Programme Book

2018 ACMBMB

3rd ASEAN Congress on Medical Biotechnology and Molecular Biosciences

In conjunction with

2nd Conference of Malaysian Society of Human Genetics

July 9 – 10, 2018

The Gurney Resort Hotel & Residences Penang, Malaysia

Breaking The Frontiers of Biosciences Towards The Era of Precision Medicine

Professor Dr. Sultana MH Faradz | Professor Dr. Ramdan Panigoro | Professor Sir John Burn | Professor Dr. Nazalan Najimudin | Associate Professor Dr. Lai Poh San | Emeritus Suthat Fucharoen | Dr. Carsten Lederer | Dr. Domenico Coviello

Organized by

Jointly Organized by:

Malaysian Society of Human Genetics (MSHG) | Mahidol University | Universitas Padjajaran (UNPAD) | University of The Philippines | Persatuan Genetik Malaysia

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MESSAGE FROM: YBrs. PROFESSOR DATUK DR.ASMA ISMAIL VICE-CHANCELLOR, UNIVERSITI SAINS MALAYSIA

Assalamualaikum WBT and Salam Sejahtera.

Welcome to the 3rd ASEAN Congress on Medical Biotechnology and Molecular Biosciences (ACMBMB 2018) in conjunction with the 2nd Conference of Malaysian Society of Human Genetics in Penang, Malaysia. I applaud the efforts of Universiti Sains Malaysia to host the Congress this year after Mahidol University in 2015 and Universitas Padjadjaran in 2016.

I am pleased that “Breaking Frontiers of Biosciences Towards the Era of Precision Medicine” was selected as the theme of this congress. The concept of precision medicine underlies on the basis of genetics, environmental and lifestyle factors, to transform the diagnosis and prevention of diseases. With the advances in biomedical and biosciences technologies, and contribution of researchers from diverse backgrounds, precise and personalized patient care might possibly be turned into a clinical reality.

It is hoped that this congress will act as a podium for local and international students, researchers, industries, government agencies and other professional to disseminate, discuss and share information on the latest advances in medical biosciences.

I would like to acknowledge the organizing committee for their commitment and dedication in organizing this congress especially to Universiti Sains Malaysia, Malaysian Society of Human Genetics, Indonesian Society of Human Genetics, Mahidol University, Universitas Padjadjaran and University of Philippines. I am delighted to see such a large convergence of participants for this two-day event in sharing priceless knowledge and discoveries attained in research. My acknowledgement also goes to all the speakers who would be sharing their expertise with us.

I also would like to thank all the sponsors from the industry and private sector in supporting this congress. With your continued interest and support, I am sure this congress would be a huge success and an unforgettable experience.

To all participants, do take some time to visit the Universiti Sains Malaysia main campus as well as to walk around the heritage city of Georgetown, which was named as a Unesco’s World Heritage Sites in 2008 for having many well-preserved collection of historical buildings in Malaysia.

Finally, I wish everyone a wonderful time here and may your participation in this congress be productive and truly rewarding.

Thank you.

PROFESSOR DATUK DR. ASMA ISMAIL, FASc.
Vice-Chancellor
Universiti Sains Malaysia

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MESSAGE FROM: PROFESSOR DR. ZILFALIL ALWLI CHAIRMAN OF THE ORGANANISING COMMITTEE ACMBMB AND VICE PRESIDENT OF MALAYSIA SOCIETY OF HUMAN GENETICS (MSHG)

Welcoming Message from 3rd ACMBMB Chairman

Dear friends and colleagues,

With great pleasure and honour, I would like to welcome all of you to the 3rd ASEAN Congress in Medical Biotechnology and Molecular Biosciences (ACMBMB), held in conjunction with the 2nd annual conference of MSHG 2018. I would like to extend a special welcome and appreciation to all the speakers, especially those who have travelled thousand miles away from abroad, to be here in the spirit of sharing their expertise with us. I would also like to thank all young and senior researchers for supporting the conference with much enthusiasm. I am very impressed by the high quality of the submitted abstracts for this congress.

This year, Universiti Sains Malaysia hosts the 3rd ACMBMB under the title “Breaking The Frontiers of Biosciences towards The Era of Precision Medicine”. The congress features two keynotes lectures, six plenary lectures, four guest speakers and a long list of selected oral and poster presentations. The congress has attracted a heart-warming response from regional researchers and clinicians and a wide range of participants from five neighbouring ASEAN countries.

The main purpose of ACMBMB 2018 is to provide a platform for researchers and students from various disciplines to socialise and exchange valuable information. I hope this two-day congress will be fully utilised to foster in-depth discussions and collaborations between scientists, clinicians and researchers as we strive towards advancing the field of biomedicine and biotechnology for the betterment of current clinical practices and general wellbeing.

I believe this congress will contribute to the development of regional advancement of precision medicine through an exchange of the latest trends and key findings in biosciences research.

Last but not least, I sincerely hope that all of you will take this opportunity to experience the warm hospitality of unique cultural heritage and diverse ethnic background in Malaysia. On behalf of the organising committee, I wish you a fruitful and enjoyable meeting.

Thank you

Professor Dr. Zilfalil Alwi
Chairman of the Organising Committee
ACMBMB, Penang 2018

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MESSAGE FROM ASSOC. PROF DR. SARINA SULONG PRESIDENT OF MALAYSIA SOCIETY OF HUMAN GENETICS (MSHG)

Assalamualaikum wrbt and Salam Sejahtera.

On behalf of Malaysian Society of Human Genetics (MSHG) I am pleased to welcome all delegates to the 3rd ASEAN Congress of Medical Biotechnology & Molecular Biosciences (ACMBMB) 2018 which is held in conjunction with the 2nd Conference of Malaysian Society of Human Genetics (MSHG) which runs from 9th to 10th July here in Penang, Malaysia. It gives me great pleasure to wish all delegates "Selamat Datang" and warm welcome. We are very proud to be co-organizing the conference this year as our first inaugural conference of MSHG took place in May 2013. It is high time that such an event took place to highlight the remarkable contribution of important topics related with medical biotechnology, molecular biosciences and human genetics.

I very much look forward to welcoming and seeing you at this conference. I'm sure all of us will have fruitful, rewarding exchanges and productive intellectual session at this ACMBMB in the next few days. I wish you every success with this important conference and I would like to express my sincere for all participants to discover new opportunities in the still growing area of research.

I would like to extend my gratitude and that of the entire ACMBMB conference organising committee, their efforts in developing such a stimulating and interesting conference programme. Well done and thanks all team member including the Universiti Sains Malaysia (USM) as the host for this event. Also, I am warmed and encouraged by your overwhelming support as participants of this conference and let us all meet again in the next conference.

Sarina

Assoc. Prof. Dr. Sarina Sulong
President
Malaysian Society of Human Genetics (MSHG)

Malaysian Society of Human Genetics (MSHG)

The Malaysian Society of Human Genetics was established in 2012. It is a professional body that provides a platform for the human genetics throughout Malaysia to meet and exchange ideas.

Aims & Objectives:

- Promote networking among scientists working in the field of human genetics in Malaysia.
- Encourageand promote activities that will increase knowledge in the field of human genetics.
- Encourage and promote training in human genetics in Malaysia.
- Provide expert advice in human genetics to the government and statutory bodies and non-statutory bodies in Malaysia.
- Promote collaboration with other organizations, whether national or international in the field of human genetics.
- To encourage and promote research grants in the field of human genetics.
- To collect and receive contributions from certain parties in order to achieve the goal of association through programs or activities organized by the association.

Membership:

- Open to Malaysian citizens residing in Malaysia, regardless of gender, race, religion, who have attained the age of 18 years and above.

Member Type	Annual Fee
Ordinary Members (Individuals involved in area related to genetics and live in Malaysia)	**RM30**
Lifetime Member (Ordinary members who pay fees for life)	**RM200** **(Once in a lifetime)**
Associate Member (Malaysian citizens residing overseas and non-Malaysian citizens residing in Malaysia and outside Malaysia)	**RM150**
Student (Malaysian students and non-Malaysians studying in institutions of higher learning in Malaysia)	**RM10**
Honorary Members (Appointed if the association can contribute towards the development of society, especially at the international level. Appointments are suggested and agreed upon during the annual general meeting)	

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CONGRESS AT A GLANCE

ACMBMB 2018

CONGRESS AT A GLANCE

ASEAN Congress on Medical Biotechnology and Molecular Biosciences (ACMBMB) is an annual joined forces event organised by Universiti Sains Malaysia, Mahidol University, Universitas Padjajaran and University of the Philippines. Universiti Sains Malaysia is going to host the Congress this year after Mahidol University (2015) and Universitas Padjajaran (2016). ACMBMB 2018 is to be held in conjunction with the 2nd Conference of Malaysian Society of Human Genetics from July 9th to 10th this year at the Gurney Resort Hotel & Residences, Penang, Malaysia. The theme for this year is "Breaking the frontiers of biosciences towards the era of precision medicine".

Similar to the previous ACMBMB, ACMBMB 2018 is a multidisciplinary effort to improve and break barriers among basic and translational research in the field of biosciences, especially in medical biotechnology and molecular biosciences. It will feature distinguished plenaries, interactive symposia, poster and platform opportunities for free papers, state-of-the-art review course, and ample of networking opportunities.

The Congress aims to create a platform where researchers across the globe, especially from the Asia Pacific region, can reflect on advances made in the scientific fields of human genetics, medical biotechnology and molecular biosciences. Thus, this conference is the best place to learn about the contemporary research progress and anticipate future development of those disciplines. The congress will bring together over 100 delegates, among the local and international healthcare professionals across all sectors including doctors, scientists, technicians and postgraduate students.

The congress will also serves the delegates with inspiring and up-to-date knowledge from eminent scientists such as from the Newcastle upon Tyne Hospitals NHS Foundation Trust. (United Kingdom), Mahidol University (Thailand), The Cyprus Institute of Neurology & Genetics (Cyprus), Diponegoro University (Indonesia), Galliera Hospital, Genova (Italy), Universitas Padjadjaran (Indonesia) and National University of Singapore and Khoo Teck Puat-National University's Children Medical Institute (KTP-NUCMI).

The added value for this event is, ACMBMB 2018 will witness a Memorandum of Understanding (MoU) exchange ceremony between Malaysian Society of Human Genetics and Indonesian Society of Human Genetics that may bridge the gap between the two countries through the two scientific societies. Besides that, the selected abstracts will be suggested to be published as full articles in the Asian Journal of Medicine and Biomedicine (AJMB).

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FACULTY OF SPEAKERS

Keynote I
Professor Sir John Burn (Newcastle Upon Tyne, United Kingdom)

Biography

Professor Sir John Burn obtained an MD with distinction, a first class honours degree in human genetics from Newcastle University, where he has been Professor of Clinical Genetics since 1991 and a consultant specialist since 1984. He led the regional NHS Genetics Service for 20 years and helped to create the Centre for Life which houses an education and science centre alongside the Institute of Genetic Medicine and Northgene Ltd, the identity testing company he launched in 1995. He chairs DNA device company QuantuMDx. He was knighted in 2010, chosen as one of the first 20 'local heroes' to have a brass plaque on Newcastle Quayside in 2014. He received the Living North award in 2015 for services to the North East 2000 – 2015. He is also Executive Chairman of the international organisation, the Human Variome Project, which seeks to share knowledge of genetic variation for clinical benefit. In 2017, John became Chairman of the Newcastle upon Tyne Hospitals NHS Foundation Trust.

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Keynote II
Professor Emeritus Dr Suthat Fuchareon (Mahidol University, Thailand)

Biography

Dr. Suthat Fuchareon is Professor Emeritus at the Institute of Molecular Biosciences, and previous Director of the Thalassemia Research Center, Mahidol University, Nakornpathom, Thailand.
Dr. Fuchareon is internationally recognized for his work on thalassemia in Thailand and Southeast Asia. His scientific interests encompass the spectrum of basic, translational, clinical and epidemiological research. Work from the Thalassemia Research Center at Mahidol University has set the standard for defining the molecular genetics, genomics, and genotypic/phenotypic correlations of thalassemic syndromes. His many publications include clinical trials on the use of inducers of fetal hemoglobin and iron chelation, the use of MRI imaging for assessment of iron overload, as well as landmark epidemiologic studies defining the genetic diversity and public health burden of these diseases.

Dr. Fuchareon has been the recipient of many honors, among them the Outstanding Researcher Award from the National Research Council of Thailand, Outstanding Scientist Award from the Foundation for the Promotion of Science Award and Technology under the Patronage of His Majesty the King of Thailand, the Royal Dusdhi Mala Medal in Science, awarded by H.M. the King of Thailand. Golden Silk Award, Guangxi Medical University, PRC. He served as the Co-Chair for the 2009 NHLBI/NIDDK Thalassemia Workshop: Clinical Priorities and Clinical Priorities. Dr. Fuchareon had delivered the Berend Houwen Lecture at the International Society for Laboratory Hematology in New Orleans on May 6, 2011.

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Plenary I
Dr. Carsten Lederer
Cyprus Institute of Neurology and Genetics & The Cyprus School of Molecular Medicine

Biography

Dr Lederer received his PhD from the University of East Anglia for work in plant virology at the John Innes Centre, Norwich, UK. He is now a group leader at the Molecular Genetics Thalassemia Department (MGTD; head: Marina Kleanthous) of the Cyprus Institute of Neurology and Genetics (CING), where he heads the MGTD Gene Therapy and Genome Editing unit. Dr Lederer is Assistant Professor and course coordinator at the Cyprus School for Molecular Medicine, executive board member of the Global Globin 2020 Challenge, curator of the ITHANET Portal, board member of the Cyprus Society of Human Genetics and a member of the European Society of Human Genetics and the Hellenic Society of Gene Therapy and Regenerative Medicine
.

Research Interests

Dr Lederer's current research focus is the gene therapy of β-globinopathies and particularly of β-thalassaemia by three different approaches, a) mutation-specific RNAi-supplementation of gene addition, b) genome editing of disease modifiers and c) homology-independent gene repair. Additional scientific activities include involvement in the development of the ITHANET Portal and its classification of pathological variants, as well as the promotion of the Global Globin 2020 Challenge for prevention, disease management and comprehensive epidemiology in low and middle-income countries.

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Plenary II
Professor Dr. Sultana MH Faradz
MSHG-InaSHG Plenary Lecture): Sex Development Disorders: Mistaken Identity

Biography

In 1978, she graduated from the Faculty of Medicine Diponegoro University (FMDU) and was recruited as a lecturer at FMDU. In 1988 and 1990 she joined cytogenetics course in Tottori University Japan, followed with cancer cytogenetics at the Prince of Wales Hospital Sydney in 1992 and her PhD on Medical Genetics with the title of her thesis on Fragile X syndrome (1994-1998) at the UNSW, Sydney during which she did her course on Clinical Genetics at Sydney Children Hospital (1994-1995), Clinical epidemiology at School of Community Medicine, UNSW Sydney (1995), a research fellow at Queen's University Kingston Canada (1996 and 1997). She was a research fellow at AMC Hospital, Amsterdam (2000), RUNMC Nijmegen (2000) and the MIND Institute at the University of California, Davis (2002). Since 2001, she started working as a genetic counselor in 2 hospitals. In 2003, she has been appointed as a professor of medical science. She is a member of National Research Council in 2005-2011. In 2005, she awarded the best lecturer at MFDU. In 2006 she led the establishment of the First Master Program on Genetic counseling in Indonesia in collaboration with several centers in overseas. Since 2007 she has been appointed as a director of Center for Biomedical Research at FMDU. In 2008 she awarded a best scientist from the Mayor of Semarang Municipality and in 2009 awarded Australian Alumni Finalist in Research and Innovation from the Australian Embassy in Jakarta. In 2010 she awarded a Program Scheme Academic Mobility Exchange fellowship for research project on DSD at Murdoch Children Research Institute/ University of Melbourne for 3 months. She was appointed as a vice Rector for development and collaboration in 2011-2015. In 2012 she awarded as National best researcher in Medicine from Bakrie foundation. Since 2013 she is a member of Indonesian Academy of Science (AIPI). In 2013, 2015 and 2016 she received an award from Diponegoro University for the achievement of International scientific article publication with major research in intellectual disability and disorders of sex development. She published her research in >50 peer reviewed journal. She is a member of APSHG, HGSA, ASHG and president of InaSHG (Indonesian Society of Human Genetics).

FACULTY OF SPEAKERS

Plenary III
Dr Domenico Coviello
Galliera Hospital, Genova

Biography
Domenico Coviello (MD, PhD) is a medical geneticist, Director of Laboratory of Human Genetics, Galliera Hospital, Genoa, since 15/3/2010. Head of Medical Genetics Laboratory, Ospedale Maggiore Policlinico, University Hospital Milan, Italy (2000-2010), teaching responsability at Milan University, Faculty of Biotechnology. He has been working at Modena University (1998-2000), and Genoa University 1989-1997).

He worked in international labs: 1978-Cytogenetic Laboratory, Guy's Hospital Medical School, London; 1984-William Dunn School of Pathology, University of Oxford; 1987-1988-Department of Molecular Genetics, M.D. Anderson Cancer Centre, University of Texas, Houston, Texas, USA; 1991-Human Genetics Branch, NICHD, NIH, Betesda, MD, USA (Dr. J Marini); 1995 and 1997 - Department of Genetics, Harvard Medical School, Boston, MA, USA.

EU projects: 1995-1997- EC Concerted Action Biomed I on Hypertrophic Cardiomyopathy; 1997-1999- EC Concerted Action Biomed II EUROSCREEN. 2005-2009 Eurogentest. 2007-2009 Eurogenguide. 2008-2010 Eurogene. 2009-2012 PHGEN II.

Scopus author ID: 7003921883,
ResearcherID: J-9477-2016

1999-2005 Member of ESHG Public and Professional Policy Committee;
2004-2009 Chairman of ESHG Educational Committee;
2009-2014 Board member of the ESHG
2013-2018 Board member of Italian Society of Human Genetics (SIGU)
2016-2018 Board member of EMQN (European Molecular Genetics Quality Nerwork)
2014-2018 Secretary General of European Board of Medical Genetics (EBMG)
2015-2020 Human Variome Project Member– Co-Leader Italian Node

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FACULTY OF SPEAKER

Plenary V
Assoc. Prof Dr. Lai Poh San
National University of Singapore

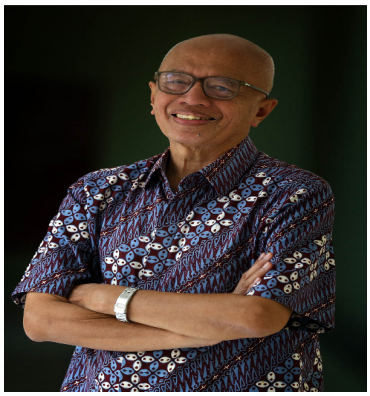
Biography

Associate Professor Poh-San LAI heads the Human Molecular Genetics Lab of the Dept of Paediatrics at Yong Loo Lin School of Medicine, National University of Singapore and Khoo Teck Puat-National University's Children Medical Institute (KTP-NUCMI). Her interests are in genetics and genomics. She has several projects related neuromuscular disorders, congenital diseases and undiagnosed disorders. She has been involved in the development of genetic tests for molecular analysis, prenatal diagnosis and genetic counseling of pediatric genetic diseases in Singapore having first set-up the laboratory for molecular studies and diagnosis in pediatric disorders such as Duchenne muscular dystrophy, retinoblastoma, hemophilia, spinal muscular atrophy, etc. in 1990. For this work, she was the recipient of the Singapore National Youth Award for Science and Technology in 1998 (the highest accolade in the country for Science and Technology in youth category). Poh San was also the sole recipient of the Young Investigator's Award given by the Singapore Academy of Medicine in 1992 in recognition of outstanding original research work presented at the 26th Singapore-Malaysia Congress of Medicine. Currently, she has special interests in harnessing novel next-gen technologies to understand disease pathogenesis for clinical applications. Her other research interests are in behavior genetics and exploring psychosocial, ageing, lifestyle, behavioural, biological and genetic determinants related to various traits.

Poh San serves on a number of international consortiums, advisory committees, editorial journal boards and societies. She is President of the Biomedical Research and Experimental Therapeutics Society of Singapore (BRETSS) and immediate Past President of Asia-Pacific Society of Human Genetics (APSHG), She is an executive committee member of the International Federation of Human Genetics Societies (IFHGS), and member of the American Society of Human Genetics (ASHG) and American College of Medical Genetics (ACMG). She is Co-Chair for the HUGO Pan-Asian Population Genomics Initiative (HUGO-PAPGI) and has served as a member of the Scientific Advisory Board for health program at the Philippines Genome Center.

She is an adjunct Faculty member of the Genome Institute of Singapore (GIS), Adjunct Principal Member of Technical Staff with the Defence Medical and Environmental Research Institute (DMERI), Defence Science Organization, Singapore and Scientific Advisor to the Molecular Diagnostic Centre (MDC) at NUH. She sits on a number of local committees such as the Local Review Panel (OF-YIRG) of the National Medical Research Council, Institutional Review Board for the Defence Science Organization (DSO) and Singapore Armed Forces (SAF) and Institutional Review Board for Lee Kong Chian School of Medicine, Nanyang Technological University. At National University of Singapore, Poh San is the Deputy Chair of the Institutional Biosafety Committee and Vice-Chair of MD1 Tahir Foundation Building Research Safety Committee. She also serves as Technical Assessor/Expert for the Singapore Accreditation Council (Singapore Laboratory Accreditation Scheme). She sits on the Advisory Board on several genetics and genomics-related organizations and serves on the Editorial Boards of several journals. She also serves on Ph.D. committees, and has trained both graduate students and interns. She has extensive collaborations with overseas and local institutes, holding or having held more than 25 national grants awarded as Principal Investigator or Co-Principal Investigator. She has two international patents filed in the field of diagnostics and therapeutics.

FACULTY OF SPEAKER

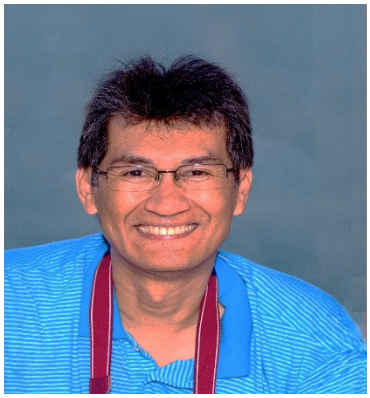


Plenary IV
Prof Dr. Ramdan Panigoro
Faculty of Medicine, Universitas Padjadjaran UNPAD

Biography

Ramdan Panigoro is a Professor of the Department of Biomedical Sciences in the Faculty of Medicine, Universitas Padjadjaran (UNPAD). He has passion in the field of biochemistry and molecular biology, immunogenetic, and human leucocyte antigen (HLA). He is also a member in Medical Genetics Research Center UNPAD and Research Ethics Committee of the university. After finishing his study on Medical Doctor program (UNPAD, 1973), his passion in immunogenetic has brought him to complete both MSc and PhD degree in University of The New South Wales, Australia focusing on human red blood cells, HLA, and ABO gene polymorphisms in the Indonesian people. At the moment, Prof Panigoro leads a multidisciplinary research team bridging the biomedical, clinical, and social science of thalassemia. His latest research, entitled "The Implementation of Biomedical and Psychosocial Management Approach to Improve the Quality of Life in Thalassemia Patients" has produced several articles on various international journals. This research has successfully seized public attention since his research team is actively educating people about thalassemia and its complications, how to deal with thalassemia, and advocating with local government in West Java Province, mainly in the District of Bekasi, Pangandaran, Tasikmalaya, Sukabumi, and Garut, where most of thalassemia patients in Indonesia reside in. Prof Panigoro showed his concern on improving biomedical science as he is the Chairman of the Third, Fourth, and Fifth Bandung International Biomolecular Medicine Conference (BIBMC) in 2014, 2016, and 2018. He was also pioneered the partnership and research collaboration between UNPAD, Universiti Sains Malaysia, Mahidol University, and University of the Philippines.

