Workshop Abstracts

The gene in prevention of disease?

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The degree to which disease risk is influenced by either genetic factors or environmental factors varies along a continuum. Some diseases have well-established genetic causes: sickle cell anaemia, hereditary haemochromatosis and Huntington's chorea, for instance, and can be attributed entirely to the inheritance of specific gene variants with particular functional consequences. In contrast, the development of other, more complex, multi-factorial diseases, such as Type 2 diabetes, obesity and rheumatoid arthritis is characterized by a less clearly defined combination of genetic, environmental, and behavioural influences.

The explosion of information generated by the development and use of a variety of gene-based technologies is having a profound and enduring impact on our basic understanding of the biological mechanisms underlying disease aetiology and pathophysiology. Accordingly, applied genetic research is increasingly focussed on how this expanding knowledge of gene structure, function and expression can be employed in practical approaches to prevent or reduce the risk of specific diseases.

This presentation focusses primarily on how information derived from genetic studies can be utilized in both the prevention and control of complex diseases. It describes how applications of current techniques in the fields of nutrigenetics, pharmacogenomics and toxicogenomics are being used to identify and assess disease risk. It also alludes to some of the more pressing conceptual, ethical, legal and social questions that are raised by information generated by genetic testing. The presentation concludes by speculating on the important roles that gene-derived knowledge could play in the future prevention and treatment of disease.

Lessons from sickle cell disease research: where are we now?

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Since sickle cell disease (SCD) was first described in the Western medical literature in 1910 – the result of astute bedside observations and use of a microscope – research has led to major innovations in clinical practice that, where available, have decreased morbidity and improved median survival rates. The scope and variety of sciences providing evidence that impact the lives of persons living with SCD has continued to increase. Basic sciences such as Physiology and Pharmacology continue to answer questions regarding pathophysiological mechanisms and molecular targets of therapeutic relevance. Genetic research seeks to identify modifiers of severity and mildness. All clinical specialties, including Nephrology, Neurology, Obstetrics and Gynaecology and Dentistry are collaborators in the effort to, through research, prevent or attenuate various clinical complications. Other sciences, such as Child Development, Psychology and Economics, are integral to developing and/or assessing the impact and validity of current interventions. The Sickle Cell Unit is uniquely placed to develop, pilot, implement and assess the effectiveness of interventions which can impact the lives of persons living with SCD locally, regionally and globally. Current collaborative efforts with the Ministries of Health, Jamaica and Brazil, the Pan American Health Organization, the Caribbean Network of Researchers in Sickle Cell Disease and Thalassemia and the Sick Kids Caribbean Initiative are examples of how synergy can be harnessed to make the most of available resources in a way that, in the final analysis, provides reliable data, which evidence can be used to inform policy-makers and colleague clinicians for the benefit of patients.

Plenary Lecture

Ethnicity and cardiometabolic risk paradox: lessons from the African Diaspora

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The global epidemic of diabetes has extended to the developing countries in Sub-Saharan Africa (SSA), the United Kingdom (UK) and the West Indies. In this context, black persons with Type 2 diabetes in the African Diaspora continue to manifest 1.5–2 times higher prevalence rates than in the white populations. We have demonstrated that blacks with and without Type 2 diabetes have alterations in hepatic and peripheral insulin sensitivity, beta cell function and insulin clearance (IC) when compared to whites. In addition, non-diabetic blacks in the African American Diaspora manifest multiple metabolic modifiers that predict Type 2 diabetes and its subtypes in blacks. These pathogenic modifiers include differences in subclinical inflammation, adipocytokines and oxidative stress burden in blacks in the African Diaspora prior to clinical diagnosis.

Despite greater insulin resistance, blacks have a lower prevalence of the metabolic syndrome (MetS) that can be partly ascribed to the lower prevalent rates of some major components of the MetS, namely the lower serum triglycerides and higher high-density lipoprotein cholesterol (HDL-C) levels in blacks when compared to whites. Moreover, the relationships among insulin resistance and cardiovascular disease (CVD) risk factors are weaker in blacks than whites. However, with the emerging inconsistencies in the association of insulin resistance and CVD risk factors in blacks, the use of current definitions and the cut-off points for MetS have become problematic. These can be explained in part by the multitude of metabolic abnormalities in the African Diaspora. These abnormalities include but are not limited to i) defective hepatic insulin extraction, ii) resultant secondary hyperinsulinaemia and peripheral resistance, iii) hepatic glucose dysregulation and iv) paradoxical relations between insulin resistance and triglycerides and HDL-C, visceral adiposity and the MetS. These metabolic alterations have strong genetic components which appear to play a pivotal and primary role in the pathogenesis of insulin resistance, Type 2 diabetes and CVD in blacks in the African Diaspora.

