



1st International Symposium and Workshop

MEDICAL GENETICS UPDATE: FROM GENOMICS TO CLINIC

PROGRAMS AND ABSTRACTS BOOK

MAY 16 – 18, 2024

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**INTERNATIONAL SYMPOSIUM AND
WORKSHOP MEDICAL GENETICS
UPDATE: FROM GENOMIC TO CLINIC
(ISMGU 2024)**

**May 16 – 18, 2024
Universitas YARSI, Jakarta
INDONESIA**

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ISMGU 2024 Organizing Committee

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Table of Contents

Welcome Note	6
Background	7
About Univeritas YARSI	8
Symposium and Workshop Venue	9
Symposium Venue – Ar – Rahim Hall	9
Workshop Venue - Workshop Room	9
Venue Floor Plan	10
Ground Floor	10
11 th Floor.....	11
12 th Floor.....	11
Committee	12
Steering Committee:	12
Organizing Committee.....	12
Schedule	14
Workshop.....	14
Symposium	15
Scope of Posters and Oral Presentations	16
Guidelines	16
Abstract	16
Posters.....	17
Workshop Session 1 - Tips-Tricks on Chromosome and Molecular Analysis on Genetic Disease	18
Dr. Aziati Azwari Annuar (Human Genome Center, Universiti Sains Malaysia)	18
Prof. dr. Sultana MH. Faradz, PAK, Ph.D (Universitas YARSI).....	19
Workshop Session 2 - Genetic Counselling Practice	20
Dr. dr. Nurin Listyasari, MSimed (KG) (RSAB Harapan Kita, Jakarta)	20
Prof. dr. Sultana MH. Faradz, PAK, Ph.D (Universitas YARSI).....	21
Workshop Session 3 - How Do We Know Pathogenic Variants? (Cancer Cases).....	22
Prof. dr. Sofia Mubarika Harjana M.Med.Sc,PhD (Universitas Gadjah Mada) & Ahmad Rusdan Handoyo Utomo, Ph.D (Universitas YARSI)	22
Workshop Session 4 - Resolving Issues on Hematologic Disorder: Case Discussion on Thalassemia and G6PD Deficiency	23

Prof. Dr. Zilfalil Alwi, MBBS, MMed, MSc, Ph.D, FMM (Universiti Sains Malaysia, WHO TAG-G, Unesco Chair on Human Genetics of Thalassemia)	23
Prof. dr. Narazah Mohd Yusoff, MBBS, DCP, M.Med Sci, Ph.D (Advanced Medical and Dental Institute, Universiti Sains Malaysia)	24
Dr. dr. Anggraini Iriani, SpPK, Subsp HK (K) (Universitas YARSI)	24
Plenary Talks	25
Yosr Hamdi, Ph.D (Institut Pasteur of Tunis, Tunisia, WHO TAG-G)	25
Prof. Dr. Zilfalil Alwi, MBBS, MMed, MSc, Ph.D, FMM (Universiti Sains Malaysia, WHO TAG-G, Unesco Chair on Human Genetics of Thalassemia)	26
Prof. dr. Pratiwi Pujilestari Sudarmono, Ph.D, Sp.M.K (K) (Universitas YARSI)	27
Scientific Symposium 1	28
Prof. Dr. Liza Sharmini Ahmad Tajudin, MBBS, MMed, Ph.D, FAM (Departement of Ophtalmology, Universiti Sains Malaysia)	28
Nur Aizati Athirah Daud, Ph.D (Human Genome Center, Universiti Sains Malaysia) ..	29
Prof. dr. Gunadi, Ph.D., Sp.BA, Subsp.D.A.(K) (Universitas Gadjah Mada)	30
Scientific Symposium 2	31
Dr. Nazihah Mohd Yunus (Human Genome Center, Universiti Sains Malaysia)	31
Prof. Thanyachai Sura MD, FRCP (Department of Internal Medicine, Mahidol University, Thailand).....	32
Scientific Symposium 3	33
Dr. Catherine Lynn T. Silao (National Institutes of Health, University of the Philipines)	33
dr. Mila Maidarti, SpOG-KFER, Ph.D (Universitas Indonesia).....	34
Prof. dr. Sultana MH. Faradz, PAK, Ph.D (Universitas YARSI).....	35
Scientific Symposium 4	36
Prof. dr. Narazah Mohd Yusoff, MBBS, DCP, M.Med Sci, Ph.D (Advanced Medical and Dental Institute, Universiti Sains Malaysia)	36
Dr. Norafiza binti Mohd Yasin (Institute for Medical Research, Setia Alam, Selangor, Malaysia)	37
Prof. Dr. Raja Zahratul Asma Raja Sabudin (Departement of Pathology, Universiti Kebangsaan Malaysia).....	38
Oral Presentation Abstract	39
Genetics	39
Cancer Biology and Cancer Therapeutics	53

Cellular and Molecular Research, Immunology	58
Pharmaceutical Sciences, Phytopharmacy and Herbal Medicine	62
Health Promotion, Health Policy and Education	67
Others	84
Special Industry Session	92
3Billion.....	92
Pandu Biosains - Illumina	94
Prodia	95
Poster Presentation Abstract	96
Genetics	96
Cancer Biology and Cancer Therapeutics	105
Cellular and Molecular Research, Immunology	105
Pharmaceutical Sciences, Phytopharmacy and Herbal medicine	111
Health Promotion, Health Policy, and Education	127
Others	129

Welcome Note

Assalamu'alaikum warahmatullahi wabarakatuh,

Dear our respectful colleagues,

Universitas YARSI, on its 57th anniversary, is proud to hold the 1st International Symposium and Workshop, "Medical Genetics Update: From Genomic to Clinic." This activity is a collaboration between the Postgraduate School's Biomedical Science Master's and Doctoral Study Program, the Faculty of Medicine, and the Research Center of Genomic/Genetic Universitas YARSI. This activity begins with a workshop to expand participants' knowledge and skills about genetic diseases, from molecular, cellular, laboratory, and clinical aspects to practical genetic counseling. The symposium was held over two days, presenting extraordinary speakers from Indonesia and abroad. The speakers will share their expertise and experience regarding genetic disorders, oncology, personal and precision medicine, diagnosis, and genetic counseling.

We also invite students, lecturers, clinicians, and researchers to present their latest research results in oral sessions and poster presentations. This particular event is also an occasion for the initiation of the formation of the Asian-African Medical Genetics Society (AAMGS), which is expected to become a means for Asian and African researchers to exchange knowledge and conduct joint research in the field of genetics. Allow us to take this opportunity to express our sincere thanks to Dr. Dra. Lucia Rizka Andalucia, Apt, M.Pharm, MARS Director General of Pharmaceuticals and Health Devices. Ministry of Health of the Republic of Indonesia for being pleased to deliver the keynote speech. We also express our infinite gratitude to the incredible speakers who made the effort to reveal their expertise and experience in this scientific meeting.

We are very grateful for the financial support of sponsors 3billion, Inc, Seoul, Korea; PT. Pandu Biosains- Illumina Jakarta and Prodia Widyahusada Jakarta, so this event can run smoothly and successfully. Hopefully, this collaboration will continue in the future. Our sincere thanks to the Chairman of the Yarsi Foundation, Prof. dr. H. Journalis Uddin, PAK., and dr. Shanti Jurnal, Sp.A., M.Kes., and Chancellor of Universitas YARSI, Prof. dr. Fasli Jalal, Ph.D., for the guidance and support in organizing this event. We also express our deepest thanks to the Steering Committee, Organizing Committee, and students who have worked hard to organize this event. May Allah SWT reward you with abundant goodness.

We hope that all participants and speakers enjoy this event and get an advantage from friendship, exchanging knowledge, experience, and insight, as well as continue collaborating in education and research.

Wassalamu alaikum wa rahmatullahi wa barakatuh

Warm regard,

Wening Sari
Chairman



Background

Reports of congenital anomalies in newborns in Indonesia are increasing. The causes of these anomalies should be investigated for prevention. This condition is often overlooked by health workers due to ignorance and inadequate of examination and diagnosis facilities in accordance with scientific development. The discovery of Next Generation Sequencing (NGS) in 2006 enables scientists to establish the diagnosis especially in rare disease and cancer.

Universitas YARSI is a renowned private university in Jakarta. YARSI has employed networks of international and national lecturers, won competitive grants in genomic and biomedical sciences. YARSI Genomic Facility has run two NextGen Sequencing platforms (both short and longreads supported by Illumina and Oxford Nanopore, respectively) to support training of graduate students in Genetic Counselling Master Program of Biomedical Sciences.

In connection with the development of genomic and genomic diagnosis facilities, Universitas YARSI will hold The 1st International Symposium and Workshop "Medical Genetics Update: From Genomic to Clinic" on May 16-18, 2024 in Jakarta, Indonesia in collaboration with several human genetics experts from abroad (Malaysia, Philippines, Thailand, Egypt and Tunisia). This activity is a forum for researchers to present the latest research results, to discuss, to improve participants' knowledge and skills in the health sector, especially in the fields of genetics at the cellular stage, molecular biology, genomics, clinical genetics, cancer, metabolic diseases and infectious diseases.

About Univeritas YARSI

Universitas YARSI (<http://yarsi.ac.id>) is an Indonesian premier research university which strives to enhance and strengthen its educational programs and has taken various initiatives to complement its educational excellence. Universitas YARSI started its history more than 50 years ago with medical school for undergraduate students. It now has 6 faculties: medicine, dentistry, law, economy, information technology and psychology. Additionally, it also established several post graduate programs: Master of Management, Master of Notary, Master of Biomedical Sciences, Master of Hospital Administration and of Biomedical Sciences. Setting the goal to be in 500 top universities in the world, it is now committed to research more than ever. YARSI research institute was established in 2012 to support its mission to be a top-notch research university. It consists of six core research centers: genetics, stem cell, herbal, telomere, e-health and halal research centers.

Symposium and Workshop Venue

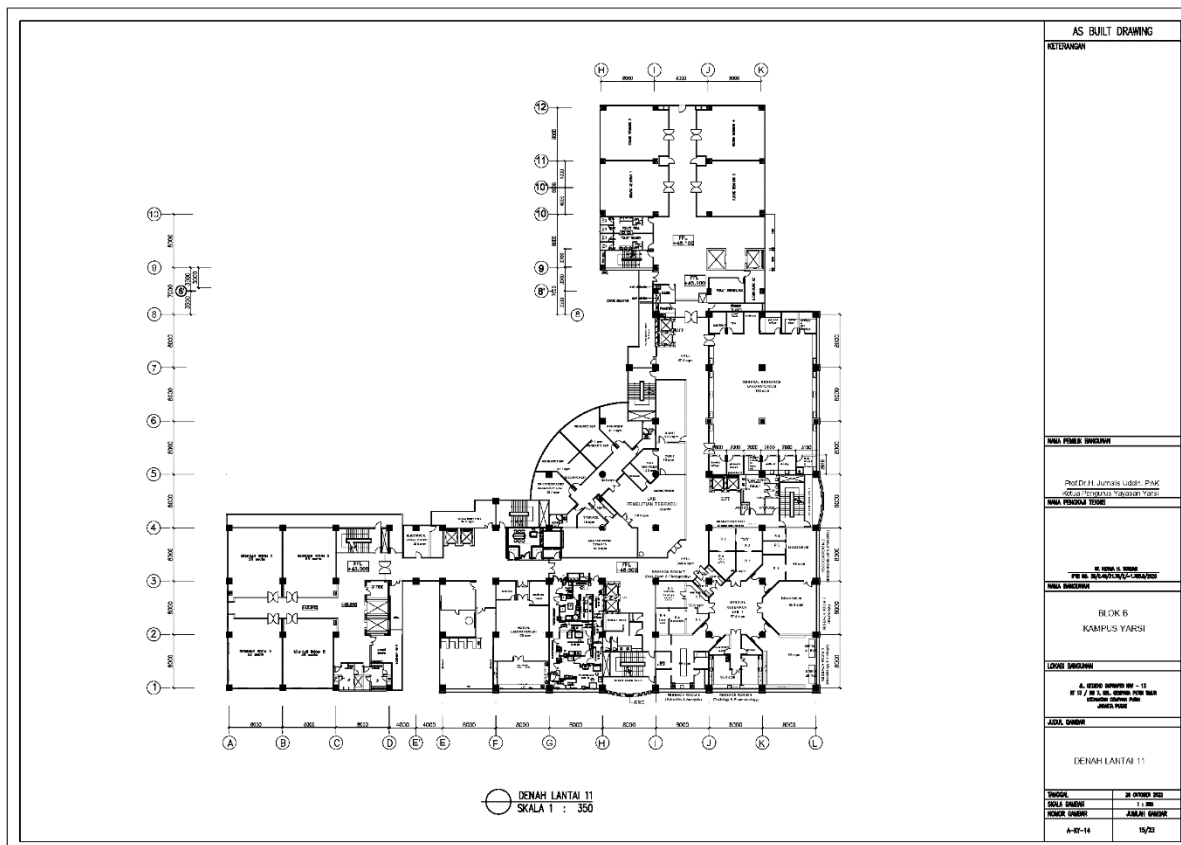
Symposium Venue – Ar – Rahim Hall



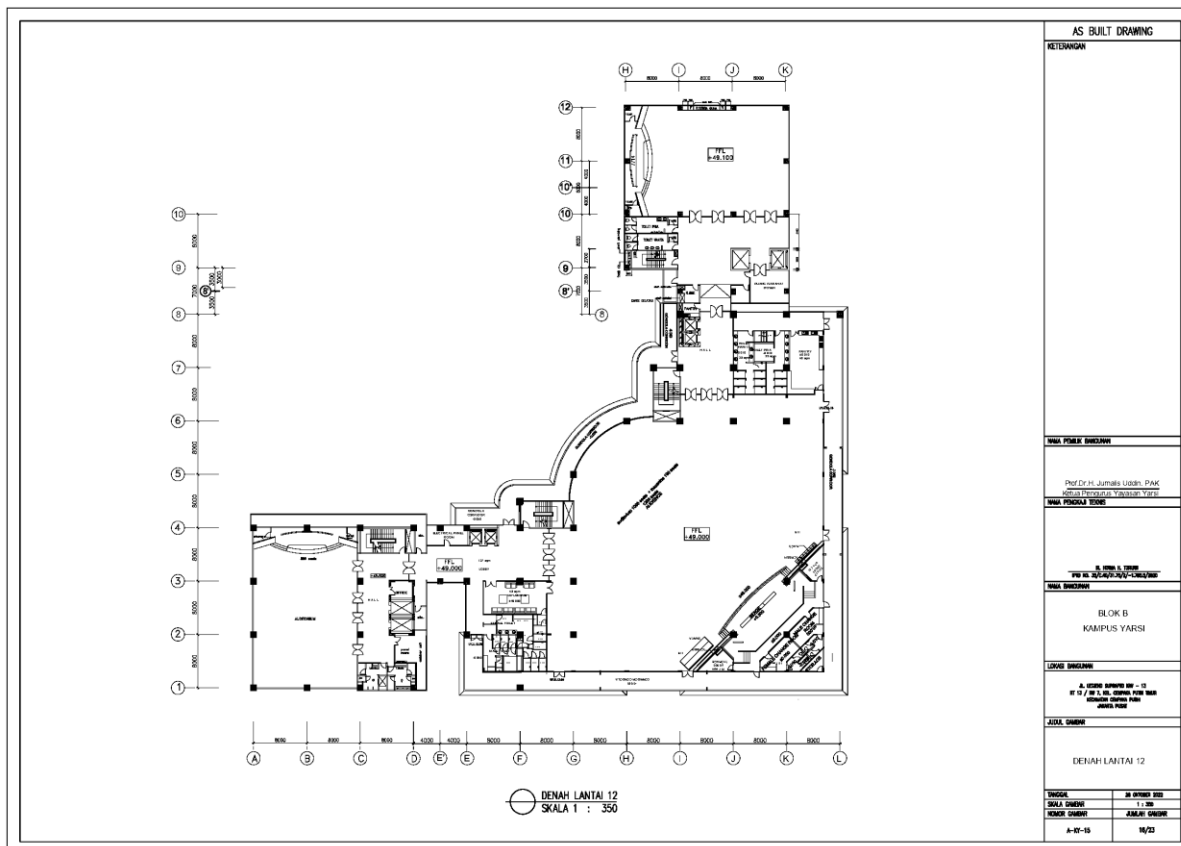
Workshop Venue – Workshop Room



11th Floor



12th Floor



Committee

Steering Committee:

1. Prof. dr. H. Jurnalís Uddin, P.A.K.
2. dr. Shanti Jurnalís, Sp.A., M.Kes.
3. Prof. dr. Fasli Jalal, Ph.D.
4. Prof. dr. Tjandra Yoga Aditama, Sp.P(K), DTM&H., MARS., DTCE., FISR.
5. Prof. dr. Sultana MH. Faradz, PAK, PhD
6. Prof. dr. Pratiwi Pujilestari Sudarmono, Ph.D, Sp.M.K(K)
7. Prof. Dr. dr. Ratna Sitompul, Sp.M, Subsp.I.I

Organizing Committee

Chairman	: Dr. dr. Wening Sari, M.Kes
Vice Chairman	: Dr. Dra. Ndaru Andri Damayanti, MSc
Secretary	: Nabila Pyrerina Thamrin, B.Eng, M.Eng Biomed, Ph.D
Treasurer	: 1. Dr. Juniarti, S.Si, M.Si 2. A'immatun Nadrifah, SE

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4. Nur Ilham Rishma, SKM, M.Biomed
5. dr. Sabrina Azmi M.Biomed
6. dr. Sri Rahayu M.Biomed
7. Dhana Putri Utami, S.DV

Symposium:

1. dr. Miranti Pusparini, M.Pd (Ked). (Coordinator)
2. dr. Yurika Sandra, M.Biomed.
3. dr. Aan Royhan. MSc
4. Silviatun Nihayah, SSi, M.Biomed
5. dr. Rika Ferlianti, M.Biomed
6. Salsha Nur Alfidza (MC-Master's student)
7. dr. Juliana Gozali (MC-Master's student)

Workshop:

1. Ahmad Rusdan Handoyo Utomo, Ph.D. (Coordinator)
2. Kinasih Prayuni, S.Si, M.Si
3. dr. Citra Fitri, Sp.K.J.
4. Dr. dr. Wan Nedra. SpA
5. Dr. dr. Elsyé, SpA
6. Intan Razari, S.Si

Scientific (Oral and Poster Presentation):

1. Dr. dr. Anggraini Iriani, Sp.PK, Subsp HK (K) (Coordinator)
2. dr. Dhinasty Armenia Wirakusumah Sp.PK

3. dr.Yenni Zulhamidah, M.Sc
4. Prof. Dr. Dra. Endang Purwaningsih, MS
5. Dr. Drs. Restu Syamsul Hadi, M.Kes.
6. Dr. Dra. Risdawati Djohan, MKes. Apt.
7. Dr. dr. Diniwati Muktar, M.Kes., AIFM.
8. Dr. Dra. Himmi Marsiati, M.S.
9. Dr. drh. Titiiek Djannatun
10. Dian Widiyanti, S.Si., M.Si., Ph.D
11. Dr. dr. Indra Kusuma, M.Biomed.
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13. Maya Genisa, S.Si., M.T., Ph.D.
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15. Harliansyah, PhD
16. Indra Bachtiar, Ph.D
17. Dr. dr. Fatimah Eliana, SpPD, KGH
18. Moch Abdussalam S.Si., M.Si

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5. Fahad ST
6. dr. Lusy Indranita
7. dr. Nurmayani
8. dr. Juliana Sp.PA
9. dr. Nursin Sp.PA
10. Djonady MSc

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1. Iman Dwi Almunandar, S.Psi. (Coordinator)
2. Sujasno SE
3. Riski Gemara, ST
4. Ratim
5. Ridho S. Farm

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1. Ambar Hardjanti, S.Si., M.Si (Coordinator)
2. Konconoviyati, S.Si., M.Biomed.
3. Ike Irmawati Purbo Astuti, S.Si., M.Si., Med.
4. Heni Mudiawati, ST
5. Habibah S.SM

Funding (Exhibition and Sponsorship):

1. Ahmad Rusdan Handoyo Utomo, Ph. D (Coordinator)

2. Dr. Drs. Restu Symasul Hadi, M.Kes
3. Dr. dr. Indra Kusuma, M.Biomed
4. Indra Bachtiar, Ph. D
5. Mohamad Agung Sadeli, S.Si
6. Fajar Rojak, S.Si
7. drh. Jasir Hakim Hidayah
8. Pendrianto, S.Si, M.Pharm

Schedule

Workshop

Session 1

Program: Tips-Tricks on Chromosome and Molecular Analysis on Genetic Disease

1. Dr. Aziati Azwari Annuar

Analysis and Clinical Application of Human Chromosomes and Cytogenetics in Medicine

2. Prof. dr. Sultana MH. Faradz, PAK, Ph.D

Case Discussion on Some Common Genetic Diseases: From Chromosomal to Molecular Diagnosis

Session 2

Program: Genetic Counseling Practice

1. Dr. dr. Nurin Listyasari, MSimed (KG)

2. Prof. dr. Sultana MH. Faradz, PAK, Ph.D

Sponsorship Speech

1. Dr. Seung Woo Ryu

Introduction of Variant Interpretation in Rare Diseases

Session 3

Program: How Do We Know Pathogenic Variants? (Cancer Cases)

1. Ahmad Rusdan Handoyo Utomo, Ph.D

2. Prof. dr. Sofia Mubarika Harjana M.Med.Sc,Ph.D

Session 4

Program: Resolving Issues on Hematologic Disorder: Case Discussion on Thalassemia and G6PD Deficiency

1. Prof.Dr. Zilfalil Alwi, MBBS, MMed, MSc, Ph.D, FAMM

Quick Method for Screening and Diagnosis Thalassemia

2. Prof. dr. Narazah Mohd Yusoff, MBBS, DCP, M.Med Sci, Ph.D

Diagnosis and Clinical Manifestation of G6PD Deficiency

3. Dr. dr. Anggraini Iriani, SpPK(K), Subsp HK (K)

State of The Art in Hematology Genetic Diagnosis

Symposium

Plenary Lecture Day 1

1. **Dr. Dra. Lucia Rizka Andalusia, Apt., M.Pharm., MARS** (Ministry of Health Republic of Indonesia)
Keynote speech
2. **Yosr Hamdi, Ph.D**
Human Genomics and Precision Oncology: Towards A Data Driven Healthcare System in LMICs
3. **Prof.Dr. Zilfalil Alwi, MBBS, MMed, MSc, Ph.D, FAMM**
Global Globin Network (GGN)
4. **Prof. dr. Pratiwi Pujilestari Sudarmono, Ph.D, Sp.M.K (K)**
The Prospect of Microbiome in Genetic Disorder