FACULTY OF SPEAKERS



Plenary VI
Prof Dr. Nazalan Najimuddin
Universiti Sains Malaysia

Biography

Nazalan Najimudin is currently a Professor in Molecular Genetics at the School of Biological Sciences, Universiti Sains Malaysia (USM). He earned his Bachelor of Science (Honours) degree in Biochemistry at the University of Melbourne in 1982. He then pursued his graduate study at Cornell University (USA) in Genetics and Development under the supervision of Prof Stanley A. Zahler. He returned to Malaysia in 1989 to join Universiti Sains Malaysia where he maintained his interest in gene regulation and evolution. His laboratory team is currently studying the nitrogen fixation systems in Gram positive bacteria, especially the Paenibacillus group. The genomes of several species of this group have been completed to understand the evolution of their nitrogen fixation regulons. In depth investigations are being performed in order to understand how the nitrogen fixation genes are being regulated. He was also the Life Science Research Dean of USM from 2006-2012 before returning full time to his laboratory at the faculty. He has recently been appointed the Director of Nexus (Science) which is tasked with driving the R D C & I of the university into translational activities. He is also a life member of Genetics Society of Malaysia. He is strongly of the view (and is convinced) that by educating all life science undergraduates in GENETICS, they will have a wholesome view of biology, right from the level of organismal biodiversity to cellular and molecular functions.

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SPEAKER'S ABSTRACT

Keynote I
Importance of human genetics towards precision medicine
Professor Sir John Burn
Newcastle Upon Tyne, United Kingdom

Abstract

Caring for the sick and studying traits that run in families are both as old as our civilisations. For the last two centuries diagnoses and treatment have been transformed by science. Human genetics has medical and scientific origin over 100 years old and has often been overlooked in the transformation; discovery of blood groups, eradication of rhesus disease and new born screening of inborn errors all have their origins here. Now that whole genome sequencing is becoming routine, the stage is set to change an individual's life course. Managing 8000 rare disease and understanding cancer are only the beginning. This presentation will explore the technical and organisational barriers to be overcome as we use genomics to drive a global move towards precision medicine.

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SPEAKER'S ABSTRACT

Keynote II

Molecular biology of thalassemia for personalized medicine

Professor Emeritus Dr Suthat Fuchareon
Mahidol University, Thailand

Abstract

Thalassemia is one of the most common congenital blood disorder worldwide. The defect in the globin genes decreases production of - or - globin chain causing chronic anemia. The complex gene-gene interaction leads to many thalassemic diseases including homozygous -thalassemia, -thalassemia/Hb E and Hb Bart's hydrops fetalis. Prevention and control of thalassemia is possible with the combination of providing the best care for existing cases and preventing the birth of new cases. Treatment for thalassemia major is regular blood transfusion to maintain adequate levels of the hemoglobin concentration. Iron overload in thalassemia patients is secondary to either multiple blood transfusions or increased iron absorption or the combination of both. Patients with severe iron overload need iron chelator. Cure for thalassemia is possible by stem cell transplantation and recently by gene therapy.

The basic concept in medicine includes diagnosis, therapeutic intervention and preventive measure. Advances in biomedical research, especially molecular biology, have certain impact in medicine. Thalassemia is a good example in using molecular biology to identify globin gene mutation causing - and - thalassemia. This leads to molecular diagnosis and prenatal diagnosis which are major parts of thalassemia control program. Moreover, knowing mutation of the gene has led to gene therapy which is the cure for the disease. All of these are the proof of concept of "personalized medicine" in the new era.

Inthe near future mass thalassemia screening may be carried out by using next generation sequencing (NGS) technology. Both NGS screening and gene therapy will affect the health care system of a country. At current cost it is not possible for a family to absorb the expensive cost of NGS and gene therapy. Moreover, using NGS for accurate diagnosis of thalassemia will need a group of bioinformaticians to analyze data. This is a challenge to the society how to cope up with personalized medicine.

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SPEAKER'S ABSTRACT

Plenary I

Gene editing for genetic disorders; therapy development in the fast lane

Dr. Carsten Lederer
Cyprus Institute of Neurology and Genetics & The Cyprus School of Molecular Medicine

Abstract

Gene editing and in particular the advent of CRISPR/Cas9 technology may turn the tide on therapy development for many genetic disorders. As pharmaceutical companies have begun to embrace gene therapy for the treatment of human disease, the principle of gene editing fundamentally changes the scope and speed of development for molecular medicines. Gene therapy by gene addition took decades to reach the clinic by incremental disease-specific refinements of vectors and methods, whereas gene therapy by genome editing in its basic form merely requires certainty about the causative mutation and access to target cells of interest. CRISPR/Cas9 technology in particular went from concept to first clinical trial in three years instead of thirty: therapy development in the fast lane. This presentation will summarise key achievements to date in therapy development based on different gene editing tools and will give a perspective on their employment for the treatment of genetic disorders. With emphasis on CRISPR/Cas9 technology, this talk will then highlight ongoing improvements to specificity, delivery and efficiency of existing tools, discovery of new enzymes, and ingenious reengineering and reemployment of gene editing tools as genome disruptors, transcriptional regulators, epigenetic modifiers and base editors. Thousands of orphan diseases are up for adoption, and recent developments in the gene therapy field finally conspire to find many of them a good home.

Keywords

Anemia, Sickle Cell, Beta-Thalassemia, Genetic Therapy, Gene Editing, Fetal Hemoglobin

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SPEAKER'S ABSTRACT

Plenary II
Disorders of Sex Development: Mistaken Identity
Professor Dr. Sultana MH Faradz
Universitas Diponegoro, Indonesia

Abstract

Disorders of sex development (DSD) are defined as congenital conditions in which development of chromosomal, gonadal or anatomical sex is atypical, while in clinical practice this term means any abnormality of the external genitalia. This term replaced hermaphrodite, pseudohermaprodite, intersex and ambiguous genitalia. DSD is classified based on karyotype, hormone assessment and imaging; and molecular and/or histopathology examination into 3 group i.e. DSD associated with sex chromosome abnormalities (sex chromosome DSD) and DSD with a normal chromosome complement (46, XX DSD and 46, XY DSD).

The majority of children born with DSD are identified in the new-born period due to visible genital differences; however, detection can occur later when expected features of pubertal development do not progress on time. The most common cause of 46, XX DSD is Congenital Adrenal Hyperplasia which is inherited as autosomal recessive manner, while androgen Insensitivity Syndrome (AIS) is a X linked inheritance and the most common identified of 46, XY DSD.

Management of DSD is challenging especially if identified at later age and when gender change issue is considered. Problems which may occur if one DSD patient is missed or under diagnosed, and eventually mistreated involve not only medical aspect, mainly dealing with sex assignment and consecutive management of therapy, but also psychosexual aspect. It is really hard for the patients and their families when they find that their choice of gender and sexual rearing have been incorrect.

Thorough phychological and genetic counseling as well as cultural adaption and religion consultation need to be done in orderly manner. Comprehensive multidisciplinary management with the proper diagnosis for USD help patient in gender assigment and support patient to improve quality of life.

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SPEAKER'S ABSTRACT

Plenary III
Synopsis of the talk "Human Genetics: Beyond Boundaries Technology"
Dr. Domenico Coviello
Galliera Hospital, Genova

Abstract

Medical biotechnology applied to DNA has dramatically changed the molecular diagnosis approach in the genetic field. From "Medical Genetics" that classically include mainly the mendelian diseases and congenital malformations syndromes we are now completely absorbed by "Medical Genomics" that involve all fields of medicine dealing not only with genes that presents causing disease mutations, but with a tremendous amount of gene "variants" that contribute, in all the complex diseases, in determining the phenotype variation or disease predisposition. DNA analysis today require a large contribution from bioinformatics, the many DNA variants need to be investigated on large number of subjects and needs to be validated with in vitro or in vivo functional studies. Sharing clinical and molecular data is one of the most important challenge that we are facing. Specific database needs to be used in conjunction with big data obtained from normal population or from complete life records of single patients. The vision is not only precision medicine to cure disease at early stage but we are in the phase of reverse medicine where, from nucleic acid analysis (coding and non-coding sequences), we will define the disease in the earliest phase of its presentation or even before the clinical manifestation.

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SPEAKER'S ABSTRACT

Plenary IV

Building Awareness of Thalassemia Through Interprofessional Collaborative Practice

Professor Dr. Ramdan Panigoro
Universitas Padjajaran, Bandung, Indonesia

Abstract

Thalassemia, a hemoglobinopathy caused by absence or defect in globin chain synthesis, ranks as the most common monogenic disorders in the world. Thalassemia patients require continuous blood transfusion and iron chelator agents through their whole life. Chronic transfusion and inadequate chelator consumption may raise issues such as iron overload induced multiple organ failure and quality of life in the context of caregiving and social life problems. Therefore, optimizing multidisciplinary approach is clearly needed to counter the arising bio-psycho-social problems of thalassemia, especially in young adults. Thalassemia working group Universitas Padjadjaran is a research group that focus on bench to bedside research to improve the outcome of thalassemia patients. Our study has mapped complications of cardiomyopathy, neurology and cognitive disorders, immune system response as well as developing iron chelators from natural resources (Caesalpinnia sappan L.) and creating animal model bridging biomedical and clinical science. We also collaborate with nurse and social worker to address caregiving and social problems faced by thalassemia patients. Therefore, strengthening partnerships between academic, business, government, and media sectors is needed to solve global challenge of thalassemia.

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SPEAKER'S ABSTRACT

Plenary V
The future of genetics screening - 4th industrial revolution
Associate Professor Dr. Lai Poh San
National University of Singapore

Abstract

It is undisputed that the last three industrial revolutions have changed the way mankind live, work and play. With the arrival of the 4th industrial revolution, we are witnessing the breathtaking transformation of individual physical and biological domains, such genomics, bioengineering, nanotechnology and digital analytics, into powerful merging technologies that can impact healthcare and precision medicine. Indeed, the mapping of human genome and discoveries of genes that are responsible for disease susceptibility have changed the way in which we are able to better manage and treat many genetic disorders. Genetic screening offers opportunities to perform diagnosis, carrier screening, disease stratification or prognosis, and treat or cure disorders that are caused by abnormalities in the functioning of gene(s). Predictive medicine may also offer opportunities to prevent diseases before they occur if we identify them early enough through genetic screening. Genetics screening is not a novel concept as we have been performing genetic testing for conditions such as G6PD deficiency, IEMs, thalassemia, SMA, DMD, etc. for quite some time. However, conventional methods such as biochemical, cytogenetics or single gene testing are slowly being replaced with faster and more efficient screening methods. As technology-driven discoveries are being made at an unprecedented pace, we are at the cusp of new approaches that harness AI, IoT, cloud computing, robotics, social media, smart-phones, fit-bit wearables and other digital technologies with whole genome-level data and personal health-information that will shape how we will deliver healthcare to the public in unimaginable ways. This talk will discuss on how current technology breakthroughs may shape the way genetic screening will be conducted in future as novel genetic testing methods are being increasingly adopted into routine laboratory and clinical practice. Next – gen sequencing approaches, for example, have already moved into the realm of clinical-grade sequencing in offering precise data-driven diagnosis for many patients. Increasingly too, new modalities for personalized genetics screening have been spun off such as in the realms of pharmacogenomics, wellness programmes, diet management, etc. As the 4th industrial revolution starts generating tangible out comes for improved healthcare, we should also be aware of the concerns and potential pitfalls generated by these technologies.

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SPEAKER'S ABSTRACT

Plenary VI

Searching for Remedies of Ageing-Associated Disorders Using Tiny Animals as Models

Prof Dr. Nazalan Najmuddin (Universiti Sains Malaysia)

Abstract

Tiny invertebrates have become popular models for research because of their relatively short lifespan and ease of maintenance. While the fruit fly Drosophila melanogaster has a long history as a model organism and the nematode worm Caenorhabditis elegans is fast catching up since the early 1960s. When the sequence of the entire genome of C. elegans became available, it became apparent that the similarity of genes in this microscopic nematode and those in humans is remarkable. The nematode is not only a good model for studying general biological processes as well as elucidating the pathogenesis of diseases. Alzheimer's disease (AD) is a chronic, irreversible brain disease attributed to the accumulation of extracellular senile plaques comprising β-amyloid peptide (Aβ). A transgenic C. elegans containing overexpressed human - amyloid proteins strain offers a nice model as a screening system to look for natural products that will potentially improve its lifespan. This transgenic Caenorhabditis elegans exhibited severe paralysis and a shorter life span of about a week when the □-amyloid protein is expressed. In this study, this particular strain of nematode was used to study the anti-paralysis effect of Danshen (root of Salvia miltiorrhiza Bunge). The potential of Danshen water extract, danshensu and salvianolic acid A as therapeutics for AD based on the response of this strain will be discussed. It will be interesting to see how these tiny animals can help improve the aging problems that we face as human. The mechanism that we can uncover using these model organisms could shed some light into the pathways that is required to slow down the process of aging hence improving the quality of life at older age.

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PROGRAMME

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"BREAKING THE FRONTIERS OF BIOSCIENCES TOWARDS THE ERA OF PRECISION MEDICINE"

Hall I and II, Level 2
The Gurney Resort Hotel & Residences Penang, Malaysia

Day 1: 9th July 2018, Monday

0730	Registration
0815	Arrival of VIPs and Invited Guests
0830	Arrival of YBrs. Professor Ir. Dr. Abdul Rahman Mohamed, FASc Deputy Vice-Chancellor Research & Innovation, Universiti Sains Malaysia *representing the Vice-Chancellor, Universiti Sains Malaysia*
0840	Negaraku Menara Ilmu USM Transformation Video Recitation of Doa
0850	*Welcome Address* — *Chair: Dr. Norhayati Yusop & Dr. Norsyahida Arifin* **Professor Dr. Zilfalil Alwi** Organising Chairman of ACMBMB2018 & Vice President of Malaysian Society of Human Genetics (MSHG)
0900	Opening Ceremony **YBrs. Professor Ir. Dr. Abdul Rahman Mohamed, FASc** Deputy Vice-Chancellor Research & Innovation, Universiti Sains Malaysia representing the Vice-Chancellor, Universiti Sains Malaysia
0915	MSHG-InaSHG MoU Signing/Exchange Ceremony
0930	Keynote 1 — *Chair: Professor Dr. Zilfalil Alwi* ***Importance of human genetics towards precision medicine*** Professor Sir John Burn (Newcastle University, United Kingdom)
1030	Photo Session Refreshment and poster viewing
1100	Plenary I — *Chair: Dr. Azlina Ahmad* ***Gene editing for genetic disorders: therapy development in the fast lane*** Dr. Carsten Lederer (Cyprus Institute of Neurology and Genetics & The Cyprus School of Molecular Medicine)
1145	Plenary II: MSHG-InaSHG Lecture — *Chair: Assoc. Prof. Dr. Sarina Sulong* ***Sex development disorders: mistaken identity*** Professor Dr. Sultana MH Faradz (Universitas Diponegoro, Indonesia)
1230	Lunch
1400	Free communication: Lead speaker I — *Chair: Dr. Tan Huay Lin* ***Development of albumin glycation detection platforms for diabetes mellitus monitoring*** Dr. Deanpen Japrung (National Nanotechnology Center, Thailand)
1415	Free communication: Lead speaker II ***The role of Il-8 immunomodulatory pathway in the differentiation of stem cells from human exfoliated deciduous teeth (SHED) into odontoblast-like cells*** Dr. Khairul Bariah Ahmad Amin Noordin (Universiti Sains Malaysia)
1430	Oral presentation I
1645	Refreshment

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Day 2: 10th July 2018, Tuesday

0830	Keynote II ***Molecular biology of thalassaemia for personalised medicine*** Professor Emeritus Dr. Suthat Fuchareon (Mahidol University)	*Chair: Assoc. Prof. Dr. Nazia Majid*
0930	Plenary III ***Human genetics: beyond boundaries technology*** Dr. Domenico Coviello (Galliera Hospital, Genova)	*Chair: Dr. Khairul Bariah Ahmad Amin Noordin*
1015	Tea break	
1045	Free Communication : Lead speaker I ***miRNA as a promising specific biomarkers for ovarian cancer and targeted therapy*** Professor Dr. Sofia Mubarika Haryana (Universitas Gadjah Mada, Indonesia)	*Chair: Dr. Wan Rohani Wan Taib*
1100	Free Communication : Lead speaker II ***High resolution single nucleotide polymorphism analysis of genomic aberrations in childhood acute lymphoblastic leukaemia*** 1) Dr. Zubaidah Zakaria (Institute for Medical Research, Malaysia) 2) Dr. Jeyanthy Eswaran (Newcastle University Medicine Malaysia)	
1120	Oral presentation II	
1300	Lunch	
1400	Plenary IV ***Building Awareness of Thalassemia Through Interprofessional Collaborative Practice*** Professor Dr. Ramdan Panigoro (Universitas Padjajaran, Bandung, Indonesia)	*Chair: Dr Surini Yusoff*
1445	Plenary V ***The future of genetics screening - 4th industrial revolution*** Associate Professor Dr. Lai Poh San (National University of Singapore)	*Chair: Dr. Muhammad Hamdi Mahmood*
1530	Plenary VI ***Searching for remedies of ageing-associated disorders using tiny animals as models*** Professor Dr. Mohd Nazalan Mohd Najimudin (Universiti Sains Malaysia)	*Chair: Dr. Farizan Ahmad*
1615	Awards presentation and closing ceremony	*Chair: Dr. Tan Huay Lin & Dr. Siti Aishah Zainal*
1700	Refreshment and adjourn	

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LIST OF ORAL PRESENTATION

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"BREAKING THE FRONTIERS OF BIOSCIENCES TOWARDS THE ERA OF PRECISION MEDICINE"

Hall 1 and 2, Level 2
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List of Oral Presentations: Applied Sciences
Day 1: 9th July 2018, Monday

Code	Time	Paper Title / Presenter
LP-01-01	1400	Free communication: Lead speaker I ***Development of albumin glycation detection platforms for diabetes mellitus monitoring*** Dr. Deanpen Japrung (National Nanotechnology Center, Thailand)
LP-01-02	1415	Free communication: Lead speaker II ***The role of Il-8 immunomodulatory pathway in the differentiation of stem cells from human exfoliated deciduous teeth (SHED) into odontoblast-like cells*** Dr. Khairul Bariah Ahmad Amin Noordin (Universiti Sains Malaysia)
OP-01-1	1430	***Compressive strength, bioactivity and biocompatibility properties of chitosan-coated carbonate apatite scaffold enriched by gentamicin drug*** Ms. Normahira Mamat (Universiti Sains Malaysia)
OP-01-2	1440	***Anti-cancer activity of Luvunga scandens extracts against human malignant melanoma cells*** Ms. Sama Naziyah Shaban (International Islamic University Malaysia)
OP-01-3	1450	***Development of SERS based biosensor for cancer screening*** Dr. Suwussa Bamrungsap (National Nanotechnology Center, Thailand)
OP-01-4	1500	***Osteogenic Performance of MC3T3-E1 Cells on Granular Hydroxyapatite Scaffold*** Dr. Farinawati Yazid (Universiti Kebangsaan Malaysia)
OP-01-5	1510	***Mode of action of cinnamon bark (Cinnamomum verum) essential oil and the combinatory bactericidal activity with meropenem against KPC-producing Klebsiella pneumoniae*** Mr. Shun-Kai Yang (Universiti Putra Malaysia)
OP-01-6	1520	***2-Methoxy-1,4-Naphthoquinone (MNQ) inhibits glycolytic activities in breast cancer cell (MDA-MB-231)*** Ms. Syukriyah Mat Daud (Universiti Sains Malaysia)

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LIST OF ORAL PRESENTATION

Code	Time	Paper Title / Presenter
OP-01-7	1530	***The effect of the extract Singawalang (Petiveria alliaceae) leaves in decreasing cholesterol blood level and the profile of the liver and the blood vesssel in hypercholesterolemia rats*** Dr. Arifa Mustika (Universiti Airlangga, Indonesia)
OP-01-8	1540	***Tualang Honey and Its Methanolic Fraction Protect Against LPS-Induced Neuroinflammation and Amyloid Deposition in Male Rats*** Mr. Wan Muhammad Hilmi Wan Yaacob (Universiti Sains Malaysia)
OP-01-9	1550	***Application of Malva Nut Gum as Bioreductor and Stabilizer on Gold Nanoparticle Synthesis: Preparation of Chikungunya Immunoassay Biosensor*** Ms. Bevi Lidya (Universiti Padjajaran, Indonesia)
OP-01-10	1600	***NFAT Molecular Profiles of VEGF-induced-dental-stem-cells Cultured on Human Amniotic Membrane*** Ms. Siti Nurnasihah Md Hashim (Universiti Sains Malaysia)
OP-01-11	1610	***DNMT1 is expressed in actively-proliferating lymphoid blast cells and significantly associated with E2F gene set in malignancies*** Dr. Kah Keng Wong (Universiti Sains Malaysia)
OP-01-12	1620	***Establishing Fragile X Research Center: An Experience from Developing Country, Indonesia*** Dr. Tri Indah Winarni (Universitas Diponegoro, Indonesia)
OP-01-13	1630	***Personalised genomics for healthy lifestyle and wellness*** Prof. Dr. Amir Feisal Merican Bin Aljunid Merican (Universiti Malaya, Malaysia)

