Under the Auspices of

COLLEGE OF CHEMICAL PATHOLOGISTS OF SRI LANKA

8th ANNUAL ACADEMIC SESSIONS 2023

"Advancing Through Challenges"

14th AND 15th JULY, 2023
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JULY 2023 | ISSUE NO. 8

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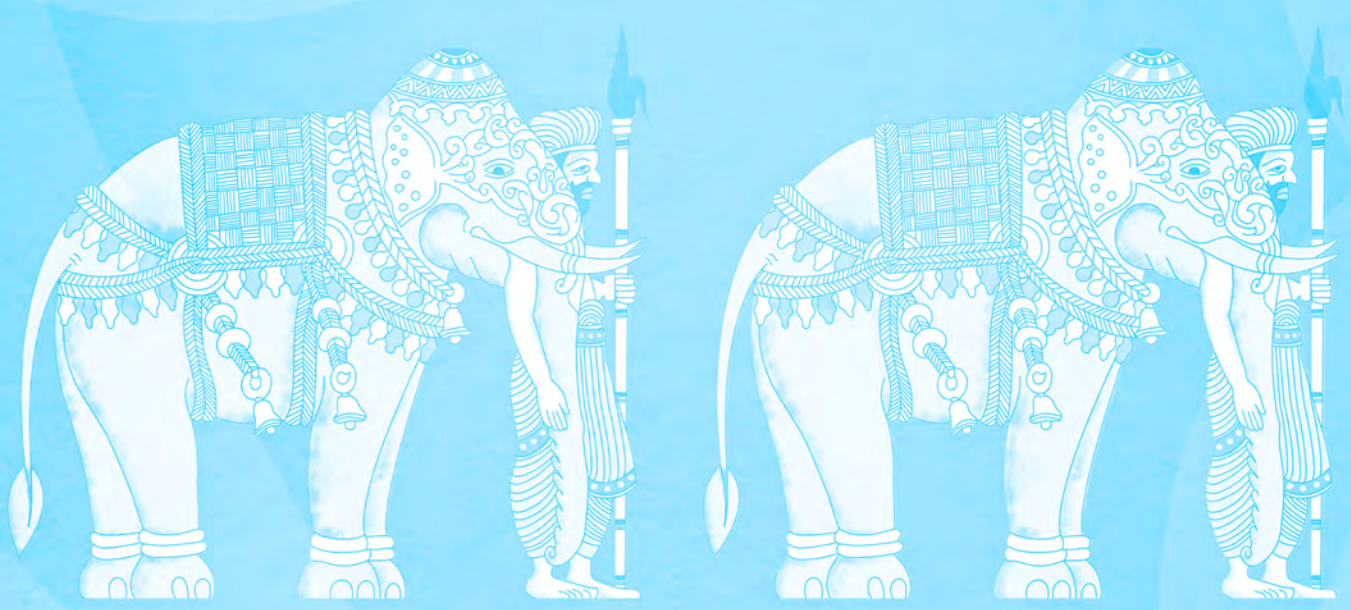
COLLEGE OF CHEMICAL PATHOLOGISTS OF SRI LANKA

8th Annual Academic Sessions 2023

"Advancing Through Challenges"

14th and 15th July, 2023
Kandy, Sri Lanka

CONTENTS



MESSAGES

MESSAGE FROM THE PRESIDENT

Dr Dulani Jayawardana
MBBS, Dip Path, MD (Chemical Pathology)
Consultant Chemical Pathologist
National Hospital Kandy
Sri Lanka

I am pleased to extend to you an invitation to the 8th Annual Academic Sessions of the College of Chemical Pathologists of Sri Lanka (CCPSL AAS 2023), which will be held on July 14th and 15th July, 2023 at the Grand Kandyan Hotel in Kandy, Sri Lanka. This year's theme is "Advancing Through Challenges", and we aim to highlight the crucial role that laboratories play in clinical care and the development of a sustainable quality health system.

CCPSL has always been at the forefront of advancing the field of laboratory medicine in Sri Lanka, and I am committed to ensuring that we continue to lead the way. Under the auspices of International Federation of Clinical Chemistry (IFCC) and Asia Pacific Federation of Clinical Chemistry (APFCB), the two-day annual academic sessions will provide an excellent opportunity for medical and laboratory professionals to update and share their knowledge. We are delighted to have renowned foreign delegates join us and share their insights on novel topics, which will undoubtedly contribute to the advancement of the laboratory field.

Moreover, we will also have a two-day industrial exhibition that will showcase the latest technology for the field of laboratory medicine in Sri Lanka. The exhibition aims to provide a platform for industry experts to interact with medical and laboratory professionals, share their knowledge, and explore new opportunities to advance the field.

On behalf of my council, I invite you to participate in this important event and contribute to the advancement of the laboratory field for quality and sustainable healthcare systems in Sri Lanka. We look forward to your presence and active participation. We must recognize that the laboratory is not just a service provider, but a critical component of a sustainable quality health system. Let us embrace the challenges ahead with confidence and determination, knowing that we have the expertise and dedication to advance our field and improve healthcare for all Sri Lankans.

Dr Dulani Jayawardana
8th President
College of Chemical Pathologists of Sri Lanka

MESSAGE FROM THE CHIEF GUEST

Dr Asela Gunawardena
MBBS, MSc (Med Admin), MCMA, MBA (Common Wealth), Dip BS, MA
Director General of Health Services
Ministry of Health, Nutrition and Indigenous Medicine
Sri Lanka

I am delighted and honoured to be the chief guest of the 8th Annual Academic Sessions of College of Chemical Pathologists of Sri Lanka.

The theme selected this time "Advancing through challenges" is a timely and appropriate one which would open discussions among the laboratory professionals in continuing and improving Chemical Pathology services in our country during this challenging period.

Since the establishment of the college in 2015, it has grown rapidly into a dynamic professional organisation that provides leadership to the Chemical Pathology profession in Sri Lanka by improving the quality of laboratory services and training laboratory professionals. The college performs these multiple tasks through academic sessions addressed by both local and international speakers, organising workshops and case discussions, working in collaboration with the Ministry of Health, developing national guidelines and generating quality laboratory reports amidst the current financial restrictions.

Quality laboratory practice is essential for the provision of optimal healthcare to patients. To achieve this requirement, it is important for the clinical laboratory professionals to meet regularly to share their knowledge and experience and to discuss best laboratory practice to support the health system in our country. Hence, I believe that this session would provide education to the laboratory professionals for their continuing professional development and provide networking opportunities to all in the field.

I appreciate the dedication and commitment of Chemical Pathologists as a group of experts in laboratory medicine who strive in achieving better patient care through quality laboratory services.

While conveying my warm wishes to the president and the council of the College of Chemical Pathologists of Sri Lanka for the success of this event, I also wish to assure continuous support for the present and future endeavours under taken by the college.

Dr Asela Gunawardena
Director General of Health Services
Ministry of Health, Nutrition and Indigenous Medicine

MESSAGE FROM THE PRESIDENT IFCC

Professor Khosrow Adeli
PhD, FCACB, DABCC, FAACC
The President
International Federation of Clinical Chemistry and Laboratory Medicine (IFCC)

On behalf of the IFCC organization, it is my great pleasure to extend a warm welcome to all attendees of the Annual Academic Sessions 2023, hosted by the College of Chemical Pathologists of Sri Lanka. We are thrilled to collaborate with and support this year's conference, which fosters scientific exchange and close interactions among pathologists, laboratory scientists and professionals from the diagnostic industry. By bringing us all together in forums like this, we ensure that our organizations and the field of laboratory medicine remain at the forefront of advancements.

Encouraging a culture of innovation is crucial, as it leads to technological and process advancements in all aspects of clinical laboratory operations. The current decade presents an exciting and rapidly evolving time for the field of clinical laboratory medicine. This timely conference provides an excellent opportunity to explore the opportunities and challenges in Chemical Pathology and laboratory medicine services amidst these dynamic innovations and developments. The realm of in vitro diagnostics and clinical laboratory medicine has experienced significant growth and expansion following the pandemic. There is now a greater recognition of the vital role played by clinical laboratories and laboratory professionals in public health and patient care worldwide. The CCPSL 2023 conference will serve as a platform to showcase the latest developments, scientific breakthroughs and technological innovations in this important field.

I invite colleagues locally, regionally and internationally to participate in the academic sessions, industry exhibits and other offerings of this premier scientific event, taking place in Sri Lanka, July 14-15th, 2023. I extend my heartfelt wishes to the College of Chemical Pathologists of Sri Lanka as they organize a productive and enjoyable conference in the beautiful city of Kandy. May the event be filled with fruitful discussions, valuable insights and memorable experiences for all participants.

Professor Khosrow Adeli
IFCC President

MESSAGE FROM THE PRESIDENT APFCB

Professor Tony Badrick
The President
Asia Pacific Federation for Clinical Biochemistry and Laboratory Medicine (APFCB)

On behalf of the Asia Pacific Federation of Clinical Biochemistry Executive Board and Committees, and the broader membership I would like to express my support for the Annual Academic Sessions of the CCPSL meeting. Organising these meetings is always onerous, but they are an essential component of the ongoing education and professional development of pathologists and scientists. Interacting with each other and discussing common issues is a critical aspect of professionalism and these meetings provide the venue for that to occur. I thank those involved with the preparation and organisation of the meeting, and I hope everyone who attends will be able to take back to their laboratory something that they have learned that will lead to improved patient outcomes.

Professor Tony Badrick
APFCB President

MESSAGE FROM THE DEPUTY DIRECTOR GENERAL (LABORATORY SERVICES)

Dr Sudath K Dharmaratne
MBBS, MSc, MD (Medical Administration), DIPPCA, FMCA
Deputy Director General Laboratory Services
Ministry of Health, Nutrition and Indigenous Medicine
Sri Lanka

I send this message to convey my best wishes to the College of Chemical Pathologists of Sri Lanka on the occasion of the 8th annual academic sessions.

Chemical Pathologists play a pivotal role in both preventive and curative medicine by providing numerous test facilities which are crucial in diagnosis and management of patients. The CCPSL is well known for its commitment to improving the standards of laboratory services in the field of Chemical Pathology in Sri Lanka.

The annual academic session brings the specialists in Chemical Pathology and other specialties together with current trends and concepts. This time CCPSL has chosen "Advancing through challenges" as the theme for their annual academic sessions. I am sure that members of this prestigious college have the courage and capacity to achieve it.

The continuous support extended by the CCPSL to the Ministry of Health through collaborative work on policy making and other important matters is a great strength to uplift the health care services of the county.

College of Chemical Pathologists Sri Lanka remains committed in advancing best practices and demonstrating the value of the laboratory professionals to the entire healthcare system. Let me extend my warmest thanks to the organizers of this important conference.

I congratulate and wish the CCPSL all the success.

Dr Sudath K Dharmaratne
Deputy Director General (Laboratory Services)
Ministry of Health, Nutrition and Indigenous Medicine

MESSAGE FROM THE JOINT SECRETARIES

Dr Dilanthi Warawita
MBBS, Dip Chem Path, MD (Chemical Pathology)
Consultant Chemical Pathologist
Senior Lecturer
Department of Biochemistry and Molecular Biology
Faculty of Medicine
University of Colombo

Dr Maduri Vidanapathirana
MBBS, Dip Chem Path, MD (Chemical Pathology), FRCPath(UK)
Senior Lecturer, Consultant Chemical Pathologist
Department of Pathology
Faculty of Medical Sciences
University of Sri Jayewardenepura

A sustainable quality health system relies heavily on the accurate and timely information provided by laboratory tests which enables healthcare providers to deliver the best possible care to their patients. In light of the current economic crisis in Sri Lanka, medical laboratories have faced numerous obstacles and hurdles. Nevertheless, our primary objective remains to provide the best possible healthcare services to our patients, regardless of these difficulties. Hence we selected the theme "Advancing through Challenges" for our 8th Annual Academic Sessions of the College of Chemical Pathologists of Sri Lanka (CCPSL).

We believe that this conference is an excellent opportunity for us to come together and address these challenges, highlighting new developments and innovations that can help us to overcome them.

As part of the conference, we are also organizing a two-day workshop for medical laboratory technologists and scientists to help address gaps in knowledge and highlight new developments, equipping participants with the necessary skills to overcome the challenges we face in the current healthcare landscape.

While thanking the foreign and local faculty for their invaluable contribution to the various symposia plenaries and lectures, we are hopeful that the 8th Annual Academic Sessions of the CCPSL which is organized for the first time in Kandy would turn out to be a very productive and enjoyable learning experience for all the delegates.

Dr Dilanthi Warawita
Dr Maduri Vidanapathirana
Joint Secretaries
CCPSL

COUNCIL

COLLEGE OF CHEMICAL PATHOLOGISTS OF SRI LANKA, THE COUNCIL 2023

Sitting (Left to right)	: Dr Dilanthi Warawita, Dr Ganga Withana Pathirana, Dr Saroja Siriwardene, Dr Thushara Hewageegana, **Dr Dulani Jayawardena,** Dr Kisali Hirimuthugoda, Dr Thathsarani Vithana Pathirana, Dr Neranjana Vithanage, Dr Maduri Vidanapathirana
Standing – 1st row (Left to right)	: Dr Nangai Kularatnam, Dr Rajitha Samarasinghe, Dr Vithegi Kesavan, Dr B.K.T.P.Dayanath, Dr Manjula Dissanayake, Dr Chandrika Meegama, Dr Gaya Katulanda, Dr Thamara Herath, Dr Eresha Jasinge
Standing – 2nd row (Left to right)	: Dr Thushari Vithanage, Dr Gawri Abeynayake, Dr Sakunthala Jayasinghe, Dr Ushani Jayawardane, Dr Nadeen Senanayake, Dr S.I. Majitha, Dr Deepani Siriwardhana, Dr Dilinika Perera
Absent	: Dr Homathy Sivakumar

COLLEGE OF CHEMICAL PATHOLOGISTS OF SRI LANKA COUNCIL - 2023

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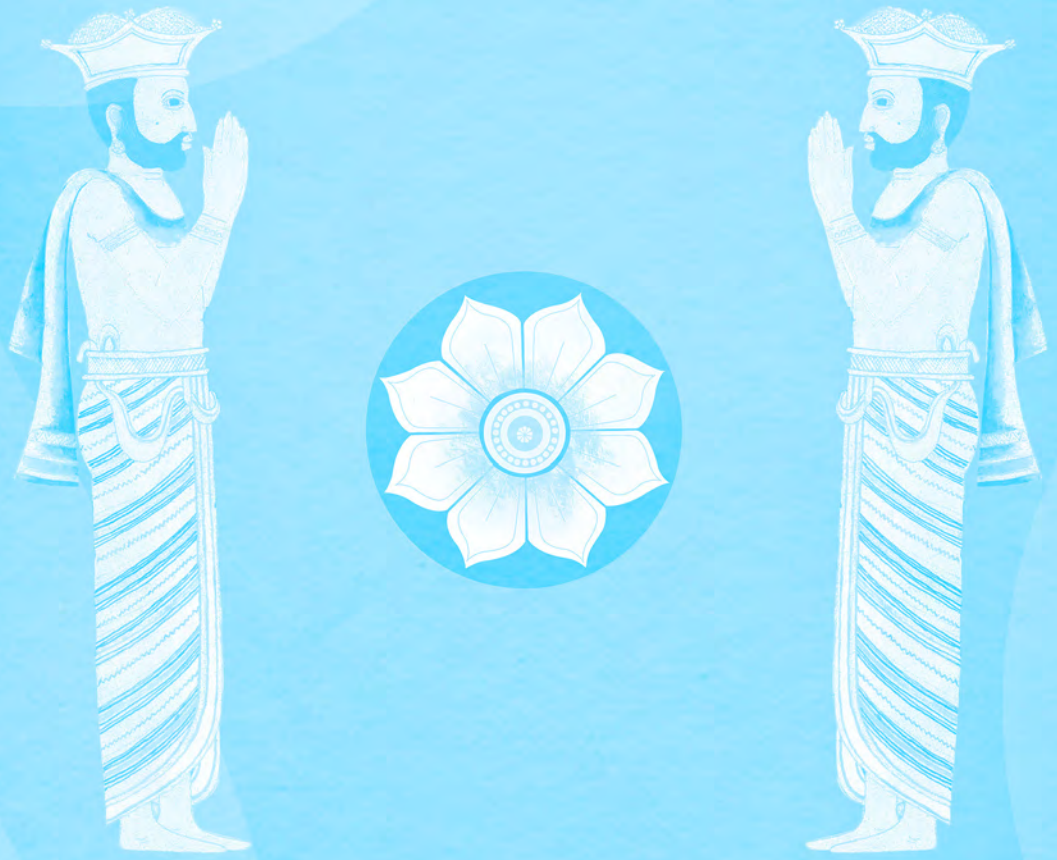
	Dr Rajitha Samarasinghe
	Dr Gaya Katulanda
	Dr B.K.T.P. Dayanath
	Dr Thamara Herath
	Dr Manjula Dissanayake
	Dr S.I. Majitha
	Dr Vithegi Kesavan
	Dr Homathy Sivakumar
	Dr Sakunthala Jayasinghe
	Dr Dilinika Perera
	Dr Ushani Jayawardane
	Dr Nangai Kularatnam
	Dr Thushari Vithanage
	Dr Gawri Abeynayake
	Dr Nadeen Senanayake
Advisor	Dr Saroja Siriwardene



PROGRAMMES

ACADEMIC PROGRAMME

COLLEGE OF CHEMICAL PATHOLOGISTS OF SRI LANKA Annual Academic Sessions 2023 (CCPSL AAS 2023) Academic Programme		
Day 1 : 14th July 2023		
TIME	**TOPIC**	**RESOURCE PERSON**
8.00 – 9.00 am	**Registration**	
9.00 – 9.30 am	**Plenary 1** **Lipoprotein (a) in clinical and laboratory practice**	Prof John Burnett
9.30 – 10.30 am	**Symposium 1: Quality assurance in total testing process**	
9.30 – 10.00 am	Quality control: Current problems and future trends	Prof Tony Badrick
10.00 – 10.30 am	Adding value in the post analytical phase	Dr Samuel Vasikaran
10.30 – 11.00 am	**Tea**	
11.00 – 11.30 am	**Plenary 2** **Autoimmune and neuroimmune markers**	Dr Barnali Das
11.30 – 12.30 pm	**Symposium 2: Investigations in renal diseases**	
11.30 – 12.00 pm	Biochemical challenges of evaluation of patient before and after transplant	Dr Udana Rathnapala
12.00 – 12.30 pm	Providing a service for therapeutic drug monitoring	Prof Graham Jones
12.30 – 1.00 pm	**Plenary 3** **Future models for clinical biochemistry training and development**	Dr Adrian Park
1.00 – 2.00 pm	**Lunch**	
2.00 – 2.30 pm	**Plenary 4** **Provisionally alkaline phosphatase isoenzyme analysis**	Dr Rajeev Srivastava
2.30 – 3.30 pm	**Symposium 3 Inborn errors of metabolism (IEM)**	
2.30 – 3.00 pm	Disorders of amino acid metabolism	Dr Charlotte Dawson
3.00 – 3.30 pm	IEM : Analytical aspects	Dr James Pitt
3.30 pm onwards	**Tea**	
Industrial Exhibition Closes at 5.00 pm		

ACADEMIC PROGRAMME CONTD.

COLLEGE OF CHEMICAL PATHOLOGISTS OF SRI LANKA Annual Academic Sessions 2023 (CCPSL AAS 2023) Academic Programme		
Day 2 : 15th July 2023		
TIME	**TOPIC**	**RESOURCE PERSON**
8.00 – 8.30 am	**Registration**	
8.30 – 9.00 am	**Plenary 5** **FGF-23 and phosphate metabolism**	Dr Samuel Vasikaran
9.00 – 10.00 am	**Symposium 4 Laboratory management**	
9.00 – 9.30 am	Data mining as a laboratory management tool	Prof Ken Sikaris
9.30 – 10.00 am	Analytical performance specifications for clinical laboratory	Prof Tony Badrick
10.00 – 10.30 am	**Plenary 6** **Getting the right answer, metrological traceability in laboratory medicine**	Prof Graham Jones
10.30 – 11.00 am	**Tea**	
11.00 – 11.30 am	**Plenary 7** **Cancer molecular biomarkers: Filtering the music from the noise**	Dr Sanjeeva Gunasekara
11.30 – 12.30 pm	**Symposium 5: Hypertension**	
11.30 – 12.00 pm	Young hypertension-when is it considered secondary?	Dr Suranga Manilgama
12.00 – 12.30 pm	Laboratory role and pitfalls in investigating a young hypertensive	Dr Manjula Dissanayake
12.30 – 1.00 pm	**Plenary 8** **Role of laboratory in clinical care pathway**	Dr Charles Antonypillai
1.00 – 2.00 pm	**Lunch**	
2.00 – 2.30 pm	**Plenary 9** **Closing the evidence gaps in pediatric reference intervals: The CALIPER initiative**	Prof Khosrow Adeli
2.30 – 3.30 pm	**Oral presentations**	
3.30 – 4.00 pm	**Closing ceremony and prize giving**	
4.00 pm onwards	**Tea**	
Industrial Exhibition Closes at 5.00 pm		

MEDICAL LABORATORY SCIENCE PROGRAMME

COLLEGE OF CHEMICAL PATHOLOGISTS OF SRI LANKA Annual Academic Sessions 2023 (CCPSL AAS 2023) Medical Laboratory Science (MLS) Programme		
Day 1 : 14th July 2023		
TIME	**TOPIC**	**RESOURCE PERSON**
8.00 – 8.45 am	**Registration**	
8.45 – 9.00 am	**Inauguration**	
9.00 – 9.30 am	Glucose measurement and clinical utility	Dr Nangai Kularatnam
9.30 –10.00 am	Understanding the clinical importance of measurement uncertainty and analytical goals	Prof Ken Sikaris
10.00 –10.30 am	**Tea**	
10.30 –11.00 am	Biochemical parameters in extremes of age	Dr Dilinika Perera
11.00 –11.30 am	ISO 15189:2022: Challenges and opportunities	Dr Deepani Siriwardhana
11.30 –12.00 pm	Basic laboratory tests in inborn errors of metabolism	Dr Eresha Jasinge
12.00 – 1.00 pm	**Lunch**	
1.00 – 1.30 pm	How to evaluate laboratory performance - Quality indicators	Dr Vithegi Kesavan
1.30 – 2.00 pm	Variations in results of thyroid profile	Dr Gaya Katulanda
2.00 – 2.30 pm	Analysis of body fluids	Dr Nadeen Senanayake
2.30 – 3.00 pm	Use of POCT & related problems	Dr Thushari Vithanage
3.00 pm onwards	**Tea**	
Industrial Exhibition Closes at 5.00 pm		

MEDICAL LABORATORY SCIENCE PROGRAMME CONTD.

