board Member Thalassaemia International Federation (TIF), Malaysia

Jeehan Saleem - Vice Chairperson, Maldivian Thalassaemia Society, Male’, Maldives
Faculty Members

Dr Androulla Eleftheriou

Dr Eleftheriou obtained her graduate and postgraduate degrees (BSc Hons, MSc, PhD) from the University of London, in the fields of Biochemistry, Microbiology and Virology. She has been awarded a number of scholarships by the Cyprus government, the World Health Organization and the Fulbright Commission. Her postdoctoral fellowship was completed at the Centre for Disease Control in Atlanta, GA, USA. Dr Eleftheriou has also obtained a Diploma in Business Management from the University of Leicester, UK.

From 1990 until August 2006, Dr Eleftheriou was Head of the Virus Reference Centre of the Cyprus Ministry of Health - a centre she was closely involved in establishing. Since 2005 she is the Director of the Cyprus WHO Collaborating Centre. She was working with Thalassaemia International Federation (TIF) on a voluntary basis since 1993 as a Scientific Coordinator of TIF’s educational programme, and since 2006 she is the Executive Director of the Federation. Dr Eleftheriou regularly acts as a WHO consultant on issues related to her field of expertise.

She is the author of a number of publications published by TIF; as well as a number written in collaboration with WHO and other international bodies on a wide range of scientific topics. Dr Eleftheriou is the Chief Editor of the Magazine of the Thalassaemia International Federation (TIF Magazine), issued quarterly and distributed to hundreds of subscribers in more than 40 countries worldwide.

Prof. Nguyen Anh Tri

Professor Nguyen is the Director of the National Institute of Haematology and Blood Transfusion (NIHBT) and leads the National Blood Safety Programme in Vietnam. He is also the President of the Vietnamese Thalassaemia Association (ViTA). In 1982, he graduated from the Hanoi University Medical School. He specialized in Haematology and Blood Transfusion, receiving his PhD degree in 1993. He published three books on Haematology. He is currently the Chairman of Scientific Committees of National and International conferences on Haematology and Blood Transfusion, organized in Vietnam.

Prof. Anuja Premawardhena

MBBS (Peradeniya), MD - General Medicine (Col), MRCP (UK), FRCP (Lond), D.Phil (Oxon), FCCP (Sri Lanka)

Anuja Premawardhena is a specialist in General Medicine and works at present as Professor of Medicine at the University of Kelaniya, Sri Lanka. He graduated from the University of Peradeniya, Sri Lanka, with MBBS in 1992, being placed first in the all-island merit list. He completed his MD (General Medicine) in 1997 winning the John Stokes Gold Medal as the top performer at the examination. After securing a Commonwealth Scholarship, he studied for his D.Phil in Oxford University, UK, under Prof. Sir David Weatherall, where he developed his interest in thalassaemia and the haemoglobinopathies.

He has set up the first ever dedicated thalassaemia diagnosis centre in Sri Lanka in 2003 and also the first and only Adolescent and Adult Thalassaemia Care Unit in 2006. A Molecular Diagnostic Laboratory was set up in 2009.

Professor Premawardhena has developed an ongoing collaboration with the University of Oxford and the University of Toronto, spanning over 19 years, studying patients with Haemoglobin E / b-thalassaemia, which has become the main area of his research and publications. His other research interests include iron metabolism and hereditary disorders of iron loading.

He has been awarded the Presidential Award for Research in all years from 2003 to 2012 and in 2013 he was elected as Fellow in the National Academy of Sciences of Sri Lanka for his contribution to scientific development.

*in alphabetical order by first name
Dr Aparna Singh Shah

Dr Aparna Singh Shah is currently working in the South East Asia Regional Office of the World Health Organization (WHO SEARO) holding the position of the Regional Adviser for Health Laboratory Services and Regional Focal Point for Antimicrobial Resistance.

She has completed her M.B.B.S.: (Bachelor of Medicine & Bachelor of Surgery) at King George Medical College in Lucknow, India, her Medical Degree at Kasturba Medical College in Manipal, Karnataka, India, and her Postdoctoral Fellowship at the International Vaccine Institute (IVI) in Seoul, South Korea.

Prof. Athanasios Aessopos

First Dept. of Internal Medicine, University of Athens Medical School

Heart involvement in haemoglobinopathies was professor Aessopos’ main subject of investigation. Being a partner with Prof. F. Fessas and Prof. D. Loukopoulos, both prominent haematologists and pioneers in thalassaemia and the haemoglobinopathies, professor Aessopos established, as a cardiologist, in the early 1980s, the first in the world dedicated outpatient clinic for heart problems in thalassaemia, sickle cell disease and other haemoglobinopathies, in the First Department of Internal Medicine, University of Athens Medical School at Laiko Hospital, Athens, Greece. In this clinic, that revolutionized the cardiovascular clinical management and research in haemoglobinopathies, more than 1000 recorded patients have been followed regularly and treated, several doctors have been trained, doctoral theses have been developed and a considerable bulk of research work has been conducted with more than a hundred publications in international peer-reviewed journals, among them some pioneering ones that changed the perspectives and management strategies in the field.

Professor Aessopos is nowadays a recognized world expert in cardiovascular disorders of thalassaemia and haemoglobinopathies and he is frequently invited to give lectures in national and international congresses and asked for patient consultation in Greece, Cyprus, and other countries. Overall, his work contributed substantially to the impressive improvement of the cardiac condition currently observed in thalassaemia and to the understanding of the pathophysiology of heart injury. This has led to the prevention, amelioration and the reversal of compromised heart function. Moreover, his work has stimulated young doctors to be involved in this field, paved the way to further investigation, enhanced patients’ hope and helped them comply better with therapy. Pulmonary hypertension in haemoglobinopathies was established by his work.

Assoc. Prof. Chanane Wanapirak

Associate Professor Chanane Wanapirak graduated as Doctor of Medicine from the Faculty of Medicine, Chiang Mai University, Thailand, in 1985, followed by Certification from the Thai Board of Obstetrics & Gynecology and the Thai Sub-Board of Maternal Fetal Medicine. After that, he has completed his training as a Clinical Fellow in Prenatal Diagnosis and Genetics at the Simpson Memorial Maternity Pavilion, Edinburgh, Scotland, UK, in 1991, and as a Clinical Fellow in Obstetrics & Gynecology Ultrasound and Prenatal Diagnosis Techniques at Munster, Germany, in 1994. In 1998 he was trained as a Fellow in Science & Technology Agency (STA) in the field of molecular biology at the National Institute of Bioscience and Human technology (NIBH), Tsukuba, Japan. He has served as the head of the Department of Obstetrics and Gynecology and at present, he is the head of the Maternal and Fetal Medicine Unit.

He has taken responsibility in the Programme of Prevention and Control of Severe Thalassemia and recently in the Down Syndrome Project at a nationwide level. In 2009 he received the “Chiang Mai University Award” for the best researcher in health science. In 2011, he received the “Guest Master in Academic Award” from the Department of Health, Ministry of Public Health and in 2013 he received the, “Chiang Mai University Award” for the best instructor in health science.

Associate Professor Chanane Wanapirak has published more than 100 publications.
Faculty Members

Prof. Ping Chen, MD, Ph D
Vice Director of Thalassemia Research Institute, Guangxi Medical University, Nanning, Guangxi, P.R. China
Director of Hemoglobin Research Laboratory, the First Affiliated Hospital, Guangxi Medical University, Nanning, Guangxi, P.R. China

Chen Ping obtained her Medical degree from Sun Yat-Sen Medical University in Guangzhou, Guangdong Province, P.R. China. She completed her postdoctoral degree at Guangxi Medical University in Nanning, Guangxi Province, P.R. China.

She was further trained at the Thalassemia Research Center of Mahidol University, Bangkok, Thailand, from 1996 to 1998 and in 1999 at the First Department of Medicine, University of Athens School of Medicine, Athens, Greece. She began her career in 1984 where she was Resident in the Department of Pediatrics of Guangxi Medical University until 1990. From 1990 until 1997 she served as attending doctor in the same department. She became an Associate Professor in 1998 and Professor in 2004.

Professor Chen Ping is a member of four professional societies, namely:
− the Guangxi Branch of Medical Genetics, Chinese Medical Association, of which she is the Vice Chairperson
− the Guangxi Branch of Pediatrics, Chinese Medical Association
− the Medical Genetics Branch of Chinese Medical Association
− the Editorial Board of Chinese Journal of Medical Genetics

The unit where she currently works is the center for screening, diagnosis, prenatal diagnosis and treatment of thalassemia in Guangxi Province.

She has received several research grants, among which one from the Ministry of Health of China in 2010, one from the Department of Science &Technology of the Government of Guangxi Province in 2009, one from the Ministry of Science & Technology in 2008 and one from the National Natural Science Foundation of China in 2007.

She has received the Second Award of Guangxi Scientific and Technological Progress for Prenatal Diagnosis of Thalassemia in 2008 and the National Song Qing Ling Pediatrics Medical Award for “The prevention and treatment on thalassemia in Guangxi province” in 2009.

Dr Cheuk-Kwong Lee

Dr Lee is currently the deputizing Chief Executive and Medical Director of the Hong Kong Red Cross Blood Transfusion Service. He received his undergraduate and medical training at the University of Hong Kong and became specialist in Haematology and Haematological oncology at the Queen Mary Hospital, the University of Hong Kong, Hong Kong. After 8 years of clinical work, he moved to Hong Kong Red Cross Blood Transfusion Service where he became the head of Blood Collection and Donor Recruitment. Apart from securing safe and adequate blood supply to patients in Hong Kong, Dr Lee is also the head of Hong Kong’s Bone Marrow Donor Registry, which provides unrelated haematopoietic stem cells to patients undergoing transplantation.

Dr Lee has developed interest at transfusion microbiology, donor health and medicine. He has made extensive contribution in understanding and implementing preventive measures in minimizing transfusion-transmitted bacterial sepsis. Besides, as healthy and committed donors are a success to the blood service, Dr Lee has been actively involved in studies to understand and minimize donation-related vasovagal reaction and donors’ haemoglobin and iron.

In the thalassaemia community, Dr Lee is the immediate past chairman of the Hong Kong Society for the Study of Thalassaemia. He works closely with the thalassaemia patient group of Hong Kong.
**Prof. Chi Kong Li, MBBS, MD**

Dr. Li graduated from The University of Hong Kong in 1981. After completion of paediatric training, he further sub-specialized in paediatric haematology/oncology and bone marrow transplantation with overseas training in the UK and the US. Currently he is the Chief of the Paediatric Haematology/Oncology/BMT Division and also the Chief of Service of the Department of Paediatrics at Prince of Wales Hospital, and Honorary Clinical Professor of The Chinese University of Hong Kong. His main research interest is in acute leukaemia, thalassaemia and haematopoietic stem cell transplantation. He is now the Vice-Chairman of the Hematology Committee of the Chinese Pediatric Society, Board member of the International-BFM Study Group, and Advisor of the Guangdong Province Thalassemia Society. He has published 246 peer-reviewed papers and he has served as the editor of 1 book and authored 3 chapters.

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**Dimitrios A. Kountouras, MD, PhD**

Internist - Hepatologist, Head of the Hepatology Department at “HYGEIA” Hospital, Athens Greece, Fellow at the Academic Department of Gastroenterology at “Laikon Hospital”, University of Athens, Greece

Dr Dimitrios Kountouras graduated from the Medical School of Athens University, Greece. He completed his internship in Internal Medicine in the 2nd Academic Department of Internal Medicine, University of Athens, at “Hippocrates” Hospital in Athens, Greece. In 2013 he supported the Ph.D. thesis entitled: “Epidemiological and clinical features, natural course and therapeutic interventions in adult patients with homozygous beta-thalassaemia major and chronic HCV infection”.

Currently, he is the Head of the Hepatology Department at “HYGEIA” Hospital, and Director of the 2nd Department of Internal Medicine - Hepatology at “MITERA “ Hospital, in Athens, Greece.

Dr Kountouras participated in national and international clinical research protocols. His fields of interest are: Liver Disease in Patients with Thalassemia, HCV Infection, and Medical Education.

Dr Kountouras is a fellow at the Academic Department of Gastroenterology at “Laikon Hospital”, University of Athens, Greece, and a Member of the Research Team for Thalassemia.

Dr Dimitrios Kountouras has many peer-reviewed publications and has delivered many lectures relevant to his principal interest of liver disease in thalassaemia but also in internal medicine and clinical hepatology.

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**Dr. Duong Ba Truc**

Department of Clinical Hematology, The National Hospital of Pediatrics, Hanoi, Vietnam

Dr. Duong Ba Truc obtained his MD in General Medicine from the Medical University of Sofia, Bulgaria, in 1978. He further completed his PhD from Hanoi University in 1996.

He has held the position of Chief of the Hematology Laboratory of the National Hospital of Pediatrics in Hanoi, Vietnam through 1996-2003 and the position of Chief of the Department of Clinical Hematology at the same hospital through 2003-2013.

Between 2002 and 2006 he also served as Chief of the Study Group on Thalassemia Diagnosis at Community Level in the North of Vietnam and between 2009 and 2014 as Chief of the Study Group on Thalassemia Prevention in the Hoa Binh Province of Vietnam. He has also been a Member of the Study Group on Bone Marrow Transplantation in Thalassemia from 2007 to 2014.

Dr Duong Ba Truc has published 20 publications on Thalassemia and the Hemoglobinopathies in Vietnam, between 1983 and 2003.
**Faculty Members**

**Assoc. Prof. Erica Wood**

Associate Professor Erica Wood is a transfusion medicine specialist, and Head of the Transfusion Research Unit in the Department of Epidemiology and Preventive Medicine at Monash University in Melbourne, Australia. Erica works as a consultant haematologist at Monash Health and holds an honorary appointment at Peter MacCallum Cancer Centre.

Erica is vice-president of the International Society of Blood Transfusion (ISBT), and a member of its working parties on haemovigilance, clinical transfusion practice and transfusion-transmissible diseases. She is also president of the International Haemovigilance Network, and a past-president of the Australian and New Zealand Society of Blood Transfusion. Erica is a member of the World Health Organization’s Expert Advisory Panel on Transfusion Medicine and has served as a WHO regional advisor in blood safety.

Erica is a member of the Executive and Advisory Committees of the Blood Matters Transfusion Practice Improvement Collaborative (a partnership between the Victorian Department of Health and the Blood Service) and past chair of its Serious Transfusion Incident Reporting (STIR) regional haemovigilance program. She is actively involved in teaching, currently serving as Chief Examiner in Haematology for the Royal College of Pathologists of Australasia, and on the RACP/RCPA Haematology Joint Specialist Advisory Committee.

**Dr Frédéric B. Piel, PhD**

Dr Fred Piel graduated in Geographical Sciences with 1st Honours at the Université Libre de Bruxelles (ULB) in 2000. He completed a PhD in the Biological Control and Spatial Ecology Lab (LUBIES, lubies.ulb.ac.be) at ULB in 2006, before joining the Malaria Atlas Project (MAP, www.map.ox.ac.uk) at the Department of Zoology, University of Oxford in 2007 to lead a new global initiative on inherited haemoglobin disorders in humans. In 2012, Dr Piel moved to the Evolutionary Ecology of Infectious Disease (EEID, www.eeid.ox.ac.uk) within the same Department before becoming Departmental Lecturer in Disease Genetics in 2013.

His research focuses on the epidemiology and health burden of haemoglobinopathies, sickle-cell disease in particular but also the thalassaemias, haemoglobin C, the Duffy Blood Group and G6PD Deficiency. Dr Piel’s work aims at using innovative and rigorous quantitative methods (including geostatistics and spatial models) to assemble contemporary evidence for informing public health policies to prevent and manage these disorders, and ultimately improving the quality of life of patients.

In the last 9 years, Dr Piel has developed a large global network of international collaborators and published in prestigious medical journals including The New England Journal of Medicine, The Lancet, The Lancet Global Health and PLOS Medicine. He is an expert of haemoglobinopathies for the Global Burden of Disease (GBD) Study and a member of the Editorial Board for the Transactions of the Royal Society of Tropical Medicine & Hygiene.

**Dr Huaying Liu**

Huaying Liu is a doctor at Nanfang Hospital, Southern Medical University, Guangzhou, Guangdong province, PR China, with expertise in children’s blood disease diagnosis and treatment. Currently, she is in charge of the Hematopoietic Stem Cell Transplantation Unit for thalassaemia patients. She is conducting research on iron overload in thalassaemia, funded by Guangdong Science Foundation.

Until June 2015, 570 patients with thalassaemia underwent HSCT in this Unit. Overall survival for patients treated with HSCT of matched donor is around 90%, as published also in Blood journal on 11 Sep 2012.
Dr Hishamshah Mohd Ibrahim

Hishamshah Mohd Ibrahim, M.D., is a Senior Consultant Paediatrician and Senior Consultant Paediatric Haematologist & Oncologist with clinical responsibilities and research interest in childhood malignancies, stem cell transplantation, haematological disorders and infections in the immunocompromised. He received his M.D. and Master of Medicine (Paediatrics) from Universiti Kebangsaan Malaysia and begun his paediatric professional career in Hospital Kuala Lumpur (HKL), Prince of Wales Children’s Hospital, Sydney, Australia, Fred Hutchinson’s Cancer Research Center, Seattle, Oregon and at Children’s Hospital of Los Angeles, California, in the United States. He has been with the Paediatric Department of HKL since 1994 as a clinician but has also assumed numerous administrative and senior advisory roles for the Ministry of Health, Malaysia. He is part of the core team collaborating with other stakeholders in formulating national strategies and policies for Haematology, Oncology and Transplantation services for the country. In the field of Thalassaemia, Dr Hishamshah is a key member in the National Steering Committee for the Thalassaemia prevention and control programme, which started in 2005 and he is the chairman for the Malaysian Clinical Guidelines for the Management of Transfusion Dependent Thalassaemia. He is also an active member of numerous medical societies and non-governmental organisations and writes as well as lectures regularly in the fields of paediatric haematology, oncology and stem cell transplantation, both locally and abroad.

Fathmath Jeehan Saleem

Jeehan Saleem was born in Male, Maldives. On scholarship from the New Zealand government she completed a Master’s in Public Health in 2009 at the University of Otago, New Zealand. She also has a Graduate Diploma in Applied Science, specialising in Molecular Genetics from University of Waikato, New Zealand in 2000 and a BSc in Medical Laboratory Science from the RMIT University, Melbourne Australia in 1998.

In 1992 she started her professional career at Society for Health Education (SHE), a local NGO, becoming Head of Laboratory Services in 2003 and thus servicing 3 full-fledged laboratories: a Thalassaemia and haemoglobinopathies laboratory, Biochemistry, and Molecular Genetics laboratory - in which she played a significant and crucial role in its set up. At the same time, Ms Saleem also coordinated the national Thalassaemia prevention programme, including raising public awareness, screening general public and genetic counselling. In 2010 Ms Saleem joined the United Nations Population Fund (UNFPA) in Maldives, first working as a technical consultant on the Youth HIV/AIDS programme and then (2011 - 2014) as the National Program officer for Reproductive Health working on the Youth and Adolescent Sexual Reproductive Health (ASRH) programme. She has undertaken several scientific research with Thalassaemia, Molecular characterization and mapping, waterborne diseases and cervical cancer are her areas of expertise.

Ms Saleem is presently the Chairperson of the Maldivian Thalassaemia Society (MTS) after starting volunteering as a general member in 2006, fulfilling a desire to serve directly to patient and parent community. She is actively involved in achieving the goals of the organisation particularly in finding ways and means for thalassaemias to be healthy and productive members of society. She has initiated numerous patient oriented programs that have benefitted the patient community immensely. Ms Saleem continues to strive to improve the quality of care for patients collaborating with national and international institutions. She has also served as a member of the National Thalassaemia advisory committee of the Ministry Health.

She has received 3 scholarships and several notable awards such as the Junior Chamber International’s (JCI) Ten Outstanding Young Person (TOYP) world award for humanitarian and voluntary leadership (2012), the Maldives President’s Award for her international recognition (2013), and National Youth Award (2007) for the dedication and humanitarian work in the area of Thalassaemia prevention and support for Thalassaemia patients and their families.
Faculty Members

Prof. John B. Porter, MD, FRCP, FRCPath
Department of Haematology, University College London, London, UK

John Porter is Professor of Haematology and Consultant Haematologist at the University College London Hospitals in London, UK, and head of the joint Red Cell Unit for UCLH and Whittington Hospitals. He graduated from the University of Cambridge in 1974. He was awarded an FRCP by the Royal London College of Physicians in 1995 and an FRCPath in Haematology by the Royal College of Pathologists in 1996. He was awarded the Lionel Whitby Medal for MDs with exceptional merit.

The treatments of thalassaemia and sickle cell disorders have been Professor Porter’s main clinical and research fields, with particular reference to iron overload in these conditions. He has received funding from many sources, including the Medical Research Council (MRC), the Welcome Foundation and National Institutes of Health (NIH), for this work. His research has focused on the mechanisms of iron chelation, the speciation and uptake of non-transferrin-bound iron (NTBI) species and their relevance to iron-mediated toxicity, the molecular basis of iron homeostasis in health and disease, and the actions and toxicities of mixed-ligand chelation therapy. Ongoing clinical studies aim to clarify the importance of NTBIs in the monitoring and design of deferoxamine regimens, the reversal of cardiac dysfunction with chelation regimens, the role of red cell microvesiculation in the prothrombotic state, and the treatment of bone disease in thalassaemia. Professor Porter is also the principal UK investigator in the ongoing multicentre randomized controlled trials of the orally active iron chelator Deferasirox.

Professor Porter has published more than 100 peer-reviewed articles. He has also made numerous contributions to books, as well as clinical guidelines and other medical articles. Professor Porter has served as scientific adviser to the British Society of Haematology, the UK Thalassaemia Society, the Thalassaemia International Federation (TIF), and to grant review and advisory panels at the NIH in Bethesda, MD, USA. He is a consultant and co-investigator for the Thalassaemia Clinical Research Network, also at the NIH.

Khanh Quoc Bach, MD, MSc
National Institute of Hematology and Blood Transfusion (NIHBT), Hanoi, Vietnam

Dr Khanh Quoc Bach serves as a physician at the National Institute of Hematology and Blood Transfusion in Hanoi, Vietnam, since 1995. He trained through a Hospital Internship (fellowship) in Hematology at the Edouard Herriot Hospital, Lyon, France. He was a Guest Researcher in Hematology and Blood Transfusion, at the National Institute of Health, Maryland, United States. He completed a study visit in Bone Marrow Transplantation at St Jude Children’s Research Hospital, Memphis, Tennessee, United States and another study visit in Leukemia and Bone Marrow Transplantation at MD Anderson Cancer Center, Houston, Texas, United States.

In his major administrative responsibilities he serves as a physician of the Department of Clinical Hematology since 1994, as the Deputy Director of NIHBT since June 2005, as the General Secretary of the Vietnamese Society of Hematology and Blood transfusion since 2012, and currently he is the Vice-President of Vietnamese Thalassemia Association and the Vice-President of the Vietnamese Hemophilia Association.

He is a Member of the American Society of Hematology (ASH) and a Member of the International Society of Thrombosis and Hemostasis (ISTH) since 2008.
**Prof. Dr. Mahmood Ahmed Chowdhury (Arzu)**

Professor Dr. Mahmood Ahmed Chowdhury (Arzu) is now working as Professor of Paediatrics at the Institute of Child Health, Maa (Mother) & Shishu (Children) Hospital in Chittagong, Bangladesh. He is also the Honorary Chief Physician of Thalassaemia Welfare Center - Bangladesh, located in Chittagong.

He graduated from Chittagong Medical College in 1985 and obtained a Diploma of Child Health from the same institution. He received FCPS in Paediatrics from Bangladesh College of Physicians and Surgeons, Dhaka, in 1996. He further received FRCP from the Royal College of Edinburgh, UK, in 2011. He has extensive contribution in the field of thalassaemia and based on his contributions, Thalassaemia International Federation nominated him for the Renzo Galanello Training Fellowship Program in Whittington Hospital, London, under Dr. Farrukh Shah. During his training program he also worked with other prominent consultants on thalassaemia at different institutions around London.