LIST OF ORAL PRESENTATION

List of Oral Presentations: Basic Sciences
Day 2: 10th July 2018, Tuesday

Code	Time	Paper Title / Presenter
LP-02-01	1045	Free Communication : Lead speaker I ***miRNA as a promising specific biomarkers for ovarian cancer and targeted therapy*** Professor Dr. Sofia Mubarika Haryana (Universitas Gadjah Mada, Indonesia)
LP-02-02	1100	Free Communication : Lead speaker II ***High resolution single nucleotide polymorphism analysis of genomic aberrations in childhood acute lymphoblastic leukaemia*** Dr. Zubaidah Zakaria (Institute for Medical Research, Malaysia) Dr. Jeyanthy Eswaran (Newcastle University, United kingdom)
OP-02-01	1120	***Ultrasensitive detection of circulating miRNA by branched-rolling circle amplification coupled with graphene fluorescence-based sensor*** Mr. Krissana Khoothiam (Mahidol University, Thailand)
OP-02-02	1130	***Bioinformatics Analysis of Differentially Expressed Genes in Liver Cancer for Identification of Key Genes and Pathways*** Ms. Aisya Fathiya Che Rosli (Universiti Sains Malaysia)
OP-02-03	1140	***Identification mRNA MAGE A1-A10 from testicular tissue using new technique*** Dr. Gondo Mastutik (Universiti Airlangga, Indonesia)
OP-02-04	1150	***Identification and characterization of the VPE gene family and its expression in response to Fusarium oxysporum infection in Musa acuminata*** Mr. Wan Muhamad Asrul Nizam Wan Abdullah (Universiti Putra Malaysia)
OP-02-05	1200	***Dopamine receptors (DRD4 and DRD5) mRNA expression in peripheral blood lymphocytes of healthy Malay men subjects*** Dr. Nur Khadijah Muhamad Jamil (Universiti Sultan Zainal Abidin, Malaysia)

LIST OF ORAL PRESENTATION

Code	Time	Paper Title / Presenter
OP-02-06	1210	***Gene Expression Analysis of Stem Cell Migration in Local Angiogenesis of Tissue Repair*** Ms. Nur Syazwani Aziz (Universiti Sains Malaysia)
OP-02-07	1220	***Increased Relative Lymphocyte Number with Reduced Mature Activated T-Lymphocytes (CD3+CD69+) in Stunted Iron Overloaded Major Beta-Thalassemia Patients*** Dr. Mohammad Ghozali (Universiti Padjajaran, Indonesia)
OP-02-08	1230	***Genetic Associations Study of rs12745968 and rs4822752 Between Rheumatoid Arthritis and Schizophrenia Patients*** Ms. Nur Shafawati Ab Rajab (Universiti Sains Malaysia)
OP-02-09	1240	***Family Profile of Down Syndrome Patients in West Java Province, Indonesia*** Mr. Bremmy Laksono (Universiti Padajajaran, Indonesia)
OP-02-10	1250	***Expression of SOX 10 gene in U87 cell treated with siRNAs*** Mr. Mohd Khairi Zahri (Universiti Sultan Zainal Abidin, Malaysia)

LIST OF POSTER PRESENTATION

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List of Poster Presentations: Applied Sciences

Code	Paper Title / Presenter
PA-01	***Combinatorial effect analysis of Peppermint (Mentha x piperita L. Carl) essential oil and meropenem against plasmid-mediated resistant E. coli.*** Ms. Moo Chew Li (Universiti Putra Malaysia)
PA-02	***Molecular and physiological responses of recalcitrant indica rice to lignosulfonates during callus proliferation.*** Mr. De Xian Andrew Kok (Universiti Putra Malaysia)
PA-03	***Oriented antibody conjugation on fluorescence dye-doped silica nanoparticles for targeted in vivo imagin.*** Ms. Kiatnida Treerattrakoon (National Nanotachnology Center, Thailand)
PA-04	***The effect of Flaxseed extract on skin elasticity of the healing wound in rabbit.*** Dr. Omar Abdul Jabbar (International Islamic University Malaysia)
PA-05	***Cytotoxicity assay of nilotinib and nicotinamide in a myeloid cell line, K562.*** Ms. Nur Rasyidah Muhammad (Universiti Sains Malaysia)
PA-06	***Wazir: a VCO based bioproduct developed for Haemorrhoids.*** Mr. Mohd Razip Asaruddin / Dr. Muhammad Hamdi Mahmood (Universiti Malaysia Sarawak)
PA-07	***Cytotoxicity of betel quid and areca nut aqueous extracts on mouse fibroblast and mouth-ordinary-epithelium 1 cell lines.*** Dr. Badr Abdullah Al-Tayar (Universiti Sains Malaysia)
PA-08	***Association of PDFGRA Gene Polymorphisms at Exon 12, 14 and 18 with Clinical Response to Imatinib Mesylate Treatment Among Chronic Myeloid Leukemia Patients.*** Ms. Nur Sabrina Rashid (Universiti Sains Malaysia)
PA-10	***Risk Factors for Psychopathology Experienced by Caregiver of Thalassemia Children.*** Dr. Sjarief Hidajat Effendi (Universitas Padjajaran, Indonesia)
PA-11	***Rapid Aneuploidy Detection of Chromosome 21 by Segmental Duplication - High Resolution Melting Analysis for Prenatal Diagnosis.*** Dr. Annisa Utami Tihnulat (Universitas Diponegoro, Indonesia)

LIST OF POSTER PRESENTATION

Code	Paper Title / Presenter
PA-12	***Generation of induced pluripotent stem cells from normal human dermal fibroblast using non-integrative Sendai virus.*** Ms. Lihui Tai (National Cancer Council, Malaysia)
PA-13	***Identification of Bacteria by Amplification Gene Encoding 16S rRNA and Antibiotics Resistence Test from Outpatients Pneumonia.*** Dr. Tina Rostinawati (Universitas Padjadjaran, Indonesia)
PA-14	***Stability of Glycated Albumin in Human Serum Analyzed by Developed Apta-sensor*** Ms. Chayachon Apiwat (National Nanotechnology Center, Thailand)
PA-15	***Optimization of DNA Isolation Method from Formalin-Fixed-Paraffin-Embedded (FFPE) Tissues and Comparative Performance of Four Different Polymerase Chain Reaction (PCR) Kits.*** Dr. Almira Zada (Universitas Padjadjaran, Indonesia)
PA-16	***A molecular approach in establishing evidence of asymptomatic submicroscopic malaria among the orang asli population in RPS Pos Kemar.*** Adela Ida Jiram / Ms. Nurnadiah Mohd Sukor (Institute for Medical Research, Malaysia)
PA-17	***RET mutation screening in Hirschsprung Patients at a Tertiary Hospital in Indonesia: Mutation Rate and in Silico Analysis.*** Dr.Yunia Sribudiani (Universitas Padjadjaran, Indonesia)
PA-18	***Interaction between sensory dendrite and epithelial cells in Drosophila.*** Mr. Chee Wei Tee (Universiti Sains Malaysia)
PA-19	***Challenges in Diagnosis of Mitochondrial Respiratory Chain Complexes Disorder in Human Skin Fibroblasts.*** Ms. Rosnani Mohamed (Institute for Medical Research, Malaysia)

LIST OF POSTER PRESENTATION

List of Poster Presentations: Basic Sciences

Code	Paper Title / Presenter
PB-01	***DNA Damage Response profiles in reprogrammed osteosarcoma cell lines.*** Ms. Pei Feng Choong (National Cancer Council, Malaysia)
PB-02	***Epithelial-Mesenchymal Transition (EMT) Deregulation in Reprogrammed Oral Squamous Cell Carcinoma (OSCC-iPSCs).*** Ms. Nalini Devi Verusingam (National Cancer Council, Malaysia)
PB-03	***Mutations of FLT3 and CKIT Genes in Core Binding Factor with Acute Myeloid Leukemia: IMR Experience.*** Ms. Zahidah Abu Seman (Institute for Medical Research, Malaysia)
PB-04	***Identification of Chromosomal Translocations in Leukaemia Using Multiplex Reverse Transcriptase Polymerase Chain Reaction: A Retrospective Ten Years Single Institution Study in Multi-Ethnic Malaysia.*** Ms. Nor Rizan Kamaluddin (Institute for Medical Research, Malaysia)
PB-05	***Evaluation of MGMT methylation status among HUSM glioma patients: a preliminary study.*** Ms. Revathy Murali (Universiti Sains Malaysia)
PB-06	***Chromosomal instability in human osteosarcoma is mediated through Hypoxic inducible factor 1 (HIF-1).*** R. Rozi / Dr. Muhammad Hamdi Mahmood (Universiti Malaysia Sarawak)
PB-07	***Impact of Promoter Polymorphisms of Apoptotic Signalling Regulatory Genes Fas/FasI on Chronic Myeloid Leukaemia Susceptibility Risk.*** Dr. Aziati Azwari Annuar (Universiti Sains Malaysia)
PB-08	***An enigmatic hyperdiploid multiple myeloma with novel and complex cyto-genetic abnormalities - A rare occurrence.*** Dr. Foong Eva (Universiti Sains Malaysia)
PB-09	***Sox 6, SOX 13 and SOX 9 Expression Pattern in Meningioma in East Coast Malaysia.*** Wan Rohani Wan Taib / Nurul Balqis Md Dzali (Universiti Sultan Zainal Abidin, Malaysia)
PB-11	***Determination of Drugs Resistance Molecular Markers of Plasmodium falciparum in Malaysia.*** Ms. Afiqah Saleh Huddin (Institute for Medical Research, Malaysia)
PB-12	***A case of allelic dropout in SLC25A13 gene in a patient with Citrin Deficiency.*** Ms. Siti Aishah Abdul Wahab (Institute for Medical Research, Malaysia)
PB-13	***Kennedy Disease: The First Case Report in Malaysia.*** Ms. Nor Azimah Abdul Azize (Institute for Medical Research, Malaysia)
PB-14	***Recognition of Genital Ambiguity as an Unusual Presentation of Klinefelter Syndrome in Childhood.*** Dr. Nurin Aisyiyah Listyasari (Universitas Diponegoro, Indonesia)
PB-15	***Novel mutations in SLC16A2 gene in four unrelated Malaysian boys with MCT8 deficiency.*** Dr. Nur Jannah Arifin (Institute for Medical Research, Malaysia)

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Code	Paper Title / Presenter
PB-16	***Prevalence of Lysosomal Storage Diseases in Malaysia.*** Mr. Affandi Omar (Institute for Medical Research, Malaysia)
PB-17	***RUNX2 Single Nucleotide Polymorphism (rs6930053) In Class II Malocclusions Patients: A Preliminary Study.*** Assoc. Prof. Khairani Idah Mokhtar (International Islamic University Malaysia)
PB-18	***Association study of Janus Kinase 2 gene polymorphisms in Malaysian patients with Crohns disease.*** Ong Shin Yee / Dr. Kee Boon Pin (University of Malaya, Malaysia)
PB-19	***Contribution of GDF-15 and FGF-21 as potential diagnostic biomarkers for mitochondrial respiratory chain disorders.*** Dr. Dyg Pertiwi Abg Kamaludi (Institute for Medical Research, Malaysia)
PB-20	***A Case Report of a Malaysian with WAGR Syndrome*** Dr. Fatimah Azman (Universiti Sains Malaysia)
PB-21	***Sex Chromosomal Mosaicism among DSD patients in Indonesia*** Ms. Mahayu Dewi Ariani (Universitas Diponegoro, Indonesia)
PB-22	***Clinical Profile of Autosomal Dominant Hereditary Ataxia*** Dr. Siti Aminah Soba (Universitas Padjadjaran, Indonesia)
PB-23	***Mitochondrial DNA Hypervariable Segment Region (HVS-I) Analysis of Semoq Beri Population in Peninsular Malaysia.*** Mr. Muhamad Aidil Zahidin(Universiti Malaysia Terengganu, Malaysia)
PB-24	***Genetic variants of BMP4/HpHI and IRF6 /MboI gene in two families with non-syndromic cleft lip and palate patients.*** Prof. Dr.Ani Melani Maskoen (Universitas Padjadjaran, Indonesia)
PB-25	***Complications in adult thalassemia patients due to iron overload*** Dr. Pandji Irani Fianza (Universitas Padjadjaran, Indonesia)

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ABSTRACT OF ORAL PRESENTATION

LP-01-01

Development of albumin glycation detection platforms for diabetes mellitus monitoring

Deanpen Japrung*, Chayachon Apiwat, Kiatnida Treerattrakoon and Wireeya Chawjiraphan

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ABSTRACT

Aptamers are short ssDNA or RNA that specifically bind to target molecule using three-dimensional structure. Their target molecules could be cells, proteins, metal ions, and toxin. Aptamers are more stable and easy to produce compared with the antibody and can be selected from the large aptamer library using the method called "Systematic Evolution of Ligands by Exponential Enrichment" or "SELEX". Our group have selected and modified DNA aptamers specifically bound human serum albumin (HSA) and glycated human serum albumin (GHSA), which is a marker for non-communicable diseases such as diabetes mellitus, kidney dysfunction and Alzheimer. In this study, three sensor platforms, which are electrochemical, nanopore and graphene-based aptasensor have been developed for diabetes mellitus detection. The fluorescent quenching graphene oxide (GO) and Cy5-labeled aptamers could be used for GHSA and HSA detection in both human serum and urine samples (both DM patients and normal group). These indicate that our aptasensor has a potential for diagnosis and monitoring of diabetes mellitus.

Keywords: Aptamer; diabetes mellitus; glycated human serum albumin; aptasensor

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ABSTRACT OF ORAL PRESENTATION

LP-01-02

The role of interleukin-8-immunomodulatory pathway in the differentiation of stem cells from human exfoliated deciduous teeth into odontoblast-like cells

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ABSTRACT

Interleukin-8 (IL-8), a pro-inflammatory cytokine, has been reported to be implicated in odontogenesis and involved in odontoblast-mediated immune responses but the exact mechanism remains unclear. Hence, our group conducted a study to investigate the role and mechanism of IL-8 in the differentiation of stem cells from human exfoliated deciduous teeth (SHED) into odontoblast-like cells. SHED were seeded on human amniotic membrane (HAM) and treated with bone morphogenetic protein 2 (BMP-2), and following treatment, SHED were harvested on day 1, 7, 10, and 14. Odontogenic differentiation potential was assessed by the expression of odontogenic markers using reverse transcriptase PCR, and calcium deposition was analysed by Alizarin Red S staining. Thereafter, the optimal concentrations of reparixin, an IL-8 inhibitor, and recombinant human IL-8 (rhIL-8), an IL-8 inductor, were determined using Real Time PCR. The effects of IL-8 inhibition and induction were then analysed using Real-Time PCR and Western blotting. The levels of IL-8 protein secretion of SHED with and without IL-8 induction and inhibition during odontogenic differentiation were analysed using ELISA, while the effect of IL-8 in calcium deposition of SHED was determined using Alizarin red S staining. The results of our present study showed that odontoblast specific markers DSPP, DMP1,and OPN were highly expressed on day 7 onwards as odontogenic differentiation occurred. For IL-8 downstream pathway analysis, PI3K/AKT/mTOR and JAK2/STAT3 signalling pathways were suggested to be involved in odontogenic differentiation as the expression of all the markers were high, whereas inhibition of IL-8 using reparixin caused significant reduction of their expression. In conclusion, our study suggests that inhibition of IL-8 receptor by reparixin promotes odontogenic differentiation of SHED when cultured on HAM and treated with BMP-2.

Keywords: Interleukin-8 (IL-8); SHED; odontogenic differentiation; reparixin

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ABSTRACT OF ORAL PRESENTATION

OP-01-01

Compressive strength, bioactivity and biocompatibility properties of chitosan-coated carbonate apatite scaffold enriched by gentamicin drug

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ABSTRACT

Chitosan-coated carbonate apatite (CA) scaffold -with and -without antibiotic enrichment were investigated for their compressive strength, biodegradability and cytotoxicity. Three different scaffolds which were chitosan-coated carbonate apatite, chitosan-coated carbonate apatite enriched by gentamicin loaded poly-lactic acid (PLA) microsphere and chitosan-coated carbonate apatite enriched by gentamicin, denoted as CS-CA, CS-GENMS-CA and CSGEN-CA, respectively were fabricated through the dipping and infiltration methods. Neat CA and neat microsphere were used as control for comparison on compressive strength, biodegradability and drug release behaviour. Biodegradability of scaffold in Hank's Balance Salt Solution (HBSS) was characterized by weight loss whereas apatite mineralization on scaffold surface was examined using Scanning Electron Microscope (SEM). CS-GENMS-CA showed the increasing of 3-fold compressive strength than neat CA scaffold and also has lower degradation rate among other coated scaffolds. Apatite formation on CS-GENMS-CA represented the presence of gentamicin-loaded PLA microsphere, not hindered the mineralization process for 28-day immersion in HBSS. Infiltration of gentamicin into the coated scaffold, prolonged the effective antibiotic release for up to 28 days. Cell proliferation using Presto Blue assay was observed after 7-day incubation, indicated that all scaffolds cause no cytotoxicity effect towards human osteoblast cell (hFOB). CS-GENMS-CS enhanced more cell proliferation, followed by CSGEN-CA and CS-CA. These results showed that the coated CA with gentamicin has contributed to the favour of cell growth. Thus, suggesting CS-GENMS-CA which exhibited good cell proliferation, sustained drug release and improvement of compressive strength; is a potential candidate for use in bone tissue engineering.

Keywords: Carbonate apatite scaffold; biodegradation; compression; proliferation; drug release

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ABSTRACT OF ORAL PRESENTATION

OP-01-02

Anti-cancer activity of Luvunga scandens extracts against human malignant melanoma cells

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ABSTRACT

Human malignant melanoma cells are characterized by abnormalities in cell differentiation and growth. Skin cancer is reported as one of the most common types of cancer with increasing numbers of occurrence. Luvunga scandens (L. scandens) is one of the medicinal plants found in Malaysia. This plant is known to possess many bioactivities and general health effects, yet its anti-proliferative effect is generally under reported and need to be scientifically evaluated. The aim of this study is to evaluate the anti-proliferative and apoptotic effects of L. scandens plant extract against human malignant melanoma cell line. 3-(4,5-dimethylthiazol-2-yl)-2,5-diphenyltetrazolium bromide (MTT assay) was used to test the cytotoxicity effect caused by L. scandens on malignant melanoma cells, in addition to measuring the safety levels of L. scandens against normal cell lines (HaCaT and HDF). Scratch assay was carried out to monitor the cell growth. The morphological changes of L. scandens treated malignant melanoma cells was confirmed by scanning electron microscopy, and the apoptotic effect of the plant against malignant melanoma cells was tested using caspase 3/7 assay, followed by cell cycle analysis done using a flow-cytometer on skin cancer cells treated L. scandens plant. The results showed that the extracts (Methanol, Dichloromethane, Hexane) possess cytotoxic effect against skin cancer cells, and no cytotoxic activity on both HaCaT and HDF cells. The scanning electron microscopy results demonstrate that L. scandens treated cells showed overall changes in the cell shape, alteration of surface morphology, absence of microvilli and appearance of blebs. Caspase 3/7 assay results showed that L. scandens dichloromethane (DCM) extract exhibited the highest level of apoptosis against malignant melanoma cells. For cell cycle analysis, the L. scandens treated malignant melanoma cells show high readings in the sub-G1 phase. This in vitro study has proven that L. scandens plant extract exhibit anti-proliferative effects against human malignant melanoma cell line, hence, it can be considered as a new promising potential anti-cancer therapy.

Keywords: Luvunga scandens; MTT; SEM

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ABSTRACT OF ORAL PRESENTATION

OP-01-03

Development of SERS based biosensor for cancer screening

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ABSTRACT

The demand for specific and accurate cancer screening has driven the development of novel diagnostic platforms having high selectivity and sensitivity. Surface-enhanced Raman scattering (SERS) based biosensors have been continuously developed as new biodiagnostic tools for highly sensitive molecular and cellular detection due to advantages in the richness of spectroscopic information together with high spatial resolution. The key to accomplishing highly effective SERS-based detection in bioanalysis depends on the strong electromagnetic enhancement of SERS substrate and probe-target interactions. To achieve high signal enhancement, Raman active molecules need to be in close proximity to roughened metallic surface in nanometer-scale. Gold nanorods (GNRs) have been utilized for SERS tags fabrication to detect biomolecules such as cells, proteins, and oligonucleotides, due to their strong light absorption and scattering. To realize targeting property, DNA probes can be attached to GNRs surface to provide specific cells and nucleic acid detection. Therefore, the selectivity and sensitivity of the detection could be achieved by the specificity of DNA probes towards their targets and the strong detectable signal from SERS technique.

Keywords: Surface enhanced Raman scattering (SERS); gold nanorod; biosensor; cancer screening

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ABSTRACT OF ORAL PRESENTATION

OP-01-04

Osteogenic performance of MC3T3-E1 cells on granular hydroxyapatite scaffold

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ABSTRACT

The initial seeding density plays a crucial role in determining the effectiveness of bone tissue regeneration utilising scaffold. One of the factors that influence cell osteoblast differentiation is cell seeding related factor. Therefore, this study aims to determine the effect of different initial seeding densities on the osteoblast performance of MC3T3-E1 cells in hydroxyapatite granular scaffold. MC3T3-E1 cells at different seeding density (5x105 and 1x106 cells/cm2) were cultured in the 2-dimensional flask and 3-dimensional hydroxyapatite scaffold. Cells in both cultures were analysed in term of morphology using CellB software while biochemical activity was assessed using Alkaline Phosphatase (ALP) assay. MC3T3-E1 showed a mononucleated, fibroblast-like shape cell with extended cytoplasmic projection. Cell seeding density of 1x106 cells/cm2 reached 90% confluency while 5x105 cells/cm2 reached 70% confluency on day six of culture. Variations in cell seeding density significantly influenced the cell osteodifferentiation. During 2-Dimensional osteoblast differentiation, lower initial seeding density resulted in higher ALP activity. However, cells were seeded on hydroxyapatite scaffold, by increasing the seeding density from 5x105 to 1x106 cells/cm2 resulted in decreased of ALP activity. This study has shown that the seeded cell population in the 3-dimensional hydroxyapatite scaffolds clearly affected the degree of osteoblast cell differentiation in which a higher seeding density was not necessarily better. The seeding density played a major role in influencing the corresponding cell differentiation.

Keywords: Seeding density; osteoblast; Granumas®; alkaline phosphatase

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ABSTRACT OF ORAL PRESENTATION

OP-01-05

Mode of action of cinnamon bark (Cinnamomum verum) essential oil and the combinatory bactericidal activity with meropenem against KPC-producing Klebsiella pneumoniae

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ABSTRACT

Antimicrobial resistance is an ongoing challenge in the clinical setting at present. To address this issue, combinatorial therapies have been applied in the clinical setting to tackle multi-drug resistant bacterial infections and these have frequently proven to be effective. In addition to the wider acceptance amongst the general public for nature-based products, the mining of novel antimicrobials is moving towards the direction of greener plant-based compounds from synthetic chemical compounds such as essential oils. Thus, this study was undertaken to look at the combinatory effects of cinnamon bark essential oil (CBO) and meropenem against KPC-producing K. pneumoniae. We found that CBO had a relatively low MIC of 0.16% (v/v) when used against KPC-producing K. pneumoniae. When used in combination, the MIC is further reduced to 0.08% (v/v). Furthermore, we also found that mode of action of CBO involved the disruption of the bacterial membrane, as determined in the zeta potential measurement, outer membrane permeability assay and scanning electron microscopy. Comparative proteomic via label free LC-MS/MS had also been performed on the non-treated and CBO-treated KPC-KP cells which further validated the disruption in the bacterial membrane as well as in the membrane and cell wall- repairing mechanism. Hence, results obtained from our studies strongly suggests that CBO may potentially play a promising role with regards to the mining of novel antimicrobial compounds, which inevitably may help eradicate antimicrobial resistance infections in the clinical setting.