COLLEGE OF CHEMICAL PATHOLOGISTS OF SRI LANKA Annual Academic Sessions 2023 (CCPSL AAS 2023) Medical Laboratory Science (MLS) Programme		
Day 2 : 15th July 2023		
TIME	**TOPIC**	**RESOURCE PERSON**
8.30 – 9.00 am	**Registration**	
9.00 – 9.30 am	EQA : Interpretation of results and trouble shooting	Dr Thamara Herath
9.30 – 10.00 am	Practical application of IQC	Dr S I Majitha
10.00 – 10.30 am	**Tea**	
10.30 – 11.00 am	New trends in lipid profile – Essentials to know	Dr Neranjana Vithanage
11.00 – 11.30 am	Urine analysis and its clinical utility	Dr Kisali Hirimutugoda
11.30 – 12.00 pm	Harmonization and standardization of biochemical and immuno assays	Dr Barnali Das
12.00 – 1.00 pm	**Lunch**	
1.00 – 1.30 pm	Risk management in clinical laboratory	Prof Praveen Sharma
1.30 – 2.00 pm	Chemical Pathology quiz	Dr Thurairatnam Inthujah Dr Rajika Jinasena Dr Hiruni Amarasekara Dr Shanika Sandaruwani
2.00 – 2.30 pm	Electrolytes: What you need to know in laboratory practice	Dr Gawri Abeynayake
2.30 – 3.30 pm	Oral presentations	
3.30 – 4.00 pm	Closing ceremony and prize giving	
4.00 pm onwards	**Tea**	
Industrial Exhibition Closes at 5.00 pm		

INAUGURATION PROGRAMME

6.15 pm	Invitees take their seats
6.30 pm	The ceremonial procession
6.35 pm	National anthem
6.40 pm	Lighting of the traditional oil lamp
6.45 pm	Welcome address by **Dr Dilanthi Warawita**, Joint Secretary CCPSL
6.50 pm	Induction of the new President by immediate Past President, **Dr Kisali Hirimutugoda**
6.55 pm	Address by the President CCPSL, **Dr Dulani Jayawardana**
7.10 pm	Address by the Chief Guest, **Dr Asela Gunawardana,** Director General of Health Services
7.20 pm	Award of the medal to Past President
7.25 pm	Award of CCPSL Felicitation
7.30 pm	Award of CCPSL Fellowship
7.40 pm	CCPSL Oration 2023 **"A lifetime of learning"** by **Prof Ken Sikaris**, Consultant Chemical Pathologist, Melbourne Pathology, Australia
8.10 pm	Vote of thanks by **Dr Maduri Vidanapathirana**, Joint Secretary, CCPSL
8.15 pm	Cultural show
8.45 pm	Ceremonial procession leaves the hall
8.50 pm	Reception

AWARDS

FELLOWSHIP AWARD

FELLOWSHIP AWARD

Professor Neelakanthi Vajira Ratnatunga
Consultant Histopathologist
Faculty of Medicine
University of Peradeniya
Sri Lanka

It is our great pleasure and honour to present this Fellowship to Professor Neelakanthi Vajira Illangakoon Ratnatunga, in recognition of her exceptional contributions to the field of Pathology and her dedicated service as a healthcare leader, distinguished academic, and exemplary teacher. Professor Neelkanthi Ratnatunga's remarkable career and unwavering commitment have left an indelible mark on the medical community in Sri Lanka.

Born on July 22nd, 1953, in Kandy, Professor Neelakanthi Ratnatunga embarked on her academic journey at Girls High School, Kandy. She qualified in Medicine from the University of Peradeniya, in 1978, where she excelled in her undergraduate exams, ultimately topping her batch in the finals.

After completing her internship and post-intern appointments, she joined the Department of Pathology at the University of Peradeniya as a lecturer in 1980. In 1982 she commenced her training for the Diploma in Pathology. Equipped with her extensive knowledge and expertise, she became a board-certified specialist in Histopathology in 1992 and was appointed as Professor in Pathology in 2000. Her dedication to her craft was further recognized when she was honored as Professor Emeritus at the University of Peradeniya.

She obtained a Common Wealth Medical Fellowship and was trained in Immunohistochemistry techniques related to renal disease at St. Thomas' Hospital London. She undertook training in muscle histochemistry and immunohistochemistry related to muscle disease at the Dubowitz Neuromuscular Center, Imperial College School of Medicine, Hammersmith Hospital London UK through a senior Commonwealth Fellowship.

The gained knowledge and extensive training paved her to provide selfless service as the renal Pathologist to various Transplant and Dialysis Units, significantly impacting the healthcare of countless patients across Sri Lanka.

In addition to that, she played a pivotal role in introducing and establishing advanced techniques in laboratories in Sri Lanka, including the first referral laboratory for immunohistochemistry, renal histopathology and immunofluorescent laboratory, and muscle histochemistry.

FELLOWSHIP AWARD CONTD.

Her involvement in numerous committees and boards unveil her exceptional leadership abilities and her commitment to improving medical education and research, including Senate representative to the Council University of Peradeniya, 2009-2012, visiting lecturer, faculty of Dental Sciences, University of Peradeniya, Co-editor of Journal of Diagnostic Pathology 2011/2012, member of editorial board of Ceylon Journal of Science and Sri Lanka Journal of Medicine till 2011 and also in 2013, editor in chief of Peradeniya University research sessions 2012 and editor in chief of proceedings of the International Medical Congress of PeMSAA 2017.

She is a member of many professional bodies, including Hon: Secretary College of Pathologists of Sri Lanka, President of the College of Pathologists of Sri Lanka in 2001, President of the Kandy Society of Medicine in 2015 and President of the Peradeniya Medical School Alumni Association 2011/2012. She is a Fellow of the Royal College of Physicians of Edinburgh, a Fellow of the College of Pathologists of Sri Lanka and a Fellow of the Sri Lanka College of Surgeons.

She was appointed as the Head of the Department of Pathology and Director Laboratory at Teaching Hospital Peradeniya at a very young age and had to bear numerous responsibilities in the department with hardly any staff. With all these hurdles, she was able to nurture a significant number of excellent future Pathologists. Besides, she was a Chairperson of the Board of Study of Pathology for two terms from 2004 to 2007 and 2010 to 2011.

Professor Neelakanthi Ratnatunga extended her tremendous support to the field of Chemical Pathology recognizing the value of specialty. She introduced automation in the Chemical Pathology laboratory at Teaching Hospital Peradeniya, ensuring accurate and consistent results. She introduced colour-coded tubes for blood collection and strictly enforced quality controls. Despite not being a trained Chemical Pathologist, she checked all control runs and results daily, encouraging the technical staff to maintain quality control charts and relevant records. She placed great importance on further training of technologists and promoted attendance at training programs, both locally and internationally.

Her commitment to training the next generation of pathologists has had a lasting impact on the field and has contributed to developing highly competent professionals. Of many trainees, seven trainees successfully completed their diplomas and went on to become skilled and accomplished Chemical Pathologists. As the chairperson of the Board of study in Pathology at the post graduate institute of Medicine, Professor Neelakanthi Ratnatunga played a vital role in resolving issues related to trainees in Chemical Pathology with patience and tolerance. Her expertise and leadership were instrumental in addressing challenges and finding practical solutions in this specialized area of Pathology.

Furthermore, Professor Neelakanthi Ratnatunga has made significant contributions to form the Chemical Pathology division at the Faculty of Medicine, University of Peradeniya. Her efforts have paved the way for advancements in diagnostic and therapeutic practices, benefiting countless patients and furthering the progress of medical science in Sri Lanka.

Professor Neelakanthi Ratnatunga's research endeavours, primarily focused on renal pathology, have garnered significant recognition. She has published over 25 papers since 2016 and has been a frequent presenter at national associations, earning numerous prizes and awards for her exceptional

FELLOWSHIP AWARD CONTD.

contributions. Notable among these accolades are the President's Research Bonus in 1999, the National Science Foundation of Sri Lanka Award for her project on IgA nephropathy in Sri Lanka in 2011, and the NRC Merit Award for scientific publications on CKDu in 2014.

In addition to her research and academic pursuits, Professor Neelakanthi Ratnatunga has actively participated as an examiner for undergraduate and postgraduate examinations, ensuring the development of future medical professionals in Sri Lanka. Her commitment to nurturing young minds is further exemplified by her supervision of four MPhil and PhD student research projects in CKDu.

In addition to her professional pursuits, Professor Neelakanthi Ratnatunga has a rich and diverse range of interests and talents. She demonstrated a deep passion for the performing arts from an early age. She received the prestigious Best Actress award for an English drama and actively participated in numerous school dramas, showcasing her love for the stage. Her involvement in the school choir and her proficiency as a piano and organ player in school and church further underscores her passion for music. During her time at High School in Kandy, she served as the Head Prefect and Senior House Captain, exhibiting her leadership skills and commitment to excellence. She was also involved in various activities, such as Girl Guides.

As a teenager, she enjoyed pop music and even won a Beatles competition, surprising her parents, who never expected her to go beyond the Ordinary Levels. Despite these initial doubts, she proved herself academically. She has always had a keen interest in ballroom dancing too.

Her fun-loving and mischievous nature, coupled with her love for music and dancing, have remained integral aspects of her personality, shaping her character and contributing to her achievements. While her aspirations may have initially leaned toward a career in the film industry, fate led her to become a medical professor instead. Nevertheless, her vibrant and multifaceted qualities have continued to guide her throughout her career, infusing her work with creativity, passion, and a zest for life.

In addition to her professional pursuits and being a busy surgeon's wife, Professor Neelakanthi Ratnatunga finds joy and fulfilment in her role as a mother of two children. Their elder son is a Consultant Surgeon in the Department of Surgery at the Faculty of Medicine, University of Peradeniya, and the younger daughter holds the esteemed position of Professor of Immunology at the Department of Microbiology, Faculty of Medicine, University of Peradeniya. She says she is surprised they turned out OK because she thought she was a full-time academic and a part-time mother! Behind every remarkable individual is a strong support system, and Professor Neelakanthi Ratnatunga's achievements are no exception. Her husband has been an unwavering pillar of support throughout her illustrious career, enabling her to make remarkable contributions to the field of Pathology. I believe she is currently enjoying a serene and heartwarming time with her grandchildren, remembering her childhood and also feeling a sense of nostalgia. Therefore, it is with great pleasure and profound appreciation that we confer upon Professor Neelakanthi Vajira Illangakoon Ratnatunga this Fellowship. Her remarkable accomplishments, selfless dedication, and significant contributions to the field of Pathology in Sri Lanka make her a true inspiration to all aspiring Pathologists. We extend our heartfelt congratulations and wish her continued success in all her future endeavours.

ORATION

CCPSL ORATION 2023

Professor Kenneth (Ken) Sikaris
Consultant Chemical Pathologist
Melbourne Pathology
Australia

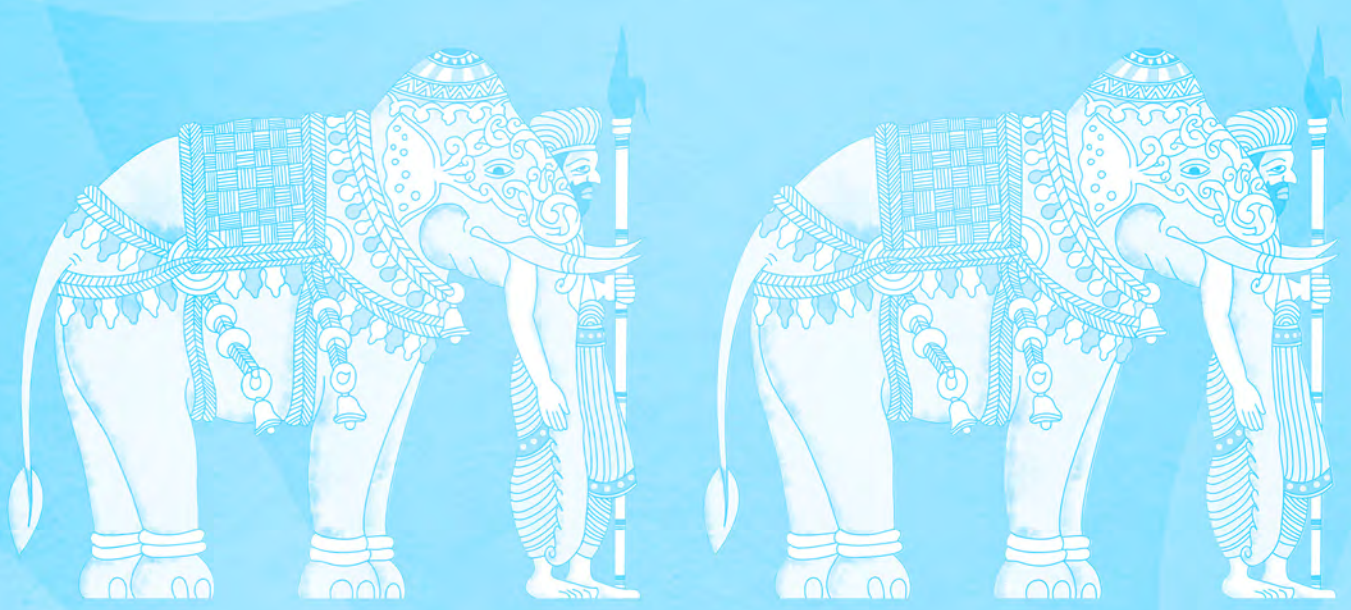
Ken has always been a good student and his secondary school performance allowed him to attend Melbourne University, although not in the ways he imagined. He completed his honors degree in science and rather than continuing with a PhD scholarship, he then transferred and completed a medical degree.

His interest in scientific enquiry inevitably lead him back to the laboratory and he completed his Chemical Pathology training and worked as a Chemical Pathologist for over thirty years.

Despite working predominantly for private Pathology companies, he has continued to teach and conduct research having hundreds of peer reviewed publications and conference abstracts. He has held countless expert committee roles at both national and international level and he is proud, like many of our lecturers at this meeting, to be listed as an associate editor of the Tietz textbook, the "Bible" for serious students of Clinical Chemistry.

Although notorious for his interest in PSA, lipids, analytical quality and reference intervals, his publications show investigations into almost all aspects of Chemical Pathology. Now transitioning to retirement, colleagues cannot fathom how such an intense career could draw to a close, but perhaps they do not appreciate the underlying challenge that has motivated his lifelong interests.

FACULTIES



INTERNATIONAL GUEST FACULTY

INTERNATIONAL GUEST FACULTY

Professor John Burnett

MB ChB, MD Otago, PhD Western, FRCPA, FFSc, FACB, FCSANZ, FAHA
Consultant Chemical Pathologist
Department of Clinical Biochemistry, PathWest Laboratory Medicine
Royal Perth Hospital and Fiona Stanley Hospital Network
School of Medicine, University of Western Australia
Australia

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Professor Tony Badrick is Currently the CEO of the RCPAQAP. Adjunct Professor School of Pharmacy and Pharmacology, Griffith University, Honorary Associate Professor, National Centre for Epidemiology and Public Health ANU College of Health and Medicine and ANU College of Science, Honorary Associate Professor, Faculty of Medicine, Bond University, Gold Coast, Visiting Fellow, Australian Institute for Health Innovation, Macquarie University. He was President of the Australasian Association of Clinical Biochemists (2003-2007) and Vice President of the Australian Institute of Medical Scientists (2011-2018), is the President of the Asian Pacific Federation of Clinical Biochemistry.

INTERNATIONAL GUEST FACULTY CONTD.

Dr Samuel Vasikaran
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Dr Sam Vasikaran is a medical graduate of the University of Colombo and Fellow of the Royal College of Pathologists of Australasia. He is a Chemical Pathologist at PathWest and has been based at Royal Perth Hospital for the last thirty years.

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Executive Member, Scientific Division, International Federation of Clinical Chemistry & Laboratory Medicine (IFCC), 2018 – 2023.Chair, American Association of Clinical Chemistry (AACC), Indian Section.

National Accreditation Board for Testing & Calibration Laboratories (NABL) Assessor & College of American Pathologist Inspector.

Adjunct Faculty, Kasturba Medical College, MAHE, Manipal. Editorial Board of many Journals & Reviews Editor, Practical Laboratory Medicine, Elsevier. Recipient of three oration awards and seven international and eight national awards.

INTERNATIONAL GUEST FACULTY CONTD.

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Professor Jones is a Chemical Pathologist at SydPath, St Vincent's Hospital, Sydney overseeing the chemical pathology laboratory services and Conjoint Professor, University of NSW.

He is active in professional activities at national and international levels in the fields of traceability of laboratory results (member of JCTLM executive), external quality assurance (advisor for RCPAQAP), testing for kidney disease (Australia and IFCC), Analytical performance Specifications (EFLM and Australia) diagnosis of diabetes, units for therapeutic drug monitoring, common reference intervals, and harmonisation of laboratory reporting (Australia). He is active in teaching and research with over 100 peer reviewed papers.

Dr Adrian Park
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Dr Park trained in Chemical Pathology (Metabolic Medicine) at Imperial College, London and has been a Consultant at Addenbrooke's Hospital, Cambridge, since 2008. Dr Park is actively involved in Chemical Pathology Training in the UK and is currently Chair of the Chemical Pathology Steering Committee at the Royal College of Pathologists, UK. Dr. Park is the Chair of IFCC Taskforce on Global eLearning/ academy.

INTERNATIONAL GUEST FACULTY CONTD.

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Dr Rajeev Srivastava is a Consultant Chemical Pathologist at the Queen Elizabeth University Hospital, Glasgow, UK. He graduated from Delhi University and did higher specialist training in Clinical Biochemistry at Ninewells Hospital in Dundee. He has authored various peer-reviewed publications in leading medical journals. He is co-author of the best-selling textbook *Clinical Biochemistry: An Illustrated Colour Text,* which has been translated into many languages, *Case Studies in Clinical Biochemistry* and the chapter on Metabolic Diseases in *Hutchison's Paediatrics.* His special interests lie in osteoporosis and bone metabolism, diagnosis of inborn errors of metabolism and parenteral nutrition.

Dr Charlotte Dawson
Consultant in Chemical Pathology / Metabolic Medicine
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United Kingdom

Since 2012 **Dr Charlotte** has been a Consultant in Chemical Pathology / Metabolic Medicine at University Hospitals Birmingham UK, specializing in adult inherited metabolic disorders (IMD) and lipid disorders. The Adult IMD service in Birmingham cares for more than 1200 patients in the Midlands and South West England. Charlotte and colleagues run over 30 clinical trials including 'first in man' gene and mRNA therapy trials. She is an honorary senior lecturer at the University of Birmingham and a senior examiner for the FRCPath diploma, Royal College of Pathologists UK.

INTERNATIONAL GUEST FACULTY CONTD.

Dr James Pitt
PhD, FHGSA, FFSc (RCPA)
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Royal Children's Hospital, Melbourne
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Dr James is a Biochemical Geneticist and head of the Metabolic and Newborn Screening Laboratories at Victorian Clinical Genetics Services. These laboratories provide a unique service for the diagnosis and monitoring of inborn errors of metabolism, servicing a state population of 6.6 million.

Professor Khosrow Adeli
IFCC (International Federation of Clinical Chemistry)
and the Hospital for Sick Children/University of Toronto
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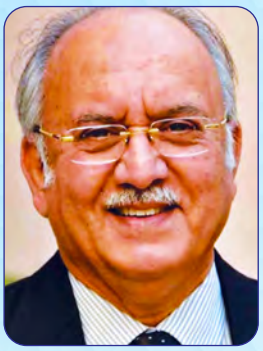
Professor Adeli is a senior scientist and academic clinical biochemist with over 30 years of experience in clinical chemistry service, education, and research. Numerous leadership positions in the field of clinical biochemistry and laboratory medicine over the past three decades. Established track record in both basic and clinically oriented research in the fields of metabolic health and disease as well as clinical biochemistry and pediatric laboratory medicine. Published over 600 articles and abstracts with an h-index of 78 and >33,400 citations. Academic achievements have been recognized nationally and internationally through several prestigious research awards. Elected as the new president of IFCC (International Federation of Clinical Chemistry and Laboratory Medicine) (2020-2023), a worldwide organization with >120 member societies and >50,000 laboratory physicians and scientists around the world.

INTERNATIONAL GUEST FACULTY CONTD.

Professor Kenneth (Ken) Sikaris
Consultant Chemical Pathologist
Melbourne Pathology
Australia

Professor Ken Sikaris received his science and medical degrees at Melbourne University before continuing to train as a Chemical Pathologist at several university Teaching Hospitals. His career as a consultant Chemical Pathologist has been predominantly in the private Pathology sector and he has worked at Melbourne Pathology for the last 20 years. With specific interests in quality assurance and interpretation, especially reference limits and decision limits, he has served on numerous national and international committees. Having given hundreds of lectures, as an associate editor of the Tietz textbook and as an examiner for the RCPA and AACB in Australia, Ken has been heavily involved in teaching during his long career.



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Professor Sharma is the Chair, IFCC-Committee on Clinical Laboratory Management (CCLM), Secretary, Asia Pacific federation for Clinical Biochemistry (APFCB), Director, South East Asia on WASPaLM Board, Editor-in-Chief, Indian Journal of Clinical Biochemistry (IJCB), Scientific Consultant to Snibe, Director, FOUNDATION FOR QUALITY (INDIA) for Rajasthan State, Director, National Referral Centre for Lead Projects India (NRCLPI), Jodhpur.

President, Indian Society of Lead Awareness and Research, Assessor (NABL), International Lead Assessor (AERSSC), President ACBI (2003-2004 and 2014-2015), Chairman, APFCB Congress and Conferences (2019 to 2022), Chairman, APFCB Communication committee (2010-2019), Chief Editor, APFCB News (2010-2019).

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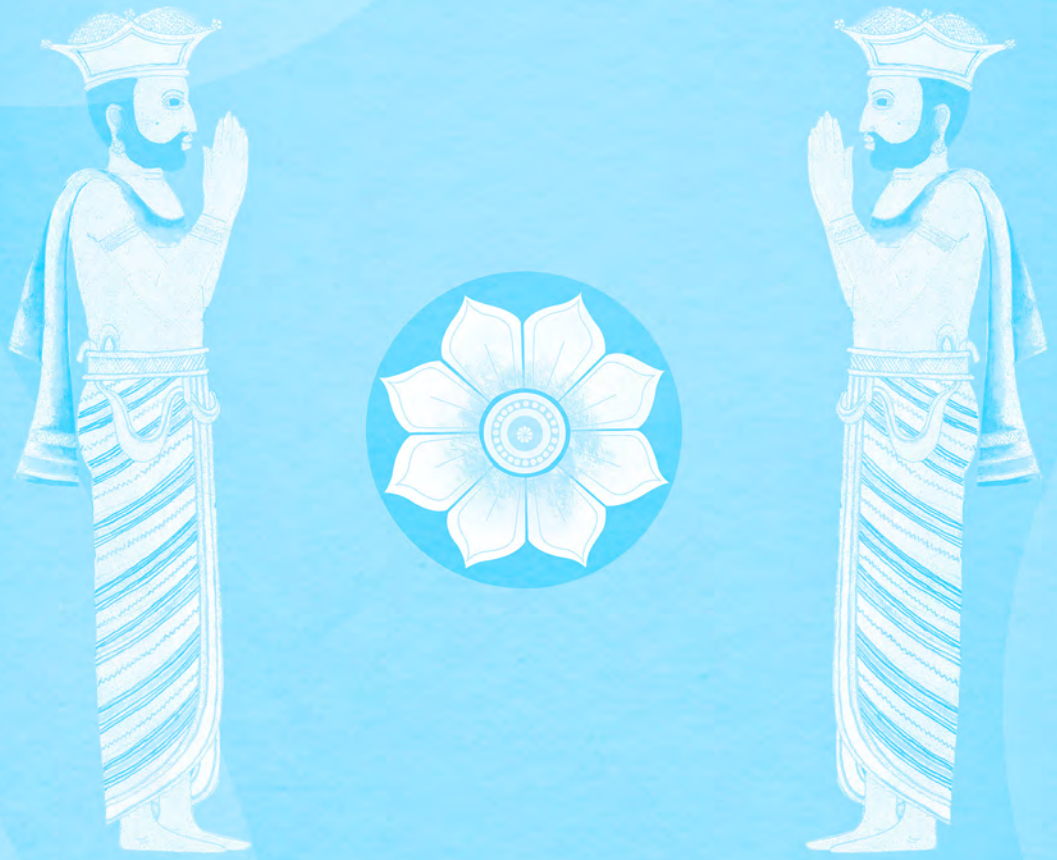
ABSTRACTS



SPEAKER ABSTRACTS

DAY 1

Lipoprotein (a) in Clinical and Laboratory Practice

Prof John Burnett

Lipoprotein (a) [Lp(a)] is a highly heterogeneous, cholesterol ester-rich, apoB-containing lipoprotein of unknown physiological function, formed by the binding of LDL to the plasminogen homologue apo(a). Genetic, epidemiologic and Mendelian randomization studies have established that Lp(a) is an independent and causal risk factor for atherosclerotic cardiovascular disease (ASCVD) and calcific aortic valve disease, through mechanisms associated with increased atherogenesis, inflammation, and thrombosis. However, at this juncture, we lack a) standardized and harmonized assays to measure Lp(a), b) universal guidelines for diagnosing and providing risk assessment, and 3) targeted treatments to lower Lp(a). Circulating Lp(a) concentrations are largely determined by genetic factors, but are also influenced by ethnicity, hormones, kidney function, and acute inflammatory events.