Professor Chowdhury started the first thalassaemia carrier detection program of Bangladesh in Chittagong. His primary goal was to bring awareness about the severity of thalassaemia and to raise this issue to the policy makers of Bangladesh so that the government can initiate a national thalassaemia program. His wife, Dr. Razia Sultana, who is a Consultant Pathologist, is also actively involved with him in order to improve the patients’ well-being. He has a significant number of publications on thalassaemia, both nationally and internationally. He also attended thalassaemia conferences in Abu Dhabi and New Delhi, and gave poster presentations on the socio-economic situation of thalassaemia in Bangladesh.

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**Mahmoud Hadipour Dehshal, Pharm.D**

Born in the green zone of Iran, Dr. Mahmoud Hadipour Dehshal faced the challenges of living with thalassaemia. After graduating from high school, he moved to Iran’s capital city, Tehran, where he obtained a Pharm.D degree from Tehran University of Medical Sciences.

He worked for both private and governmental sectors and run his own business in the field of pharmacy. In addition, as an active member of Iranian Thalassaemia Society, he has experienced fighting for persons with thalassaemia in an unstable atmosphere.

He wrote many articles both as a professional journalist in Iranian newspapers and as a scientist in scientific journals. His main interest is to investigate on the prevention of thalassaemia, ethical marketing in the field of health, drugs’ quality, and the role of NGOs in the health system.
Faculty Members

Maria Liza T. Naranjo, M.D.
Hematologist, Dr. Fe del Mundo Medical Center

Her involvement with thalassemia began in 2003, during her first year of hematology fellowship training at the Hematology-Oncology Unit of Dr. Fe del Mundo Medical Center in Greater Manila, Philippines. Under the dynamic and inspiring mentorship of the late Dr. Alendry P. Caviles Jr. and Dr. Ernesto d’J Yuson, Dr. Naranjo was assigned the task of strengthening the thalassemia patient support group “Balikatang Thalassemia (Ba-Tha)”\(^1\). From conducting regular medical and lay fora on thalassemia and other patient activities, Dr. Naranjo re-connected Ba-Tha with the Thalassemia International Federation in 2005 and spearheaded the local celebration of World Thalassemia Day (May 8th every year) in 2006 up to the present day.

Together with Dr. Yuson, she is presently involved in the HPLC screening for thalassemia and serves as a collaborator for the ongoing genetic research on thalassemia with the Institute of Human Genetics, National Institute of Health. She is interested in strengthening her knowledge in carrier detection in the hope of contributing to the establishment of a National Thalassemia Screening Program and Registry. She has also dedicated herself in networking with the Philippine Blood Center to address the challenges of blood availability for thalassemia, the Quezon City Health Department to improve thalassemia awareness, detection and care at the community level and has had opportunities to advocate for a comprehensive thalassemia support package that would cover family screening, blood transfusion assistance and iron overload management under the Government’s Universal Health Care Program.

She is a member of the Philippine Alliance of Patient Organizations (PAPO) and is known among the thalassemia patient community as a devoted thalassemia supporter with a strong belief in the capacity of empowered patients to be part of the equation in solving the health needs of thalassemia in the Philippines.

\(^1\)Balikatang Thalassemia (Ba-Tha) is a thalassemia patient support group in the Philippines, founded in 1993 by Dr. Caviles and Dr. Yuson along with thalassemia families, with the aim to address the special medical needs of thalassemia patients while providing a supportive and caring community. The group’s name translates to “having a shoulder to lean and depend on”.

Prof. Mehran Karimi

Mehran Karimi has been a Professor of Pediatric Hematology-Oncology of Shiraz University of Medical Sciences since 1997. He is also the chief of the Hematology Research Center and works in the field of thalassemia, hemophilia, homeostasis and thrombosis in Shiraz University of Medical Science, Iran, for a period of 17 years.

After graduating in medicine from Shiraz University of Medical Sciences, he completed sub-specialty in hematology with more focus on thalassemia and hemophilia, from Shiraz and Ahvaz Universities of Medical Sciences as well as through separate 6-month fellowships in Leeds, UK, in 1996, and in Milan, Italy, in 2001 & 2003.

Professor Karimi’s research interests are in the fields of thalassemia, hemophilia, and thrombophilia. Professor Karimi is director of programs in these disease areas at the referral Hospital in Shiraz. He is a member of the International Society on Thrombosis and Haemostasis (ISTH), the American Society of Hematology (ASH) and the Iranian Pediatric Hematology-Oncology Society, as well as a reviewer of some peer-reviewed journals. He also received two awards as a best researcher from Razi Festival in 2003 and 2008 in Iran and the 15th Abu Reyhan Biruni Research Festival Award in 2014. He was also selected as the best researcher in Shiraz University of Medical Sciences from 2004 to 2014. Professor Mehran Karimi has authored more than 263 articles and more than 274 abstracts in peer-reviewed journals and has been an invited speaker at many national and international meetings.
Dr Michael Angastiniotis

Dr Angastiniotis was born in 1941 in Famagusta, Cyprus. He graduated in Medicine from the University of Aberdeen, Scotland, in 1966 and received his graduate training in Paediatrics in Scottish and Oxford hospitals. After returning to Cyprus, he attended various courses such as Thalassaemia (Biochemistry-Prenatal Diagnosis) at the University College London, London, UK, Genetics and Haematology/Oncology at the Duncan Guthrie Institute, Glasgow, Scotland, UK. Until retirement he was Director of the Paediatric Department of Archbishop Makarios III Hospital as well as the Cyprus Thalassaemia Centre at the same hospital. Currently he holds the position of the Medical Advisor for Thalassaemia International Federation (TIF). He is a Member of the Committee for the Control of Hereditary Anaemias since 1983 and a Member of the WHO Expert Advisory Panel on Human Genetics since 1990. In addition, he has been a Special Advisor of the WHO for the control of haemoglobinopathies in the Eastern Mediterranean region, including the UNRWA fields.

Michele Rhee

Michele Rhee has served as Bluebird Bio’s head of patient advocacy since January 2015. Formerly, Michele was Director of Strategic and Program Initiatives of the National Brain Tumor Society where she played an integral role in the overall development and strategy of the organization. Prior to joining the National Brain Tumor Society, she worked at Health Advances, a consulting firm specializing in life sciences’ new product marketing and commercialization. As a rare disease and cancer survivor, Michele also founded and serves on the Board of Costs of Care, an advocacy organization dedicated to transforming American healthcare delivery by empowering patients and their caregivers to deflate medical bills. She received her MBA from the Yale School of Management and her MPH from the Yale School of Public Health. She currently serves on the Consumer Health Council of Massachusetts Health Quality Partners.

Assoc. Prof. of Radiology, Pairash Saiviroonporn, PhD
Department of Radiology, Faculty of Medicine Siriraj Hospital, Mahidol University, Bangkok, Thailand

Pairash Saiviroonporn completed his MS and PhD educational training in biomedical engineering at Boston University, MA, USA. His dissertation title was “Non-Fourier Encoding for intervention MRI”. Afterwards he become an academic staff at the Radiology Department, Siriraj Hospital, Faculty of Medicine, Mahidol university, Bangkok, Thailand. Currently he is an Associate Professor of Radiology at the same institute, teaching radiology residents and fellows. His interest is on cardiac and liver T2* measurements for thalassemia patients, which involves not only MR acquisition, but also post-processing analysis. He has published papers regarding the reduction of inter-observer variations of the analysis.
Faculty Members

Panos Englezos

Mr Panos Englezos is a graduate of London University in Economics and Accounting. He is the Chairman and Chief Executive of a group of companies which he established in 1961. The birth of his son, George, with β-thalassaemia major led to his deep lifelong involvement in the development and promotion of the Pancyprian support groups of patients and parents, since 1969. He was elected the first president of the Cyprus Thalassaemia Association with numerous initiatives and activities aimed to create support services and government recognition, and the importance of developing national strategies for the control of thalassaemia in Cyprus, as a priority. He held this post until June 1995.

In 1986, as an extension of the work of a very few countries’ patients organizations experiences, including the Cyprus Thalassaemia Association, the Thalassaemia International Federation (TIF) was established with the vision and mission to strengthen the medical care and the prevention programmes worldwide. He was elected as TIF’s first chairman, and is regarded by the international thalassaemia community as its ‘founding Father’. He has travelled extensively promoting TIF’s objectives, mainly establishing Thalassaemia associations, promoting improved medical care, international research and prevention programmes. He has received several awards for his pioneer role in the fight against thalassaemia in Cyprus and the world, such as the District Governor’s Appreciation by Lions International Gold Medal for continuous and long-term services in fighting for patients with thalassaemia in 1982, the Greek Haematological Society and Greek Thalassaemia Association’s Silver medal “Kilikas” in 1993, the King Abdulazis University Award for contribution in establishing a thalassaemia association in Saudi Arabia in 1993, the Gold Medal of the Cyprus Thalassaemia Association in 1996 and the Rotary Paul Harris Fellow for outstanding contribution to society in 1997, among others.

Panos Englezos has been a most successful business professional in Cyprus and has held important public and political offices, including the chair of the Cyprus Telecommunications Authority, the Rotary Club Nicosia, the Cyprus Publishers Association, and the Cyprus Tourism Organisation just to name a few. Currently, Panos Englezos is serving also as the President of the Board of the newly established Cyprus Alliance for Rare Disorders (C.A.R.D.), the Pan-European Blood Safety Alliance (PBSA).

Dr. Ngo Manh Quan

Dr Ngo Manh Quan graduated from Hanoi Medical University in 1999 and has worked for the National Institute of Hematology and Blood Transfusion (NIHBT) in Hanoi, Vietnam. He obtained an MSc in 2008 and a PhD in 2015 on Hematology and Blood Transfusion at the same university. Currently, he is the Head of the Donor Recruitment Department, working at the Thalassemia Center of NIHBT.

Dr Nguyen Thi Thu Ha

Dr Nguyen Thi Thu Ha graduated in 1995 from Hanoi Medical College. She obtained her MSc degree in hematology and blood transfusion in 2009 from the same College. She has been studying for her PhD in Hematology and Blood Transfusion since 2013, with specialization in thalassaemia.

Between 2005 and 2011, she has been working in several clinical departments of the National Institute of Hematology and Blood Transfusion (NIHBT) in Hanoi, Vietnam.

From 2011 until the present day, she holds the positions of General secretary of the Vietnamese Thalassemia Association and Vice Director of the Thalassemia Center in the National Institute of Hematology and Blood Transfusion.
Prof. Paul Kwo

Paul Kwo is currently Professor of Medicine and Medical Director of Transplantation at the Indiana University School of Medicine, Indianapolis, USA. He joined the faculty in 1995 after receiving gastroenterology and hepatology training at the Mayo Clinic in Rochester, Minnesota, USA. Since that time, he has distinguished himself in the field of chronic hepatitis C and has a large practice devoted to current and novel therapies for the treatment of hepatitis C. He has won multiple awards at the university, local, and national levels.

Prof. Philippe Leboulch

Philippe Leboulch is a professor of medicine and cell biology at the University of Paris-Sud and director of France’s institute of Emerging Diseases and Innovative Therapies (iMETI) of the CEA. He is also a visiting professor both at Harvard Medical School in Boston in the Genetics Division of the Brigham & Women’s Hospital, and at Mahidol University in Bangkok in the Hematology Division of Ramathibodi Hospital.

He received his MD degree from the University of Paris and became a resident of the University Hospitals of Paris. He then pursued a postdoctoral fellowship at the Massachusetts Institute of Technology before joining Harvard Medical School where he served on the faculty and headed a research laboratory since 1993.

Professor Leboulch has made pioneering contributions to the field of gene therapy and to the treatment of the beta-hemoglobinopathies, from bench to bedside. With regard to the gene therapy of beta-thalassemia and sickle cell disease, he has published on the stable retroviral transfer of beta-globin/LCR derivatives (EMBO J 1994), the design of chimeric genes that inhibit sickle globin polymerization (PNAS 1995) and the first correction of mouse models of sickle cell disease (Science 2001). He then led an international effort to bring these advances to the clinic and became the scientific director of the first worldwide gene therapy trial for the gene therapy of the beta-hemoglobinopathies, which resulted in the first conversion to transfusion-independence of a thalassemia patient 8 years ago (Nature 2010). He is a founder of bluebird bio, the leading gene therapy company, of which he remains co-Chairman of the Scientific Advisory Board. He directed bluebird bio’s scientific effort that resulted in multi-center international clinical trials for the gene therapy of beta-thalassemia and sickle cell disease.

Professor Lebouch has received numerous awards, including a Grand Prize from the French National Academy of Sciences and France’s Legion of Honor. He has served on the Editorial Board of Blood and as Member and/or Chairman of multiple NIH panels including as sub-committee Chairman of NHLBI “Strategic review and recommendation panel for the 21st century”.

Pustika Amalia Wahidiyat, MD, PhD

Pediatric Hematology-Oncology, Department of Child Health, Faculty of Medicine, University of Indonesia, Cipto Mangunkusumo National Hospital

Pustika Amalia Wahidiyat studied Medical Science and Medicine at the University of Indonesia in Jakarta, Indonesia. Afterwards, she became a pediatrician at the same institution. She took her fellowship in Pediatric Hematology-Oncology at Amsterdam Medical Centre (AMC), Emma Children’s Hospital, in The Netherlands. In 2009, she received her doctoral degrees in Medicine from the University of Indonesia. Her dissertation title is “Genetic factors modifying the phenotype of β-thalassemia/HbE: Interaction between β-, α-thalassemia mutations, XmnI-γ polymorphisms, and SNPs on the β-globin gene cluster”.

Currently, she is a lecturer and senior staff of the Hematology-Oncology Division and Undergraduate Program Coordinator, Department of Child Health, Faculty of Medicine, University of Indonesia, Cipto Mangunkusumo Hospital, Jakarta, Indonesia, where she is teaching medical students and pediatric residents. She is also responsible for thalassemia patients since she has been appointed as Acting Chairman of the Thalassemia Centre in Jakarta. She has published papers regarding thalassemia patients.
Faculty Members

Dr. Roshan Colah

For the last 36 years, Dr. Roshan Colah has been working in various positions at the National Institute of Immunohaematology under the Indian Council of Medical Research in Mumbai and she just retired in June 2015 as Scientist G and Director-in-Charge of the Institute. She had been the Head of the Department of Haematogenetics at the Institute since 1988. Her group worked on Red Cell Disorders, and in particular on Hemoglobinopathies, Red Cell Enzymopathies and Membranopathies. Her main area of research has been on the epidemiology of the thalassemias and sickle cell disease, understanding the molecular basis of the thalassemias and developing prevention programmes in India, as well as on establishing newborn screening programmes for sickle cell disorders.

She has been involved in many teaching and CME programmes and has also given lectures for the “Master of Science in Haemoglobinopathies” e-learning course, conducted by the University College London, London, United Kingdom.

She has received several awards and has been invited to participate in many national and international meetings. She has published over 225 papers in national and international journals.

Dr. Sein Win

Dr Sein Win graduated from the University of Medicine, Yangon, Myanmar, in 2000 and completed his Master Course in Internal Medicine at the University of Medicine, Yangon, Myanmar, from 2004 to 2006. Afterwards, he attended a specialist training course for Doctor of Medical Science in Clinical Haematology at the University of Medicine, Yangon, Myanmar, from 2008 to 2010. He studied Bone Marrow Transplantation at the Christian Medical College, Vellore, Tamil Nadu, India, from 2012 to 2013. He is a current General Secretary of the Myanmar Society of Haematology.

Dr. Shau-Yin Ha

Consultant, Department of Paediatrics & Adolescent Medicine, Queen Mary Hospital, The University of Hong Kong

Dr. Shau-Yin Ha is currently a Consultant of the Department of Paediatrics and Adolescent Medicine, Queen Mary Hospital, Hong Kong. He is Honorary Clinical Associate Professor of both the Departments of Paediatrics and Pathology at the University of Hong Kong.

He graduated from the University of Hong Kong in 1981 and subsequently obtained the Fellowship of Royal College of Physicians, Pathologists and Paediatrics and Child Health from UK. He is doubly qualified with fellowships in both Paediatrics and Pathology (Haematology).

He has been looking after children with haematological and oncological disorders for the past 25 years. His main areas of interests are thalassaemia, haematology, acute myeloid leukaemia, and haematopoietic stem cell transplantation.

He has previously had the opportunity to extend his services as chairman of the Hong Kong Paediatric Haematology & Oncology Study Group and the Hong Kong Society for the Study of Thalassaemia. He is currently the Board Director of two charitable organisations in Hong Kong, namely Children’s Thalassemia Foundation and Children’s Catastrophic Disease Foundation. He is also a Board Member of the Asia Pacific Iron Academy.
Shobha Tuli

Shobha Tuli is the founding member of Thalassemics India, established in 1987 in New Delhi. She served as Joint Secretary of Thalassemics India until 1992. From 1993 onwards, Mrs. Tuli is serving as Secretary of the Board of Directors of Thalassemics India. In 1993 she supported the establishment of a thalassemia unit at Sir Ganga Ram Hospital in New Delhi, known as the ‘Preeti Tuli Thalassemia Unit’.

Since 1996, Mrs. Tuli has been a member from India in the Board of Directors of Thalassaemia International Federation (TIF) representing Thalassemics India as its Voting Member. Mrs. Tuli has served TIF from many officer posts, including that of the President, and she is currently holding the position of Vice President on TIF’s Board of Directors. Mrs. Tuli has travelled extensively on behalf of TIF and has represented TIF with utmost commitment and devotion. Her vast experience on the needs and challenges regarding haemoglobin disorders in low-resource countries is invaluable and contributes significantly to the work of TIF in India and abroad. Since 2000, she has been the President of the “Federation of Indian Thalassemics”. She was advisor to the Indian Red Cross Society for three years from 2003-2006. She is Advisor & Coordinator at the Bone Marrow Transplant Centre at B.L. Kapur Memorial Hospital, New Delhi, India. She has served as one of the members of the Advisory Committee at the Manav Rachana International University from 2009-2011. She has been involved in a number of activities in India under different capacities to improve patient care, thalassemia awareness and its prevention. At present, she is actively involved at the government level to draw the attention of relevant officials to thalassemia patient’s rights, their care and the prevention of the disease, in collaboration with Thalassaemia International Federation and the Federation of Indian Thalassemics.

Mrs. Tuli has received the State Award from the Department of Social Welfare, Government of N.C.T. of Delhi, in 1998, for the extraordinary work done by Thalassemics India in the field of Child Welfare. She has also received the “George Englezos Award” bestowed by the Thalassaemia International Federation in 1999 for the services provided in the field of thalassemia.

Her contribution in bringing thalassemia awareness and management to the forefront at the national level is unique and she has travelled as a volunteer extensively, assisting, supporting and guiding patients across India with unlimited patience and sincere commitment. Her passion in life is to help thalassemics.

Prof. Suradej Hongeng

Professor Suradej Hongeng received his MD in 1987 from the Mahidol University in Thailand. He is specialized in Paediatrics and Haematology-Oncology. Between 2002 and 2008, he held the position of Assistant Dean of Research Affairs in Ramathibodi Hospital at Mahidol University in Thailand. He is a member of several societies including the American Society of Hematology (ASH) and the American Society for Blood and Marrow Transplantation. Throughout his career, he received several awards including the Outstanding Paediatric Resident Award from the University of Illinois in Chicago, Illinois, USA, in 1991, and the Young Investigator Award at Ramathibodi Hospital, Bangkok, Thailand, in 1998.

His recent research focuses on: frequency of thiopurine s-methyltransferase (TPMT) genetic mutation polymorphism in Thai children with acute leukaemia.
Faculty Members

Prof. Suthat Fucharoen

Professor Suthat Fucharoen is an active researcher in genetics, especially in the area of thalassemia. In addition to his clinical and academic duties, he has been a prolific researcher dedicated to furthering the understanding of the pathophysiology and therapeutic management of thalassemia, particularly the molecular genetics of the disease. As a result of many years of research, he has authored or co-authored more than 250 articles published in peer-reviewed local and international journals, including Hemoglobin, the British Journal of Haematology and Blood. He is also an editorial board member for Hemoglobin and the Journal of the Medical Association of Thailand. He is an active member of numerous local and international professional associations including the Society of Hematology of Thailand, the Genetic Society of Thailand, the New York Academy of Sciences, the American Society of Hematology, and the International Molecular Biology Network (IMBN), Asia-Pacific Division and HUGO.

For the last few years he has been involved in the setting of the Asian Network for Thalassemia Control aiming to help neighboring countries to prevent and control of thalassemia. Dr. Fucharoen had been involved in many local and international conferences by serving as member of the International Advisory Committee and/or scientific committee. He served as a secretary general of the first meeting of the Asia-Pacific Society of Human Genetics held in Bangkok in 1989 and secretary general of the combined meeting of HUGO Pacific and Asia-Pacific Society of Human Genetics in 2002. Dr. Fucharoen is well known among hematologists, molecular biologists and geneticists worldwide, especially in Asia.

Prof. Tim St Pierre

The University of Western Australia, Perth, Australia

Professor Tim St Pierre heads the Medical Physics programme at The University of Western Australia, Perth, Australia. He trained at the University of Liverpool, UK, gaining a BSc with Honours in 1983 and a PhD in 1986. Following postdoctoral positions at Murdoch University in Western Australia, he was appointed to the faculty at the University of Western Australia in 1995.

Professor St Pierre’s main scientific interests are in the application of physics to medicine and biotechnology. He leads a research group at the University of Western Australia that focuses on applications of magnetic measurement techniques for the non-invasive characterization of iron in biological systems. He and his team developed the non-invasive liver iron measurement technology, FerriScan®. Much of his research has been related to iron overload in thalassaemia patients.

He has published over 150 peer-reviewed papers in the fields of iron and magnetism in biology, biotechnology, and medicine, in journals such as Blood, Circulation, Magnetic Resonance in Medicine, Gastroenterology, and Magnetic Resonance Imaging. In 2010 he won a Clunies Ross Award from the Australian Academy of Technological Sciences and Engineering for his work on non-invasive measurement of tissue iron deposits and in 2014 he was an IEEE Magnetics Society Distinguished Lecturer giving lectures on Magnetic Materials in Medicine at 55 academic and industrial research institutions worldwide.
Assoc. Prof. Vip Viprakasit

Associate Professor Vip Viprakasit graduated in 1994 from the Faculty of Medicine, Siriraj Hospital, Mahidol University and from 1994 until 1999 he has received his residency in Paediatrics and Paediatric Haematology at Chiang Mai University and Mahidol University, respectively. He is currently Associate Professor in Haematology and also the Programme Coordinator for thalassaemia research at Siriraj Thalassaemia Centre since 2008.

Prof. Viprakasit has published over 100 articles and abstracts in peer-reviewed journals at both the national and international levels, including Science, Blood, Lancet, Human Molecular Genetics, British Journal of Haematology, etc. He made numerous presentations at international meetings and conferences. In 2006, he received the prestigious Thailand Research Fellowship from Thailand Research Fund. Since 2008, he has been the Asia-Pacific Medical Collaborator and member of the International Scientific Advisory Committee for Thalassemia International Federation (TIF). At present, he is running a research group focusing on molecular genetics in human diseases, especially thalassaemia, for which he received granting supports from several local and international funding agencies.

Viresh Piplani

Viresh is a technology professional from New Delhi, India. He has completed his Masters Degree in Computer Science and an MBA degree in Information Technology, from prestigious institutes.

He has over 12 years of experience in software development and is currently working in one of the largest software company in India as Technology Specialist.

In his free time he enjoys reading books and articles, going out with friends, travelling, photography, cricket and exploring things.

Prof. Wirawit Piyamongkol, MD, MSc, PhD, FRTCOG

Wirawit Piyamongkol obtained his Medical Degree from Chiang Mai University, Chiang Mai, Thailand, in 1998 and fulfilled Obstetrics and Gynaecology training in 1992 and obtained Sub-Board of Maternal and Fetal Medicine in 2002 from the Medical Council of Thailand. He also obtained an MSc in Prenatal Genetics & Fetal Medicine from University College London, UK, in 1998 and a PhD in Obstetrics and Gynaecology (Molecular Genetics) from University College London, UK, in 2001. His thesis topic was on single cell PCR for PGD of single gene disorders.