Keywords: Cinnamon bark essential oil; combinatorial therapy; comparative proteomic; label free LC-MS/MS; membrane disruption.

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ABSTRACT OF ORAL PRESENTATION

OP-01-06

2-Methoxy-1,4-Naphthoquinone (MNQ) inhibits glycolytic activities in breast cancer cell lines

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ABSTRACT

2-Methoxyl-1,4-Naphthoquinone (MNQ) is one of the quinones that can be extracted from garden balsam (Impatiens balsamina). MNQ was reported to induce apoptosis, increase DNA damage by reactive oxygen species (ROS) production and suppress the cells' invasion and migration in several cancer cell lines, including highly metastatic breast cancer cells MDA-MB-231. It is known that cancer cells exhibit high levels of glycolysis as well as lactate production which resulted in metastatic activity. This study was conducted to investigate the effect of MNQ on glycolytic activities in MDA-MB-231 cells. Initially, the cells viability were tested at various doses of MNQ (5-100 μM) using MTT proliferation assay. As the half maximal inhibitory concentration () was obtained, the cells were tested with dose of MNQ for glucose uptake and lactate assays. The results showed, MNQ decreased the percentage of MDA-MB-231 cell viability in a dose-dependent manner with value of 43 μM/ml. The percentage of glucose uptake into the cells and lactate production decreased significantly after treatment with the MNQ as compared to untreated controls. Our findings indicated the ability of MNQ to inhibit the glycolytic activities in MDA-MB-231 cells, suggested the potential of MNQ to be further developed as an effective agent/adjuvant against highly metastatic breast cancer.

Keywords: 2-Methoxy-1,4-Naphthoquinone (MNQ); glucose metabolism; glycolytic activity; breast cancer cells; MDA-MB-231 cells

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ABSTRACT OF ORAL PRESENTATION

OP-01-07

The effect of Singawalang (Petiveria alliaceae) leaves extract on the blood cholesterol level, profile of liver and blood vessel in Hypercolesterolemic rats

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ABSTRACT

This study has been conducted to determine the effects of Singawalang (Petiveria alliaceae) leaves extract on cholesterol blood level, profile of liver and blood vessel in hypercholesterolemic rats. The research is essential to attain a scientific evidence, for the use of Singawalang leaves extract, as treatment for hypercholesterolemia. Twenty-five hypercholesterolemic rats were randomly assigned into five different groups. Group 1 received the drug vehicle, group 2 were treated with simvastatin at a dose of 0.18mg / head / day/ orally. Whereas groups 3, 4, and 5 were treated with the Singawalang leaves extract at doses of 90 mg / kg bw; 180 mg / kg bw and 360 mg / kg bw respectively. The treatment was administered once-a-day, orally for 14 days. On the 15th day, those rats were sacrificed for organs and blood investigations. The results showed a decrease in cholesterol levels in all groups following the treatment. Differences in cholesterol levels before and after treatment were significant ($p = 0.000$; $p<0.05$). In addition, there were no improvement in the profile of liver and blood vessel following the treatment. This research concluded that the extract of Singawalang leaves decreased total cholesterol level in hypercholesterolemic rats.

Keywords: Petiveria aliacea; hypercholesterolemic; liver; blood vessel

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ABSTRACT OF ORAL PRESENTATION

OP-01-08

Tualang honey and its methanolic fraction protect against LPS-induced neuroinflammation and amyloid deposition in male rats

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ABSTRACT

Tualang honey (TH) exhibits a number of pharmacological actions including antioxidant, anti-inflammatory, anticonvulsant and cognitive enhancer. The purpose of this study is to investigate the effects of TH and its methanolic fraction (MTH) on the expression of proinflammatory cytokines and amyloid deposition in the hippocampus of systemic lipopolysaccharide (LPS)-injected rats. Sixty male Sprague Dawley rats were divided into 5 groups (n=12): (i) control rats, (ii) untreated LPS rats (5 mg/kg) (iii) LPS rats treated with TH 200 mg/kg, (iv) LPS rats treated with MTH 150 mg/kg and (v) LPS rats treated with memantine 10 mg/kg. All treatments were administered intraperitoneally once daily for 14 days. The rats were sacrificed and hippocampal tissues were carefully dissected out. Determination of proinflammatory cytokines expression and amyloid deposition in the hippocampus were carried out by immunohistochemistry staining method and ELISA, respectively. The COX-2 and TNF-α expression, as well as amyloid deposition, were highest in untreated LPS rats compared to other experimental groups. TH and MTH significantly reduced the concentration of COX-2 and TNF-α expression, as well as amyloid deposition in the hippocampus of LPS rats comparable to memantine group. In conclusion, the findings suggest that TH and MTH protect the hippocampus against LPS-induced neuroinflammation and amyloid deposition as effective as memantine.

Keywords: Tualang honey; lipopolysaccharide; amyloid deposition; proinflammatory cytokines

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ABSTRACT OF ORAL PRESENTATION

OP-01-09

Application of malva nut gum as bioreductor and stabilizer on gold nanoparticle synthesis: Preparation of Chikungunya immunoassay biosensor

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ABSTRACT

Gold nanoparticles can be used as detectors in various diagnostics and therapeutics of diseases. Currently, we are developing a diagnostic kit for Chikungunya virus by using gold nanoparticle as biosensor on immunoassay. One of the preliminary steps of the research is to synthesize gold nanoparticles by natural product as bioreductor. Green-gold nanoparticle was chosen as the candidate of biosensor because of its low toxicity. Gold nanoparticle was produced by using malva nut gum (Scaphium macropodum (Miq.) Buem.) as bioreductor and biostabilizer. Tetrachloroauric acid ($HAuCl_4$) solution was mixed with malva nut gum; and stirred until the gold nanoparticle's colour turn into red. The optimum reaction time and concentration of malva nut gum as bioreductor were 4 hours and 3% w/v, respectively. Moreover, the product was stable for 23 days. It was found that the speed of stirring of synthesis also influenced the particle size. Particle size ranged from 74-106 nm was produced by 500 rpm of stirring for 4 hours. The product was characterized by spectrophotometer UV-Vis, SEM / EDS, and FTIR.

Keywords: Malva nut gum; bioreductor; biostabilizer; gold nanoparticle; Chikungunya

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ABSTRACT OF ORAL PRESENTATION

OP-01-10

NFAT molecular profiles of VEGF-induced-dental-stem-cells cultured on human amniotic membrane

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ABSTRACT

Nuclear factor of activated T-cells (NFAT) signalling pathway involved in endothelial differentiation have been described in the literature. However, the information about the involvement of NFAT pathway on the stem-cell-endothelial differentiation is limited. Thus, we investigated the NFAT pathway using an in-vitro 3D-construct. The construct that mimicked the endothelial-induced environment were made of extracellular matrix (ECM)-rich scaffold, human amniotic membrane (HAM) and pro-angiogenic growth factor, vascular endothelial growth factor (VEGF). In this study, stem cells from human exfoliated deciduous teeth (SHED) were cultured on HAM with an addition of VEGF. Cyclosporine A (CsA) inhibitor was used to inhibit the NFAT pathway. The expression level of genes associated with the NFAT signalling pathway was quantified using One-Step Real-Time PCR. SHED induced with VEGF (SHEDi) acted as a control group, while the treatment groups were SHEDi cultured on HAM (SA), SHEDi cultured on HAM treated with VEGF (SAV), and SHEDi cultured on HAM treated with VEGF and CsA (SAVC). The result showed that CsA boosted the gene expression of all gene markers (Cox-2, IL-8 and RCAN1.4) at day 1, but not ICAM-1. While at days 7 and 14, IL-8, ICAM-1 and RCAN1.4 were down-regulated by CsA inhibitor. The cell morphology was undetermined, but the presence of filament-like structures could be seen through an inverted microscope. This study provides an insight on the effect of angiogenic microenvironment contributed by HAM scaffold on SHED-endothelial differentiation. Besides, this study accomplishes in showing the involvement of the NFAT pathway of the current in-vitro 3D-construct.

Keywords: Endothelial gene markers; human amniotic membrane; nuclear factor of activated T-cells pathway; stem cells from human exfoliated deciduous teeth; vascular endothelial growth factor

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ABSTRACT OF ORAL PRESENTATION

OP-01-11

DNMT1 is expressed in actively-proliferating lymphoid blast cells and significantly associated with E2F gene set in malignancies

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ABSTRACT

DNA methyltransferase 1 (DNMT1) is a maintenance methyltransferase crucial for cellular proliferation and cell cycle activation. DNMT1 is required for the formation of follicular germinal centre (GC) structure and B-cell differentiation into plasma cells. However, little is known on the expression profile and functions of DNMT1 in B cells residing in the non-GC areas of interfollicular regions. In this study, utilising a validated anti-DNMT1 monoclonal antibody (clone 2B5), we demonstrated that in all human lymphoid tissues investigated, DNMT1 was negative in plasma cells but highly expressed in centroblasts residing in the GC region with a number of DNMT1-positive cells residing in the non-GC interfollicular regions. We hypothesised that these cells might be immunoblasts, a type of lymphoid cells that directly transform into plasma cells independent of the GC structure, due to its cellular features i.e. large cells containing round nucleus and large nucleolus. Thus, we proceeded to investigate whether the following immune cells in the interfollicular non-GC regions expressed DNMT1 through double immunostaining: T cells (CD3+), immunoblasts (CD30+), natural killer cells (CD56+), macrophages (CD68+) and plasma cells (CD138+). DNMT1 was absent in all these populations except CD30+ immunoblasts. We have previously shown that DNMT1 was frequently expressed in diffuse large B-cell lymphoma (DLBCL), whose cellular origin was thought to arise from mature B cells (e.g. centroblasts, immunoblasts, plasma cells), and associated with high Ki-67 (proliferation marker) expression. Both centroblast and immunoblast cell populations are actively proliferating cells. To elucidate these fundamental roles of DNMT1 further (i.e. proliferation and cell cycle activation), we examined DNMT1 transcript expression profile in 31 different types of cancers where it was most highly expressed in DLBCL followed by other malignancies. We subsequently performed Gene Set Enrichment Analysis (GSEA) on the top 15 cancers with the highest DNMT1 transcript values, and showed that the "E2F targets" gene set involved in cell cycle activation were most significantly enriched ($p<0.05$; FDR<0.01) in all 15 cancer types. Taken together, these suggest that the fundamental roles of DNMT1 in cellular proliferation and cell cycle activation occur in both normal and malignant cell types.

Keywords: DNMT1; immunoblast; diffuse large B-cell lymphoma; proliferation; cell cycle

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ABSTRACT OF ORAL PRESENTATION

OP-01-12

Establishing Fragile X Research Center: An Experience from Developing Country, Indonesia

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ABSTRACT

Fragile X syndrome (FXS) is the most common cause of monogenic inherited intellectual disabilities (ID) with X-linked pattern of inheritance. The prevalence of ID in developing countries was calculated as much as 3%, of which genetic was the underlying mechanism in about 20% of the cases. In Indonesia, fragile X syndrome accounted for 2% of the ID population. Center for Biomedical Research (CEBIOR), Faculty of Medicine Diponegoro University is the only Fragile X Research Center in Indonesia located in Semarang, Central Java, is the only center that offered FXS molecular diagnosis. Based on the number of the Indonesian population, 28,000 cases of FXS should be diagnosed in our center. However, the phenotype of FXS is less obvious when compared to Down syndrome (the most common cause of genetic ID) resulting in indistinguishable FXS. Coupled with the fact that the knowledge of FXS in healthcare providers concerning several aspects of FXS is yet to be fully understood, hence the awareness of FXS is unsatisfactory. Another obstacle that may have caused low detection rate is due to the lack of attention in genetic disorders from the government. In Indonesia, infectious disease is still the most important issue for national insurance health coverage followed by metabolic syndrome and cancer which may yield a direct impact on the mortality rate. FXS screening program based on research in high risk ID population had been done using various techniques, ranging from conventional cytogenetics to advanced molecular analysis in our center. Concerted effort is needed to improve the general awareness of the syndrome and encouragement from the government may shift national insurance health coverage in genetic disorders to be more favorable.

Keywords: Fragile X syndrome; Indonesia

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ABSTRACT OF ORAL PRESENTATION

OP-01-13

Personalised genomics for healthy lifestyle and wellness

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ABSTRACT

Our genome is a roadmap to health and wellness. Human genomics data demonstrated a significant impact in the diagnosis of a wide range of diseases or treatment for specific types of cancer. Advances in genomics and bioinformatics technologies have given us the opportunity to peer into our genetic futures in ways we might never have imagined. The present work illustrates the current sequencing- and genotyping-based services offered by commercial providers to generate personal genome and wellness profiles. The services offered fall into various categories namely fitness, health and performance; allergy and healthy diet; side-effects of some drugs; talent and skills as well as susceptibility to non-inherited life-threatening diseases. Upon submission of an online order to a service provider, a Saliva Collection Kit will be posted directly to the consumer. The kit offers an easy, convenient and safe method for the collection, stabilization, transportation, and storage of saliva samples. This direct-to-consumer (DTC) service use cells found in the saliva to obtain the required DNA for analysis. Each customer will finally receive a personalised report with straightforward, genetically tailored recommendations for optimizing their lifestyle and well-being. DNAKU is a local initiative by the Malaysia Genome Institute (MGI) of the National Institutes of Biotechnology Malaysia providing services for Malaysian newborns, children, and adults. This initiative utilises high throughput, next-generation genotyping array and an in-house built bioinformatics pipeline for a large scale data analysis. This presentation will also cover on the challenges and possible solutions regarding wellness and lifestyle genomics including the lack of accuracy and clarity of information over which specific genes or variants are being tested; consent, privacy and ownership of the personal genomic data; lack of universally accepted guidelines and legislation; availability of extensive genetic counseling and psychological support to families for the interpretation of the genetic data and availability of interventions that could be initiated as early as possible that would reduce morbidity or mortality. Genomic information can be a powerful tool and would allow consumers to make informed health decisions and personal lifestyle choices that will improve their quality of life.

Keywords: Genomics; bioinformatics; healthy lifestyle; wellness; DNAKU

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In conjunction with
2nd Conference of Malaysian Society of Human Genetics

ABSTRACT OF ORAL PRESENTATION

LP-02-01

miRNA as a promising specific bio marker for ovarian cancer and targeted therapy

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ABSTRACT

Epithelial ovarian cancer (EOC) accounts for 25% of all malignancies in women and considered the most lethal gynecological malignancy, accounting for 4.2 % of all cancer-related deaths in women. Most EOC patients are diagnosed at late stages, leaving little chance for survival due to the lack of effective treatments. During the past century, incidence of EOC has been slowly yet steadily increasing, while development of more effective treatment has lagged behind, leading to little, if not none, improvement in overall survival. Current standard treatment for EOC includes a combination of surgical resection and chemotherapy, which acts efficiently as initial treatment. However, most EOC patients recur after a few years and turn to be resistant to existing treatments. Despite the use of aggressive treatment, recurrence is frequently seen among EOC patients, and cancerous metastasis is one of the predominant causes of mortality. Therefore, exploration of novel biomarkers for early diagnosis, prognosis prediction, and effective therapies will definitely contribute to current EOC treatment and management. Recently, noncoding RNA molecules, microRNAs (miRNAs) are drawing a lot of attention for both physiological and pathological processes because of its stability, differ from RNA that is very fragile. By imperfect complementary sequence pairing between miRNA seed region and the 3'-untranslated region (UTR) of target genes, miRNAs negatively regulate target genes by either mRNA degradation or translational repression, thus directly or indirectly affecting almost all cellular pathways. Our group have analyzed the profiles of ovary cancer using Nano string. Almost 800 miRNA genes were analyzed, it showed the expression profiles of 10 most upregulated and 1 most down regulated. For the predicted signaling pathways, we have had validation on several miRNA from plasma of Ovary cancer for several oncomiR and tumor suppressor. The targeted development using chitosan to develop Ago-miR and mimicking miRNA are underway to be tested in in vitro study SKOV3 cell lines.

Keywords: ca ovary; miRNA expression profile; targeted therapy

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ABSTRACT OF ORAL PRESENTATIONS

LP-02-02

High resolution single nucleotide polymorphism analysis of genomic aberrations in childhood acute lymphoblastic leukaemia

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ABSTRACT

Leukaemia is the sixth most frequent cancer in Malaysia and the commonest cancer in children. Acute Lymphoblastic Leukaemia (ALL) is one of the major causes of death in children. Acquired chromosomal abnormalities are the hallmark of ALL, which define biologically distinct subtypes of the disease. The strong association of many chromosomal abnormalities and prognosis has been utilized in risk stratification for treatment in a large number of protocols worldwide. High-resolution single nucleotide polymorphism (SNP) 6.0 array analysis was carried out on 55 Malaysian childhood precursor B-ALL (BCP-ALL) patients diagnosed between 2016 and 2017. The raw data was analyzed using Genotyping Console Software v4.2.0.26. SNP array results were validated using Multiplex Ligation-dependent Probe Amplification (MLPA) and Fluorescence in situ Hybridization (FISH). Our objective was systematically characterize genomic aberrations in childhood BCP-ALL among Malaysian populations. SNP array analysis revealed 31 copy number variant regions within the 55 samples. The most frequent copy number gains were on chromosome regions 22q11.22 (91%), 2p11.2 (64%), 15q11.2 (64%), 14q11.2 (51%) and Xq21.31 (44%) and the most frequent copy number losses were 4q13.2 (51%), 8p11.2 (33%) and 3q26.1 (31%). Gain of 22q11.22 and loss of 4q13.2 were the most frequent alterations found in this study. The recurrently targeted copy number abnormalities involved several leukaemia-related genes-CDKN2A/B, MLL, IKZF1, PAX5, RUNX1, ERG, CRLF2, SHOX, CSFR2A, BTG3 and ETV6. We identified several new recurrent aberrations with possible new target genes: Gain of 22q11.22 and loss of 4q13.2 were the most frequent alterations found in this study. These chromosomal regions contain genes such as POM121L1P, IGLL5 and UGT2B17. These potential genes may contribute to the leukaemogenesis in childhood BCP-ALL. Integrating the findings of this study with the clinical, cytogenetic and molecular biology data would allow us to propose new strategies that would improve the diagnosis, prognostication and treatment of Malaysian childhood BCP-ALL patients.

Keywords: BCP-ALL; SNP; MLPA; FISH.

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ABSTRACT OF ORAL PRESENTATIONS

OP-02-01

Ultrasensitive detection of circulating miRNA by branched-rolling circle amplification coupled with graphene fluorescence-based sensor

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ABSTRACT

In this study, we developed an isothermal microRNAs (miRNAs) detection platform based on high efficient branched-rolling circle amplification (BRCA) system coupled with graphene oxide (GO)-based fluorescence assay. In the strategy, cancer-associated miR-21 was selected as a model target. Target miRNA complementary binds a linear single-stranded DNA (ssDNA) probe and turns to circularized conformation under reaction containing T4 RNAligase2. The BRCA products were amplified after adding secondary primers in the present of Phi29 DNA polymerase, then quantified by measuring fluorescent level after adding GO-sensing complexes. The limit of detection of our developed platform was 0.87 fM, which is lower than that of the original BRCA method. Our platform also can discriminate a single mismatch miRNA mutant and can be applied to determine amounts of target miRNA in total RNA extracts from cancer cell lines. These indicate that our platform has a potential for miRNA detection in clinical diagnosis.

Keywords: MicroRNA; graphene oxide; branched-rolling circle amplification; isothermal amplification

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ABSTRACT OF ORAL PRESENTATIONS

OP-02-02

Bioinformatics analysis of differentially expressed genes in liver cancer for identification of key genes and pathways

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ABSTRACT

Liver cancer is among the main leading cause of mortality in Malaysia and the world. To identify the key genes and pathways in liver cancer, mRNA microarray dataset GSE84402, GSE64041, GSE60502, GSE29271 and GSE19665 were downloaded from the Gene Expression Omnibus database. Differentially expressed genes (DEG)s from all datasets were obtained by GEO2R web tool and assembled manually with the cut-off criteria set at adj. $P < 0.01$ and $|logFC| > 1$. Functional and pathway enrichment analysis were performed for the DEGs using DAVID database. STRING database was utilised to construct a protein-protein interaction network. Cytoscape 3.6 software and associated plug-ins were used to visualise and analyse protein-protein interaction (PPI) network. A total of 681, 1564, 1040 and 2265 DEGs were identified from GSE84402, GSE64041, GSE60502, GSE29271 and GSE19665 datasets, respectively. 184 DEGs were screened out in at least four datasets consisting of 70 up-regulated genes and 114 down-regulated genes. These genes were mainly enriched in the terms related to mitotic cell cycle and regulation of nuclear division. Putative PPI network was established with confidence score 0.7 comprising 184 nodes and 1021 edges. Using MCODE plug-in, four modules were detected from the PPI network with module 1 being the most significant. The enriched functions of module 1 included sister chromatid cohesion and mitotic spindle assembly checkpoint. In conclusion, these results identified the key genes and pathways, which could improve understanding of the molecular mechanisms and provide potential targets for liver cancer diagnosis and treatment.

Keywords: bioinformatics; liver cancer; differentially expressed genes; functional enrichment analysis; protein–protein interaction

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ABSTRACT OF ORAL PRESENTATIONS

OP-02-03

Identification of MAGE A1-A10 mRNA from testicular tissue using the common primer for RT-PCR

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ABSTRACT

One of the cancer that expressed the MAGE A mRNA is lung cancer. The specimen for diagnose of lung cancer are usually just a few of tissue from core biopsy, forcep biopsy, Fine needle aspiration biopsy, or bronchoalveolar lavage. The melanoma antigen (MAGE) gene is the only gene that is expressed in cancer cells and in testicular tissue. The MAGE A gene is consist of 12 variants called MAGE A1, A2, A3, A4, A5, A6, A7 (pseudo gene), A8, A9, A10, A11, and A12. The MAGE A mRNA can be detected using the Reverse Transcription-Polymerase Chain Reaction (RT-PCR), but it is required a common primer for detecting the most of MAGE A variants. The objective of this study was to develop the common primer for detecting the MAGE A1-A10 mRNA. The specimen used in this research was testicular tissue taken from patients who had received the orchidectomy therapy at Dr Soetomo Hospital, Surabaya, Indonesia, in 2017. Detection of MAGE A1-A10 mRNA was performed by RT-PCR technique using the common primer for MAGE A1-A10. MAGE A1, A2, A3, A4, A5, A6, A8, A9, and A10 variants mRNA were detected using primer set of GMF10/GMR10 for the first round and primer set of GMF10/GMR12 for the second round. Primer set of MMRP1/MMRP2 was used as a comparison for MAGE A1-6 mRNA identification. The mRNA from testicular tissue was extracted, followed by RT-PCR. The PCR products were analysed by 2% agarose gel electrophoresis. Result showed that the PCR products of primer pair GMF10/GMR10 ranges from 823-919 base pair (bp) whereas products of GMF10/GMR12 were 461-557bp in size depending on the variants of MAGE A1-A10. The next optimization test was RNA total dilution test. A total of 133.4 ng/ml of RNA was used, and was diluted with a ratio of 1:10, 1:100, 1:1000. All of the MAGE A1-A10 mRNA can be detected together in the same tube PCR. This common primer can be used as a biomolecular tool for identification of all variants of MAGE A1-A10 mRNA in cancer cells.