Scientific Symposium 1 Day 1

1. **Prof. Dr. Liza Sharmini Ahmad Tajudin, MBBS, MMed, Ph.D, FAM**
Personalized Medicine of Glaucoma
2. **Nur Aizati Athirah Daud, Ph.D**
Navigation Pharmacogenomics Practice in Malaysia: Addressing Realities and Challenges
3. **Prof. dr. Gunadi, Ph.D., Sp.BA, Subsp.D.A.(K):**
Personalized Medicine Development in Indonesia
4. **Prof. Ghada El-Kamah, MD, Ph.D:**
Precision Medicine and Genomic Education for Better Health Service in Developing Countries

Scientific Symposium 2 Day 2

1. **Prof. dr. Herawati Sudoyo, MS, Ph.D**
Indonesian Genome: Exploring the Rich Tapestry and Its Impact on Phenotypic Landscape
2. **Dr. Nazihah Mohd Yunus**
Genetic Disorders in North-Eastern Peninsula Malaysia: the USM Experience
3. **Prof. Thanyachai Sura MD, FRCP**
Rare and Undiagnosed Diseases: From Diagnosis to Management in Thailand

Scientific Symposium 3 Day 2

1. **Dr. Catherine Lynn T. Silao**
Screening of Rare Disease in The Philippines
2. **Prof. Dr. dr. Budi Wiweko, Sp. OG(K), MPH**
Prenatal Diagnosis in Monogenic Disorders
3. **Prof. dr. Sultana MH. Faradz, PAK, Ph.D**
Genetic Counseling in Infertility and Prenatal Diagnosis

Industrial Symposium Day 2

1. Vendor: **Illumina, Inc.**

Scientific Symposium 4 Day 2

1. Prof. dr. Narazah Mohd Yusoff, MBBS, DCP, M.Med Sci, Ph.D

Deciphering SKP2 Mediated Regulation of TERT in AML

2. Dr. Norafiza binti Mohd Yasin

Laboratory Approach for Thalassemia Genotyping

3. Prof. Dr. Raja Zahratul Asma Raja Sabudin

Molecular Analysis of G6PD Gene Mutations; the Malaysia Experience

Scope of Posters and Oral Presentations

1. Molecular Genetics and Techniques
2. Counselling Genetics and Social Aspects
3. Clinical Genetics
4. Immunology
5. Communicable and Non-Communicable Diseases
6. Health Technology and Medical Treatment
7. Pharmaceutical Sciences and Phytopharmacy
8. Health Promotion, Health Policy and Education
9. Mental Health
10. Environmental and Occupational Health
11. Sport Sciences and Physical Education
12. Cancer Biology and Cancer Therapeutics
13. Cellular and Molecular Research
14. Stem cell and Herbal medicine
15. Anti-aging
16. Reproduction
17. Others

Guidelines

Abstract

1. Abstract written using Microsoft Word, using Times New Roman font size 12 pt with 1.5 line spacing.
2. Consists of a maximum of 2 A4 pages.
3. Written in Indonesian or English.
4. The title consists of a maximum of 20 words.
5. Accompanied by 5-6 keywords.
6. The names of the authors are written in full, including the institution of origin and email for the corresponding author.
7. The abstract is divided into the following sub-headings:
 - Introduction
 - Method
 - Results
 - Conclusion
 - Bibliography

Posters

Posters displayed at the 1st International Symposium and Workshop are made with the following provisions:

1. Each participant can only bring one A1 poster size (width 594 mm x length 841 mm) which will be displayed vertically.
2. The poster must include the title, name of the researchers and logo of the Institution, background/ introduction /abstracts, methods, results (text and images / photography / schemes), conclusions, references (additions), sponsors / institutions (+ logos), contact details, the background should be short and straight to the purpose of the problem (purpose-method- Findings- Conclusions and Suggestions).
3. Posters should be made with using computer software (with graphs, tables, providing it with photography is highly recommended).
4. The minimum resolution of poster is 300 dpi.
5. The poster is printed by the participant and handed over on the morning of May 17, 2024 (first day of the 1st International Symposium).

Workshop Session 1 - Tips-Tricks on Chromosome and Molecular Analysis on Genetic Disease

Dr. Aziati Azwari Annuar (Human Genome Center, Universiti Sains Malaysia)

ANALYSIS AND CLINICAL APPLICATION OF HUMAN CHROMOSOMES AND CYTOGENETICS IN MEDICINE

Aziati Azwari Annuar

With the emergence of cutting-edge genetic technologies, the exploration of human chromosomes and cytogenetics has gained significance in the medical field. Cytogenetics are the study of chromosomes, which are long strands of DNA and protein that the genetic information in a cell. This process entails analyzing tissue, blood, amniotic fluid, or bone marrow samples in a specialized laboratory to identify any changes in chromosomes, whether numerical or structural, such as deletion, translocation, inversion, insertion, ring formation, or other abnormalities. The practice of cytogenetics demands expertise from trained medical laboratory professionals. The application of cytogenetics has expanded the scope of genetic disorder testing, encompassing prenatal screening, diagnosis of postnatal infants with syndromic characteristics, assessment of disorders of sexual differentiation (DSD), and plays a crucial role in the field of oncology, specifically in hematological malignancies and solid tumors. Detecting chromosomal anomalies early in prenatal stages allows for well-informed decision-making regarding pregnancy management and preparation for caring for infants with genetic disorders. Furthermore, it plays a vital role in determining gender assignment and managing individuals with DSD. The cytogenetic analysis of cancer cells aids oncologists and clinicians in predicting the prognosis of the disease, selecting appropriate treatment approaches, and monitoring treatment efficacy.

Prof. dr. Sultana MH. Faradz, PAK, Ph.D (Universitas YARSI)

CASE DISCUSSION ON SOME COMMON GENETIC DISEASES: FROM CHROMOSOMAL TO MOLECULAR DIAGNOSIS

Sultana MH Faradz

Genetic testing can help to identify the cause or confirm a diagnosis associated with developmental delay, intellectual disability, dysmorphic features and multiple congenital malformations. Many cases cannot be diagnosed in the past with only clinical symptom, dysmorphism and chromosome analysis. In low middle income countries (LMIC) such as Indonesia with lack of laboratories facilities have been still used cytogenetics testing as a routine testing for some syndromes especially for sex identification.

Since 2005 when genomic analysis has been started and developed rapidly, many genetic diseases including some syndromes and cancer as well as inherited or familial diseases can be diagnosed. Microdeletion syndromes as part of rare diseases such as Prader Willi syndrome, Cri du chat syndrome sometimes can be detected in chromosome analysis however most of them have to be confirmed with chromosomal microarrays. Di George syndrome which is the most common cases of microdeletion syndrome that cannot be detected using chromosome analysis now days the diagnosis easily established using chromosomal microarrays.

Disorders of Sex Development or Differences in sex development (DSD) are conditions with an atypical chromosomal, gonadal or phenotypic sex, which leads to differences in the development of the urogenital tract and different clinical phenotypes. Chromosomal DSD such as Klinefelter is quite common and well-known chromosomal DSD. Though the diagnosis of many DSD cases cannot be established using only chromosome analysis except for sex identification. Advanced molecular techniques either using Sanger sequencing or targeted gene panel using NGS will help the diagnosis and management. Some cases will be presented here.

Workshop Session 2 - Genetic Counselling Practice

Dr. dr. Nurin Listyasari, MSimed (KG) (RSAB Harapan Kita, Jakarta)

GENETIC COUNSELING PRACTICE

Nurin Aisyiyah Listyasari, Sultana MH Faradz

Genetic counselor is a healthcare provider that provide information about how a genetic condition could affect an individual or family, including the pattern of inheritance, interpret genetic tests and help estimate the risk of a disease. Genetic counseling in family with genetic diseases should be followed by establishing the pedigree, carriership testing, which is essential to find carriers in the extended family, to calculate recurrence risks and to do early prevention. A rare disease is a disease that affects a small percentage of the population. In the US affect about fewer than 200,000 people with 7000 forms of rare diseases. Rare diseases often caused by a gene mutation or pathogenic variant with the pattern of inheritance follow Mendelian manner or denovo, however many rare diseases is still unknown and need many years to establish the certain diagnosis.

We will present some rare diseases cases with inherited genetic diseases such as autosomal recessive, autosomal dominant and X—linked inheritance with the diagnosis from the simple testing and more complicated either by chromosomal and or advanced molecular testing. Participants will be invited to involve in the discussion for the typical dysmorphology of the syndromes, pattern of inheritance based on pedigree analysis, what kind of testing needed, pre and post testing counseling, variant interpretation, the possible diagnosis, risk of having more affected in the family and option for the further management.

Prof. dr. Sultana MH. Faradz, PAK, Ph.D (Universitas YARSI)

GENETIC COUNSELING PRACTICE

Nurin Aisyiyah Listyasari, Sultana MH Faradz

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Workshop Session 3 - How Do We Know Pathogenic Variants? (Cancer Cases)

Prof. dr. Sofia Mubarika Harjana M.Med.Sc,PhD (Universitas Gadjah Mada) & Ahmad Rusdan Handoyo Utomo, Ph.D (Universitas YARSI)

HOW DO WE KNOW PATHOGENIC VARIANTS? (CANCER CASES)

Sofia Mubarika Harjana & Ahmad R. Utomo

Biomedical Genome Science Initiative (BGSI) is Indonesian Ministry of Health to sequence 10K genome in variety of diseases including cancer. Nasopharyngeal cancer (NPC) is among the top five most common cancer cases affecting males in their productive age (median 40-50 years old). Although latent infection of Epstein Barr Virus (EBV) is an established risk factor in NPC, screening programme to detect NPC early has been elusive. Serological and nucleic acid-based detection methods have been proposed but not yet adopted widely in clinical practises. Recent data suggests that germline genetic variants in NPC patients with strong family history may also contribute to high risk of NPC occurrence. However, the interpretation of genetic variants being pathogenic or benign may be complicated in the absence of other data such as frequency of variants in general population, functional studies in vitro, computational prediction, and evidence of co-segregation of variants exclusively in affected family members.

Workshop Session 4 - Resolving Issues on Hematologic Disorder: Case Discussion on Thalassemia and G6PD Deficiency

Prof. Dr. Zilfalil Alwi, MBBS, MMed, MSc, Ph.D, FAMM (Universiti Sains Malaysia,
WHO TAG-G, Unesco Chair on Human Genetics of Thalassemia)

QUICK METHODS FOR SCREENING AND DIAGNOSIS OF THALASSEMIA

Bin Alwi Zilfalil

Thalassaemia, a group of inherited blood disorders characterized by abnormal haemoglobin production, present a significant global health burden. Early detection and accurate diagnosis are crucial for effective management and prevention of complications associated with the disease. Thalassaemia requires molecular analysis for predicting disease severity and ensuring accurate diagnosis, especially in cases with complex variant interactions. There are various molecular techniques for thalassemia detection, methods based on conventional Polymerase Chain Reaction (PCR) such as gap-polymerase chain reaction (gap-PCR), single-tube multiplex amplification refractory mutation system PCR (ARMS-PCR), multiplex ligation-dependent probe amplification (MLPA) and others. The drawback of conventional methods is long run times, low resolution, and the inability to detect certain mutations. However, the emergence of Next Generation Sequencing (NGS) technology has spurred its wider adoption in routine diagnostics of thalassaemia. NGS, whether through whole-genome sequencing, exome sequencing, or targeted gene-panel analysis, offers higher throughput and accuracy compared to conventional methods. It has significantly improved thalassemia diagnostics, detecting a higher percentage of carriers and enabling characterization of known and novel mutations across all globin genes. The integration of NGS with PCR-based methods has further enhanced variant detection, as demonstrated by recent studies screening for thalassemia variants. Despite its benefits, NGS requires substantial infrastructure and expertise, posing challenges for implementation in low- and middle-income countries (LMICs). Nevertheless, the combination of NGS with PCR-based techniques holds promise for improving at-risk couple detection at a relatively low cost. Continued efforts to overcome resource constraints and enhance accessibility will be critical for maximizing the potential of NGS in thalassemia screening and diagnosis on a global scale. In conclusion, the development of a rapid and reliable screening and diagnostic method for thalassemia represents a significant advancement in the field of haematology. By improving the diagnostic process and facilitating early identification of at-risk individuals, this approach holds promise for reducing the disease burden and improving patient outcomes on a global scale.

Prof. dr. Narazah Mohd Yusoff, MBBS, DCP, M.Med Sci, Ph.D (Advanced Medical and Dental Institute, Universiti Sains Malaysia)

Dr. dr. Anggraini Iriani, SpPK, Subsp HK (K) (Universitas YARSI)

STATE OF THE ART IN HEMATOLOGY MOLECULAR GENETIC DIAGNOSIS

Anggraini Iriani

Benign and malignant hematological disorders are heterogeneous in both biological and clinical aspects. The alterations of genomic profile associated with these diseases are complex and variable including mutations, translocations, karyotypic rearrangements and post-translational modifications. This evidence in association with the evolution of molecular techniques has led to a modification of the existing dogma focusing on a single gene or single pathway analysis. The development of new methods in molecular biology has not only allowed the individualized molecular diagnosis of diseases but has also led to the discovery of genetic or targeted therapeutic schemes. The concept of this review is to analyze the molecular basis of hematological diseases, as well as to present some new molecular technology and how they can affect overall survival. Using polymerase chain reaction (PCR), karyotype analysis, fluorescence in situ hybridization (FISH) and next-generation sequencing (NGS) it is possible to design better risk stratification categories and determine minimal residual disease (MRD). Immune check points inhibitors, antibodies and chimeric antigen receptor (CAR)-T cells can guide most efficient therapeutic strategies.

Plenary Talks

Yosr Hamdi, Ph.D (Institut Pasteur of Tunis, Tunisia, WHO TAG-G)

HUMAN GENOMICS AND PRECISION ONCOLOGY: TOWARDS A DATA DRIVEN HEALTHCARE SYSTEM IN LMICs

Yosr Hamdi

Major advances in genetics and recent growing availability of health data present an opportunity to make precise personalized patient care a clinical reality. The goal of precision medicine is to deliver the right treatment to the right patient at the right dose and the right time.

Here we are presenting our experience in applying research knowledge in the implementation of precision oncology in Tunisia, a North African country that represents a central hub of population admixture and human migration between African, European and Asian populations.

We started by characterizing the genetic architecture of the Tunisian population in the framework of several national and international pilot research projects starting with rare and monogenetic diseases and up to complex diseases such as cancer. We used traditional and next generation sequencing technologies (NGS) to identify the mutational spectrum of several hereditary diseases. Novel and specific genetic mutations have been identified for each disease and population-specific genomics databases were set up. Then, we identified the steps towards building sufficient capacity to effectively integrate genomic medicine and molecular diagnostics in clinical practice. Several training activities in bioinformatics, bio-statistics, NGS data analysis, Omics data annotation and interpretation have been carried as a part of the H3ABioNet and H3Africa projects. Finally, we promoted the precision oncology ecosystem by involving all engaged stakeholders including health care providers, clinicians, pathologists, radiologists, oncologists, academic researchers, bioinformaticians, diagnostic companies, policy makers and advocacy groups.

Currently, an Oncogenetics Diagnosis Unit is implemented at Institut Pasteur of Tunis for several cancer types including breast, ovarian, cervical, prostate, melanoma, pancreatic and colorectal cancers. For each cancer patient, a genetic and molecular profiling is performed to identify actionable biomarkers. Based on the profiling report, our oncologists individualize disease prevention, detection, diagnosis, treatment and management.

In conclusion, our experience highlights the importance of conducting innovative translational research to enable greater precision in disease prevention, diagnosis and treatment. However, the degree of clinical annotation, data interpretation and data sharing remain challenging. Additional efforts are now made to advance personalized medicine in patient care by educating consumers and providers, accelerating research and supporting necessary changes in policy and regulation.

Prof. Dr. Zilfalil Alwi, MBBS, MMed, MSc, Ph.D, FMM (Universiti Sains Malaysia, WHO TAG-G, Unesco Chair on Human Genetics of Thalassemia)

GLOBAL GLOBIN NETWORK (GGN)

Bin Alwi Zilfalil

The Global Globin Network (GGN), is a project-wide initiative of the Human Variome/Global Variome Project (HVP), was established as a collaborative effort to address the significant health burden posed by hemoglobinopathies, particularly thalassemia and sickle cell disease. These genetic disorders are highly prevalent, with approximately 80% of cases occurring in countries with low and middle incomes (LMICs). The urgency to combat the impact of these disorders on affected populations prompted the establishment of the GGN in 2015, which focuses on harnessing recent developments in human genomics and facilitating systematic data collection and sharing in LMICs.

The primary objective of the GGN is to provide support for countries facing the challenges associated with hemoglobinopathies. The network engages in various activities, including research collaboration, knowledge sharing, capacity building, and advocacy. One of the critical aspects of the GGN's work is its emphasis on collaborative research and knowledge exchange, which contributes to the identification of rare variants and a deeper understanding of genotype-phenotype correlations in diverse global populations. This knowledge, in turn, enables the development of personalized interventions for affected individuals.

To achieve its objectives, GGN collaborate with several organizations, including ITHANET, ClinGEN, INHERENT, and Global Variome. Through its collaboration with INHERENT, it aims to conduct a large, multi-ethnic genome-wide association study (GWAS) to explore the role of genetic modifiers in hemoglobinopathies. This collaboration addresses challenges from previous studies, such as small sample sizes and low statistical power, while promoting the participation of diverse populations worldwide.

In LMICs, the GGN extends its support to establish comprehensive variant databases to enhance diagnosis and management. The network facilitates research collaborations to develop innovative treatments and advocates for crucial measures such as newborn screening, prenatal counselling, and carrier screening to reduce the prevalence of thalassemia. By addressing these aspects comprehensively, the GGN aims to empower LMICs in their efforts to combat hemoglobinopathies.

In summary, the Global Globin Network plays a pivotal role in empowering LMICs to tackle the challenges caused by hemoglobinopathies. Leveraging cutting-edge genomics, the GGN enables a proactive approach to understanding, preventing, and managing these disorders, ultimately contributing to the improved health and well-being of affected populations worldwide.

Prof. dr. Pratiwi Pujilestari Sudarmono, Ph.D, Sp.M.K (K) (Universitas YARSI)

THE PROSPECT OF MICROBIOME IN GENETIC DISORDER

Pratiwi P. Sudarmono

The human microbiota comprises 10–100 trillion microorganisms including bacteria, viruses, protozoa, and fungi, among which bacteria are the most abundant, with a density of 10¹¹–10¹² cells/ml. More than 99% of the bacteria belong to the phyla Firmicutes, Bacteroides, Proteus, and Actinomycetes, whereas Firmicutes and Bacteroides are dominant in the gut flora of the healthy host.

The relationship between the microbiome and genetic disorders is a complex and evolving. Genetic disorders are caused by abnormalities or mutation in human DNA, causing a wide range of health condition, from relatively mild to severe. Research in this area are progressing very fast. One among others with increasing incidence is Inflammatory Bowel Disease (IBD), including Crohn's disease and ulcerative colitis.

Dysbiosis in the gut microbiome characterized by an imbalance of good and pathogenic microbes can trigger inflammatory responses in genetically susceptible individuals. The dysregulated immune responses in IBD patients also can be caused by microbial metabolites, such as short-chain fatty acids produced by gut bacteria.

The gut microbiota can decompose carbohydrates and indigestible oligosaccharides in food, synthesize short-chain fatty acids (SCFAs), such as butyric acid, propionic acid, and acetate, and provide abundant energy for the intestinal epithelium. Beneficial bacteria in the gut microbiota can play an immunosuppressive role by regulating host immune cells. Some harmful bacteria can also induce inflammatory cytokines by immune cell interactions or their metabolites to promote the intestinal damage. Herein, we review the pathogenic interaction of the microbial communities with the intestinal epithelial barrier, metabolome, and immune system of patients with IBD, and discuss the practical strategies used by microbiota-based therapies to treat IBD patients.

Scientific Symposium 1

Prof. Dr. Liza Sharmini Ahmad Tajudin, MBBS, MMed, Ph.D, FAM (Departement of Ophthalmology, Universiti Sains Malaysia)

PERSONALIZED MEDICINE OF GLAUCOMA

Liza Sharmini Ahmad Tajudin

Glaucoma is one of the main causes of irreversible blindness in the world. It is estimated that the prevalence of glaucoma is increasing significantly in Asia due to improvement in health care in Asia. Early detection and prompt management reduced the potential of blindness significantly. Currently, management of glaucoma is based on reduction of target pressure. However, there are many cases that achieved target pressure but still progress significantly.

Glaucoma is a group of complex diseases characterized by progressive optic neuropathy with specific visual field defect. Thus, so far there is no definitive pathogenesis of glaucoma. Genetics is believed to play an important role due to the strong evidence in those with family history of glaucoma. However, due to the complexity of the disease with the interplay of endophenotypes, there are no specific genes strong enough to associate with glaucoma. In this talk, the potential genetic markers will be discussed including potential markers for endophenotypes. The discussion on single gene such Myocilin to the future role of probability risk score will be highlighted with specific focus for patients in Asia.

Nur Aizati Athirah Daud, Ph.D (Human Genome Center, Universiti Sains Malaysia)

NAVIGATING PHARMACOGENOMICS PRACTICE IN MALAYSIA: ADDRESSING REALITIES AND CHALLENGES

Nur Aizati Athirah Daud

Pharmacogenomics, the study of how genetic variations affect an individual's response to drugs, holds immense promise for precision medicine. In Malaysia, the integration of pharmacogenetics/ pharmacogenomics into clinical practice presents both opportunities and challenges. This abstract explores the current landscape of pharmacogenomics practice in Malaysia, highlighting the research outcomes among this population, the realities faced by healthcare professionals and patients, along with the challenges encountered in implementing pharmacogenetic testing and interventions.