Wirawit Piyamongkol started his carrier as a lecturer at the Department of Obstetrics and Gynaecology, Faculty of Medicine, Chiang Mai University, Chiang Mai, Thailand, in 1992. He gained experience working at the UCL Centre for Pre-implantation Genetic Diagnosis, The Galton Laboratory, Department of Obstetrics & Gynaecology, University College London, during 1997-2001. He was promoted to be an assistant professor in 1997 and an associate professor in 2006. He is currently the head of the Department of Obstetrics and Gynaecology, Faculty of Medicine, Chiang Mai University, Chiang Mai, Thailand, since 2013.

Wirawit Piyamongkol is specialised in Molecular and Clinical Genetics in addition to Pre-Implantation Genetic Diagnosis and Prenatal Diagnosis. He has set up the first PGD Centre in Thailand and South East Asia. He is the principal investigator and collaborator of several research projects, holding several research grants. He published as the principal investigator and collaborator in several scientific papers. He is the member of the Medical Council of Thailand, the Medical Association of Thailand and the Royal College of Obstetricians and Gynaecologists of Thailand. He is also the reviewer of several International Journals, including Prenatal Diagnosis, Human Reproduction and Blood.
Prof. Yesim Aydinok, MD  
Department of Paediatric Haematology, Ege University Children’s Hospital, and Ege University Hospital Blood Bank, Izmir, Turkey

Yesim Aydinok graduated from Ege University Medical School in 1986. She was appointed in 1992 as a paediatrician at the Ege University Medical School, Izmir, Turkey. In 1999 she moved to the position of paediatric haematologist and proceeded with studies on haemoglobinopathies at the Royal Free Hospital School of Medicine in London, UK. She was first Visiting Lecturer and Senior Lecturer for clinical haematology at the Great Ormond Street Hospital in London, UK and afterwards she became Associate Professor at the Ege University Children’s Hospital. Dr. Aydinok has been appointed as Director of the University Hospital Blood Bank in 1999 and as Professor in Ege University Children’s Hospital in 2005.

Thalassaemia is the main focus of Professor Aydinok’s clinical studies and research. She has been involved in clinical trials for new iron chelators since 2003. Professor Aydinok has published 87 peer-reviewed articles, mostly in the field of thalassaemia.

She is an associate editor of Hematology, a member of the American Society of Hematology (ASH) and the European Hematology Association (EHA). She is a member and scientific advisor to the Committee of Haemoglobinopathies at the Ministry of Health in Turkey and Scientific Director of the Turkish Thalassaemia Federation. Professor Aydinok has been the Director and a member of the Committees on Haemoglobinopathies in the Turkish Paediatric Haematology Association and the Turkish Haematology Association.

Prof. Yingyao Chen

Yingyao Chen is Professor of Health Services at the School of Public Health, Fudan University (FUSPH), Director of the Key Lab of Health Technology Assessment (Ministry of Health) at Fudan University, and Chair of the Department of Hospital Management at FUSPH. He is also the Associate Dean of the School of Public Health, responsible for international collaborations.

He received his Bachelor of Medicine at Shanghai Medical University in 1991, his Master of Public Health at Shanghai Medical University in 1997, and earned his Ph.D. in Management at Fudan University in 2006. He took part in a visiting scholar program at the University of California, Los Angeles, United States, from 1999 to 2001.

His academic interests focus on health technology assessment, health policy, health economics, and hospital management. He was a PI of several projects funded by the World Health Organization, World Bank, China Medical Board, Ministry of Health, Ministry of Science and Technology, National Natural Science Foundation of China, and provincial health authorities of China. He has published 150 papers in Chinese or in English, of which 100 in Chinese and 6 in English are with first authorship and corresponding authorship. He is the editor-in-chief of Health Services Evaluation and Disease Burdens of Main Birth Defects and Economic Evaluation of Their Preventive Strategies in China. He is a co-author of 14 books.
With its lentiviral-based gene therapy and gene editing capabilities, bluebird bio has built an integrated product platform with broad potential application to severe genetic diseases and T cell-based immunotherapy. bluebird bio’s clinical programs include LentiGlobin® and Lenti-D™. LentiGlobin is currently in three clinical studies: a global Phase 1/2 study, called the Northstar Study, for the treatment of beta-thalassemia major; a single-center Phase 1/2 study in France (HGB-205) for the treatment of beta-thalassemia major and severe sickle cell disease; and a U.S. Phase 1 study for the treatment of severe sickle cell disease (HGB-206). Lenti-D is currently in a Phase 2/3 study, called the Starbeam Study, for the treatment of childhood cerebral adrenoleukodystrophy. bluebird bio also has ongoing preclinical CAR T immuno-oncology programs, as well as discovery research programs utilizing megaTALs/homing endonuclease gene editing technologies.
**DAY 1 - SATURDAY, 26 SEP 2015**

**Chairs: Nguyen Anh Tri, Vip Viprakasit**

08:30 - 08:55 Overview of Transfusion Dependent Thalassaemia management (TDT) - Chi Kong Li

08:55 - 09:20 Overview of Non-Transfusion Dependent Thalassaemia management (NTDT) - Anuja Premawardhena

09:20 - 09:45 Haematopoietic Stem Cell Transplantation (HSCT) current progress - Suradej Hongeng

09:45 - 10:00 - Panel Discussion

10:00 - 10:30 Coffee Break

**Chairs: Androulla Eleftheriou/ Aparna Singh Shah/Erica Wood**

10:30 - 10:55 Blood adequacy and safety - Challenges in the Asian Region - Aparna Singh Shah

10:55 - 11:15 Blood adequacy, safety and recruitment: the Vietnam model - Ngo Manh Quan

11:15 - 11:40 Blood safety international standards-the Australian Model - Erica Wood

11:40 - 12:05 Blood transfusion in Thalassaemia - Cheuk Kwong Lee

12:05 - 12:30 Mitigating infectious risks in transfusion practice - Yesim Aydinok

12:30 - 14:00 Lunch

**Chairs: Bach Quoc Khanh/Suthat Fucharoen**

Presentations selected from submitted abstracts:

14:00 - 14:06 α-Thalassaemia: The Maldivian story - Zileena Zahir

14:06 - 14:12 α-Thalassaemia Gene Mutations in neonates from Mazandaran, Iran (2012) - Hossein Jalali

14:12 - 14:18 Sanguinate™ as a Therapeutic Agent for thalassaemia - Kittiphong Paiboonsukwong

14:18 - 14:24 Prevalence of alloimmunization in Major Beta Thalassaemia in Northern Iran (2010) - Mehrnoush Kosaryan

14:24 - 14:30 Efficacy and Safety of Deferasirox in single vs divided dosage in Thalassaemic Children - Shruti Kakkar

14:30 - 14:36 Evaluation of iron overload in Thalassaemia patients - Nguyen Thi Thu Hà

14:36 - 14:42 Endocrine Complications in patients with Transfusion-Dependent Thalassaemia after Haematopoietic Stem Cell Transplantation - Wing-Shan Queenie See

14:42 - 14:48 Haematopoietic stem cell transplant (HSCT) outcome for thalassemia major in Amirkola Bone Marrow Transplantation (B.M.T) ward - Ahmad Tamaddoni

14:48 - 14:54 Factors affecting physical, emotional and social Health related quality of life of patients who are more than 12 years and diagnosed with BETA-Thalassaemia - MBKC Dayasiri

14:54 - 15:00 Thalassaemia micro-particles induced platelets activation - Saovaros Svasti
## DAY 1 - SATURDAY, 26 SEP 2015

### 15:00 - 15:30  Coffee Break

**Chairs:** Androulla Eleftheriou/Suthat Fucharoen  
**Country report presentations:**  
- 15:48 - 15:57  India - Roshan Colah  
- 15:57 - 16:06  Indonesia - Pustika Amalia Wahidiyat  
- 16:06 - 16:15  Philippines - Maria Liza Naranjo  
- 16:15 - 16:24  Bangladesh - Mahmood Chowdhury (Arzu)  
- 16:24 - 16:33  Vietnam - Nguyen Anh Tri  
- 16:33 - 16:42  Myanmar - Sein Win  
- 16:42 - 16:51  People’s Republic of China - Chen Ping  
- 16:51 - 17:00  Panel Discussion

### 15:30 - 17:00  Country reports

**Chair:** Nguyen Anh Tri  
17:00 - 17:05  Welcome and introduction - Nguyen Anh Tri  
17:05 - 17:30  Optimal Management of iron overload: targeting the liver, heart and beyond - Vip Viprakasit  
17:30 - 17:55  Establishing a comprehensive thalassaemia care programme: successes and challenges - Ibrahim Mohamed Hishamshah  
17:55 - 18:00  Questions and Answers

### 17:00 - 18:00  Novartis Oncology Satellite Symposium

**Chairs:** Nguyen Binh Minh/ Le Thanh Hang  
**Messages by:**  
- TIF - Panos Englezos  
- NIHBT/ViTA - Nguyen Anh Tri  
- ASEAN - Fernando Ferdinal  
- Ministry of Health of Vietnam

**Key Note Presentations:**  
- Haemoglobin Disorders: Access to care and prevention: The global picture - Androulla Eleftheriou  
- Gene Therapy - Philippe Leboulch  
- Traditional Music Show

### 18:00 - 20:00  Opening ceremony
### DAY 2 - SUNDAY, 27 SEP 2015

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<tr>
<th>Time</th>
<th>Session</th>
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<tr>
<td>08:30 - 10:00</td>
<td><strong>Symposium 3: Iron</strong>&lt;br&gt;Chair: John B. Porter/Vip Viprakasit&lt;br&gt;08:30 - 08:55 Iron metabolism and iron load in Haemoglobin disorders - Vip Viprakasit&lt;br&gt;08:55 - 09:20 Iron chelation in Transfusion Dependent Thalassaemia (TDT) - John B. Porter&lt;br&gt;09:20 - 09:45 Iron chelation in Non-Transfusion Dependent Thalassaemias (NTDT) - Ibrahim Mohamed Hishamshah&lt;br&gt;09:45 - 10:00 Panel discussion</td>
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<td>10:00 - 10:30</td>
<td>Coffee Break</td>
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<td>10:30 - 12:30</td>
<td><strong>Symposium 4: Common complications - disease and treatment related</strong>&lt;br&gt;Chair: Shau Yin Ha/ Dimitrios Kountouras&lt;br&gt;10:30 - 10:55 Liver Disease: Viral Hepatitis: Treat or Cure - Dimitrios Kountouras&lt;br&gt;10:55 - 11:20 Heart Complications - Athanassios Aessopos&lt;br&gt;11:20 - 11:45 Endocrine complications - Shau Yin Ha&lt;br&gt;11:45 - 12:10 Infections - Suthat Fucharoen&lt;br&gt;12:10 - 12:30 Silent cerebral ischaemia in Transfusion Dependent Thalassaemia (TDT) patients - Mehran Karimi</td>
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<td>12:30 - 14:00</td>
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<td>16:15 - 16:35</td>
<td>Screening and Counselling - Roshan Colah</td>
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<td>16:35 - 16:55</td>
<td>Obstetric approaches - Chanane Wanapirak</td>
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<td>16:55 - 17:15</td>
<td>Pre-implantation Genetic Diagnosis (PGD) - Wirawit Piyamongkol</td>
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<td>Panel Discussion</td>
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<td>16:15 - 16:35</td>
<td>Symposium 6: Prevention</td>
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<td>17:30 - 17:50</td>
<td>Epidemiology studies of the thalassaemia syndromes in Asia - Fred Piel</td>
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<td>17:50 - 18:10</td>
<td>Health technology assessment - Ying Yao Chen</td>
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<td>18:10 - 18:30</td>
<td>Generic or Substandard Drugs - Mahmood Hadipour Dehshal</td>
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<td>12:30 - 14:00</td>
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<td>14:00 - 15:00</td>
<td>Symposium 2: Treatment updates</td>
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<td>15:30 - 17:15</td>
<td>Symposium 3: Common Complications: Endocrine, Cardiac and Liver</td>
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<td>Haemoglobin Disorders: Access to care and prevention:</td>
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<td>The global picture - Androulla Eleftheriou</td>
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<td>Traditional Music Show</td>
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## DAY 2 - SUNDAY, 27 SEP 2015

**Chair: Bach Quoc Khanh/ Suradej Hongeng**

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<tr>
<td>09:00 - 09:25</td>
<td>Haematopoietic Stem Cell Transplantation (HSCT) - Who can benefit - Suradej Hongeng</td>
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<td>09:25 - 09:50</td>
<td>Quality and efficacy of life-saving drugs in thalassaemia - Mahmood Hadipour Dehshal</td>
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<td>09:50 - 10:00</td>
<td>Panel Discussion</td>
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**Coffee Break**

**Chair: Duong Ba Truc/Mahmood Hadipour Dehshal**

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<tr>
<td>10:30 - 11:00</td>
<td>Psychosocial support - Michael Angastiniotis</td>
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<td>11:00 - 11:30</td>
<td>New advances in the treatment of thalassaemia - John B. Porter</td>
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<td>11:30 - 12:00</td>
<td>MRI Technologies for Measurement of Tissue Iron Concentrations - Tim St. Pierre</td>
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<td>12:00 - 12:30</td>
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**Lunch**

**Chairs: Shobha Tuli/ Androulla Eleftheriou**

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<tr>
<td>14:00 - 14:25</td>
<td>The role of associations in education and advocacy - Androulla Eleftheriou</td>
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<td>14:25 - 14:40</td>
<td>Organising an association: Building up infrastructure - Shobha Tuli</td>
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<td>14:40 - 15:00</td>
<td>The Maldives Patients’ Survey - Jeehan Saleem</td>
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**Coffee Break**

**Chairs: Mahmood Hadipour Dehshal/Michele Rhee**

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<td>A day in a patient’s life - Viresh Piplani</td>
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<td>15:55 - 16:20</td>
<td>Clinical Trials: Understanding clinical trials - Androulla Eleftheriou</td>
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<td>16:20 - 16:45</td>
<td>The patients’ perspective on clinical trials - Mahmood Hadipour Dehshal</td>
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<td>16:45 - 17:10</td>
<td>Patient Advocacy and update on recent gene Therapy trial results - Michele Rhee</td>
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<td>17:10 - 17:30</td>
<td>Panel Discussion</td>
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**Closing remarks**

Panos Englezos, Nguyen Anh Tri
Poster Abstracts

● Epidemiology and prevention:


Bahrain success story in controlling sickle Cell Disease (1984-2015) - Al-Arrayed S.

● α-thalassaemia syndromes:

Haematological Characteristics of a Cohort of Sri lankans with - α^37 and - α^42 deletions - Jayawardana SMA, Wettasinghe KT, Sumathipala D, Goonasekera HWW, Dissanayake VHW.

α-thalassaemia: The Maldivian Story - Zahir Z, Saeed M, Asha AK, Umar A, Fiureen F, Zila M.

Mapping the frequency distribution and burden of α-Thalassaemia across South East Asia: A call for Data - Hockham C, Penman M, Gupta S, Weatherall D, Piel F.


● Blood transfusion: adequacy & safety.

SANGUINATE™ as a Therapeutic agent for Thalassaemia - Paiboonsukwong K, Sirankapracha P, Rattanaporn P, Thamprasert W, Fucharoen SM, Misra H, Bainbridge J.


● Iron overload and management:

Can Hydroxyurea also work as an iron chelator in β-thalassaemia patients? - Italia K, Ghosh K, Colah R.

Efficacy and safety of Deferasirox in single vs divided dosage in thalassaemic children - Kalra A, Kakkar S, Sobti PC.

Evaluation of iron over load in thalassaemia patients - Nguyen TTH, Bach QK, Nguyen AT, Le XH.

Cardiac and liver T2* improvements in β-thalassaemia major twin patients with severe iron-overloaded and Hepatitis C treated with Deferoxamine and deferiprone: a case report - Rahmartani LD, Wahidiyat PA.

Deferasirox in Thai children with thalassaemia: 4.5 years’ experience - Juntharaniyom M, Wijitphun W, Muangkao B, Thungtakoon N, Sinpugsa Y.


Endocrine:

Endocrine complications in patients with transfusion-dependent thalassaemia after haemopoietic stem cell transplantation - Wing-Shan QS, Yuet-Ling T, Cheuk DKL, Shau YH.

Clinical profiles and medical complications of patients who are more than 12 years and diagnosed with beta thalassemia major in Sri Lanka - Dayasiri MBKC, Mudiyanse RM, Kulathilake S.

Hepatological:

Assessment of liver function test in thalassemic patients with iron overload - Promson R, Wijitphan W, Tomanakan K, Juntharaniyom M.

Gene therapy and regulation:

Haematopoietic stem cell transplant (HSCT) outcome for thalassaemia major in Amirkola bone marrow transplantation (BMT) ward - Tamaddoni A, Mahmoodi H, Behkar M, Shirkosh S, Babak Tamaddoni B.

Use of multiplex ligation-dependent probe amplification (MLPA) in the identification of deletional mutations causing raised HBF in Singapore population - Tan GP, Lai AHM, Ng I, Law HY.

Fertility and Pregnancy:

Marriage and child bearing in patients with transfusion-dependent thalassaemia major - Zafari M, Kosaryan M, Aily A.

Quality care/Quality of life:

Factors affecting physical, emotional and social health related quality of life of patients who are more than 12 years and diagnosed with β-thalassemia major in Sri Lanka - Dayasiri MBKC, Mudiyanse RM, Kulathilake S.

Marriage and child bearing in patients with transfusion-dependent thalassemia major - Zafari M, Kosaryan M, Aliasgharian A.

Psychosocial support:

Perceptions of β-thalassemia major patients and their parents about medical students history taking behaviour - Mudiyanse RM, Dayasiri MBKC, Kulathilake.

Effectiveness of a patient-parent empowerment program among β-thalassemia patients in improving doctor-patient communication - Mudiyanse RM, Dayasiri MBKC, Kulathilake S.
Poster Abstract Sessions

- **Bone disease:**


- **Non-transfusion dependent thalassaemia:**

  A study on effectiveness of Hydroxyl-urea in a thalassaemia treatment centre in Eastern-India - Mukhopadhyay T.

  Arterial thrombosis in four Myanmar Non-transfusion dependent HbE-β-thalassaemia intermedia patients - Sein W, Hein HO, Kyaw TA, Kyaw Z, Moe H, Aye AG, Htun LN, Rai M.


  Thalassaemia microparticles induced platelet activation - Svasti S, Phongpao K, Kheansaard W, Morales NP, Fucharoen S, Pattanapanyasat K, Chaichompoo P.


  Report on patients with non-transfusion-dependent β-thalassemia major being treated with Hydroxyurea attending the Thalassaemia Research Centre, Sari, Mazandaran Province, Islamic Republic of Iran in 2013 - Kosaryan M, Karami H, Zafari M, Yaghobi N, Aliasgharian A.

  The effect and side effect of Hydroxyurea therapy on patients with β-thalassemia: a systematic review to December 2012 - Kosaryan M, Mandana Zafari M, Alipur A, Omran AH, Aliasgharian A.

- **Molecular Control:**

  Analysis of the mechanisms underlying the developmental regulation of embryonic and fetal β-like globin genes - Vadolas J, Mark Roosjen M, Betty Kao B, Gearing LJ, Blewitt ME, McColl B.

  Analysis of β-thalassemia mice to understand innate immune abnormalities in β-thalassemia patients - Siwaponanan, P Siegers J, Ng T, Svasti S, Fucharoen S, Wijburg O, Vadolas J.

= Oral & Poster
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Actively investigating new treatments for patients with beta thalassemia

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**Title:** BLOOD ADEQUACY, SAFETY & RECRUITMENT - THE VIETNAMESE MODEL

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**ABSTRACT:**

The voluntary non-remunerated blood donation (VNRBD) program was launched as national campaign in Vietnam in 1994. During more than 20 years, the efforts of Ministry of Health, Vietnam Red Cross and other stakeholders in the whole country have led to improvement of blood transfusion service. The total blood collection increased nearly 9 times in 20 years from about 138,000 in 1994 to more than one million units in 2014. The percentage of voluntary blood donation increased from 14% to 97%, which is the proof of success. Vietnam has centralized and modernized its blood transfusion network to improve blood safety; besides, Steering Committee of VNRBD was established in 2007 and now developed at all levels. Furthermore, the creative events and communication campaigns, good models of blood collection and good service for blood donors are the effective solutions to a safe, stable donor resource in Vietnam.
ABSTRACT:

TDTM includes beta TM, beta thalassaemia intermedia (TI) converting to TM, beta-E thalassaemia and alpha TM (Hb Bart’s) whom survive the neonatal period. Management of TDTM starts with correct diagnosis which is made by clinical, laboratory and family history. Genotyping is more commonly used to confirm the type of severe thalassaemia and predicts the clinical course. In this part of the world, the differentiation of TM from TI is important as the degree of anaemia in TI may be precipitated by intercurrent infection. TI may not require life-long transfusion but they are now facing other new challenges such as thromboembolism and late iron overload. TDTM requires regular blood transfusion once every 2-5 weeks. The goal of regular transfusion is to maintain adequate haemoglobin level even before each transfusion, usually above 9.5 g/dl. The child will be spared from medullary hyperplasia (skeletal deformity), extramedullary erythropoiesis (hypersplenism), and achieve normal growth and development. However TDTM may develop complications including allo-immunisation, transfusion-transmitted infection (HIV, HBV, HCV), immune modulation (susceptible to infection). They will invariably develop iron overload as the body cannot excrete the excess iron infused with each transfusion. Without iron chelation, TDTM patients will develop organ damages as result of the iron toxicity. Endocrine organs are particularly prone to iron-induced damage, they usually develop endocrinopathies after age of 10 years such as diabetes mellitus, hypothyroidism, hypo-parathyroidism, hypogonadism and growth hormone deficiency. Cardiomyopathy is the leading cause of death even with iron chelation therapy. To prevent iron overload and its related complications, excessive body iron must be removed by effective chelation therapy. The principle of chelation is to achieve low body iron without toxicity from chelation therapy. There are now three iron chelators available in the market, namely deferrroxamine (DFO), deferiprone (DFP) and deferasirox (DFX). DFO is the first iron chelator introduced for treatment of TDTM. It is effective to maintain low body iron, prevent organ damage and extend survival. Side effects include local reaction, impairment of bone growth, hearing and visual impairment. Drug compliance is the most challenging issue in using DFO as it has to be given as continuous subcutaneous infusion over 8-10 hours, at least 5 days per week. Adolescents may refuse the drug treatment and develop significant impairment of various organs. Oral chelators may overcome the problem of drug compliance. DFP is the first oral chelator licensed to be used in TDTM. It is taken 3 times per day and can achieve effective iron chelation, and especially beneficial in removing cardiac iron and prevent cardiomyopathy. Arthralgia and gastrointestinal upset are common side effects of DFP. Neutropenia and agranulocytosis is the most worrying side effects and patients may develop severe infection. If agranulocytosis occurs, the patient should not be re-challenged with DFP and alternative chelators should be considered. DFX is the most new iron chelator. It is taken once per day and has been shown to efficacious in removing iron from liver and heart, and lowering serum ferritin. There may be mild increase in serum creatinine, proteinuria is common and Fanconi Syndrome has been reported. Combination therapy with DFO and DFP has been shown to be more effective in iron removal. Very intensive iron chelation may also reverse some organ dysfunction such as heart failure, DM, liver disease and other endocrine dysfunction. To maintain a low body iron status, iron status should be monitored regularly. Serum ferritin is simple and can be monitored frequently, but it is also a reactant to many conditions such as infection. Serial monitoring is necessary to observe the trend. Liver iron reflects the total body iron, and was previously assessed by liver biopsy. It is now replaced by MRI liver which does not carry the risk of liver biopsy, e.g. pain, bleeding. MRI of heart, called T2*, is very useful to give non-invasive monitor of cardiac iron. T2* less than 10 ms should be managed with urgent intensive iron chelation as the chance of developing heart failure within one year is high. MRI of pituitary and pancreas also gives information of endocrinopathies. With serial monitoring of body iron, optimal iron chelation therapy can be planned. Good compliance to chelation therapy is still the
cornerstone for success of treatment, and TDTM is now having survival beyond 5th and 6th decades. Allogeneic stem cell transplant is still the only curative treatment for TDTM. HLA identical sibling donor transplant at young age (<10 years) is associated with thalassaemia-free survival over 90%. Recently unrelated donor transplant in selected patients also showed encouraged result with high survival rate. Gene therapy is under active research and hoping that it will be under clinical trial soon.