Plasma Lp(a) concentrations should be measured using an assay independent of apo(a)-isoform size with appropriate calibrators and the results reported in molar units. International guidelines recognize elevated Lp(a) as a risk enhancing factor for risk reclassification. Targeted screening strategies of high-risk patients are recommended, whereas universal population screening is not advised.

Testing for elevated Lp(a) is recommended in all patients with premature ASCVD and those at intermediate-to-high risk of ASCVD. Elevated Lp(a) should be used to assess and stratify risk and to guide decision-making on initiation or intensification of preventative treatments, such as cholesterol lowering therapy. Lipoprotein apheresis should be considered in patients with progressive ASCVD. There are currently no approved specific therapies that target elevated Lp(a), although limited clinical outcome data using PCSK9 inhibitors is consistent with the beneficial effect of such reduction..

Quality Control: Current Problems and Future Trends

Prof Tony Badrick

The talk will describe the development of quality control in clinical chemistry and focus on the fundamental assumptions made in creating the models that are routinely used. The basis of a QC strategy is an understanding of the errors that can arise, the material used to identify an error has occurred, an algorithm to flag when one of these errors is detected, and a process to follow to correct the error and amend any compromised patient results.

Generally there have been two main strategies used in QC over the last forty years, each differing in the material used as the sample. Conventional QC uses a sample that substitutes for a patient, whereas patient-based QC uses patient results directly. Both systems have advantages and disadvantages. In the talk we will explore these differences to obtain a better understanding of quality control

DAY 1

Adding Value in the Post Analytical Phase

Dr Samuel Vasikaran

The accurate analysis of blood and other body fluids and timely reporting of those results to the requesting clinicians form the foundation on which the clinical biochemistry laboratory service is based. However, the laboratory profession also has a crucial role in facilitating appropriate and optimal application of those results in patient management in order to ensure the best outcomes for the patient. The laboratory should ensure life-threatening (critical value) results are communicated promptly to the appropriate clinician; hence, appropriate mechanisms should be in place for this to be done every time in a timely manner. On the other hand, for significant-risk results that are not time critical, such as first time-raised tumour markers, what is crucial is ensuring that 100% of these results are seen by the appropriate caregiver. Patient specific reference intervals and, where appropriate, diagnostic cut-offs, decision limits or treatment targets should accompany all reported results.

The leading causes of breakdown in the diagnostic process are the failure to order appropriate diagnostic tests or their incorrect interpretation. The addition of personalised and high-quality interpretative comments to laboratory results are welcomed by clinicians and is useful in improving patient outcomes. This activity should form an important part of the training of Pathologists and Clinical Scientists as well as the subject of continuing professional development and quality assurance.

Autoimmune and Neuroimmune Markers

Dr Barnali Das

Autoimmune diseases are a family of chronic, and often disabling, illnesses that develop when underlying defects in the immune system lead the body to attack its own organs, tissues, and cells. They often endure debilitating symptoms, loss of organ function, reduced productivity at work, and high medical expenses. This talk focuses on the detection of autoimmune markers in patients with different diseases and finding the clinical significance of the particular disease and the autoimmune markers found in them during clinical diagnosis.

Autoimmune encephalitis (AE) encompasses a group of immune-mediated heterogeneous disorders of the central nervous system associated with neuronal autoantibodies. In clinical neurology there is a consensus among experts that early identification of AE can lead to an effective immunotherapy response and thus a better prognosis and management of the disease for which understanding its prevalence and characteristics is crucial.

The epilepsy, encephalopathies and movement disorders are associated with autoantibodies against neuronal proteins (LGI1, CASPR2 and VGKC-complex antibody, NMDA receptor antibody); antibody against Aquaporin-4/NMO-IgG, anti-MOG antibodies and paraneoplastic profile auto antibodies (anti Hu, CRMP5, Yo, Ri, Ma, CV2 and ampiphysin).

DAY 1

Autoimmune and Neuroimmune Markers Contd.

The method used here is indirect immunfluorecence assay which is considered a gold standard for detection of autoimmune markers. The test detects the presence of ANA in the blood of the patient which adheres to reagent test cells (substrate), forming distinct fluorescence patterns that are associated with certain autoimmune diseases. Other markers detected by immunfluorecence assay and immunoblotare ANCA, dsDNA and ASMA, LGI1/ CASPR2/ VGKC-Complex antibody, NMDA receptor antibody, antibody against Aquaporin-4/ NMO-IgG and paraneoplastic profile auto antibodies (anti Hu, CRMP5, Yo, Ri, Ma, CV2 and Ampiphysin).

In the case series, we have compared the characteristics of seropositive and seronegative cases of autoimmune encephalitis. Inferential analysis of seropositive and seronegative groups revealed statistically significant differences pertaining to cognitive disturbances (p=0.04), speech disorders (p=0.049) and tumor association (p=0.029) while rest of the characteristics were nearly the same. The study also discusses some illustrative clinical vignettes and highlights a case of concurrent anti-NMDAR encephalitis along with anti-MOG antibodies.

Biochemical Challenges of Evaluation of Patient Before and After Transplant

Dr Udana Rathnapala

Clinical biochemistry remains an inseparable and essential component of organ transplantation. Its role in pre and post-transplant evaluation and management of any organ transplantation remains crucial.

Therapeutic drug monitoring (TDM) for immunosuppressive drugs is an essential, but at the same time insufficient tool due to low predictability of drug exposition and marked pharmacokinetic variability caused by different factors, including genetic polymorphism of metabolizing enzymes and drug transporters. Tacrolimus, a calcineurin inhibitor, is a mandatory medicine in many organ transplants yet has a narrow therapeutic window making drug level monitoring fundamental.

Further, utilization of biomarkers to assess organ dysfunction remains controversial. Serum creatinine, the most commonly used biomarker to measure renal dysfunction, depends on age, gender, muscle mass, muscle metabolism, co-administered drugs and hydration status. Also, serum creatinine concentrations may not change until a significant amount of kidney function has already been lost. Although serum cystatin C is regarded as more sensitive in evaluating renal dysfunction, its role in transplantation is not well established. Other novel biomarkers also have not come into routine clinical practice.

The sudden shift of individual biochemical profile upon organ transplantation remains a challenge. The detection of change and appropriate maneuvering of the serum potassium, magnesium and calcium is critical in the immediate post kidney transplant period as the patient becomes polyuric immediately upon transplantation.

Hence, clinical biochemistry should be an integral part of a multidisciplinary team handling organ transplantation throughout its course.

DAY 1

Providing a Service for Therapeutic Drug Monitoring

Prof Graham Jones

For some medications measurement of the amount of drug in the blood is a key component of medical care, known as therapeutic drug monitoring (TMD). This is recommended for drugs which show wide variation in drug absorption and metabolism, and have small differences between effective and toxic concentrations. TDM has a number of factors which are different to usual laboratory testing and attention needs to be paid to all phases of the testing. In general TDM is applied to drugs where there is a guideline indicating the need, and providing specific advice.

In the pre-analytical phase key factors include knowing the dose administered and the time given, the required sample timing (e.g. peak, trough, other) and the actual timing of the sample collection. Generally it is necessary to ensure steady state conditions for appropriate testing, i.e more than 5 half-lives of the drug. Some drugs are bound by gel separators or are unstable and, so choice of collection tube and sample handling can be important. Drug assays need to provide accurate results across the measurement range, but especially near the clinical decision points. Immunoassay kits are available for many common drugs, but HPLC or mass spectrometry can be used for almost any drug with the correct instrumentation and technical expertise. Interpretation is based on a therapeutic interval from the lowest limit that will achieve a clinical response up to a higher limit which may indicate a risk of toxicity or lack of further benefit at higher concentrations. These limits come from clinical studies and, unlike usual reference intervals, cannot be developed or validated locally. Reporting needs to be done using units that match interpretive information. Errors in any of the components listed above may lead to the wrong interpretation and incorrect dosing of the patient.

Future Models for Clinical Biochemistry Training and Development

Dr Adrian Park

Traditional training and development of Chemical Pathology/Clinical Biochemistry trainees has involved significant face-to-face components, which have been difficult to provide to all trainees who need this. Technological advances enable us to teach, train and develop trainees in new ways which complement existing techniques. In this presentation, I will discuss this further, highlighting our local training experience, as well as discussing the development of UK and international teaching resources.

DAY 1

Provisionally Alkaline Phosphatase Isoenzyme Analysis

Dr Rajeev Srivastava

Alkaline phosphatase is an enzyme which is found in many tissues. It catalyses hydrolysis of phosphate esters at alkaline pH, though its exact clinical function is still not clear. In the blood it mainly originates from liver or bone, but is also present in intestine, kidney, leucocytes as well as the placenta. Post-translational modification leads to formation of different isoenzymes, which differ in their chemical and electrophoretic properties. Different glycosylation produces tissue-specific isoforms e.g. liver, bone. Identification of the isoenzymes helps to determine the tissue source of alkaline phosphatase elevations in serum and therefore the potential pathology.

Disorders of Amino Acid Metabolism

Dr Charlotte Dawson

Disorders of amino acid metabolism are caused by pathogenic DNA variants affecting the activity of an enzyme, co-factor or transport protein required for the metabolism of an amino acid. The gene defect leads to accumulation of unmetabolised amino acids which may itself be toxic or be converted to toxic by-products such as ammonia or organic acids. The brain, liver and kidneys are the most commonly affected organs. Some disorders present acutely when a patient is in a catabolic state causing release of large amounts of endogenous amino acids from muscle. Some conditions cause progressive neurological damage without acute decompensation.

Treatment of amino acid disorders usually involves a low protein diet with specific restriction of the precursor amino acid, supplementation of the other essential amino acids and detoxification medication if indicated.

The major groups of amino acid disorders are the urea cycle disorders, organic acidurias, disorders of branched chain amino acid metabolism, disorders of sulphated amino acid metabolism and disorders of phenylalanine and tyrosine metabolism. As it is not possible to cover all of these in detail I will focus on the urea cycle disorders.

Urea cycle disorders (UCD) are the commonest inherited metabolic disorders and the X-linked condition, ornithine transcarbamylase deficiency (OTC), is the commonest UCD. They are caused by a metabolic defect in one of the urea cycle enzymes required for detoxification of the nitrogenous waste products of amino acid metabolism. They usually present acutely with neurological features of hyperammonaemia including confusion, seizures, lethargy and behavioural disturbance. UCD may be overlooked as the cause of the patient's symptoms and measurement of plasma ammonia should therefore be considered in patients of any age presenting with unexplained encephalopathy. Treatment involves protein restriction and ammonia-scavenging medication. Newer treatments in clinical trials include gene replacement therapy and mRNA-based technologies.

DAY 1

Inborn Errors of Metabolism (IEM): Analytical Aspects

Dr James Pitt

Provision of a comprehensive laboratory service for the diagnosis of inborn errors of metabolism (IEMs) is a significant undertaking given the complexity of the human metabolome and the hundreds of IEMs that are known. It is impractical for a single laboratory to provide comprehensive quantitation of all the relevant biomarkers but a wide range of IEMs can be diagnosed or suspected with a small number of panel-based and screening tests covering a sub-set of biomarkers with quantitative and qualitative reporting. My presentation will focus on plasma amino acids, urine organic acids and urine metabolite screening as tests that can cover a wide range of IEMs, including those that present acutely. Mass spectrometry is a key technology for these tests as it provides the specificity and sensitivity to detect low levels of biomarkers in complex biological matrices. Tandem mass spectrometry has been used to greatly expand the scope of urine metabolic screening to include IEMs affecting purine and pyrimidine, oligosaccharide, bile acid and carnitine metabolism. Attention to pre-analytical specimen errors is important as incorrect sample collection or handling can lead to spurious and misleading results. The rarity of many IEMs is also a challenge to maintaining interpretive expertise which underlines the importance of participation in EQA and diagnostic proficiency programs.

DAY 2

FGF-23 and Phosphate Metabolism

Dr Samuel Vasikaran

Phosphate plays important roles in nucleotide synthesis, cell signalling, energy metabolism, and mineralization of bone in humans. Phosphate metabolism is controlled by parathyroid hormone (PTH), 1,25 dihydroxyvitamin D, and fibroblast growth factor 23 (FGF-23) through their actions on the gastrointestinal tract, the kidneys and bone. FGF-23 which is synthesised mainly by osteocytes and osteoblasts in bone, inhibits 1,25 dihydroxyvitamin D synthesis in the kidneys leading to a reduction in intestinal phosphate absorption. FGF-23 also has a direct phosphaturic action on proximal renal tubule together with PTH, to cause hypophosphataemia.

Chronic hypophosphataemia causes impaired mineralization of bone leading to development of rickets in children and osteomalacia in adults. In muscle, hypophosphataemia leads to tissue hypoxia resulting in proximal myopathy.

The commonest genetic FGF-23 related disorder is X-linked hypophosphatemia (XLH, previously known as vitamin D resistant rickets), caused by a mutation in the PHEX gene leading to increased FGF-23 in blood resulting in renal phosphate wasting. In the rare syndrome of tumour-induced osteomalacia (TIO), tumoral secretion of FGF-23 results in chronic hypophosphataemia and osteomalacia. Measurement of FGF-23 in blood may be useful in identifying FGF-23 mediated hypophosphataemic disorders.

These conditions are conventionally treated with oral phosphate and vitamin D analogues. More recently, the development of therapy with the anti-FGF-23 monoclonal-antibody burosumab has led to the healing of rickets in children and osteomalacia in adults with XLH and similar genetic disorders as well as in patients with TIO.

DAY 2

Data Mining as a Laboratory Management Tool

Prof Ken Sikaris

In the past, we relied on the teachings of experienced masters (e.g. Hippocrates) to instruct the practice of medicine. In the 20th century, there was a flourishing of medical literature and opinions so we developed an evidence based scientific approach to defining the practice of medicine, or at the very least, relied on a consensus group of experts to summarise the evidence to date for us. We also appreciate the concept of personalized medicine and the idea that what is good for the majority may not be optimal for an individual. But how can we understand everyone?

Our IT systems, Laboratory Information Systems (LIS) and Electronic Health Records (HER) are accumulating the data on "Everyone" so in theory we know what happens to everyone. As far as our laboratory interests are concerned we can use the data to explore the entire patient pathway through the laboratory from request, to sample preparation and transport, analysis, reporting and the clinical actions that are triggered by our reports. The hardest part is getting access to the data, followed by knowing exactly the question to ask. In comparison, the data analysis by spreadsheet or modern statistical packages is relatively easy for anyone with an interest in exploring what exactly is happening to our patients when they have laboratory tests. While there are ethical challenges in the use of patient data, it is also a quality assurance obligation that we ensure that opportunities for improving quality are freely available.

Analytical Performance Specifications for Clinical Laboratory

Prof Tony Badrick

Analytical Performance Specifications (APS) are used in many areas of laboratory medicine, including EQA interpretation and method evaluation. In this presentation, we will discover the basis of the APS, which are goals for performance based on acceptable imprecision and accuracy. The Milan hierarchy has given us three models of determining the evidence for those goals: patient outcomes, biological variance and expert opinion. We will look at how these APS are selected and used in EQA schemes and method evaluation.

DAY 2

Getting the Right Answer, Metrological Traceability in Laboratory Medicine

Prof Graham Jones

In the laboratory we aim to provide accurate results to allow best care of our patients. The starting point for this is to consider how laboratory tests are interpreted. They are all interpreted by comparison. This may be with a population reference interval, a clinical decision point, or a previous result from the same patient. These comparisons are only valid provided that the data from the routine laboratory is unbiased relative to the source of the results used for the comparison. Then we need to understand how we assign values for patient samples.

For all numerical results this is also by comparison. This is done by comparing the signal from the instrument with the signal from a material with a known value, i.e. the assay calibrator. The calibrator itself has a value assigned by measurement and comparison with another calibrator. This sequence is known as the calibration hierarchy or traceability chain, and leads up to a pure material or reference method which defines the "True value" for a test. This process involves reference material producers, manufacturers and routine laboratories and provides the metrological traceability of our results. Variations which occur throughout the complete process are described as the assay measurement uncertainty.

Materials or methods at the top of traceability chains may be defined by the JCTLM, or obtained from other sources, e.g. the WHO. Manufacturers must select appropriate reference materials and transfer the values with minimum uncertainty. Laboratories should select traceable methods and perform assays with good laboratory procedures.

External quality assurance is required to assess between laboratory and between method differences. In order to make good clinical decisions, we need all data, including different routine laboratories and research organisations to use methods which are metrologically traceable, i.e. they give the right answer.

DAY 2

Cancer Molecular Biomarkers: Filtering the Music from the Noise

Dr Sanjeeva Gunasekara

A biomarker can be defined as a measurable biological entity which is produced by a physiological or pathological process. These biomarkers can be chemical agents. Identification of such a biomarker can be useful in the cancer setting to diagnose, prognosticate, and detect recurrence of various cancers. These biomarkers can be found in blood, urine, soft tissue etc. They can be molecular, physiological, histological or radiological.

With the recent development of many diagnostic tools there has been an explosion of biomarkers proposed to use in cancer care. However, evidence of their clinical correlation is often lacking. Therefore, there is an urgent need to identify real practice changing biomarkers early and roll them out at bedside as soon as possible. This has got an added importance as Sri Lanka battles a crippling economic crisis with an urgent need to identify where we have to prioritize the very limited resources and procuring medicines and medical equipment.

In this presentation, I will be discussing the background of use of biomarkers, the evolution of biomarkers in cancer care, as well as the selection of biomarkers for clinical use in Sri Lanka considering the epidemiological profile of cancer patients of Sri Lanka, the impact on patient survival or improvement of quality of life, the future direction of biomarker researches in cancer care etc.

Young Hypertension-When is it Considered Secondary?

Dr Suranga Manilgama

The age cut-off defining the young hypertension varies in guidelines as <50, <40, or <30 years. The presence of hypertension at a young age increases the risk of cardiovascular events in middle age. Young people are exposed to the longer duration of high blood pressure on organs such as the brain and kidneys and it may produce irreversible changes. It contributes to an earlier onset of coronary heart disease, heart failure, stroke, and transient ischemic attacks. Though, secondary causes are identified more frequently in younger patients, the young hypertension is not always due to a secondary cause.

Predictors of a secondary cause for hypertension include young age (<30 years) with no other risk factors, drug resistant hypertension, severe hypertension (>180/110 mm Hg), sudden deterioration in blood pressure control, non-dripping status on ambulatory blood pressure monitoring, or the presence of hypertension-mediated organ damage. Investigation in young adults is advocated across most of the guidelines for the identification of secondary causes of hypertension and evidence of organ damage.

It is important and beneficial to recognize secondary causes of hypertension, as this might direct specific treatment strategies, achieve better blood pressure control and potentially cure high blood pressure in young patients. This beneficial impact needs to be balanced against the risks such as cost, patient burden, and incidental diagnoses from imaging investigations. The management of young-onset hypertension is the same as that of older individuals.

DAY 2

Laboratory Role and Pitfalls in Investigating a Young Hypertensive

Dr Manjula Dissanayake

The medical laboratory plays a major role in finding secondary causes of hypertension and evidence of organ damage. The most common causes of secondary hypertension among young adults are hypothyroidism (1.9%) renovascular disease (1.7%), renal insufficiency (1.5%), primary hyperaldosteronism (1.2%), cushing syndrome (0.5%) and pheochromocytoma (<0.3%). Organ damage is mainly seen in the brain and kidneys.

Most of these secondary causes are endocrine. There are other rare endocrine causes of secondary hypertension for example hyperthyroidism, hyperparathyroidism, acromegaly, insulin resistance and CAH-17-α hydroxylase deficiency. The laboratory investigations and pitfalls related to most common endocrine causes and routine investigations performed to identify organ damage are discussed in detail during the presentation.

Role of Laboratory in Clinical Care Pathway

Dr Charles Antonypillai

Modern-day medicine is anchored in Evidence-Based Practice (EBP). Clinical Care Pathways (CPWs) are tools that translate this evidence into a structured process, taking into account patients' culture and the healthcare institution's environment. They detail the sequence and timing of steps in the CPW, standardizing healthcare, minimizing variation, improving coordination across specialties, reducing wastage, and facilitating process evaluation and improvement. CPWs have been the norm in many European countries for decades, and introducing them into Sri Lanka's healthcare system would bring immense benefits.

Laboratories play a pivotal role in healthcare. Integrating laboratories into CPWs would improve disease diagnosis, minimize wastage, streamline healthcare, and reduce pre-analytical and post-analytical errors. Enhanced communication between laboratories and clinicians would also lead to faster investigation turnaround times and improved quality of care.

Developing CPWs is a challenging task, as healthcare professionals may resist change and may be unaware of the benefits of these pathways. The lack of data infrastructure is another issue. However, these challenges can be overcome through proper education and training of stakeholders and investments in information technology. Chemical Pathologists can play an active role in the development of CCPWs in Sri Lanka, as cost-effective healthcare is a pressing need for the country.

DAY 2

Closing the Evidence Gaps in Pediatric Reference Intervals: The CALIPER Initiative

Prof Khosrow Adeli

Clinical laboratory reference ranges serve as health-associated benchmarks that enable clinicians to interpret laboratory test results and facilitate clinical decision-making. Unfortunately, critical gaps currently exist in accurate and up-to-date pediatric reference ranges for accurate interpretation of laboratory tests performed in children and adolescents, which may contribute to erroneous diagnosis or misdiagnosis of many diseases. Several initiatives have been established internationally to address these gaps, including the KiGGS initiative in Germany, the Aussie Normals in Australia, the AACC- National Children Study in the USA, the NORICHILD Initiative in Scandinavia, and the Canadian Laboratory Initiative on Pediatric Reference Intervals (CALIPER) program in Canada (www.caliperproject.ca).

Since 2009, CALIPER has recruited more than 12,500 healthy children and adolescents, thereby establishing a comprehensive database of pediatric reference ranges for over 200 biomarkers of health and disease (www.caliperdatabase.org). However, evidence gaps continue to exist for special markers and new laboratory instruments.

To address these gaps, our team has recently completed or is currently undertaking studies to establish pediatric reference ranges for: 1) chemistry and immunological markers on new analytical systems, 2) hematological markers on multiple platforms, 3) critical care markers on point of care testing platforms, 4) markers of inflammatory disease (cytokines, calprotectin, autoimmune), 5) markers of nutritional deficiency (essential trace elements), and 6) markers of environmental toxicity (heavy metals). CALIPER is also embarking on specific sub-studies regarding maternal, child, and adolescent health, such as the Mother and Child Health Initiative, COVID-19 Seroprevalence Study, and Lipid Metabolism in Adolescents with obesity. In this presentation, I will review the recent worldwide initiatives on pediatric reference ranges as well as discuss the concept and feasibility of common reference ranges. I will also discuss the progress made by the CALIPER program, the CALIPER database, and future research directions.

ABSTRACTS OF ORAL PRESENTATIONS

RESEARCH AND AUDITS

ABSTRACTS OF ORAL PRESENTATIONS

RESEARCH AND AUDITS

OP - RP 01 Assessment of the Correlation of Haemoglobin A1c and Serum Fructosamine with Mean Blood Glucose Level in Diabetic Patients with Chronic Kidney Disease in a Tertiary Care Hospital of Sri Lanka

OP - RP 02 The Association between Inflammatory Markers and Outcomes among Covid-19 Patients Admitted to Intensive Care Units at a Tertiary Care Hospital in Sri Lanka

OP - RP 03 Relationship between Serum Total Prostate Specific Antigen and Sonographic Findings of the Prostate in a Group of Men with Benign Prostatic Hyperplasia

OP - AR 01 An Evaluation of the Internal Quality Control Performance of the General Biochemistry Analytes in a Laboratory of a Tertiary Care Hospital of Sri Lanka

OP - AR 02 Biggest Challenge in Sweat Testing; Quantity not Sufficient Samples - Audit on Sweat Collection Procedure in Sweat Chloride Testing in Lady Ridgeway Hospital for Children

OP - AR 03 Efficacy and Effectiveness of Adrenal Venous Sampling in the Management of Primary Aldosteronism; Single Centered Cohort Study at a Tertiary Care Hospital in Sri Lanka

ABSTRACTS OF ORAL PRESENTATIONS

RESEARCH AND AUDITS CONTD.