One of the foremost challenges is the limited awareness and understanding of pharmacogenetics among healthcare professionals. This lack of knowledge can lead to hesitance in incorporating genetic testing into routine clinical practice. Moreover, the cost and accessibility of genetic testing infrastructure and technologies pose significant hurdles in the widespread adoption of pharmacogenomics in Malaysia. The existing healthcare infrastructure may not be fully equipped to handle the implementation and interpretation of pharmacogenetics results, further complicating the integration process. There is also the need for standardization of evidence-based guidelines and protocols for pharmacogenetic testing and interpretation. Additionally, ethical considerations surrounding patient consent, data privacy, and the potential for genetic discrimination necessitate careful navigation in the incorporation of pharmacogenomics in clinical settings. These challenges must be addressed to ensure the successful implementation of pharmacogenomics in Malaysia.

Moving forward, addressing these realities and challenges requires a multifaceted approach. Key initiatives highlighted include the establishment of a clear policy agenda and strategic roadmap for precision medicine. Additionally, there is a need for collaborative efforts among stakeholders, including policymakers, healthcare providers, researchers, and industry partners, to address these challenges and facilitate the integration of pharmacogenomics into routine clinical practice.

Prof. dr. Gunadi, Ph.D., Sp.BA, Subsp.D.A.(K) (Universitas Gadjah Mada)

PRECISION MEDICINE ADVANCEMENT IN INDONESIA

Gunadi

Next-generation sequencing (NGS) has contributed to reducing the time and cost associated with genomic research and healthcare services, thereby fostering the growth of precision medicine.

During the COVID-19 pandemic, genomic technologies have played crucial roles in identifying pathogens, devising diagnostic and therapeutic strategies, and developing vaccines.

In 2022, the Indonesian Ministry of Health initiated the Indonesian Precision Medicine Initiative under the Biomedical and Genome Science Initiative (BGSi) to advance precision medicine in the country. It seeks to bolster diagnostic capabilities through NGS technology, determine personalized treatment, and assess the disease risks using polygenic risk scores. Indonesia, a developing nation comprising around 1,340 ethnic groups spread across more than 17,000 islands, faces unique challenges in implementing precision medicine due to its diverse demographics and geography.

One of the remarkable advancements of precision medicine in Indonesia is diagnosing genetic rare diseases using genomic testing and the possibility of clinical trials of personalized treatment for rare diseases. As one of the genomic hubs for the rare disease of BGSi, Dr. Sardjito Hospital has a potential clinical trial of gene therapy or PPMO for Duchenne muscular dystrophy.

Scientific Symposium 2

Dr. Nazihah Mohd Yunus (Human Genome Center, Universiti Sains Malaysia)

GENETIC DISORDERS IN NORTH- EASTERN PENINSULA MALAYSIA: THE USM EXPERIENCE

Nazihah Mohd Yunus

North-Eastern region of Peninsula of Malaysia covers the states of Kelantan, Terengganu and Pahang, which predominantly consists of Malays. Other races are Chinese, Indian and Siamese. Human Genome Centre (HGC) which is located in Kubang Kerian, Kelantan has been established in 2001 as a centre for diagnostic services, research innovation as well as teaching and learning for undergraduate and postgraduate students. It was known as Medical Genetic Unit since 1994. As the main centre and laboratory for genetic testing, HGC receives samples for diagnostic tests from North-Eastern of Peninsula of Malaysia for genetic disorders. Commonly genetic disorders cases receive in this region include trisomies, disorders of sex development, infertility, recurrent miscarriage as well as haematological disorders and solid tumor (breast cancer). Genetic testing that are been offered by HGC encompassing of cytogenetic, molecular cytogenetic and molecular genetic tests. HGC has been actively in service since for more than 2 decades, covering patient samples from Hospital Universiti Sains Malaysia (Hospital USM) as well as from local Ministry of Health as well the private hospitals, mainly from North-Eastern Peninsula Malaysia. HGC received different types of samples ranging from peripheral blood, bone marrow to tissue. For the past 20 years, a variety of cases have been diagnosed in HGC and published by the staff and researchers in reputable journals. HGC also actively collaborates with clinicians from various background including obstetrics and gynecology, pathology and pediatrics, which enable comprehensive analysis and diagnosis of the patient. From this, HGC has established collaborations with internal and external collaborators or laboratories for diagnostic services, research and medical genetic program. In conclusion, HGC has grown as one of genetic service providers in Malaysia and the only laboratory which provides the genetic testing in North- Eastern Peninsula Malaysia.

Prof. Thanyachai Sura MD, FRCP (Department of Internal Medicine, Mahidol University, Thailand)

RARE AND UNDIAGNOSED DISEASES: FROM DIAGNOSIS TO MANAGEMENT IN THAILAND

Thanyachai Sura

The global average prevalence of rare disease (RD) is 1 in 2,500 people (Richer et al., 2015). More than 80% of RD are inherited and can manifest in adults, some are nonspecific leading to a major reason why rare diseases go undetected. RD can affect any organ system and clinical manifestations could vary from those of acute life-threatening situation to subacute and chronically ill. Disease progression causes wide range of effects, with rapid life threatening deteriorate on over hours, episodic with intermittent decompensations and asymptomatic intervals, or insidious with slow degeneration over decades. Adult physicians who are interested in RD and inborn error of metabolism (IEMs) are faced with the fact that most if not all existing books and diagnostic algorithms refer to pediatric forms of these diseases. Some of the patients were classified into undiagnosed diseased (UND) group which are having limitation for diagnosis in many country members. More than 50% of these patients are adult form of RD in which many of them are ultrarare. A portion of the population cannot access RD diagnosis due to the lack of funds and specialized resources and/or limits in the organization/coordination among the existing centers. Clinical presentations and clues for diagnosis may take time, 1–6 years in average, in which some affected organs already passed the point of no return. With the development of biomedical technologies, the possibility of RD treatment has been established which is proved to reduce both the morbidity and mortality of RD.

A total number of 185 unconfirmed diagnosis patients, with range from 4-10 years follow-up, at our Adult Medical Genetics clinic. All of these patients were having thoroughly investigations and consultations, then considered for the corresponding gene panels study and whole exome sequencing. With the genomic study we could identify 148 confirmed diagnosis. Twelve patients were identified with lysosomal storage diseases (LSD) with low level of enzyme essay study. A number of 120 of the identified patients are diagnosed and have been treated, but 107 of these group already have the complications in multisystem. Twenty -eight of them get the confirmed diagnosis with no treatment available or continue the surveillance program.

Scientific Symposium 3

Dr. Catherine Lynn T. Silao (National Institutes of Health, University of the Philippines)

SCREENING OF RARE DISEASES IN THE PHILIPPINES

Catherine Lynn T. Silao

Rare diseases or orphan disorders are life-threatening or chronically debilitating diseases, which are of low prevalence that special combined efforts are needed to address them. The term orphan disorder describes a rare disease whose rarity results in little or no funding nor research for treatments, without financial incentives from the government or other agencies. In the Philippines, a disease is considered rare when it affects one patient in every 20,000 people. The thousands of rare diseases identified globally are typically genetic in origin. These diseases affect children who are often at risk of delayed diagnosis or are even misdiagnosed due to poor understanding of their condition. Patients afflicted with rare diseases often have their quality of life reduced and need lifelong medical care varying between medications and multidisciplinary therapies to alleviate the symptoms and effects of the disease. The lack of public awareness and a lack of existing network of support have affected Filipino patients afflicted with these disorders. Medical assistance is hard to get hold of under the conditions of the country's health priority, and raising awareness for people affected by rare disease still prevails.

Newborn screening (NBS) is a national public health program for the early identification of disorders. It describes various tests that are done during the first few hours or days of a newborn's life thereby preventing severe health problems including death. The Philippine NBS program, that started as a research in 1996 with a screening panel of 6 disorders, has since expanded to more than 28 additional hereditary disorders including thalassemias. As of 2022, around 17 million babies have been screened with 294,130 babies confirmed positive for the different disorders included in the panel. The integration of NBS in the public health system has been an opportunity for introduction of genetic services, especially with the regional comprehensive follow-up/ treatment centers and the implementation of expanded screening, in the different regions of the country. Early detection has allowed treatment to begin immediately, which reduces or even eliminates the effects of the condition.

dr. Mila Maidarti, SpOG-KFER, Ph.D (Universitas Indonesia)

PRENATAL DIAGNOSIS IN MONOGENIC DISORDERS

Budi Wiweko, Mila Maidarti

Monogenic disorder is a disorder that occurs because of single gene mutation. It could be autosomal or sex-linked and could occur naturally or due to environmental changes. At least 80% of all rare diseases are caused by more than 4000 monogenic mutations. From more than 10000 monogenic disorders, around 17% are neurological. One of the solutions for this is prenatal diagnosis. Some of the monogenic disorders that could be detected through prenatal diagnosis includes, but not limited to cystic fibrosis, Duchenne Muscular Dystrophy, and Huntington's disease.

Prenatal diagnosis is a medical technique used to determine whether a fetus has a genetic disorder or other abnormalities before birth. This technique is commonly used for genetic diseases that cannot be cured and where termination becomes a consideration, or used in cases that require immediate prenatal management and where maternal complications or fetal mortality are of concern. Another indication for prenatal diagnosis for monogenic disorders is when the parents have a history of severe genetic disease within the family. The results of the prenatal testing may provide important information to guide clinicians in the clinical management.

Prenatal diagnosis can be performed through invasive and non-invasive methods. Invasive methods, such as amniocentesis and chorionic villus sampling (CVS), poses a risk of fetal damage or death. Non-invasive approaches, like ultrasound imaging, are not yet accurate enough for genetic disease diagnosis and still require fetal sampling for confirmation. Recent advancements involve non-invasive sampling of fetal cells, DNA, and mRNA from maternal circulation, offering potential clinical applications.

One of the applications that has been widely used globally is the Pre-implantation Genetic Testing for Monogenic Disorder (PGT-M) which is used to identify single pathogenic variants within the embryos before implantation in IVF patients. PGT-M is performed by taking a few trophoblast cells from the embryo through biopsy at day 5-6 which is then analyzed. PGT-M differs from PGT-A, in which it identify specific single-gene disorders as compared to testing for the correct number of chromosomes in PGT-A

Prof. dr. Sultana MH. Faradz, PAK, Ph.D (Universitas YARSI)

Genetic Counseling in Infertility and Prenatal Diagnosis

Sultana MH Faradz

Genetic counseling service is not yet common in Indonesia, but the awareness on understanding genetic disease and its management is slowly emerging. Genetic counseling is a branch of medical genetics that is needed in the management of every patient with genetic problem. Genetic counselors work as members of a health care team, which provides information and support to families with a history of genetic disorders and may be at risk for such conditions. When obstetrician refer a detected fetal condition, genetic counselor will provide information and support for the patient' decisions regarding genetic testing in pregnancy.

The main symptom of infertility is an inability to get pregnant. The cause of infertility may include inadequate levels of certain hormones in both men and women, trouble with ovulation in women and about 50% may cause by genetic defect such as structural chromosome abnormalities, aneuploidy mosaicism and single gene disorders.

Prenatal screening tests for pregnant women with history of genetic disorders or infertility can identify whether the baby is more or less likely to have certain birth defects. These tests include blood tests for chromosome and DNA analysis, a specific type of ultrasound, maternal serum screening, noninvasive prenatal cell-free DNA screening and invasive prenatal testing namely amniocentesis or chorialis villus biopsy. It is important to inform to the patients which test is only a high risk or prediction of an abnormal baby and which one is actually to established the diagnosis either chromosomal aberration or single gene disorders. So that the decision of termination pregnancy can be made precisely and without ethical dilemma.

Scientific Symposium 4

Prof. dr. Narazah Mohd Yusoff, MBBS, DCP, M.Med Sci, Ph.D (Advanced Medical and Dental Institute, Universiti Sains Malaysia)

DECIPHERING SKP2 MEDIATED REGULATION OF TELOMERASE REVERSE TRANSCRIPTASE (TERT) IN ACUTE MYELOID LEUKEMIA (AML)

Narazah Binti Mohd Yusof

Leukaemia in Malaysia is ranked as the ninth (9th) most common cancer among the Malaysian population with 1,905 cases and 1,377 deaths. Acute myeloid leukemia (AML) is one of the most prevalent types of leukemia. AML is characterized by the uncontrolled proliferation of immature myeloid blast cells.

Overexpression of TERT expression and telomerase activity can be seen in haematological malignancies including leukemogenesis. Previous studies have shown that prolonged suppression of SKP2 and AML1/ETO led to TERT down-regulation in AML t(8;21)

Nevertheless, the mechanism by which SKP2 regulates TERT is currently unknown. AML1/ETO was also found to regulate TERT via SKP2/CDKN1B axis with Rb and E2F1 involvement in AML t(8;21). c-Myc is known as a transcriptional activator of TERT in cancer cells including leukemia. This project aimed to elucidate the mechanism of TERT regulation by SKP2 in AML.

Dr. Norafiza binti Mohd Yasin (Institute for Medical Research, Setia Alam, Selangor, Malaysia)

LABORATORY APPROACH FOR THALASSAEMIA GENOTYPING

Norafiza Mohd Yasin

Thalassaemia is an inherited blood disorder that is most frequently found in Southeast Asian populations. Conventional molecular diagnosis is routinely performed for the prevention and control of thalassemia. The majority of laboratories focus on identifying common mutations, involving HBA and HBB genes. Although most patients are diagnosed with a common thalassaemia panel, a few of them are still unknown. Therefore, to increase the effectiveness of the prevention and control program for severe α and β -thalassemia in this region, in addition to common thalassemia mutations, rare or unknown mutations should also be identified. Several advanced techniques such as multiplex ligation-dependent probe amplification (MLPA) and sanger sequencing were developed for comprehensive thalassaemia genotyping. Next-generation sequencing (NGS) application in thalassemia genotyping is limited and has just recently emerged due to current demands in seeking alternative DNA screening tools that are more comprehensive, efficient, versatile, and cost-effective. This review aims to understand the approach for thalassaemia genotyping, including the most current and expanding technology uses, advantages, and limitations, along with the issues and solutions related to its integration into routine screening and diagnosis of thalassaemias. Comparison between conventional techniques with NGS will be discussed.

Prof. Dr. Raja Zahratul Asma Raja Sabudin (Departement of Pathology, Universiti Kebangsaan Malaysia)

MOLECULAR ANALYSIS OF G6PD GENE MUTATIONS: THE MALAYSIA EXPERIENCE

Raja Zahratul Asma Raja Sabudin

G6PD deficiency is the most common enzymopathy, leading to alterations in the first step of the pentose phosphate pathway, which interferes with the protection of the erythrocyte against oxidative stress and causes a wide range of clinical symptoms of which hemolysis is one of the most severe. The G6PD deficiency causes several abnormalities that range from asymptomatic individuals to more severe manifestations that can lead to death. It is estimated 500 million people affected globally. In Malaysia, G6PD deficiency is one of the commonest causes of neonatal jaundice with incidence rate of 5.1% and 5.5% among male neonates of Malay and Chinese origins respectively. Nowadays, only 9.2% of all recognized variants have been related to clinical manifestations. WHO working groups have classified G6PD variants based on enzyme activity into five classes with thresholds of 10–60% as a defined level of G6PD activity. More than 180 G6PD deficiencies have been identified at the DNA level and over 400 variants described based on biochemical properties. It is important to understand the molecular basis of G6PD deficiency to understand how gene mutations can impact structure, stability, and enzymatic function. Variation in the enzyme activity of G6PD deficiencies has an impact on malaria treatment, because antimalarials such as primaquine, sulfanilamide and sulfadoxine have been observed to cause hemolysis in G6PD deficient individuals. In Malaysia, a total of 16 known mutations have been identified and most recently two novel mutations have been found and were evaluated for biochemical and structural properties. Many types of PCR methods have been used to identify those mutations and most recently in UKM Medical Centre, PCR-based reverse dot blot flow through hybridization method that simultaneously screens 14 G6PD mutations using the HybriBio G6PD deficiency Gene Diagnostic Kit. (Chaozhou HybriBio Biochemistry Ltd, China) has been optimized. While in Clinical Genetics Unit, Advanced Diagnostics Laboratory, Advanced Medical and Dental Institute, Universiti Sains Malaysia successfully diagnosed a patient with chronic non spherocytic haemolytic anaemia using next-generation sequencing (NGS) and identified two heterozygous missense variants in G6PD: Canton c.1376G>T, p.(Arg459Leu) and Kaiping c.1388G>A, p.(Arg463His) mutations.

Oral Presentation Abstract

Genetics

Oral ID – O.G-01

AMENORRHEA ASSOCIATED WITH TURNER SYNDROME AND MULLERIAN AGENESIS: A RARE CASE REPORT

Ria Margiana,^{1,2,3} Maria Paulina Budyandini Dyah Pramesti^{1,2}, Djonady Sugiaman⁴

¹ Andrology Program, Faculty of Medicine, Universitas Airlangga, Surabaya, Indonesia.

² Dr. Soetomo General Academic Hospital, Surabaya, Indonesia

³ Department of Anatomy, Faculty of Medicine, Universitas Indonesia, Jakarta, Indonesia

⁴ Biomedical Science Program, Major in Genetic Counseling, Post Graduate School,
Universitas YARSI, Jakarta, Indonesia

*Corresponding author: pramestidyan@gmail.com

Abstract

Introduction

Turner syndrome is a genetic disorder that only affects females and is defined by the whole or partial loss of one X chromosome. Mullerian duct agenesis is a condition in which the reproductive system of females fails to develop normally during development. The objective of this case study is to enhance comprehension regarding the diagnosis and treatment of patients presenting with Turner syndrome and Mullerian duct agenesis.

Material and Methods

The present study used a case report methodology. A female patient, aged 20, presented at the hospital with a medical ailment referred to as primary amenorrhea. Additional assessment raised concerns about Turner syndrome accompanied by Mullerian duct agenesis. A karyotyping analysis was conducted to verify the diagnosis. The patient was provided with instruction regarding her medical conditions and received suitable treatment. Turner syndrome, characterized by the absence of one X chromosome in females, often presents with Mullerian duct agenesis, a condition where the structures that grow into the uterus and fallopian tubes fail to form properly.

Result and discussion

This combination of conditions is considered a rare developmental abnormality. Diagnosis necessitates a comprehensive assessment that involves multiple medical examinations, including karyotyping. Effective management entails providing knowledge about the disease and implementing suitable measures to avoid any potential difficulties.

Conclusions

Turner syndrome, accompanied by Mullerian duct agenesis, is an uncommon illness that necessitates precise diagnosis and well-coordinated treatment. The process of karyotyping is an essential and decisive measure in verifying the diagnosis. Effective education and assistance are crucial for aiding patients in effectively managing their disease and mitigating any severe problems in the future.

Keywords:

Turner syndrome, ductus Muller agenesis, amenorrhea, uterus, reproductive health

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Oral ID – O.G-02

A MOLECULAR GENETIC INVESTIGATION INTO THE *SRD5A2* VARIANTS IN AN INDONESIAN POPULATION WITH HYPOSPADIAS

Rizki Diposarosa^{1,2}, Yunisa Pamela^{2,3}, Herry Herman⁴, Sjarif H Effendy^{2,5}, Yunia Sribudiani^{2,3*}

¹Department of Surgery, Division of Pediatric Surgery, Dr. Hasan Sadikin General Hospital, Faculty of Medicine, Universitas Padjadjaran, Bandung, Indonesia, ²Research Center of Medical Genetics, Faculty of Medicine, Universitas Padjadjaran, Bandung, Indonesia, ³Department of Biomedical Sciences, Faculty of Medicine, Universitas Padjadjaran, Bandung, Indonesia, ⁴Department of Surgery, Division of Orthopedic Surgery, Hasan Sadikin Hospital, Faculty of Medicine, Universitas Padjadjaran, Bandung, Indonesia, ⁵Department of Pediatric, Hasan Sadikin Hospital, Faculty of Medicine, Universitas Padjadjaran, Bandung, Indonesia

*Corresponding author: y.sribudiani@unpad.ac.id

Abstract

Introduction

Hypospadias, characterized by an abnormal opening of the urethra on the ventral side of the penis, is one of the most common congenital anomalies of male external genitalia. Hypospadias present either as an isolated condition, or as a syndromic condition accompanied by other phenotypes. *SRD5A2* encoding steroid 5 alpha-reductase type 2 protein has been known to be linked with hypospadias. This study describes a complete molecular analysis of *SRD5A2* variants associated with the risk of hypospadias.

Materials and Methods

Peripheral blood DNA was isolated from thirty-seven isolated and thirty-seven syndromic hypospadias patients. Polymerase chain reaction (PCR) was conducted to amplify all exons and exon-intron boundaries of *SRD5A2*, followed by Sanger sequencing. Analyses of mutation and polymorphism were performed using BioEdit. In silico analysis was conducted using Mutation Taster®, Polymorphism Phenotyping-2 (PolyPhen-2) and Sorting Intolerant from Tolerant (SIFT). Statistical analysis was done using Pearson's chi-squared test.

Results and Discussion

In one patient with isolated hypospadias, a novel missense mutation c.32T>C/p.Leu11Pro was identified in *SRD5A2* and in silico analysis predicted this mutation to be pathogenic. Three polymorphisms were identified in *SRD5A2*: rs632148 (c.-62G>C) and rs522638 (c.281+15T>C) found in the non-coding region and rs523349 (c.265C>G/p.Val89Leu) found in the exon 1. Among these variants, the mutant alleles significantly increased the risk of having syndromic hypospadias more than the isolated one with relative risk (RR) ratios of 1.97, 1.78 and 1.77 respectively (p<0.005).

Conclusions

In this study, several variants in *SRD5A2* were identified in an Indonesia population with hypospadias. The novel mutation c.32T>C/p.Leu11Pro was predicted to be pathogenic while three polymorphisms (rs632148, rs523349 and rs522638) were found to be associated with an increased risk for developing hypospadias in a syndromic form rather than the isolated form.