Summary
Iron chelation can prevent iron-induced damage and prolong survival. The introduction of oral chelators improves the drug compliance and significantly reduces mortality. Combination treatment may be more effective to remove body iron. Allogeneic stem cell transplant performed in selected patients could achieve high cure rate.
Title: EPIDEMIOLOGY STUDIES OF THE THALASSAEMIA SYNDROMES IN ASIA

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ABSTRACT:

α- and β-thalassaemia are both common haemoglobinopathies in Asia. Recent evidence suggests that HbH disease might not be as benign as previously thought. HbE β-thalassemia, the common form of severe β thalassemia in Asia, has a remarkably variable phenotype ranging from a transfusion-dependent disorder to a condition that is compatible with reasonable growth and development without treatment. Furthermore, recent studies showed that the prevalence of thalassaemias can vary substantially over relatively short distances. They represent an increasing burden and it is essential to refine our knowledge of the numbers of patients affected by the disorders to define sustainable prevention and management strategies at national and regional levels. Despite huge developments in genetic testing and genomics over the last three decades, reliable prevalence data is still very sparse for most Asian countries, particularly regarding the α-thalassaemia syndromes. A better understanding of spatio-temporal changes in the distribution of thalassaemia variants, for example caused by population movements within Asia, could also help adapting national policies. The epidemiological analysis of data collected through remarkable local efforts conducted across the region, combined with modern geo-statistical methods, could certainly provide a substantial contribution to define appropriate health policies to manage the burden of haemoglobinopathies in Asia, and ultimately improve the quality of life of thalassaemia patients.
Title: BLOOD TRANSFUSION IN THALASSAEMIA

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ABSTRACT:
Thalassaemia presents a wide spectrum of clinical problems and management issues for their caring physicians. It can be of minor and asymptomatic carrier trait to serious life threatening condition. With transfusion treatment, most patients with thalassaemia major could survive and nowadays many could enjoy almost normal life expectancies. Besides, some have even been treated successfully by haematopoietic stem cell transplantation donated by siblings, cord blood or unrelated donors. However, under most circumstances, blood transfusion remains the most important treatment modality in their life time which can be of a few units to repeatedly life-long. Therefore, better understanding blood transfusion enables both patients and caretakers working toward better clinical outcome and quality of life.

In fact blood transfusion in thalassaemia can be addressed in four aspects - namely adequacy, safety, quality and outcome. For those maintained on stable and regular transfusion therapy, their primary focus would certainly be on the adequacy and timeliness of the blood supply. However, at the same time, they always look for high quality of blood with minimal adverse transfusion reaction because of the frequent occurrences of febrile and allergic reactions in the past. At the clinical side, caring physicians not only concern on their responses to blood transfusion, but also monitor risk of adverse transfusion reaction, transfusion transmitted infection and iron overload. In laboratories, blood bankers have been regularly assessed the development of the red cell allo-antibodies and /or auto-antibodies that could delay in finding suitable blood for their treatment. With our present knowledge and clinical management strategies, most of them are well addressed and formulated into standard protocols. Adherence to the best practices with regular update contributes to significant improvement of thalassaemia care. Besides, understanding of any peculiar local issue in particular patients profile would further benefit their treatment outcome.

Recently, better thalassemia care has extended further to those non transfusion dependent thalassaemia who require intermittent blood transfusion. Together with increasing life expectancy of the thalassaemia major patients, the blood demand for thalassaemia patients will be increased. Therefore, development of the blood demand modeling has to be taken into consideration to ensure the sustainability of adequate blood safety.

1 Abstracts received from Faculty members of the Scientific Programme by 28 August 2015 have been included.
2 Abstracts arranged in accordance to their appearance in the Scientific Programme.
Title: COUNTRY REPORT: EPIDEMIOLOGY AND CURRENT STATUS OF MANAGEMENT OF THALASSEMA AND SICKLE CELL DISEASE IN INDIA

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ABSTRACT:
The thalassemias and sickle cell disease pose a significant challenge both for management of the existing patients as well as for control in a vast country like India. It is estimated that there would be more than 100,000 patients with β thalassemia and around 150,000 cases of sickle cell disease.

The mainstay of management of β thalassemia major patients in India involves safe and adequate blood transfusions and iron chelation. It has been estimated that 2 million units of packed red cells would be required for transfusion of thalassemia patients in India. Due to the consistent efforts of thalassemia societies and health professionals, care and management of thalassemia has been included in the 12th Five Year Plan by the Government of India. As health care is looked after by the state, diagnosis and management of Thalassemia patients is done by respective State Governments. However, under the National Health Mission, the Central Government supplements the efforts of the State Governments in providing health care services including the establishment of blood transfusion services. According to the guidelines of the National Blood Transfusion Council under the National Aids Control Organization, blood is provided free of cost to patients with thalassemia and sickle cell disease. Yet, studies show that only around 15% of patients receive optimum management. Although many thalassemia day care centres have been established in different cities both in Government and private hospitals as well as by the Indian Red Cross Society, there is a great urban-rural divide with majority of patients in rural areas being inadequately managed.

Iron chelation using Desferrioxamine was not a feasible option for many patients with β Thalassemia major both due to the high cost and need for continuous infusion. Monotherapy with deferiprone has been used extensively since several years and now many patients have moved to deferasirox. Some experience has also been gained on combination of deferiprone and deferasirox which was found to be safe and efficacious. Iron chelators are now provided free of cost in some states.

Around 4 to 10% of multitransfused thalassemia major patients develop alloantibodies, the most common ones being anti-Kell and anti-E. Although testing for hepatitis B, hepatitis C and HIV is mandatory, transfusion transmitted infections is still a problem.

Some studies have shown that wheat grass therapy helps to increase the hemoglobin level by about 1g/dl and increases the time interval between transfusions while others did not find any significant benefit. Hydroxyurea has been used beneficially in Thalassemia intermedia patients and a good response has been seen in over 70% of these patients.

There are now 10 to 12 bone marrow transplant centres in major cities and although the cost of BMT is much lower than in western countries, it exceeds the economic reach of most families. The availability of HLA matched donors is a limitation. Haploidentical transplants have also been successfully done. Marrow donor registries and cord blood storage facilities are now available.
The clinical presentation of sickle cell disease is relatively milder in Indian patients compared to African and Carribian patients. However, systematic follow-up of patients in central India over several years have documented significant morbidity and mortality. Fixed low dose hydroxyurea has been shown to be very effective for a clinical and haematological response in sickle cell anemia patients in India having the Arab-Indian haplotype. Comprehensive care during follow-up of newborn cohorts and educating tribal communities for home care of sickle cell anemia children has reduced the frequency of complications. Delivering health care using a mobile clinical unit in an outreach programme has also been demonstrated. Under the National Health Mission, different State Governments are establishing centres in different districts for diagnosis and management of Sickle cell disease patients.

Thus, the quality of life for both β thalassemia and sickle cell anemia patients has improved and as patients are now growing older, they are receiving multidisciplinary care. However, this is still largely restricted to urban areas and the same care needs to reach patients in rural areas as well.
Title: COUNTRY REPORT: CURRENT SITUATION OF THALASSEMIA MANAGEMENT IN INDONESIA

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ABSTRACT:
Thalassemia is the most common hemolytic anemia in Indonesia with gene frequency of β-thalassemia 3-10%, α-thalassemia 2.6-11% and HbE 1.5-36%. There are 80-100 new cases every year in Thalassaemia Centre Jakarta, and 7,670 thalassemia patients registered from all over Indonesia. The economic burden of lifelong blood transfusion and iron chelation therapy is approximately 23,000 USD/patient/year. Mostly are covered by national health insurance.

According to Indonesian Society of Haematology and Blood Transfusion guidelines, pre transfusion hemoglobin has to be maintained at 9 g/dL to have a normal growth. Because of leucodepleted blood and NAT screening is not widely used in Indonesia, patients still have risk of getting transfusion reactions and infections. Iron overload are commonly measured by serum ferritin and transferrin saturation every 3-6 months. Meanwhile, cardiac and liver T2* MRI can only be done in Cipto Mangunkusumo Hospital, Jakarta. All iron chelators are available in Indonesia.

Besides infection, cardiac dysfunction is the most common cause of death (46%) in Thalassaemia Centre Jakarta. Laboratory test such as ECG, echocardiography, cardiac and liver T2* MRI is done every 12 months. A cross sectional study was performed in 166 thalassemia major patients in Thalassaemia Centre Jakarta from April 2012 to December 2014. The median of cardiac and liver T2* MRI were 34.36 ms (3.25-75.95) and 1.62 ms (0.71-12.82) [p=0.019, r=0.18]. The frequency of mild to normal myocardium based on cardiac T2* MRI was 85.5% and 70% of them used deferiprone as single therapy. In contrast, there was only 23% subject with mild to normal liver iron; most of them had severe liver iron (52.4%).

During 1970s, the survival of thalassemia major patients in Indonesia was 7-12 years of age. With recent advances in diagnostic and comprehensive management, the survival of thalassemia patients is improved which leads to other psychosocial problems. Preventing newborns with thalassemia major should also be the focus of thalassemia management in Indonesia and can be prevented by doing premarital screening, prenatal diagnosis and genetic counselling. All of these require government’s commitment in terms of socializing, creating a policy and providing the facilities. Unfortunately, there are many obstacles to bring this issue as a national program because of demographic, ethnicity, religion and diversity of mutations and phenotypes. As a start, In June 2011 The Ministry of Health launches health technology assessment on thalassemia screening protocol and prevention program and has been done as a pilot project in 20 big cities with high gene frequency. Medical associations together with the Indonesian Thalassemia Foundation, non-governmental organization and medical students association increasing public awareness on thalassemia through seminars, social events, and massive thalassemia screening.
Title: COUNTRY REPORT - BANGLADESH

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ABSTRACT:
Bangladesh is one of the most densely populated countries in the world with an area of 1, 47,570 sq/m. The population of Bangladesh is about 156.6 million. Bangladesh has achieved success in reducing infant mortality rate, but morbidity is gradually increasing due to diagnosis of genetic disorders like thalassaemia. Thalassaemia is prevalent in all areas across the country and there is no data regarding the number of patients with this disorder. The Common type is HbE beta thalassaemia. The expected number of patients with beta thalassaemia and HbE beta thalassaemia born annually is about 1040 and 6443 respectively. There is a wide variation in the carrier status. In Chittagong a screening program in 1500 Bengali school students revealed prevalence of beta thalassaemia trait and HbE trait as 4.5% and 6.1% respectively. There are some prevention programs in a small scale but no awareness program exists until now. Diagnostic facilities are also very poor. Hb Electrophoresis is available only in private laboratories in big cities. There are no idea about HPLC and prenatal diagnostic procedures. Safe blood transfusion is not yet possible and not free of charge either. Safe blood transfusion is only possible in tertiary centres and not in district hospitals. It is also observed that the frequency of blood transfusion is not the same even in family members having especially Hb E beta thalassaemia. Blood donation programs for patients with thalassaemia are in a small scale. Most of the blood donors are relatives of the patients. Most of the patients are heavily iron loaded as they are not aware of taking iron chelating drugs. Iron chelating drugs are available only in big centers, they are costly and health professionals are not well informed about them. Three iron chelating drugs are available: Desferiprone, Desferioxamine, Deferasirox. They are all costly and usually prescribed separately or with combination.

So, in conclusion it can be stated that more epidemiological studies need to be conducted in the future. The current status of case management of thalassaemia is inadequate and the majority of patients in the country cannot afford the treatment. There are no specialized centres for the management of thalassaemia in Bangladesh in government and private levels, apart from few in Dhaka and Chittagong. Prenatal diagnosis is still in nascent stage. So there should be a national health policy regarding the management of thalassaemia in Bangladesh. International organizations like the Thalassaemia International Federation (TIF) should support in manufacturing certain drugs regarding management of patients with HbE beta thalassaemia, which is more prevalent in Bangladesh as well as in ASEAN countries.
Title: EPIDEMIOLOGY AND CURRENT STATUS OF CASE MANAGEMENT OF THALASSAEMIA IN MYANMAR - COUNTRY REPORT

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ABSTRACT:
Myanmar has high incidence of important haemoglobinopathies; α-thalassaemia - 10% to 56.9%, HbE - 1 to 28.3%, β-thalassaemia - 0.54 to 4.07%, with 1 to 4.9 births per 1000 infants with a major haemoglobinopathy. In Myanmar, -α^3.7 type of alpha-thalassaemia mutation is the commonest mutation. The commonest genetic abnormalities in Hb H disease are (--SEA/-α^3.7) (53%) and (--SEA/-α^4.2) (30%). Eighteen different beta-thalassaemia mutations have been characterized. The commonest beta-thalassaemia mutations are CD41/42 (-TCCT), IVS 1-1 (G→T), CD 17 (A→T) and IVS 1-5 (G→C).

There is no national thalassaemia registry but hospital registries exist. According to hospital-based records, HbE/β-thalassaemia accounts for 46% to 58%, and HbH 6% to 37%. There is no reference expert center but four specialist haematology centers exist. Molecular diagnostic facilities are available only at National Health Laboratory but not at primary and regional level. The diagnosis and management guideline at national level does not exist but TIF’s guidelines are used in specialist haematology centers. Thalassaemia patients are managed in government and private-owned hospitals by haematologists, physicians, paediatricians and general practitioners. Joint care with obstetricians, endocrinologists etc. is being practiced.

Blood transfusion was a cost-sharing service in the past but recently becomes a free-of-charge at public institutions. At National Blood Center, NAT screening is used, and leuco-depleted blood units (by centrifugation) are available but are not irradiated. Red cell allo-immunisation is seen in 5% to 9.7%. Anti-E and anti-c rhesus antibodies are commonest. Hepatitis B and C serology positivity rate is around 3.88% and 12.6% respectively. Free-of-charge hepatitis B vaccinations for patients and blood donors are recently introduced by a local NGO.

More than half of transfused thalassaemia (52% -93.25%) has crossed the serum ferritin cut-off (> 1000 ng/ml) to start iron chelation therapy. Only 20.4% to 33% of patients needing iron chelation are taking treatment. Desferrioxamine is unregistered in Myanmar. Deferiprone is available but not deferasirox.

Increased transfusion demand and symptomatic splenomegaly are the main indications for splenectomy, and sepsis is the most common complication of splenectomy.

Hydroxyurea for fetal haemoglobin modulation is using only by specialists. Sixty percentage of patients complied one year treatment without side effects, with reduction in transfusion requirement, modest increase in pre-transfusion Hb and elevation in HbF percentage.

Bone marrow transplantation for thalassaemia has not been started yet.

Growth stunting was observed in 70.5% of β-thalassaemia patients in Yangon Children Hospital. Among beta-thalassaemia intermedia patients, delayed puberty was observed in 25% of male and 85% of female. Osteopenia in 18%, osteoporosis in 21%, enlarged left ventricle in 17%, enlarged left atrium in 27%, enlarged right ventricle 77% and deranged glucose metabolism in 13% of beta-thalassaemia intermedia patients was also observed.

About 49.5% of thalassaemia families attending day care centre has financial burden having to use 10% to 40% of total monthly income for blood transfusion visit. National prevention program, structured population screening, genetic counseling, premarital counseling and prenatal diagnosis are not available yet.
Title: LIVER DISEASE; VIRAL HEPATITIS: TREAT OR CURE

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ABSTRACT:
Liver disease in multi-transfused patients with beta-thalassemia major is the synergistic effect of iron toxicity, HCV hepatitis and to some degree of congestive disease because of heart failure. However, contradicting data exist regarding the impact of each factor on liver fibrosis, either separately or in combination. Liver disease in these patients is progressing by age and modified by chelation treatment and treatment of HCV infection.

Despite the prolonged duration of their disease, adult patients do not present with end-stage liver disease and decompensated cirrhosis as frequently as expected, due to early cardiac death. Improvement in iron overload management has contributed to an overall prolongation of survival of these patients.

HBV infection remains the leading cause of cirrhosis and HCC worldwide but it is not common in thalassemic patients. Different types of patients with chronic HBV, HBeAg(+) and HBeAg(-) define the treatment objectives. HBV cure equals to HBsAg seroconversion. Clearance or reduction of HBV DNA reduces the risk of cirrhosis or HCC independently of the the HBeAg status. Approved treatments for CHBV are Interferons (standard or Pegylated) and viral protease inhibitors (nucleotide or nucleoside analogs, (NUCs)): Lamivudine, Adefovir, Entecavir, Tenofovir and Telbivune. NUCs are potent antivirals, administered orally with few and rare adverse reactions, used as treatments of indefinite duration due to the risk of recurrence of viremia, resulting in undetectable serum HBV DNA in 94% to 98% of patients, HBeAg seroconversion in 40% to 41% and HBsAg loss in 3% to 10%, after 5 years. Long-term viral suppression has been shown to reverse fibrosis.

The current principal goal remains viral suppression and prevention with HBV vaccination.

There is no vaccine for HCV available yet neither any effective post-exposure prophylaxis because of the genetic diversity of HCV that is an RNA virus with high replication, with a lot of genotypes and subtypes. Nevertheless, current HCV treatment equals to cure, with cure rates exceeding 90%, almost independently of the previous treatment status, the fibrosis stage, the viral load or genotype, without significant resistance issues. DAAs include NS3/4A protease inhibitors, the NS5B polymerase inhibitors and the nonstructural 5A protein, or NS5A inhibitors. The duration of treatment is short (8, 12 or 24 weeks), in respect of the previous treatment response, the presence of cirrhosis and the Q80K polymorphism for the GT1a. Difficult to treat patients like GT1 null responders, cirrhotics or transplant recipients can be easily and effectively treated with excellent SVR results.

Thalassemia syndromes have been traditionally associated with high hepatitis C prevalence rates due to the regular blood transfusion requirements. Treatment with interferon and ribavirin usually leads to anemia, poor tolerance and subsequently low clearance rates, thus often failing to halt progression to cirrhosis. Although current guidelines suggest that interferon–free and ribavirin–free regimens should be used in these patients, very few data have been yet presented. Treatment of hepatitis C with all oral regimens in cirrhotic patients with thalassemia is safe. Preliminary efficacy results are promising and seem not to differ from published SVR rates in other populations.
Title: HEART COMPLICATIONS IN β THALASSEMA

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ABSTRACT:

β-thalassemia is an inherited hemoglobinopathy characterized by reduced synthesis of the β-globin chains that results in chronic dyserythropoietic anemia. The molecular basis of the disease is usually the homozygous or double heterozygous inheritance of two abnormal genes of the β-globin locus. More than 200 mutations of β chain gene has been described. Depending on the clinical severity, two forms are distinguished: the severe, transfusion-dependent thalassemia major (TM) and the milder and usually transfusion-independent thalassemia intermedia (TI).

In thalassemia, heart complications still represent significant morbidity and remain the leading cause of mortality in transfusion dependent (TM) patients.

Pathophysiology of cardiovascular disease in Thalassemia (TM/TI)

A) Due to the basic molecular defect, common features exist in the forms of Thalassemia. Among these anemia, bone marrow expansion and extra-medullar hematopoiesis, hepato-splenomegaly, transfusion needs, increased intestinal iron absorption, RBC defects provoking hemolysis, impaired immune competence, are the most common seen.

B) Common features in thalassemia, depended on the form of the disease, lead to, a different degree of severity, pathogenetic mechanisms, for cardiovascular injury. The recognized mechanisms are: high output state, iron load, elastic tissue damage (hemolysis), NO reduction, hypercoagulability, susceptibility to infections, proinflammatory environment. High cardiac output is the leading mechanism in TI, while iron overload dominates the injury in TM. All the damaging mechanisms can be modified by selected treatment.

C) Clinical Consequences: In the majority of the patients, the cardiovascular injury and the clinical consequences are the result of a combined action of different pathogenetic mechanisms to the whole spectrum of the heart’ s components, despite the fact that, one mechanism may dominate the injury in a certain heart structure. A variety of clinical phenotypes has been described.

1. Pericardium involvement (Viral infections (susceptibility to infections)-Iron load)
2. Vascular complications a) Arteries (Increased arterial stiffness - endothelial dysfunction (Oxidative stress, Elastic tissue damage)) b) Myocardial infarction (Endothelial dysfunction, Hypercoagulability, inflammatory environment, Elastic tissue damage) c) Strokes (ischemic, hemorrhagic (Endothelial dysfunction, Hypercoagulability, Valvular disorders, Arrythmias, LV dysfunction, Elastic tissue damage)) d) Veins-Thrombosis and Thromboembolic complications (Hypercoagulability, Endothelial dysfunction)
3. Valvular involvement (regurgitation - stenosis)
4. Arrhythmias (PAC’s , AT, A.F -Atrio-ventricular conduction abnormalities PVC’s, VT)
5. Right-sided Heart involvement (Iron deposition(R.V. dysfunction)-Pulmonary hypertension (increased cardiac output, increased pulmonary vascular resistance))
6. Left-sided Heart involvement, (High output state, Increased arterial stiffness (afterload), Iron deposition (systolic and diastolic dysfunction), Infections (Myocarditis), Endocrine abnormalities, Arrhythmias Valvular involvement).
Title: ENDOCRINE DYSFUNCTION IN PATIENTS WITH TRANSFUSION-DEPENDENT THALASSAEMIA

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ABSTRACT:
Despite the improvement in survival, endocrine complications are still common in patients with transfusion dependent thalassaemia (TDT). Previous studies have reported 30 to 66% of TDT patients suffered from at least one endocrine complication. The varied prevalence is partly related to the differences in methods of monitoring, age of study cohort, transfusion and chelating regimen.

The cause for endocrine dysfunction among this population is mainly attributed to excessive iron deposition in various endocrine glands secondary to chronic blood transfusion. Chelating treatment with subcutaneous desferrioxamine was used for more than four decades but compliance is often a major issue. On the other hand, there is also concern to start iron chelation with desferrioxamine early, as it could cause adverse effect on bone growth. The availability of newer oral chelators - deferiprone and deferasirox, has further improved the control of iron overload. Their efficacy in removing both cardiac and hepatic iron is proven. However, data on their efficacy in preventing or reverting iron related endocrine dysfunction are still limited. Some studies had been done to assess the prevalence of endocrine complications among TDT patients managed with different chelation regimen in different era, but conclusion was not definite. This might partly be related to the variable ferritin levels achieved in different cohorts. In fact, many studies have shown the prognostic role of ferritin for endocrinopathies. There is also evidence that, intensification of iron chelation could prevent or even revert some early endocrinopathies, including hypothyroidism, hypogonadism and impaired glucose tolerance.

Myocardial and pituitary iron loading determined by magnetic resonance imaging (MRI) have been shown to be associated with the development of hypogonadism and diabetes mellitus. There is also evidence suggesting a long prodromal state of silent iron deposition before any overt endocrine dysfunction. However, the association between diabetes mellitus/ impaired glucose tolerance and the degree of pancreatic iron loading was not established in most studies. Therefore, monitoring of myocardial and pituitary iron loading with MRI might be a good tactic to allow early recognition of preclinical iron deposition. In addition, new strategies to screen for early endocrinophathies have emerged. These include continuous glucose monitoring (CGMS) to look for early glycaemic abnormalities and use of new biomarkers e.g. anti-mullerain hormones (AMH) and inhibin B for gonadal reserve assessment. All these strategies may allow early intervention with intensified chelation. Obviously, tight control of iron overload is one of the most important strategies to prevent or delay subsequent endocrine complications.