OP - RP 01

Assessment of the Correlation of Haemoglobin A1c and Serum Fructosamine with Mean Blood Glucose Level in Diabetic Patients with Chronic Kidney Disease in a Tertiary Care Hospital of Sri Lanka

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Introduction and objectives

An impaired glycaemic control of patients with chronic kidney disease (CKD) results in disease progression to end stage kidney disease. Therefore, it is essential to use a better indicator for assessing glycaemic control of diabetic patients with CKD. Although HbA_{1c} and fructosamine are indirect markers of assessing glycaemic control, these are affected to a certain extent by effects of CKD itself or due to treatment modalities apart from the glycaemic status. Therefore, the aim of this study is to assess the correlation of HbA_{1c} and fructosamine with mean post prandial blood glucose (PPBS) level to determine its utility as markers of glycaemic control in diabetic patients with CKD.

Methods

A descriptive cross sectional study was conducted prospectively among diabetic CKD patients attending the nephrology clinic. The samples for PPBS were collected on 4^{th}, 8^{th}, 10^{th} and 12^{th} week to calculate mean PPBS and the analysis of haemoglobin, albumin, total protein, creatinine, fructosamine and HbA_{1c} was performed at the end of 12^{th} week. The results were statistically analyzed by using Pearson correlation.

Results

Out of 137 patients, 67% were male and 33% were female. The majority was in CKD stage IV (34.3%). A positive, strong and statistically significant correlation was observed between HbA1c and mean PPBS ($r = 0.77$) while a statistically significant and moderate strength relationship was noted with fructosamine ($r = 0.63$). Haemoglobin level showed no correlation with both HbA1c and fructosamine ($r < 0.1$).

Conclusions

This study shows that both HbA_{1c} and fructosamine have a good positive correlation with mean PPBS while HbA1c has a stronger relationship compared to fructosamine. It emphasizes that fructosamine still has a place in assessing the short term glycaemic control while HbA_{1c} is the better marker for monitoring the long term control in diabetic patients with CKD.

Keywords

Chronic kidney disease, Diabetes mellitus, HbA_{1c}, Fructosamine

ABSTRACTS OF ORAL PRESENTATIONS

RESEARCH AND AUDITS CONTD.

OP - RP 02

The Association between Inflammatory Markers and Outcomes among Covid-19 Patients Admitted to Intensive Care Units at a Tertiary Care Hospital in Sri Lanka

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Introduction and objectives

Covid-19 became a global pandemic that affected millions of people worldwide. Most of the patients were critically ill when admitted to Intensive Care. SARS-CoV-2 induces an extensive inflammatory response leading to critical illness. Thus, it is required to identify the inflammatory markers that could be used as predictors of patient outcomes. The main objective was to study the association between inflammatory markers and outcomes of patients admitted to the ICU with Covid-19 pneumonia.

Methods

A retrospective cohort study was conducted among patients with confirmed laboratory diagnosis of Covid-19, admitted to COVID ICUs of University Hospital, KDU from June to December 2021. Demographics, peak values of inflammatory markers (serum ferritin, lactate dehydrogenase enzyme, C-reactive protein), sepsis indicators (procalcitonin, leukocyte count, absolute neutrophil count and neutrophil-lymphocyte ratio) and patient outcomes (deceased or survived) were collected. Data were analyzed using descriptive statistics. The chi-square test and Mann-Whitney U tests were used to compare variables.

The correlation between inflammatory markers and sepsis indicators was assessed using Pearson correlation. Bivariate logistic regression was used to assess the predictive value of inflammatory biomarkers.

Results

Data from 219 patients (119-deceased, 100-discharged) were included after excluding 95. Peak levels of inflammatory markers, CRP [n=119; 233.83 mg/L, (±121.60)] and LDH [n= 102; 938.45 IU/L, (±459.71)] were higher among the deceased group and had statistically proven association with poor patient outcomes ($p<0.05$). Peak levels of CRP and LDH showed statistically significant positive correlations with sepsis indicators where CRP was correlated with all sepsis indicators at 0.01 level. Bivariate logistic regression indicated CRP as a predictor of poor patient outcomes (OR 1.005; CI 95% 1.001-1.009; $p=0.009$).

Conclusions

There was a significant association between inflammatory markers and severity and mortality in Covid-19 patients. Peak levels of CRP and LDH were predictive of sepsis. CRP was predictive of disease prognosis and poor patient outcomes in Covid-19.

Keywords

Covid-19, Inflammatory markers, Patient outcome, Sepsis

ABSTRACTS OF ORAL PRESENTATIONS

RESEARCH AND AUDITS CONTD.

OP - RP 03

Relationship between Serum Total Prostate Specific Antigen and Sonographic Findings of the Prostate in a Group of Men with Benign Prostatic Hyperplasia

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Introduction and objectives

Benign prostatic hyperplasia (BPH) is an enlargement of the prostate gland which causes lower urinary tract obstructive symptoms in men. Patients with BPH show increased volume of the prostate. Several studies have reported that there are increased serum total Prostate-specific antigen (tPSA) levels in BPH. Serum tPSA is commonly used for the monitoring and early detection of prostate cancer. But the relationship between serum tPSA and prostate volume in BPH is less studied. The most accurate method for measuring prostate volume is trans rectal ultrasonography (TRUS). But it is not feasible to perform in all patients.

The objective of this study was to observe the relationship between serum tPSA and prostate volume in a group of men with BPH.

Methods

This was a cross-sectional study conducted during November 2022 to January 2023 in genitourinary clinic, teaching hospital, Karapitiya. 92 patients with BPH were recruited for the study. Required data were obtained from their clinical records. Serum tPSA, prostate volume and age were statistically analyzed by using Pearson correlation analysis.

Results

The correlation coefficient of serum tPSA and prostate volume in overall patients with BPH is 0.695 ($p < 0.001$). The correlation coefficient of age and serum total PSA level is 0.22 ($p < 0.05$) and age and volume of the prostate is 0.195 ($p > 0.05$). The serum t-PSA level ranges from 0.15 to 8.90 ng/mL and the prostate volume ranges from 20-105 cc. The majority of the men had prostate volume less than 30 cc and serum tPSA value in the range of 0-2.50 ng/mL.

Conclusions

There is a statistically significant relationship between serum tPSA level and volume of the prostate in men with BPH. Serum t-PSA level may be considered a valuable estimate of prostate volume in BPH.

Keywords

Serum total prostate specific antigen, Prostate volume, Benign prostate hyperplasia

ABSTRACTS OF ORAL PRESENTATIONS

RESEARCH AND AUDITS CONTD.

OP - AR 01

An Evaluation of the Internal Quality Control Performance of the General Biochemistry Analytes in a Laboratory of a Tertiary Care Hospital of Sri Lanka

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Introduction and objectives

Internal quality control procedure (IQC) of a laboratory assesses the performance of the analytical phase and its evaluating parameters are mean, standard deviation (SD), coefficient of variation (CV%), bias%, total analytical error (TE%) and sigma metric (α). The common practice is to use the manufacturer assigned mean and SD for a third party IQC material for the analyzer to assess the IQC performance. Therefore, this study was aimed to assess whether the IQC performance parameters are within the internationally recommended standards (Westgard, EFLM Biological Variation Database).

Methods

Two levels of IQC material (L1 and L2) for eighteen general biochemical analytes were selected to assess the cumulative mean, SD, CV%, TE% and σ of the past one month duration. External quality control results of the same month were used to calculate the bias%. The calculated CV%, bias%, TE% and σ were compared with international standards. A σ value of ≥3 was considered as acceptable.

Results

When considering both levels, all the performance parameters were acceptable in 6 analytes (L1) and in 5 analytes (L2) out of 18. In 6 analytes (L1) and 7 analytes (L2), although the CV% and bias% were acceptable, either the TE % or σ or both were unacceptable. In one analyte (L2), only the CV% and σ were acceptable. In the rest of the 6 analytes (L1) and 5 analytes (L2), only the CV% or bias% was acceptable.

Conclusions

Routine IQC performance evaluations are focused only on CV% and bias%. This study shows that even though the CV% and bias% are acceptable, the TE% and σ can still be unacceptable. Therefore, it is essential to estimate the TE% and σ whenever IQC performance is monitored to avoid false reassurance. In such situations, CV% and bias% should be further reduced until TE% and σ are acceptable.

Keywords

Internal quality control, Coefficient of variation, Bias, Total error, Sigma metric

ABSTRACTS OF ORAL PRESENTATIONS

RESEARCH AND AUDITS CONTD.

OP - AR 02

Biggest Challenge in Sweat Testing; Quantity not Sufficient Samples - Audit on Sweat Collection Procedure in Sweat Chloride Testing in Lady Ridgeway Hospital for Children

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Introduction and objectives

Sweat chloride testing is considered as the gold standard for diagnosis of cystic fibrosis and thus laboratory performance on sweat testing is an important and crucial factor for diagnosis. The first step of sweat testing involves pilocarpine iontophoresis to stimulate sweat glands followed by collection of sweat into a macroduct coil. The sweat volume of <15µL is considered as quantity not sufficient (QNS) and cannot be analyzed and needs retesting which impacts medical, psychological and economical aspects of patient management. The objective of the audit is to assess the sweat collection procedure to minimize QNS samples.

Methods

The assessment criteria were based on "Guidelines for the performance of the sweat test for the investigation of Cystic Fibrosis in the UK (2nd Version) - March 2014. Audit was conducted by using the checklist prepared with reference to above criteria and data were collected by periodic direct observation and interviewing the laboratory personnel. The QNS% was calculated for the audit period of 2021/01/01 to 2021/12/31. The re audit was conducted after quality improvement for the period of 2022/01/01 to 2022/12/31.

Results

QNS (overall) was 36% [<3/12 age group-6.6% (<10%) and >3/12 age group 30% (<5%)] for the year 2021 and QNS (overall) was 18% [<3/12 age group-2.2% (<10%) and >3/12 age group 13.95(<5%)] for the year 2022.

Conclusions

Some deviations from standards were identified in the field of staff training, use of sweat inducer and patient preparation during audit period and appropriate actions were initiated in selection and preparation step and staff training. The re audit showed improvement QNS% after above quality improvements. As the QNS rate is still not up to the standards, further improvement is needed to optimize the sweat collection process.

Keywords

Cystic fibrosis, Sweat test, Pilocarpine iontophoresis, QNS

ABSTRACTS OF ORAL PRESENTATIONS

RESEARCH AND AUDITS CONTD.

OP - AR 03

Efficacy and Effectiveness of Adrenal Venous Sampling in the Management of Primary Aldosteronism; Single Centered Cohort Study at a Tertiary Care Hospital in Sri Lanka

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Introduction and objectives

Adrenal venous sampling (AVS) is the gold standard to discriminate unilateral primary aldosteronism (UPA) from bilateral disease (BPA). AVS is technically demanding and only performed in a limited number of centers in Sri Lanka. This study aimed to evaluate the effectiveness of AVS in the management of primary aldosteronism.

Methods

A total of 32 patients who underwent AVS at National Hospital of Sri Lanka from April 2021 to April 2023 were enrolled. Demographic, clinical and laboratory data were obtained retrospectively. A procedure was considered successful when adequate cannulation of both adrenal veins was demonstrated. Cortisol gradient across adrenal vein (AV) and peripheral vein was used to establish success of venous cannulation. Lateralization was determined by the aldosterone gradient between two sides. Continuous and categorical variables were summarized with mean, SD and proportions respectively. Mean and standard deviation of contralateral suppression index (CSI) were estimated with an intercept-only Bayesian inference model.

Results

Of the 32 patients, the average age was 52.47 + 26.14 and 19 (59.4%) were males. Both AVs were successfully cannulated in 12 (37.5%). Among them, lateralization was demonstrated in 11 (91.7%) and one was diagnosed as bilateral disease. There were no total failures. Right AV cannulation was unsuccessful in 18 (56.25%), of which lateralization was demonstrated in 9 (50%) and others were inconclusive. Left AV cannulation was unsuccessful only in 2 (6.25%); one was lateralized and other remained inconclusive. Estimated mean of the CSI was 0.33 (89% credible interval 0.11-0.86). Seven patients underwent unilateral adrenalectomy and had demonstrated significant improvement in blood pressure during follow up. Two patients await surgery. Others were treated medically.

Conclusions

Despite failure due to procedural difficulties, AVS remained useful in the management of patients with PA. Moreover, success of the procedure needs experienced hands and advanced equipment to achieve optimal outcome in PA.

Keywords

Adrenal venous sampling, Lateralization, Contralateral suppression index

ABSTRACTS OF ORAL PRESENTATIONS

CASE REPORTS AND CASE SERIES

ABSTRACTS OF ORAL PRESENTATIONS

CASE REPORTS AND CASE SERIES

OP - CR 01 Shedding Light on Parathyroid Adenoma in Pregnancy: The Success of Parathyroid Hormone Assay in Fine Needle Aspirate: A Case Report

OP - CR 02 Adenine Phosphoribosyl Transferase (APRT) Deficiency Presenting as a Rare Inherited Etiology of Nephrolithiasis: First Sri Lankan Case Report

OP - CR 03 Risk of Ignoring Chronic Diarrhoea: A Patient with Carcinoid Tumour

OP - CR 04 First Case Report of Testosterone Assay-negative Interference in a Female with Virilization

OP - CR 05 Importance of Calculating Measurement Uncertainty in Intra-operative Parathyroid Sampling

OP - CR 06 An Unresolved Case of Hypercortisolism; A Diagnostic Dilemma

ABSTRACTS OF ORAL PRESENTATIONS

CASE REPORTS AND CASE SERIES CONTD.

OP - CR 01

Shedding Light on Parathyroid Adenoma in Pregnancy: The Success of Parathyroid Hormone Assay in Fine Needle Aspirate: A Case Report

Galmangodage NE[1], Pradeepa BGD[1], Samarakoon SMPP[1], Premadasa MGTW[2], Sujeeva N[1], Balasooriya BMCM[1], Katulanda GW[1]

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Introduction

Gestational primary hyperparathyroidism (PHPT) can be difficult to diagnose. Preoperative localization of parathyroid adenoma (PA) with imaging can be challenging, once the biochemical diagnosis is made. Therefore, the measurement of parathyroid hormone (PTH) in parathyroid fine needle aspiration (FNA) washout under ultrasound (USS) guidance from neck lesions is helpful.

Case Presentation

A 33-year-old woman at the 25th week of pregnancy was admitted due to increased vomiting. A small goiter was detected.

Investigations revealed hypercalcemia with ionized calcium of 2.10 mmol/L and hypophosphatemia with serum phosphate of 1.6 mg/dL. Her intact PTH was high at 525 pg/mL, vitamin D was deficient at 16 ng/mL with high urinary calcium excretion of 354 mg/24 hours.

USS of the neck revealed a well-defined hypoechoic lesion below the right lobe of the thyroid, suggestive of a PA.

Fine needle aspiration of the adenoma under USS guidance was done to obtain an aspirate which was washed out with 1 mL of normal saline. The PTH of the washout was 1879 pg/mL.

A diagnosis of PA was made.

Total thyroidectomy with parathyroid gland exploration was done. Intraoperative PTH monitoring was performed with levels dropping from 395.4 to 49.1 pg/mL at 10 minutes. Her serum calcium returned to normal at 2.41 mmol/L on postoperative day 3.

Discussion and Conclusions

In PHPT, preoperative localization of PA is important to decide on surgery. Sestamibi and computed tomography use ionizing radiation carrying risk to the fetus. Neck ultrasound is safe but sensitivity is reduced in patients with goiter. USS guided FNA of neck lesions enables sonographic, cytological, and biochemical results to be combined to localize parathyroid tissue with greater certainty. Intraoperative PTH monitoring is used to monitor the successful removal of the lesion in Sri Lanka.

Keywords

Primary hyperparathyroidism, Parathyroid adenoma, Pregnancy, Parathyroid hormone wash-out

ABSTRACTS OF ORAL PRESENTATIONS

CASE REPORTS AND CASE SERIES CONTD.

OP - CR 02

Adenine Phosphoribosyl Transferase (APRT) Deficiency Presenting as a Rare Inherited Etiology of Nephrolithiasis: First Sri Lankan Case Report

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Introduction

Adenine phosphoribosyl transferase (APRT) deficiency is an autosomal recessive disorder of adenine metabolism& a rare inherited cause of renal stones disease leading to chronic kidney disease if left untreated. Deficient APRT enzyme activity results in the accumulation of adenine, which is then converted to 2,8-dihydroxyadenine (DHA) by xanthine oxidase. The resulting urinary hyper excretion of highly insoluble and nephrotoxic derivative, the DHA can manifests as recurrent/multiple radiolucent nephrolithiasis or crystalline nephropathy.

Case Presentation

Herein, we describe a 20-year-old male with recurrent colicky abdominal pain since childhood& came to medical attention following spontaneous passage of renal stone. His basic urine and serum investigations showed no abnormalities. Ultrasound scanning of the abdomen revealed small contracted kidney on right side with radiolucent calculus on vesico-ureteric junction. The stone was analyzed by Fourier Transform Infrared Spectroscopy after surgical removal and the composition demonstrated as 100% 2,8-dihydroxyadenine(DHA). The dried urine spot analysis also revealed high concentration of urinary adenine [0.111 mmol/L (<0.001)] and 2,8 DHA [0.181 mmol/L (<0.001)] suggestive of APRT deficiency.

The whole exome sequencing revealed a homozygous variant of uncertain significance in the APRT gene [c.270G>T p. ((=))] confirming the genetic diagnosis of APRT deficiency. The patient was successfully managed with xanthine oxidase inhibitor, allopurinol therapy, dietary restriction of adenine rich foods& good hydration. The treatment response was assessed with renal functions, USS-KUB, urinary adenine, 2,8-DHA and oxipurinol level during follow up visits & showed good compliance & response to allopurinol therapy.

Discussion and Conclusions

Radiolucent nephrolithiasis is the commonest presentation of APRT deficiency which needs to be differentiated from uric acid & xanthine stones. This case illustrates the importance of renal stone analysis which leads to an early diagnosis, timely initiation of treatment & prevention of the long-term complications associated with APRT deficiency such as progression to chronic kidney disease.

Keywords

APRT deficiency, Urolithiasis, Dihydroxyadenine

ABSTRACTS OF ORAL PRESENTATIONS

CASE REPORTS AND CASE SERIES CONTD.

OP - CR 03

Risk of Ignoring Chronic Diarrhoea; A Patient with Carcinoid Tumour

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Introduction

Chronic diarrhea is very common symptom and can be due to variety of causes such as infection, inflammation, neuropathy, hyperthyroidism, irritable bowel syndrome and neuroendocrine tumors. Colorectal neuroendocrine carcinoma (NEC) is a rare tumor that demonstrates aggressive growth pattern with ingrowth into the tract, metastasis to the other organs, and invasion to the surrounding organs.

Case Presentation

A 38-years-old male presented with nausea, loose stools and dizziness for 4 years. He had watery diarrhea which was not associated with meals. He had undergone upper and lower gastrointestinal endoscopy multiple times without localizing significant pathology. Ultrasound scan abdomen and pelvis was performed and was insignificant other than a right sided renal calculus. Thus, he was managed symptomatically each episode. Through four years his symptoms worsened despite negative general biochemistry and hematological investigation results except for mildly elevated liver enzymes. Following tumor markers was done and results were insignificant. CEA-2.17 ng/mL (<2.5), AFP 1.35 ng/mL (<10), CA 19.9 - 7.6 U/mL (0-37). A CT scan abdomen was performed which revealed multiple focal lesions in liver suspicious of tumor deposits. Patient underwent liver biopsy and histology confirmed a carcinoid tumor. Chromogranin level was performed later and was markedly elevated > 9000 µg/L (<100 µg/L). 5-HIAA level was 11.6 mg/24hr. (2-6 mg/24hr). As he had liver metastasis he was classified as stage 4 with poor prognosis. Patient was started on octreotide and followed up with gradually decreasing chromogranin levels.

Discussion and conclusions

Though endoscopy is the best screening method for gastrointestinal carcinoma, carcinoid tumor could be missed due to their submucosal location, multicentricity and indolent growth pattern. Being aggressive tumors, they will easily metastasize. Therefore, in patients with persistent diarrhea it is always better to do a serum chromogranin level or 24-hour urine for 5-HIAA to rule out neuroendocrine tumors as prompt diagnosis will improve prognosis.

Keywords

Chronic diarrhoea, Carcinoid tumour

ABSTRACTS OF ORAL PRESENTATIONS

CASE REPORTS AND CASE SERIES CONTD.

OP - CR 04

First Case Report of Testosterone Assay-negative Interference in a Female with Virilization

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Introduction

Testosterone immunoassays are not free from interference. Thus, it may cause diagnostic dilemma during interpretation of test results. Here, we report a case of a girl who was investigated for severe hirsutism with discordant testosterone levels due to such interference.

Case Presentation

A 17-year-old girl presented with a history of rapidly worsening hirsutism, virilization, and amenorrhea. Examination revealed BMI of 27 kg/m^2 with severe hirsutism and features of virilization. Her baseline serum total testosterone level (chemiluminescent microparticle immunoassay, CMIA via Abbott architect i2000 analyzer) was 456 ng/dL (14-76) with normal gonadotrophins and DHEA-S levels. CECT abdomen revealed normal adrenal glands with solid lesions in bilateral ovaries. Bilateral adrenal and ovarian venous sampling (AVOS) attempted with successful cannulation with lateralization into right ovary. However, it was found to have normal peripheral serum total testosterone of 56.14 ng/dL during AVOS which was analysed via CMIA Advia Centaur XP analyzer. Immunoassay interference with Advia was suspected. Testosterone assay is prone to interference from androgenic compounds. Reanalysis of the original serum sample using Vitros Testosterone II assay, a higher affinity assay, revealed a total testosterone level of 460 ng/dL which was compatible with Abbott architect i2000 confirming the negative interference in Advia. Laparoscopic right oophorectomy and left ovarian cystectomy were done. Intraoperative testosterone monitoring with cannulation of bilateral ovarian veins further confirmed the neoplastic source in right ovary. Histology revealed a sertoli cell tumor of the right ovary and benign serous cystadenoma of left ovary.

Her hirsutism was significantly improved, menstruation resumed. Serum testosterone was normal at 3 months post-operative visit.

Discussion and conclusions

This report illustrates a rare case of falsely low testosterone levels due to assay interference most probably due to androgenic compounds cosecreting from tumor. Close collaboration between clinicians and the laboratory is necessary to avoid unnecessary clinical investigations as well as inappropriate treatments.

Keywords

Interference, Testosterone

ABSTRACTS OF ORAL PRESENTATIONS

CASE REPORTS AND CASE SERIES CONTD.

OP - CR 05

Importance of Calculating Measurement Uncertainty in Intra-operative Parathyroid Sampling: A Case Report

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Introduction

Measurement uncertainty (MU) is the expression of the statistical dispersion of the values attributed to a measured quantity. MU helps to identify the significance of the difference between two values. It can be mathematically calculated using either standard deviation (SD) or coefficient of variation (CV).

Surgical removal of the culprit parathyroid gland is the curative treatment for primary hyperparathyroidism and the successful of removal it can be accessed by intra-operative parathyroid hormone (PTH) monitoring as it has a short half-life and turnaround time.

Case Presentation

A 61-year-old female was incidentally found to have high total calcium (11 mg/dL), low phosphate (2.5 mg/dL), high PTH (134.9 pg/mL) and low vitamin D (18 ng/mL). Her serum magnesium, ALP and renal function test were normal. With treatment, her vitamin D level became normal but still, she had persistently high calcium level. As Tc99m SESTAMIBI scan revealed a parathyroid adenoma on the left superior lobe, selective left superior parathyroidectomy was planned. During the surgery, the baseline PTH was 91.7 pg/mL, 10 minutes after selective parathyroidectomy it dropped to 89.0pg/mL and again it rose to 94.7 pg/mL. By calculating the MU, the fluctuation in the PTH measurement concluded as due to the analytical uncertainty.

Discussion and Conclusions

MU helps determine whether the difference between two results is negligible due to the uncertainty in the analysis or is it significant due to the changes in patient's condition. Assuming that the preanalytical factors were constant during the sampling, at 95% of confidence interval the MU was 69.28 – 114.12 pg/mL. Though there is fluctuation in PTH values, as it is within the MU range, it indicates that the culprit gland was not successfully removed.