The importance of case studies in informing research

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Case reports and case series are the first line of evidence and are where medical evidence begins. The act of recording and publishing observations of the patients under our care allows us to build clinical knowledge-base and has historically been to our benefit. Case reports are essential in expanding our knowledge in reporting unusual presentation of a disease or unreported side effects of a drug. Variations in the presentation, diagnosis or management of new or emerging diseases are usually reported first in case reports and importantly, they form the basis of progressing to the higher level of evidence such as randomized trials. There are rare conditions that cannot be subjected to the rigors of a meta-analysis or systematic review. In this era of evidence-based medicine, case reports and case series form the lowest level of evidence, but let us not forget, evidence nonetheless.

Clinical trials – the "gold standard" in research

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Clinical trials have long been recognized as the "gold standard" in clinical research. Since 1968, The University of the West Indies (UWI), Mona has been involved in several randomized clinical trials (RCTs), published in the world's leading medical journals. These include hyoscine butylbromide, low dose aspirin and misoprostol to improve perinatal outcomes of pregnant and delivering women; the International Rotavirus Vaccine Trial in over 1800 Jamaican children, and hydroxyurea in sickle cell disease (HbSS) patients with painful crises. In 2013, the top five Faculty of Medical Sciences research publications and awards were for clinical trials, or for natural products to reduce hypertension and hypercholesterolaemia which have the potential to enter clinical trials. During the past eight years, attempts were being made to develop a Clinical Trials Centre (CTC) within the UWI Faculty of Medical Sciences at Mona "to generate income and enhance UWI's reputation" as well as to facilitate scientific advancement by improving translational research capacity. In response to several operating challenges under the current CTC's administrative structure, a multi-disciplinary Clinical Trials Advisory Committee (CTAC) was commissioned by the Dean to review the strengths and weaknesses of the current CTC and create a workable proposal to advance the "way forward". Several recommendations are being made and if implemented, will improve intramural clinical trials, as well as encourage and attract extramural pharmaceutical industry-sponsored clinical trials and research to the UWI Mona. The basic components include building a research facility and establishing the necessary physical infrastructure, expanding staffing, revising standard operating procedures, implementing training programmes while considering financing and budgeting to maximize income. When implemented, these should better enable UWI's CTC to achieve its stated goals.

Online evidence-based resources for the practising clinicians

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Background: Clinicians are commonly in need of current information for patient care. The use of online evidence-based resources (EBRs) for answering clinical questions at point-of-care is seldom used and poorly understood by clinicians in developing countries.
Objectives: To review and appraise commonly used online EBRs for clinical care and to provide recommendation for use with practising clinicians.
Methods: A search of the Cochrane Collaboration webliography of evidence-based healthcare resources was conducted and a review of databases offering online access to medical literature. An assessment of evidence-base medicine (EBM) tools was carried out to assess five domains: 1) quality of evidence, 2) accessibility, 3) relevance to point-of-care clinical practice, 4) affordability and 5) speed and ease of interpretation. Additionally, those resources that had a mobile application to facilitate real-time answers to clinical queries and freely accessible online were documented for poor resource settings.
Results: A total of 36 EBRs were identified, of which 70% were free and six being free only with registration. One-half was not relevant to physicians for point-of-care clinical practice but suited for patient evidence aid, policy-making and allied health professionals. Ten of the evidence-based resources met three or more of the five assessment domains. Only two of the resources were free and few had mobile applications or adapted versions for smartphone use. Evidence-based resources that rank highly on the five domains and considered applicable to low or poor resource settings included PubMed Clinical Queries and the Cochrane Library. The speed and ease of interpretation of these were variable and dependent on those translated into clinical summaries using the practical evidence about real-life situations model.
Conclusion: Relevant and quality evidence-based resources exist for use by physicians; however, most of these are fee based. Free resources which are applicable for poor resource settings are few and may not be relevant at the point of clinical care, with challenges in speed and ease of interpretation of evidence.

Communicating the evidence

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As part of efforts to equip clinicians and basic scientists to bridge the gap between the bench, the bedside and beyond, this presentation is aimed at highlighting the important facets of reporting research findings to clinical and non-clinical audiences as well as empowering researchers to evaluate and interpret appropriately the published research findings. This discourse will explore the different approaches to reporting research findings so that it is appropriately targeted at the audience that is on the receiving end. The different segments of a report to a scientific audience will be explained and the content the different sections – the Introduction, Methods, Results and Discussion – will be illustrated using examples. Approaches to reporting to a non-scientific audience and steps that can be taken to prepare scientists to bridge the communication gap will also be presented. It is hoped that as a result of this presentation, workshop participants will become more cognisant of the correct manner in which research findings are to be communicated. They will also be apprised of what they should expect from scientific and non-scientific reports of research findings.