For patients who become transfusion-independent after haemopoietic stem cell transplantation (HSCT), post-transplant iron chelation by venesection or by chelator is often an integral part of the management. However, late endocrine complications, especially hypogonadism, are common among these patients. Contributing factors include previous iron overload before transplant, use of cytotoxic agents for conditioning, and corticosteroid use.
Title: INFECTION IN THALASSEMIA AND ITS MANAGEMENT

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ABSTRACT:
Infections are major complications and causes of death of severe thalassemic patients. Several studies demonstrated that thalassemic patients are more susceptible to infection than normal individuals. This may range from minor infections such as upper respiratory tract infection and diarrhea to pneumonia and septicemia. In splenectomized patients septicemia can be very acute and overwhelming leading to death in a short period. The cause of increased susceptibility to infections in thalassemia does not appear to be defective lymphocytes. Investigations have not yet been able to pinpoint the real mechanisms. Iron overload and severe anemia may be involved. However, the underlying mechanism seems to be very complex, involving reactions between thalassemic RBC vesicles, abnormal RBC surface, complement system and other adhesice molecules, platelet, coagulation factors and endothelium. An extensive study of causative organisms of such serious infections among thalassemic patients revealed Salmonella typhi, Escherichia coli, Streptococcus pneumonia, Klebsiella pneumonia, Pseudomonas aeruginosa and Staphylococcus aureus as the most common causative agents. A fungal infection by Pythium organism leads to arterial occlusion and gangrene of the legs.

Physicians must be aware of the potential life threatening infections in TDT patients who underwent splenectomy and patients should be educated for seek early care when fever develops. In patients at risk and with indicative symptoms, prompt initiation of empirical antibiotics is essential. Intravenous infusion of third generation cephalosporin, combined with gentamicin or ciprofloxacin or vancomycin.

Better understanding of underlying mechanisms and their impact on evolving infections, regional and community based differences in infectious risks and preventative measures may contribute to a reduction in infection-related mortality in thalassemia.
Title: MRI TECHNOLOGIES FOR MEASUREMENT OF TISSUE IRON CONCENTRATION: PITFALLS AND CHALLENGES

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ABSTRACT:
Magnetic resonance imaging methods have played a key research role in studies of iron overload in transfusion dependent patients giving new insights into the relationships between liver and cardiac iron loading, iron chelator dose, and morbidity. Currently there is a rapid uptake of these methods into routine clinical practice as part of the management strategy for iron overload in regularly transfused patients. Given the many different methods of data acquisition, data analysis, and published calibrations, several potential pitfalls exist in the establishment of such techniques on a given scanner. These pitfalls can result in erroneous calibration of scanners which can potentially lead to inappropriate decision making with respect to management of iron overload with chelators. Here we review the challenges and pitfalls of establishing MRI techniques that are suitable for tissue iron measurement in regularly transfused patients. The challenges are reviewed in the context of the levels of accuracy and precision required by haematologists managing iron overload with iron chelators.
Title: MRI - THE THAI EXPERIENCE

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ABSTRACT:
For thalassemia patients, treatment with iron chelation therapy is crucial for the prevention of complication and improvement quality of life. Several oral iron chelators are now available, hence, there is a need to appropriately monitor and follow up in a long term fashion to provide the best possible management of iron chelation practice. Implementation by mean of magnetic resonance imaging (MRI) on tailoring iron chelation therapy has become a standard care in iron monitoring of Thalassemia patients. The aim of our non-commercial program is to provide a technical assistant for the MRI iron measurements on both cardiac and liver and to collect anonymized clinical data on patients submitting their MRI exams for analysis under an IRB approved process. Image analysis will be accompanied by a completed data form briefly summarizing the patient’s current medical condition including medications, transfusion schedule, iron-mediated co-morbidities, and limited appropriate lab values. All data will be coded under unique study ID numbers and placed in a database linked to the MRI results. In the study, the participated sites can be category according to their available MR resource and personals as clinical, investigator, and research sites. The clinical type is the site, which prefer to refer cases to be acquired at the research site and also contribute patient data on Clinical Report Form (CRF) to be deposited into the database. After completed the analysis, the iron measurement result will be reported back to the site. For the other two types, MR acquisition will be performed at the sites and need to deposit CRF data into the database. The difference is that the investigator site still requires data to be analyzed at the center site while the research one can perform its own analysis. Currently, we have total of 6 sites in Thailand participate in the project: two clinical, two investigator, and two research sites. Up till now, there are total of 495 studies from 316 Thalassemia patient data in the database (150 males and 166 females; age, 18.3 ± 11.5 years). Most of the patients (92%) have no cardiac iron overload but have liver iron overload (72%) with serum ferritin levels of 2324 ± 2960 ng/ml. The program should benefit Thalassemia patients on proper monitoring of iron overload and increase awareness of clinicians on iron overload problem, and provide a solid database for health policy makers to allocate appropriate resources and supports to help tackle this common and important health complication.
Title: HAEMATOPOIETIC STEM CELL TRANSPLANTATION (HSTC) - THE SOUTH CHINA EXPERIENCE

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ABSTRACT:

Background
The optimization of both erythrocyte transfusion and iron chelating has resulted in a remarkable improvement in the life expectancy of patients with thalassemia major (TM). However, only curative therapy remains allogeneic stem cell transplantation (SCT). HLA matched sibling transplant (MST) has been commonly used for TM patients with well results. However, this option is unavailable to many patients as a result of a lack of compatible MS, especially in China. Matched unrelated-donor transplant (MUT) and Haploidentical-donor transplant (HIT) have be well performed in malignant disease, but few in thalassemia patients because of high risk of transplantation so far. To expand donor pool and to lower risk of HSCT from alterative donor, we designed a NF-08-TM HSCT protocol for MST, MUT and HIT for TM patients since 2009. Outcomes before June 2011 have published in “Blood” in 2012. Here we updated these outcomes.

Aims
To check stability and reliability of NF-08-TM protocol in MST, MUT and HIT for thalassemia patients.

Methods
357 consecutive patients with TM underwent SCT between January, 2009 and December, 2014 in our center, including 179 in MUT, 22 in HIT (≥ one HLA mismatch), and 156 in MST. Rate of male to female is 232:125. The median age at transplant was 6 years (range: 0.6-16), All patients were followed up until July 31, 2015. The median follow-up time is 36 months (range: 7-78). All patients received the NF-08-TM protocol, which included a new risk classification to adjust dose of IV Busulfex and Cyclophosphamide and a conditioning regimen of Bu following Cy instead of Cy following Bu. Simultaneously, a intensify graft versus host disease (GVHD) prophylaxis consisted of ATG, Cs A, MMF and short MTX was given.

Results
The estimated 5-year overall survival and TM-free survival were 92.4% and 90.1%, 84.4% and 75%, and 94.2% and 91% in the MUT, HIT, and MST, respectively. Correspondingly, the cumulative transplant-related mortality was 7.6%, 15.6% and 5.8%, respectively. The cumulative incidence of graft rejection was 4.0% in total.

Conclusion
Our large dataset of prospective study provides that MUT or HIT was comparable with MST when using NF-08-TM SCT protocol when comparing MUT or HIT with the MST for TM patients.

Keywords
Hematopoietic cell transplantation, Sibling, Thalassemia, Unrelated donor
Title: SCREENING AND COUNSELING FOR THALASSEMI A AND SICKLE CELL DISEASE IN INDIA

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ABSTRACT:
India has a huge and diverse population with around 70% of people residing in rural regions. Thus community control of hemoglobinopathies is a difficult and challenging goal.

The burden of the thalassemias and sickle cell disorders is enormous. Different studies have estimated that 7500 to 12000 children affected with β thalassemias and 5000 to 7000 children with sickle cell disease are born annually. As yet there are no registries to document the exact figures.

Awareness and screening programmes have been undertaken since the last 4 decades or more by National Institutes, the Anthropological Survey of India, few medical colleges, Rotary and Lions clubs, other NGOs, thalassemia and sickle cell societies in different regions of the country which have shown that the prevalence of β thalassemia carriers varies from <1 to 17 % in different ethnic groups with an average of 3 to 4% for the country. As screening is now extended to rural areas in different districts, a high prevalence of β thalassemia is also observed in some tribal populations ranging from 6 to 12 %. The sickle gene is mainly restricted to the scheduled tribes, scheduled castes and other backward classes in the sickle cell belt which includes the states of Gujarat, Maharashtra, Rajasthan, Madhya Pradesh, Chhattisgarh, Jharkhand and Orissa with pockets in Kerala and Tamil Nadu with the highest load in Madhya Pradesh.

Many populations in the north-eastern states have a very high prevalence of HbE carriers reaching >50% in some groups while in West Bengal the prevalence of HbE carriers is around 4%.

Screening and counselling has been undertaken in schools, colleges, antenatal clinics, community centres, corporate offices, among family members of affected children as well in rural areas by house to house surveys. Screening for the thalassemias has been done by CBC and HPLC analysis in majority of the reported studies, however NESTROFT has also been used in resource poor settings as a preliminary test. Borderline / Normal HbA2 levels with near normal red cell indices are increasingly being reported in β thalassemia carriers and they pose a diagnostic challenge especially in young couples where one of the partners is a classical β thalassemia carrier. Apart from silent β thalassemia mutations, associated δ gene mutations are increasingly being seen in β thalassemia heterozygotes reducing the HbA2 levels to normal.

Screening for sickle cell disorders is largely done using the solubility test followed by haemoglobin electrophoresis or HPLC particularly in rural regions. Newborn screening for sickle cell disease has also been initiated in tribal and non-tribal populations in Maharashtra, South Gujarat, Chhattisgarh and Madhya Pradesh during the last 4 to 5 years and cohorts of sickle cell disease babies are being followed up and given comprehensive care to reduce morbidity and mortality.
Prenatal diagnosis programmes are expanding to more centres in different regions of the country. Most of the counsellors are medical social workers with in-house training at centres undertaking screening and control programmes. Given the ethnic diversity of the population, >70 β thalassemia mutations have been identified and novel mutations continue to be reported. ARMS and reverse dot blot hybridization are the 2 approaches used for prenatal diagnosis at the 10 to 12 centres offering these services mainly in big cities.

Thus utilizing the existing public health system which reaches out to 70% of the population is important for these programmes to reach rural areas where they are most needed. The recent capacity building workshops conducted by the Thalassemia International Federation in different regions of India have provided significant motivation to many NGOs and other groups to work together. At the same time the Central and State Governments as well as the National Health Mission through various programmes are providing support for community control of the haemoglobin disorders.
Title: PREVENTION OF THALASSEMIA: OBSTETRIC APPROACHES

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ABSTRACT:
Thalassemia refers to a group of inherited hematological disorders. They occur most commonly among Asian people, especially of Southeast Asian, Southern Asian and Mediterranean populations. Complications of thalassemia can be prevented when patient with thalassemia major are treated with regular blood transfusions to keep their hemoglobin level around or more than 10 g/dl. But frequent blood transfusions also lead to an accumulation of iron in the body, which can damage the heart, liver, and other organs. Iron chelator can helps the body to eliminate excess iron and prevent or delay problems related to iron overload. While these conventional therapy involving blood transfusions and iron chelation are expensive, prenatal approaches by prenatal diagnosis and selective abortion are more economical, which should not be carried out indiscriminately. This approach needs an accurate diagnosis and very good understanding of the disease.

In Thailand, the type and frequencies of thalassemia are heterogenous, both α and β thalassemia and some abnormal hemoglobins such as Hb E and Hb Constant Spring. The frequencies are 20-30% for α thalassemia, 3-9% for β thalassemia, 10-50% for Hb E and 1-8% for Hb Constant Spring. These abnormal genes in different combinations lead to over 60 thalassemia syndromes. Due to the large number of thalassemic patients and limited medical service resources, it is not possible to give optimal blood transfusions and iron chelating agents to the majority of patients. The best approach to cope with thalassemia in developing countries, including Thailand, is to prevent birth of new cases with major thalassemic disease.

In 2004, our team reported the prevalence of thalassemia in pregnant women in northern area of Thailand, with the overall prevalence of thalassemia trait or heterozygote being 25.4%; which classified as α thalassemia 1 trait were 6.6%, β thalassemia trait 3.75, Hb E trait 11.6%, homozygous Hb E 0.8%. The combination of α thalassemia 1 trait and β thalassemia trait was 1.2%, combination of α thalassemia 1 trait and Hb E traits were 1.5% and also found β thalassemia/Hb E disease as 0.2%. This high prevalence of thalassemia trait leads to many births of severe thalassemia as previously described. Before the prevention and control program by prenatal approach was initiated in 1994, our department encountered about 20 cases of homozygous β thalassemia, 30-40 cases of β thalassemia/Hb E disease and 20-30 cases of Hb Bart’s hydrops each year, from the total births of 6000-8000.

Chiangmai Strategy for The Prevention and Control of Severe Thalassemia
Since 1998, the Chiangmai team developed a feasible method and have had great success in control of severe thalassemia, simply and at much lower cost. Later, we have modified the original strategy to be more accurate and effective. The strategy includes:

1. Genetic counseling

2. Identification of pregnancies at risk by:
   a. Retrospective screening (history review of known risk)
   b. Prospective screening for asymptomatic women
      1. Screening test: OF test and Hb E screening
      2. Diagnostic test (if both partners of the couple are positive according to the screening test):
         a. Hb A2 level and PCR for α trait 1 if OFT positive
         b. Hb A2 level if OFT negative but Hb E positive
3. Prenatal diagnosis for pregnancy at risk
   a. Prenatal counseling
   b. Cordocentesis at 18-22 weeks’ gestation
   c. Fetal blood analysis with high performance liquid chromatography (HPLC)

4. Termination of the affected pregnancy

The prospective screening consisted of OF test and Hb E screening tests in women with no risk and testing the husbands of the women with a positive result. Subsequently, the OF test was replaced by MCV when the automated cell counters became available nationwide. If both partners had a positive result, the further diagnostic test (Hb A₂ and PCR) for the carrier was needed. A pregnancy in which both partners of the couple were carriers was considered a couple at risk (CAR), the further in-detail counseling and cordocentesis was offered for prenatal diagnosis of the severe thalassemia syndrome. (Figures 1 and 2)

**Figure 1** Main scheme for the screening of thalassemia carriers, Chiangmai strategy

**Figure 2** Outline of thalassemia screening techniques.
Presently, we introduced the IC strip test (immunochromatographic strip test) for detection α thalassemia 1 (SEA type) in the area which PCR test was not available.

Since the program was started the number of new cases of severe thalassemia at our institute is gradually decreasing. In the very first few year of program, we evaluated the cost-effectiveness of the program and found that among the total pregnant population of 21,000 individuals that we screened, 80 affected fetuses had been detected and terminated. The total cost of the prevention program was about US$ 257,140 and the cost of management of these affected cases, if they had been born, would be US$ 7,200,000. The cost-benefit ratio is 1:28, which is a very highly cost-effective project.

**Conclusion**

Thalassemia is one of the major public health problems in Asia and Thailand. The strategy for a control program consists of treatment and prevention; treatment of existing cases in the most cost-effective way, and reducing the new cases. This should define the methods of treatment and prevention appropriate to each individual country. To achieve success in the prevention and control of thalassemia needs continuity and holistic approach. It is expected that with optimal collaboration and support, effective prevention and control can be achieved. This will lead to better quality of life for the next generation.

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**Table 1** Effectiveness of the prevention and control program at Faculty of Medicine, Chingmai University, between 2003-2013.

- **Estimated couple at risk 2.4% (367 couple)**
- **Missing couple at risk (CAR) 46 12.5%**
- **Efficacy of CAR detection 87.5%**
ABSTRACT:
Thalassemias are the most common single gene disorder worldwide and cause significant health problems in endemic areas. At present, the strategy to prevent new cases includes population screening, genetic counseling, offering prenatal diagnosis (PND) to the couples at risk and termination of affected pregnancy (TOP). Preimplantation genetic diagnosis (PGD) or embryo selection is an alternative to the traditional PND giving the couples at risk a chance to start a pregnancy with a disease free baby. PGD is the earliest form of PND which allows selection of unaffected embryos prior to pregnancy establishment assuring that the baby will be healthy and TOP can be eliminated. Genetic materials from embryos at preimplantation stage can be retrieved by polar body biopsy, cleavage stage embryo biopsy or blastocyst biopsy using a micromanipulator. Genetic analysis of chromosome abnormalities and sex determination can be performed using fluorescent in situ hybridization (FISH) or comparative genomic hybridization array (aCGH) while single cell PCR is used for analysis of single gene disorders. Major problems concerning single cell PCR include PCR amplification efficiency, allele drop out (ADO), contamination risk and various DNA analysis techniques are discussed. Novel PCR protocols for alpha- and beta-thalassemias were developed and tested. Clinical PGD cycles and the first baby from PGD for alpha- and beta-thalassemias using multiplex fluorescent single cell PCR in Thailand are also demonstrated. A total of 40 PGD cycles have been performed giving rise to a total of 15 successful healthy pregnancies.

Keywords: allele drop out (ADO), embryo selection, preimplantation genetic diagnosis (PGD), prenatal diagnosis (PND), single cell multiplex fluorescent PCR, thalassemia
Title: HEALTH TECHNOLOGY ASSESSMENT

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ABSTRACT:

In the present context, health technology includes drugs, devices, medical and surgical procedures used in healthcare delivery, the knowledge associated with this, as well as organisational and support systems, within which care is provided. Health technology assessment (HTA) is a multidisciplinary activity that systematically examines the technical performance, safety, cost-effectiveness, organizational implications, social consequences, legal and ethical considerations of the application of a health technology. Its main purpose is to inform technology-related policy making in health care, and thus improve the uptake of cost-effective new technologies and prevent the adoption of technologies that are of doubtful value for the health system.

HTA provides a bridge between research and decision-making, including policy and clinical decision-making. It provides evidence-based information to help make decisions on the selection and utilization of health technologies (including emerging new technologies), to promote efficient and appropriate health resource allocation, and to control costs while maximizing value for patients and the health care system. HTA has been gaining recognition internationally and has played an increasingly important role in health policy-making.

A case study to measure the economic burden of Down’s syndrome (DS) in China and cost-effective analysis of prenatal diagnosis intervention for Down’s syndrome is presented to demonstrate HTA’s application in the field of birth defects. The economic burden of DS is calculated from direct healthcare costs, direct non-healthcare costs and indirect costs. The incidence approach is employed to measure the lifetime economic burden of a new DS birth in China in 2003. The average lifetime economic burden of a new DS case from the family perspective and the societal perspective amount to US$47,000 and US$55,000, respectively. Indirect (productivity) costs are responsible for most of the total economic loss. Sensitivity analysis shows that the incidence rate, survival rate, value of productivity, labor capacity of people with DS, and utilization rate of related services are influencing factors to the economic burden of DS. The economic burden of DS is substantial for the family of a person with DS, as well as to society. It is urgent to offer DS families social support to reduce their heavy burdens, and to implement a prenatal screening strategy to reduce the incidence of DS births.

In current clinical practice in China, for a cohort of 10,000 pregnant women, the strategy which delivers karyotyping by chorionic villus sampling (CVS) or amniocentesis (AC) only to those pregnant women aged 35 and above (maternal age screening strategy) can detect 0.67 DS births. The strategy which offers the diagnostic test after maternal serum screening with α-fetoprotein (AFP) and human chorionic gonadotrophin (hCG) double test (maternal serum screening strategy) can detect 1.41 DS births. The cost per prevented DS birth by the maternal age screening strategy and maternal serum screening strategy is $13,091 US dollars and $56,048 US dollars, respectively. Sensitivity analysis shows that the maternal serum screening strategy can be cost-effective if uptake rate of CVS or AC for patients with positive serum tests increase while the cost of serum screening decreases.
As an inherited hemoglobin disorder, thalassemia is prevalent among people of Mediterranean ancestry and Southeastern Asian. A study effort will aim to evaluate the Guangxi’s thalassaemia prevention program in China, which is composed of massive campaign of public education, screening and diagnosis, three tier integrated health delivery system (prenatal diagnosis centers, prenatal diagnosis sub centers, free premarital health care and one-stop marriage registration service), human resources and capacity building, facility development (including preliminary screening laboratories), etc. A HTA study of thalassaemia and its prevention will be underway, including measuring the economic impact of patients with thalassemia, and evaluating he prevention program’s effectiveness, cost-effective, and other impacts.
Title: GENERIC AND COPY DRUGS

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ABSTRACT:

Introduction
During recent decades, raising the awareness of persons with thalassaemia and their relatives has triggered an immense demand for standard care and high quality medicines. However, the governmental subsidiary has failed to catch up with the demand especially in developing countries and the decision makers have used generic or copy drugs to compensate for the shortage of budget. The strategy boomed a few questions about the quality of generic or copy drugs.

Material and Method
The investigation aims to evaluate the outcome of switching into generic or copy drugs in treating patients with thalassaemia. To reach authentic results, we review clinical trials conducted to evaluate the efficacy of generic and copy drugs together with a few chemical analyses on the drugs. Both clinical data and physicochemical material has been utilized to indiscriminately judge about quality of the drugs.

Results
The results of the study shows that a few generic and copy drugs which are used for thalassaemia treatment are contaminated with either expected or unexpected impurities; hence, they may cause postponed adverse drug reactions in the patients.

Conclusion
Although affordability is a vital factor for providing patients with medicines in developing countries, decision makers should take more responsibility for the quality of medicines especially used for treatment of patients who are dependent on long time treatment. More restrict regulations should be imposed to assure consumers about the drugs’ quality.
Title: HAEMATOLOGICAL CHARACTERISTICS OF A COHORT OF SRI LANKANS WITH -α^3.7AND -α^4.2 DELETIONS

Abstract Category: α-Thalassaemia Syndromes

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Declaration: The results of this study was presented locally at the University of Colombo Annual Research Symposium 2014

ABSTRACT:

Background
In alpha thalassaemia deletions are commoner than non-deletional mutations. The -α^3.7 and -α^4.2 deletions are very common deletions worldwide as well as in Sri Lanka. This study describes haematological phenotypic characteristics of individuals with -α^3.7 and -α^4.2 deletions.

Methodology
This descriptive study was carried out at the Human Genetics Unit, Faculty of Medicine, Colombo. Genotyping for -α^3.7 and -α^4.2 deletions was done using gap-PCR methodology. Automated full blood counts, Bio-Rad© HPLC haemoglobin quantification data and Leishman stained blood pictures were analyzed.

Results
Thirty one individuals carrying -α^3.7 and -α^4.2 deletions were studied. Twenty three (74%) were asymptomatic and recognized incidentally. Five (16%) were symptomatic at presentation. Three were recruited for family screening. Fifteen (48.4 %) were homozygous and nine (29%) were heterozygous for the -α^3.7 deletion while the αα/-α^4.2 and -α^3.7/-α^4.2 genotypes were observed in five (16.1%) and two (6.5%) individuals respectively. Majority (n =26, 84 %) showed a phenotype compatible with the genotype.

The mean corpuscular haemoglobin(MCH) was consistently low in all the study subjects(14-25.9 pg, mean 21.65±2.98). Except for haemoglobin levels (p- 0.047) no statistically significant difference of the red cell parameters or the HPLC data were seen between the different genotypes. Similarly the presence of different morphological cell types in the blood picture did not vary significantly between genotypes.

This cohort was further analyzed according to number of defective genes [One defective gene (αα/-α^3.7 and αα/-α^4.2) and Two defective genes(-α^3.7/-α^4.2 and α^3.7/-α^4.2)]. There was no statistically significant difference in either in RBC parameters, HPLC parameters or the presence of different red cell morphological types except target cells. The presence of target cells was significantly higher (p- 0.045) in individuals with two defective genes.

Conclusions
Majority showed typical haematological features of α thalassaemia carriers. Patients with atypical features need further genetic studies.

1 Abstracts received by 28 August 2015 have been included.
2 Abstracts arranged by topic area.
Title: ALPHA THALASSAEMIA: THE MALDIVIAN STORY

Abstract Category: α-Thalassaemia Syndromes

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ABSTRACT:

Background
The α-thalassemia is one of the most common monogenetic hereditary disorders worldwide. Molecular diagnosis for α-thalassaemia was initiated in the Maldives as part of a population screening program for the prevention of β-thalassaemia.