This case shows the importance of determining the MU to check the clinical significance in the fluctuation of analytical measurement.

Keywords

Measurement uncertainty, Standard deviation, Coefficient of variation, Intra operative parathyroid hormone monitoring

ABSTRACTS OF ORAL PRESENTATIONS

CASE REPORTS AND CASE SERIES CONTD.

OP - CR 06

A Case of Unresolved Hypercortisolism; A Diagnostic Dilemma

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Introduction

Physical and psychological stressors can derange the hypothalamo-pituitary-adrenal axis and could lead to non-neoplastic physiological hypercortisolism. Differentiating Cushing's syndrome (CS) from pseudo-Cushing's syndrome (PCS) is challenging.

Case Presentation

A 38-year-old woman, newly diagnosed diabetes mellitus and hypertension with body mass index of 45.5 kg/m2 was investigated for hypercortisolism. She did not have highly discriminative clinical features of CS such as easy bruising, thin skin, facial plethora, proximal myopathy, purple striae, osteoporosis or hypokalemia except moon face and supraclavicular fat pads. Investigations revealed non-suppressed overnight and low dose dexamethasone suppression tests (ODST and LDDST) with elevated adrenocorticotrophic hormone (ACTH) of 83 pg/mL (4.7-48.8). High dose dexamethasone suppression test showed >50% reduction of basal cortisol. Magnetic resonance imaging pituitary revealed hemorrhage into pituitary macroadenoma. Her other pituitary hormones were normal except prolactin which was 1864 mIU/dL (110-564). Midnight to morning cortisol ratio of 0.19 and midnight serum cortisol (awake) of 96 nmol/L favored PCS. Pre-operative vasopressin stimulation test revealed 140 % rise in ACTH from basal level with the absolute rise of 6.93 pmol/L favoring CS. Also, insulin tolerance test, significantly elevated ACTH, dexamethasone suppression test (DST) and urinary free cortisol supported CS. She underwent bilateral inferior petrosal sinus sampling which confirmed ACTH origin of pituitary gland.

She underwent trans-sphenoidal surgery twice and histology revealed pituitary hyperplasia and normal pituitary in 1st and 2nd surgeries respectively. Post-operatively, she had persistently non-suppressed DST and high ACTH level (183 pg/mL). Post-operative vasopressin stimulation test was suggestive of CS (337%, 17.70 pmol/L rise in ACTH). Finally, it has been decided to treat the patient by radiotherapy with the bridging therapy of fluconazole.

Discussion and Conclusions

Obese patients presenting with cushingoid features can be a case of Cushing's disease or PCS. Clinicians should pay attention to physical and psychological stressors which could lead to non-neoplastic hypercortisolism.

Keywords

Cushing's syndrome, Pseudo-Cushing's syndrome

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RESEARCH AND AUDITS

FIRST PLACE

OP - RP 01 – Assessment of the Correlation of Haemoglobin A1c and Serum Fructosamine with Mean Blood Glucose Level in Diabetic Patients with Chronic Kidney Disease in a Tertiary Care Hospital of Sri Lanka

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SECOND PLACE

OP - CR 03 – Efficacy and Effectiveness of Adrenal Venous Sampling in the Management of Primary Aldosteronism; Single Centered Cohort Study at a Tertiary Care Hospital in Sri Lanka

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THIRD PLACE

OP - RP 02 – The Association between Inflammatory Markers and Outcomes among Covid-19 Patients Admitted to Intensive Care Units at a Tertiary Care Hospital in Sri Lanka

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CASE REPORTS AND CASE SERIES

FIRST PLACE

OP - CR 01 – Shedding Light on Parathyroid Adenoma in Pregnancy : The Success of Parathyroid Hormone Assay in Fine Needle Aspirate: A Case Report

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SECOND PLACE

OP - CR 02 – Adenine Phosphoribosyl Transferase (APRT) Deficiency Presenting as a Rare Inherited Etiology of Nephrolithiasis: First Sri Lankan Case Report

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THIRD PLACE

OP - CR 03 – Risk of Ignoring Chronic Diarrhoea; A Patient with Carcinoid Tumour

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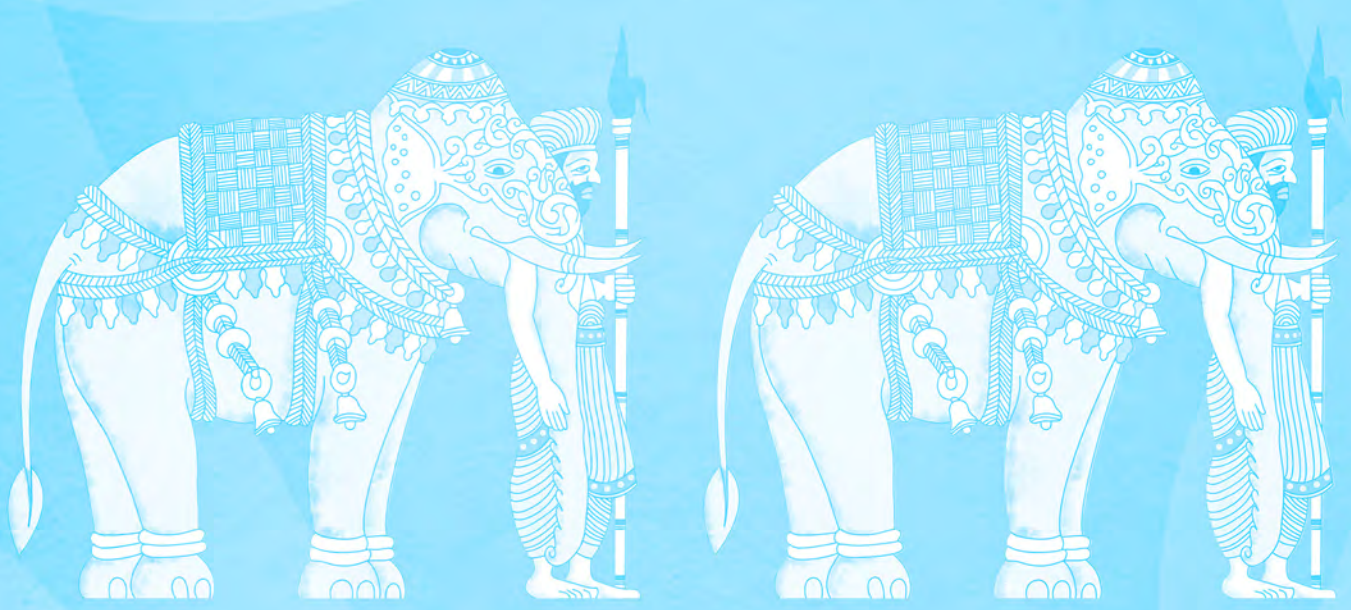

ABSTRACTS OF E-POSTERS

RESEARCH AND AUDITS

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RESEARCH AND AUDITS

RP 01 Determination of the Proportion of Biochemical Euthyroidism just before the Surgery and Assessment of Free T_4 Level in Postoperative Day 01 in Patients Undergoing Total Thyroidectomy Presented to the Base Hospital Avissawella, Sri Lanka

RP 02 Association between HbA_{1c} and Oral Glucose Tolerance Test (OGTT) in Patient with Fasting Plasma Glucose Values between 100 mg/dL and 126 mg/dL (Impaired Fasting Glycaemia) Presented to a Tertiary Care Center in Sri Lankan Setting

RP 03 Comparison of Serum Levels of Klotho in Patients with Nephrotic Syndrome with Age and Gender-Matched Non-Nephrotic Syndrome Controls

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AR 03 An Audit on Assessing the Completeness in the Request forms for Cortisol Analysis at National Hospital of Sri Lanka

AR 04 The Cost Saving by Performing Serum Thyrotropin Initially as the Only Thyroid Function Test in Teaching Hospital Karapitiya: An Exploratory Study

ABSTRACTS OF E-POSTERS

RESEARCH AND AUDITS CONTD.

RP 01

Determination of the Proportion of Biochemical Euthyroidism just before the Surgery and Assessment of Free T_4 Level in Postoperative Day 01 in Patients Undergoing Total Thyroidectomy Presented to the Base Hospital Avissawella, Sri Lanka

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Introduction and objectives

Total thyroidectomy is a common major surgery which is done mainly for chronic multinodular goiters, thyroid malignancies and for chronic thyroiditis resulting lifelong exogenous oral thyroxine therapy. Preoperative biochemical euthyroidism before total thyroidectomy irrespective of the indication for the surgery is very important to reduce the anesthetic complications. Therefore, the aim of this study was to determine the proportion of biochemical euthyroidism just before the surgery and assessment of free T4 level in postoperative day 01 in patients undergoing total thyroidectomy by measuring the circulating free T_4 (FT_4) level in the postoperative day 01 to identify transient increment in the circulating FT_4 level as it can be correlated with the indication.

Methods

This was a hospital and analytical laboratory based cross sectional study. The participants were those who presented for the total thyroidectomies for December 16th 2017 to March 31st 2018, irrespective of the gender and the indication for the total thyroidectomy. Samples were collected just before the surgery for TSH and FT_4 and postoperative day 01 for FT_4. Statistical analysis was done using Microsoft Excel using Minitab (16th version).

Results

There was proportion of 97.56% of patients have shown biochemical euthyroidism and 2.44% showed hyper or hypothyroidism just before the thyroidectomy according TSH and FT4. 88.64% of patients showed biochemical euthyroidism day 01 post operatively. There was no significant increment in the circulating free T4 level measured in the postoperative day 01 due to the handling of the thyroid gland irrespective of the indication for thyroidectomy (p=0.8862).

Conclusions

There was a significant proportion of patient who had biochemical euthyroidism just before thyroidectomy and postoperative day 01. There was no significant association between pre and postoperative day 01 FT_4 levels (p=0.8862) irrespective of the indications.

Keywords

Euthyroidism, Total thyroidectomy

ABSTRACTS OF E-POSTERS

RESEARCH AND AUDITS CONTD.

RP 02

Association between HbA_{1c} and Oral Glucose Tolerance Test (OGTT) in Patient with Fasting Plasma Glucose Values between 100 mg/dL and 126 mg/dL (Impaired Fasting Glycaemia) Presented to a Tertiary Care Center in Sri Lankan Setting

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Introduction and objectives

Diabetes mellitus is a metabolic disorder associated with multiple etiologies. Patients with impaired fasting glycaemia and impaired glucose tolerance are important clinical aspects as they can predict the future diabetes and its complications. Fasting plasma glucose and OGTT require individuals to fast. In contrast HbA_{1c} is an indicator of chronic hyperglycemia and the test needs a non-fasting sample. Therefore, the aim of this study was to evaluate the association between OGTT and HbA_{1c} in patients with IFG.

Methods

This was an analytical laboratory based cross sectional study. One hundred twenty three (123) participants were selected. The selected participants were those who were having fasting plasma glucose value of 100-126 mg/dL (5.5-7.0 mmol/L), previously not diagnosed to have diabetes mellitus, presenting to the general medical clinic as cases for OGTT and HbA_{1c}. Selected populations were separated into 2 groups according to WHO and American Diabetes Association IFG cut off values. The two groups were fasting plasma glucose between 100 – 109 mg/dL and 110-126 mg/dL. OGTT and HbA_{1c} were performed to two groups. Statistical analysis was done using SPSS, Microsoft office excels 2016 and ANOVA.

Results

High risk of impaired OGTT was observed when FPG >110 mg/dL (Odd ratio=4.148) as these patient populations had significantly increased 2 hour OGTT values (p=<.001) and increased HbA_{1c} values of >6.1% (mean value of 5.7% and 6.5%) (Odd ratio=10.06). The linear regression model was determined with FPG, OGTT values to predict HbA1c values and r square was 0.369.

Conclusions

There was a significant association between OGTT and HbA_{1c} values in patients who were having FPG values of >110 mg/dL.

Keywords

HbA_{1c}, OGTT, Impaired fasting glycaemia

ABSTRACTS OF E-POSTERS

RESEARCH AND AUDITS CONTD.

RP 03

Comparison of Serum Levels of Klotho in Patients with Nephrotic Syndrome with Age and Gender-Matched Non-Nephrotic Syndrome Controls

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Introduction and objectives

Klotho has significant beneficial effects on the kidney by alleviating oxidative stress and its antiapoptotic properties. This protective effect of klotho in different types of nephropathy, including diabetic nephropathy, has been demonstrated in multiple studies. Further, Klotho levels are also affected by factors such as steroids. The pathologic alterations viz, increased oxidative stress, and inflammation, observed in different types of nephropathy have also been demonstrated in Nephrotic syndrome. However few studies have explored the role of Klotho in Nephrotic syndrome.

Methods

The current study aimed at evaluating Klotho levels in serum of children with Nephrotic syndrome when compared with controls. The study consisted of 40 patients of nephrotic syndrome whose Klotho levels have been compared with 40 age and gender matched healthy controls. Klotho protein levels were evaluated by enzyme linked immunosorbent assay using commercially available kits.

Results

The results indicated significantly higher Klotho protein levels in nephrotic syndrome patients with mean and SD of 83.92 ± 112.28 as compared to the controls with mean and SD of 27.15 ± 27.56 with p value of =0.001. In this study we tried to find out the relation between the Klotho protein with steroid taking and steroid non taking patients. We found higher Klotho protein levels in steroid taking patients (mean and SD of 125.68 ± 144.47) than not on steroid treatment (mean and SD of 30.14 ± 49.98) with p value of 0.05.

Conclusions

This study is the first to assess the klotho protein levels in nephrotic syndrome patients in the Indian population. In conclusion, this higher levels of klotho protein might be contributed by the steroid drugs given to the patients while on relapse. There are few studies that mentioned this type of behavior in klotho levels.

Keywords

Klotho Protein, Nephrotic syndrome, Oxidative stress, Proteinuria

ABSTRACTS OF E-POSTERS

RESEARCH AND AUDITS CONTD.

RP 04

Association of Lead and Redox Sensitive Transcription Factors in Occupationally Pb Exposed Population

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Introduction and objectives

Lead (Pb) is an omnipresent environment pollutant that has deleterious effects on various organ systems in the human body. One of the proposed underlying mechanisms of Pb toxicity is oxidative stress. Nuclear factor (erythroid-derived 2) – like 2 (Nrf2) and Nuclear factor – kappa B (NF-κB) are two key redox sensitive transcription factors that are found to modulate cellular redox homeostasis. The current study aimed to evaluate the association of blood lead levels (BLL) in occupationally exposed individuals with gene expression of Nrf2 and NF-κB.

Methods

The study comprised of 80 occupationally Pb exposed workers with age and gender matched healthy controls. Blood levels of Pb was measured in whole blood using atomic absorption spectrometry (GFAAS) with Zeeman correction using iCE 3500 AAS (Thermo Fischer Scientific, Waltham, USA) and mRNA expression was determined by RT-PCR in a BioRad CFX96 System by using miRCURY LNA SYBR® Green PCR Kit (Qiagen).

Results

The findings indicated significantly higher levels of Pb in the exposed group (5.91±4.13 µg/dL) down regulation of Nrf2 gene expression (fold change=0.58, p value=0.002) which showed negative correlation with BLL (r = -0.31, p value = 0.03). An increasing albeit non-significant trend was noted in NF-αB gene expression in the exposed group.

Conclusions

This is the first study to assess the expression of these transcription factors in a human cohort, wherein the dysregulated gene expression is suggestive of their involvement in attenuating Pb induced ROS and hence their potential role in regulation of cellular redox damage.

Keywords

Blood lead level, NF-κB, Nrf2

ABSTRACTS OF E-POSTERS

RESEARCH AND AUDITS CONTD.

RP 05

The Combined Effect of Neurogenic Locus Notch Homolog Protein 4 and Brain-Derived Neurotrophic Factor Variants on Schizophrenia

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Introduction and objectives

Schizophrenia is a chronic neuropsychiatric illness affecting 1% of the world population. Many genetic and environmental factors lead to schizophrenia. So, analyzing just one SNP in a single gene to explore the correlation between susceptibility genes and schizophrenia does not reflect the exact relationship between the disease and the gene. As a result, the current study sought to investigate the association of neurogenic locus notch homolog protein 4 and brain-derived neurotrophic factor variants combined effects on schizophrenia.

Methods

Hundred schizophrenic patients and 100 healthy controls were recruited from the department of Psychiatry. PANSS and GAF were used to assess the severity of schizophrenia. BDNF (rs6265) and NOTCH4 polymorphism (rs367398 and rs387071) were genotyped using PCR-RFLP. The inheritance models and haplotype analysis were analyzed using the software SNPStats. The strength of association was assessed using the OR with 95% CI.

Results

According to the study, the NOTCH4 gene variants rs367398 (G/G) and rs387071 (T/T) were associated with an increased risk of developing schizophrenia, with OR of 2.26 (CI: 1.01-5.05) and 0.55 (CI: 0.23-1.33) respectively. Both the NOTCH4 rs367398 and rs387071 genotypes were significantly associated (p=0.041 and p=0.019) with schizophrenia. Further, the haplotype analysis has also supported the association of NOTCH4 locus with schizophrenia but after joining the rs6265 SNP of the BDNF gene, recombination analysis revealed a combined effect on the susceptibility to schizophrenia, with GA-TT decreasing and CT/CC-GG/GA increasing risk of schizophrenia (Global haplotype association p=0.002).

Conclusions

rs367398 in NOTCH4 and rs6265 in BDNF are significantly associated with schizophrenia. Due to the allele-allele interaction, the combined effects of both the genes has an increased susceptibility to schizophrenia and play an important role in the symptomatology of the disease.

Keywords

BDNF, NOTCH4, SNP

ABSTRACTS OF E-POSTERS

RESEARCH AND AUDITS CONTD.

AR 01

A Comparison of Coefficient of Variant (CV%) of General Biochemistry Analytes among Laboratories in Sri Lanka

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Introduction and objectives

The internal quality control (IQC) assesses the imprecision of an assay via CV%. As there is no manufacturer assigned value for our analyzer, CV% is calculated using cumulative SD and mean routinely. Some of those were unacceptable compared to the international standards, but were acceptable when compared to the same manufacturer's assigned values for different analyzers. Therefore, this study intended to assess whether the CV% of different analyzers meet the international standards.

Methods

The CV% of 18 biochemical analytes in 12 different analyzers around Sri Lanka, not having the same assay method, were calculated using past one month IQC means and SDs. The CV% was compared with international standards (Westgard, European Federation of Laboratory Medicine and Royal College of Pathologists of Australasia) and manufacturer's assigned CV% values.

Results

Although the CV% of all analytes was acceptable compared to the manufacturer's assigned CV% for the respective analyzers, the CV% of some analytes was not acceptable in certain analyzers compared to any of the standards. Out of the 12 analyzers, CV% of albumin was acceptable in only 2, alkaline phosphatase was acceptable in 4, creatinine was acceptable in 8, total protein was acceptable in 6, lactate dehydrogenase was acceptable in 10 and high density lipoprotein was acceptable in 10 analyzers.

Conclusions

This study indicates, although all the analytical procedures achieved their respective manufacturer's assigned CV%, that CV% were not within the international standards. These standards are based on the fact that the analytical CV% should always be within the desirable limit of intra individual biological variation (CVi). However, the common practice is to maintain the analytical CV% according to the manufacturer's claim or a laboratory derived CV%. This can falsely reassure that the assay imprecision is acceptable, unless it is compared to the international standards based on CVi.

Keywords

Internal quality control, Coefficient of variation, Intra individual biological variation

ABSTRACTS OF E-POSTERS

RESEARCH AND AUDITS CONTD.

AR 02

An Evaluation of Cost per Test in a Government Laboratory to Determine the Cost Effectiveness of Outsourcing the Laboratory Tests

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Introduction and objectives

The prevailing financial crisis in the country has created a remarkable price hike in cost for analytical reagents and other laboratory consumables. The allocated hospital budget is inadequate to purchase the required amount of reagents with this increased price. Hence, there is a delay in supply of reagents which results in an interruption to the continuous service provision. The aim of this study is to evaluate the difference between cost per test in the government and private sector to determine whether the outsourcing to the private sector is beneficial to provide an uninterrupted service to patients.

Methods

The costs for reagents, other consumables, water and electricity, printing papers, waste disposal, accreditation and personal salaries (including overtime payments) were calculated for fifty analytes for a period of six months (July-December 2022) including both day and night laboratories. The number of tests performed during that period was calculated from laboratory monthly statistics. The calculated cost per test was compared with the cost per test in private sector laboratories. The cost minimization analysis was used to determine the least expensive alternative.

Results

Out of 50 analytes, 49 of analytes (general biochemistry, HbA1c, serum protein electrophoresis, hormones, hs-troponin I, tumor markers) were found to be less expensive when performing in the government sector.

When compared with the cost in the private sector, 19 analytes were >80% less expensive, 09 analytes were >70-79% less expensive, 14 analytes were >50-69 % less expensive, 6 analytes were >20-49% less expensive and one analyte was 5% less expensive in the government sector. Only the antinuclear antibody level was cheaper in the private sector compared to the government sector.

Conclusions

This evaluation showed that performing tests in the government sector is more cost effective than outsourcing laboratory tests even in the current crisis situation even with the huge price increment of reagents.

Keywords

Cost per test, Cost effectiveness, Outsourcing of tests

ABSTRACTS OF E-POSTERS

RESEARCH AND AUDITS CONTD.

AR 03

An Audit on Assessing the Completeness in the Request forms for Cortisol Analysis at National Hospital of Sri Lanka

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Introduction and objectives

Cortisol level in blood has diurnal variations and is influenced by certain physiological and pathological conditions. Assay related interference can be caused by certain metabolites and drugs when it is measured by chemiluminescence competitive immunoassay. For dynamic function tests (DFT) patients need to be prepared promptly.

Inappropriate sampling and patient preparation may lead to unnecessary clinical suspicion and inappropriate further investigations and management. Objective of this audit was to assess the appropriateness in ordering cortisol tests.

Methods

Three hundred and twenty request forms of cortisol tests during the month of March in radioimmunoassay laboratory, NHSL were analyzed retrospectively.

Results

One hundred and forty (43.8%) of the analyzed request forms were incomplete. Out of them, 79% lacked clinical history including medications, timing of sampling or the name of DFT were not mentioned in 24.3% and 21% were devoid of patient's identification (namely, age 16%, sex 14% and requesting ward 1%).

Female predominance of 53.7% was found amongst total request forms. Majority, 42.4%, belonged to patients in the 41-60 years group. Most of the requests, 48.1%, were received from medical units.

9 am and random cortisol were requested in 74.3% and 10.2% respectively. Others were analysed as a part of DFT. Indications for requests were hyponatremia in 18.9%, adrenal insufficiency in 15.4%, young hypertension in 13.1% and the remaining were documented as pituitary tumor, post pituitary adenectomy, cushinoid features and recurrent hypoglycemia.

Conclusions

Relevant patient identification details, relevant clinical history, sample timing and name of the DFT (if indicated) need to be mentioned in the request form in order to provide a report with correct results to the patient on time. This audit showed the inadequacy in ordering the cortisol analysis. Therefore, a checklist need to be followed when receiving the samples for cortisol analysis to avoid the wastage.

Keywords

Cortisol assay, Pre analytical errors, Request form

ABSTRACTS OF E-POSTERS

RESEARCH AND AUDITS CONTD.

AR 04

The Cost Saving by Performing Serum Thyrotropin Initially as the Only Thyroid Function Test in Teaching Hospital Karapitiya: An Exploratory Study

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Introduction and objectives

Clinicians order only Thyroid Stimulating Hormone (TSH) or both TSH and free thyroxine (fT_4) or all TSH, fT_4, and free triiodothyronine (fT_3) for evaluation of thyroid functions. Chemical Pathology laboratory (CPL) Teaching Hospital Karapitiya (THK) reviews all these requests as a policy to decide on the most relevant thyroid function test/s to perform. TSH alone is performed initially as the policy with some exceptions for all thyroid requests. Both TSH and fT_4 are performed for risk groups such as pediatric patients, pregnant women and patients from endocrine and diabetes clinics and peripheral hospitals. After evaluation of TSH results, fT_4 is performed in the second line depending on its necessity for clinical management. The laboratory does not offer fT_3 tests. This study is to determine the cost saving by adhering to the local laboratory policy (LLP) already described.