Keywords:

Hypospadias, mutation, polymorphism, *Steroid 5 alpha-reductase type 2*, urethra

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Oral ID – O.G-03

THE POTENTIAL CLINICAL UTILITY OF MOLECULAR GENETIC TECHNOLOGIES IN UNVEILING THE SPECTRUM ETIOLOGY OF MISCARRIAGES: A SYSTEMATIC REVIEW

Ilham Utama Surya*, Muhammad Dwi Priangga, Riyan Hari Kurniawan, Cepi Teguh Pramayadi, Herbert Situmorang, Eka Rusdianto Gunardi

Kintani Health Reproduction Integrated Unit, Dr Cipto Mangunkusumo National Hospital, Jakarta, Department Obstetric and Gynecology, Dr Cipto Mangunkusumo National Hospital, Medical Faculty Universitas Indonesia

*Corresponding author: ilham.surya@ui.ac.id

Abstract

Introduction

Karyotyping is well recognized as one of the examinations to detect genetic disorder with a resolution greater than 5 Mb and can identify the genetic causes of spontaneous abortion in 49% of cases. A more advanced molecular genetic testing, such as next generation sequencing (NGS), offers an appealing alternative to conventional karyotyping as it possesses a resolution of >100 Kb. Thus, NGS holds the potential to reveal genetic variations associated with miscarriages. Therefore, we conducted review studies the additional diagnostic yield of NGS in miscarriage.

Materials and Methods

We conducted literature search on PubMed for studies published between 2016-2023 using the query “abortion”, “pregnancy loss”, “miscarriage”, and “next generation sequencing”. From 13 obtained articles, 7 articles were included in this study. We categorized genetic disorders into distinct groups such as aneuploidy, polyploidy, copy number variant (CNV), and mosaicism. To identify the CNV, we mapped the affected chromosomes using UCSC genome browser GRCh37/hg19. The classification of CNV adhered to ACMG guidelines.

Results and Discussion

The overall diagnostic rate of NGS was 59%. Among the genetic causes of miscarriage, aneuploidy accounted for 67%, CNV for 24%, mosaicism for 5%, and polyploidy for 3%. We also found trisomy 16 and monosomy X to be the most frequent causes of aneuploidy. Microdeletions in 1p36.2 and microduplication 21q22.3 were the predominant CNV. Furthermore, trisomy 16 and 22 were the most affected form of mosaicism.

Conclusions

The diagnosis yield of NGS in miscarriage was 59%. The etiological spectrum of miscarriages encompassed aneuploidy, CNV, mosaicism, polyploidy, and mosaicism.

Keywords:

Abortion, miscarriage, pregnancy loss, next generation sequencing, genetic

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THE ROLE OF DNA METHYLATION ON ISCHEMIC STROKE OUTCOME

Loury Priskila

Biochemistry Department, Faculty of Medicine Duta Wacana Christian University
Jl. Dr. Wahidin Sudirohusodo no. 5-25 Yogyakarta 55224, Indonesia

*Corresponding author: loury@staff.ukdw.ac.id

Abstract

Introduction

Epigenetic variation is a phenomenon that causes modifications to the genome without disrupting the DNA sequence but can affect the development of a disease. One of the mechanism of epigenetic is DNA methylation which is an epigenetic modification to genes will affect the structure of DNA as well as DNA expression.

Material and Methods

This study is a literature review using Pubmed and Google Scholar. The keywords used were 'DNA methylation' and 'stroke outcome' from 2018 until 2023.

Result and Discussion

DNA methylation is associated with transcriptional inactivation, this is caused by the addition of methyl groups to the CpG Island and causes inhibition of DNA transcription mediated by the enzyme DNA methyltransferase (DNMT). DNA methylation in LINE-1 will cause deregulation of the LINE-1 gene, which can affect the metabolism of lipids and carbohydrates, which can affect the metabolic system. In chronic conditions, this will increase the levels of vascular cell adhesion molecular (VCAM-1), which will also play a role in BBB permeability and leukocyte recruitment. DNA methylation of EXOC4 is associated with the modulation of pathways leading to endocytosis and NK cell regulation, which will cause changes in NIHSS scores. Hypermethylation of Brain-Derived Neurotrophic Factor (BDNF) in the promoter region will be associated with long-term outcomes related to physical disability, cognitive function.

Conclusion

DNA methylation strongly influences the inflammatory promoter genes. It also can trigger inflammation with various pathways such as increased platelet aggregation, activation of inflammatory pathways, recruitment of NK cells and so on, positively correlating with worsening stroke outcomes.

Keywords

Epigenetic, DNA Methylation, Stroke Outcome.

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DIAGNOSTIC SIGNIFICANCE OF CIRCULAR RNA IN PULMONARY ARTERIAL HYPERTENSION: SYSTEMATIC REVIEWFajri Marindra Siregar^{1,2}, Jajah Fachiroh³, Anggoro Budi Hartopo⁴, Sofia Mubarika Haryana³

¹Doctorate Program of Medical and Health Science, Faculty of Medicine, Public Health and Nursing, Universitas Gadjah Mada, Yogyakarta, Indonesia, ²Department of Biochemistry, Faculty of Medicine, Universitas Riau, Pekanbaru, Indonesia, ³Department of Histology and Cell Biology, Faculty of Medicine, Public Health and Nursing, Gadjah Mada University, Yogyakarta, Indonesia, ⁴Department of Cardiology and Vascular Medicine, Faculty of Medicine, Public Health and Nursing, Gadjah Mada University–Dr. Sardjito Hospital, Yogyakarta, Indonesia

*Corresponding author: fajrifkunri@gmail.com / fajrimarindrasiregar@mail.ugm.ac.id*Abstract**Introduction*

Documenting the global incidence of pulmonary arterial hypertension (PAH) is still challenging due to the requirement of invasive modalities. The number of published circular RNA (circRNA) research has increased significantly in recent years, making circRNAs some of the most renowned molecules in RNA biology. CircRNA concentrations are stable under normal conditions in biological fluids; hence, they may serve as diagnostic biomarkers. This systematic review aims to provide a concise summary of the diagnostic significance of circRNA in PAH.

Material and Methods

A systematic search was conducted in PubMed, SCOPUS, Clinical Key, and BiomedCentral databases to identify relevant observational studies that measured circRNA expression in PAH. The search was limited to publications published in English up to July 2022. The QUADAS-2 tools were utilized to evaluate the methodological quality of the original study. The assessment of publication bias was conducted through the utilization of Egger's test. The data synthesis was performed using MedCalc. This systematic review adheres to the PRISMA guidelines. The review protocol was registered in the PROSPERO database (reference CRD42022360617).

Results and Discussion

Ten different forms of circular RNA were found to be dysregulated, with six exhibiting upregulation and four demonstrating downregulation, based on the evaluation of seven articles in the final stage (out of 483 titles initially). The diagnostic biomarker circRNA exhibits a sensitivity of 61.9%-90.48% and a specificity of 60.0%-98.78%. The area under the receiver operating characteristic curve (AUC) for circRNA ranges from 72.1% to 94.1%. CircRNAs have closed-loop structures with covalent bonds and don't have 5'-3' polarities or polyadenylated tails. This makes them stable, specific, and conservative. Due to their unique properties, circRNAs are superior to other types of RNAs for diagnosing certain disorders.

Conclusions

These findings suggest that circRNA, as reported, shows promise as a diagnostic biomarker.

Keywords

Biomarker, circular RNA, diagnostic value, pulmonary arterial hypertension

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CHROMOSOMAL ABNORMALITIES IN PRE AND POSTNATAL COHORT: A SINGLE CENTER STUDY IN WEST JAVA

Almira Zada^{1,2}, Bremmy Laksono², Riksa Parikrama², Nur Amalia Rosyada², Merry Afni², Ida Parwati^{2,3}

¹Department of Biomedical Sciences, Faculty of Medicine, Universitas Padjadjaran, Bandung, Indonesia, ²Biomedical Laboratory, Faculty of Medicine, Universitas Padjadjaran, Bandung, Indonesia, ³Department of Clinical Pathology, Faculty of Medicine, Universitas Padjadjaran, Bandung, Indonesia

*Corresponding author: a.zada@unpad.ac.id

Abstract

Introduction

Chromosomal analysis or karyotyping is a laboratory technique used to examine the chromosomes of an individual, offering insights into chromosome numbers, structures, and arrangements to diagnose genetic diseases. Our study aims to investigate the distribution by indications and the frequency of chromosomal abnormalities in different clinical populations in West Java.

Materials and Methods

This is a cohort retrospective study recruited 1142 patients with various clinical indication for karyotype evaluation from the year 2015 until 2023. Samples were obtained from peripheral blood (n=1065), amniotic fluid (n=67), umbilical cord blood (n=1), and product of conception (n=9). Giemsa-banding (G-banding) karyotyping and Fluorescence in situ hybridization (FISH) were performed to identify chromosomal abnormalities.

Results and Discussion

The overall successful identification of chromosomal abnormalities in the samples is 43.3%. The disorders of sex development (DSD) being the most common indication (48.9%), followed by congenital anomalies and developmental delay, infertility, and recurrent pregnancy loss. From all DSD indication, 46,XY DSD, 46,XX DSD and sex chromosomal DSD were accounted for 61.9%,25.7% and 12.4% respectively. Numerical chromosomal abnormalities accounting for the majority of cases (80.2%). Trisomies, particularly trisomy 21, were the most frequent numerical abnormalities (58.3%). Mosaicism was identified in 4.8% cases. Translocations and isochromosome were the most commonly identified structural chromosomal abnormalities. There are significant differences between males and females in the presence of chromosomal abnormalities across certain indications. Males consistently exhibited a higher incidence of chromosomal abnormalities in cases of Down syndrome, Edward syndrome, and Patau syndrome.

Conclusions

This study provides valuable insights into the frequency and distribution of chromosomal abnormalities in West Java. Nevertheless, these findings emphasize the importance of karyotype analysis in diagnosing chromosomal disorders, providing appropriate management and genetic counseling.

Keywords

Chromosomal abnormality, Karyotype, Distribution, Frequency, West Java

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SINGLE-CELL SEQUENCING COMPARISON STUDY FOR MOUSE RETINA RESEARCH

Ayudha Bahana Ilham Perdamaian^{1,2}, Dwi Jami Indah Nurhasanah³, Sri Swasthikawati⁴,
Gunadi⁵, Supanji¹, Muhammad Bayu Sasongko^{1*}

¹Department of Ophthalmology, Faculty of Medicine, Public Health and Nursing, Universitas Gadjah Mada, Yogyakarta, Indonesia, ²Doctorate Program of Medicine and Health Science, Faculty of Medicine, Public Health, and Nurse, Universitas Gadjah Mada, Yogyakarta, Indonesia, ³Master Program of Biomedical Science, Faculty of Medicine, Public Health, and Nurse, Universitas Gadjah Mada, Yogyakarta, Indonesia, ⁴Research Center for Genetic Engineering, Research Organization for Life Sciences and Environment, National Research and Innovation Agency (BRIN), Bogor 16911, Indonesia, ⁵Pediatric Surgery Division, Department of Surgery/Genetics Working Group/Translational Research Unit, Faculty of Medicine, Public Health and Nursing, Universitas Gadjah Mada, Dr. Sardjito Hospital, Yogyakarta 55281, Indonesia

Abstract

Introduction

Mouse retina which consists of many cell types, i.e. photoreceptor, bipolar, ganglion and RPE, makes it difficult to study the transcriptomic profile at the cell level. Many studies conducted single-cell sequencing using distinct procedures, from single-cell tagging (FACS sorting, droplet-based or microwell-based) to the sequencing step (sequencer type and sequencing depth) and ultimately yielded varied outcomes. Furthermore, scientists also use mouse retina as starting material to examine their single-cell sequencing modification. In this review, the authors want to classify and investigate the methods for single-cell sequencing from retina samples. All relevant papers were acquired from PubMed, Semantic Scholar, and Google Scholar.

Material and Methods

Papers were grouped according to tissue dissociation, cell tagging, and sequencing procedure (instrument, deep, and coverage). Protocols were sorted based on the sequencing results i.e. the number of acquired cell types, PCA.

Results and Discussion

The review indicates that papain enzyme, FACS-Sorted cell before loaded into Chromium, and short-read NGS as the most used methods yield the best performance.

Conclusions

This review will assist future research in determining appropriate strategies for performing single-cell sequencing of retina samples.

Keywords

Single Cell Sequencing, Next Generation Sequencing, Retinal cell, Mouse eye, FACS

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Cancer Biology and Cancer Therapeutics

Oral ID – O.C-01

SOMATIC MUTATION PROFILE OF HIGH-GRADE SEROUS OVARIAN CANCER (HGSOC) PRIMARY TUMOURS ASSOCIATED WITH MALIGNANT ASCITES AND METASTASIS

Paranita Ferronika^{1*}, Haryo Wicaksono¹, Nuril M. Maghfiroh¹, Risky Oktriani², Addin Trirahmanto³, Irianiwati Widodo¹.

¹Department of Anatomical Pathology, ²Department of Biochemistry, ³Department of Obstetrics and Gynecology, Faculty of Medicine, Public Health, and Nursing, Universitas Gadjah Mada / Dr. Sardjito General Hospital, Yogyakarta City, Indonesia.

*Corresponding author: paranita.ferronika@ugm.ac.id

Abstract

Introduction

High-grade serous ovarian cancer (HGSOC) typically spreads via the trans-coelomic route, often involving the accumulation of ascites, which is a known prognostic factor for poorer outcomes. Despite this, there has been limited research conducted on the mutated genes that are present in primary tumors of HGSOC patients who develop malignant ascites. Our study aimed to compare the mutation profiles of HGSOC tumors in patients with positive malignant ascites, negative malignant ascites, and those with and without metastasis.

Materials and Methods

We analyzed tissue samples from eight HGSOC patients, including five patients with positive malignant ascites and two patients with metastasis, using whole exome sequencing. We also integrated data from The Cancer Genome Atlas (TCGA) to validate our findings.

Results and Discussion

Our analysis revealed a total of 734 somatic mutations across 724 genes in all eight patients. Out of 724 genes, 523 genes were present in patients with positive malignant ascites. Our study found that 59 mutated genes were shared between HGSOCs with positive malignant ascites and TCGA stage T1c tumors, including well-known cancer-related genes such as TP53 and AKAP9. Enrichment analysis indicated that those 59 mutated genes are mostly involved in actin cytoskeleton organization and insulin receptor signaling pathways. Similarly, among patients with metastasis, we identified 137 mutated genes, with 40 genes overlapping with TCGA stage M1 tumors, including notable genes like IRS4 and CHD4. Enrichment analysis showed that those 40 mutated genes are mostly involved in non-integrin membrane-ECM interaction and axon guidance pathways.

Conclusion

Our findings suggest that identifying the mutated genes that are associated with positive malignant ascites and metastasis could potentially serve as predictive markers for assessing the malignancy of ascites and the likelihood of metastasis in HGSOC patients. This information could significantly enhance prognostic capabilities in clinical settings.

Keywords

Somatic mutation, high-grade serous ovarian cancer, malignant ascites, metastasis

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A COMBINED RISK MODEL SHOWS VIABILITY FOR PERSONALIZED BREAST CANCER RISK ASSESSMENT IN THE INDONESIAN POPULATION: A CASE/CONTROL STUDY

Bijak Rabbani*¹, Sabrina Gabriel Tanu¹, Kevin Nathanael Ramanto¹, Jessica Audrienna¹, Fatma Aldila¹, Eric Aria Fernandez¹, Mar Gonzalez-Porta¹, Margareta Deidre Valeska¹, Jessline Haruman¹, Lorina Handayani Ulag¹, Yusuf Maulana¹, Kathleen Irena Junusmin¹, Margareta Amelia¹, Gabriella Gabriella¹, Feilicia Soetyono¹, Aulian Fajarrahman¹, Salma Syahfani Maudina Hasan¹, Faustina Audrey Agatha², Marco Wijaya², Stevany Tiurma Br Sormin², Levana Sani¹, Soegianto Ali³, Astrid Irwanto¹, Samuel J Haryono²

¹Nalagenetics Pte Ltd, 1093 Lower Delta Road, #04-05/06/07/08, Singapore 169204, ²SJH Initiatives, MRCCC Siloam Hospitals Semanggi, Jakarta, Indonesia, ³Biomedicine Department, School of Medicine and Health Sciences, Atma Jaya Catholic University of Indonesia, Jakarta, Indonesia

*Corresponding author: clinicalstudies@nalagenetics.com

Abstract

Introduction

Breast cancer continues to be a noteworthy global issue, with its occurrence increasing in Indonesia. This study seeks to assess the suitability of risk-based screening methods within the Indonesian population through a case-control investigation involving 305 women.

Material and Methods

We developed a personalized breast cancer risk assessment workflow, which combines multiple risk factors, including clinical (Gail model) and polygenic (Mavaddat model) risk predictions, to form a unified risk category.

Results and Discussion

Each individual risk model predictive accuracy within the Indonesian context is assessed through the assessment of the area under the receiver operating characteristic curve (AUC) (AUC for clinical risk: 0.67 [0.61,0.74]; AUC for polygenic risk: 0.67 [0.61,0.73]). More importantly, our integrated risk approach improved the AUC to 0.70 (0.64, 0.76), underscoring the benefits of a comprehensive model.

Conclusions

Our results unveil, for the first time, the suitability of the Mavaddat and Gail models for Indonesian populations, highlighting that within this demographic, combined risk models offer a more effective and superior predictive framework than single-factor approaches.

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Keywords

Breast cancer, polygenic risk score, monogenic risk, clinical risk, gail model

Oral ID – O.C-03

***BRAF* p.VAL600GLU, *KRAS* MUTATIONS AND CLINICOPATHOLOGICAL CHARACTERISTIC AMONG COLORECTAL CANCER PATIENTS IN A TERTIARY HOSPITAL IN WEST JAVA, INDONESIA**

Reno Rudiman¹, Ahmad N. Alfariy², Kiki Lukman¹, Prapanca Nugraha¹, Yophie Setiawan³, Yunia Sribudiani^{4,5*}

¹Division of Digestive Surgery, Department of Surgery, Dr. Hasan Sadikin General Hospital, Faculty of Medicine, Universitas Padjadjaran, Bandung, Indonesia, ²Undergraduate School, Faculty of Medicine, Universitas Padjadjaran, Bandung, Indonesia, ³Master of Biomedical Sciences, Faculty of Medicine, Universitas Padjadjaran, Bandung, Indonesia, ⁴Department of Biomedical Sciences, Faculty of Medicine, Universitas Padjadjaran, Bandung, Indonesia, ⁵Research Center of Medical Genetics, Faculty of Medicine, Universitas Padjadjaran, Bandung, Indonesia

*Corresponding author: y.sribudiani@unpad.ac.id

Abstract

Introduction

The colorectal cancer (CRC) remains a major burden in cancer worldwide, ranking third and second in incidence and mortality respectively. Detection of biomarkers including *BRAF* p.Val600Glu and *KRAS* mutations can help in predicting prognosis and response to therapy in CRC. This study aims to evaluate frequency of *BRAF* p.Val600Glu and *KRAS* mutations among Indonesian patients and their associations with clinicopathologic characteristics.

Materials and Methods

Fifty-three CRC samples were collected from January to September 2022 in the Department of Surgery, Dr. Hasan Sadikin General Hospital, Bandung, Indonesia. *BRAF* p.Val600Glu and *KRAS* mutations were analyzed using PCR followed by Sanger sequencing. Associations between *BRAF* p.Val600Glu and *KRAS* mutations were evaluated using binary logistic regression analysis.

Results and Discussion

None of the samples were positive *BRAF* p.Val600Glu mutation. *KRAS* mutations were detected in 52.8% patients (n=28), of which 3.6% were p.Gly12Ser (n=1), 32.1% were p.Gly12Asp (n=9), 7.1% were p.Gly13Asp (n=2), 3.6% were p.Gln61His (n=1), 3.6% were p.Asp126His (n=1), 3.6% were p.Lys169Glu (n=1), and polymorphism of p.Asp73= were detected in 57.1% of the samples (n=16). *KRAS* mutation status did not differ significantly between the groups of age of onset, sex, tumor location, tumor histology, stage, and family history.

Conclusions

BRAF p.Val600Glu mutations were not observed in any of these CRC patients, while *KRAS* mutations were present in high frequency. *KRAS* mutations status do not associate with age of onset, sex, tumor location, tumor histology, stage, and family history.

Keywords

BRAF p.Val600Glu, Colorectal Cancer, Early Onset, *KRAS*, Late Onset

Cellular and Molecular Research, Immunology

Oral ID – O.M-01

THE UNTOLD STORY OF IGM LEVELS IN MALARIA-INFECTED SPLENECTOMY PATIENTS

Leily Trianty¹, Aisah R. Amelia², Desandra A. Rahmayenti^{3,4}, Nur I. Margyaningsih³, Labibah Qotrunnada³, Fauziyah Fadllan Hasrini³, Prayoga⁴, Leo Leonardo⁴, Nurjati C. Siregar⁵, Rintis Noviyanti¹, Nicholas M. Anstey⁶, Steven Kho^{4,6}

¹Eijkman Research Center for Molecular Biology, Jakarta, ²Exeins Health Initiative, Jakarta, ³Eijkman Institute for Molecular Biology, Jakarta, ⁴Timika Malaria Research Program, Papuan Health and Community Development Foundation, Timika, ⁵Department of Anatomical Pathology, Rumah Sakit Cipto Mangunkusumo and Universitas Indonesia, Jakarta, ⁶Menzies School of Health Research, Darwin

*Corresponding author: trianty.leily@gmail.com

Abstract

Introduction

Recent research discovered the presence of a large biomass of viable malaria parasites within the spleen of splenectomy patients, highlighting a hidden *Plasmodium* reservoir. However, the spleen is also responsible for mounting immune responses against the parasite. Exaggerated IgM production is a crucial factor in hyper-reactive malarial splenomegaly syndrome (HMSS), which, if not treated, be fatal. We hypothesized that HMSS is the endpoint of a progressive increase in IgM levels, spleen size and abnormal immune responses from regular exposure to malaria parasites. Prompt antimalarial treatment is crucial, yet routine management strategies for HMSS risk are lacking in endemic areas.

Materials and Methods

We enrolled splenectomy patients between 2015-2023 at the Timika Regional Hospital. Sixty-five plasma samples were collected, and had IgM levels examined, alongside recording the spleen weight. HMSS was characterized by plasma IgM levels 2SD greater than the mean of baseline in the Timika population.

Results and Discussion

Of the 65 patients examined in the study, 35.4% were infected with *Plasmodium falciparum*, 26.1% with *Plasmodium vivax*, 3.1% with *Plasmodium malariae*, 16.9% with a mixed infection, and the remainder were uninfected negative controls. The median spleen weight was 319 grams [Range: 39 – 3611 grams], while the median IgM concentration was 103.4 ng/mL [Range: 25 – 2557.2 ng/mL]. Among the 65 patients, 4 individuals were suspected of having HMSS (Range IgM level: 1396.1-2557.2 ng/mL). Of these, three were infected by *P. falciparum*, and one tested negative. Spearman's correlation coefficient of $r=0.6$ ($p=1.54 \times 10^{-7}$) indicated a positive correlation between spleen weight and IgM concentration.