Method
Results of 7253 individuals referred to our genetics laboratory between the years 2005 and 2014 were evaluated. Analysis for α-thalassemia gene mutations were carried out using Multiplex GAP polymerase chain reaction (PCR) for three common α-gene deletions (-α(3.7), -α(4.2), and --α(SEA)) and for--α (Fil), --α (Med) and --α (Thai) mutations in certain instances. Also 529 cases of known β-thalassaemia carriers were analyzed for co-inheritance of α-thalassemia.

Results
Most common gene abnormality observed was αα/α 3.7 (49.1%), followed by α 3.7/α3.7 (18.8%), αα/α 4.2 (1.2%), α 3.7/α4.2 (0.4%) and α4.2/α4.2 (0.1%). Thus α+-thalassaemia carriers accounted for 69.6% of the total cases analyzed. Conversely, 27.9% of the referral cases did not have any of the mutations (-3.7, -4.2 or -SEA). There were 6 suggestive cases of HbH disease where Hb Bart’s peak was observed with HPLC, together with HbH inclusions detected with brilliant cresyl blue stain, although only α3.7/α3.7 mutations were observed. Additionally, 2.5% of the cases did not have any of the mutations while they had abnormally low red cell parameters. Furthermore, 40.6% of β-thalassaemia carriers had co-inherited α+-thalassaemia mutations.

Conclusion
Our findings suggest that α+-thalassaemia (namely, -α (3.7) single gene deletion) is highly prevalent in the population. Results are suggestive of the possible presence of undetected α0-thalassaemia mutations in the population. It is essential to expand our molecular facility to enable diagnosis of a wider range of alpha mutations, particularly α0-thalassaemia mutations. Furthermore, the relatively high prevalence of co-inherited α-thalassaemia among β-thalassaemia carriers indicates the importance of molecular analysis to diagnose double heterozygous cases.
**Poster Abstract**

**Title:** MAPPING THE FREQUENCY DISTRIBUTION AND BURDEN OF α-THALASSAEMIA ACROSS SOUTH AND SOUTHEAST ASIA: A CALL FOR DATA

**Abstract Category:** α-Thalassaemia Syndromes

**Authors:** Carinna Hockham¹, Bridget Penman¹, Sunetra Gupta¹, David Weatherall², Fred Piel¹

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**ABSTRACT:**

**Background**

α-thalassaemia is the commonest monogenic disorder in humans. The severe forms, haemoglobin H disease and Hb Bart’s hydrops fetalis syndrome, are particularly common in Southeast Asia where they represent a neglected health problem. Milder forms, although clinically benign, are important genetic modifiers of other inherited conditions, such as β-thalassaemia, and reach extremely high frequencies in certain populations in India and Nepal. Yet despite this, our understanding of the prevalence, genetic diversity and health burden of α-thalassaemia in South and Southeast Asia remains limited. To address this gap, we have begun to compile a systematic database of epidemiological information on α-thalassaemia, which will serve as a useful tool for addressing both basic science questions and applied public health queries.

**Methods**

We conducted a comprehensive search of online bibliographic databases to identify published surveys of α-thalassaemia and its genetic variants across South and Southeast Asia. Specific protocols were developed to deal with the genetic complexity of α-thalassaemia, compared with structural haemoglobin variants.

**Results**

We identified 1,336 unique references on α-thalassaemia in South and Southeast Asia. Amongst these, <10% contained informative data for this project, covering 58 specific geographical areas. Accounting for changes in diagnostic methods over time, the data indicates marked geographic heterogeneities in α-thalassaemia prevalence within and between countries, highlighting the importance of finer scale data.

**Conclusion**

Suitable data from internationally referenced published literature is limited, hindering efforts to refine our epidemiological knowledge of this disorder in Asia. It is clear that a large amount of data is present in local journals and unpublished reports. As a result, collaboration with international institutions, national screening programmes, and field researchers and clinicians is paramount to assembling the most complete evidence-base for the region. Such a resource is critical for developing appropriate and sustainable public health policies to better prevent and manage α-thalassaemia.
Title: ALPHA THALASSEMA GENE MUTATIONS IN NEONATES FROM MAZANDARAN, IRAN, 2012

Abstract Category: α-Thalassaemia Syndromes

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ABSTRACT:

Aim
Alpha thalassemia is one of the most prevalent disorders worldwide and carrier frequency of the disease is varied in different parts of the world. Although different studies in Iran and Mazandaran province have been carried out to identify different mutations of alpha globin gene among people with low hematological indices, frequencies of these mutations were unknown in general population, and thus the aim of this study was to evaluate the carrier frequencies of alpha globin gene mutations among neonates in Mazandaran.

Material and methods
Four hundred and twelve neonates were collected from a delivery ward of a hospital in Sari. DNA was extracted from their cord blood samples using phenol-chloroform-based method. For the detection of five common alpha thalassemia gene mutations, multiplex-GAP-PCR and PCR-RFLP methods were applied.

Results
Sixty three (15.29%, confidence interval, CI 95%: 11.81 -18.77) of investigated neonates had at least one of the five evaluated mutations. The -α3.7 deletion had the highest frequency (9.7%, CI 95%: 6.84 -12.56) and none of the neonates had -Med double gene deletion. The -α4.2 deletion, αααanti3.7 triplication, and α−5nt mutations had frequencies of 4.1% (CI 95%: 2.19 -36.01), 2.2% (CI 95%: 0.78 -3.62), and 0.49% (CI 95%: −0.18 -1.16), respectively.

Discussion
Our study showed that in most of the alpha thalassemia carriers just one copy of alpha globin gene was absent and they are not at risk of having children with Hb H disease or hydrops fetalis; however, up to 2.2% of neonates were carriers for αααanti3.7 triplication and they will be at risk for having a child with thalassemia intermediate if they marry a person which is a carrier of beta thalassemia.

Keywords
Alpha thalassemia, Allelic frequency -α3.7 deletion, αααanti3.7 triplication
Title: SANGUINATE™ AS A THERAPEUTIC AGENT FOR THALASSEMA

Abstract Category: Blood transfusion - Adequacy and Safety

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ABSTRACT:

Background
Chronic blood transfusions given to Transfusion Dependent Thalassemia (TDT) patients increase the risk of infection, iron overload and alloimunization. PEGylated carboxyhemoglobin bovine (SANGUINATE) is a therapeutic agent that can improve anemic condition by transferring oxygen to anemic tissues, reduce inflammation and inhibit vasoconstriction to avoid above problems. SANGUINATE has been proved to be safe and well tolerated in healthy human and stable sickle cell disease patients, and might be used to treat the hypoxia condition in thalassemia. This study presents results from Phase IIa trial of SANGUINATE on 12 beta-thalassemia patients with non-transfusion dependent thalassemia (β-NTDT).

Methods
Twelve β-NTDT patients between the ages of 18-55 year old were recruited into a Phase IIa trial. Eight patients received 4 weekly infusions of SANGUINATE followed by 2 monthly infusions. A total of 6 doses of 160 mg/kg of SANGUINATE were infused within 4 months. Four patients served as controls to receive Standard of Care (SOC). Patients’ safety, pharmacokinetics, erythropoiesis, oxidative stress, iron status and cardiac functions were evaluated.

Results
There were no serious adverse events that related to SANGUINATE. Hemoglobin and hematocrit level did not increase; however, patients had no anemic symptoms. Systolic and diastolic pressure were slightly increased in some patients following administration of drug and returned to baseline within 24 hours. One patient had an increase in right ventricular systolic pressure (RVSP) after infusion without any cardiac symptoms and returned to normal level within 72 hours. Iron status remained constant throughout study.

Conclusions
SANGUINATE was demonstrated to be safe for thalassemia patients. Higher doses of SANGUINATE should be studied. SANGUINATE will be a good option for thalassemia patients who developed alloantibodies to blood transfusions. Other hypoxic conditions related to thalassemia such as pulmonary hypertension and chronic leg ulcers may be also a good target for SANGUINATE treatment.
Title: PREVALENCE OF ALLOIMMUNIZATION IN MAJOR BETA THALASSEMIA IN NORTHERN IRAN (2010)

Abstract Category: Blood transfusion - Adequacy and Safety

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ABSTRACT:

Background and Objective
Repeated blood transfusion is the major treatment for patients with major thalassemia. However due to antigen encounters, it may initiate body reactions, including alloantibodies against red blood cell antigens. This study was done to determine the Prevalence of alloimmunization in major beta thalassemia patients in northern Iran.

Method
This descriptive - analytic study was carried out on 218 thalassemic patients (100 males and 118 females) with average age of 22.5±7 years in northern Iran during 2010. Each sample was tested for the presence of Alloantibodies including C, Cw, Lea, E, Lua, Leb, K, Jkb, N, P1, D, Jka, M, S, Xga, e, Fya, s, c, Fyb, k, Kpa, Jsb, Lub and Coa.

Results
Eighty eight cases (40.4%; 95% CI: 33.9–46.9) were positive for the presence of alloantibodies. Alloantibodies against C, Cw, Lea red blood cell surface antigens were the most prevalent (40%). No significant correlation was found between emergence of alloantibody with the age of initial, frequency and duration of blood transfusion.

Conclusion
Alloimmunization is a common observation in thalassemic patients and should be prevented by transfusing compatible blood.

Keywords
Beta thalassemia, Blood transfusion, Alloimmunization, Antibody
Title: IRON CHELATION AND OSTEOPOROSIS IN THALASSEMA. DOES IT HAVE ANY ROLE?

Abstract Category: Bone disease

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ABSTRACT:

Background:
To investigate the beneficial role of iron depletion on bone mineral density (BMD) of thalassemia patients suffering from iron overload, we compared the efficacy of 3 different iron chelation regimens.

Methods
In 52 consecutive patients with thalassemia major, BMD was measured by dual energy X-ray absorptiometry of lumbar spines and femur neck. The effects of treatment with 3 iron chelation regimen including deferoxamine, combination of deferoxamine and deferiprone, and deferasirox for a period of 12 months on BMD was compared. Results: Prevalence of patients with BMD below the expected range for age showed a borderline significant higher value in the lumbar spines compared to femur neck at chelation point 1 (80.8% vs 61.5%, P= 0.05) and was significantly higher at chelation point 2 (88.5% vs. 61.5%, P=0.001). Serum ferritin significantly reduced after 12 months iron chelation therapy (p= 0.001). The most effective regimen was combination of deferoxamine and deferiprone. None of the used regimens could alter BMD in the studied population. Overall prevalence of vitamin D deficiency was 57.7% in the studied population. Conclusion: Iron overload can adversely affect bone mineralization in patients with thalassemia. However, iron chelation cannot solely reverse this effect. Correction of other possible etiologic factors in the context of effective and long term iron chelation may improve BMD status of these patients.

Keywords
Beta-thalassemia, Bone mineral density, Chelation therapy, Iron overload
Title: ENDOCRINE Complications IN patients WITH transfusion-dependent thalassaemia AFTER haemopoietic stem cell transplantation

Abstract Category: Endocrine complications

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ABSTRACT:

Background
Endocrinopathies are common in patients with transfusion-dependent thalassaemia (TDT). Patients who had undergone haemopoietic stem cell transplantation (HSCT) successfully are also at risk of the endocrinopathies due to iron overload and sequela of conditioning chemotherapy.

Objective
To evaluate the prevalence and risk factors of endocrine complications in TDT patients after HSCT.

Method
We performed a retrospective review of endocrine complications in all TDT patients who underwent HSCT successfully from 1992 to 2013 at our hospital.

Results
Forty patients were included. Thirty had HLA-matched related donor transplant (cord blood: 10, cord blood+ marrow: 1, marrow: 19) and ten had unrelated donor transplant (marrow: 4, peripheral blood stem cell: 6). Busulfan/ Cyclophosphamide/ ATG as backbone were used for conditioning. The median age at HSCT was 8 years (range: 1-24 years). The median follow-up time was 11 years (range: 2-23 years). After transplant, seven patients did not receive any chelation, twenty-four had repeated venesection, three had desferrioxamine and six had deferasirox.

The commonest endocrine complication was hypogonadism (68.2%) followed by growth failure (25%). Older age at HSCT was associated with higher risk of hypogonadism (p=0.008). One female patient achieved motherhood after natural pregnancy. Eight patients (18.2%) developed hypothyroidism and required thyroxine replacement (primary 6.8%, secondary 11.4%). Risk factors for hypothyroidism included older age at HSCT (p=0.008) and post-transplant cardiac iron overload measured by MRI T2* (p=0.04). Other endocrine complications were less prevalent (adrenal insufficiency 9%, diabetes mellitus 2%). Serum ferritin levels, liver pathology grading and chronic graft-versus-host disease were not significantly associated with endocrine complications.

Conclusion
Hypogonadism, growth failure and hypothyroidism are the commonest endocrine complications post-HSCT in TDT patients. They should be regularly screened and treated accordingly. Performing HSCT earlier and better pre-transplant and post-transplant iron chelation might reduce the endocrine complications.
Title: CLINICAL PROFILES AND MEDICAL COMPLICATIONS OF PATIENTS WHO ARE MORE THAN 12 YEARS AND DIAGNOSED WITH BETA THALASSEMA MAJOR IN SRI LANKA

Abstract Category: Quality care for quality of Life / Endocrine complications

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ABSTRACT:

Background
The life expectancy of patients with thalassemia major has significantly increased in recent years, as reported by several groups in different countries. However, complications are still frequent and affect the patients' quality of life. This study aims to identify clinical profiles and medical complications among patients with beta thalassemia major in view of optimizing the currently provided care.

Methods
The study was carried out prospectively at university paediatric Thalassemia centre at TH Peradeniya, Sri Lanka. Forty patients with beta thalassemia major and who were more than twelve years were enrolled to the study. All patients were interviewed regarding their clinical profiles and medical complications by trained medical graduates over a period of three months using a pretested, semi-structured questionnaire.

Results
Forty patients were available for analysis and mean age was 17 years (range 12-24 years). 50% were male patients. 75% (30/40) of fathers and 70% (28/40) of mother had received secondary education. Most fathers (30%, 12/40) were manual labourers and most mothers (87.5%, 35/40) were house wives. 17.5% (7/40) had a family history of beta thalassemia major. 80% (32/40) were nuclear families. Mean age at diagnosis was six months. 42.5% (17/40) had been splenectomised. All were on regular iron chelation therapy. Endocrine related complications included hypoparathyridism (67.5%, 27/40), hypothyroidism (27.5%, 11/40), diabetes (12.5%, 5/40), pubertal delay (40%, 16/40) and short stature (42.5%, 17/40). Other complications included chronic liver disease (5%, 2/40), hypersplenism (25%, 10/40), arthropathy (10%, 4/40) and cardiac hemosiderosis (5%, 2/40). None had transfusion transmitted infections. One patient died during the study period.

Conclusions
Most patients with beta thalassemia major had difficult socio-economic backgrounds. Most complications were related to endocrine dysfunction following iron overload and were comparable to other studies in the region. None had transfusion transmitted infections.
Title: MOLECULAR EPIDEMIOLOGY OF HEMOGLOBINOPATHIES IN CAMBODIA

Abstract Category: Epidemiology and Prevention

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ABSTRACT:

Background
Determination of the magnitude of thalassemia problem in a country is important for implementation of a national prevention and control program. In order to acquire accurate thalassemia prevalent data, the gene frequency either of α- or β-thalassemia in different regions of a country should be determined as variation in the thalassemia gene frequencies over relatively short geographical distances had been reported.

Methods
The molecular basis of thalassemia in Cambodia was carried out in a community-based cross-sectional survey of 1,631 unrelated individuals from 3 regions of Cambodia; Battambang, Pheah Vihear and Phnom Penh. Detection of thalassemia mutations was performed by PCR-based technique i.e. gap-PCR for deletional α-thalassemia, dot blot hybridization for nondeletional α-thalassemia and reverse dot blot hybridization for β-thalassemia.

Results
Thalassemia was found in 62.7% of the 3 studied population composing of two most common β- and α-globin gene mutations, HbE and α-thalassemia 2 (-α3.7), respectively. A regional specific thalassemia gene frequency was observed. Pheah Vihear had the highest prevalence of HbE (55.9%), α-thalassemia 2 (46.0%) and non-deletional α-thalassemia (15.1%) genes, whereas Phnom Penh had the lowest frequency of thalassemia genes. Interestingly, in Pheah Vihear, the frequency of Hb Paksé (0.0357) was very high, almost equivalent to that of Hb Constant Spring (0.0438).

Conclusion
The result of this study indicates an importance of micromapping and epidemiology study of thalassemia, which will assist in the establishment of the prevention and control program of thalassemia in Cambodia.

Acknowledgements
This study was supported by Research Chair Grant, National Science and Technology Development Agency, and Office of the Higher Education Commission and Mahidol University under the National Research University Initiative.
Title: PATTERN OF HAEMOGLOBINOPATHIES WITH CAPILLARY ELECTROPHORESIS - A SINGLE CENTRE STUDY AT ICDDR, B

Abstract Category: Epidemiology and Prevention

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ABSTRACT:

Background
Haemoglobinopathies represent significant frequencies across almost all ethnic groups in Southeast Asia including Bangladesh posing a major genetic and public health problem. In Bangladesh; we have very limited data about the pattern and carrier frequency of haemoglobinopathies. Thus, the present study was thus undertaken to find out the pattern and spectrum of haemoglobinopathies in Dhaka city, the capital of Bangladesh.

Method
Retrospective analysis of 978 samples were carried out for haemoglobinopathies screening from January 2007 to May 2015 which has been submitted at diagnostic labs of icddr,b. Capillary electrophoresis (CE) was used as a first line of investigation for all patients who had a clinical or familial suspicion of haemoglobinopathies.

Results and conclusion
There were 978 abnormal chromatograms detected of 3640 population being studied. Of these, males were 457 (46.7%) and females were 521 (53.3%). Among these, 430 (43.96%) were HbE trait, 364 (37.21%) β-thalassemia trait, 91 (9.3%) HbE/β-thalassaemia, 68 (6.95%) HbE disease, 8 (0.81%) HbS trait, 6 (0.61%) Hemoglobin D Punjab and 3 (0.31%) cases were homozygous β-thalassaemia. Single case of heterozygous state with Hb Hope, Hb J-Kaohsiung, Hb Lepore and Hb C were also found. Overall, heterozygous (or carrier) states were 816 (83.4%), and homozygous or double heterozygous (or disease) state were 162 (16.6%) respectively.

Since the study populations were mostly referred cases, where the diagnostic facility is not much available, this may not be true prevalence of haemoglobinopathies; further work is needed to find out the pattern of haemoglobinopathies, in particular high-frequency populations and to provide solid evidence of health burden posed by the haemoglobin disorders in Bangladesh and other developing countries in Southeast Asia.
**Title:** BETA GLOBIN GENE HAPLOTYPES ASSOCIATED WITH HEMOGLOBIN D-PUNJAB IN NORTHERN IRAN

**Abstract Category:** Epidemiology and Prevention

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**ABSTRACT:**

**Background and Objective**
Hemoglobin D-Punjab is one of the variant of hemoglobin caused by a mutation on position 121 of beta globin gene which is frequent in India, Pakistan and Iran. Heterozygote form of this variant is mainly asymptomatic while in combination with hemoglobin S, severe form of anemia occure. This study was carried out to determine the beta globin gene haplotypes associated with hemoglobin D-Punjab in Northern Iran.

**Methods**
This descriptive study was carried out on families of 18 individuals whom were carriers of hemoglobin D-Punjab in Sari in Northern Iran. Genomic DNA was extracted from peripheral blood samples using Phenol-chloroform standard protocol. In order to identify different haplotypes associated with hemoglobin D-Punjab, PCR-RFLP method and family linkage analysis were used.

**Results**
In 17 subjects hemoglobin D-Punjab was linked to [+ - - - - + +] haplotype and in one case association with [- + + - + + +] haplotype was observed.

**Conclusion**
The hemoglobin D-Punjab alleles have mainly unicentric origin and [- + + - + + +] rare haplotype may have different genetic origin or is created as a result of gene recombination.

**Keywords:** Hemoglobin D-Punjab, Haplotype, PCR-RFLP, Iran.
Title: BAHRAIN SUCCESS STORY IN CONTROLLING SICKLE CELL DISEASE (1984-2015)

Abstract Category: Epidemiology and Prevention

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ABSTRACT:
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 1984-2015.

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 30 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.

Prenatal diagnosis is available since 2002 Common testing procedures include chorionic villi biopsy, amniocentesis, ultra sonography, and serum marker testing, followed by genetic screening.

In 1990, an Islamic 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. 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 days.
Results
Total number of patients who undergo PND during the last 13 years in our clinic was 200 patients. Fifty patients were for chromosomal testing and 150 patients for genetic blood diseases.

Percent of affected fetuses was low: 8%. Genetic counseling provided prior to testing and after getting the results.

Conclusion
PND service aim is to ensure the wellbeing 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.
Title: MARRIAGE AND CHILD BEARING IN PATIENTS WITH TRANSFUSION-DEPENDENT THALASSEMIA MAJOR

Abstract Category: Fertility and Pregnancy

Authors: Mandana Zafari, Mehrnoush Kosaryan, Aliasgharian Aily

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ABSTRACT:

Aim
With good care, patients with transfusion-dependent thalassemia major (TDTM) can reach older ages, marry and reproduce. This study was conducted by the Thalassemia Research Center.

Material and Methods
Medical notes of all TDTM patients and all non-transfusion-dependent thalassemia major (NTDTM) patients were reviewed from July to December 2012. Also, patients were interviewed. The questionnaire was made in consultation with research methodology experts and reliability was achieved by a pilot study of 12 patients, by the test-retest method (r = 0.9). Epidemiologic characteristics of patients and the pregnancy outcomes were recorded. Descriptive statistics were used with spss 17.

Results
Four hundred and nineteen medical records were reviewed. Three hundred and forty-five (82.5%) were TDTM. One hundred and seventy-five (50.7%) were female with a mean age of 25.4 ± 7.05 years and 42 (25%) had been married. Mean age of menarche and marriage was 15.4 ± 1.6 and 21.8 ± 4.5 years, respectively. Total number of live children is nine so far. Mode of delivery in female patients was cesarean section. Almost 78% of newborns weighed 2500 –4000 g. Almost 22% of pregnancies were assisted. Male patients consisted of 170 (49.3%) and 55 (32.3%) of them had been married. Mean age at marriage was 24.27 ± 3.5 years.

Conclusions
With better management, patients with TDTM can reach the age of reproduction. Medical teams should be prepared for this possibility.

Key words: fertility, infertility, pregnancy, thalassemia, transfusion-dependent thalassemia major.

Title: EXPERIMENTAL CHARACTERIZATION OF HB FLURLINGEN (HBA2: C.177C>G, p.HIS>Gln) AND HB BOGHÉ (HBA2: C.177C>A, p.HIS>Gln) REVEALS CONTRADICTORY HBA2 EXPRESSION AND TRANSLATION PATTERNS DESPITE IDENTICAL AMINO ACID SUBSTITUTIONS

Abstract Category: Gene Regulation and Therapy

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ABSTRACT:

Background
In this study, we describe the clinical features and provide experimental analyses of Hb Flurlingen (HBA2: c.177C>G, p.His>Gln) that contrasted with Hb Boghé (HBA2: c.177 C>A, p.His>Gln). Despite the identical amino acid substitution in both variants, Hb Flurlingen shows the phenotype of α-thalassemia (α-thal), whereas Hb Boghé has no impact on α2-globin (HBA2) production.

Methods
For in vitro transcription analysis, HBA2 expression constructs carrying the HBA2-WT (wild type), Hb Flurlingen and Hb Boghé sequences were generated and expressed in human bladder carcinoma 5637 cells for downstream analyses by quantitative real time-polymerase chain reaction (qReTi-PCR), and immunofluorochemistry (IFC). In silico analysis of secondary folding structures of the HBA2-WT, Hb Flurlingen and Hb Boghé mRNA sequences was performed using Mfold software.