Methods

This was a retrospective analysis of TSH and fT_4 tests ordering pattern and performing pattern during 2021 in CPL, THK. The study was conducted using 2677 patient request forms from 01st of June to 14th of October 2021. The total number of fT_4 and fT_3 tests which were not performed under LLP was calculated and the cost saving was analyzed by using arithmetics.

Results

Out of 2677 total requests, 276 fT_4 and 151 fT_3 tests were not performed along with TSH in 1st or 2nd line during 136 days in 2021. Therefore, it can be predicted that 740 fT_4 and 405 fT_3 tests could be saved per year. It is approximately LKR 1,273,000 worth saving per year.

Conclusions

Limiting fT_4 and fT_3 tests under LLP, THK saves approximately LKR 1,273,000 per year. The laboratory has reduced the reagent expenses significantly by this practice. It is a rational practice especially in a country like Sri Lanka to face current economic challenges.

Keywords

Thyroid function tests, Cost saving

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ABSTRACTS OF E-POSTERS

CASE REPORTS AND CASE SERIES CONTD.

CR 01

Tumor Lysis Syndrome in a Patient with Secondary Leukemia Following Primary Mediastinal Germ Cell Tumor

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Introduction

TTumor lysis syndrome (TLS) is an oncological emergency characterized by hyperuricemia, hypocalcemia, hyperkalemia and hyperphosphatemia which lead to clinical manifestations of renal insufficiency, cardiac arrhythmia, seizures, multiorgan failure and ultimately death. We present a patient with primary mediastinal germ cell tumor (PMGCT) who developed TLS.

Case presentation

A 24-year-old gentleman, who presented with shortness of breath, cough and body aches, found to have a large anterior mediastinal mass with involvement of surrounding organs and neurovascular compartment. Biopsy revealed mixed germ cell tumor with components of immature teratoma and yolk sack tumor cells. Elevated alfa fetoprotein and human chorionic gonadotrophin levels were responded well following chemotherapy. Massive hepatosplenomegaly and severe anemia was noted 8 months after follow up. Atypical cells resembling myeloblasts were noted in the blood picture. Bone marrow aspiration suggested possibilities of myelodysplastic syndrome with excess blasts II or therapy related acute myeloid leukemia.

On the 5th day following chemotherapy, he was found to have hypocalcemia and hyperuricemia with rising of serum creatinine >26.5 µmol/L within 48 hours.

He fulfilled two laboratory and one clinical criteria according to the Cairo Bishop definition which favors TLS though phosphate was not available. Despite he was managing with adequate fluids, uric acid reducing therapy, and antibiotics, he expired after 8th day of chemotherapy.

Discussion and conclusions

It is a well-known fact that patients with PMGCT can have secondary leukemia. Cairo Bishop definition aid in early diagnosis of TLS in high risk patients.

Keywords

Tumor lysis syndrome, Primary mediastinal germ cell tumors, Cairo-Bishop definition

ABSTRACTS OF E-POSTERS

CASE REPORTS AND CASE SERIES CONTD.

CR 04

Atypical Presentations do Exist: A Child with Infantile Nephropathic Cystinosis

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Introduction

Cystinosis is a rare (prevalence-1-9/100000) autosomal recessive genetic disorder caused by CTNS gene (17p13) encoding the lysosomal membrane cystine transporter, cystinosin, resulting in excessive intra-lysosomal accumulation of cystine damaging various tissues/organs. The typical presentations of infantile cystinosis are failure to thrive, polyuria, polydipsia, refractory rickets, proteinuria and hypokalemic paralysis attributing to renal Fanconi syndrome. Extra-renal cysteine accumulation can manifest as photophobia, neurocognitive impairment, hepatosplenomegaly and endocrinopathy. Atypical initial presentations such as Bartter syndrome & distal renal tubular acidosis (dRTA) has also reported in the literature.

Case presentation

Herein, we describe an 8-year-old girl born to consanguineous parents presented with polyuria and polydipsia and severe failure to thrive since 11 months of age. The initial evaluation revealed the presence of normal anion gap metabolic acidosis, alkaline urine and hypercalciuria which directed towards the diagnosis of dRTA. During the course of the illness, she developed short stature, hypophosphatemia with phosphaturia and elevated ALP for which she was diagnosed and treated as rickets. Presence of phosphate wasting and rickets revised the diagnosis to Fanconi syndrome. Developmental regression was noted with time and CT brain revealed cerebral atrophy.

The patient also complained of photophobia which had been present since late infancy. The ophthalmic evaluation revealed cystine crystals in cornea. Considering the clinical presentation, the diagnosis of nephropathic cystinosis was raised which was finally confirmed by the presence of homozygous likely pathogenic variant in *CTNS* gene.

Discussion and conclusions

The nephropathic cystinosis is considered as the commonest etiology for childhood Fanconi syndrome. Thus it should be considered in all patients present with clinical features and laboratory evidence of Fanconi syndrome as early initiation of cysteamine therapy can retard the disease progression. Our case highlights the fact that cystinosis may present as diverse clinical features including atypical presentations making the diagnosis challenging.

Keywords

Cystinosis, CTNS gene, Fanconi syndrome

ABSTRACTS OF E-POSTERS

CASE REPORTS AND CASE SERIES CONTD.

CR 05

Thyroxine Absorption Test Differentiates Malabsorption from Pseudo-Malabsorption: A Case Series

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Introduction

Refractory hypothyroidism (RH) is defined as the persistence of clinical and/or biochemical hypothyroidism despite levothyroxine replacement beyond 1.9 µg/kg/day. The most common cause is non-adherence to treatment, referred to as pseudo-malabsorption. However, defective absorption of the drug from the small intestine should also be excluded. The thyroxine absorption test is a valuable tool which differentiates pseudo-malabsorption from malabsorption. Different protocols for thyroxine absorption tests are available.

Case presentation

A 65-year-old woman diagnosed with primary-hypothyroidism was evaluated for RH. While on levothyroxine 350 µg daily, her TSH and fT_4 were 19.7 mIU/L (0.465-4.65) and 8.67 pmol/L (10-28.2) respectively. Her drug compliance was good. Her thyroxine absorption test revealed basal TSH and fT_4 21.8 mIU/L and 8.39 pmol/L respectively. fT_4 was analysed hourly up to 8 hours and at 24 hours. Her maximum post-ingestion fT_4 was only 9.48 pmol/L at 24 hours which indicated malabsorption of the drug and warranted further evaluation.

A girl diagnosed with hypothyroidism at the age of 7 years was referred to the endocrine opinion on persistently high TSH with fluctuating fT_4 levels at the age of 12 years. While she was on levothyroxine 100 µg daily, her TSH and fT_4 were >100 mIU/L (0.465-4.65) and 7.45 pmol/L (10-28) respectively. She reported adherence to levothyroxine.

Her thyroxine absorption test revealed basal TSH and fT4 at 32.9 mIU/l and 6.13 pmol/L respectively. Her maximum post-ingestion fT_4 was 27.6 pmol/L at 4 hours which clearly indicated pseudo-malabsorption. She was advised on the importance of adherence to the treatment.

Discussion and conclusions

Poor adherence is the most common cause of refractory hypothyroidism. However, it is very difficult to elicit compliance from some patients. A two-fold increase of post-ingestion fT_4 from the basal indicates proper absorption. Patients diagnosed to have malabsorption warrant further evaluation.

Keywords

Hypothyroidism, Levothyroxine, Malabsorption, Pseudo-malabsorption

ABSTRACTS OF E-POSTERS

CASE REPORTS AND CASE SERIES CONTD.

CR 06

Hair Depigmentation and Dermatitis: A Rare Presentation of Cystic Fibrosis

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Introduction

Cystic fibrosis (CF) is a multisystem disorder following defective CFTR protein, leading to ductal obstruction and subsequent organ dysfunction. The common presentations of CF are recurrent respiratory symptoms, exocrine pancreatic insufficiency, meconium ileus, failure to thrive, obstructive jaundice and male infertility. Rectal prolapse, pseudo-Bartter syndrome, dermatitis and hair depigmentation are considered as uncommon initial presentations where the diagnosis can be challenged.

Case presentation

A 2-months old baby boy, second surviving child of non-consanguineous parents developed poor weight gain and found to have hepatomegaly, an erythematous, maculopapular rash over lower limbs and trunk and hypo-pigmented hair. Laboratory investigations revealed iron deficiency anemia, hyperproteinemia, mildly elevated liver transaminases and the presence of fat globules in stool. The first child died at six months of age following pneumonia and was under investigation for failure to thrive, hepatomegaly and rash. For skin rash and hair abnormality, the possibility of CF as the underlying metabolic disorder was raised by the dermatology team.

The sweat testing revealed sweat chloride concentration of 90 mmol/L (>60 mmol/L- CF suggestive) suggestive of CF. Two heterozygous pathogenic mutations identified in the *CFTR* gene [c.1099del p.(cys343fs*) and c.53+1G>C] supported the genetic diagnosis of CF. Genetic analysis of healthy female sibling also revealed one heterozygous similar pathogenic variant in *CFTR* gene [c.53+1G>C] confirming the carrier status. With pancreatic supplementation and pulmonary rehabilitation not only he achieved satisfactory growth, but also his skin rash disappeared and hair regained normal color.

Discussion and conclusions

Cystic fibrosis usually presents with respiratory conditions and pancreatic insufficiency which makes the diagnosis easy. But in some instances, very rare and unusual symptom could be the initial presentation of this condition such as in our case. Even though, the dermatitis and hair hypopigmentation considered as very rare initial presentation of CF, it has already been reported in <20 cases worldwide.

Keywords

Cystic fibrosis, *CFTR*, Sweat testing

ABSTRACTS OF E-POSTERS

CASE REPORTS AND CASE SERIES CONTD.

CR 07

Case Series - Two Children with Self-Mutilation Behaviour and Hyperuricemia

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Introduction

Lesch–Nyhan syndrome (LNS) is a rare X - linked recessive disorder. LNS is caused by a deficiency in hypoxanthine-guanine phosphoribosyltransferase in the purine salvage pathway. LNS is characterized by hyperuricaemia, hyperuricosuria, neurologic dysfunction, and cognitive and behavioral disturbances including self-mutilation.

Case presentation - Case 1

A 3-year-10-month-old boy, born to non-consanguineous parents presented with global developmental delay (GDD), irritability and dystonia since birth, and self-mutilation behavior since 2 years of age. Clinical suspicion of LNS was supported by hyperuricaemic hyperuricosuric state: serum uric acid 525 µmol/L (119-327), and urine uric acid/creatinine ratio 2.22 µmol: µmol (0.26-0.65) with normal serum creatinine 35 µmol/L (30-60). Red cell HPRT activity was undetectable. MRI brain revealed cerebral atrophy. Mutation analysis revealed hemizygosity for the variant HPRT1 (C.402+1G>A). Xanthine oxidase (XO) inhibitor (allopurinol) was initiated on confirmation of diagnosis. 2 years post initiation of allopurinol he developed renal stones with preserved renal functions. Follow up urine Xanthine 4.332 mmol/L and hypoxanthine 5.616 mmol/L were above the limits of solubility, warranted dose reduction in allopurinol.

Case 2

An 8-year-old boy with parental non-consanguinity, presented with GDD since birth and self- mutilation behavior since 2 years of age. Investigations revealed marginally elevated serum uric acid 329 µmol/L (119-327), elevated urine uric acid/ creatinine ratio 1.65 µmol: µmol (0.26-0.65) with serum creatinine within age-matched limits. Red cell HPRT activity was deficient, 1 nmol/h/mg Hb (95-175). MRI brain revealed cerebral atrophy. He had a history of ammonium urate (80%) renal stones at 6 years of age. At the time of diagnosis, he had the feature of gout arthritis and gouty tophi. Allopurinol started after diagnosis.

Discussion and conclusions

Delayed diagnosis and treatment will result in complications of hyperuricaemia. Marginal hyperuricaemia warrants further investigation in a suggestive history. Furthermore correct dose adjustment of XO inhibitor is crucial in preventing xanthine stones formation.

Keywords

Lesch Nyhan Syndrome, Hyperuricaemia, Self-mutilation

ABSTRACTS OF E-POSTERS

CASE REPORTS AND CASE SERIES CONTD.

CR 08

Case Report - Postpartum Bone Crisis in a Mother with Gaucher Disease

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Introduction

Gaucher disease (GD) is an autosomal recessive lysosomal storage disease (LSD), caused by a deficiency in lysosomal α-glucocerebrosidase enzyme resulting in accumulation of its substrate, glucosylceramide in macrophages. Infiltration of the bone marrow by Gaucher cells experiences structural changes in the bone because of inadequate blood supply and remodeling. Pregnancy can exacerbate GD posing a higher risk of osteopenia, osteoporosis, osteonecrosis, and fractures. Thus, the bone disease is a particular concern in the postpartum period.

Case presentation

A 22-years old diagnosed lady with GD presented with difficulty in walking and severe bilateral lower limb pain at 3 months postpartum. She had severe pain in her bilateral lower limbs. Though she was on initial enzyme replacement therapy (ERT), she had been defaulted for around 1 ½ years duration. Investigations revealed low albumin corrected calcium 6.7 mg/dL (8.4-10.2), elevated phosphorus12 mg/dL (2.3-4.7), normal ALP 52 U/L (40-150), low 25-OH-vitamin D level 9 ng/mL (< 20 deficient) with inappropriately normal intact Parathyroid Hormone (iPTH) 35.2 pg/mL (15-68). Her dual-energy x-ray absorptiometry (DEXA) scan indicated a low bone mineral density (BMD) T score of -4.3 (< -2.5 osteoporosis).

Discussion and conclusions

Enzyme Replacement Therapy (ERT) for GD improves symptoms and prevents further complications. Without treatment, pregnancy with GD has a risk of significant bone crisis. Deficiencies in vitamin D and calcium together with poor nutrition of this patient provoked bone disease. Inappropriately normal iPTH warrants further investigations for underline hypoparathyroidism. After assessing the compatibility with breastfeeding, the continuation of ERT should be considered. Calcium and vitamin D supplementations should be recommended to promote bone health. Close monitoring and multidisciplinary care throughout pregnancy and the postpartum period for all mothers with GD is mandatory to improve the pregnancy outcome and prevent complications.

Keywords

Gaucher Disease, Postpartum, Bone crisis

ABSTRACTS OF E-POSTERS

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CR 10

Solitary Bone Plasmacytoma of Appendicular Skeleton

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Introduction

Solitary plasmacytoma is a localized proliferation of monoclonal plasma cells without systemic involvement. It is rare and account only for less than 10% of plasma cell neoplasm. Median age of diagnosis is 60 with male predominance. This is of two types, solitary bone plasmacytoma and extramedullary plasmacytoma. Solitary bone plasmacytoma commonly involves the axial skeleton of bones.

Case presentation

A 42 year-old male was investigated for right side shoulder pain with restricted movements for 3 months. His basic investigations were normal with white blood cells $8x10^3$, Haemoglobin 14 g/dL, platelet 294 x 10^3, serum creatinine 76 µmol/L, total calcium 2.3 mmoKel/L. His X-ray of the shoulder joint was normal. Symptoms were worsening therefore they proceeded with Magnetic Resonance of imaging (MRI) of cervical spine. It reveals a 1.4 cm x 1.6 cm mass involving right scapular body, bony labrum and coracoid process. True-cut biopsy of the lesion suggested a plasmacytoma. His bone marrow has only 1% plasma cells. Whole body CT reveals no other lesions.

Serum protein electrophoresis shows monoclonal band in gamma region with paraprotein level of 1.8 g/L.

Urine Bence Jones negative with serum kappa 24.2 mg/L, lamda 41.3 mg/L and **κ/λ** ratio of 0.59.

Discussion and conclusions

Solitary bone plasmacytoma of appendicular skeleton is very rare at this age. Myeloma protein is detected only in 33% to 60% of plasmacytoma patients at the time of diagnosis and it is usually less than 1 g/dL. However our patient who had paraprotein level of 1.8 g/dL at the time of diagnosis responded well to treatment and his follow up serum protein electrophoresis detected no abnormal bands indicating good prognosis.

Keywords

Plasma cell neoplasm, Solitary plasmacytoma, Serum protein electrophoresis

ABSTRACTS OF E-POSTERS

CASE REPORTS AND CASE SERIES CONTD.

CR 12

Elevated CA 125 in a Patient without an Ovarian Pathology: A Diagnostic Dilemma

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Introduction

CA 125 is a glycoprotein which is expressed by epithelial ovarian tumours and other pathologic and normal tissue of mullerian duct origin. Though it is the most widely used tumour marker for epithelial ovarian cancers many benign conditions and other malignancies are associated with elevated CA 125. Therefore, careful interpretation of results in the clinical context is of Paramount importance.

Case presentation

A 65-years-old, unmarried, previously healthy female presented with abdominal pain for 1month duration which was associated with anorexia, fatigue and weight loss. Physical examination was unremarkable except for distended abdomen. General biochemical and hematological investigations were normal. USS abdomen revealed free fluid in the abdomen without ovarian pathology. Peritoneal fluid analysis was insignificent. Tumour markers were done to exclude malignancy and results were as follows, CA 125-5150 U/mL (0-35), CEA-5.42ng/mL(<2.5) and CA 19.9-4.4 U/mL (0-37) . Despite elevated CA 125 CECT abdomen and pelvis showed irregular enhancing soft tissue lesion of the peritoneum with omental thickening and normal ovaries. Colonoscopy didn't reveal anything abnormal. Due to inconclusive results a diagnostic laparotomy was performed and found to have many peritoneal deposits. Histology of these lesions revealed features of adenocarcinoma suggestive of primary peritoneal carcinoma. Though the patient was started on chemotherapy promptly she passed away due to sepsis.

Discussion and conclusions

Patient's clinical history and markedly elevated CA 125 were suggestive of a malignancy. Initial workup was focused on finding an ovarian pathology. Primary peritoneal carcinoma is a rare malignant tumour which has a poor prognosis and spreads transperitoneally. Optimal treatment of primary peritoneal carcinoma is surgical resection and chemotherapy. But prognosis is very poor with advanced disease. Therefore, it is very important to exclude it as a cause of elevated CA 125 in patients with unremarkable ovarian pathology.

Keywords

CA 125, Primary peritoneal cancer

ABSTRACTS OF E-POSTERS

CASE REPORTS AND CASE SERIES CONTD.

CR 13

A Young Lady with Progressive Weakness, Weight Loss, Shortness of Breath and Polyneuropathy

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Introduction

POEMS syndrome is a rare multisystem disease associated with an underlying plasma cell disorder. Its name is an acronym for Peripheral neuropathy, Organomegaly, Endocrinopathy, Monoclonal gammopathy and Skin involvement.

Case presentation

A 38-year-old female presented with bilateral lower limb weakness, difficulty in breathing for 3 months duration, oligomenorrhoea and decreased libido for 6 months. Despite the general body swelling, she had a significant weight loss of 10-15 kg over a year.

She was found to have enlarged cervical, axillary, inguinal lymph nodes, pigmented dark skin, bilateral pleural effusion, hepatosplenomegaly and reduced power in all 4 limbs with glove and stocking type sensory loss. Initial investigations revealed roulex formation with increased ESR. Further confirmed hypothyroidism, Adison disease, hypogonadotropic hypogonadism. Lymph node biopsy revealed Castleman disease. Bone marrow aspiration identified 5% plasma cell proliferation. Serum electrophoresis and immunofixation electrophoresis were suggestive of IgA λ monoclonal gammopathy. Serum free light chain assay revealed increased λ light chains with κ/λ ratio of 0.59 (0.22-1.74). The diagnosis of POEMS syndrome was made according to the IMWG (International Myeloma Working Group) criteria's, with presence of polyneuropathy and plasma cell disorder, one major criteria (Castleman disease) and presence of one or more minor criteria's (organomegaly, endocrinopathy, effusion and skin lesions). Systemic therapy was given. Long term monitoring was assessed with serum free light chain levels. Patient succumbed to her illness before the planned bone marrow study of flowcytometric immunotyping.

Discussion and conclusions

POEMS syndrome is a rare multisystem disorder that has high morbidity and mortality if left untreated. Early diagnosis and treatment prevent irreversible damage. But the disease could mislead and delay the diagnosis because of its complicating and varying clinical features. Serum Vascular Endothelial Growth Factor (VEGF) level is a major diagnostic criteria of POEMS syndrome, could not perform due to non-availability.

Keywords

POEMS, Serum free light chains, VEGF

ABSTRACTS OF E-POSTERS

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CR 14

Young Man with Testicular Choriocarcinoma

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Introduction

Testicular choriocarcinoma is a non-seminomatous germ cell tumor and is the rarest of all testicular tumors, accounting for less than 1% of all germ cell tumors and only 0.19% of all testicular tumors. It affects men between the ages of 15 and 35.

Case presentation

A 27-year-old man presented with 2 months history of enlarging right testicular lump. Physical examination revealed diffuse enlargement of the right testicle, which was firm and tender to palpation. Basic laboratory investigations were normal. A malignancy was suspected and tumour marker analysis was done to rule out a testicular malignancy. Serum β hCG (human Chorionic Gonadotrophin) level was elevated 85184 mIU/ml (<5.0) with slightly elevated lactate dehydrogenase (LDH) level 397 U/L (135-214) and normal alpha-fetoprotein (AFP) level. Imaging studies revealed a right scrotal mass with features consistent with malignancy. He underwent radical right side orchidectomy and chemotherapy afterwards. Histology confirmed the diagnosis of pure testicular choriocarcinoma of the right testis. The response to treatment is being monitored by the tumour marker β hCG and it has declined 210 mIU/ml and then 32 mIU/ml, one month and two months after the surgery respectively.

Discussion and conclusions

Pure testicular choriocarcinoma is a highly aggressive malignant tumor with early multiorgan metastasis and poor prognosis. Laboratory investigation involves tumor markers such as β hCG. β hCG is significantly high in pure choriocarcinoma (>1000 IU/L) and does not produce AFP. Imaging studies should be performed to localize distant metastasis.

Keywords

Testicular choriocarcinoma, β hCG, AFP, LDH

ABSTRACTS OF E-POSTERS

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CR 15

Biliary Atresia Biochemically Mimicking Carnitine Palmitoyl Transferase 1-A Deficiency

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Introduction

Elevated free carnitine to C16 ratio in whole blood analyzed by tandem mass spectrometry (TMS) is currently being used to screen for Carnitine Palmitoyl Transferase 1A deficiency (CPT-1A), which is a rare fatty acid oxidation defect (FAOD). Here we present a case where similar findings were seen in a patient diagnosed with biliary atresia (BA).

Case presentation

A two-month-old baby boy, second born to non-consanguineous parents with an uncomplicated birth was noted to be having yellowish discoloration of body since day 6 and intermittent passage of pale stools from day 12 of life. General biochemistry revealed moderately elevated transaminases, ALP 898 U/L (60-425), GGT 361 U/L (12-132), total bilirubin 155 µmol/L (3-20), and direct bilirubin 125.2 µmol/L (0-3) with total cholesterol of 8.67 mmol/L (1.71-5.91). Serial abdominal ultrasonography showed rapidly progressing hepatosplenomegaly with portal hypertension. BA and FAOD were considered as differential diagnosis. The hepatobiliary iminodiacetic acid scan performed was suggestive of BA and intraoperative cholangiogram revealed absent biliary tree confirming the diagnosis of BA. Thus, the patient underwent the Kasai procedure. During the intervening period, the acylcarnitine profile performed in an overseas laboratory in whole blood by TMS revealed elevated free carnitine (FC) 134.67 (5-125 µmol/L) and FC/C16 of 61.0 (4.2-52 µmol/L) which are also seen in CPT 1A deficiency.

Discussion and conclusions

Elevation in free carnitine with normal long chain acylcarnitine concentrations in blood as in our patient has been seen in most patients with BA. Although the exact reason is not highlighted, the literature suggests that the reduced expression of peroxisome proliferator-activated receptor α (PPARα) a nuclear receptor that stimulates genes involved in fatty acid oxidation including CPT-1A could be the cause. The case is evidence for the importance of correlating laboratory findings with the clinical history in the diagnostic process.