Keywords: MAGE A1-A10 mRNA; common primer; testicular tissue; cancer testis antigen; RT-PCR

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ABSTRACT OF ORAL PRESENTATIONS

OP-02-04

Identification and characterization of the VPE gene family and its expression in response to Fusarium oxysporum infection in Musa acuminata

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ABSTRACT

Panama disease, caused by Fusarium oxysporum f. sp. Cubense tropical race 4 (Foc TR4) has been the major threat for global banana production. The only effective measure to counteract the disease is by planting edible resistant cultivars which only limited to resistant Cavendish banana generated through somaclonal variation. Hence, understanding on the molecular events during Foc infection are crucial in order to develop protection strategies against panama disease. This research was undertaken to study the molecular characteristic of Musa acuminata vacuolar processing enzyme (MaVPE) – a cysteine proteinase that mediates programmed cell death; during FocTR4 infection. A total of seven MaVPE genes (designated as MaVPE1 through MaVPE7) were successfully identified through systemic in-silico analysis of DH-Pahang (AA group) banana genome. Phylogenetic study and tissues specific analysis showed that MaVPEs could be divided into seed type or vegetative type. Real-time quantitative polymerase chain reaction (RT-qPCR) further revealed that most of MaVPE genes expression was induced after FocTR4 infection, specifically at 24 and 48 hours post inoculation (hpi). Moreover, induction of caspase-1 activity through caspase assay studies and increase of MaVPE expression through comparative proteomic analysis were also detected after inoculation with FocTR4. Consistently, inhibition of MaVPE activity through caspase-1 inhibitor reduced vacuolar membrane disintegration and decreased lesion formation of FocTR4 infected banana root. Further functional analysis in Arabidopsis VPE-null mutant, exhibited higher tolerance to FocTR4 infections and decreased cell death incidence. Taken together, our findings suggest that VPE act as a key molecule in modulating susceptibility response in FocTR4-infected plant.

Keywords: Musa acuminata (M. acuminata); Fusarium oxysporum f. sp. Cubense (Foc); vacuolar processing enzyme (VPE); programmed cell death (PCD); panama disease.

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ABSTRACT OF ORAL PRESENTATIONS

OP-02-05

Dopamine receptors (DRD4 and DRD5) mRNA expression in peripheral blood lymphocytes of healthy Malay men subjects

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ABSTRACT

Evidence suggests that dopamine systems peripherally reflect central dopamine systems activity. To recent, studies have been carried out widely on central dopamine systems, while investigation of dopamine system in various organs peripherally is still little. In this study we investigated dopamine receptors (DRD4 and DRD5) mRNA expression in peripheral blood lymphocytes of healthy Malay men subjects. Blood samples were collected from 40 healthy Malay men subjects. Lymphocyte was isolated from whole blood using isolation media, followed by RNA extraction and cDNA synthesis using commercially available kits. The mRNA expression of DRD4 and DRD5 were assessed by RT-PCR using specific primers for dopamine receptors mRNAs and β-actin as internal control. Descriptive statistic was applied for data analysis. Results showed that both DRD4 and DRD5 mRNAs were expressed in peripheral blood lymphocyte of healthy Malay men subjects. Even though the conclusive function of these receptors is still being investigated, these results may suggest that the peripheral dopamine systems might play important tools to reflect central dopamine physiology, pathology and pharmacologically.

Keywords: Dopamine receptors; mRNA Expression; lymphocytes; RT-PCR

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ABSTRACT OF ORAL PRESENTATIONS

OP-02-06

Gene expression analysis of stem cell migration in local angiogenesis for tissue repair

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ABSTRACT

Dental stem cells offer great potential benefit for application in stem cell-based therapy. Stem cells from exfoliated deciduous teeth (SHED) exhibit mesenchymal stem cell characteristics, with the ability to form various cell types of different lineages. Migration of endothelial cells plays a critical role in angiogenesis, to further support blood vessel formation during tissue repair and regeneration. However, the exact cellular characteristics of SHED undergoing angiogenesis, in term of its migratory capacity and gene expression pattern have not been fully understood. Hence, this study aims to assess the differential gene expression of SHED following angiogenesis and migratory induction. SHED migration capacity under different seeding density, and between undifferentiated state and the angiogenically induced group are investigated. Briefly, in-vitro cultured SHED was induced to form endothelial cells by supplementation of angiogenic factors. Scratch test assay was used to estimate the rate of cell migration, whereas transwell migration assay was performed to collect RNA samples on day 1,3,7,10 and 14. Following RNA extraction, the samples were further analysed by RT-PCR for detection of stem cell markers, migration markers and angiogenic markers. The gathered findings indicated that SHED is highly capable of forming endothelial cells. SHED was found to maintain stem cell markers expression (CD73, CD90 and CD 105) throughout the 14 days of angiogenic induction. Meanwhile, the expression of migratory markers; CCR1, CXCR4 and CCL28 were found to be downregulated in comparison to the angiogenic markers; Ang1, IL8 and VE Cadherin. SHED also demonstrated a higher capacity to undergo cell migration under immature state in comparison to the angiogenically induced environment. In conclusion, SHED undergoing angiogenesis is postulated to have much more lower capacity to migrate to the healing site. The knowledge gained from this study can be used to plan a strategic approach for stem cell-based tissue repair.

Keywords: dental; stem cell; repair; angiogenesis; migration; SHED

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ABSTRACT OF ORAL PRESENTATIONS

OP-02-07

Increased relative lymphocyte number with reduced mature activated T-lymphocytes (CD3+CD69+) in stunted iron overloaded major beta-thalassemia patients

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ABSTRACT

Alteration of immune response as either the consequence or cause of growth retardation is one of the common complication found in major beta-thalassemia patients, which may lead them to be susceptible to infection. T-lymphocytes harboring CD69 following their activation are central elements of the immune system. A cross-sectional analytical study applying multicolor flowcytometry aimed to characterize T-lymphocytes surface protein of asymptomatic 51 pediatric major beta-thalassemia patients routinely visit Hasan Sadikin General Hospital for routine blood transfusion linked to their growth status, iron level, and hematology profile were done. Nutritional status was assessed by height-for-age z-score. Serum iron, total iron□binding capacity (TIBC), serum transferrin, and serum ferritin were measured. Hemoglobin concentration, mean corpuscular volume, mean corpuscular hemoglobin and mean corpuscular hemoglobin concentration were also measured. Respectively, 41% and 22% of the population showed stunted and severely stunted growth. Stunted major beta-thalassemia patients showed a significant higher differential count of lymphocyte ($p = 0.02$) than well-nourished patients. Compared to well-nourished major beta-thalassemia patients, significant reduced CD3+CD69+ T-lymphocyte population ($p = 0.04$) while higher TIBC level ($p < 0.0001$) were found. However, TIBC level of stunted major beta-thalassemia patients was significantly lower ($p < 0.0001$) than in severely stunted patients. In conclusion, a chronic inflammatory disorder accompanied with cellular immunological defect revealed in undernourished besides double burdened by iron overload is a serious health problem in major beta-thalassemia patients and this preliminary study makes it even more pronounced. Further investigation in characterizing the T-lymphocyte subsets and cytokines involved in malnourished major beta-thalassemia patients is imperative to be carried out.

Keywords: Thalassemia major; T-lymphocyte; CD3+CD69+; stunting

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ABSTRACT OF ORAL PRESENTATIONS

OP-02-08

Genetic associations study of rs12745968 and rs4822752 between rheumatoid arthritis and schizophrenia patients

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ABSTRACT

The association between rheumatoid arthritis (RA) and schizophrenia (SZ) is not a new issue. Since 1936, studies of the relationship between these two diseases have been received in many fields including genetics. SZ is a psychiatric disorder and RA is an autoimmune disease of the joints that occurs when the body's immune system attacks its own cells. Both diseases are thought to be influenced by multiple genetic risk factors which were modified by the environment. We postulated that individual genetic variant may have either opposing effects or same effects on the risk of schizophrenia and rheumatoid arthritis. In this study, two significant SNPs were selected from an analysis of two large databases; rs12745968 and rs4822752 represent FAM69A and CRYBB1 genes, respectively. The SNPs were genotyped using Taqman SNP Genotyping as it was high throughput and highly accurate, precise, time-efficient, and cost-effective. A total of 270 (90 RAs, 82 SZs and 98 controls) blood samples from consented subjects were extracted for DNA and genotyped for both markers. The genotyped data was validated using sequencing for selected sampels. In addition to assessing allele and genotype frequencies, the results were also calculated for association and odds ratio test using SPSS software (version 22) based on chi-square calculation with 95% confidence interval (CI) and p-value <0.05. AA genotype of rs12745968 was significantly found in SZ (p=0.023) and RA (p=0.001) patients, and CC genotype of rs4822752 was also statistically significant in SZ (p=0.002) and RA (p=0.00006) compared to controls. Likewise, susceptibility risk of AG was significantly shown in rs12745968 in RA (OR=3.0175, CI=1.059-8.598, p=0.032) and SZ groups (OR=6.0208, CI=1.6997-21.338, p=0.0019). Moreover, the heterozygous genotype, CT of rs4822752 revealed the significant risk to both diseases, RA (OR=7.8245, CI=2.8177-21.7276, p=0.000016) and SZ (OR=5.6676, CI=2.288-14.0395, p=0.00007). As a conclusion, this preliminary results provided evidences for the engangement of rs12745968 and rs4822752 as susceptibility risk to RA and SZ. However, further correlation analysis is warranted to genetically explore the risk alleles among RA/SZ which lead to important insights of pathogenesis of the disease.

Keywords: Rheumatoid arthritis; schizophrenia; SNP genotyping

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ABSTRACT OF ORAL PRESENTATIONS

OP-02-09

Family profile of Down syndrome patients in West Java Province, Indonesia

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ABSTRACT

Down syndrome is the most common chromosomal abnormality characterised by complete chromosome 21 trisomy (classical), or partial chromosome 21 trisomy (mosaicism), or chromosome rearrangement involving chromosome 21 (translocation). The study was conducted to describe the family and social profile of Down syndrome patients who were referred to a research centre in Bandung, the capital city of West Java Province, Indonesia. This descriptive study provides picture of residential location and surrounding area of their dwellings around West Java Province region, which previous studies showed it as risk factors. This study involved people with Down syndrome in various ages whose chromosomes were examined by conventional karyotyping and Fluorescent In-Situ Hybridization (FISH) method. Data were collected from 205 patients with Down syndrome, during the period of September 2015 to March 2018, who were referred to Cell Culture and Cytogenetics Laboratory, Faculty of Medicine Universitas Padjadjaran, Indonesia. Results showed that the most common type of Down syndrome was classical 195 patients (95%), followed by mosaic 6 patients (3%) and translocation 4 patients (2%). Most of them live in urban area 84 patients (41%) while the rest live in the suburban area 76 patients (37%), and only a few came from rural area 15 patients (7%) and the rest were unknown area or uncompleted data. They live mostly in dense area (51%), in mid-density area (31%) and the rest live in sparse populated area (18%). Their houses are mostly located in residential estate area 107 patients (52%), nearby industrial area 38 patients (18%), and around agricultural area 30 patients (15%). For maternal age, there were 84 mothers (40%) with 35 years old and more, and the rest under 35 years old. Based on the study, it could be concluded that classical Down syndrome is the most common type, where the majority live in dense residential area. This study offered several new information regarding frequency, average age, area of Down syndrome patients' residence and the condition of neighbourhood in West Java where they live.

Keywords: Chromosome; descriptive; Down syndrome; prevalence

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ABSTRACT OF ORAL PRESENTATIONS

OP-02-10

Expression of SOX 10 gene in U87 cell treated with siRNAs

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ABSTRACT

Gliomas, tumours arise from glial cells, are the most common primary brain tumours of the central nervous system. Its incidence and mortality rates are still rising. An increasing number of studies have reported that SOX 10 plays an important role in various cancers. However, the role of SOX10 in gliomas remains inadequately appreciated. In this study, we aimed to investigate the biological role and potential molecular mechanism of SOX10 in gliomas. We found that the mRNA and protein expression levels of SOX10 were prevalently and significantly overexpressed in human glioma cell lines. We performed gene knockout experiments by transfecting glioma cell line, U87 with SOX10 small interfering RNAs (siRNA). The treatment was done using three types of siRNAs. It showed that SOX10 siRNA transfection significantly suppressed mRNA and protein expression of SOX10 in glioma cells. Furthermore, knockdown of SOX10 significantly inhibited cell proliferation and invasion, but promoted apoptosis in glioma cells. Our data suggest that knockdown of SOX10 inhibits glioma cell growth and invasion, possibly by downregulating downstream oncogenic proteins, providing novel insights into the development of glioma therapy through targeting of SOX10.

Keywords: Glioma; SOX10; siRNA; U87; gene expression

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ABSTRACT OF POSTER PRESENTATIONS

PA-01

Combinatorial effect analysis of Peppermint (Mentha x piperita L. Carl) essential oil and meropenem against plasmid-mediated resistant E. coli

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ABSTRACT

This study was carried out to investigate the bactericidal mechanism of peppermint essential oil (PEO) when PEO (1 % v/v) used individually and PEO (1 % v/v) in combination with meropenem (0.5 µg/mL) against multidrug resistant Escherichia coli. Chemical composition of PEO were identified via GC-MS, followed by time-kill analysis which was performed to evaluate the antibacterial activities of PEO and meropenem. In order to assess the ability of PEO in disrupting the bacterial membrane, zeta potential measurement, outer membrane permeability test and scanning electron microscopy were performed. Next, anti-quorum sensing assay was performed to assess the ability of PEO in quorum sensing inhibition. A complete killing activity was observed within five minutes of treatment with PEO and meropenem at sub-lethal concentrations. In addition, the zeta potential measurement and outer membrane permeability test performed indicated increase in the membrane permeability and membrane disruption which can be observed in the scanning electron micrograph. Furthermore, significant decrease in the light production of E. coli pSB1075 treated with PEO indicates the presence of quorum sensing inhibitors within PEO. The findings suggested that PEO possesses the capability to disrupt the bacterial outer membrane, thus increasing membrane permeability, in addition to possible inhibition of bacterial quorum sensing ability in multidrug resistant E. coli, elucidation of the actual mechanism will, greatly assist the mitigation of reversal of antibiotic resistance.

Keywords: Escherichia coli; essential oil; membrane permeability; Mentha x piperita L. Carl; quorum sensing; combinatorial effect.

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ABSTRACT OF POSTER PRESENTATIONS

PA-02

Molecular and physiological responses of recalcitrant indica rice to lignosulfonates during callus proliferation

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ABSTRACT

Lignosulfonate (LS) is commonly used as stimulant to enhance plant growth. To date, the effects of LS on regeneration of recalcitrant Oryza sativa indica L. CV. MR219 has not been reported. Therefore, this study was undertaken to evaluate the effects of LS on callus proliferation of recalcitrant MR219 rice. The MR219 calli were proliferated on MS media supplemented with different ion-chelated (NaLS and CaLS) and concentrations (50, 100, 150, 200 mg/L) of LS. Optimum callus proliferation rate of 88% was successfully obtained on MS media supplemented with 100 mg/L CaLS. In addition, presence of CaLS also increased adventitious root formation of MR219 callus by 62% Further expression analysis of adventitious root-related genes (OsWOX11, OsAUX1 and OsIAA23) recorded a 1.7-fold increment of OsWOX11 expression in CaLS treated calli, implying a positive role of CaLS in adventitious root development. Besides, CaLS-treated calli also recorded a 1.2-fold higher endogenous indole-3-acetic acid (IAA) content and enhancement of nutrient ions (Na+, K+, Ca2+, Mg2+, Fe2+, Mn2+, Zn2+ and Cu2+) uptake as compared to non-treated calli. Consistently, expression analysis of auxin-related genes (OsASA1, OsTAA1 and OsYUC1) and nutrient uptake-related genes (OsAKT1, OsHAK5, OsCBL, OsCIPK23 and OsCamk1) also showed a similar increment trend. Interestingly, the Ca2+ increment was observed throughout four weeks, but the major increment of K+ was only detected starting from week two. The observed rise of Ca2+ following the enhancement of endogenous K+ content, further suggest the possible cross-talk between these ions. Subsequent, proteomic profiling analysis revealed, an increase of carbon and nitrogen metabolisms in CaLS treated callus. Taken together, our results suggest that the presence of CaLS enhance proliferation, and adventitious root formation of MR219 callus through up-regulation of endogenous auxin synthesis, enhance nutrients uptake, and carbon-nitrogen metabolisms.

Keywords: Adventitious root development; callus growth; carbon and nitrogen metabolism; lignosulfonates; nutrient uptake; Oryza sativa

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ABSTRACT OF POSTER PRESENTATIONS

PA-03

Oriented antibody conjugation on fluorescence dye-doped silica nanoparticles for targeted in vivo imaging

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ABSTRACT

This work demonstrates the use of Cy5-doped silica nanoparticles (Cy5-SiNPs) with oriented conjugation of monoclonal antibodies against epithelial cell adhesion molecule (EpCAM), a biomarker for colorectal cancer, for targeted in vivo imaging. The antibody conjugation was performed on Cy5-SiNPs that were previously coated with a layer of protein G. The Protein G serves as a linker controlling antibody orientation due to their site-specific interaction with the constant domain (Fc) of Immunoglobulin G (IgG). This conjugation method allows the binding sites (Fab) of the antibodies to be facing outward, thus maintaining the conjugates affinity to bind to the target (EpCAM). The affinity of the oriented conjugates was compared with that of conjugates derived from a conventional EDC coupling by in vitro analysis using confocal fluorescence imaging and flow cytometry. The result demonstrated that the oriented conjugates provided 12 times higher sensitivity to bind to the target cells (HT-29) than that of the conjugates prepared by conventional EDC method. In vivo fluorescence imaging in mice bearing HT-29 tumor xenograft indicated time-dependent accumulation of the oriented conjugates at the tumor site and prolong fluorescence signal retention up to 14 days post-injection. This research demonstrated that the Cy5-SiNPs with oriented antibody conjugation developed herein can improve the sensitivity of in vitro analysis and can be successfully applied for targeted fluorescence in vivo imaging.

Keywords: antibody conjugation; protein G; fluorescence imaging; in vivo imaging; colorectal cancer detection

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ABSTRACT OF POSTER PRESENTATIONS

PA-04

The effect of flaxseed extract on skin elasticity of the healing wound in rabbit

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ABSTRACT

Management of disturbed wounds, large skin defects and the areas where skin tension precludes wound closure is of high clinical importance. Healing in such wounds occurs through epithelization and contraction processes (second-intentions healing) that may result in certain undesirable complications including keloid formations, a poor final cosmetic appearance and the formation of a fragile epithelial layer. The objective of this study is to evaluate the effectiveness of flaxseed gel in enhancing connective tissue firmness and improving skin texture during wound healing by means of viscoelasticity parameter. 30-male white neozealand rabbit were included in this study divided into 3 groups; one group of 10 rabbit received Flax seed gel topically for three times intervals (1, 3, and 7 days); a second group received Fucidin cream as positive control, while third group have not received any treatment as negative control, Skin elasticity measurements were performed using the DermaLab system (Cortex Technology). Photos of treated areas were taken during the procedure. Throughout the study skin elasticity was significantly greater in the Flaxseed group than in other groups. Flaxseed increase elasticity value from (4.2±1.02) to (4.7±1.9) after 7 days treatment (P=0.003). while no significant differences were evident in both Fucidin treated group (positive) control (1.0±0.34) and non-treated group (negative) control (1.8±1.4) (p=0.068) group. Hence, Young's modulus of skin elasticity in flaxseed group was more reproducible than other groups demonstrating the comparable efficacy of flaxseed extract in skin elasticity and distensibility. This study showed clearly the therapeutic effect of flaxseed extract on biologic tissue, including stimulation of microcirculation and improvement of fibroblastic cell activity. Elasticity evaluation demonstrated increased density and firmness in the network of collagen/elastic fibers in the dermis and subcutis during wound healing process promise in generating therapeutic gel to be used in wound healing process.

Keywords: Flaxseed; elasticity; skin wound

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ABSTRACT OF POSTER PRESENTATIONS

PA-05

Cytotoxicity assay of nilotinib and nicotinamide in a myeloid cell line, K562

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ABSTRACT

Nicotinamide is an active form of vitamin B3 with many physiological and pharmacological functions in various organisms. Its function as an NAD+ precursor and also a substrate for PARP-1 play an important role in DNA repair and also maintenance of the genomic to improve the capacity in DNA repair. Nicotinamide also inhibits poly (ADP-ribose) polymerase (PARP-1) that reassembles the DNA strand breaks caused by radiation and chemotherapy. Drug resistance towards the first-line treatment for Chronic Myeloid Leukemia has forced clinicians to switch to second- generation treatment such as nilotinib. The role of nicotinamide as a cancer preventive agent will be studied by investigating its effect in K562 cell line. This study aims to determine the IC50 of nicotinamide and nilotinib in a K562 cell line. The cell was cultured in a sterile environment with RF10 as the medium before being treated with nicotinamide and nilotinib for 72 hours. Concentration of dilution was done to obtain the desired concentration of nicotinamide (0.02 M, 0.04 M, 0.06 M, 0.08 M and 0.1 M) and nilotinib (5.0x10-9 M, 1.0x10-8 M, 2.0x10-8 M and 4.0x10-8 M) with the final percentage of 0.1% DMSO in 96- well plate. Cell cytotoxicity assay was carried out by using Cell Counting Reagent SF at 450nm. These cell treatments have shown that more than 50% of the cell viability have been inhibited as the concentration increases. Nicotinamide and nilotinib were able to inhibit the proliferation of K562 cell line at 4.02-83.44% and at 7.95-66.32%, respectively. IC50 value was successfully determined by using software GraphPad Prism for nicotinamide (0.03433 M) and nilotinib (8.788x10-9 M). These data will be used for the future study to investigate the effect of nilotinib supplemented with nicotinamide associated with poly(ADP-ribose) polymerase-1 (PARP-1) on K562 cell line.