Conclusions

In conclusion, the IgM production is increased progressively in association with increased spleen size, with HMSS being the extreme endpoint. Thus, elevated IgM concentrations may be used as an indicator of potential HMSS. Further research is needed to better understand its mechanisms and improve clinical management strategies.

Keywords

Malaria, splenectomy, splenomegaly, IgM

Oral ID – O.M-02

PHYLODYNAMIC INSIGHTS INTO THE TRANSMISSION AND SPREAD OF SARS-COV-2 XBB 1.5 IN INDONESIA

Dhithram Tenrisau^{1,5}, Yeni Indriyani^{2,5}, Moch Thoriq Assegaf Al Ayubi³, Abdul Haris Ibrahim^{4,5}, Rahmat Saputra⁶, Arie Ardiansyah Nugraha⁷, Noor Alis Setiyadi²

¹MSc Health Data Science, London School of Hygiene of Tropical Medicine, ²Departement of Public Health, Universitas Muhammadiyah Surakarta, Indonesia, ³Faculty of Medicine and Health Sciences, UIN Syarif Hidayatullah Jakarta, Indonesia, ⁴ASEAN Bioad中国家 Virtual Center, ⁵Public Health Literature Club, Indonesia, ⁶Institut Kesehatan Payung Negeri, Pekanbaru, Indonesia, ⁷National Health Laboratory Biology, Ministry of Health, Indonesia

*Corresponding author: yeni.indriyani@ums.ac.id

Abstract

Introduction

The COVID-19 pandemic has presented challenges such as difficult contact tracing and a high prevalence of asymptomatic cases. This study uses a molecular clock framework to trace the evolutionary trajectory of SARS-CoV-2 variant XBB 1.5 from October 2022 to November 2023.

Materials and Methods

We analyzed 226 sequences of XBB 1.5 from Indonesia and 8 from other countries from GISAID. The Wuhan sequence from GenBank became the reference. The countries were chosen based on the top tourist arrivals reported by National Statistics (BPS). Sequence alignment was conducted using MAFFT v7.490, and phylogenetic analysis employed 1000 bootstrap replications in IQ-TREE version 2.3.3. We used an Uncorrelated Clock Model with TreeDater 0.5.0 to estimate the timeline of viral divergence. Transmission clusters were identified based on geographic and temporal data, supported by bootstrap values over 90%, with reconstruction done via epicontacts 1.2.0.

Results and Discussion

The XBB 1.5 variant was first detected in East Java in November 2022, with significant branches traced to samples from Singapore and Australia. The temporal analysis showed a negligible delay (approx. 0.47 days) between molecular dating and sample collection. Jakarta reported the highest number of clusters (29), with Bandung City identified as the major contributor to inter-city spread (22%). Limitations of the study include incomplete contact tracing data and inadequate sequencing resources in Indonesia.

Conclusions

Our findings illustrate a geographic spread pattern of XBB 1.5 within Indonesia, concentrated in major urban centers like Jakarta, Java Island, and Bali, likely facilitated by air travel and human mobility. Genomic surveillance should be considered as an integral part of contact tracing.

Supplemental Materials

The full visualization of the study can be accessed at https://dhithram.github.io/XBB_Phylo/.

Keywords

Molecular clock, genomic, surveillance, COVID-19, phylogenetic, outbreak

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Pharmaceutical Sciences, Phytopharmacy and Herbal Medicine

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IDENTIFICATION OF NUTRIENTS AND PHYTOCHEMICALS IN MIRABILIS JALAPA FLOWER EXTRACT FOR DEVELOPMENT OF ANEMIA TREATMENT

Fera Widyanti¹, Dono Indarto^{1,2,3}, Yuliana Heri Suselo^{1,2,3}

¹Postgraduate Program of Nutrition Sciences, Universitas Sebelas Maret, Jl. Ir. Sutami 36A, Surakarta, Central Java 57126, Indonesia, ²Biomedical Laboratory, Faculty of Medicine, Universitas Sebelas Maret, Jl. Ir. Sutami 36A, Surakarta, Central Java 57126, Indonesia, ³Departemen of Physiology, Faculty of Medicine, Universitas Sebelas Maret, Jl. Ir. Sutami 36A, Surakarta, Central Java 57126, Indonesia

*Corresponding author: fera_1983@student.uns.ac.id

Abstract

Introduction

Anemia is one of the most common health problems around the world, especially in developing countries. Adolescent girls are one of the age groups that are susceptible to anemia, resulting in low concentration and achievement in their studies. The prevalence of anemia aged 15-24 years increased from 18.4% in 2013 to 32.2% in 2018. Iron supplementation is recommended to alleviate anemia in adolescent girls, but it frequently has side effects. *Mirabilis jalapa* is recognized as an ornamental plant that consists of *Indicaxanthin*, *Miraxanthin-V*, and *Boeravinone-F*. the alkaloid fraction of combined *M. jalapa* flowers is able to increase intracellular iron levels in the hepatocarcinoma cell line through regulation of matriptase-2-hepcidin pathway. Therefore, the aim of this study was to analyze the nutrient and chemical compositions of yellow *M. jalapa* flower extract.

Materials and Methods

We used dried yellow *M. jalapa* flowers in this study, which were obtained from the Merapi Farma Herbal Center in Sleman Yogyakarta and extracted using a maceration method with n-hexane and 96% (volume/volume) ethanol solvents. Nutrition and chemical compositions of *M. jalapa* extracts were evaluated using the proximate and liquid chromatography-mass spectrometry (LC-MS) methods and conducted at the Integrated Research and Testing Laboratory, Gadjah Mada University, Yogyakarta.

Results and Discussion

The extract of *M. jalapa* flowers contained 18.71% (weight/weight) water, 12.08% ash, 25% proteins, 4.74% fats, 39.47% carbohydrates, 0.11% Fe, 8.95% Zn and 5.51% vitamin C. Furthermore, *Betaxanthin* group compounds were found in the positive peak of the LC-MS chromatogram.

Conclusions

The extract of yellow *M. Jalapa* flowers contains high iron levels and *Betaxanthin* group compounds, which are potentially developed for anemia treatment. Further investigation is required for identification of phytochemicals in the extract of *M. jalapa* flowers.

Keywords

Anemia, *Mirabilis jalapa*, Flower extract, *Betaxanthin*, Iron levels

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THE ROLE OF ETHANOL EXTRACT OF KEBIUL SEEDS (*CAESALPINIA BONDUCELLA*) AS AN ANTI-INFLAMMATORY IN MALE WHITE RATS OF THE SPRAGUE DAWLEY STRAIN INDUCED BY THE FIVE-SIXTH SUBTOTAL NEPHRECTOMY METHOD (5/6 SN)

Leony Hestoria^{1,2}, Juniarti³, Linda Armelia⁴

¹Master of Biomedicine, YARSI University Graduate School, Jakarta, Indonesia, ²Department of Anatomy and Histology, Faculty of Medicine, YARSI University, Jakarta, Indonesia, ³Department of Biochemistry, Faculty of Medicine, YARSI University, Jakarta, Indonesia, ⁴Department of Internal Medicine, Faculty of Medicine, YARSI University, Jakarta, Indonesia
*Corresponding author: juniarti@yarsi.ac.id

Abstract

Introduction

Chronic kidney disease (CKD) is still a major and serious health problem in the world. Kebiul seeds (*Caesalpinia bonducella*) contain flavonoids which can inhibit the activity of the Renin Angiotensin Aldosterone System (RAAS) which plays a role in the mechanism of CKD. The use of kebiul seeds empirically needs to be proven by using experimental animals that were induced by five-sixth subtotal nephrectomy (5/6 SN). The aim of this research was to determine the effect of ethanol extract of kebiul seeds as an anti-inflammatory in improving kidney function and to see how it reduces inflammatory mediators by looking at the levels of Interleukin-6 (IL-6) and Interleukin-8 (IL-8).

Material and Methods

This research was an in vivo experiment (posttest design) carried out on 30 male white rats of the Sprague Dawley strain, 2 months old, weighing 200-250 grams, divided into 6 groups that received the following treatment: K1, K2, K3, 5/6 SN with ethanol extract of kebiul seeds at doses of 150, 300, 500 mg/kg BW. CKD induction was carried out with 5/6 SN before treatment. After that, the treatment was carried out for 14 days and then the kidney tissue to be examined was taken to see the levels of IL-6 and IL-8 using the ELISA method. Data will be processed using oneway ANOVA and Kruskal Wallis, data significance is indicated by $p < 0.05$.

Result and Discussion

Based on the research results, it was found that IL-6 levels in negative control 1 (K1) kidney samples were 29.71 ± 1.52 pg/ml, negative control 2 (K2) were 36.83 ± 4.99 pg/ml, positive controls (K3) was 35.00 ± 1.31 pg/ml, the kebiul seed ethanol extract group at doses of 150, 300 and 500 mg/kg BW was 40.39 ± 6.71 , 30.65 ± 2.79 , and 32.49 ± 3.03 pg/dl. IL-8 levels in K1 were 16.85 ± 1.20 pg/ml, K2 were 32.32 ± 1.75 pg/ml, K3 were 16.17 ± 1.87 pg/ml, kebiul seed ethanol extract group dose 150, 300

and 500 mg/kg BW were 20.95 ± 0.62 , 21.15 ± 3.37 and 26.68 ± 5.80 pg/ml. It can be concluded that administration of kebiul seed ethanol extract significantly reduced IL-8 levels at doses of 150 mg, 300 mg, 500 mg, and IL-6 levels were not significant at doses of 300 mg, 500 mg.

Conclusions

This shows that the ethanol extract of kebiul seeds can improve kidney function and suppress inflammatory mediators.

Keywords

Chronic Kidney Disease, boils, *Caesalpinia bonducella*, anti-inflammatory, IL-6, IL-8, 5/6 SN

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Health Promotion, Health Policy and Education

Oral ID – O.H-01

ANIMAL PROTEIN INTAKE IN CHILDREN AGED 4-6 YEARS IN BEKASI

Virjinea Hawani Reine, Nur Asiah, Firman Firman Arifandi

Faculty of Medicine YARSI University Jalan. Letjen. Suprpto Cempaka Putih Jakarta

*Corresponding author: nur.asiah@yarsi.ac.id

Abstract

Introduction

Stunting in Indonesia is still high (27.7%) based on the 2019 Indonesian Toddler Nutrition Status Survey (Survei Status Gizi Balita Indonesia/ SSGBI) (Ruswati, et al, 2021). Children who consume quality protein have a lower risk of nutritional problems, such as stunting. Animal protein is superior to vegetable because it contains complete essential amino acids. calcium, iron, magnesium, zinc as bone builders, nervous system. To build a strong bone and muscle mass of children growth , we need optimal animal protein in their daily food intake

Material and Methods

This study was to determine animal protein intake in 32 children aged 4-6 years at Al-Muhajirin Bekasi Kindergarten in December 2023, with a cross-sectional design. Data were from 2x24 hour food recalls. The questionnaires consisted of the amount of their total protein intake, including vegetable protein and animal protein.

Results and Discussion

Most mothers are aged 25-35 years, undergraduates and housewives. As many as 96% of children have sufficient protein intake. Average protein intake was 49.5 grams, consisted of 28.48 grams of animal origin and 20.92 grams of vegetable intake. The average protein intake is greater than the recommended intake for children 4-6 years (35 grams). It was supported by the head of the Bekasi district health service who was promoting the movement of eating eggs and drinking milk together among school children (Gebyar Bekasi Dare Zero New Stunting).

Conclusions

The average protein intake is greater than the recommendation but the quality of protein consumption still needs to be researched.

Keywords

Animal protein, children.

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EFFECT OF FLOWER EXTRACT CREAM TELANG (*CLITORIA TERNATEA* L) ON VEGF EXPRESSION AND ANGIOGENESIS ON INCISION WOUND HEALING: AN INVIVO STUDY

Nurmayani Irwandi¹, Juniarti², Nunung Ainur Rahmah*³

¹Biomedical Science Study Program, Graduate School of Universitas YARSI, Jakarta, ²Department of Biochemistry, Faculty of Medicine, Universitas YARSI, Jakarta, ³Department of Anatomical Pathology, Faculty of Medicine, Universitas YARSI, Jakarta

*Corresponding author: nunung.ainur@yarsi.ac.id

Abstract

Introduction

The number of injury cases continues to increase from year to year. However, treatment still uses antiseptics which are irritating and toxic. Therefore, it is necessary to look for a solution that can be used safely and at an affordable price. The research aims to determine the sensitivity and effect of butterfly pea flower (*Clitoria ternatea* L) extract cream on the expression of *Vascular Endothelial Growth Factor* (VEGF) and the amount of angiogenesis in the healing of incision wounds in male white rats of the *Sprague dawley* strain compared with betadine.

Materials and Methods

In vivo experimental research was carried out at the integrated laboratory at YARSI University, Jakarta in 2023. The administration of *Clitoria ternatea* L extract cream to incision wounds is based on different doses, namely concentrations of 5% (5 grams), 10% (10 grams), and 15% (15 grams). Sensitivity test using a skin prick test. Examination of VEGF expression by immunohistochemical staining and the amount of angiogenesis by hematoxylin-eosin staining. Data analysis used the Kruskal-Wallis test followed by the Post Hoc Mann-Whitney test.

Results and Discussion

The results showed that the sensitivity test of *Clitoria ternatea* L. cream extract was safe to use. *Clitoria ternatea* L extract cream significantly increased VEGF expression on day 3 ($p=0.006$), and increased the amount of angiogenesis on day 3 ($p=0.000$), day 7 ($p=0.002$), and day 15 ($p=0.000$) at the most optimal concentrations of 5%, 10%, and 15% with a wound healing percentage rate of 0% compared to betadine. There was a correlation between VEGF expression and the amount of angiogenesis on days 3.

Conclusions

Clitoria ternatea L extract cream is safe to use, and can increase the expression of VEGF and the amount of angiogenesis same with betadine. *Clitoria ternatea* L extract cream can be a safe and affordable choice for treating wounds.

Keywords

Angiogenesis, *Clitoria ternatea* L, VEGF, Wounds

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Oral ID – O.H-03

FTO GENE, MACRONUTRIENT RECOMMENDATION AND ADIPOSITY: THE DEVELOPMENT OF PERSONALIZED DIET FOR OBESITY PREVENTION BASED ON GENE VARIATION

Harry Freitag Luglio Muhammad¹, Risa Shofia¹, Harry Kasuma Kiwi Aliwarga¹, Josano Rehan Dhani¹, Annisaa Widyasari¹, Muflihatul Muniroh², Gunadi³

¹PT. Widya Genomic Nusantara, ²Faculty of Medicine, Universitas Diponegoro, ³Faculty of Medicine, Public Health and Nursing, Universitas Gadjah Mada

*Corresponding author: harry.freitag@gmail.com

Abstract

Introduction

The interplay between genetic predisposition and dietary habits contributes significantly to obesity. Tailoring dietary interventions based on individual genetic variations can optimize weight management strategies. We present an algorithm utilizing genetic markers, particularly single nucleotide polymorphisms (SNPs) on the FTO gene, to mitigate obesity risk and devise personalized weight loss regimens. This study aimed to assess the correlation between dietary recommendations for carbohydrate, protein, and fiber intake, guided by FTO gene variations, and clients' body mass index (BMI).

Materials and Methods

This is an observational study of data originated from clients of Widya Genomic undertook GenKu program to test for gene variation. Age, body weight and height was obtained through online questionnaire. Genetic variation of FTO (rs11075990, rs8050136, rs3751812, rs11075990) was obtain using microarray. Analysis was done using ANCOVA with correction for age and sex.

Results and Discussion

Data from a total of 39 clients (41% women) were obtained and analyzed anonymously. The mean age is 47.6 years old and body mass index (BMI) is 24.2 kg/m². Though our algorithm for risk of obesity, we showed that the BMI those high risk for obesity (n=33) were not significantly different from those with lower risk for obesity (n=5) (24,3 ± 0,7 vs 23,8 ± 1,1 kg/m², respectively) (p=0,676). Those with recommendation for a lower carbohydrate diet based on genetic algorithm had higher BMI (26,2 ± 1,1 kg/m²) than those with standard carbohydrate diet (22,9 ± 0,6 kg/m²) (p=0,011). Those with recommendation for higher fiber intake based on genetic algorithm also had higher BMI (25,8 ± 1,1 kg/m²) than those with standard fiber intake (23,1 ± 0,6 kg/m²), (p=0,031). Recommendation for high/ standard protein diet was not associated with BMI (p=0,528). We combine all three macronutrient recommendation and those with low carbohydrate, high protein and high fiber diet based on genetic algorithm had higher BMI (26,7 ± 1,5 kg/m²) than those without all three combination (23,2 ± 0,6 kg/m²) (p=0,013).

Conclusions

Genetic variation in obesity risk alone is not association with BMI. We developed specific dietary recommendation based on variation at FTO gene, those include low carbohydrate, high protein and high fiber. In this study we confirmed that those with altered diet was associated with higher BMI.

Keywords

Genetic, obesity, FTO, diet, body mass index

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NUTRITION AND CHEMICAL ANALYSIS OF BUTTERFLY PEA FLOWERS COMBINED WITH LEMON AS AN HERBAL DRINK FOR OBESITY TREATMENT

Agustiarini Dewi Anugrahani¹, Dono Indarto^{1,2,3}, Eti Poncorini P.^{1,4}, Lilik Wijayanti^{1,5}¹Postgraduate Program of Nutrition Sciences, Universitas Sebelas Maret, Jl. Ir. Sutami 36 A Surakarta, Central Java 57126, Indonesia,²Biomedical Laboratory, Medical Faculty, Universitas Sebelas Maret, Jl. Ir. Sutami 36 A Surakarta, Central Java 57126, Indonesia,³Physiology Department, Medical Faculty, Universitas Sebelas Maret, Jl. Ir. Sutami 36 A Surakarta, Central Java 57126, Indonesia,⁴Public Health Department, Medical Faculty, Universitas Sebelas Maret, Jl. Ir. Sutami 36 A Surakarta, Central Java 57126, Indonesia,⁵Clinical Pathology Department, Medical Faculty, Universitas Sebelas Maret, Jl. Ir. Sutami 36 A Surakarta, Central Java 57126, Indonesia[*Corresponding author: agustiarini23@student.uns.ac.id](mailto:agustiarini23@student.uns.ac.id)

Abstract

Introduction

Obesity is a global health problem that become a main risk factor for cardiovascular diseases. Our previous study showed that a combination of butterfly pea flowers and lemon (*Litrusia* herbal drink) could reduce body weight (BW) and lipid profiles in obese rats. In addition, *hesperidin* has anti-obesity properties, which are often found in citrus fruits. Administration of 55 mg/kg/day *hesperidin* for 30 days reduced BW significantly compared to the administration of 120 mg/kg/day orlistat in obese rats. *Hesperidin* could increase lipolysis and inhibit lipogenesis through up-regulation of Peroxisome Proliferator Activator Receptor- γ gene expression. However, *hesperidin* levels of butterfly pea flowers have not been established. Therefore, this study aimed to analyze the macronutrient composition and *hesperidin* levels of *Litrusia* herbal drinks.

Materials and Methods

Butterfly pea flowers and lemon fruits were obtained from Pasuruan and Malang plantation, respectively. We made *Litrusia* herbal drinks by combining the dried butterfly pea flowers and fresh lemon juice at an 85:15% ratio and adding liquid stevia sweetener. We analyzed the macronutrient contents of the *Litrusia* herbal drink at the Laboratory of Food Chemistry and Biochemistry, Faculty of Agriculture, UNS. *Hesperidin* levels in the dried butterfly pea flower, fresh lemon juice, and *Litrusia* herbal drink were measured using a High-Performance Liquid Chromatography (HPLC) method, and conducted at the Laboratory of Integrated Biology UNS.

Results and Discussion

200 ml of *Litrusia* herbal drink contained 0.68 g of proteins, 1.64 g of fats, 0.20 g of carbohydrates, and 18.28 kcal of energy. The highest *hesperidin* levels were found in dried butterfly pea flowers (113.02 mg/200ml), followed by *Litrusia* herbal drink (105.14 mg/200ml) and fresh lemon juice (29.52 mg/200ml) respectively.

Conclusions

Litrusia herbal drink has low energy and high levels of *hesperidin*, which may become a good supplement for obesity treatment.

Keywords

Obesity, Butterfly pea flower, Lemon fruit, *Hesperidin*, Herbal drink

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NUTRITIONAL AND PHYTOCHEMICAL ANALYSIS OF SNAKE FRUIT PONDOK PEELS FOR DEVELOPMENT OF OBESITY TREATMENT

Puri Dwi Andina¹, Dono Indarto^{1,2,3}, Lusi Oka Wardhani^{1,4}

¹Postgraduate Program of Nutrition Sciences, Universitas Sebelas Maret, Jl. Ir. Sutami 36A, Surakarta, Central Java 57126, Indonesia, ²Biomedical Laboratory, Medical Faculty, Universitas Sebelas Maret, Jl. Ir. Sutami 36A, Surakarta, Central Java 57126, Indonesia, ³Physiology Department, Medical Faculty, Universitas Sebelas Maret, Jl. Ir. Sutami 36A, Surakarta, Central Java 57126, Indonesia, ⁴Clinical Pathology Department, Medical Faculty, Universitas Sebelas Maret, Jl. Ir. Sutami 36A, Surakarta, Central Java 57126, Indonesia

*Corresponding author: puridwiandina1511@student.uns.ac.id

Abstract

Introduction

Obesity is caused by fat accumulation in the body with high mortality rate related to other metabolic disorders. Some obese patients failed to reduce their body weight through diet and physical activity. Natural products from Indonesian plants have been developed for an alternative treatment of obesity, but their efficacy needs further investigation. *N-hexadecanoic acid* derived from ethyl acetate extract of CcR8 fungi has a potential benefit for obesity treatment. Snake fruit pondok is one of the famous cultivar fruits in Indonesia which its peels are thrown away as rubbish. This study aimed to analyze the nutritional and chemical contents of snake fruit pondok peels for development of obesity treatment.