Keywords

CPT1A, Biliary atresia, PPARα, Acylcarnitine profile

ABSTRACTS OF E-POSTERS

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CR 16

A Patient with Primary Hyperparathyroidism with Controversial Imaging Findings

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Introduction

Primary hyperparathyroidism is a clinical entity presenting commonly with hypercalcemia and hypophosphatemia. The commonest cause for primary hyperparathyroidism is parathyroid adenoma but rarely four-gland hyperplasia and parathyroid carcinoma should also be considered. Localization of parathyroid lesion is important to decide on minimal invasive surgery.

Case presentation

A-58-year old female presented with heart burn, constipation and generalized body aches. She had hypercalcemia (2.67 mmol/L) and high ALP (201 U/L) with persistently normal phosphate levels. Her PTH level was elevated (104.9 pg/ml). Her ultrasound scan neck, Contrast Enhanced Computed Tomography (CECT and 4 D CT neck did not reveal a parathyroid lesion. Sestamibi scan revealed left inferior parathyroid adenoma .Selective parathyroid venous sampling localized lesion along middle thyroid vein. The patient underwent left inferior parathyroid adenoma excision and intra operative PTH was reduced to 10.2pg/ml. Histology of the lesion was compatible with parathyroid adenoma. Her calcium and phosphate normalized following surgery.

Discussion and conclusions

After biochemical confirmation of primary hyperparathyroidism radiological confirmation should be done. This may include imaging modalities such as ultrasound scan neck, Magnetic Resonance Imaging (MRI), Contrast Enhanced Computed Tomography (CECT), (99m) Tc-sestamibi scintigraphy and 4D CT. Selective parathyroid venous sampling is used when other imaging modalities are inconclusive for the localization of the lesion. Depending on the localization of the parathyroid adenoma minimal invasive surgical procedure can be decided by the surgical team to decide on the minimally invasive surgery.

Keywords

Primary hyperparathyroidism, Selective parathyroid venous sampling, Parathyroid adenoma

ABSTRACTS OF E-POSTERS

CASE REPORTS AND CASE SERIES CONTD.

CR 17

A Patient Diagnosed Multiple Myeloma with Negative Monoclonal Band on Serum Protein Electrophoresis

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Introduction

Monoclonal gammopathies have wide spectrum of presentations from monoclonal gammopathy of undetermined significance (MGUS) to overt multiple myeloma (MM). Multiple myeloma accounts for 1% of the total malignancies and 10% of the total hematological malignancies. Among all the multiple myeloma, 15% composed of light chain myeloma.

Case presentation

A 64-year-old male was evaluated for severe backache and difficulty of walking for two months. He had anemic symptoms and loss of weight. On evaluation his ESR was high (100 mm first hour) with anemia and moderate roulex formation. His serum creatinine was elevated (1.48 mg/dL) and corrected Ca was at upper reference range (10.5 mg/dL.). Serum electrophoresis revealed no monoclonal bands, but serum immunofixation revealed abnormal band in lambda region. When proceeded with urine electrophoresis, there was abnormal monoclonal band in gamma region and urine electrophoresis was suggestive of light chain myeloma with abnormal monoclonal band in free lambda region. Serum free light chain assay was high with serum free lambda value 1392 mg/dL (4.23-27.69). Serum free light chain involved to uninvolved ratio was 68.23. Patient had multiple lytic lesions in skeletal survey and bone marrow aspiration and trephine biopsy with > 80% plasma cells confirmed the diagnosis of light chain myeloma. He was started on bortazomib and dexamethasone.

Discussion and conclusions

It is important to go ahead with serum immunofixation, urine immunofixation and light chain assay even when there is no evident monoclonal band in serum protein electrophoresis, when clinical history is suggestive of multiple myeloma to exclude light chain myeloma.

Keywords

Light chain myeloma, Serum protein electrophoresis, Serum immunofixation

ABSTRACTS OF E-POSTERS

CASE REPORTS AND CASE SERIES CONTD.

CR 18

Positive Benedict Test in a Patient with Cataract

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Introduction

Galactosemia is caused by deficiency of enzymes in carbohydrate metabolism that converts galactose to glucose. This is due to, defect in converting galactose to glucose due to deficiencies of enzymes in Leior pathway. This causes accumulation of galactose in tissues leading to cataract, renal and liver dysfunction and cognitive impairment and even may lead to lethal consequences. Early diagnosis and management can lead to better prognosis of these patients.

Case presentation

A 16-day-old baby with an uncomplicated antenatal and birth history, presented with jaundice and progressive darkening of urine with a 20% of weight loss and one episode of hypoglycaemia. Examination revealed icterus, hepatosplenomegaly and bilateral cataract. Biochemical investigation showed direct hyperbilirubinemia with very high alpha-fetoproteins in serum. Positive urine Benedict test and negative glucose by strip method favoured galactosemia. High galactose levels in a dried blood specimen done at a reputed laboratory overseas by fluorimetry, confirmed the diagnosis. There was a gradual reduction of bilirubin and undetectable reducing substances on second day after starting soya- based formula.

Discussion and conclusions

Analysing urine for reducing substances by Benedict test is valuable in a patient suspected of having galactosemia in a developing country like ours.

Keywords

Galactosemia, Direct hyperbilirubinemia, Cataract

ABSTRACTS OF E-POSTERS

CASE REPORTS AND CASE SERIES CONTD.

CR 19

Case Report of a Child with Seizures and Skin Lesions- What to do With Biotin???

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Introduction

Biotinidase is needed for the recycling of biotin to maintain the free biotin pool, which acts as a cofactor for carboxylases such as propionyl Co-A (PCC), methylcrotonyl Co-A (MCC), pyruvate (PC) and acetyl Co-A carboxylase (ACC). Biotinidase deficiency an autosomal recessive disorder manifests as seizures, hypotonia, alopecia, skin lesions, vision and hearing impairment. It can be effectively treated with pharmacological doses of biotin and early detection improve prognosis.

Case presentation

A 2-month-old baby boy of consanguineous parents presented with seizures following vaccination. His immediate postnatal period which was complicated by sepsis responded to intravenous antibiotics. However much attention has not been paid to repeated short lasting jittery movements noted since day 6 of life. Examination demonstrated spasticity, generalized acrodermatitis and alopecia though growth was not affected. Routine investigations revealed mild anemia with elevated plasma lactate and uric acid. Oral biotin was initiated for the clinical suspicion of biotinidase deficiency.

Analysis of urine for organic acids confirmed the diagnosis as the chromatogram depicted lactic acid, 3-hydroxypropionic acid, 3-hydroxyisovaleric and 3-methylcrotonylglycine the compounds of deficiency of PC, PCC and MCC respectively. Later on analysis of dried blood spots for acylcarnitines by tandem mass spectrometer (high C5OH) and fluorometry assay for biotinidase activity of 15.84 U (normal >40) in an overseas laboratory supported the diagnosis of biotinidase deficiency. With biotin child's clinical state improved and urine collected after 1 month of biotin showed disappearance of abnormal compounds detected before indicating the miracle activity of the cofactor, biotin. Now the child is thriving well without seizures.

Discussion and conclusions

Clinical suspicion of biotinidase deficiency in patients with seizures and skin changes warrants initiation of biotin therapy that will change the overall clinical outcome. The laboratory plays a vital role in diagnosing biotinidase deficiency.

Keywords

Biotinidase, Seizures, Urine organic acid

ABSTRACTS OF E-POSTERS

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CR 20

A Young Male with Hypertension: Cushing Disease

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Introduction

Cushing disease remains one of the most diagnostically challenging endocrine pathologies due to the non-specificity of symptoms. The prevalence of hypertension in patients with Cushing syndrome is approximately 80%. Trans-sphenoidal excision is the first line treatment in functional pituitary adenomas. Mortality and morbidity remains high in Cushing disease despite early success and disease recurs in approximately 50% of patients.

Case presentation

A 22-year-old male was referred to the clinic due to high blood pressure. He complained weight gain, abdominal distention, facial puffiness over five months duration. He was plethoric, had moon face with central obesity. Baseline biochemical evaluation, thyroid function tests, follicular stimulating hormone,luteinizing hormone, prolactin levels were normal. 9.00 am cortisol and midnight serum cortisol were very high, levels were 902 nmol/L (118.8-618.0) and 861 nmol/L respectively. Both overnight dexamethasone suppression test (cortisol 878.7 nmol/L) and low dose dexamethasone suppression test (cortisol 946.0 nmol/L) were not suppressed establishing the diagnosis of Cushing syndrome. Plasma adrenocorticotropic hormone (ACTH) was 73 pg/mL (10-60). MRI brain showed 5 mm×6 mm size pituitary lesion, indicative of a pituitary microadenoma. Bilateral inferior petrosal sinus sampling (IPSS) confirmed pituitary source of ACTH production by central to peripheral plasma ACTH ratio of >2. Patient underwent trans-sphenoidal resection of the pituitary adenoma.

However, post operatively he developed polyuria and excessive thirst with elevated serumsodium (149 mmol/L), high serum osmolality (312 mOsmol/Kg) with low urine osmolality (161 mOsmol/kg). IM Desmopressin was given due to the probable diagnosis of diabetes insipidus. Ultimately, he made a complete recovery

Discussion and conclusions

IPSS is the most accurate procedure in differentiating hypercortisolism of pituitary or ectopic origin as compared with clinical, biochemical, and imaging analysis. Central Diabetes insipidus is a common complication following trans-sphenoidal hypophysectomy and occurs in up to 20% of patients. This case highlights the importance of careful evaluation of a young patient with hypertension.

Keywords

Cushing disease, Bilateral inferior petrosal sinus sampling, Diabetes insipidus

ABSTRACTS OF E-POSTERS

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CR 21

Three Patients with Adrenoleukodystrophy

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Introduction

X linked adrenoleukodystrophy, a rare inherited neurodegenerative disorder has always been a diagnostic and therapeutic challenge. It is caused by a defect in the ABCD1 gene that affects the central nervous system, adrenal cortex and testis. Up to now, six distinct phenotypes have been described, however no correlation has been found between the phenotype and the mutations. Currently there is no promising treatment to prevent the onset or modify the disease progression.

Case presentation - Case 1

A 10-year-old boy born to nonconsanguineous parents presented with lethargy, salt craving and darkening of the skin from the age of 3 years. He was hyperpigmented and basic biochemical investigations showed no abnormality. His 9 am cortisol was 60 nmol/L and there was inadequate response to short Synacthen test (SST) (cortisol 0 mins–76.5 nmol/L, 30 mins–78.6 nmol/L, 60 mins–82.5 nmol/L). Magnetic resonance imaging (MRI) of brain showed high T2/ FLAIR signal changes indicative of adrenoleukodystrophy. Genetic analysis identified a targeted variant in the ABCD1 gene in hemizygous state establishing a genetic diagnosis of X linked adrenoleukodystrophy.

Case 2

A 6-year-old brother of the previous patient, although relatively asymptomatic had hyperpigmentation on his lips and palms. His ACTH level was 1495 pg/mL(10-60) and SST showed a subnormal response (cortisol 0 mins-88.5 nmol/L, 30 mins-89.2 nmol/L, 60 mins-105.0nmol/L). Genetic analysis identified the same variant in the ABCD1 gene as his brother's.

Case 3

Mother of above siblings had a sister and her son developed recent onset behavioural changes and one episode of afebrile convulsions at the age of 8 years.Imaging revealed LOES MRI severity score of 11. Genetic analysis identified the same targeted variant in the ABCD1 gene. All the three patients were offered stem cell transplant (HSCT).

Discussion and conclusions

In adrenoleukodystrophy although the most common presentation is neurological, any combination of symptoms is a possibility. Adrenal failure being irreversible even after HSCT, is best treated with hormone replacement. These cases demonstrated the typical X linked recessive pattern of inheritance of the condition.

Keywords

Adrenoleukodystrophy, Hemizygous variant in ABCD1 gene

ABSTRACTS OF E-POSTERS

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CR 22

A Case Report of Bisalbuminaemia Associated with Chronic Inflammatory Demyelinating Polyneuropathy

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Introduction

Bisalbuminaemia is a rare protein anomaly characterized by the presence of two distinct fractions of albumin on serum protein electrophoresis (SPE).This benign condition may be inherited or secondary. We report a case of bisalbuminaemia with chronic inflammatory demyelinating polyneuropathy (CIDP).

Case presentation

Previously healthy 69-year-old male presented with progressive numbness and pain of both hands and feet for six months. Two of his brothers have been dead at 50s due to gastrointestinal carcinomas. He is an ex-alcoholic for 6 years with no significant medical or surgical history. Nerve conduction studies of limbs were suggestive of demyelinating pathology. SPE revealed bisalbuminaemia (36.6 g/L) and polyclonal increase of gamma globulins in the gamma region (13.9 g/L). All other biochemical investigations and imaging were normal. He was initially treated with i.v. immunoglobulins and is followed up with oral prednisolone up to date. SPE after 3 months revealed same bisalbuminaemia and polyclonal gammopathy.

Discussion and conclusions

Bisalbuminaemia is a rare dysproteinaemia characterized by the presence of an abnormal albumin in addition to normal albumin. The hereditary form is autosomal co dominantly inherited with a cumulative frequency varying from 0.7/1000 to 1/1850. Prevalence of acquired form is unknown. Acquired form has been reported in patients with high doses of beta lactams, pancreatic pathologies, monoclonal gammopathies, hepatopathies, hyperamylasemia, hypothyroidism, diabetes, Alzheimer disease, chronic renal failure and nephrotic syndrome. A case associated with CIPD was not found in the literature. Screening of his son and a brother revealed no bisalbuminaemia. Hereditary bisalbuminaemia which is due to a point mutation in albumin gene on chromosome 4 leading to autosomal codominant inheritance is a possibility and target gene mutation analysis is the definitive diagnostic method. We planned long term follow-up with SPE to decide whether bisalbuminaemia is CIDP associated or inherited.

Keywords

Bisalbuminaemia, Serum protein electrophoresis (SPE), Chronic inflammatory demyelinating polyneuropathy (CIDP)

ABSTRACTS OF E-POSTERS

CASE REPORTS AND CASE SERIES CONTD.

CR 24

Primary Pigmented Nodular Adrenocortical Disease (PPNAD): A Rare Cause of ACTH-Independent Cushing Syndrome

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Introduction

Primary pigmented nodular adrenocortical disease (PPNAD) is a rare cause of ACTH independent Cushing's syndrome. It can be associated with Carney's complex and has a characteristic gross and microscopic appearance. We report a case presenting with hypercortisolism due to PPNAD in her fourth decade.

Case presentation

A 33-year-old woman presented with secondary amenorrhea, weight gain of 6 kg over 2 months. She had a history of diabetes, hypertension, osteoporosis and severe depression with suicidal thoughts for 7 months duration. Examination revealed moon face, buffalo hump and central obesity. Cortisol following overnight dexamethasone suppression was 583 nmol/L (<50 nmol/L) and cortisol after low dose suppression was 543 nmol/L (<50 nmol/L). Her high dose dexamethasone suppression test didn't show suppression of cortisol while her ACTH of was undetectable. CECT adrenal protocol revealed right adrenal nodule (6 mm x 5 mm) and 9 mm enlargement of medial limb of left adrenal gland with preserved shape. Adrenal venous sampling (AVS) was performed and left adrenal vein cortisol was 4087 nmol/L with an inferior vena cava cortisol of 742 nmol/L though the right adrenal vein sampling was unsuccessful. Bilateral adrenalectomy was performed and histology suggestive of PPNAD. Patient recovered uneventfully and put on oral hydrocortisone.

Discussion and conclusions

PPNAD, as a cause of ACTH-independent Cushing's syndrome, may be difficult to diagnose. Adrenal imaging can be normal or there may only be minor or minimal adrenal nodularity that can hardly be distinguished from normal glands. Adrenal hyperplasia is described histologically not specific and nondescriptive itself.

Selective venous sampling has a ravishing advantage during successful cannulation to identify the functioning over nonfunctioning gland. AVS alone cannot confirm the diagnosis of PPNAD, yet it can be used as a surgical indication for adrenal gland resection.

Keywords

Cushing syndrome, PPNAD, AVS

ABSTRACTS OF E-POSTERS

CASE REPORTS AND CASE SERIES CONTD.

CR 25

Case Report-A Young Female with Primary Hyperaldosteronism

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Introduction

Primary hyperaldosteronism is characterized by elevated plasma aldosterone and suppressed renin release, with hypertension and hypokalemic alkalosis. This can be due to an adenoma of one adrenal gland, idiopathic adrenal hyperplasia, adrenal carcinoma, or glucocorticoid suppressible hyperaldosteronism.

Case presentation

A 34-year-old female was referred for evaluation of young hypertension due to persistent elevated blood pressure following delivery. Her past medical history was uneventful until the diagnosis of pregnancy induced hypertension at 27 weeks in her first pregnancy. Baby was delivered by emergency cesarean section due to high blood pressure. Her investigation findings revealed metabolic alkalosis with serum potassium 2.9 mmol/L. Ultrasound scan and the renal duplex was normal and 24-hour urine metanephrines were in normal range. While on verapamil, prazosin and oral KCl plasma aldosterone concentration was 19.74 ng/dL (7-30) and plasma renin concentration was 1 mU/L (4.2-4.5) with aldosterone to renin ratio of 19.7 ng/dL/mU/L (<3.7), which was highly suggestive of primary hyperaldosteronism. Plasma aldosterone following saline loading was 30.3 ng/dL (<5) which confirmed the diagnosis.

In the Adrenal protocol of the CECT abdomen, there was a 1.5 cm x1.1 cm x1 cm lesion compatible with a benign right adrenal adenoma. Laparoscopic right adrenalectomy was performed. Histology revealed adrenocortical adenoma. Post operatively, the patient's blood pressure normalized without antihypertensives.

Discussion and conclusions

The aldosterone to rennin ratio is a screening test for primary hyperaldosteronism. Failure of aldosterone suppression with saline loading confirms the diagnosis. Lesion localisation with imaging may be helpful and adrenal venous sampling is considered as the gold standard for differentiation of an adenoma from bilateral idiopathic adrenal hyperplasia. Patients below 35 years as in this case with spontaneous hypokalemia, marked aldosterone excess and CT evidence of unilateral adrenal adenoma may not need adrenal catheterization before proceeding to unilateral adrenalectomy. Patient was free from all clinical features following surgery.

Keywords

Primary hyperaldosteronism, Aldosterone to renin ratio, Saline loading

ABSTRACTS OF E-POSTERS

CASE REPORTS AND CASE SERIES CONTD.

CR 27

Role of Measurement Uncertainty in Clinical Decision Making

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Introduction

As all measurements are imperfect, an estimate of measurement of uncertainty provides an interval of values within which the true value of a test is believed to lie with a stated probability. So it improves the quantitative reliability of a laboratory measurement which is used in clinical decision making.

Case presentation

A 33 year old female was investigated for young hypertension with hypokalemia suspecting primary hyperaldosteronism. Her blood pressure was 160/100 mmHg with potassium level of 3.3 mmol/L. Apart from low potassium all other investigations were within normal range.

She had aldosterone renin ratio of 18 with plasma aldosterone concentration of 10.2 ng/ml and plasma renin activity of 0.6 ng/ml/hr. Her saline loading test revealed plasma aldosterone concentration of 23.91 ng/dL.

She underwent adrenal venous sampling as CECT adrenal protocol was normal.

Site	Aldosterone (ng/dL)	Cortisol (nmol/L)	Aldo/Cort Ratio	Selectivity Index
R/Adrenal	1300	738.18	1.760	1.93
L/Adrenal	56.5	886.12	0.063	2.32
Femoral	3.23	382.02	0.008	-

Lateralization Index: 27.93

As R/adrenal vein selectivity index was less than 2, measurement of uncertainty (MU) was calculated using coefficient of variation (CV) of each cortisol level.

Level of Cortisol	Mean	Standard deviation	Coefficient of variation
02 (femoral vein cortisol)	433	58.5	13.51%
03 (R/Adrenal vein cortisol)	759	101	13.30%

Since selectivity index is R/adrenal vein cortisol level into femoral vein cortisol level, measurement of uncertainty is calculated using the equation of $\sqrt{}$ (CV^2_2 +CV^2_3) × coverage factor of 02.

It was 0.3786. So dispersion of lab result was 1.93 ± 0.379

Hence 1.93 + 0.379 > 2, R /Adrenal vein considered as a cannulated side.

Discussion and conclusions

Patient may have an aldosterone secreting lesion in the right adrenal gland.It is difficult to come to a conclusion on adrenal venous sampling unless measurement of uncertainty is calculated. Measurement uncertainty should be calculated, evaluated, displayed and finally used to improve the quality and reliability of test reports.

Keywords

Measurement uncertainty

ABSTRACTS OF E-POSTERS

CASE REPORTS AND CASE SERIES CONTD.

CR 28

Optimizing the Hypoglycemia Cutoff in Insulin Tolerance Test: A Case Report

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Introduction

The principle of Insulin Tolerance Test (ITT) is increased cortisol level in response to insulin induced hypoglycemia stimulating the adrenocorticotropic hormone (ACTH) secretion. A normal cortisol response to ITT is a strong predictor of intact hypothalamic-pituitary-adrenal axis. An adequate cortisol response means the rise of cortisol to >550 nmol/L following adequate hypoglycemia (< 40 mg/dL). Here, adequate hypoglycemia should be achieved to interpret the test.

Case presentation

An 18-years-old diagnosed women with a giant prolactinoma underwent trans-sphenoidal pituitary adenectomy on last year. Her post-operative cortisol was 7.54 nmol/L. It increased only up to 119 nmol/L with hydrocortisone treatment. She was otherwise clinically stable and her thyroid function test was normal. Thirty minutes after commencing the ITT, her capillary glucose level dropped to 40 mg/dL and she had hypoglycemic symptoms. Blood was drawn for cortisol and plasma glucose analysis before correcting hypoglycemia.

Plasma glucose value at 30 minutes sample checked in lab was 43mg/dL. The cortisol values were 557.18 nmo/L and 584.33 nmo/L in in 45 minutes and 60 minutes respectively. With the determination of measurement uncertainty (MU) of plasma glucose, it was concluded as the patient was having adequate cortisol response.

Discussion and conclusions

To check the adequacy in cortisol response, the patient must reach adequate hypoglycemia with the plasma glucose value < 40 mg/dL. But in this case, the maximum drop in plasma glucose was 43 mg/dL. To eliminate the possibility of analytical variation, MU for that glucose value was calculated. As the MU (37.75 to 48.25 mg/dL) at 95% confidential interval (1.96 SD) is < 40 mg/dL, it was taken as the patient had reached adequate hypoglycemia. With the cortisol values > 550 nmol/L, it was concluded as the patient had adequate cortisol response.

This case has enlighten the need of determining MU to exclude the analytical variation, when a measured value has reached near standard cutoff target.

Keywords

Insulin tolerance test, Adequate hypoglycemia, Measurement uncertainty

ABSTRACTS OF E-POSTERS

CASE REPORTS AND CASE SERIES CONTD.

CR 29

A Rare Case of Severe Ectopic Cushing Syndrome due to a Thymic Carcinoma Recurrence

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Introduction

Endogenous or exogenous hypercortisolism gives rise to Cushing syndrome. ACTH dependent endogenous Cushing syndrome has two subtypes; Cushing disease and ectopic Cushing syndrome. Ectopic Cushing syndrome (ECS) is rarer compared to Cushing disease and is caused by ectopic secretion of ACTH/CRH. The main culprits include small cell carcinoma of lung, tumors of thymus and pancreas.

Case presentation

A 60-year-old woman with a past history of hypertension and diabetes mellitus, presented with hyperpigmentation of skin involving knuckles and palms along with symptomatic hypokalemia. There were no features of cushingoid appearance including moon facies, dorsal fat pad or striae. 10 years ago, she had undergone complete resection of a thymic tumor followed by chemotherapy. Histology had revealed a well differentiated neuroendocrine carcinoma. This time, patient had a high cortisol burden (9 am cortisol 1489 nmol/L) with unsuppressed overnight dexamethasone suppression test (ODST) and significantly high ACTH (146.8 pg/mL) confirming ACTH dependent Cushing syndrome. Contrast enhanced computerized tomography of chest and abdomen identified a recurrence of thymic neoplasm with local extension. Patient underwent bilateral adrenalectomy to reduce the cortisol burden following IV etomidate therapy with cortisol monitoring. However, due to a postoperative lower respiratory tract infection, the patient succumbed to death.