Keywords: cytotoxicity assay; nicotinamide; nilotinib; K562 cell line

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ABSTRACT OF POSTER PRESENTATIONS

PA-06

Wazir; a VCO based bioproduct developed for haemorrhoids

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ABSTRACT

Haemorrhoids occur when blood vessels at the lowest anal region swell abnormally. There are various different ointments currently used to treat haemorrhoids such as herbal based products to ease of inflammation moisturize and lubricate the affected region. However, scientific evidence supporting the effectiveness for those herbal based product is still lacking. Virgin coconut oil (VCO) contains lauric acid and its derivative, monolaurin which displayed anti-microbial, anti-oxidant and anti-inflammatory properties. Furthermore, VCO contains natural humectant and property which expedite wound healing. In this present study, we described biotechnological approach in development of a product to treat haemorrhoids. The aim of this study was to compare our VCO haemorrhoids cream prototype (proposed trade named; Wazir) to other haemorrhoids creams. For methodology, Wistar albino rats (n=28), age 6-8 weeks old, weighing 160-180 g, were used in this study. Those animals were divided into four groups namely, negative control, croton oil (5%) induced positive control for haemorrhoids, control treated with hydrocortisone 1% (as used for clinical treatment) and induced animals treated with herbal preparation a prototype cream that contained the following leaves extracts; fig leaves (5%), artichoke (5%), walnut (5%) and horse chestnut fruit (5%). After 3 days of treatment with the prototype cream, those animals were sacrificed, histology & blood investigations namely, Myeloperoxidase, Malondyaldehyde, nitrate/nitrite and nitrotyrosine levels and Superoxide Dismutase activity were performed. For results, we observed, the treated group showed significant histological change towards haemorrhoids healing (one-way ANOVA & Tukey's multiple comparison tests; $p=0.000$; $p<0.01$). In conclusion, the herbal preparation demonstrated a marked improvement for haemorrhoids. Therefore, this strongly suggests that our group had developed an effective potential treatment for haemorrhoids.

Keywords: Virgin coconut oil (VCO); haemorrhoids; herbal cream; bioproduct

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ABSTRACT OF POSTER PRESENTATIONS

PA-07

Cytotoxicity of betel quid and areca nut aqueous extracts on mouse fibroblast and mouth-ordinary-epithelium 1 cell lines

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ABSTRACT

Betel quid chewing is a traditional habit associated with oral cancer. The composition of betel quid varies, but typically consists of areca nut and slaked lime wrapped in betel leaf. Although betel quid is associated with oral cancer, its role in the initiation and promotion stages of carcinogenesis is not fully clear. This study aims to investigate the cytotoxicity of crude betel quid and areca nut aqueous extracts on mouse fibroblast (L929) and mouth-ordinary-epithelium 1 (MOE1) cell lines. Selected concentrations of betel quid and areca nut (0.1 g/ml, 0.2 g/ml, 0.4 g/ml) were used in the study. Cytotoxicity analysis using MTT assay was performed in triplicates, whereby L929 and MOE1 were treated with each of the extract for 24 hours, 48 hours, and 72 hours respectively. The results were analysed using one-way ANOVA with Scheffe and Games-Howell Post hoc test and Kruskal Wallis complemented by Mann Whitney U-test for comparison of means at $p<0.05$. Both betel quid and areca nut extracts at all concentrations significantly resulted in reduced cell viability against L929, in comparison to the control. In betel quid and areca nut-treated MOE1, betel quid at all concentrations significantly resulted in increased cell viability, in comparison to the control, whereas areca nut at the highest concentration significantly resulted in reduced cell viability of MOE1 compared to control group at 48 hours and 72 hours incubation period.

Keywords: Areca nut; betel quid; cytotoxicity; L929; MOE1; oral cancer

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ABSTRACT OF POSTER PRESENTATIONS

PA-08

Association of PDFGRA gene polymorphisms at exon 12, 14 and 18 with clinical response to Imatinib mesylate treatment among chronic myeloid leukemia patients

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ABSTRACT

Platelet-derived growth factor receptor alpha (PDGFRA), a type III tyrosine kinase receptor has been implicated in some cancers. Mutation of PDGFRA gene which leads to constitutive activation of the receptor was associated with the development of resistance toward Imatinib mesylate (IM). This study was aimed to detect the PDGFRA gene polymorphisms at exon 12 (c.1701 A>G), exon 14 (c.1664 A>G) and exon 18 (c.2525 A>T) in association with treatment response among chronic myeloid leukaemia (CML) patients who treated with IM. A total of 86 patients (43 CML responses and 43 CML resistances) treated with IM for more than 12 months were recruited. High resolution melt (HRM) analysis was performed to detect PDGFRA gene polymorphism at exon 12, 14 and 18. The HRM curve was able to distinguish the genotypes by three difference clusters (homozygous wild-type, heterozygous and homozygous variant). Homozygous wild-type was used as the reference. HRM curve analysis for exon 12 was successfully differentiating the wild-type with the homozygous variant by a different melting curve, and no heterozygous variant was found. While for exon 14 and 18 the curve showed no difference with the wild-type. Few selected representative samples from different HRM clusters were sent for DNA sequencing for validation. The association of the genotypes with IM treatment response was assessed by means of odds ratio (OR) with 95% CI calculated by logistic regression analysis. The study revealed that CML patients carrying the homozygous variant (GG) genotype of exon 12 PDGFRA gene showed a higher risk of acquiring resistant; however, the association was not statistically significant with OR: 1.597 (95% CI: 0.681-3.745, P = 0.281). For exon 14 and 18, all the analyzed samples showed no polymorphism that associated with IM treatment response. Thus, the results were concluded that CML patients with exon 12 (c.1701 A>G) polymorphism are posing higher risk towards developing resistance to IM treatment but not for exon 14 and 18 which showed no significant findings.

Keywords: Chronic myeloid leukaemia; PDGFRA; Imatinib mesylate; polymorphisms

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ABSTRACT OF POSTER PRESENTATIONS

PA-10

Risk factors for psychopathology experienced by caregiver of thalassemia children

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ABSTRACT

Caregivers of thalassemia children must have good knowledge, physical, and psychological well-being to cope stress from burden they carry on. The occurrence of psychological changes of caregiver can lead to the emergence of symptoms of psychopathology that disrupts their quality of life. Those psychological changes can also be caused by psychosocial stressors. Therefore, the aim of this study was to know the distribution of psychopathology scores and the related factors among caregivers of thalassemia children. This is an analytic cross-sectional non-paired comparative study. A total of 54 subjects of study were caregivers who attended World Thalassemia Day event 2017 in Bandung, consented and recruited using consecutive sampling method. Caregivers who participated in screening test and was not illiterate. All subjects filled in questionnaire on socio-demographic information, knowledge of thalassemia, and SCL-90 questionnaire, by themselves (self-administered). Subjects who had done a psychological assessment was excluded. The study estimated that more than 75.9% subjects have good knowledge of thalassemia and 42.6% had psychopathologic symptoms. The risk factor that showed significant association with psychopathologic symptoms was occupation (p=0.045, OR 3.3735, 95%CI 1.001 to 11.383). There was no significant relationship found between other risk factors (sex, age, income, marital status, formal education, and knowledge of thalassemia). Thalassemia does not only affect the persons with the disorder but also their caregivers in many aspects including psychosocial well-being. Occupation was associated with psychopathology manifestations among caregivers of thalassemia children. Hence it is important for physicians to give health care management not only to the patients but also their caregivers.

Keywords: Caregiver thalassemia; risk factors; psychopathology manifestations

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ABSTRACT OF POSTER PRESENTATIONS

PA-11

Rapid aneuploidy detection of chromosome 21 by segmental duplication – high resolution melting analysis for prenatal diagnosis

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ABSTRACT

Chromosomal aneuploidy causes a number of syndromes and the most common is Down Syndrome (trisomy 21). These syndromes cause severe intelectual disability, and many abnormalities for lifetime. Thus, prenatal diagnosis is important to predict these aneuploidy. Karyotyping has been known as a gold standard for chromosomal abnormality detection. However, it has some disadvantages such as time consuming, culture failure, external contamination, and labour intensive. Molecular methods have been developed for rapid aneuploidy detection, such as Segmental Duplication-High Resolution Melting Analysis (SD-HRM). The purpose of this study was to propose and investigate a SD-HRM as screening method for rapid aneuploidy detection. This study used 1 primer set containing 2 pairs of primer for segmental duplication, which are similar sequences located on different chromosomes (chromosome 21,7, and 12). Dosage of segmental duplication targeted on aneuploidy (trisomy) will affect the melting profile and produce different melting curve compare to the euploid sample (normal) when amplified. This study used peripheral blood DNA of unaffected control (n=30) and Down syndrome individuals (n=57) which had been confirmed as classic trisomy (n=53), and mosaic (n=4) with karyotyping. SD-HRM attained high sensitivity (100%) and specificity (100%) (CI 95%=1.0), equal accuracy with karyotyping as diagnostic gold standard. Trisomy 21 samples were clearly differentiated with unaffected control. Mosaic trisomy 21 also was detected as positive with SD-HRM. Quantification analysis using mixed mosaic samples approach showed that SD-HRM could detect mosaic sample until 20% abnormal DNA. SD-HRM enables to detect aneuploidy with fast, sensitive, and cost-effective thus meet the demand to be used as method for prenatal diagnosis screening.

Keywords: Aneuploidy; karyotyping; prenatal diagnosis; SD-HRM

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ABSTRACT OF POSTER PRESENTATIONS

PA-12

Generation of induced pluripotent stem cells from normal human dermal fibroblast using non-integrative Sendai virus

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ABSTRACT

Induced pluripotent stem cells (iPSC) exhibited embryonic-like properties with unlimited self-renewal and multilineage differentiation properties, which is a potential cell source in regenerative medicine and transplantation therapy. Although retroviral and lentiviral transduction methods to generate iPSC are well established, the risk of mutagenesis limited the use of the cells in therapeutic applications. This study aims to generate iPSC from normal human dermal fibroblast (NHDF) cell line via non-integrative Sendai virus (SeV) transduction. When NHDF reached 70-80% confluency, SeV vectors expressing Yamanaka factors (Oct4, Sox2, Klf4 and c-Myc) were added and incubated for 24 hours. On day 6, the transduced cells were re-plated on a vitronectin-coated plate and daily medium change was performed. On day 18-26, colonies with embryonic stem cell (ESC)-like cellular morphology were observed and transferred to a new vitronectin-coated plate. After Passage 10, the iPSC generated were free of SeV as confirmed with RT-PCR. NHDF-derived iPSCs expressed multiple pluripotency markers by immunofluorescence staining and qRT-PCR as compared to parental NHDF. Following suspension culture in low attachment plate for 8-10 days, iPSC formed embryoid body-like spheres, similar to ESC. NHDF-derived iPSC also demonstrated the ability to undergo directed differentiation into cells of different germ layers. Taken together, NHDF were successfully reprogrammed into iPSC using non-integrating SeV. Further characterisation, such as teratoma formation and genome-wide sequencing, are required to elucidate the molecular profile and function of these cells.

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ABSTRACT OF POSTER PRESENTATIONS

PA-13

Identification of bacteria by amplification gene encoding 16s rRNA and antibiotics resistance test from pneumonia outpatients

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ABSTRACT

The bacteria that cause upper respiratory tract infections (URTI) pneumonia have become resistant due to the use of various antibiotics. The aims of this study were to identify the bacteria in URTI pneumonia outpatient samples and test resistance to some antibiotics. The bacteria were isolated from sputum sample of outpatients in a hospital in Garut West Java Indonesia and tested for gram staining. Then, isolation of bacterial chromosome and amplification of gene encoding 16s rRNA was performed using polymerase chain reaction method. The isolated bacteria were analyzed for antibiotic resistance to amoxicillin 30 µg/10 µg, cefadroxil 30 µg/10 µg, trimethoprim 5 µg/10 µg, sulfamethoxazole 300 µg/10 µg, ceftriaxone 30 µg/10 µg and cefotaxime 30 µg/10 µg. The PCR primers for the gene encoding 16s rRNA of Pseudomonas aeruginosa, Enterobacter cloacae, Klebsiella variicola, Pseudomonas geniculata, and Serratia marcescens were designed based on gene database; http://blast.ncbi.nlm.nih.gov. The result of resistance test showed that ceftriaxone and cefotaxime inhibited those bacteria while there were no inhibition zones of amoxicillin and cefadroxil except against to K. variicola. In addition, trimethoprim had no activity on P. aeruginosa and P. geniculata. Likewise, sulfamethoxazole had no activity against E. cloacae and P. geniculata. The isolated bacteria from URTI pneumonia outpatients are believed to cause hospital acquired pneumonia which had become resistant to amoxicillin, cefadroxil, trimethoprim and sulfamethoxazole.

Keywords: URTI pneumonia bacteria; 16s rRNA; PCR; sequencing; resistance test

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ABSTRACT OF POSTER PRESENTATIONS

PA-14

Stability of glycated albumin in human serum analyzed by developed aptasensor

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ABSTRACT

Glycated human serum albumin (GHSA) is a product from non-enzymatic reaction called “Glycation” in blood stream. The glycation involves the addition of reducing blood sugar to human serum albumin (HSA); the most abundant transport protein in blood, to form a freely reversible Schiff base or Amadori products. GHSA has been reported as a biomarker for glycemic control in diabetes patients, Alzheimer and inflammation, therefore, accurate concentration of GHSA in serum/plasma has been considered. Previously, we successfully developed aptasensor for glycated albumin detection in diabetes mellitus (DM) serum/plasma. This work we studied the stability of GHSA and HSA in serum/plasma samples using our developed aptasensor. We found that GHSA and HSA concentrations are stable at least 8 hours at room temperature, at least 7 days at 4OC and at least 30 days at -80OC after blood drawing. However, we observed significant changes of the GHSA and HSA concentrations in serum/plasma stored at -80OC for longer than 30 days due to serum osmolality. Interestingly, GHSA concentrations from DM serum/plasma were highly fluctuated comparing with those from normal serum/plasma. It is possible that reversible glycation rate in the DM serum/plasma was higher than that in normal samples. Therefore, in order to get accurate GHSA and HSA concentrations for diabetes monitoring, human serum/plasma samples are not allowed to be kept at room temperature for no longer than 8 hours or at 4OC for no longer than 7 days and at -80OC for no longer than 30 days. In order to prolong the sample storage time, glycation inhibition should be considered.

Keywords: Glycated albumin; human serum albumin; serum; plasma; stability; aptasensor

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ABSTRACT OF POSTER PRESENTATIONS

PA-15

Optimization of DNA isolation method from Formalin-Fixed-Paraffin-Embedded (FFPE) tissues and comparative performance of four different Polymerase Chain Reaction (PCR) kits

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ABSTRACT

Cancer tissue may have important information useful for diagnostic, prognostic and genetic counselling of cancer patient. Formalin-Fixed-Paraffin-Embedded (FFPE) is a routine method used for preserving tissues including cancer tissue. However, genomic DNA isolated from FFPE tissue is often difficult to be amplified using PCR method due to DNA fragmentation and DNA-protein crosslinks induced by formalin preservation. This study aimed to optimize DNA isolation method from FFPE tissue and subsequently be used to compare the performance of four different PCR ready-to-use kits. Genomic DNA was isolated from FFPE colon and prostate cancer tissue using Quick-DNA™ FFPE Kit (Zymo Research) with and without pre-heating treatment in KOH/NOH solution. Four different PCR kits: MyTaq HS Red Mix 2X (BioLine), FastStart Taq DNA Polymerase (Roche), KAPA2G fast PCR Kit 2X (KAPA Biosystem) and KOD FX Neo (Toyobo) were used to amplify the Androgen Receptor (AR) gene from the genomic DNA. DNA electrophoresis was performed to compare the PCR results. The results showed that BioLine and Toyobo kits gave better PCR results than Roche and KAPA Biosystem. Increasing amount of Taq polymerase and dNTPs of Roche kit by two fold could increase the quality of PCR results. Toyobo could amplify DNA fragment to a maximum of 417 bp, however none of the PCR kits could amplify DNA fragment up to 450 bp. Pre-heated treatment of FFPE tissue in NaOH/KOH did not improve the DNA quality and PCR results. However, designing the primers producing amplicon of not more than 450 bp is suggested.

Keywords: DNA-FFPE; isolation; PCR; performance

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ABSTRACT OF POSTER PRESENTATIONS

PA-16

A molecular approach in establishing evidence of asymptomatic submicroscopic malaria among the Orang Asli population in RPS Pos Kemar

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ABSTRACT

Submicroscopic cases are often associated with asymptomatic malaria parasite carriers especially among adults. Asymptomatic submicroscopic parasitaemia may serve as a reservoir for infection even when very efficient rapid diagnosis and treatment programmes have been implemented. The objective of this study was to uncover the evidence of submicroscopic malaria among asymptomatic Orang Asli in RPS Pos Kemar, Hulu Perak. Study samples were collected from 4 villages located within Pos Kemar and were selected based on the number of previous malaria cases. A total of 751 villagers were consented and participated in this study. All these blood samples were examined by microscopic examinations (749) and nested PCR (751). Our findings showed that 16 samples (1.6%) were positive by microscopy and 8 samples (2.4%) were positive for P.vivax by PCR. Seven (0.9%) samples were submicroscopic malaria. A study described that many of these asymptomatic infections are present at densities below than the limit for microscopic detection and, therefore, the use of microscopy is likely to lead to underestimation of the malaria burden. Although microscopy remains the gold standard for the diagnosis of malaria and quantification of Plasmodium parasites, the rapid advances in molecular biology and nucleic acid testing methods and their routine application in clinical studies and epidemiological surveys have enabled the detection of low-density submicroscopic infections as reported in several other studies. Moreover, the development of highly sensitive, specific and quantitative molecular diagnostic tests for malaria are becoming increasingly important as control strategies to eliminate asymptomatic infections that serve as reservoirs for transmission.

Keywords: Malaria; asymptomatic; submicroscopic; Orang Asli; PCR

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ABSTRACT OF POSTER PRESENTATIONS

PA-17

RET mutation screening in Hirschsprung patients at a tertiary hospital in Indonesia: Mutation rate and in silico analysis

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ABSTRACT

Hirschsprung disease (HSCR) is a congenital intestinal malformation characterized by absence of enteric ganglia in the gastrointestinal tract. Mutations in RET gene have been known to be the major cause of both sporadic and familial HSCR cases. On average, 40 to 60 HSCR cases a year had been admitted to a tertiary hospital in West Java, in Indonesia. This study aims to screen for RET mutations in HSCR patients from a tertiary hospital in Indonesia and perform in silico analysis to predict the pathogenicity of the RET coding mutations. Thirty five patients enrolled in this study and DNA was isolated from 3 ml of peripheral blood. We performed a targeted-deep sequencing approach using MiSeq-Illumina Platform on the UTRs, exon-intron boundaries and the exonic regions of RET. One polymorphism on coding region was identified heterozygously in 5 patients (c.2071G>A/p.Gly691Ser) and three RET mutations were identified heterozygously in 3 patients. One of RET mutation is located in the coding region (c.1271A>G/p.Lys424Arg) and the other two mutations are located in 3utr and splicing site. In silico analysis using Polyphen and SIFT for RET coding mutation showed that the variant was consider to be benign/tolerated. This study shows that the mutation rate is around 8.6% (3/35) in this group of patients which is smaller than that of previously described studies in different population (13-15%).

Keywords: Hirschsprung disease (HSCR); in silico; mutation; polymorphism; RET

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ABSTRACT OF POSTER PRESENTATIONS

PA-18

Interaction between sensory dendrite and epithelial cells in Drosophila

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ABSTRACT

Contact-mediated self-avoidance/repulsion restricts dendrite in a 2D space. Defect in dendrite-extracellular matrix (ECM) adhesion disrupts the confinement and results in self-crossings in 3D space. In this study, epithelial genes underlying dendrite-ECM affecting larval dendrite patterning were identified through genetic screen using mutants and RNAi knockdown. In-vitro and in-vivo assays were performed to characterize the epithelial gene function. To understand their mechanism, further in-depth study is required to elucidate their roles in coordinating sensory dendrite arborization.

Keywords: Dendrite; epithelial cells; Drosophila

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ABSTRACT OF POSTER PRESENTATION

PA-19

Challenges in Diagnosis of Mitochondrial Respiratory Chain Complexes Disorder in Human Skin Fibroblasts

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ABSTRACT

Mitochondrial respiratory chain (MRC) disease is a heterogeneous group of disorders characterized by impaired energy metabolism due to genetically-based oxidative phosphorylation (OXPHOS) dysfunction. The diagnosis is challenging as the preliminary diagnosis is solely based on the clinical basis. The present study describes the biochemical and molecular approaches used to confirm MRC disease. Skin biopsies were taken from 24 infants suspected of having inborn errors of metabolism and further being processed to acquire fibroblasts for all experimental work. Biochemical approaches in MRC diagnosis involved kinetic enzymatic assay and Western-Blot analysis of MRC proteins; namely Complex 1 to Complex V. The kinetic activity of each enzyme complex was captured using microplate spectrophotometer at a specific wavelength. The enzyme activities were expressed as a rate (nmol/min) per mg of protein. In Western-blot analysis, proteins were separated by electrophoresis and electro-blotted onto PVDF transfer membrane, followed by immuno-binding reaction of antibody against targeted proteins subunit. The protein immune-blots were visualised and estimated through a Protein Image Analyser. In any case of abnormality detected by both analyses, the mitochondria DNA sequencing would be performed to detect for any mutation. Out of 24 patients, 4 patients were found to have MRC complex I and IV deficiencies by enzymatic assay, with a moderately low protein detection through Western blot analysis. 2 out of these 4 abnormal patients were preliminarily diagnosed with fatty acid oxidation deficiencies, suggesting a secondary to OXPHOS defects. Through DNA sequencing, one patient was concluded for a heterozygous mutation for SURF-1 (gene encodes an assembly factor of mitochondrial complex IV), therefore the patient is a carrier. Despite the success in getting a conclusive and reliable diagnosis, the study also recorded few important challenges during the process, which includes difficulties in obtaining skin biopsy samples from the patients, the requirement for a large number of cells and fresh samples, and also the preferences of using isolated intact-mitochondrial for enzymatic assay analysis.

Keywords: Mitochondrial respiratory chain; skin biopsy; fibroblasts

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ABSTRACT OF POSTER PRESENTATION

PB-01

DNA Damage Response profiles in reprogrammed osteosarcoma cell lines

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ABSTRACT

Osteosarcoma (OS) is a malignancy of the bone occurring mostly in children and adolescents. Alterations and mutations to genes associated with proliferation and differentiation increase the risk of OS tumourigenicity. Reprogramming of OS cells to a more primitive stage could be useful to study and understand OS pathogenesis. By using retroviral OSKM, the Yamanaka factors, two OS cell lines, G-292 and Saos-2, were reprogrammed to pluripotency. Colonies from the reprogrammed OS, designated as iG-292 and iSaos-2, showed ESC-like morphology, expressed pluripotency markers, formed embryoid body-like spheres, expressed markers from three germ layers and showed the ability to differentiate into adipocytes and osteocytes. However, in vivo study showed teratoma formation only in iG-292. Hierarchical clustering analysis from global gene expression profile of both parental and reprogrammed OS demonstrated distinctive separation of two clusters of population. Differentially expressed genes (DEGs) were further grouped into DNA repair, cell cycle and apoptosis pathways. Our data showed that iG-292 displayed more DEGs than iSaos-2 in these three pathways. The ability to repair DNA damage in cells is regarded as a crucial process to protect genome integrity and to suppress tumourigenesis. There are no reports on DNA Damage Response (DDR) pathways of reprogrammed cancer cells. OS has been linked to DNA mutation and are known to be resistant to DDR. Thus, this study is essential to gather valuable and novel information on OS pathogenesis, in particular DDR, for future therapeutic intervention.