Results
The gene transcription and translation analyses revealed that cells transfected with the Hb Flurlingen construct had significantly lower HBA2 transcription (~55.4%, p ≤0.01) and reduced protein synthesis when compared to the wild type group. In contrast, cells transfected with the Hb Boghé construct showed no significant changes in HBA2 transcription or translation activities when compared to the wild type group. The in silico prediction of possible effects of these mutations on the folding structures of the HBA2 transcripts showed a change of secondary folding pattern in the Hb Flurlingen transcript when compared to those of HBA2-WT and Hb Boghé.

Conclusion
Our experimental findings support the clinical presentation of an α-thalassemic phenotype for Hb Flurlingen in contrast with Hb Boghé, despite identical amino acid substitutions. The results confirm the importance of experimental analysis in establishing the impact of novel base substitutions.
**Title:** HEMATOPOIETIC STEM CELL TRANSPLANT (HSCT) OUTCOME FOR THALASSEMA MAJOR IN AMIRKOLA BONE MARROW TRANSPLANTATION (B.M.T) WARD

**Abstract Category:** Gene Regulation and Therapy

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**ABSTRACT:**

**Background**
Hematopoietic stem cell transplant is the main and definitive treatment for thalassemia major. For this purpose we tried to treat some of our thalassemia major patients who had suitable completely match donor with HSCT in Amirkola B.M.T ward.

**Methods and materials**
31 thalassemia major had underwent to HSCT from august 2010 to November 2014 in Amirkola B.M.T ward.14 Patients were female and 17 male, age distribution was between 3-26 years. The patients divided in two groups on the basis of Lucarreli classification (on the basis of portal fibrosis, hepatosplenomegaly and serum ferritin level). Group I were patients in class I and II and group 2 were patients in class III. Age distribution in group I was between 3 to 17 year and age distribution in group II was between 18 to 26 year. we collected all information about engraftment and B.M.T failure and rejection and survival and event free survival, also mortality and some side effect for instance GVHD. All information collected and analyzed.

**Findings**
Event free survival in group I was 75% and in group II was 40%. 2 out 16 patients in class I died and 5 patients in class III died. Severe GVHD happened in 6 cases, but only one happened in group I patient. Non engraftment patients happened in 13% cases and rejection happened in 4 patients in group II.

**Conclusion**
HSCT is a choice way to relieve thalassemia major patient from their problems but in class I and II specially in young patients has the best result.

**Keywords:** HSCT, major thalassemia, out come
Title: USE OF MULTIPLEX LIGATION-DEPENDENT PROBE AMPLIFICATION (MLPA) IN THE IDENTIFICATION OF DELETIONAL MUTATIONS CAUSING RAISED HBF IN SINGAPORE POPULATION

Abstract Category: Gene Regulation and Therapy

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ABSTRACT:

Introduction
Raised HbF is associated with polymorphisms in ⁶γ-globin gene or deletions in the β-globin gene cluster. The latter are known to cause δβ- thalassaemia or Hereditary Persistence of Fetal Haemoglobin (HPFH). With the high incidence of β-thalassaemia in Singapore, it is important to identify patients with raised HbF caused by β-globin gene deletions. Current strategy is limited to using Gap-PCRs to detect known deletions or the laborious and expensive southern blotting.

Objective
To evaluate using MLPA for the detection of novel and known deletions in the β- globin gene cluster in patients with high HbF levels.

Methods and Materials
DNA samples from 24 patients screened in Genetics Service were analysed. All patients had HbF >5% (range 5 - 28.7%) and were negative for 100kb Chinese ⁶γ(⁴γδβ) deletion and 28kb South East Asian (SEA) HPFH deletion. MLPA analysis was performed using SALSA MLPA KIT P102- B2 HBB kit (MRC Holland). It detects the gene dosage of the Locus Control Region (LCR) and HBE, HBG2, HBG1, HBD and HBB genes in the β- globin gene cluster.

Results
Eleven patients were found to have deletions in the β-beta globin gene cluster. One had a deletion of HBB gene (MCV 78.5fL and HbF 23.6%). Six had deletions from HBBP1 to downstream of HBB region (MCV: 68.2 -79.4fL; HbF: 20.9 -25.1%). Four had deletions from HBG1/G2 to downstream of HBB region (MCV: 73-78.3fL; HbF: 22.7-28.7%). Thirteen patients with no deletion had HbF ranged from 5-21.2%. Seven had HbF of 5-9.7%, four with 10.7-18.1% and two with 20.8-21.2%.

Conclusion
MLPA is efficient in detecting deletions in the β- globin gene cluster causing HPFH or δβ-thalassaemia in individuals with raised HbF of more than 20%. It provides an alternative to targeted screening for known deletional mutations and has the potential in identifying novel deletions.
Title: MICROARRAY ANALYSIS OF ERYTHROID PROGENITORS IN INDIVIDUALS WITH β-THALASSAEMIA

Abstract Category: Gene Regulation and Therapy

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ABSTRACT:

Background
β- thalassaemia is a disorder of globin gene synthesis resulting in reduced or absent production of the β-globin chain in red blood cells. Traditionally, the pathogenesis of β-thalassaemia has been attributed to ineffective erythropoiesis with intramedullary apoptosis of the erythroid progenitors. Recently, studies in mouse models have challenged this hypothesis with the concept of delayed progenitor maturation sited as a contributing factor to the ineffective erythropoiesis.

This study uses microarray technology to examine the erythroid progenitor mRNA of patients with transfusion dependent β-thalassaemia and compare it to erythroid progenitor mRNA from healthy controls.

Methods
Haematopoietic stem cells were isolated from the peripheral blood of 6 healthy controls and 6 transfusion dependant β-thalassaemia patients. Following 7 and 14 days in culture early- and late- erythroblasts were isolated and purified. After RNA isolation and linear amplification, gene expression analyses were performed using microarray technology. The generated data were analysed by two methods; the brb-ArrayTools platform and with the Bioconductor platform using bead level data.

Results
Morphological difference in maturation was not observed following 7 days in culture, while a pronounced delayed maturation was observed in the patient group after 14 days. For both analyses, following 7 days in culture there was no difference in gene expression between the control and patient groups. After 14 days in culture, 431 differentially expressed genes were identified by each method including 47 genes identified by both methods. Interrogating these gene lists with gene ontology tools a subset of 86 genes was selected whose results were confirmed by Quantitative-Real-Time-PCR.

Conclusion
The changes in gene activity and development associated with the phenotype of β-thalassaemia occur late in the maturation process of erythroid-lineage cells. We believe that these changes in gene expression are due to delayed erythropoiesis in erythroblasts of β-thalassaemic patients as a result of their reduced β-globin expression.
Title: ASSESSMENT OF LIVER FUNCTION TEST IN THALASSEMIC PATIENTS WITH IRON OVERLOAD

Abstract Category: Hepatological

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ABSTRACT:
Iron overload is thalassemia complication that has regular blood transfusion. Thalassemic patients were transfused more than 12 to 15 times always affect with iron overload. Though its necessary to examine serum ferritin in order to assess liver functions. The definite of iron overload is serum ferritin over than 1000 ng/mL. necessary to take iron chelating tablets for reducing iron accumulation. Iron accumulating damages many organs. In case serum ferritin over than 2500 ng/mL such more damage many organs such as liver, heart etc. The aim of this study for assessment association feritin level and liver functions, liver enzyme, aspartateaminase(AST), alanineaminase(ALT), alkalinephosphatase(ALP) were examined in thalassemia with iron overload. This study was retrospective study, studied thalassemia from adolescences ward in Khonkaen hospital between January to December 2014. Using unpaired t test, percentage, mean and standard derivation. In this study, finding thalassemia with iron overload 106 cases and 2 group serum ferritin levels. First group, serum ferritin 1000 to 2500 ng/mL(1156± 175.4) 85 cases, 80.2 %. The mean of liver enzyme level, aspatatetransaminase(AST), alaninetransaminase(ALT) and alkalinephosphatase(ALP) were 37.7 ±21.9 U/L, 28.8 ± 22.6 U/L and 154 ± 52.1 U/L respectively. Second group, serum ferritin over than 2500 ng/mL(3370± 1271) 21 cases, 19.8 %. The mean of liver enzyme level, aspatatetransminase (AST), alaninetransaminase (ALT) and alkalinephosphatase (ALP) were 67.9 ±48.9 U/L, 63.0 ±55.7 U/L and 159.5 ±58.6 U/L respectively. Serum ferritin 1000 to 2500 ng/mL showed signification positive correlation with alkalinephosphatase (p < 0.0001) and serum ferritin over than 2500 ng/ml showed significant positive correlation with aspatatetransaminase (AST), alaninetransaminase (ALT) and alkalinephosphatase(ALP) (p< 0.0001). In addition, amount of blood transfusion were various depends on age, weigh, the period of diagnosis and Iron chelating tablets. In consequence liver functions examination is necessary for all thalassemic patients with iron overload.

Keywords: Iron overload, serum ferritin, Aspatatetransaminase (AST), Alaninetransaminase (ALT), Alkalinephosphatase (ALP)
Title: CAN HYDROXYUREA ALSO WORK AS AN IRON CHELATOR IN β-THALASSAEMIA PATIENTS?

Abstract Category: Iron Overload and Management

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ABSTRACT:

Background
Hydroxyurea is now increasingly used in the management of different haemoglobinopathies. This has added to the cost of management of the disease. However, if hydroxyurea provides an additional benefit of iron chelation, it will be cost effective in the long run and could be helpful in optimum removal of iron from the body.

Aim
Based on our earlier study on iron overloaded mice, who on treatment with hydroxyurea showed a reduction in serum ferritin levels and tissue iron, we hypothesise this phenomenon in regularly transfused β-thalassaemia patients.

Methods
17 β--thalassaemia intermedia patients and 4 HbE-β-thalassaemia patients (requiring transfusion), were enrolled for hydroxyurea therapy (10-20 mg/kg/day) for 12 months. All patients, (age 4-32 years) had presented after 2-3 years of age. The patients were evaluated clinically, haematologically and molecularly before starting therapy. Blood was collected for complete blood count, haemoglobin electrophoresis by HPLC, serum ferritin by ELISA, cellular labile iron pool (LIP) using calcein acetoxymethyl ester (CAAM) and flow cytometry, externalization of phosphotidylserine using annexin V by flow cytometry, the reactive oxygen species (ROS) using 2’-7’-dichlorofluorescin diacetate (DCF), reduced glutathione levels (GSH) using 1-(4-chloromercuryphenylazo-2-naphthol) by flow cytometry.

Results
12 of the β-thalassaemia patients and 2 of the HbE-β-thalassaemia patients responded to the therapy with haemoglobin maintained above 7.5g/dl without transfusions. An increase in mean Hb, MCV and HbF and a mean decrease in serum ferritin was observed among the responders. Also a statistically significant decrease in LIP, externalization of phosphotidylserine and ROS along with a statistically significant increase in GSH was also seen after hydroxyurea therapy among the responders.

Conclusion: Hydroxyurea apart from its effect on increasing HbF synthesis in haemoglobinopathies like sickle cell anaemia, β-thalassaemia and HbE-β-thalassaemia, has an additional role in iron chelation. However, a larger study is required.
Title: EFFICACY AND SAFETY OF DEFERASIROX IN SINGLE VS DIVIDED DOSAGE IN THALASSEMIC CHILDREN

Abstract Category: Iron Overload and Management

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ABSTRACT:

Background
Dose-dependent effect of DFX had been observed in many studies. Ferritin levels tended to be high even on DFX (30 - 40 mg/kg/day), so safety and efficacy of higher doses (40 - 50 mg/kg/day) in once daily vs two daily doses needed to be compared.

Aim
To study the efficacy and safety of DFX (40-50 mg/kg/day) in single vs divided doses in thalassemic children.

Methods
The prospective study was conducted in thalassemia unit of Dayanand Medical College and Hospital over 18 months. Patients were randomized to two groups. Both groups received DFX in doses of 40 - 50 mg/kg/day. Group A OD dose and group B in BD dose. Efficacy parameters studied were ferritin levels, cardiac & liver MRI T2 *. Patients were monitored for gastrointestinal side effects, rash and change in serum creatinine values, SGPT and GFR.

Results
Serum ferritin levels reduced in both the groups viz. Group A (2692.5 ± 1232.35 ng/ml to 1959.5 ± 696.19 ng/ml p value = 0.07), Group B (2766.2 ± 897.51 to 2569.9 ± 762.6 ng/ml ;p value = 0.38) with significant difference between the two groups at the end of study (p=0.05). Liver MRI T2* values improved in group A (2.1 ± 1.50 to 3.1 ± 1.84 ms ; p= 0.096) as compared to group B (2.8 ± 1.84 to 2.2 ± 2.71 ms; p=0.70). Overall mean cardiac MRI T2 * values decreased (30.5 ± 10.30 ms to 24.5 ± 4.94 ms ; p=0.202 ) in group A and ( 31.7 ± 14.78 ms to 20.5 ± 11.78 ms ; p=0.167) in group B. No significant adverse effects were noted with 40 -50 mg/kg/day dose of DFX apart from mild increase in liver transaminases (SGPT) levels.

Conclusion
DFX is safe at 40 - 50 mg/kg/day. Once daily dosage is more efficacious than twice daily dosage schedule.
Title: EVALUATION OF IRON OVER LOAD IN THALASSEMIA PATIENTS

Abstract Category: Iron Overload and Management

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ABSTRACT:
Iron overload is a leading cause of morbidity and mortality in patients with thalassemia and related complications include liver cirrhosis and cardiac disease. It is therefore essential to evaluate iron overload condition in thalassemia patients. Objectives: To determine iron overload in a thalassaemia patients. Methods: A cross sectional study of 299 transfused thalassaemia patients was conducted. The variables of interest included age, gender, Hb typing, globin mutations, serum ferritin, LIC, T2*. Results: Of 299 thalassemia patients, 46 (15.4%) were alpha thalassemia (HbH), 57 (19,1% ) were beta thalassemia and 196 (65,5%) were beta-thalassemia/HbE. Regarding severity, 31 (10,4%) were thalassemia minor, 232 (77,6%) were thalassemia intermediate (TI) and 36 (12,0%) were thalassemia major (TM). There were 295 (98,7%) thalassemia patients with serum ferritin over 600ng/ml, among them 58.9% over 2500ng/ml. The average serum ferritin in the thalassemia minor group was 1236 ± 568 ng/ml, 3280 ± 1171 ng/ml in TI and 4336 ± 1938 in TM, the difference in the mean ferritin among the three groups was statistically significant (p < 0.001). There were 256 (85,6%) patients with LIC ≥ 3mg /g, among them 222 (74,2%) patients with LIC ≥ 15mg /g, the average LIC was 9.5 ± 5.9 mg/g in thalassemia minor, 20.2 ± 6.8 mg/g in TI and 24.1 ± 5.0 in TM group, with a significant difference among the 3 groups (p <0.001). Serum ferritin showed a significant positive correlation with LIC (R = 0.514; p <0.001), and a ferritin increase of 125ng/ml was associated with a LIC increase of 1mg/g.

There were only 44 patients (15.1%) with T2* < 20ms, among them 15/36 (41.7%) were TM and 29/232 (12.5%) were TI, without any case in the thalassemia minor group. Serum ferritin had a significant negative correlation with T2* in TI (R = - 0.345; p <0.001), no correlation was found between T2* and ferritin in TM (R = - 0.057, p=0.741), no correlation between ferritin and T2* in group of T2*< 20ms. In this study, almost of thalassemia patients developed iron overload and consequences of iron over load which is alarming to healthcare professionals who take care of thalassemia patients in Vietnam.
Title: CARDIAC AND LIVER T2* IMPROVEMENTS IN BETA THALASSEMA MAJOR TWIN PATIENTS WITH SEVERE IRON-OVERLOADED AND HEPATITIS C TREATED WITH DEFEROXAMINE AND DEFERIPRONE: A CASE REPORT

Abstract Category: Iron Overload and Management

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ABSTRACT:

Background
The combination of Deferiprone/Deferoxamine (DFP/DFO) as iron chelator agents for thalassemia patients has been reported since many years, particularly in patients who do not respond adequately to a single chelating agent.

Objective
The aim of this case report is to investigate the efficacy, tolerability and safety of combined DFP/DFO in Thalassemia patients with severe iron overload.

Case presentation
These twins were diagnosed beta thalassemia major 20 years ago and have been receiving blood transfusion regularly since then. Eight years ago, they were also got several complications such as Hepatitis C, diabetes mellitus, and one of them also suffered of liver abscess. That time only DFO available. A year ago based on clinical presentation and MRI T2* examination, they were diagnosed with cardiomyopathy, which was most likely caused by lack of efficacy in removing liver/cardiac iron with monotherapy. We started the combination therapy with DFO (30 mg/kg/day) 2-3 x/week and DFP (100 mg/kg/day) every day. After 1 year, the cardiac MRI of twin 1 is now improved from 9.43 ms to 10.83 ms (15%); and the liver MRI increases from 1.18 ms to 1.70 ms (40%). The cardiac MRI of twin 2 is also improved from 13.45 ms to 15.89 ms (18%); and the liver MRI increases from 1.57 ms to 2.19 ms (39%). The ferritin level for both twins 1 and 2 declined from 5,992 mg/dL to 2,310 mg/dL and 6,039 mg/dL to 2,639 mg/dL, respectively.

Conclusion
Combined DFX/DFO can be considered as an alternative option when monotherapy fails to remove iron overload in cardiac and liver tissues.

Keywords
chelation therapy; deferoxamine; deferiprone; cardiac liver iron overload; thalassemia; hepatitis.
**Title:** DEFERASIROX IN THAI CHILDREN WITH THALASSEMIA; 4.5 YEARS EXPERIENCE

**Abstract Category:** Iron Overload and Management

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**ABSTRACT:**

**Objective:**
To evaluate the efficacy and safety of the oral iron chelator deferasirox in retrospective cohort of Thai children with thalassemia with high iron load.

**Materials and Methods:**
Thalassemia children 24 case from 120 case (1-18 yrs) at thalassemia clinic were enrolled and followed up for a period 54 months between July 2010 and December 2014. The dose of deferasirox was determined by their baseline serum ferritin and was adjusted to a maximum of 40 mg/kg/day depending of response, Ferritin, SGOT, SOPT, Alk, Cr, BUN, Urine examination and adverse effect were regularly monitored.

**Results:**
They were 2 groups analysis devised by first group age <5 yrs, 20 case with TDT 9 cases, NTDT 11 cases and second group age ≥ 5 yrs 4 cases who had fail first line drug due to complication or poor chelation, TDT 3 cases and NTDT 1 case.

In first group 9 cases of TDT found that a significant decline in serum ferritin of mean dose deferasirox 24.14 mg/kg/day. The mean serum ferritin at 12, 24, 36, 48 months was 1661, 1862.79, 1644.55, 1472.2 and significant decline SGOT, SGPT, Alk from mean serum SGOT 40.53 to 31.5, SGPT 32.83 to 13.5 and Alk 176.99 to 159.5. In 11 case with NTDT < 5 yrs, we found that increase mean serum ferritin from 1432.81 to 1910.28 at mean dose deferasirox 22.75 mg/kg/day but significant decline SGOT, SGPT, Alk from mean serum SGOT 56.73 to 33.67, SGPT 48.7 to 15.33 and Alk 188.77 to 115.67. In this study one case Down’s syndrome with βthal/HbE was death due to fungus pneumonia and respiratory failure.

In second group with age ≥ 5 yrs with TDT 3 cases and NTDT 1 case that showed, only one NTDT was good response decline mean serum ferritin from 1893 to 1607.9 and serum SGPT 59 to 42, SGOT 74 to 48, Alk 190 to 118 all of liver enzyme were decline by mean dose deferasirox at 14.25 mg/kg/day duration 36 months. All of TDT poor response to chelation two cases ferritin raising (mean ferritin 1601.3 to 2879, 3968 to 5299.2) due to poor habit, iron food loading from eating. The last TDT who had longest duration up to 54 months because he had A/E SNHL from deferiprone, mean ferritin at 12, 24, 36,48 months was 3136.2, 3675, 5702.3, 4359 and poor response of SOPT 101 to 172, SGOT from 111 to 125 Alk from 273 to 227 at mean dose 25 mg/kg/day of Deferaxirox. In study showed that only on had N/V but can continuous therapy.

**Conclusions:**
Deferasirox was safe for thalassemia children with iron overload only one had N/V and still continuous and least age to given was 20 months (3 cases). In TDT may need higher dose of deferasirox than NTDT. In adolescence patient were need multiple factor to success of chelation and higher dose of deferasirox must to be considered.
Title: THE HOMOZYGOUS HEMOGLOBIN EE GENOTYPE IS ASSOCIATED WITH INCREASED SERUM FERRITIN AND SOLUBLE TRANSFERRIN RECEPTOR CONCENTRATIONS AMONG CAMBODIAN WOMEN: IMPLICATIONS FOR THE DIAGNOSIS OF IRON DEFICIENCY

Abstract Category: Iron Overload and Management

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ABSTRACT:

Background
Genetic hemoglobin (Hb) disorders are common in Southeast Asia and are associated with a low Hb concentration. Our aim was to determine if Hb disorders were also associated with ferritin and soluble transferrin receptor (sTfR) concentrations (biomarkers of iron deficiency [ID]).

Methods
We recruited 450 women (18-45 y) from Prey Veng province, Cambodia. Venous blood was collected in July 2012 and assessed for Hb, serum ferritin, sTfR, retinol binding protein, folate, vitamin B12, C-reactive protein and α-1 acid glycoprotein. Hb typing was determined by capillary electrophoresis and genotyping by PCR analysis. Geometric mean ratios calculated using linear regression were used to determine the proportional change in the outcome variables (95% CI).

Results
Overall, we found contradictory evidence of ID prevalence among women using ferritin (<15 µg/L [n=9; ~2%]) and sTfR (>8.3 mg/L [n=79; ~18%]) biomarkers. Ferritin and sTfR were significantly elevated among women with the homozygous Hb EE genotype (n=31; 7%). The Hb EE genotype was associated with a 50% (14, 96%) and 51% (37, 66%) increase in the geometric mean ferritin (µg/L) and sTfR concentration (mg/L). Using sTfR for ID diagnosis, prevalence was higher among women with the Hb EE genotype (n=17; 55%) as compared to the normal Hb AA genotype (n=20; 10%); however, no differences were found among groups using ferritin.

Conclusions
No differences in ID prevalence were found among groups using ferritin; however, women had surprisingly high mean ferritin concentrations (~95 µg/L). Further research is needed to investigate if these factors would have a more substantive impact on ID prevalence in a population with ferritin concentrations closer to the cut-off for ID diagnosis. We conclude that sTfR may not be an accurate biomarker of ID among individuals with the Hb EE genotype. An iron supplementation trial is warranted to determine if women are truly iron deficient.
Title: EVALUATION OF EFFICACY OF ORAL DEFERASIROX BY TWICE-DAILY DOSING IN PATIENTS WITH TRANSFUSION-DEPENDENT BETA THALASSEMIA

Abstract Category: Iron Overload and Management

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ABSTRACT:

Background
Beta thalassemia major patients need consistent blood transfusion from the early years of their life. Deferasirox is an oral chelating agent used routinely as once daily dose to excrete excess iron. This study aimed to compare the efficacy and safety of deferasirox two times daily (BD) with the usual once daily dosing.

Method
Patients of an, who were receiving only deferasirox as their chelating agent were enrolled into the study. All have received deferasirox at least 6 month as once-daily dosing. The last ferritin before entering the study and the mean deferasirox daily dose during the previous six months was considered as baseline ferritin and deferasirox dose, respectively. Ferritin was checked at 1, 2, 3, 4, 5, and 6 months for patients who returned regularly to the hospital. Laboratory tests including CBC-diff, Creatinine (Cr), Aspartate Aminotransferase (AST) and Alanine Aminotransferase (ALT) were measured to evaluate the safety of deferasirox BD dosing.

Results
A total of 21 transfusion-dependent patients were included. The men ferritin level was significantly decreased from 1815 ng/ml to 1473 ng/ml (P= 0.02). There were not any significant changes in AST, ALT and Cr levels compared to baseline values.

Conclusion
Twice daily dosing of deferasirox was associated with more decrease in ferritin level compared to baseline single daily dose values without any hepatic or renal adverse effects.