Materials and Methods

Fresh peels of snake fruit pondok were obtained from Sleman, Yogyakarta. Dried peels of snake fruit pondok were extracted using the maceration method with 70% ethanol. The ethanol extracts were determined for their nutrient compositions at the Laboratory of Food Technology, Faculty of Agriculture, Universitas Sebelas Maret while the chemical analysis was performed at the Laboratory of Integrated Research and Testing Analysis, Universitas Gadjah Mada using a GC-MS method.

Results and Discussion

The extract of snake fruit pondok peels contained higher percentages of carbohydrates (41.33%), minerals (28.37%), and water (25.31%), and lower percentages of crude fiber (3.68%), fats (2.62%), proteins (2.37%), and vitamin C (0.88%). We identified 75 phytochemicals in the extract from which the five higher proportions were *hexadecanoic acid, methyl ester, 9,12-octadecadienoic acid (Z,Z)-, methyl ester, 9,12,15-octadecatrienoic acid, methyl ester, (Z,Z,Z)-, n-hexadecanoic acid, and linoelaidic acid* with retention time at 16.66, 18.26, 18.33, 17.12, and 18.72 minutes respectively.

Conclusions

The ethanol extract of snake fruit pondok peels might have a potential benefit for obesity treatment. *In vitro* and *in vivo* studies are needed to evaluate the anti-obesity properties of snake fruit pondok peels extract.

Keywords

Obesity, Snake fruit pondok peel, Phytochemical, Nutritional, *n-hexadecanoic acid*

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COMPREHENSIVE MANAGEMENT OF RIFAMPICIN-RESISTANT TUBERCULOSIS BASED ON HOLISTIC DIAGNOSIS AND PRECISION MEDICINE PRINCIPLES: A CASE REPORT FROM PRIMARY CARE IN INDONESIA

Diana Batara Munti^{1,2}, Erlina Wijayanti^{1*}

¹ Medical Faculty Universitas YARSI, Jakarta Indonesia, ²STIKES AN NASHER, Cirebon Indonesia

*Corresponding author: erlina.apri@gmail.com

Abstract

Introduction

Rifampicin-resistant tuberculosis (RR-TB) necessitates a multidimensional, patient-centered approach integrating genomic medicine principles for optimal management.

Material and Methods

A comprehensive holistic assessment was conducted on a 48-year-old man with RR-TB through medical history, examination, laboratory/imaging studies, genogram, host-environment analysis, genetic counseling, and testing recommendations. Diagnosis incorporated biological, psychological, social, spiritual, and genomic aspects per WHO guidelines.

Results and Discussion

Assessment revealed treatment adherence, support, nutrition, psychosocial, and infection control issues. Pharmacogenomic and nutrigenomic testing guided personalized interventions. Family history and comorbid diabetes indicated hereditary susceptibility. TB, diabetes, and drug response markers were evaluated for risk stratification and therapy.

Conclusions

This case underscores primary care's role in delivering holistic, person-centered RR-TB care integrating genomic medicine. Comprehensive management with advanced diagnostics, personalized interventions, and multidimensional support can improve outcomes, quality of life, and adherence in resource-limited settings.

Keywords

Genomic Medicine, Personalized Medicine, Tuberculosis, Social Determinants, Diabetes.

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Oral ID – O.H-07

PATIENT CASE STUDY INTERMITTENT ASTHMA IN CHILDREN THROUGH A PRIMARY SERVICE FAMILY MEDICAL APPROACH

Bayu Raharjo¹, Erlina Wijayanti², Zwasta Pribadi Mahardhika³

¹Primary Care Family Medicine Study Program, Medical Faculty Universitas YARSI

*Corresponding author: drbayuraharjo@gmail.com

Abstract

Introduction

Asthma is a non-infectious chronic respiratory disease with bronchospasm in the airways which results in obstruction and hyperreactivity of the respiratory tract as a result of interactions between genetic factors, host factors and the environment. This case report describes a child aged 4 years 10 months with intermittent asthma symptoms in primary care.

Materials and Methods

With a holistic, comprehensive and genomic approach to family medicine in carrying out management based on Evidence Based Medicine which is family-approach, patient centered and community oriented. This study is primary data obtained through history taking, physical examination, child growth and development and home visits.

Results and Discussion

The assessment reveals genetic factors in the patient's family who suffer from asthma, psychosocial challenges, medication compliance, family support, nutritional problems, controlling the risk of asthma, and recommends genetic examination of the patient's siblings who have a potential risk of asthma with specific genes that are generally involved in asthma. is that ORMDL3 is strongly associated with asthma causing high levels of IgE, ADAM33 is strongly expressed in bronchial smooth muscle cells and lung fibroblasts and is involved in airway hyperresponsiveness and decreased lung function. Other genes frequently involved include IL33 – Interleukin-33 (cytokine).

Conclusions

In this case, internal factors were obtained from the patient's family, namely a family history of asthma and external factors that trigger asthma such as house dust, dust mites, pet dander, cigarette smoke, air pollution and extreme weather changes. Parents need to know the risk factors for asthma and recommendations for allergy testing to identify specific asthma triggers. In the role of precision medicine in primary care in a holistic and comprehensive family approach, it can predict the risk of someone suffering from asthma based on the principles of genomic medicine and environmental factors, so that more effective prevention of risk factors for asthma can be achieved.

Keywords

Intermittent asthma, Precision Medicine, Genomic factors, holistic, comprehensive

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TOWARDS THE IMPLEMENTATION OF PRECISION MEDICINE IN INDONESIAN PRIMARY CARE

Erlina Wijayanti^{1*}, Dian Mardhiyah¹, Zwasta Pribadi Mahardhika¹, Yusnita¹, Dini
Widianti¹ ¹ Faculty Medicine, YARSI University, Jakarta, Indonesia

*Corresponding author: erlina.apri@gmail.com

Abstract

Introduction

Current medicine still uses 1 size for all while each individual's condition can be different. Precision medicine provides therapies and preventions that consider individual variations such as genes, environment, and lifestyle (1). The importance of precision medicine has been realized by the Indonesian government and outlined in the 2023 Health Law. Another support is the Biomedical and Genome Science Initiative (BGSI) has been built to develop research and service in several hospitals. Integrated electronic medical records are being developed to improve the quality of data that helps decision making (2). The ease for customers to carry out genetic tests has also been facilitated by several laboratories so that the customers can access directly.

Results and Discussion

The weaknesses that need to be anticipated are public knowledge about potential ethical issues (3), costs that tend to be expensive and have not been financed by BPJS insurance, and follow-up of genetic examination results. Primary care has an important role because it is the first contact with the patient. The primary care physician is the coordinator who further determines the service plan including disease prevention.

Conclusions

The ecosystem that supports the implementation of precision medicine in primary care needs to be well prepared. As people come into more contact with primary care, precision medicine has the potential to improve quality.

Keywords

Precision medicine, primary care

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Oral ID – O.H-09

SCHIZOPHRENIA WITH DISORDERS OF FAMILY FUNCTIONING THROUGH A FAMILY MEDICINE APPROACH

Jamilah^{1,2}, Zwasta Pribadi Mahardhika^{1*}, Dian Mardhiyah^{1*}

¹Medical Faculty Universitas YARSI, Jakarta Indonesia

*Corresponding author: zwasta.pribadi@yarsi.ac.id

Abstract

Introduction

Precision medicine has an important role in risk prediction and treatment of schizophrenia. Doctors can predict an individual's risk for schizophrenia based on analysis of DNA, genome, epigenetic responses, and environmental and lifestyle factors. This allows for more effective and personalized preventive measures, as well as more targeted treatment. This case report illustrates 39-year-old woman with schizophrenia with disorders of family functioning through a primary care family medicine approach.

Materials and Methods

The data obtained includes primary data. Primary data was obtained through anamnesis (autoanamnesis and alloanamnesis with the patient's mother and siblings), physical examination and home visits, filling in patient files obtained through in-depth interviews to complete family data, psychosocial and environmental data and conducting physical examinations and mental status examinations on patients. The assessment is carried out based on a holistic diagnosis from the beginning, process and end of the study.

Results and Discussion

The assessment revealed that there were genetic factors in the patient's family who suffered from schizophrenia, a history of past trauma, lack of knowledge which made the family not take the patient to see a doctor and receive routine treatment, and lack of family support. Personal plans emphasize education about the disease & treatment, family support and eliminating bad stigma in society. Chromosome Microarray Analysis (CMA) examination has been suggested to be a first-line genetic test for schizophrenia. The Genesight psychotropic test can analyze medications and neuropsychiatric genes, detect the genes themselves and can help with the selection and dosing of medications for psychiatric patients. Scientific evidence supports the idea that genetic factors play an important role in the cause of schizophrenia, studies show that the risk of the disease is approximately 10% in first-degree relatives and 3% in second-degree relatives.

Conclusions

In the treatment of schizophrenia, precision medicine can help in several ways. Precision medicine can predict an individual's risk for schizophrenia based on analysis of genetics and environmental factors, allowing for more effective preventive measures and determine the most effective medication options for each patient, based on individual response to medications. Precision medicine can help in improving the quality of schizophrenia treatment in a more effective way.

Keywords

Schizophrenia, Precision Medicine, Genetics, Effective preventive, Medications.

Others

Oral ID – O.O-01

PREVALENCE AND NATURE OF POTENTIAL DRUG-DRUG INTERACTIONS IN HOSPITALIZED DIABETIC PATIENTS: A CROSS-SECTIONAL STUDY

Muhammad Adil Khan¹, Nadia Farhanah Syafhan^{*1,2}, Retnosari Andrajati^{1,2}, Bambang Wispriyono³, Sidra Noor⁴

¹Department of Clinical Pharmacy, Faculty of Pharmacy, Universitas Indonesia, Depok, West Jawa-16424, Indonesia, ²Installation of Pharmacy, Universitas Indonesia Hospital, Depok, West Jawa-16424, Indonesia, ³Department of Environmental Health, Faculty of Public Health, Universitas Indonesia, Depok, West Jawa-16424, Indonesia, ⁴Department of Pharmacy Practice, Shifa College of Pharmaceutical Sciences, Shifa Tameer-e-Millat University, Islamabad, Pakistan

*Corresponding author: nadia.farhanah@farmasi.ui.ac.id

Abstract

Introduction

Diabetes mellitus is a chronic condition commonly associated with complications and comorbidities that often require hospitalization to manage them. Such patients are frequently subjected to complex medical regimens which increase the likelihood of potential drug-drug interactions (pDDIs). The study aimed to assess the prevalence, severity levels, risk ratings, and the association of factors affecting pDDIs among hospitalized diabetes mellitus patients.

Materials and Methods

A retrospective cross-sectional study was conducted on 200 adult diabetic inpatients of Universitas Indonesia Hospital admitted from January 2023 to December 2023 who received more than one prescribed drug. Pregnant, intensive-care patients were excluded. Lexi-Interact was used for screening pDDIs in all prescribed drugs for each patient. The Chi-square test was used to check the association of different factors with pDDIs prevalence.

Results and Discussion

The prevalence of pDDIs in our study population was 89%, with a total of 966 pDDIs. The number of identified pDDIs per patient was 4.83 ± 4.45 . Most of the pDDIs were categorized as moderate (75.6%), followed by minor (16.2%), and major (8.1%). The risk rating of C category pDDIs was found mostly (69.2%) followed by D (18.4%) and B (11.1%). The association of number of prescribed medications, comorbidities, and hospital stay was statistically significant with pDDIs having $p < 0.001$ each.

Conclusions

The findings of this study revealed that pDDIs are highly prevalent in adult inpatients with diabetes. Most of the pDDIs are of moderate severity and C risk rating category. Polypharmacy, comorbidities, and a longer hospital stay are key factors associated with pDDIs prevalence. This emphasizes the critical need for appropriate monitoring and management strategies to reduce pDDIs and their related adverse consequences.

Keywords

Diabetes mellitus, drug-drug interaction, medication safety, risk factor, Indonesia

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Poster ID – O.O-02

THE POTENTIAL OF SECRETOME IN ANTI-AGING: AN ANALYSIS OF TELOMERE LENGTH

Reza Aditya Digambiro¹, Dyah Ayu Woro Setyaningrum¹, Florinda Ilona¹,
Julian Chendrasari¹, Indah Widya Lestari¹

¹Department of Anatomical Pathology, Faculty of Medicine Universitas Trisakti,
Jl. Kyai Tapa No.1, RT.5/RW.9, Tomang, Kec. Grogol Petamburan, Kota Jakarta Barat,
Daerah Khusus Ibukota Jakarta, 11440, Indonesia

*Corresponding author: drdigambiro@trisakti.ac.id

Abstract

Introduction

In the relentless search for anti-aging therapies, there comes the stem cell-derived secretome therapy. In that line, therefore, the present project will explore the potential of secretome from mesenchymal stem cells (MSCs) in anti-aging, specifically with reference to telomere length, which is a principal determinant of cellular aging.

Materials and Methods

We systematically reviewed the papers that published from January 1, 2017 until February 20, 2024 from PubMed, Google Scholar, and Scopus search databases using keywords that included the following: "mesenchymal stem cells," "secretome," "anti-aging," and "telomere length." Both human and animal studies were considered, with outcomes ranging from functional results from cellular rejuvenation to systemic aging markers and structural changes in biomolecular or histochemical parameters.

Results and Discussion

A total of 13 animal studies, with 3 human clinical trials and 10 animal studies, provided evidence for general therapeutic potential of the secretome, with marked improvements in skin rejuvenation, reduction in inflammation, and positive modulation of telomere length. The secretome from multiple sources, including adipose tissue, bone marrow, and umbilical cord, was administered through tailored delivery. The functional outcomes referred to enhancement in skin texture and elasticity, while the structural referred to upregulation of regenerative markers and reduction in inflammatory cytokines.

Conclusions

The MSC secretome represents a new and really promising approach to anti-aging therapy, particularly in view of the modulation of telomere length. More studies will be needed in order to have deep clarity with respect to the mechanisms and long-term safety. This further underscores the importance of more research to enable comprehensive insight into methods of therapeutic use of MSC secretome for anti-aging applications.

Keywords

Anti-aging, mesenchymal stem cells, secretome, telomere length, cellular senescence, tissue regeneration.

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The Fate of Zona-Free Oocyte Following Intra Cytoplasmic Sperm Injection: A Case Report

Zakiyatul Faizah^{1,2,*}, Geraldo Laurus³, Vellyana Lie^{3,4,6}, Andri Rezano⁵, Widjiati⁶, Hendy Hendarto^{7,8}

¹Doctoral Program of Medical Science, Faculty of Medicine, Universitas Airlangga, Indonesia, ²Department of Biomedical Science, Faculty of Medicine, Universitas Airlangga, Indonesia, ³Andrology Study Program, Faculty of Medicine, Universitas Airlangga, Indonesia, ⁴Wahidin Sudiro Husodo General Hospital, Indonesia, ⁵Department of Biomedical Science, Faculty of Medicine, Universitas Padjadjaran, Indonesia, ⁶Faculty of Veterinary Medicine, Universitas Airlangga, Indonesia, ⁷Department of Obsetric and Gynaecology, Faculty of Medicine, Universitas Airlangga, Indonesia, ⁸Clinic Tiara IVF, Graha Amerta, Dr. Soetomo General Academic Hospital, Indonesia

*Corresponding author: zakiyatul.faizah-2021@fk.unair.ac.id

Abstract

Introduction

In humans, oocytes are surrounded by an extracellular matrix called the zona pellucida (ZP). The ZP, which is synthesized, produced, processed, secreted, and shed by oocyte cells, consists of four glycoprotein fibrils interconnected to form a layered structure. ZP has function such as: spermatozoa selection for fertilization process, preventing polyspermy occurrence; Preventing blastomere separation in embryos during the phase; Inhibiting blastomere attachment to the tube wall during the pre-compaction phase.

Material and Methods

This case report showed the potential of free zone oocytes can development like the others.

Results and Discussion

This case report presents a case of a 44-year-old woman undergoing ICSI. Only one oocyte was obtained, but during the zona pellucida denudation process, it detached, leaving behind a zona-free oocyte (ZFO). The available ZFO underwent ICSI and culture; the cells developed into 4 cells on the third day and embryo transfer was performed. Unfortunately, there was no pregnancy. In the above case, ICSI was performed with ZFO still allowing fertilization to occur, it can also be observed that there is horizontal cell development due to ZFO. There are not many studies comparing outcomes between embryos without ZP and embryos with intact ZP. A retrospective study was conducted in Japan comparing outcomes of both types of embryos from the same patients. The study showed no significant differences between the two types of embryos.

Conclusions

This case report presents a case of a 44-year-old woman undergoing ICSI with a zona-free oocyte (ZFO). Unfortunately, there was no pregnancy.

Keywords

Oocyte, Zona free oocyte, Zona pellucida, ICSI, Good Health and well being.

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RETHINKING CLINICAL PRACTICE IN THE GENOMIC ERA

Austin (Seung Woo) Ryu, Ph.D.

With the advent of next-generation sequencing (NGS), clinical genetic testing was revolutionized by being able to screen a large number of genes simultaneously and provide an efficient molecular diagnostic tool for rare disease patients. Currently, the two most widely used NGS test methods are gene panel tests which sequence mostly coding regions of a selected set of genes, and exome tests which sequence coding regions of almost all known human genes.

Between the two tests, panel tests have been the preferred test in clinical settings, predominantly driven by their cost and insurance coverage. However, there are additional advantages of panel tests compared to exome. Panel tests tend to have a higher depth-of-coverage (DOC) for the targeted genes, allowing for greater sensitivity of specificity for both smaller copy number variants (CNV) and mosaic variants. In addition, panel tests are free from ethical considerations such as carrying risks for incidental findings being unveiled. This means that, compared to gene panels, exomes have lower mean DOC and are accompanied by the limitation of lower resolution when calling CNVs and also, exomes come with a risk of identifying incidental findings and because of that risk, the patients need to be consented before the test can be ordered and this imposes a burden on the ordering physicians.

However, exomes offer greater benefits when it comes to genetically heterogeneous cases. While some diseases such as peroxisomal disorders or ciliary dyskinesia have hallmark clinical presentations with relatively limited sets of known causal genes, many genetic disorders have overlapping clinical features with each other and high genetic heterogeneity. Because of this reason, it is not uncommon to find cases for which the causal gene identified is completely different from the suspected gene based on a clinical diagnosis and that is why exomes yield a higher diagnostic rate than panels. Another key advantage of exomes is the possibility of reanalyzing all genes periodically. As novel gene discovery continues, reanalysis of existing data is essential for undiagnosed cases and reanalysis of existing exome data has been demonstrated to increase the diagnostic rate by 10% when performed at least after a year of the initial analysis because the novel genes had been already sequenced. Conversely, most gene panels fail to incorporate novel genes in a timely manner and therefore patients need to receive new tests such as another panel, exome, or genome to continue the diagnostic workup.

Generally, exome is becoming more popular than panels as the first-tier genetic test in more countries worldwide for the reasons described above and because the test cost is continuously decreasing. However, ultimately, the ordering physician needs to determine if panel or exome is more appropriate for each patient with a suspected rare disease.

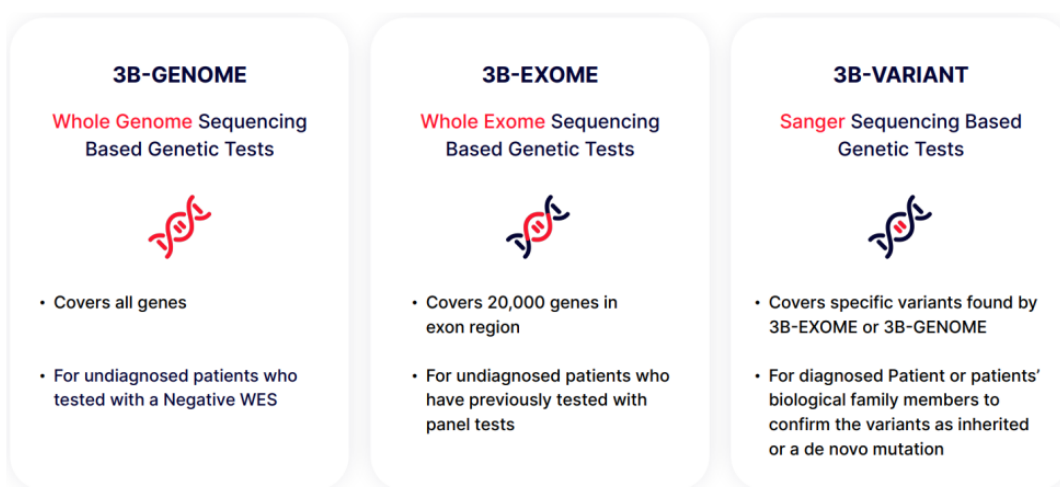
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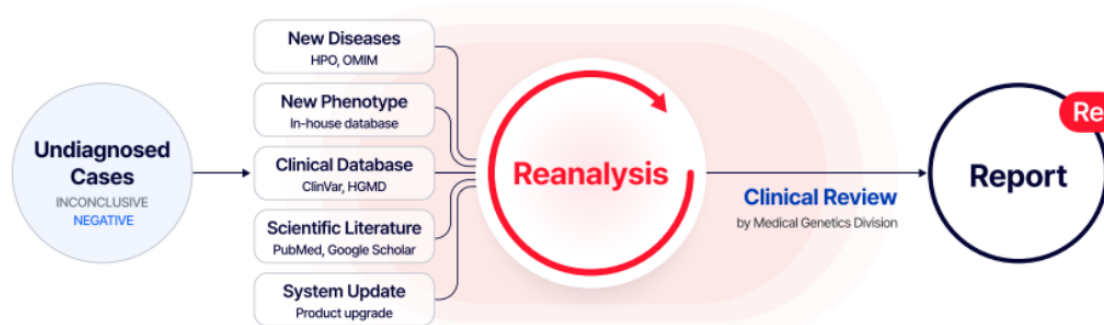
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Thyroid cancer
Diabetes type I
Diabetes type II

Gestational diabetes
Diabetic peripheral neuropathy
Diabetic nephropathy
Chronic kidney disease
Obesity
Lipid metabolism disorders
Stroke
Coronary heart disease
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Myocardial infarction
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Familial Hypercholesterolemia

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Poster Presentation Abstract

Genetics

Poster ID – P.G-01

CASE REPORT: COMPREHENSIVE PHYSICAL EXAMINATION AND GENETIC COUNSELING IN A PATIENT WITH EEC SYNDROME

Siti Farhanah Aulia¹, Sultana Faradz²

¹Master's Student in Genetic Counseling, Biomedical Sciences, YARSI University, Jakarta, ²Head of Genomic/Genetic Research Center of YARSI University Jakarta

*Corresponding author: farhanaaulia@rocketmail.com

Abstract

Introduction

EEC Syndrome (Ectrodactyly-Ectodermal Dysplasia-Cleft Lip/Palate Syndrome) EEC syndrome is very rare genetic disease characterized by limb abnormalities, ectodermal dysplasia, and cleft lip/palate. Mostly caused by mutations of the TP63 gene and can be autosomal dominant inheritance or spontaneous mutations.