Discussion and conclusions

ECS due to thymic neuroendocrine tumor secreting ACTH is a rare but a recognized entity. Diagnosis of such, needs high index of suspicion and prompt investigations including biochemical and imaging studies. Typical cushingoid phenotype may not be apparent in ECS although rapid development of metabolic derangements may arise with extensive cortisol burden. It is mandatory to continue uninterrupted monitoring for recurrence after initial treatment together with patient education due to high recurrence rate of the tumor.

Keywords

Ectopic Cushing syndrome, Thymic carcinoma

ABSTRACTS OF E-POSTERS

CASE REPORTS AND CASE SERIES CONTD.

CR 30

Assessment of Insulin Resistance Using Quantitative Insulin Sensitivity Check Index (QUICKI): A Case Report

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Introduction

Insulin resistance is defined as "A subnormal biological response to normal insulin concentrations". It is usually associated with obesity, stress, certain medications, pregnancy, lipodystrophy, insulin antibodies, genetic defects in insulin-signaling pathways and blocking autoantibodies against the insulin receptor.

Hyperinsulinaemic euglycaemic clamp (HEC) is the "Gold standard" for insulin sensitivity measurement. As it is time and money consuming, several insulin sensitivity indices were developed using fasting plasma insulin, glucose and triglycerides to detect insulin resistance.

Case presentation

A 34-year-old man with known Budd-Chiari Syndrome body mass index of 23 kg/m2, was newly diagnosed with diabetes mellitus. His HbA1c and fasting plasma glucose were 9.9% and 351 mg/dL respectively. The rest of his biochemical investigations, including the lipid profile, were normal.

Despite increasing the dose of insulin step wise (more than 100 IU of Mixtard insulin/day), his blood glucose remained over 300 mg/dL persistently throughout the day. His Quantitative insulin sensitivity check index (QUICKI) was 0.2874 and insulin resistance was diagnosed.

Discussion and conclusions

This patient had QUICKI value less than the expected cut off for diabetic patients which further gave the room to access the cause for insulin resistance. In the recourse-limiting setting, we were unable to do insulin antibody levels or genetic studies to identify the course for resistance.

However, this case report provides a scope to review literature on using QUICKI to identify the insulin resistance. QUICKI is a novel empirically derived index that has better linear correlation with HEC, especially in obese and diabetic subjects. It is mathematically derived from fasting blood glucose (85 mg/dL) and fasting insulin levels (35.5 mU/L). To increase the accuracy of interpretation, different cut off values were used for obese, non-obese and diabetic patients.

Keywords

Insulin resistance, Hyperglycemia, QUICKI index

ABSTRACTS OF E-POSTERS

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CR 33

A Case Report: A Woman with Cushing's Disease Presenting with Pulmonary Embolism, Severe Refractory Hypokalemia and Hypercortisolism Induced Secondary Hypopituitarism

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Introduction

Cushing's disease is caused by ACTH-secreting pituitary adenoma leading to cortisol excess & occurs more frequently in females. Severe refractory hypokalemia can be a presenting feature in ectopic ACTH secretion but rare in Cushing's disease. Hypercortisolism have been reported to have 10 fold increased risk of venous thromboembolism, mostly during the active phase of the disease and even after biochemical remission. Thrombophlic state is caused by hypercoagulability [high levels of fibrinogen and VWF], impaired fibrinolysis and also by associated obesity & hypertension. Hypercortisolism itself can cause secondary hypogonadism and hypothyroidism, due to the inhibitory effects of glucocorticoids on hypothalamus and pituitary. Mass effect of the adenoma can also contribute to secondary hypopituitarism.

Case presentation

A 54-years-old obese female presented with worsening dyspnoea over past 3 days. On admission she was hypoxic and revealed typical clinical features of hypercortisolism. She had a history of recent onset diabetes mellitus, hypertension and depression for which treatment was started. Her serum potassium level was 2.1 mmol/l, 9 a.m. cortisol was >1000 nmol/L and ACTH levels were high, TSH, T_4 and FSH, LH all were low with elevated D-dimer levels. CT pulmonary angiogram revealed right sided pulmonary emboli while magnetic resonance imaging of the brain revealed a right sided pituitary macroadenoma without mass effect.

She was started with ketoconazole without going in to overnight (ODST) and low dose (LDDST) dexamethasone suppression tests and cortisol day curve was done to assess the treatment response. Later she underwent transsphenoidal removal of the tumor but found to have recurrence.

Discussion and conclusions

Although not common, high cortisol burden and hypercortisolism related life-threatening complications can be seen in Cushing's disease. Biochemical findings are very much important in the diagnosis, assessing severity, complications& in identifying recurrences in severe Cushing's disease.

Keywords

Cushing's disease, Pulmonary embolism, Hypokalemia, Secondary hypothyroidism

ABSTRACTS OF E-POSTERS

CASE REPORTS AND CASE SERIES CONTD.

CR 34

Clinical Presentation and Genetic Heterogeneity Including Two Novel Variants in Sri Lankan Patients with Infantile Sandhoff Disease: A Case Series

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Introduction

Sandhoff disease is a rare autosomal recessive neurodegenerative disorder due to the lack of α-Hexosaminidase activity which further subdivided into infantile, juvenile, and adult forms depending on the age of onset.

Case presentation

Eight diagnosed infantile Sandhoff disease (iSD) patients who were referred to the Chemical Pathology Department of Lady Ridgeway Hospital for Children during 2017 to 2021 were analyzed retrospectively.

The mean age of their presentation was 12.5 months, with female predominance. Most died at or around two years of age. All presented with developmental delay or regression associated with other typical features (seizures, abnormal head circumference and hepatosplenomegaly). Cherry-red spots on fundoscopy and hyperacusis played a crucial role in diagnosing iSD clinically. Café-au-lait spots also presented in all. Atypical features like mitral regurgitation and atrial septal defects were found in our patients. All had low total hexosaminidase activity.

We found c.1417 +5G>A and c.1303_1304insCT p.(Arg435Thrfs*10) novel variants of HEXB gene among the nine different gene mutations that were identified. Two HEXB gene variants were found in more than two patients, and two patients had more than two mutations in the HEXB gene.

Discussion and conclusions

This is the first Sri Lankan study that expands the clinical and molecular basis of iSD with its novel findings. Neurological features, hyperacusis and Cherry red spots on fundoscopy were consistent with the literature. iSD should be considered a differential when a child presents with cardiac involvement or café-au-laid spots with other striking features.

Though there were nine HEXB variants, including two novel variants, the genetic variance or hexosaminidase enzyme activity did not influence the clinical pattern of iSD.

In contrast to other studies, all our patients died within the age of two years, thereby proper management guidelines and follow-up methods needed to be established.

Keywords

Infantile Sandhoff disease, Atrial septal defect, Café-au-lait spots, HEXB gene novel variants

ABSTRACTS OF E-POSTERS

CASE REPORTS AND CASE SERIES CONTD.

CR 35

Presence of Calcium Oxalate Crystals in Cerebrospinal Fluid: Case Report

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Introduction

Cerebrospinal fluid (CSF) is an aqueous medium, present in the ventricles of the brain and the central canal of the spinal cord and flows around these organs in subarachnoid space. It transports substances like glucose, immunoglobulins and electrolytes and cells like lymphocytes and monocytes. Usage of CSF full reports for diseases involving the central nervous system is increasing. Sample collection errors are commonly occurring but correctable preanalytical errors that could mislead the diagnosis and delay in decision making.

Case presentation

A 35-year-old male presented with slurred speech and difficulty of complete closure of eyes for two days of duration followed by a five day history of fever and loose stools. He also complained of pain and numbness over fingers and toes and later pain in flexor muscles mainly biceps and hamstrings and lower back pain. He was apparently healthy before this illness. In the second of hospital stay, knee reflexes were diminished even though they were equivocal on admission. On microscopic examination of CSF, extracellular calcium oxalate crystals were noted. The test was repeated in a preserved sample that was taken at the same time and confirmed the same.

Discussion and conclusions

Hematoidin crystals in CSF suggest presence of old (~ 2 weeks) blood from intracranial bleeding while oxalate crystals suggest ethylene glycol poisoning or primary hyperoxaluria type 1. Clinically, the patient presented with features suggestive of atypical Guillain-Barre syndrome. Presence of crystals in CSF in the context of a given neurological picture could not dismiss the possibility of sample collection error.

Making a specific mark in the container than in the screw cap of the container to identify the "Glucose" tube and pre- labelling the containers before starting the lumbar puncture procedure are simple steps to avoid mixing up containers.

Keywords

Cerebrospinal fluid, Crystals

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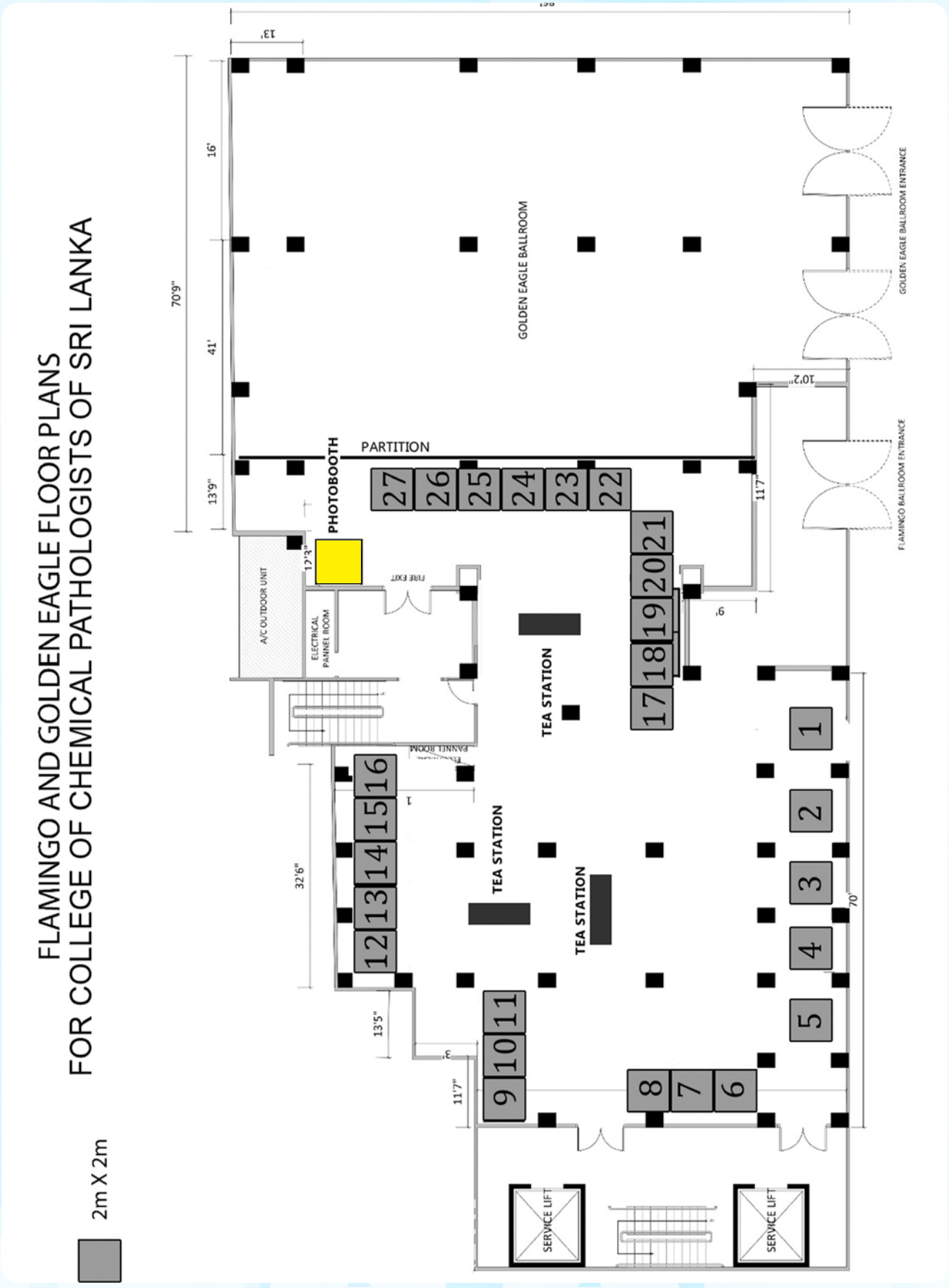

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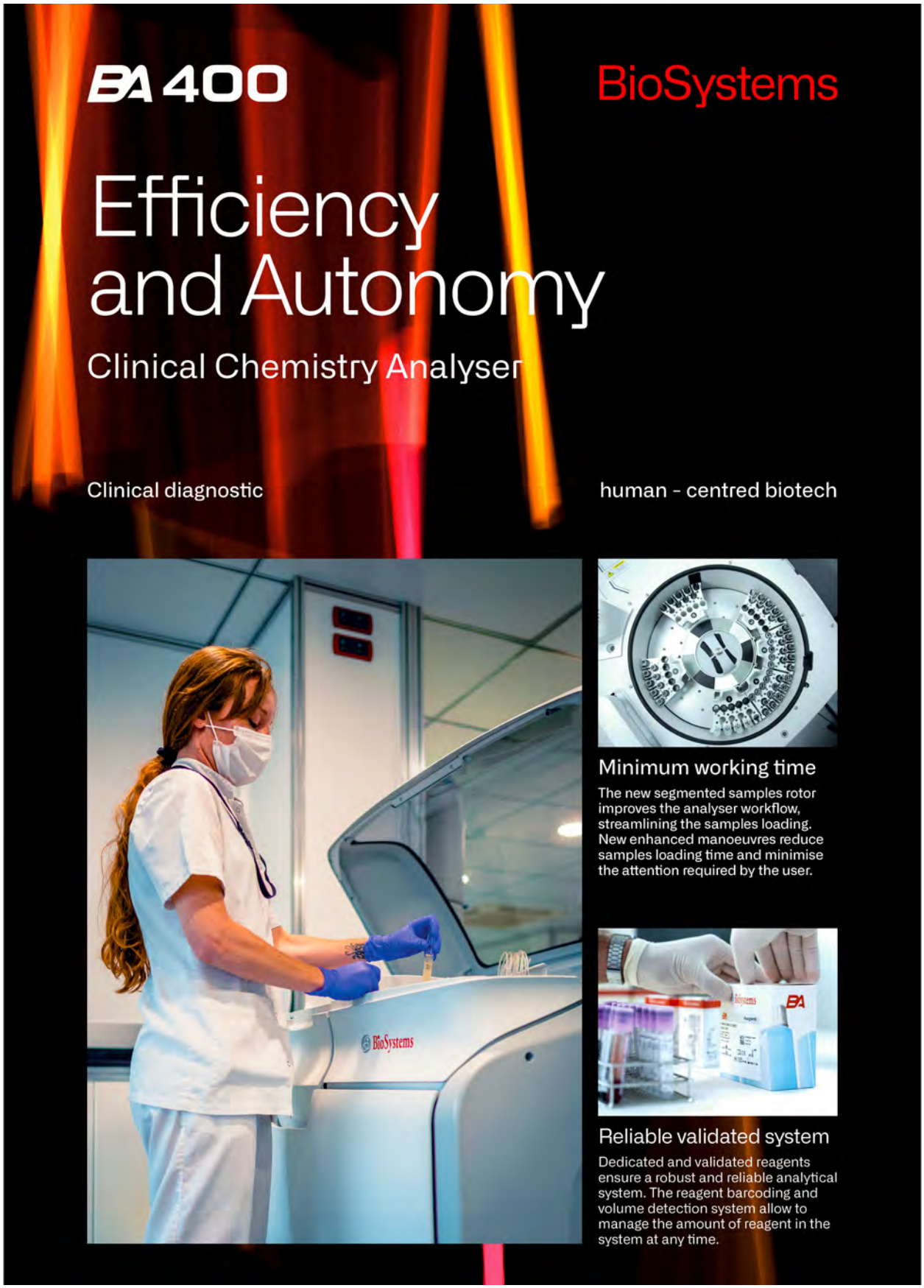
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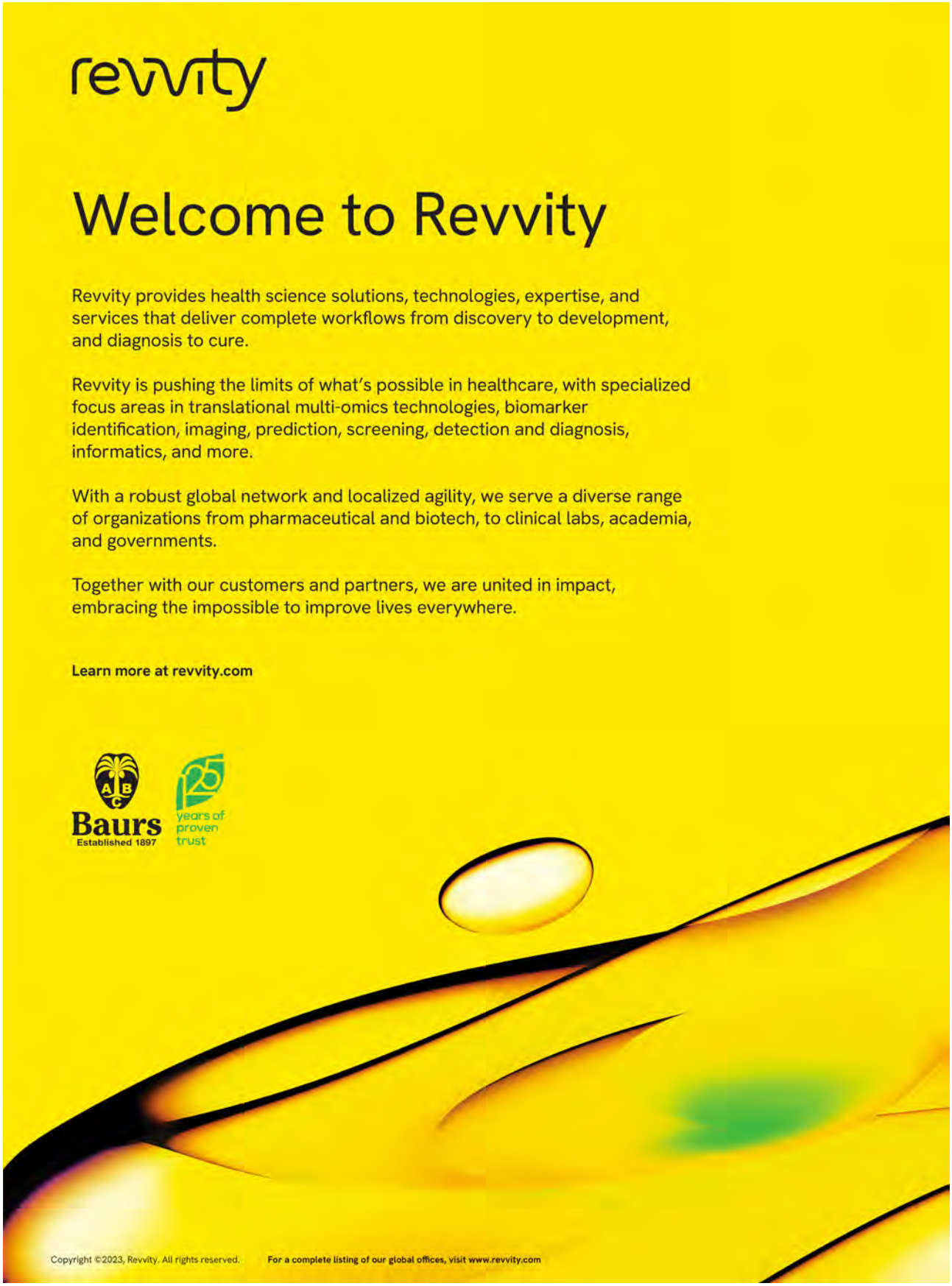
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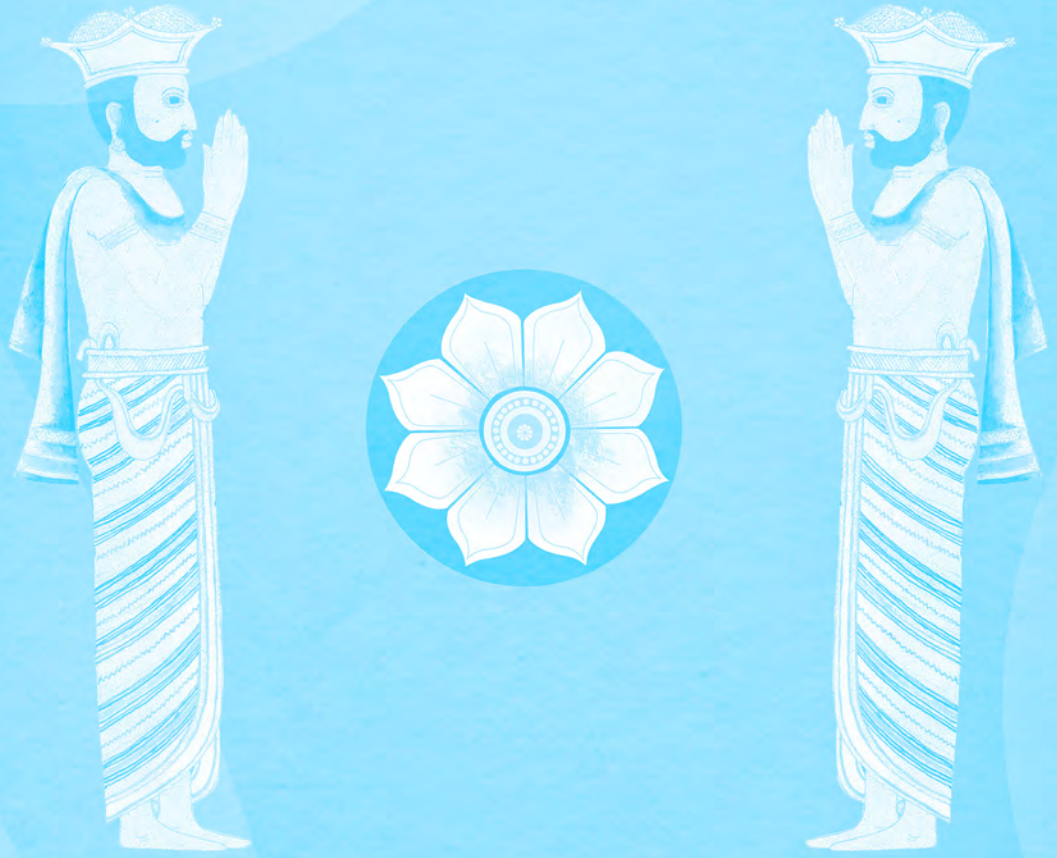

ACKNOWLEDGEMENTS

ACKNOWLEDGEMENTS

The President and the Council of CCPSL 2023, gratefully acknowledge the support and assistance given by;

- The Chief Guest, Dr Asela Gunawardena, Director General of Health Services, Ministry of Health Nutrition and Indigenous Medicine
- The Orator, Prof Ken Sikaris
- International Federation of Clinical Chemistry (IFCC)
- Asia Pacific Federation for Clinical Bio Chemistry and Laboratory Medicine (APFCB)
- International and local faculties
- Chairpersons of the plenaries and symposia
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- Diamond, Platinum, Gold, Silver, Bronze and other sponsors and all exhibitors
- Dr Chandula Fernando and Mrs Madhushi Uthpala, Coordinators, CCPSL
- Mr Kamal Dissanayake and the team of Global Events and Conventions Services, Event Organizer
- Mr Dinesh Gamage, Pasilta Graphics (PVT)Ltd, Printer
- Mr Dinesh Martis, Designer
- Mr Hemal Dissanayake, TEXTIT. BIZ, IT personnel
- Mr Amal Ranawaka, Photographer
- Management and Staff of Grand Kandyan Hotel, Kandy
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