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ABSTRACT OF POSTER PRESENTATION

PB-02

Epithelial-Mesenchymal Transition (EMT) deregulation in reprogrammed Oral Squamous Cell Carcinoma (OSCC-iPSCs)

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ABSTRACT

Reprogramming, pluripotency and malignant transformation are interrelated processes controlled by epigenetic. The epigenetic mechanism during cancer cells reprogramming, somehow results in reversing the 'cancer state' by silencing oncogenes or activates tumour suppressor genes. Not only reversing the cell fate, suppression of epithelial to mesenchymal transition (EMT) related genes were also reported in recent studies. EMT is a biological process known to induce migratory phenotype of cancer cells, and greatly collaborates in the pathogenesis of cancer. EMT reversion acquired from reprogramming, which in turn could be used to explore the cancer epigenome. Herewith, we have successfully demonstrated reprogramming of H103 (OSCC - STNMP Stage I) as evident by pluripotent characterisations. Further downstream analysis was carried out to investigate the differential gene expression (DGE) patterns via microarray platform between parental H103 and H103-iPSCs. The overall DGE pattern showed deregulation of EMT genes indicating H103-iPSCs may have reduced tumorigenic properties upon reprogramming. The encouraging result of this in-vitro study has therefore confirmed its worthiness to study the tumour-suppression effect upon reprogramming, where H103-iPSCs might be of value in the future as in modelling OSCC pathogenesis and drug screening.

Keywords: induced pluripotent stem cells; OSCC; reprogramming; EMT

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ABSTRACT OF POSTER PRESENTATION

PB-03

Mutations of FLT3 and CKIT genes in core binding factor with acute myeloid leukemia: IMR experience

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ABSTRACT

Core binding factor (CBF) is a heterodimeric transcription factor made up of two subunits, CBFα (also known as RUNX-1) and CBFβ. CBF plays a role in the transcriptional activation of a number of genes required for normal hematopoietic differentiation. CBF acute myeloid leukemia (AML) include 2 major subtypes, respectively associated with translocation t(8;21) (CBFα) and inversion inv(16)/t(16;16) (CBFβ), which show a high rate of complete remission (CR) and prolonged CR duration. FLT3 internal tandem duplication (ITD) and D835 mutations is frequently observed aberration associated with poor prognosis in acute myeloid leukaemia (AML). CKIT mutations have been reported in core binding factor (CBF) AML. In CBF AML patients, frequently detected second mutations are FLT3 and c-KIT. This study aims to explore FLT3 and c-kit mutations in patients with acute myeloid leukaemia (AML). In this study, we retrospectively analysed the prevalence of FLT3 and CKIT mutations in 108 AML patients with t(8;21) and inv (16). The bone marrow and peripheral blood samples were extracted using QIAamp DNA MiniPrep Kit. The multiplex RT-PCR assay was performed using FLT3 Mutation Assay (Invivoscribe, USA). CKIT mutation was performed using AmoyDx CKIT Mutation and Human CKIT Gene D816V Mutation Detection Kit and run via Applied Biosystems 7500 Real-time PCR. The frequencies of FLT3-ITD, FLT3-D835, and CKIT mutations were 1.98%, 5.94%, and 14.8%, respectively. Double mutations of CKIT and FLT3-D835 were detected in 3 cases (2.97%). The occurrence rate of FLT3-ITD and CKIT mutations increased in adult with 21.4 % and 15.8% respectively compared to paediatric patients. Notably, the prevalence of FLT3-D835 and double mutations of FLT3-D835 and CKIT were observed only in adults. The frequencies of FLT3-ITD, FLT3-D835, and CKIT mutations in our AML patients are lower compared to other study. This may suggests that mutations of other genes also involve in in stimulating proliferation of leukaemia cells. Identification of these mutations are important for prognostication and optimization of patient care.

Keywords: FLT3-ITD; FLT3-D835; CKIT; acute myeloid leukaemia; core binding factor

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ABSTRACT OF POSTER PRESENTATION

PB-04

Identification of chromosomal translocations in leukaemia using multiplex reverse transcriptase polymerase chain reaction: a retrospective ten years study in Malaysia

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ABSTRACT

Leukaemia is the sixth most common cancer in Malaysian population and seventh most common in males and eighth most common in females. It is the most common cancer among children. Recent advances in genomics have contributed significantly towards a better understanding of the genetic landscape of leukaemia. Identification of recurrent chromosomal translocations which can be detected in a substantial number of these patients are important for classification of the disease, prognostication, treatment monitoring and also to guide targeted therapy. A total of 3433 bone marrow or peripheral blood samples were collected from patients who were newly diagnosed with leukaemia. Their demographic data, bone marrow morphology and also immunophenotyping results were recorded. Multiplex reverse-transcriptase polymerase chain reaction was performed using the HemaVision®-28N protocols for detection of 28 common translocations. This study aims to report the incidence of leukaemia-specific translocations in Malaysian patients who were admitted in tertiary care hospital from 2008 to 2017. Among the 3433 patients, 1500 patients were diagnosed with Acute Lymphoblastic Leukaemia, 1160 for Acute Myeloid Leukaemia, 581 for Chronic Myeloid Leukaemia and 192 patients for Acute Promyelocytic Leukaemia. We found that 34.7% of these patients have chromosomal translocations. Overall, 23 fusion gene transcripts were detected. The most common genetic aberration found were BCR-ABL1 47.8%, PML-RARA 13.1% and RUNX1-RUNX1T1 11.4%. Interestingly, based on our study, we found one CML case with unique breakpoint with larger PCR product compared to common breakpoint within BCR gene at exon e14 (B3) and ABL1 gene at exon e2 (A2). In this case, the PCR product have additional 124bp and sequencing confirmed the extra nucleotide derived from ABL1 gene at exon 1 chromosome 9. Multiplex RT-PCR is an effective and rapid screening tool for detection of recurrent chromosomal translocations in leukaemia. A comprehensive sub classification of leukaemia by molecular technique is very useful not only for diagnostic purpose, but also for risk stratification, prognostication and targeted therapy.

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ABSTRACT OF POSTER PRESENTATION

PB-05

Evaluation of MGMT methylation status among HUSM glioma patients: A preliminary study

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ABSTRACT

Growing evidences show that understanding the roles of biomarkers has significantly increased our current perception of gliomagenesis, prognostic evaluation, and treatment planning for patients. For instance, identification of the promoter methylation status of O6-methylguanine-DNA-methyltransferase **(MGMT)** gene encodes MGMT, a protein with DNA repair activity may improve the efficacy of current standard care in glioma as the methylation status has been recently introduced to be a predictive biomarker for stratification of the treatment plan. To further understand the roles of MGMT, the present study aims to evaluate the status of MGMT promoter methylation status of glioma patients in HUSM. In this study, 21 samples of paraffin-embedded glioma tissue (FFPE) were obtained based on their grading from Grade II (n = 5), III (n = 4) and IV (n = 12). Subsequently, DNA extraction and methylation status was validated by methylation-specific PCR (MSP) using two pairs of primers specifically targeting the unmethylated (UM) and methylated (M) regions of the MGMT gene respectively. MSP results identified high intratumoral heterogeneity of the samples in all grades of the tumours. In Grade II glioma, 20% were M and 80% were both UM and M while in Grade III and IV glioma, 25% were M and 75% were both UM and M. None of the samples exhibited UM status alone of the MGMT promoter. Nevertheless, analysis using Fisher's exact test found no statistical association between the MGMT methylation status and any of the tested clinicopathological parameters such as tumour grading, age, gender, and race of the patients ($p > 0.05$). Similar to our results, intratumoral diversity of MGMT promoter methylation status has also been previously demonstrated in some studies. Besides highlighting the existence of the intratumoral heterogeneity, it may also suggest future challenges for defining personalized treatment based on the epigenetic status of MGMT.

Keywords: MGMT; MSP; MGMT methylation status

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ABSTRACT OF POSTER PRESENTATION

PB-06

Chromosomal instability in human osteosarcoma is mediated through Hypoxic inducible factor 1α (HIF-1α)

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ABSTRACT

Osteosarcoma is a rare bone malignancy with fast tumour progression disrupting tumour mass oxygen supply. As to ameliorate the hypoxic environment, new blood vessel developed from the existing vasculature through angiogenesis. However, in the literature, the role of Hypoxic inducible factor 1α (HIF-1α) in human osteosarcoma is unclear. Although osteosarcoma tissue cultures study suggested that the chromosomal instability is due to disturbance of the chromosomal segregation mechanisms and a defective mitotic checkpoint affecting cellular proliferation. However, not much is known about the underlying signalling pathways mediating chromosomal instability in human osteosarcoma. The aim of this study was to investigate the involvement of HIF-1α as the mediating signalling pathway leading to chromosomal instability in human osteosarcoma. The methods used were micronuclei staining, Fluorescent in situ hybridization (FISH) and immunohistochemistry. Our results showed increased micronuclei formation (24/31) 77.4% and amplification of 6p21 chromosome region (16/22) 72.7% in human osteosarcoma. This was related to increased HIF-1α and VEGF protein expression ($p<0.05$). In addition, there were also occurrence of PAS positive blood vessel (vasculogenic mimicry, VM) in those cases (12/29) 41.4% compared to (6/26) 20.7% non-VM blood vessel ($p<0.05$). It was concluded that hypoxic tumour microenvironment partly contributes in chromosomal instability, leading to increase angiogenic factor which further decreased patient's survival.

Keywords: Osteosarcoma; chromosomal instability; Hypoxic Inducible Factor 1α (HIF-1α);Vascular Endothelial Growth Factor (VEGF).

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ABSTRACT OF POSTER PRESENTATION

PB-07

Impact of promoter polymorphisms of apoptotic signaling regulatory genes FAS/FASL on chronic myeloid leukemia susceptibility risk

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ABSTRACT

Chronic myeloid leukemia (CML) is a stem cell disorder in which Philadelphia (Ph) chromosome is implicated in the etiopathogenesis. Ph chromosome translocation results in the formation of BCR-ABL oncogene which is known to deregulate different downstream pathways which ultimately lead to cell proliferation, defective DNA repair, and inhibition of apoptosis. But evidence show that the Ph chromosome alone is insufficient for development of CML. Host genetic susceptibility factors which favours CML development are not clear. Fas cell surface death receptor (FAS) is a member of tumor necrosis factor superfamily which interacts with its ligand, FASL to initiate apoptosis. Promoter polymorphisms in FAS/FASL genes are known to influence the apoptotic signaling and influence carcinogenesis. So this study was undertaken in a total of 191 subjects (93 CML patients and 98 normal control) to investigate the frequencies and impact of FAS-670A>G and FASL-844 T>C polymorphism on CML susceptibility risk. After getting written consent, blood samples of study subjects were collected, DNA extraction done from blood samples and genotyping was performed using PCR-RFLP technique. The genotypes were categorized into homozygous wild type, heterozygous and homozygous variant. The association of the genotypes with CML susceptibility risk was assessed by means of logistic regression analysis and deriving odds Ratio with 95% CI. Homozygous wild genotype was used as reference. Few representative genotypes were validated using DNA sequencing. Data was analyzed using SPSS Version 22. On evaluating the impact, the study revealed a significant association of FASL−844T>C polymorphisms with CML susceptibility risk. Both variant C allele (OR 1.756, CI 1.163,2.652, p=0.007) and variant genotype CC of FASL-844 (OR 2.261, CI 1.013,5.047, p=0.047) carried significantly higher risk for CML development. The heterozygous genotype TC significantly conferred lower risk for CML susceptibility (OR 0.379, CI 0.176,0.816, p=0.013) For FAS-670 A>G polymorphism, both heterozygous genotype AG and variant genotype GG showed higher values (OR 1.642 CI, 0.719-3.750 and OR 1.133, CI 0.464-2.768, respectively) but were statistically insignificant (p= 0.239 and p= 0.784 respectively). Our results highlight the impact of FASL-844T>C polymorphism on CML susceptibility risk. This novel finding might be helpful in early identification of individuals who are at higher risk for development of CML.

Keywords: Chronic myeloid leukemia; susceptibility risk; FAS/FASL; promoter polymorphism

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ABSTRACT OF POSTER PRESENTATION

PB-08

An enigmatic hyperdiploid multiple myeloma with novel and complex cytogenetic abnormalities – A rare occurrence

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ABSTRACT

Multiple myeloma (MM) is a cytogenetically heterogenous plasma cell malignancy. Based on the hallmark cytogenetic abnormalities, MM can be divided into hyperdiploid and non-hyperdiploid subtypes. The hyperdiploid subtype is characterized by trisomies of certain odd numbered chromosomes namely 3, 5, 7, 9, 11, 15, 19 and 21 whereas non-hyperdiploid subtype is characterized by translocations of the immunoglobulin heavy chain alleles at chromosome 14q32 with various partner chromosomes with the most important of which being t(4;14), t(6;14), t(11;14), t(14;16), and t(14;20). In general, hyperdiploid patients are considered a better prognostic group while non-hyperdiploid patients are considered a high risk group. Here we report one myeloma case presented with hyperdiploid karyotype along with other complex high risk abnormalities and also a novel abnormality which was not previously described. A 74 years old Malay lady presented with abnormal biochemical profile namely reversed albumin globulin ratio with very high globulin level, normochromic normocytic anaemia and acute kidney impairment. As part of MM diagnostic work up, bone marrow cytogenetic analysis was performed using conventional cytogenetics and fluorescent in situ hybridization (FISH). Conventional cytogenetic analysis showed hyperdiploid metaphases with 51-53 chromosome range and involving translocation (2;3)(q21;p21), trisomies 5,7,11,15,17,19 and 21. FISH analysis revealed presence of del(13)(q14.3), t(11;14)(q32;q13.3), t(4;14)(p16.3;q32.2) and del(17)(p13). The genetic profile of MM can acts as a determinant of patient survival and response to treatment. The reported patient's tumour cells exhibited a hyperdiploid chromosome count and translocation t(11;14) which are associated with good prognosis. However some of the tumour cells exhibited translocation t(4;14), deletions of chromosome 13q and 17p13. These abnormalities are associated with adverse prognosis. A novel abnormality translocation t(2;3) whose significance is unknown also was detected in the tumour cells. Hence this patient who is currently undergoing treatment (on dexamethasone, thalidomide, intravenous immunoglobulin and intravenous Zoledronic acid) is being closely follow up to determine whether the presence of these high risk cytogenetic abnormalities along with good prognosis abnormalities will confer her a more aggressive disease course. This case is presented because of the rare and simultaneous occurrence of these good prognostic, adverse prognostic abnormalities and also one novel abnormality in one patient.

Keywords: Multiple myeloma; hyperdiploid; non-hyperdiploid; complex cytogenetic abnormalities; interphase FISH

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2nd Conference of Malaysian Society of Human Genetics

ABSTRACT OF POSTER PRESENTATION

PB-09

Sox 6, Sox 13 and Sox 9 expression pattern in meningioma in East Coast Malaysia.

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ABSTRACT

Sox (Sry-related high- mobility group box) family plays significant functions in human development from embryonic development, organogenesis and the most recent findings, tissue regeneration. As cell key modulators, deregulation of these genes has been associated with several human diseases including cancer. Sox D and Sox E, two of the nine SOX subgroups and their molecular mechanisms in carcinogenesis are widely studied. Eventhough there are a considerable number of studies done on SOX gene in glioma, such studies in meningioma, which is the most frequent brain tumor type in East Coast Malaysia are still lacking. Thus, this study is opted to determine the expression levels of Sox 6 (SOX D), Sox 13 (SOX D) and Sox 9 (SOXE) in both low and high-grade meningioma in Malaysia population. Formalin-Fixed Paraffin-Embedded (FFPE) samples of low-grade meningioma, high-grade meningioma and a normal brain FFPE tissue were sectioned by using microtome. RNA extraction was then performed according to manufacturer's instruction. cDNA conversion was then completed by using reverse transcription technique. Finally, Sox6, Sox13 and Sox9 expression pattern in meningioma were achieved by q-PCR assay and normalised to nonneoplastic brain tissues. Each target gene was normalized with beta Actin as internal control or housekeeping gene. The data was analysed statistically with One way ANOVA by using Graphpad Prism 6. The results displayed that Sox6, Sox13 and Sox9 gene were downregulated in all low-grade meningioma in comparison of normal tissue. In addition, there were upregulation observed in both Sox6 and Sox9 expression but downregulated in high-grade meningioma for Sox13. Sox6, Sox13 and Sox9 expression levels in selected brain tumours in Malaysia population provide new insights of SoxD and SoxE expression in this population. The well-known varsity functions of Sox genes and the canonical interaction of Sox genes with their co-factors may elucidate the fluctuations of Sox gene expression level across diseases and genetic backgrounds. Thus, functional studies are recommended to be carried out to observe the selected genes' functions and mechanisms whether they should reflect their diverse roles in specific Malaysia population.

Keywords: Sox6; Sox13; Sox9; brain tumours; qPCR; meningioma

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ABSTRACT OF POSTER PRESENTATION

PB-11

Determination of drugs resistance molecular markers of Plasmodium falciparum in Malaysia

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ABSTRACT

Continuous monitoring of the development and spread of malaria parasite resistance towards anti-malarial drugs is fundamental in progressing towards eliminating malaria from Malaysia. However, the updated data on the distribution of drug resistance molecular markers in Peninsular Malaysia is limited, involving mostly only from East Malaysia. Thus, the aim of this study was to determine the drug resistant molecular markers and their distribution in Peninsular and East Malaysia. A total of 67 diagnostic blood samples from malaria infected individuals were received from district health centres and confirmed as P. falciparum by nested PCR for species specific identification. Samples were then subjected for drug resistance genes analysis for artemisinin (PfKelch-13; K13) and chloroquine (Pfcrt and Pfmdr1) using Restriction Fragment Length Polymorphism (RFLP) and validated by sequencing. Out of 67 samples, 2 (3%) samples showed a presence of K13 mutations at P553L and A675V which has been implicated to be associated with delayed parasite clearance for artemisinin. Meanwhile, 44 samples (65.7%) showed mutation for Pfcrt genes at K78T site and 28 samples (41.8%) have mutation for Pfmdr-1 at N86Y. Although artemisinin combination-based therapy is used as first line anti malaria treatment in Malaysia, continuous monitoring of the distribution of Pfcrt and Pfmdr1 mutations alongside the emergence of K13 mutations is vital in order to facilitate national policy makers in governing and managing the burden of the disease to the country.

Keywords: Drug resistance; molecular markers; Plasmodium falciparum; malaria

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In conjunction with
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ABSTRACT OF POSTER PRESENTATION

PB-12

A case of allelic dropout in SLC25A13 gene in a patient with Citrin Deficiency

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ABSTRACT

Citrin deficiency is an autosomal recessive disorder caused by biallelic pathogenic variants in SLC25A13 gene. PCR and Sanger sequencing of the SLC25A13 gene is the most reliable method for definitive diagnosis of citrin deficiency. However, an incidental finding of a case with allelic dropout; a phenomenon which does occur occasionally was identified recently. Allele dropout is an amplification failure of one of two alleles caused either due to allele-specific sequence variations where primer cannot hybridize to sequence binding sites or preferential amplification of an alternate allele. This study described a case of allelic dropout in a citrin deficiency patient and strategies used to detect and to solve the case. One-month-old male infant presented with neonatal intrahepatic cholestasis was referred to Molecular Diagnostics & Protein Unit, Institute for Medical Research for SLC25A13 gene mutation analysis. The diagnostic strategy is by using long-range PCR to screen for a recurrence IVS16ins3kb. Next, PCR and sequencing of all the18 exons were performed using specific primers. Parental samples were obtained and tested for the origin of the mutations identified in the proband. Long-range PCR showed that the proband was heterozygous for IVS16ins3kb mutation and parental testing demonstrated that the allele was maternally-inherited. Sequencing analysis to search for a second mutation discovered 23bp duplication in exon 16, as homozygous in the proband and heterozygous only in the father; suggesting allele dropout in the proband. A PCR and sequencing with redesigned primers at exon 16 revealed a heterozygous c.1638_1660dup, p.(Ala554Glyfs*17) in both of them concluded that allele from the maternal was dropped by the original primer. No sequence variations at maternal allele that inhibit primer to hybridize, nevertheless it was the 3kb insertion in the maternal allele that caused the amplification failure with PCR and resulted in erroneous sequencing readout of a homozygous genotype. The case was successfully identified and an allelic dropout issue in SLC25A13 gene using specific primers avoiding the 3kb insertion was solved. As a conclusion, analysis of parents' sample is important to confirm the mutation and to troubleshoot obscure findings in the proband that may lead to misdiagnosis.

Keywords: Allele dropout; SLC25A13 gene; Citrin Deficiency

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ABSTRACT OF POSTER PRESENTATION

PB-13

Kennedy Disease: The First Case Report in Malaysia

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ABSTRACT

Spinal and bulbar muscular atrophy also known as Kennedy Disease (KD) (MIM #313200) is a gradually progressive neuromuscular disorder, in which degeneration of lower motor neurons resulting in muscle weakness, muscle atrophy and fasciculation. These symptoms usually begin in adulthood and worsen slowly over time. KD is inherited in an X-linked pattern, caused by abnormal expansion of CAG repeat in exon 1 of Androgen Receptor (AR) (NM_000044.3) gene. Normal individual has CAG repeats up to 36, whereas KD patient's has more than 40 repeats. We reported here a 61 years old male patient with progressive nasal speech, impairment of speech clarity, facial asymmetry and generalised weakness for over 3 years. These symptoms were noticed by his family and friends and he was advised to seek medical attention. The aim of this study is to characterize the pattern of CAG repeats in AR gene of this patient by PCR and fragment analysis. Clinical examination of this patient confirmed palatal muscle weakness, velopharyngeal insufficiency, proximal myopathy, facial myokinesia and gynacomastia. The nerve conduction study showed absent sensory and smallest motor response with diffuse neurogenic changes of chronic nature and MRI brain showed no significant changes. He was then referred to Hospital Kuala Lumpur for diagnosis of Kennedy disease and whole blood EDTA was sent to our laboratory for molecular testing. DNA was extracted using a standard protocol. PCR amplification was carried out using primers flanking the trinucleotide repeat region, followed by capillary electrophoresis and sizing of the PCR products. Genemapper software was used to determine the number of CAG repeats. One expanded allele was detected in our first KD patient in Malaysia with approximately 46 CAG repeats. Family screening was suggested for all asymptomatic male relatives as KD is an X-linked disease. As a conclusion, we have successfully determined the CAG repeats expansion in KD patient for confirmation of the disease. Capillary electrophoresis has provided accurate sizing of fragment which is very important in determination of repeats.