Material and Methods

The study utilized a case report methodology, an 11-year-old girl diagnosed with EEC Syndrome, presented with abnormalities in her fingers and toes, abnormal teeth, as well as a cleft lip and palate. The study focused on her dysmorphology analysis and the genetic counseling process. The suspicion of a diagnosis was based on physical and clinical examinations, including dysmorphology examination, syndrome identification using Face2Gene software, and syndromic analysis using the Smith Recognizable Pattern book.

Results and Discussion

Based on physical examination and pedigree analysis, the patient shows a distinct set of characteristics. Her hair is light-colored, sparse, and thin, while dental issues include partial anodontia and microdontia. Ocular features consist of blepharophimosis and dry eyelids. Also Cleft lip and cleft palate. In the upper extremities, oligodactyly is observed, and ectrodactyly in the lower extremities. There are no similar complaints within her family across three generations. These physical findings have been confirmed using Face2Gene and syndromic analysis through the Smith Recognizable Pattern book. Genetic counseling played a crucial role in managing EEC Syndrome, informing the family about the condition, inheritance patterns, genetic factors, pregnancy risks, and treatment options. It also offered psychological support, acknowledging the emotional impact.

Conclusions

Managing EEC Syndrome involves a meticulous dysmorphology examination followed by genetic analysis and comprehensive genetic counseling. Pathogenic variants analysis especially for P63 is still on going. It is hoped that the patient and her family can face this condition with better understanding, receive appropriate care, and obtain the necessary support throughout the disease journey.

Keywords

EEC Syndrome, Dysmorphology, Genetic Counseling, p63, Rare Disease.

Poster ID – P.G-02

UNRAVELLING FAMILIAL MUSCULAR DISORDER: A JOURNEY THROUGH GENETIC COUNSELING AND CLINICAL INSIGHTS

Mentari Amir¹, Wan Nedra², Sultana Faradz³

¹Master's Student in Genetic Counseling, Biomedical Sciences, YARSI University, Jakarta, ² Pediatrician, YARSI Hospital, Jakarta, ³ Head of Genomic/Genetic Research Center of YARSI University Jakarta

*Corresponding author: mentariamir@gmail.com

Abstract

Introduction

Familial muscular disorder is a complex disease that often presents without a clear familial history, which makes diagnosis challenging. This study focuses on a familial case involving three brothers, all of whom display symptoms suggestive of a genetic muscular disorder. This emphasizes the need for detailed genetic counseling and a thorough clinical evaluation.

Materials and Methods

The approach included comprehensive pre-test genetic counseling, detailed clinical examinations, and diagnostic testing such as electromyography and serum enzyme assays. Clinical features assessed included muscle weakness, walking pattern, and presence of symptoms like pseudohypertrophy and Gower's sign. Genetic analysis targeting the neuromuscular disorder panel was planned to confirm the diagnosis.

Results and Discussion

The clinical evaluation suggested characteristics of Spinal Muscular Atrophy (SMA) Type 3, particularly considering the symptom onset and progression. Elevated creatine kinase (CK) levels were noted, especially in the second child, which suggested possible Duchenne Muscular Dystrophy (DMD). However, the absence of classical DMD signs complicated the diagnosis. Electromyography did not conclusively support a diagnosis of either SMA or DMD, indicating the necessity for genetic analysis to achieve a definitive diagnosis.

Conclusions

This case underscores the critical role of genetic counseling combined with a multidisciplinary clinical approach in diagnosing muscular dystrophy, particularly in familial contexts where the genetic basis is not immediately apparent. It emphasizes the importance of a meticulous diagnostic process to effectively manage and counsel families affected by such hereditary neuromuscular disorders. Additionally, the findings suggest a potential overlap in clinical manifestations among different types of muscular dystrophies, demonstrating the complexity involved in diagnosing these conditions.

Keywords

Familial Muscular Disorder, Genetic Counseling, Muscle Weakness, Spinal Muscular Atrophy, Duchenne Muscular Dystrophy

Poster ID – P.G-03

BIALLELIC VARIANTS IN *GLB1* CAUSES GM1 GANGLIOSIDOSIS DISEASE IN A FAMILY WITH TWO SIBLINGS

Shankar Aissvarya¹, Chong-Teik Lim¹, Meow-Keong Thong², Noraishah Mydin Abdul-Aziz³, King-Hwa Ling^{1,4}, Karuppiah Thilakavathy^{1,4*}

¹Department of Biomedical Science, Faculty of Medicine and Health Sciences, Universiti Putra Malaysia, 43400 UPM Serdang, Malaysia, ²Department of Population Medicine, M. Kandiah Faculty of Medicine and Health Science, Universiti Tunku Abdul Rahman, 43000 Kajang, Malaysia, ³Associate Research Fellow, Faculty of Medicine and Health Sciences, Universiti Putra Malaysia, 43400 UPM Serdang, Malaysia, ⁴Genetics and Regenerative Research Group, Faculty of Medicine and Health Sciences, Universiti Putra Malaysia, 43400 UPM Serdang, Malaysia

*Corresponding author: thilathy@upm.edu.my

Abstract

Introduction

GM1 gangliosidosis, is a lysosomal disease characterised by a build-up of GM1 ganglioside due to deficiencies in the β -galactosidase class of enzymes¹. It is caused by variants within the *GLB1* gene, resulting in production of a defective form of the β -galactosidase enzyme². This subsequently impairs cell physiology causing progressive destruction of the nerve cells in the central nervous system³. Hence, this study aims to identify the genetic variants responsible for GM1 gangliosidosis in a family with two affected children. The proband, a 27-year-old man (age at diagnosis: 16-years-old), presented with learning difficulties and dystonic movement with severely reduced β -galactosidase activity. His 37-years-old brother, affected by the same condition, presented with a more severe phenotype. Both affected brothers showed progressive deterioration while both parents, older sister and older brother are unaffected.

Materials and Methods

Buccal swabs were collected from the proband, affected brother and parents. The proband's DNA was sent for whole-exome sequencing (WES) followed by variant identification through bioinformatics analyses. The identified variants were validated through Sanger sequencing, performed on proband and family members.

Results and Discussion

WES genetic analysis revealed the proband has a heterozygous pathogenic variant, (NM_000404.4:c.1325G>A, p.Arg442Gln), and a heterozygous variant of uncertain significance (VUS) (NM_000404.4:c.1022G>T, p.Gly341Val), in the *GLB1* gene. Sanger sequencing validated the variant found via WES. The affected brother carried the biallelic variants as the proband. The mother is heterozygous for the pathogenic variant while the father is heterozygous for the VUS. Interestingly, a previous study⁴, found the same compound heterozygous variants in two GM1 gangliosidosis patients within the same family.

Conclusions

These findings not only confirmed the variants involved in GM1 gangliosidosis within this family but also emphasise the importance of comprehensive genetic analyses, particularly in unravelling inherited variants to facilitate appropriate genetic counselling and treatment options, including gene therapy.

Keywords

GM1 gangliosidosis, *GLB1*, biallelic variant, whole exome sequencing, β -galactosidase

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Poster ID – P.G-04

FAMILIAL INTELLECTUAL DISABILITY WITH SUSPECTED FRAGILE X SYNDROME

Meutia Lieska Urfa¹, Sultana Faradz²

¹Master's Student in Genetic Counseling, Biomedical Sciences, YARSI University, Jakarta, ²
Head of Genomic/Genetic Research Center of YARSI University Jakarta

Email: lieska279@gmail.com

Abstract

Introduction

Familial Intellectual Disability (FID) is a complex genetic condition within families and often has associations with Fragile X Syndrome (FXS), a genetic disorder causing developmental brain impairments and limited cognitive abilities. Fragile X Syndrome (FXS) is one of the major causes of FID, but there are cases where similar symptoms occur without mutations in the *FMRI* (Fragile X Messenger Ribonucleoprotein) gene responsible for FXS.

Materials and Methods

We conducted a case study on an 11-year-old male patient exhibiting typical clinical symptoms of FXS, including learning disabilities, behavioral issues, and physical characteristics associated with the disorders. Based on the medical history and pedigree we gathered that the patient's cousin also has the same typical clinical symptoms of FXS, and the patient's aunt also has behavioral disorders and learning disabilities, resulting in her inability to engage in independent activities. A test was performed to search for mutations in the *FMRI* gene.

Results and Discussion

The patient exhibited clinical manifestations consistent with FXS, however no mutations in the *FMRI* gene were detected. This indicates the possibility of gene variants or other genetic mechanisms underlying the clinical manifestations of FXS in the patient.

Conclusions

This finding suggests that despite the absence of mutations in the *FMRI* gene, patients can exhibit the typical clinical manifestations of FXS. The limitation of this case study did not conduct cytogenetic testing before examining *FMRI*. It is important to rule out the possibility of chromosomal abnormalities that could cause symptoms similar to FXS, especially considering there are family members of mother site also exhibit similar symptoms. The use of advanced diagnostic methods such as Chromosomal Microarray Analysis (CMA) probably confirming the diagnosis and guiding appropriate patient management.

Keywords

Familial Intellectual Disability, Fragile X Syndrome, *FMRI* gene mutation, cytogenetic testing, Chromosomal Microarray Analysis

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Poster ID – P.G-05

FREQUENCY OF IS6110 AND IS1081 RECOMBINASE POLYMERASE AMPLIFICATION IN MYCOBACTERIUM TUBERCULOSIS COMPLEX POSITIVE PATIENTS AT KOLONEL ABUNDJANI REGIONAL HOSPITAL

Derallah Ansusa Lindra^{1,2}

¹Pulmonologist at RSUD Kolonel Abundjani Bangko, ²Biomedical Science Doctoral Program
of Yarsi University

*Corresponding author: ansusadera@yahoo.co.id

Abstract

Introduction

Tuberculosis (TB) caused by *Mycobacterium Tuberculosis* (MTB) weighs heavily on world's public health security. Tuberculosis is an infectious disease that is often difficult to diagnose. An efficient, specific, convenient and inexpensive MTB test is needed to prevent and control TB. Recombinase Polymerase Amplification (RPA) is isothermal DNA amplification reaction which uses low temperatures, fast and sensitive compared to other amplification technologies. The aim of this research is to assess frequency of IS6110 and IS1081 Recombinase Polymerase Amplification in Mycobacterium Tuberculosis complex positive patients at Kolonel Abundjani Regional Hospital

Materials and Methods

This research used a retrospective descriptive cross-sectional study. Secondary were taken at pulmonology clinic from August 2023 until April 2024. TB is diagnosed by using Xpert MTB-RIF Ultra to evaluate the IS6110 and IS1081 Polymerase Recombinase Amplification.

Results and Discussion

There were 60 positive MTB cases during August 2023 to April 2024, consisting of 42 patients (70%) men and 18 patients (30%) women. The highest age range was 36-55 years and > years as many as 27 patients (45%). Total of 47 patients (78.3%) had no history of previous MTB treatment. Eighty percent (80%) or 48 patients have no comorbidities in positive MTB patients. The median endpoint value of the growth curve (EndPt) rpoB1, rpoB2, rpoB3 dan rpoB4 were 449,5; 302,5; 213; and, irrespectively. 136. IS6110 and IS1081 Polymerase Recombinase Amplification are the most common medium Cycle threshold, that is 54 patients (90%), consist of 54 patients (90%), male gender was 70% and the age range from 36 to 55 was 46.3%.

Conclusions

Mycobacterium tuberculosis DNA complex detection using IS6110 and IS1081 Polymerase Recombinase Amplification are most frequently detected in cycle threshold medium (16-22). The gender group that is mostly detected is men that takes 70%, with age range between 36 – 55 years as many as 46.3%.

Keywords

Recombinase Polymerase Amplification (RPA), IS6110 and IS1081 RPA, *Mycobacterium Tuberculosis* kompleks, RPA Cycle threshold, DNA *Mycobacterium Tuberculosis* detection. .

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Poster ID – P.G-06

THE IMPORTANT ROLE OF GENES IN DETERMINING TREATMENT IN GRAVES' PATIENTS

Fatimah Eliana¹

¹Faculty of Medicine, Universitas YARSI, Jakarta

*Corresponding author: fatimaheliana@yarsi.ac.id

Abstract

Introduction

Graves' Disease (GD), also known as toxic diffuse goiter, is inherited as a complex multigenic disorder, with the presence of antibodies acting as agonists against the thyrotropin receptor in thyroid gland. This study aimed to evaluate the role of genetic factors affecting the relapse of patients with Graves' disease in Indonesia, included protein tyrosine phosphatase-22 (PTPN22), cluster of differentiation 40 (CD40), the cytotoxic T lymphocyte- associated factor-4 (CTLA4 or CD152), thyrotropin receptor (TSHR), thyroglobulin (Tg) and FCRL3 (FC receptor-like-3).

Material and Methods

This was a retrospective study that were collected from 140 GD patients, 72 GD patients who recurred and 68 GD patients who did not relapse after no longer taking anti thyroid drugs for more than 1 years. Genetic polymorphism examination was performed using PCR-RFLP. The logistic regression was used since the dependent variables were categorical variables.

Results and Discussion

The analysis of this study demonstrated that there was a correlation between relapse of disease and C/C genotype of PTPN22 gene, C/C genotype of CD40 gene, G/G genotype of CTLA-4 gene, C/C genotype of TSHR gene, C/C genotype of thyroglobulin gene ($p=0.012$), and G/G genotype of FCRL3 gene

Conclusions

This study confirms the usefulness of genotype in predicting recurrence and determining treatment in Graves' patients.

Keywords

Graves' Disease; PTPN22 gene; CD40 gene; CTLA-4 gene; TSHR gene; Thyroglobulin gene; FCRL3 gene

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Cancer Biology and Cancer Therapeutics

Poster ID – P.C-01

Cellular and Molecular Research, Immunology

Poster ID – P.M-01

DERMAL FIBROBLAST PROLIFERATION CHARACTERISTIC IN A LONG-TERM CONTINUOUS CULTURE SYSTEM

Marisa Riliani, Indra Kusuma, Endang Purwaningsih, Muhammad Samsul Mustofa

*Corresponding author: marisa.riliani@yarsi.ac.id

Abstract

Introduction

It is still unclear how we can indicate cell behaviour in every passage. The passage numbers can be a good barometer but it is misleading. We should not rely on guess work and predict when we should expect to start a batch for preservation, or the optimal passage for experiment. The amount of time for one cell to double known as population doubling (PD) should allow us to characterise the cellular behaviour between every passage. This is a good indication of a cellular behaviour, whether it should be preserved as a master cell bank (MCB), to be used as a working cell bank (WCB), or has been degenerate as an unstable cell bank (UCB).

Materials and Methods

The human fibroblast cells collected from the preputium of a 10-year-old male was cultured in Dulbecco's Modified Eagle's Medium (DMEM), supplemented with 10% fetal bovine serum (FBS) and Antibiotic-Antimycotic (AA). Cells were maintained at 37°C with 5% CO₂ in a humidified chamber and sub-cultured at 80–90% confluency. Culture media was replaced every two days. Cells were sub-cultured from passage 1 to passage 18. The amount of harvesting cells were counting every week with the formula of PD [$\text{Log}_{10}(\text{harvesting cell No}/\text{seeding cell No})/\text{Log}_{10}(2)$]. This descriptive study was analyzed by one-way ANOVA.

Results and Discussion

The mean value of PD for passages 1-5 was 52.92, passages 6-11 was 61.48, and passages 12-18 was 98.03. Statistically shows that there is a significant difference in the mean value of PD based on the three groups, $P=0.003$. The post-hoc shows that there are significant differences between the MCB with UCB groups and the WCB with UCB groups, but there is no difference between the MCB with WCB groups. It is indicated that below passage 11 the cells are in the optimal condition for preservation and working batch, but above it, the cell has been changing in the behavior of doubling capacity, so it is only can be used for restricted applications.

Conclusions

The application of fibroblast cells can be classified based on population doubling criteria which shows that in passages 1-5 they can be preserved as a master cell bank (MCB), passages 6-11 can be used as a working cell bank (WCB) and in passages 12-18 there has been a change in behavior, as an unstable cell bank (UCB).

Keywords

Fibroblast, Passage, Population, Doubling, Characterization

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Poster ID – P.M-02

IMMUNE RESPONSE PROFILE OF SARS-COV-2 VACCINATION AGAINST ANIMAL MODEL AND HUMAN USING ELISPOT METHOD

Astria Novitasari Nidom^{1,2}, Balqis Afifah¹, Dhani Prakoso¹, Ilsan Arvan Nurgas³, Nor Laela Ramadhaniyah¹, Setyarina Indrasari¹, Chairul Anwar Nidom^{1,3}, Reviany Vibrianita Nidom^{1*}

¹Professor Nidom Foundation, Surabaya, Indonesia, ²Doctoral Program, Faculty of Medicine, Airlangga University Surabaya, Indonesia, ³Doctoral Program, Faculty of Veterinary Medicine, Airlangga University Surabaya, Indonesia

*Corresponding author: reviany@pnfinstitute.org

Abstract

Introduction

Severe acute respiratory syndrome coronavirus 2 (SARS-CoV-2) infection has afflicted millions of people in a worldwide pandemic. ELISpot is a highly quantitative method that can measure various response magnitudes and assess important cellular immune-related activities such as IFN- γ secretion. This study aimed to find out the profile of immune response after COVID-19 vaccination in mice and humans by ELISpot method.

Material and Methods

An experimental study with a post-test-only control group design using 90 mice (*Mus musculus* L.). The test group consisted of 9 groups (P1-P9) also 100 blood samples with details that were vaccination only, vaccinated and had infected with COVID-19 or survivors, unvaccinated only, and unvaccinated but survivors. Furthermore, we analyzed the IFN- γ profile in mice and human PBMC using ELISpot reader by Mabtech.

Results and Discussion

Animal model study

The highest mean IFN- γ profile was obtained in the treatment group injected with 50 μ g VLPs (P5) and the lowest in the treatment group injected with PBS (P1). Human immune profile study. The data were obtained from the lowest and the highest value of ELISpot measurement in each of groups.

Conclusion

IFN- γ is a promoter of all immune system regulations when there are viral antigens that enter the body. ELISpot can be considered a method to analyze cellular immune profiles in animal models and humans.

Keywords

Sars-cov-2, vaccination, immune response, ELISpot

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Poster ID – P.M-03

THYMOQUINONE EFFECT ON THE CELL VIABILITY AND REACTIVE OXYGEN SPECIES (ROS) LEVEL IN BRIN-BD11 INDUCED BY ALLOXAN

Weni, Linda¹ ; A.Jusman, Sri Widia² ; Sadikin, Mohamad² ; Iskandriati Diah³

¹Department of Biochemistry, Faculty of Medicine, YARSI University, Jakarta 10510, Indonesia, ²Department of Biochemistry, Faculty of Medicine, Indonesia University, Jakarta, 10430, Indonesia, ³Primate Research Center, Bogor Agricultural University, Bogor 16151, Indonesia

*Corresponding author: lindaweni@yarsi.ac.id

Abstract

Introduction

Oxidative stress is known to play a crucial role in the pathogenesis of diabetes and its complications. The study aimed to evaluate the impact of Thymoquinone (TQ) on cell viability and reactive oxygen species (ROS) level in diabetes mellitus. This study designed a way to investigate BRIN-BD11 cells by using electrofusion of rat pancreatic β -cell line RINm5F with a cell line from pancreatic islet cell of mice from New England Deaconess Hospital (NEDH) induced by alloxan to generate reactive oxygen species (ROS).

Material and Methods

The in-vitro study used BRIN BD11 cell culture, which was incubated for 48 hours with TQ at a dose of 1 to 20 μ M after 48 hours of incubation with 8.8 mM ALX. The cell viability was measured using the trypan blue dyeing method, and the level of oxidative stress assays used DCFDA (2',7', dichlorofluorescein diacetate) kit with spectrofluorometry method.

Result and discussion

The results showed that incubation with alloxan (ALX) 8.8 mM for 48 hours decreased the viability of BRIN BD11 cells. However, administration of TQ in doses 1-20 μ M increased the cell viability significantly. Treatment with TQ 1;5 μ M increased significant cell viability compared to the ALX group ($p = 0.009$; $p < 0.05$). Moreover, treatment with TQ after 48 hours of incubation with alloxan 8.8 mM decreased ROS level but not significantly ($p > 0.05$).

Conclusion

Thymoquinone acted as an antioxidant and helped overcome the effects of oxidative stress on pancreatic β -cells due to ALX induction.

Keywords

Cell viability, ROS, BRIN-BD11, Alloxan, TQ

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Poster ID – P.M-04

THE EFFECTIVENESS LOTUS LEAF (*Nymphaea pubescen L*) EXTRACT ON THE GROWTH OF SHIGELLA DYSENTERIAE AND SALMONELLA ENTERICA SEROVAR TYPHIMURIUM IN VITRO

¹Intan Keumala Dewi, ²Akram Ushaim Zuhdi, ²Adira Hayyu Putri Hidayah, ¹Ike Irmawati Purbo Astuti, ³Firman Arifandi

¹Department of Microbiology, Faculty of Medicine, YARSI University, Jakarta, Indonesia, ²Faculty of Medicine, YARSI University, Jakarta, Indonesia, ³Department of Islam, Faculty of Medicine, YARSI University, Jakarta, Indonesia

Abstract

Introduction

Diarrhea can be caused by *Shigella dysenteriae* and *Salmonella enterica* serovar typhimurium. One of the traditional treatments for diarrhea is lotus leaves. Considering that several antibiotics are currently experiencing resistance, lotus leaves with their antimicrobial content are expected to be able to inhibit the growth of *Shigella dysenteriae* and *Salmonella enterica* serovar typhimurium. The aim of this research was to determine the effectiveness of lotus leaf extract (*Nymphaea pubescen L*) as an antibacterial against *Shigella dysenteriae* and *Salmonella enterica* serovar Typhimurium.