Key Words: Beta Thalassemia major, Deferasirox, Ferritin, Efficacy
Title: ANALYSIS OF THE MECHANISMS UNDERLYING THE DEVELOPMENTAL REGULATION OF EMBRYONIC AND FETAL B-LIKE GLOBIN GENES

Abstract Category: Molecular Control

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ABSTRACT:
The clinical symptoms of hemoglobin disorders such as β-thalassaemia and sickle cell anemia are significantly ameliorated by the persistent expression of γ-globin after birth. This knowledge has driven the discovery of important regulators that silence γ-globin perinatally. Improved understanding of the γ- to β-globin switching mechanism holds the key to devising targeted therapies for β-haemoglobinopathies. To further investigate this clinically important developmental switch a novel fluorescent-based cellular reporter assay system was developed. Two fluorescent reporter genes, DsRed and eGFP, were inserted into an intact 183 kb intact human β-globin locus, replacing the coding regions of the γ-globin and β-globin genes, respectively and was stably transfected into adult murine erythroleukemic (MEL). Following RNA interference (RNAi)-mediated knockdown of two key transcriptional regulators, Myb and BCL11A, we observed a derepression of γ-globin, measured by DsRed fluorescence and RT-qPCR. Interestingly, double knockdown of Myb and DNA methyltransferase 1 (DNMT1) resulted in a robust induction of ε-globin, (up to 20% of total β-like globin species) compared to single-knockdowns. Conversely, double-knockdowns of BCL11A and DNMT1 enhanced γ-globin expression (up to 90% of total β-like globin species) compared to single-knockdowns. Moreover, following RNAi treatment, expression of human β-like globin genes mirrored the expression levels of their endogenous murine counterparts. These results demonstrate that Myb and BCL11A cooperate with DNMT1 to achieve developmental repression of embryonic and fetal β-like globin genes in the adult erythroid environment.
Title: ANALYSIS OF B-THALASSEMIA MICE TO UNDERSTAND INNATE IMMUNE ABNORMALITIES IN B-THALASSEMIA PATIENTS

Abstract Category: Molecular Control

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ABSTRACT:

β-Thalassemia is associated with several abnormalities in the immune system, including defective neutrophil functions, which predisposes patients to infections with both Gram-positive and Gram-negative bacterial pathogens. The molecular mechanisms involved in impaired neutrophil function during bacterial infection in thalassemia patients are not completely understood. To better understand the impact of thalassemia on the innate immune system, we investigated susceptibility to bacterial infection and neutrophil function in the Hbbth3/+ β-thalassemia mouse model. We demonstrate that Hbbth3/+ β-thalassemia mice are highly susceptible to infection with Streptococcus pneumoniae, a Gram-positive extracellular pathogen that causes sepsis, pneumonia and meningitis. Our results showed that blood and splenic neutrophils from Hbbth3/+ β-thalassemia mice show defective chemotaxis, reduced opsonophagocytosis and decreased reactive oxygen species (ROS) production compared with neutrophils from normal mice. In addition, we used gene expression studies and quantitative RT-PCR to demonstrate that genes that regulate neutrophil chemotaxis, CXCR2 and CD11b expression, opsonophagocytosis, and reactive oxygen species (ROS) production (p22phox, p67phox and p91phox) are significantly repressed during systemic infection with S. pneumoniae. The outcome of this study provides direct molecular evidence that changes in gene expression in Hbbth3/+ β-thalassemia mice contribute to the deficiencies in antimicrobial neutrophil functions observed in β-thalassemia.
**Title:** A STUDY ON EFFECTIVENESS OF HYDROXYL-UREA IN A THALASSAEMIA TREATMENT CENTRE IN EASTERN-INDIA

**Abstract Category:** Non-Transfusion Dependent Thalassaemia Corresponding / Presenting Author: Dr. Taraknath Mukhopadhyay

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**ABSTRACT:**

**Purpose**
There are lots of Thalassaemia patient in eastern-India. One of the very interesting facts about Thalassaemia in this region is that, there are different types of Thalassaemia and different types of haemoglobinopathy in this region. Also a large no. of E-beta Thalassaemia patients can be seen here. Due to diversity of mutation in Thalassaemia patient, blood requirements of the patients also vary and many clinically Thal-intermedia patient can be seen. The drug hydroxyl-urea has a very good role in these patients to increase their transfusion interval.

**Methods**
Patients’ status was recorded from patient file which is kept in the centre. From patient records the no. of the patient taking the hydroxyl-urea at present was recorded. Patients details were obtained from file and counseling of patient and guardian.

**Results**
Study was based on 35 patients of the treatment centre (Total no. of the pt. 162), who are treated successfully on hydroxyl-urea i.e. transfusion interval increased. Doses of hydroxyl-urea are between 15-20 mg/kg/day. Duration of treatment:
- 2yrs: 4pts
- 2-5 yrs: 11pts
- 5-10yrs: 20 pts.

Interval of transfusion after successful transfusion is:
- less than 1month: 4pts
- 1-2 months: 5 pts
- 2 to 6 months: 6 pts.
- 6 months to 1 yr: 11pts.
- Generally without transfusion: 9pts.

Fetal hemoglobin at the time of diagnosis varies widely from 35%-54%. Also HbE level at the time of diagnosis of E-beta patient varies from 12.5% to 62.1%. It is also strange that in 5 pt out of 35 the interval of transfusion decreases compared to early phage. In 3 pts there are wide range of fluctuation of blood requirement.

**Conclusion**
HU-is very effective drug in clinically Thal-intermedia patients. It can be effective at dose of 15—20 mg/kg/day. The drug is more or less safe within this dose of 15—20mg/kg/day. Effectiveness of hydroxyl-urea has no relationship with the HbF & Hb E level of the patient at the time of diagnosis.
Title: ARTERIAL THROMBOSIS IN FOUR MYANMAR NON-TRANSFUSION DEPENDENT HbE-BETA THALASSAEMIA INTERMEDIA PATIENTS

Abstract Category: Non-Transfusion Dependent Thalassaemia

Authors: Sein Win, Hein Htet Oo, Kyaw Thet Aye, Kyaw Zay-ya, Moe Hein, Aye Aye Gyi, Htun Lwin Nyein, Rai Mra

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ABSTRACT:

Background
Thalassaemia intermedia (TI) patients are extremely diverse in phenotypic expression and there is a wide variation in clinical complications. Thromboembolic events (TEE) are among the clinical complications of TI that were found to occur at a higher rate than in patients with thalassaemia major (TM).

Method
Retrospective analysis of case records of the department of Clinical Haematology, Yangon General Hospital (YGH) form January 2008 to December 2014 were done to find out TI patients with arterial thrombosis.

Results
Among 203 HbE-Beta TI patients admitted to the department of Clinical Haematology, YGH during study period, four had arterial thrombosis (three males and one female) (1.97%). Age at presentation ranged from 22 to 39 years. Below knee amputation was observed in two patients and above knee amputation in another two patients. Haemoglobin phenotypes were AEF in 3 patients and EF in another one. HbF ranged from 8 to 21.7%. Only one patient had past history of one unit packed cell transfusion before TTE and other three patients had no history of transfusion before TTE. All received perioperative blood transfusion. Baseline Hb ranged from 3.1 to 8.4g/dl, retic count from 0.0305 to 0.0661x 10^6/µL, total white cell count from 7.37 to 24.6 x 10^3/µL and platelet from 75 to 542 x 10^3/µL. All were non-diabetics and non-smokers. Ferritin level ranged from 1146 to 2000 ng/ml. All had raised nucleated red cell on blood film and normal coagulation parameters. All were negative for Hepatitis B, C and HIV serology. Thrombophilic screen was not done.

Conclusion
Arterial thrombosis is a rare but disabling complication in Myanmar non-transfusion dependent HbE-beta TI patients.
Title: ROLE OF GENETIC MODIFIERS IN CLINICAL OUTCOME OF HOMOZYGOUS - THALASSEMA PATIENTS FROM INDIA

Abstract Category: Non-Transfusion Dependent Thalassaemias

Authors: Priya Hariharan, Pratibha Sawant , Manju Gorivale , Roshan Colah, Kanjaksha Ghosh, and Anita Nadkarni

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ABSTRACT:

Background
The clinical manifestations of β-thalassemia are extremely variable ranging from a severe disorder (thalassemia major) to a mild presentation (thalassemia intermedia). This heterogeneity may be due to the nature of the beta-globin gene mutations, associated α-thalassemia or inheritance of genetic factors leading to raised HbF levels. We investigated the effect of genetic factors linked to raised HbF XmnI polymorphism, BCL11A SNP rs11886868 and rs 7557939, sequence alterations in the KLF1 gene and α-gene deletions on the phenotypic severity of patients with β-thalassemia.

Methods
We studied 20 thalassemia intermedia patients and 20 severe thalassemia major (TM) patients. Haematological analysis was done on a Sysmex K-1000 analyser. The HbA2 and HbF levels were measured by HPLC (Variant II-Biorad). β-genotyping was done by reverse dot blot hybridization, ARMS or DNA sequencing. Genotyping was also done for the XmnI polymorphism, β-gene deletions and 2 SNPs within the BCL11A gene. KLF1 gene variations were detected by DNA sequencing.

Results
Nine different β-thalassemia mutations were detected and 20% of TI patients showed presence of milder mutations. 2 TI patients showed presence of KLF1 variations (-187 G→A, CD 211 A→G) which were absent in TM patients. The mutant allele A of SNP rs7557939 in the BCL11A locus was significantly higher in TI patients [f (A)=0.57], whereas the variant allele C of SNP rs 11886868 did not show any correlation. The presence of the XmnI polymorphism and alpha gene deletion were more frequent in the TI group.

Conclusion
This pilot study gives a potential insight of the impact of genetic modifiers on the clinical severity of the disease. However, larger number of patients need to be studied. This may help the clinicians for better management of the disease.
ABSTRACT:

Background
Thrombosis is a major complication in β-thalassemia/HbE especially in splenectomized patients. The mechanism of hypercoagulation in thalassemia is not fully understood. Microparticles (MPs), small membrane vesiculated from various cell types, have been shown to have strong procoagulant activity in many diseases because MPs harbor phosphatidylserine and other procoagulant proteins. The increased amount of MPs was previously observed in splenectomized thalassemia patients when compared to normal subjects. However, the effect of thalassemia MPs on platelet function in β-thalassemia/HbE has not been elucidated.

Methods
MPs were isolated from 15 blood samples each from normal subjects, non-splenectomized and splenectomized β-thalassemia/HbE patients. The ability of MPs to induced platelet aggregation was examined by coincubation the isolated MPs with normal platelet-rich plasma and determination by aggregometer. Platelets activation by MPs was performed by adding MPs into whole blood of the 3 subject groups and assessing the CD41a and CD62P expression using flow cytometer.

Results
Both normal and patient MPs induced completed platelet aggregation at physiological platelet:MP ratio of 1:10. Flow cytometry analysis of activated platelets (CD41a+/CD62P+) showed a dose dependent activation of both normal and thalassemia platelets by all groups MPs. Interestingly, thalassemia MPs had a higher platelet activation potential when compared to normal control. The MPs obtained from splenectomized patients induced platelet activation in all groups at the platelet:MP ratio as low as 1:0.01. In contrast, higher number of normal MPs (platelet:MP ratio 1:10) was required to induce platelet activation. Moreover, platelets from splenectomized patients were prone to be activated by MPs when compared to normal platelets. Co-incubation of splenectomized patient MPs at the platelet:MP ratio of 1:10 resulted in the increased platelet activation, at 3.7±1.7%, 7.5±3.6% and 9.2±3.4% with normal, non-splenectomized and splenectomized platelets, respectively (P<0.05).

Conclusion
Increases of the number and platelet activation potential of thalassemia MPs play role in the risk of thrombosis in β-thalassemia/HbE.
Title: CORRELATION OF GENOTYPE WITH PHENOTYPE IN BETA THALASSAEMIA INTERMEDIA IN SRI LANKA

Abstract Category: Non-Transfusion Dependent Thalassaemia

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ABSTRACT:

Background & Aims
Previous studies have shown that a third of all patients attending thalassaemia centers in Sri Lanka have non transfusion dependant thalassaemia (NTDT); the majority of whom have Hb E β thalassaemia. Previously we attempted to study the genetic basis of non E β thalassaemia intermedia (TI) patients in Sri Lanka. In the present study we aim to correlate their genotype with the phenotype.

Methods
Fifty unrelated TI patients identified from the five main thalassaemia centers were assessed clinically and categorized into “mild”, “moderate” and “severe” groups according to their clinical severity. DNA analyses were performed by the standard techniques.

Results
Seventeen patients were homozygous or compound heterozygous for β mutations. Five of the homozygotes who carried two mild β alleles including a rare promoter mutation - 90 C>T, Hb variant alleles of Hb G-Szuhu and Hb G-Coushatta; invariably had mild disease. Nine inherited two severe β alleles with either one or two α gene deletions; despite the α deletions they had severe disease. Out of three individuals who carried two β alleles with normal α gene, one had Xmn I +/+ and had mild disease. We were unable to explain the phenotype in two individuals in this group with the existing genetic data.

Thirty three patients were heterozygous for a β mutation IVSI-5 G>C (n=12) was the commonest. Twenty eight of the heterozygotes carried excess α genes and had a mild to moderate phenotype. In this group of individuals, inherited with single β allele with normal α genes, the genetic studies could not explain the phenotype in five individuals.

Conclusion
The clinical outcomes of our TI population were mostly explained by the genotypes linked to the α and β gene cluster. However, in a minority, the existence of other causative genetic determinants remains to be molecularly defined.
Title: REPORT ON PATIENTS WITH NON TRANSFUSION-DEPENDENT B-THALASSEMIA MAJOR BEING TREATED WITH HYDROXYUREA ATTENDING THE THALASSEMIA RESEARCH CENTER, SARI, MAZANDARAN PROVINCE, ISLAMIC REPUBLIC OF IRAN IN 2013

Abstract Category: Non-Transfusion Dependent Thalassaemia

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ABSTRACT:

Hydroxyurea (HU) has been used to treat patients with non transfusion-dependent b-thalassemia major (b-TM) at the Thalassemia Research Center, Sari, Mazandaran Province, Islamic Republic of Iran since 1996. This study was performed to summarize and to share our experience. Medical records of all patients with b-thalassemia (b-thal) attending our center were reviewed in January 2013. Definition of b-TM was based on complete blood count (CBC), hemoglobin (Hb) electrophoresis, and for some patients, by the amplification refractory mutation system-restriction fragment length polymorphism (ARMS-RFLP) method. Patients who had not been transfused before, or had only occasionally had blood transfusions, were selected. Age at first blood transfusion, initial HU therapy and time of study was extracted from the records. The lowest Hb level before using HU and the last Hb value when on the HU regimen as well as the difference, were reported. Number of saved packed red cells was calculated according to duration of HU use and the usual needs of the patients. Hydroxyurea was discontinued before a planned pregnancy and during gestation and lactation periods. A p value of 0.05 was considered statistically significant. It was consistent with 1856 patients/year, and 3542 units of blood were saved. We found HU to be effective and safe in treating patients with non transfusion-dependent b-TM. We strongly recommend HU therapy.

Keywords
Non Transfusion-Dependent b-Thalassemia, Hydroxyurea
Title: THE EFFECT AND SIDE EFFECT OF HYDROXYUREA THERAPY ON PATIENTS WITH B-THALASSEMIA: A SYSTEMATIC REVIEW TO DECEMBER 2012

Abstract Category: Non-Transfusion Dependent Thalassaemia

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ABSTRACT:
Hydroxyurea (HU) is being used for patients with transfusion-dependent b-thalassemia major (b-TM) as well as non transfusion-dependent b-TM. As controversy exists regarding efficacy and safety of HU, we searched the published literature on efficacy, effectiveness and toxicity of HU in patients with b-TM. The research sources we used were: Medline, SID, PubMed, Scopus, Request, Web of Knowledge, Springer, Ovid, Cochrane searched up to October 2012. Using search terms sensitive to studies of clinical trials combined with searches on terms related to thalassemia and HU. We selected studies on randomized trials, quasi experimental trials before and after design), case reports (with 1 -5 cases), side effect studies in patients with b-TM, studies related to the mechanism of action and toxicity when used in patients with other hemoglobinopathies. We researched studies in English and Persian. Eligible articles were reviewed by two independent reviewers. Patient’s characteristics, duration of trial, outcome and side effects were extracted. The main outcomes were synthesized under a random-effects model. Heterogeneity was assessed using the Q statistic, Tau2 and I2. Subgroup analyses were performed and the statistics data (STATA) software used. More than 500 articles were reviewed. No randomized clinical trial was found. Seventeen trials with before and after designs were found, 16 case reports (1 -5 cases), 19 articles for mechanism of action and 16 studies for side effects were published from 1969 to October 2012. Hemoglobin levels after treatment showed modest but significant increase in non transfusion-dependent b-TM (p<0.0001) and in transfusion-dependent b-TM (p<0.0001).

Keywords:
Antimetabolite, b-thalassemia (b-thal), genotype, hemoglobin (Hb), Hb F inducer, hydroxyurea (HU), side effect
Title: PERCEPTIONS OF BETA THALASSEMIA MAJOR PATIENTS AND THEIR PARENTS ABOUT MEDICAL STUDENTS' HISTORY TAKING BEHAVIOUR

Abstract Category: Psychosocial Support

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ABSTRACT:

Background
Thalassemia presents with a wide range of physical signs involving entire systems of the body that demands sharp clinical skills. Their lives are full of sensitive stories related to physical, psychosocial, and economic implications of the disease. As such revealing the entire story with their ideas concern and expectations create a formidable challenge and learning opportunity for medical students. Therefore medical students and doctors approach thalassemia patients frequently with possibility of causing stress and burden. On the other hand if they are involved as patient teachers, they will be empowered and feel valued. This study focuses on perceptions of thalassemia patients and their parents about medical students’ communication skills.

Methods
The study was done at university paediatric Thalassemia centre at TH Peradeniya, Sri Lanka. Patient’s perceptions were first evaluated using a pretested questionnaire. Thirty-nine students-patients/parent encounters were independently evaluated by the respective patient/parent and a senior medical doctor by direct observation using a pre-validated check list.

Results
Parents were satisfied regarding talking with medical students in general (86%), physical examination (74%), discussing psychological (85%) and economy related (79%) problems, getting advice (89%), and learning from patients (89%). Independent evaluation by both parents and senior doctors revealed following observations respectively: self-introduction (95%, 93%), friendliness (88%, 92%), enough time to express concerns (97%, 92%), clarity of questions (95%, 95.3%), clear explanations (97%, 91.8%), addressing key issues - blood transfusions (86%, 94%), iron chelation (93%, 100%) and carrier screening (93%, 94%), addressing patients’ ideas and concerns (85.6%, 87%) and family problems (86%), and general satisfaction (92%, 90.1%). 5% of parents and 7% of senior doctors were not satisfied with overall performance of the medical student.

Conclusions
Most components in check list were rated highly by both parents and doctors and the difference was not significant; however, skills of discussing psychological and economic issues, addressing patients’ ideas and concerns, and physical examination skills had more potential for improvement. Non-satisfaction though of a minority highlights importance of teaching communication skills to medical students to enable them to communicate better with thalassemia patients.
Title: EFFECTIVENESS OF A PATIENT-PARENT EMPOWERMENT PROGRAM AMONG BETA THALASSEMIA PATIENTS IN IMPROVING DOCTOR-PATIENT COMMUNICATION

Abstract Category: Psychosocial Support

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ABSTRACT:

Background
Chronic nature of thalassemia causes changes in different aspects of life in patients and parents, including their quality of life. However, patient empowerment models have been shown to assist those with similar chronic illnesses by various study groups. Current study evaluates the effectiveness of a patient-parent communication skills training program in improving doctor-patient communication.

Methods
The study was done at university paediatric Thalassemia centre at TH Peradeniya, Sri Lanka. Twenty six children with beta thalassemia major along with parents were enrolled to the communication skills training program developed from the Calgary-Cambridge Communication Model and conducted over one day. Twenty six final year medical students who had interviewed and examined at least three patients with beta thalassemia within preceding 2 weeks were evaluated both quantitatively and qualitatively on their experience by the researchers themselves at pre-intervention stage. All students were allowed to engage with same patients over next two weeks following the intervention. Quantitative and qualitative evaluation was repeated at end of 2 weeks. Wilcoxon signed rank test was performed to identify statistically significant variables.

Results
Percentages of scores achieved from final year students regarding their commitment for selected components at pre- and post-intervention and respective significance levels were: ability to engage with thalassemia patients (69.2%, 97.6%, p<0.001), talking with patients (60%, 93.2%, p<0.001), discussing psychological issues (67.1%, 95.5%, p<0.001) and socioeconomic issues (63.1%, 95.5%, p<0.001), physical examination (75.2%, 75.8%, p>0.05), giving information (80.4%, 82%, p>0.05), giving advice (73%, 73.2%, p>0.05), learning from patients (83%, 97.6%, p<0.05), and getting feedback from patients (70.6%, 88.8%, p<0.05).

Conclusions
Empowerment of thalassemia patients/parents by training communication skills based on Calgary Cambridge Model significantly improved medical students’ commitment in engaging and talking with thalassemia patients, and discussing psychological and socioeconomic issues. It was also associated with increased commitment with learning and getting feedback from patients.
**Title:** FACTORS AFFECTING PHYSICAL, EMOTIONAL AND SOCIAL HEALTH RELATED QUALITY OF LIFE OF PATIENTS WHO ARE MORE THAN 12 YEARS AND DIAGNOSED WITH BETA THALASSEMIA MAJOR IN SRI LANKA

**Abstract Category:** Quality care for Quality of Life

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**ABSTRACT:**

**Background**  
Little data exists regarding the Health Related Quality of Life (HRQoL) of patients with beta thalassemia major and the current study aims to assess the factors determining physical, social, and emotional health of these children who are more than 12 years.

**Methods**  
Forty patients over 12 years and who were treated at Thalassemia centre of TH Peradeniya were enrolled to the study. All patients were interviewed regarding factors affecting physical, emotional and social health related quality of life by trained medical graduates over a period of three months using a pretested, semi-structured questionnaire on a likert scale ranged from 1 (least severe) to 10 (most severe). In addition, SF-36 and SF-10 HRQoL instruments were used for children who were aged >18 and <18 years respectively.

**Results**  
Forty patients were available for analysis and mean age was 17 years (range 12- 24 years). 50% were female patients. Means of each of the items tested on likert scale (1-10) among <18 and >18 years patients were: pain during work (2.5, 3.7), disturbed sleep (1.8, 2), restriction of sports (2.3, 5.2), leisure activities (2.4, 2.8), and diet (2.8, 2.8), effect of healthy peer (2.1, 1.7) and family relationships (1.7, 1.4) and academic activities (4, 5.4). Effect of family included: neglect of other children (2, 1.9), economic problems (6.1, 5.4), need for social support (5.8, 6.3), loss of employment (5.2, 4.7), and transport problems (7.1, 4.38). SF-36 assessment showed only 9% (2/21) were extremely happy that they had good health and only 7 parents were extremely happy 7/19 (36%) in SF-10 assessment of their children’s health.

**Conclusions**  
Factors underpinning the quality of life of thalassemia patients are diverse and mainly related to restriction of daily activities, school dropout and family breakdown. The study observed the effect of most factors increase with increased age though relationship with others and family breakdown was less severe in older children possibly due to increased adaptation to disease.
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MISSION
To improve the survival and quality of life of patients with thalassaemia through the promotion and support of: education, advocacy and capacity building of patients’ and their families’ awareness and education programmes for the community collaboration with national, regional and international health authorities aiming to (a) prioritise thalassaemia on national, regional and International health agendas; (b) develop and implement national disease specific programmes for its effective control, prevention and holistic care, and research programmes and studies focused on the final, total cure (c) Establish equal access of every patient with thalassaemia to high quality health and social care services provided through truly patient-centred healthcare systems.

VISION
Establishment of equal access of every patient with thalassaemia to high quality health and social care services provided through truly patient-centred healthcare systems.

BECOME MEMBERS OF THE THALASSAEMIA COMMUNITY

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