Keywords: Kennedy Disease; CAG repeats; neuromuscular disorder

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ABSTRACT OF POSTER PRESENTATION

PB-14

Recognition of genital ambiguity as an unusual presentation of Klinefelter syndrome in childhood

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ABSTRACT

Klinefelter syndrome (KS) is a sex chromosome aneuploidy caused by the presence of one or more supernumerary X chromosomes. Common clinical phenotypes of KS are comprised of tall stature with a feminine body type, gynecomastia, small testes and infertility. Cases of KS with genital abnormalities as the main evaluation in childhood are barely disclosed. We report four children presented to our clinic for assessment of ambiguous genitalia who were ultimately diagnosed with KS. The first patient was a 4 months male baby who presented with phenoscrotal hypospadias, bifid scrotal and small testes. Endocrine studies suggested a normal hypothalamic-pituitary-gonadal axis. The second patient was a 3 weeks baby born with the concern for ambiguous genitalia. He was evaluated at birth for bifid scrotum, small testes and glanular hypospadias. The third and fourth patients were three and seven years old boys with severe hypospadias, bifid scrotal and small testes. Hormonal analysis showed a low level of testosterone with normal level of FSH and LH. The chromosome analysis was 47, XXY for all of the patients confirming the diagnosis of KS. Individuals with KS have a highly varied phenotype comprising a range of physical features, however, genital anomalies are rarely described as characteristics feature of the syndrome in childhood. Clinicians need to be aware of the phenotypic variability of KS and recognize KS as one of the causes of abnormal genitalia at birth. This finding, along with appropriate genetic counselling, suggest that early detection of KS is important in monitoring potential development problems; such as hypogonadism, gynecomastia and gender dysphoria in the future.

Keywords: Klinefelter syndrome; genital; chromosome

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ABSTRACT OF POSTER PRESENTATION

PB-15

Novel mutations in SLC16A2 gene in four unrelated Malaysian boys with MCT8 deficiency

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ABSTRACT

Monocarboxylate transporter 8 (MCT8) deficiency is a disorder caused by impairment in the transcellular transport of thyroid hormones, which are essential for proper development and function of the brain. MCT8 is encoded by SLC16A2 gene located on the Xq13.2 chromosome. SLC16A2 gene is the only gene known responsible for causing the MCT8 deficiency. MCT8 deficiency is an X-linked disorder characterized by severe cognitive deficiency, infantile hypotonia, generalized muscle weakness and spasticity. Thyroid function tests in patients with MCT8 deficiency are usually abnormal with increased free 3,3′,5-triodothyronine (T3), normal to low free 3,3′,5,5′-tetraiodothyronine (T4) and normal to elevated thyroid stimulating hormone (TSH). **This study aims** to describes and characterize four novel mutations found in four unrelated MCT8 deficiency patients in Malaysia. Nineteen patients suspected with MCT8 deficiency were referred to our laboratory for SLC16A2 gene mutation analysis. PCR and direct sequencing were performed on six coding exons and flanking introns of SLC16A2 gene. Mutational analysis was then performed using Seqscape software v3.0 and the variants found were evaluated using the web-based software MutationTaster2 for pathogenicity predictions. Human Gene Mutation Database (HGMD) was used to check whether these mutations found have been previously reported. Mutational analysis of SLC16A2 gene revealed four different novel mutations which include three hemizygous frameshift mutations (c.488dupT, c.1461dupC and c.244_245insTATA) detected in three different patients and one hemizygous missense mutation (c.626G>A) found in 2 siblings. Frameshift mutations are predicted to introduce premature stop codon which will produce a truncated protein, hence rendering the thyroid hormone transporter defective. The missense mutation c.626G>A was also predicted to be disease-causing by MutationTaster2. All five patients portrayed similar clinical characteristics that include developmental delay, hypotonia and deranged thyroid function tests. Four novel mutations in SLC16A2 gene have been detected in four unrelated patients with similar MCT8 deficiency phenotype. MCT8 deficiency should be suspected in male patients with psychomotor retardation, hypotonia and deranged thyroid function tests with X-linked inheritance pattern. Genetic counselling should be provided to the parents of affected patients for future family planning.

Keywords: MCT8 deficiency; SLC16A2 gene; novel mutations

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ABSTRACT OF POSTER PRESENTATION

PB-16

Prevalence of Lysosomal Storage Diseases in Malaysia

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ABSTRACT

Lysosomal storage diseases (LSD) are a group of genetic disorders resulting from defective lysosomal metabolism and subsequent accumulation of substrates. It can be categorised into a group of lipidoses, mucopolysaccharidoses (MPS), cholesterol ester, glycogen and mucolipidoses. In Malaysia, information about the incidence of LSDs is relatively limited. The aim of this study was to determine the prevalence of LSD in Malaysia and compare them with other countries. A retrospective epidemiological study was conducted by covering the period between 2000 and 2017. The birth prevalence of LSD in Malaysia was calculated based on 83 confirmed cases by enzymatic assay and/or mutation analysis. The combined birth prevalence for all LSD in Malaysia is 0.93 per 100,000 live births compared to the Netherlands (Europe) and United Arab Emirates (Asia) which were 14 and 26.9 per 100,000 live births respectively. Within the group of lipidoses, Gaucher disease is the most frequent LSD with calculated birth prevalence of 0.31 per 100,000. Metachromatic leukodystrophy (MLD) was diagnosed in 18.07% of all diagnosed cases and the calculated birth prevalence was 0.20 per 100,000 live births. Within the group of MPS, MPS IVA has the highest calculated birth prevalence (12.2% of all cases of MPS diagnosed) of 0.33 per 100,000 live births. The birth prevalence of MPS II was 0.23 per 100,000 live births (0.45 per 100,000 male live births), representing 36.59% of all cases of MPS diagnosed. The combined birth prevalence for all MPS was 0.59 per 100,000 live births. In conclusion, this study revealed that LSD is not rare in Malaysia as compared to other countries; therefore awareness of LSD should be promoted among health care provider in Malaysia since specific therapies are available for most of the cases diagnosed.

Keywords: Prevalence; Lysosomal Storage Disease; incidence

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ABSTRACT OF POSTER PRESENTATION

PB-17

RUNX2 Single nucleotide polymorphism (rs6930053) in Class II malocclusion patients: A preliminary study

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ABSTRACT

Runt-related transcription factor 2 (RUNX2) plays important roles in osteoblast differentiation, tooth development and chondrocyte maturation; hence its involvement in craniofacial development is paramount. Mutation in RUNX2 is implicated with cleidocranial dysplasia; a bone development disorder, while single nucleotide polymorphism (SNP) in RUNX2 is associated with Class II/D2 malocclusion. Although genetic factor has been associated with the incidence of malocclusion; very limited study was conducted to determine the association of certain genes with the incidence of malocclusion in Malaysia. Thus, this preliminary study aimed to determine the presence and association of RUNX2 SNP (rs6930053) in Class II malocclusion patients. Genomic DNA was extracted from unstimulated saliva of 31 Class I (control samples) and 30 Class II malocclusion patients. Cephalometric measurements were performed prior to saliva samples collection. The DNA was amplified using the specific primers for marker rs6930053 and the genotyping was done by sequencing. Chi-square test was used to determine differences in allele and genotype frequencies. Significant difference was detected in allele ($p=3.04X10-6$) and genotype ($p=4.06X10-6$) frequencies between control (Class I) and Class II malocclusion. This result suggested there was a genetic association between RUNX2 (rs6930053) with Class II malocclusion ($p=3.04X10-6$, OR= 6.59; 95% CI=2.88~15.08). We provided preliminary observation that RUNX2 SNP (rs6930053) might contribute to Class II malocclusion in our local population. Further studies involving larger number of samples and other DNA markers of RUNX2 gene should be developed in order to understand the exact role and mechanism of RUNX2 in different classes of malocclusions and how this polymorphism affects the malocclusion cases in Malaysian population.

Keywords: Class II malocclusion; rs6930053; SNP; RUNX2

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ABSTRACT OF POSTER PRESENTATION

PB-18

Association of JAK2 gene polymorphisms in Malaysian patients with Crohn's disease

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ABSTRACT

Crohn's disease (CD) is one of the prominent subtypes of inflammatory bowel disease (IBD). It affects any part of the gastrointestinal (GI) tract, especially colon and ileum. CD is on a rising trend in Asia, with prevalence of 26 cases per 100,000 population in Malaysia. To date, there is no cure available for CD. Although the exact cause of CD remains ambiguous, genetic factor has been shown to play a very important role in the development of CD. Janus kinase 2 (JAK2) is the key protein in JAK/STAT signalling pathway that regulates inflammatory response in mucosal barrier. Alteration of the JAK2 protein due to the changes in its gene could lead to unregulated inflammations that result in the onset of CD. Thus, we aim to study the association of JAK2 gene polymorphisms in Malaysian CD patients. A total of 99 CD patients and 297 matching controls were recruited from University Malaya Medical Centre. Venous blood was drawn and genomic DNA was extracted via a conventional phenol-chloroform extraction method. A total of four selected JAK2 SNPs (rs10758669, rs7849191, rs10974944, and rs10975003) were typed via TaqMan® SNP genotyping assays in a real-time PCR system. Genotyping results were validated by PCR-resequencing approach. Genomic and allelic data were tabulated and analyzed using statistical tests to associate the SNPs with the onset of CD in Malaysian population. None of the four SNPs showed significant association with the onset of CD in the overall Malaysian population. However, in stratification analysis, the heterozygous C/G genotype of rs10974944 was found to increase risk for CD in the Chinese population (P=0.0176; OR=2.645). For genotype-phenotype association analysis, the homozygous C genotype (P=0.0247) and allele A (P=0.0268) of rs10758669 were found to associate with inflammation at the upper GI tract in CD patients. The heterozygous C/T genotype of rs7849191 was found to reduce risk for colonic CD (P=0.038; OR=0.395) and it was also observed to be associated with the extra-intestinal complication (arthritis) (P=0.0181; OR=5.238). In conclusion, JAK2 rs10974944 polymorphism was significantly associated with CD onset in Chinese population and further study could be conducted to investigate its effect on CD.

Keywords: Crohn's disease; Janus Kinase 2 gene; SNPs; genotyping.

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ABSTRACT OF POSTER PRESENTATION

PB-19

Contribution of GDF-15 and FGF-21 as potential diagnostic biomarkers for mitochondrial respiratory chain disorders.

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ABSTRACT

Mitochondrial respiratory chain disorders (MRCD) are inherited neurological disorders that occur 1 in 5000 live births. Currently, muscle biopsy is the gold standard for diagnosis of MRCD due to the lack of sensitive biomarkers discovered. Growth differentiation factor-15 (GDF-15) and fibroblast growth factor-21 (FGF-21) have been proposed as potential biomarkers for detection of MRCD. This would be a very useful for first-line diagnostic tool, instead of the invasive muscle biopsy. We aimed to validate the utility of these two factors as promising biomarkers in MRCD using both biochemical and molecular tools. We evaluated 41 plasma samples from high-risk neurological symptom of MRCD (group1), 104 samples with non-high risk of MRCD but abnormal findings through screening tests (group2) and 45 samples from healthy control (group3) by ELISA. Skin fibroblasts from 9 high risk patients were tested for their GDF-15 and FGF-21 levels, super-complex 1-V immunological activity and mutation analysis. Statistically, the level of GDF-15 and FGF-21 in group1 were significantly high compared to the group3 (mean 84560 pg/ml±24808 SEM and 33086 pg/ml±25845 SEM respectively, $p<0.05$). In group2, GDF-15 and FGF-21 were elevated between 4-34 times compared to group3 (mean1017 pg/ml±167 SEM and 1467 pg/ml±515 SEM respectively, $p<0.05$). The area under receiver-operating-characteristic curve for GDF-15 was 0.7187±0.0556 SE indicating that it has a good discriminatory power in group1 compared to FGF-21 (0.6301±0.0603 SE). The overall sensitivity and specificity of GDF-15 for cut-off value of 300 pg/ml was 90.24% and 75.56% ($p<0.05$). However, we found no significant correlation between GDF-15 and FGF-21 (p value=0.465). GDF-15 shows more precise plasma/fibroblasts quantitative biomarker in comparison to FGF-21 in diagnosing of MRCD. This was supported by fibroblasts tests in which 5 out of 9 patients showed abnormal results in their protein immunological activity and mutations were detected.

Keywords: Biomarkers; respiratory chain disorders; GDF-15; FGF-21; fibroblasts

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ABSTRACT OF POSTER PRESENTATION

PB-20

A case report of a Malaysian with WAGR syndrome

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ABSTRACT

The Wilms tumour, aniridia, genital abnormalities and mental retardation (WAGR) syndrome is a rare genetic syndrome which is caused by contiguous gene deletion of PAX6 and the adjacent WT1 genes. The signs and symptoms of WAGR syndrome are related to the loss of multiple genes on the short (p) arm of chromosome 11. Here, we report the genetic cause in the case of a 4-year-old girl with global developmental delay, bilateral cataract and ptosis, everted lower lip, and tall nasal bridge. Genomic DNA subjected to microarray analysis using Agilent CGH microarray was carried out revealing loss of 535 probes at 11p14.3 to 11p13 with size of 10,417Kb. The array showed a deleted interval containing 49 RefSeq genes / 30 OMIM genes / 7 OMIM morbid: BDNF, FSHB, PAX6, WT1, CD59, LMO2 and CAT genes which are located within the known disease region "WAGR 11p13 deletion [OMIM #194072]". Haploinsufficiencies in this region that include WT1 and PAX6 have been reported to cause major phenotypic characteristic of developmental delay and autistic features. Two studies found interstitial deletions at 11p13 including PAX6 is associated with aniridia, cataract, ptosis and mental retardation. Our findings suggested this mutation is most likely responsible for the pathogenesis of WAGR syndrome in this patient. Additionally, probands below the age of 6 years old with WT1 deletion were also found susceptible to childhood onset of Wilms tumor. While most cases result from a chromosomal deletion that occurs as a random event during gamete formation or in embryogenesis, some individuals may inherit an unbalanced translocation from an unaffected parent who carries a reciprocal translocation. Parental genetic screening is advised as risk for phenotypic abnormalities differs between familial or de novo mutation. Identification of mutation spectrum in this patient has crucial implications for understanding of a disease and its behaviour, leading to modifications of therapeutic recommendations, prognostic predictions and genetic counselling. Genetic etiology of WAGR syndrome cases in Malaysia are still scarcely reported. Hence, this case is reported for its rarity and for case collection to correlate between genotype and phenotype of WAGR syndrome in Malaysia.

Keywords: WAGR syndrome; Wilms tumour; congenital cataract

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ABSTRACT OF POSTER PRESENTATION

PB-21

Sex chromosomal mosaicism among DSD patients in Indonesia

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ABSTRACT

Chromosomal Disorder of Sex Development (DSD) is an atypical condition of external genitalia which inappropriate with sex chromosome result. The cytogenetic testing for DSD patients often revealed normal male and female karyotype. In Chromosomal DSD, some have chromosomal abnormalities, such as sex chromosome aneuploidy and mosaicism. Mosaicism is a condition when individual has more than one chromosomally different cell line due to mitotic non-disjunction during post zygotic period. Mosaicism involving different sex chromosome results broad spectrum of DSD phenotype. The aim of this study was to describe external genitalia characteristic and gender of DSD patients with chromosomal mosaicism. This research was a cross-sectional study among DSD patients who were referred to Center for Biomedical Research (CEBIOR), Faculty of Medicine Diponegoro University, Semarang, Indonesia since 2004 until 2017. Disorders of Sex Development patients were undergone physical examination and chromosomal analysis, and 22 patients who had chromosomal mosaicism were recruited. Gender assignment was done in all patients.There were nine males and three females gender of DSD patients with mosaic karyotype involving different sex chromosome (46,XY/45,X) with various percentage of cells affected. All males had severe hypospadia with six of them had bilateral cryptorchidism and two had unilateral cryptorchidism while a female patient had bilateral scrotal gonad. In addition, three males were 46,XY/46,XX had severe hypospadia and one of them had bilateral cryptorchidism. Furthermore, two males with 46,XX/47,XXY had unilateral cryptorchidism, one of them had severe hypospadia, while the other had mild hypospadia. The other results were a male with 47,XYY/46,XY/45,X had bilateral cryptorchidism;a male patient with 46,XY/46,XX/45,X hypospadia and micropenis; a female patient with 47,XXY/46,XY/45,X had labia majora hypoplastic ; a male with karyotype 48,XYYY/47,XYY/45,X had severe hypospadia and unilateral cryptorchidism; and a male with 46,Xyr/45,X had severe hypospadia. There were various phenotypic external genitalia among male and female DSD patients with sex chromosomal mosaicism. Gender assignment is important for these patients in the first childhood period.

Keywords: Disorder of Sex Development (DSD); sex chromosome; mosaicism

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ABSTRACT OF POSTER PRESENTATION

PB-22

Clinical profile of autosomal dominant hereditary ataxia

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ABSTRACT

Hereditary ataxia is a clinically and genetically heterogenous group of disorder characterized by slow progressive incoordination of gait and associated with poor coordination of hands, speech and eye movement. It can be inherited in an autosomal dominant, autosomal resessive, x linked, or mitochondrial.This is a pedigree of a 5-generation-family with gait ataxia and in combination with symptoms such as nystagmus, tremor, sensory (numbness), and dysarthria. The affected of first generation was the mother but in the 2nd, 3rd, 4th and5th generations the affected are males and females. The inheritance is autosomal dominant. The onset of gait ataxia is in between 30-50 years of age. The inheritance is autosomal dominant and the early symptom starts with sensory or dysartria. There are 8 family members with gait ataxia who have Scale for Assessment and Rating of Ataxia (SARA) between 2-28. Seven family members do not have ataxia, but have other symptoms with SARA between 0,5-2.There is anticipation within each generation. The investigation of family member no V.6.4. (male, 31 years of age) showed dysartria begins at 18 years old and gait ataxia at 30 years old. Other symptoms were nystagmus, tremor, stiffness, and numbness. Physical examination showed nistagmus, tremor, hyperreflexia, gait ataxia, and sensory disturbance. NCS study showed axonal demyelinating sensoric motoric peroneus bilateral. EMG study showed poliphasic pattern in anterior tibialis muscle. MRI showed mild dilatation of 4th ventricel, enlarge cysterna magna and with free communicating to foramen magendy (Dandy Walker Syndrome?). After 4 month physiotherapy treatment SARA score improved from 8 to 1. It need further investigation in clinical and genetic testing .

Keywords: Autosomal dominant; gait ataxia; hereditary

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ABSTRACT OF POSTER PRESENTATION

PB-23

Mitochondrial DNA hypervariable region (HVS-I) analysis of Semoq Beri population in Peninsular Malaysia

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ABSTRACT

Orang Asli is the earliest inhabitants that still practicing traditional lifestyle and has been recognised as indigenous to the country. The molecular interest on Orang Asli started from dispersal of prehistoric population by the ancient “Out of Africa” migration to other continents. In this study, DNA sequence of the mitochondrial hypervariable region I (HVS-I) was analysed among 40 unrelated Semoq Beri population in Hulu Terengganu as an initial effort to determine the sequence variation and haplogroup identification. Totally, seven haplotypes containing 18 polymorphic sites and neither deletion nor insertion were found in 481 bp long. A total of five haplogroups were identified which are haplogroups M74 (35,078.9 ± 7,152.0 ya), M21a (22,822.7 ± 7,891.2 ya), H2a3 (5,203.8 ± 3,219.3 ya), R21 (4,100 – 5,100 ya) and N9a6b (171 ya). The earliest age estimations were within Late Pleistocene until the end of Last Glacial Maximum period. The distribution of those haplogroups except H2a3 located within the Southern Mongoloid and South East Asia, thus showing bottleneck population. This study speculated that: (1) a total of five series of prehistoric migration occurred; (2) there was a possibility that the Semoq Beri ancestry was originated from Sunda shelf and (3) there was likely a minor Late Glacial/early postglacial dispersal from Sunda shelf into Peninsular Malaysia and Southern Borneo.

Keywords: Orang Asli; Semoq Beri; hypervariable segment; haplogroup; migration

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ABSTRACT OF POSTER PRESENTATION

PB-24

Genetic variants of BMP4/HpHI and IRF6 /MboI genes in two families with non syndromic cleft lip and palate patients

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ABSTRACT

Non syndromic cleft lip and palate (NS CL/P) is the most common craniofacial malformation in humans. Bone Morphogenetic Protein 4 (BMP4) and Interferon Regulatory Factor 6 (IRF6) have been consistently shown to be associated in NS CL/P from some human populations in the world. The aim of this study was to know the role of BMP4/HpHI and IRF6 /MboI gene polymorphisms in 2 families with NS CL/P and get to know whether there would be a risk factor for the rehearsal occurence of NS CL/P in the subsequent offspring through the probability analysis of the mutant genotypes. The study was laboratory descriptive design and the examination was performed in the form of pedigrees from 2 families from 3 generations with NS CL/P by using PCR-RFLP with HpHI and MboI restriction enzymes. The study results showed that the probability of TC mutant genotype of BMP4/HpHI gene polymorphism was 1/6 to be inherited in third generation of NS CL/P patients and the probablity of GA mutant genotype of IRF6/MboI gene polymorphism was 1/8 to be inherited in third generation of NS CL/P patients. The probability of the children with BMP4/HpHI and IRF6/MboI gene polymorphisms are greater when their grandparents or parents were also recognize to have BMP4/HpHI and IRF6 / MboI gene polymorphisms.

Keywords: nonsyndromic CL/P; genetic variants; inherited pattern; BMP4/HpHI; IRF6 /MboI

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ABSTRACT OF POSTER PRESENTATION

PB-25

Complications in adult thalassemia patients due to iron overload

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ABSTRACT

Thalassemia is a genetic disease caused by disruption of globin chain synthesis causing fragile red blood cells. Continuous blood transfusion every month is needed to prevent severe anemia on thalassemia patient. However, transfusions can also cause excessive iron in the blood, which can accumulate in the body tissues and interfere with the metabolism and causing tissue and organ damage. The objectives of this study were to document progress and surveillance cardiovascular and liver complications in thalassemia patients due to iron excess. This cross sectional study had research subjects of thalassemia patients who were treated at outpatient Department of Hematology Medical Oncology Hasan Sadikin General Hospital Bandung in the last 5 years, which included 62 patients. The results showed from the liver function examination, the increase of SGOT and SGPT levels occurred in 50 and 42 patients, respectively. For cardiac function examination, the most commonly found ECG abnormality was ST-T wave changes in 20 patients and in echocardiography examination was diastolic dysfunction in 15 patients.

Keywords: Thalassemia; iron overload; ferritin

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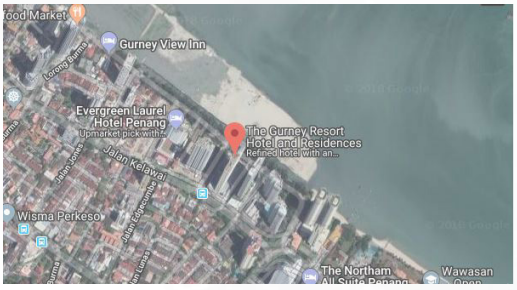

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ACKNOWLEDGEMENTS

The Organising Committee of the
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expresses gratitude to the following dignitaries and agencies for their support and commitment:
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