Material and Methods

The research conducted was an experimental laboratory study using the disk diffusion method. This research was carried out with the preparation of Lotus (*Nymphaea pubescen L*) extract with a concentration 3000 ppm, 6.000 ppm, 15.000 ppm, and 25.000 ppm as well as a positive control of the antibiotic ciprofloxacin and a negative control of the CMC emulsifier which was then tested against these two bacteria and the size of the inhibition zone formed on the MHA was measured.

Results and Discussion

The results showed that in the lotus leaf extract on *Shigella dysenteriae*, the results of measuring the diameter of the inhibition zone formed in MHA from lotus leaf extract with a concentration of 3000 ppm and 600 ppm did not form an inhibition zone. Whereas in lotus leaf extract with a concentration of 15.000 ppm and 25.000 ppm an inhibition zone was formed. In *Salmonella enterica* serovar Typhimurium lotus leaf extract, no inhibition zone was found at the four concentrations of lotus leaf extract.

Conclusions

Lotus leaf extract is effective on *Salmonella enterica* serovar Typhimurium but not effective on bacteria.

Keywords

Lotus Leaf, *Nymphaea pubescen L*, *Shigella dysenteriae*, *Salmonella enterica* serovar typhimurium

Pharmaceutical Sciences, Phytopharmacy and Herbal medicine

Poster ID – P.P-01

EXPRESSION OF HEMATOPOIETIC STEM CELLS AND STEMNESS MARKERS IN HUMAN MILK-DERIVED CELLS

Dewi Sukmawati^{1*}, Dara Yudha Nur Fadhilah²

¹Department of Histology, ²Master Program in Biomedical Science, ²Faculty of Medicine, Universitas Indonesia, Jl Salemba Raya No.6 Jakarta 10430

*Corresponding author: dewi.sukmawati@ui.ac.id

Abstract

Introduction

Human (breast) milk (HM) is important for infant nutrition, immunity, and development. Recent studies have revealed that HM also contains a heterogeneous population of living cells such as progenitor/stem cells. Hematopoietic stem cells (HSCs) as assessed by CD34, are expressed in the umbilical cord and appear to have a supportive role in fetal development due to their ability to differentiate into both hematopoietic and non-hematopoietic cells such as microglia, hepatocytes, and type-2 alveolar cells. Little is known about CD34 expression in the cellular component of HM. The aim of this study is to investigate the expression of CD34⁺ cells and stemness markers in human milk-derived cells.

Materials and Methods

This is an exploration study. All procedures in this study were approved by the Ethics Committee of the Faculty of Medicine Universitas Indonesia. We collected fresh human milk from 1- 3 months post-partum mothers who voluntarily participated in this study, and both the baby and mothers were healthy. The milk is then centrifuged, and the cellular components are obtained in pellet. Cells were then analyzed for CD34⁺ cells by flow cytometry and the stemness markers (NANOG, OCT4) by RT-qPCR.

Results and Discussion

We received and collected human milk samples from eight volunteer donor mothers with an average age of 26 years. Fresh cells derived from human milk positively express CD34 marker and the average population is approximately 1.5%. These cells also expressed the stemness markers of NANOG (1.6-folds) and OCT4 (1.05-folds).

Conclusion

Human milk-derived cells express CD34 and stemness markers. Further studies are needed to reveal the potential of human milk-derived cells as a source of hematopoietic stem cells.

Keywords

Human milk, cellular components, CD34, hematopoietic stem cell, stemness.

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Poster ID – P.P-02

EFFECT OF SERUM-FREE CULTURE MEDIUM ON SERUM RESPONSE FACTOR (SRF) EXPRESSION IN MESENCHYMAL STEM CELL (MSC) PRODUCTION CULTURE AS ADVANCED THERAPEUTIC MEDICINAL PRODUCT (ATMP)

Indra Kusuma^{1,2,3} Eko Poerwanto¹ Nur Ilham Risma Hidayati²

¹Physiology Department, Faculty of Medicine, YARSI University, Jakarta, Indonesia, ²Stem Cell Research Center, YARSI University, Jakarta, Indonesia, ³Research and Education Department, YARSI Hospital, Jakarta, Indonesia

*Corresponding author: indra.kusuma@yarsi.ac.id

Abstract

Introduction

Wharton's jelly mesenchymal stem cell (WJMSC) and human dermal fibroblast (HDF) cultures can be used as advanced therapeutic medicinal products (ATMPs). Fetal bovine serum (FBS) as a cell culture supplement derived from animals, has a composition that is not constant, causing variations in research results. Therefore, replacement supplements such as knockout serum replacement (KOSR) are needed. This study evaluated KOSR as an FBS replacement for WJMSC and HDF culture by measuring SRF mRNA relative expression and WJMSC cell proliferation.

Material and Methods

WJMSC and HDF cells were divided into an intervention group with KOSR 10% and a control group with FBS 10%. Reverse transcription polymerase chain reaction (RT-PCR) was used for SRF mRNA relative expression at the sixth-seventh passage (P6/P7) for WJMSC, and P7 for HDF. Cell counting kit-8 (CCK-8) reagent was used for the WJMSC cell proliferation at P6/P7. Data were analyzed using one-way ANOVA, or Kruskal-Wallis test.

Results and Discussion

There was no significant difference in SRF mRNA relative expression between the treatment groups of WJMSC KOSR 10%, WJMSC FBS 10%, HDF KOSR 10%, and HDF FBS 10% ($p > 0.05$). The proliferation assay results showed a significant difference in viability between the KOSR 10% and FBS 10% treatment groups ($p < 0.05$).

Conclusions

The KOSR 10% was comparable to FBS 10% in supporting SRF mRNA relative expression in WJMSC and HDF cultures, and cell proliferation in WJMSC cultures.

Keywords

WJMSC, HDF, FBS, KOSR, and SRF mRNA relative expression

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Poster ID – P.P-03

CYTOTOXICITY OF ETHANOL EXTRACT LONTAR FRUIT SEED COAT (*Borassus flabellifer L.*) AS AN ANTICANCER AGENT AGAINST HUMAN DERMAL FIBROBLAST CELL

Livia Aqilla Hamiida¹ Indra Kusuma^{2,3} Dina Almunawarah¹ Anika Rizki³

¹Faculty of Medicine, YARSI University, Jakarta, Indonesia, ²Physiology Department, Faculty of Medicine, YARSI University, Jakarta, Indonesia ³Stem Cell Research Center, YARSI University, Jakarta, Indonesia

*Corresponding author: indra.kusuma@yarsi.ac.id

Abstract

Introduction

Cancer treatment therapy using synthetic drugs has bad side effects on normal cells due to non specific tissue distribution. Therefore, herbal plants are needed as a substitute for synthetic drugs. This research is to discover the ethanol extract of lontar seed coat (*Borassus flabellifer L.*) as an anticancer agent against Human Dermal Fibroblast (HDF) cells.

Materials and Methods

HDF cells were divided into groups of Complete DMEM medium control, 3mM doxorubicin control, solvent control, medium control and lontar fruit seed coat ethanol extract at doses of 1,500 µg/mL, 750 µg/mL, 325 µg/mL, 187.5 µg/mL, 93.75 µg/mL. Cell counting reagent kit 8 (CCK-8) was used for HDF cell viability. Data were analyzed using One-Way ANOVA.

Results and Discussion

We observed that toxicity increased along with a significant increase in the concentration of lontar fruit seed coat ethanol extract. The toxicity of the ethanol extract of lontar fruit seed peel at a dose of 187.5 µg/mL did not have a significant difference with a doxorubicin concentration of 3 mM ($p > 0,05$), and the control solvent used did not have a toxic effect on HDF.

Conclusions

The use of lontar fruit seed coat ethanol extract as an anticancer agent can cause side effects on the dermal layer

Keywords

HDF, lontar fruit seeds coat (*Borassus flabellifer L.*), viability, CCK-8

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CYTOTOXICITY OF ETHANOL EXTRACT LONTAR FRUIT SEED COAT (*Borassus flabellifer L.*) AS AN ANTICANCER AGENT AGAINST HUMAN EPIDERMAL KERATINOCYTE CELL

Dina Almunawarah¹, Indra Kusuma^{1,2}, Livia Aqilla H¹, Anika Rizki³

¹ Faculty of Medicine, YARSI University, Jakarta, Indonesia, ²Physiology Department, Faculty of Medicine, YARSI University, Jakarta, Indonesia, ³Stem Cell Research Center, YARSI University, Jakarta, Indonesia

*Corresponding author: indra.kusuma@yarsi.ac.id

Abstract

Introduction

Chemotherapy for cancer treatment is very toxic to normal cells because it is given intravenously and easily causes greater damage to healthy tissues, and some side effects such as pain, nausea, hair loss, skin discoloration, and dryness. An alternative that can be used is traditional medicine using ethanol extract of *Borassus flabellifer L.* seed coat which contains flavonoids, phenols, and tannins, based on Molyneux's antioxidant properties indicator, showing very strong efficacy as an anti-cancer treatment. This study will look at the potential adverse effects of the lontar fruit seed coat (*Borassus Flabellifer L.*) extract on human epidermal keratinocytes.

Materials and Methods

Human Epidermal Keratinocyte cells were thawed and seeded 15,000 cells/well and 200 μ L volume into a 96-well multi-plate with 5 repetition groups, using 8 treatment groups of lontar fruit seed coat extract (*Borassus flabellifer L.*) at a dose of 93.75 μ g/ml, 187.5 μ g/ml, 375 μ g/ml, 750 μ g/ml, 1500 μ g/ml, positive control with no treatment, negative control with doxorubicin 3mM administration, and solvent control.

Results and Discussion

There are differences in viability that occur among the 8 treatment groups which are increased viability occurred at a dose of 1500 μ g/ml, and positive control DKSFM, then decreased viability at doses of 375 μ g/ml, 187.5 μ g/ml, 93.75 μ g/ml. We observed that the higher the dose of extract used, the lower the toxicity to the epidermis.

Conclusions

In conclusions, no toxicity was seen at the 1500 μ g/ml dose, but toxicity was seen at the 375 μ g/ml, 187.5 μ g/ml, and 93.75 μ g/ml doses. If used for cancer therapy, no adverse effects on the epidermis were seen when using a dose of 1500 μ g/ml.

Keywords

HEK, Lontar fruit seeds coat (*Borassus flabellifer L.*), CCK-8, Viability.

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POTENTIAL FOR ADMINISTERING A COMBINATION OF SULFADIAZINE AND SECRETOME IN ITS SYNERGISTIC PERFORMANCE FOR HEALING BURNS: A THEORETICAL REVIEW

Yohanes Firmansyah^{1*}, Irawan Yusuf², Sukmawati Tansil Tan³

¹Postgraduate (Doctoral) Student, Faculty of Medicine, Hasanuddin University, Makassar, Indonesia,

²Department of Physiology, Faculty of Medicine, Hasanuddin University, Makassar, Indonesia,

³Department of Dermatology and Venereology, Faculty of Medicine, Tarumanagara University, Jakarta, Indonesia

*Corresponding author: yohanesfirmansyah28@gmail.com

Abstract

Burns are among the most serious injuries, necessitating an efficient treatment strategy to accelerate healing and limit the risk of complications. Sulfadiazine, a compound with antibacterial characteristics, has long been used to treat burns. On the other hand, the secretome, which is high in growth factors and cytokines, offers promise regarding tissue regeneration. This study looks at the synergy of sulfadiazine and secretome, specifically their efficacy in lowering infection, speeding re-epithelialization, reducing inflammation, and minimizing scar formation. This study examines how sulfadiazine and growth factors (TGF- β , PDGF, KGF, FGF, Pro Collagen, and VEGF) play a crucial role in wound healing. This study found that combining sulfadiazine with secretome could be a more successful burn wound treatment. These findings suggest that innovative topical formulations could help to enhance the standard of treatment for burn victims. However, more research is needed to evaluate the efficacy of these mixes in clinical trials and to investigate their molecular mechanisms of action in greater detail.

Keywords

Sulfadiazine; growth factor; secretom; wound healing; burns

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Poster ID – P.P-06

THE ROLE OF PROPOLIS (BRITISH PROPOLIS) IN MODULATING OXIDATIVE STRESS IN ENDOMETRIOSIS: INSIGHTS INTO GLUTATHIONE PEROXIDASE (GPX) AND 8-HYDROXY-DEOXYGUANOSINE (8-OHDG) LEVELS

Dwirini Retno G^{1,2*}, Arifah Shabrina³, Nurul Ratna MM³, Eka Rusdianto G⁴

¹Department of Biochemistry and Molecular Biology, University of Indonesia-Dr. Cipto Mangunkusumo Hospital, Jl. Salemba Raya No.6, Central Jakarta, DKI Jakarta 10430, Indonesia, ²Center of Hypoxia & Oxidative Stress Studies, Faculty of Medicine, University of Indonesia, Jl. Salemba Raya no.6, Central Jakarta, DKI Jakarta 10430, Indonesia, ³Department of Nutrition, University of Indonesia-Dr. Cipto Mangunkusumo Hospital, Jl. Salemba Raya no.6, Central Jakarta, DKI Jakarta 10430, Indonesia, ⁴Department of Obstetrics and Gynecology, University of Indonesia-Dr. Cipto Mangunkusumo Hospital, Jl. Salemba Raya no.6, Central Jakarta, DKI Jakarta 10430, Indonesia

*Corresponding author: rinairret@gmail.com

Abstract

Introduction

Endometriosis is a persistent gynecological condition often exacerbated by oxidative stress, characterized by increased levels of reactive oxygen species. This imbalance leads to significant cell damage and affects the body's natural antioxidant defenses, including glutathione peroxidase (GPx). Consequently, 8-hydroxy-2'-deoxyguanosine (8-OHdG) emerges as a critical biomarker in this context. Levonorgestrel (LNG) plays a role in activating apoptosis of endometriosis lesions by regulating estrogen hormone fluctuations through follicle-stimulating hormone and luteinizing hormone. Propolis, a bee-derived product, stands out for its strong antioxidant properties that can inhibit the growth of disease-related cells. Additionally, our research explores the effect of catalase, an antioxidant enzyme, in reducing oxidative stress in endometriosis.

Materials and Methods

This study employed a clinical trial design with random allocation and double-blinding. 24 women undergoing LNG therapy for endometriosis were randomly assigned to receive either propolis containing 17.5 mg of flavonoids per drop or a placebo. The intervention was administered twice daily, in the morning and at night, with a dosage of 1 drop/10 kg body weight each time. Blood samples were taken at the initial visit and 30 days thereafter.

Results and Discussion

The results indicated a significant difference between the two groups in GPx and 8-OHdG levels ($p < 0.05$), although glutathione decreased by 0.01 (-0.01 to -0.037) $\mu\text{g/mL}$ after 4 weeks of intervention. The 8-OHdG levels showed a greater decrease in the propolis group by 17.30 ng/mL (-13.58 to 37.19) ng/mL compared to the placebo group.

Conclusion

The administration of flavonoids in propolis resulted in significant changes in GPx and 8-OHdG levels during the 4-week intervention period.

Keywords

Endometriosis, propolis, stress oxidative, glutathione peroxidase, 8-OHdG

Poster ID – P.P-07

EFFECT OF SALUANG BALUM (*LAVANGA SARMENTOSA*) ROOT EXTRACT ON STEROIDOGENESIS: FOCUS ON STAR, CYP11A EXPRESSION AND LH LEVEL

Dwi Ari Pujianto^{1*}, Ahmad Solihin², Silvani Permatasari³¹Department of Medical Biology, Faculty of Medicine, Universitas Indonesia, ²Master Programme for Biomedical Sciences, Faculty of Medicine, Universitas Indonesia,²Department of Biology, Faculty of Medicine, Universitas Palangka Raya*Corresponding author: dwi.ari@ui.ac.id

Abstract

Introduction:

Male factors contribute around 50% of infertility cases. Various efforts continue to be made to increase male fertility. Saluang balum root (*Lavanga sarmentosa*, *Blume kurz*) is one of the endemic plants in Central Kalimantan and has long been used by the Dayak people of Kalimantan to increase sexual activity and cure sexual dysfunction. Male fertility and sexual activity are greatly influenced by the steroid hormone testosterone. However, until now it is not known whether saluang balum root extract can influence the steroidogenesis pathway and the hypothalamic-pituitary-gonadal axis. The aim of this research was to analyze the effect of saluang balum root extract on StAR and CYP11A expression and LH level.

Materials and Methods:

Twenty-eight mice were used in this study and grouped into 7 groups containing 4 mice each. The groups were untreated control, vehicle and 5 groups were given saluang balum root extract at 100, 200, 300, 400, and 500 mg/Kg bw orally for 24 days. On the 25th day, the mice were euthanized, their blood was taken for measuring LH level. Testis were also isolated for total RNA isolation for analysing STAR and CYP11A mRNA expression.

Results and Discussion

LH levels increased significantly compared to the control with the highest on 100 mg/Kg bw concentration ($p < 0.05$). There was a significant increase of STAR and CYP11A mRNA expression compared to control ($p < 0.05$) with the highest at the concentration of 100 mg/Kg bw and 200 mg/Kg bw, respectively. Positive correlation was observed between increased LH levels and STAR expression.

Conclusions:

The stimulation effect of saluang balum root extract on male reproduction may work through steroidogenesis pathway activating STAR and CYP11A genes. Moreover, saluang balum also works through pituitary gonadal axis indicating by increase in LH levels.

Keywords

Steroidogenesis, saluang balum, STAR, CYP11A

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Health Promotion, Health Policy, and Education

Poster ID – P.H-01

HEMOGLOBIN LEVELS IN ELDERLY WOMEN WITH VITAMIN D SUPPLEMENTATION IN URBAN AND RURAL AREASDiniwati Mukhtar¹, Nunung Ainur Rahmah¹, Linda Weni¹, Himmi Marsiati¹,
Sri Wuryanti¹, Desi Ariyani², Sabarina Elprida Manik²¹ Biomedical Science Study Program, Doctoral Program, YARSI University, ² Doctoral
Candidate Biomedical Science YARSI University*Corresponding author: diniwati.mukhtar@yarsi.ac.id*Abstract**Introduction*

An elderly woman is someone who has reached the age of 60 years or above. Increasing age is unavoidable, and degenerative diseases will appear. Nutritional patterns play an important role in promoting healthy aging. Based on research, Vitamin D can induce erythropoiesis by increasing erythropoietin levels. This study aims to see the effect of vitamin D supplements on hemoglobin levels of elderly people in urban and rural areas.

Materials and Methods

20-day human experiment on rural and urban elderly women groups, 25 people each. The vitamin D dose were 800 IU'. Before the research began, informed consent was given. Physical examination, physiological parameters and Hb were carried out before and after the study. Statistical analysis using SPSS 25.

Results and Discussion

The values obtained between the urban and rural groups were not significantly different ($p > 0.05$) for systolic pressure and BMI, while the diastolic pressure values were statistically significantly different ($p < 0.05$). Hemoglobin values did not differ significantly, namely 12.06 ± 1.40 and 11.68 ± 1.07 ($p > 0.05$). A meta-analysis conducted to see the relationship between vitamin D supplementation and hemoglobin concentration found no significant association between them.

Conclusions

Hemoglobin levels in urban elderly women who were given Vitamin D supplements were higher than those in the rural group, statistics were not significantly different.

Keywords

Hemoglobin, urban elderly women, rural elderly women, Vitamin D

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Others

Poster ID – P.O-01

ANALYSIS OF GATROSCOPIC AND HISTOPATHOLOGICAL IMAGES OF GASTRITIS DUE TO HELICOBACTER PYLORI INFECTION IN PATIENTS WITH DYSPEPSIA SYNDROME AT YARSI HOSPITAL JAKARTA

Syafruddin A. R. Lelosutan¹, Nunung Ainur Rahmah², Ari Fahrial Syam³

¹ Program Studi Doktor Sains Biomedis Sekolah Pasca Sarjana Universitas YARSI Jakarta, Indonesia, ²Departemen Patologi Anatomi Fakultas Kedokteran Universitas YARSI, Indonesia, Staf Laboratorium Patologi Anatomi RS YARSI Jakarta, Indonesia, ³Departemen Ilmu Penyakit Dalam SubDivisi Gastroenterologi-Hepatologi RSUPN Cipto Mangunkusumo/Fakultas Kedokteran Universitas Indonesia, Dekan Fakultas Kedokteran Universitas Indonesia, Guru Besar Universitas Indonesia Jakarta.

Abstract

Introduction

The prevalence of *Helicobacter pylori* (HP) in Indonesia varies according to regional location and ethnicity. At YARSI Hospital, Jakarta, Dyspepsia Syndrome (DS) has received quite a lot of treatment procedures such as Esophagogastroduodenoscopy (EGD) and gastric mucosal endoscopic biopsies with histopathological results in the form of a number of positive images of HP. Obtain prevalence figures for HP infection as well as the results of analysis of the relationship between bacterial HP infection with the macroscopic diagnosis of EGD and histopathology of endoscopic biopsies results at YARSI Hospital, Jakarta.

Material and Methods

Retrospective study of DS cases that underwent an EGD procedure and at the same time taking endoscopic biopsies specimens for 1 year (2023–2024). Data was selected according to inclusion and exclusion criteria. Statistical processing uses SPSS version 26 and data analysis tests use the Chi-Square test within a significance limit of 0.05 and a confidence interval of 95%.

Results and Discussion

There were 72 subjects who met the requirements according to the results of their endoscopic biopsies histopathology at YARSI Hospital, Jakarta. HP bacteria were found at YARSI Hospital Jakarta with a prevalence of 18.0%. Analysis of the relationship between HP bacterial infection with the macroscopic appearance (EGD) of gastric mucosal tissue (Hyperemic-erythematous gastritis/HG, Erosive gastritis/EG and Ulcerative gastritis/UG) and with histological damage to the gastric mucosa (Active chronic gastritis/ACG and Inactive chronic gastritis/ICG) was carried out using the Chi-square test which gave significantly acceptable results with $p = 0.003$ ($p < 0.05$) and $p = 0.004$ ($p < 0.05$) respectively within the 95% confidence index; and damage to gastric mucosal tissue due to inflammation caused by HP bacterial infection in the form of ACG and ICG amounting to 8.3% and 9.7% of all subjects, respectively.

Conclusion

There is a relationship between HP bacterial infection and the macroscopic diagnosis of EGD and histopathology of endoscopic biopsies results at YARSI Hospital, Jakarta.

Keywords

Gastric mucosal biopsies, Gastritis, *Helicobacter pylori*, Histopathology

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