WORKSHOP ABSTRACTS

Meet the Professors Workshop

A comparative analysis of multi-gene testing in breast cancer patients from Jamaica and The Bahamas

PO Roberts
Department of Surgery, Radiology, Anaesthesia and Intensive Care, The University of the West Indies, Mona, Kingston 7, Jamaica

Jamaica's population has evolved from the original inhabitants (Taino), the Europeans that came during and after colonization, Africans from the slave trade predominantly of West Africa origin, Chinese and Indians who came as indentured labourers, and some of the earliest Jews to migrate to the Americas. Over decades and centuries of interracial mixing, a population has evolved with likely a unique genetic make-up. Published studies have shown a high incidence of Lynch Syndrome among Jamaican patients with colon cancer and a high incidence of partner and localizer of BRCA2 (PALB2) in Jamaican patients with breast cancer. Though The Bahamas was involved in the slave trade with slaves from the same region of Africa as Jamaica and similar European colonization, over decades and centuries, another unique set of people likely evolved. Published studies have shown The Bahamas to have one of the highest incidences of BRCA pathogenic gene variants in the world. This introductory presentation seeks to share the results of panel testing for 84 cancer susceptibility genes in Jamaican and Bahamian patients with breast cancer. It seeks to show how significantly different the causes of hereditary breast cancers are in these two populations and the importance for routine multi-gene testing in patients diagnosed with breast cancer.

Ask why: never presume. Three anecdotes that led to remarkable research outcomes

F Prendergast

Frequently, the most effective biomedical research projects evolve from informed curiosity or seeming whimsy rather than logical precepts. I will argue that by presuming we know or when we trivialize good questions, we often fail to realize or stultify great potential. Examples from two recent Nobel Prizes.

How has genomics changed gastrointestinal/liver pathology and what have I learned from genomics along the way?

R Graham

Traditional diagnostic pathology of tumours is based on morphologic assessment with the use of immunohistochemistry. Advances in molecular technologies have opened new approaches to interrogate tumours and yielded the discovery of characteristic genetic events that underlie specific tumours. This has led to the development of new diagnostic biomarkers and an improved understanding of tumour biology. As a result, new treatment approaches and prognostic models have been implemented in routine clinical care. This novel paradigm has impacted every sub-specialty of pathology including gastrointestinal/liver pathology. Along these lines, I have had the opportunity to be a part of the discovery of novel fusion gene in gastroblastoma, the development of a molecular test for fibrolamellar carcinoma and the characterization of a number of tumour types. By the end of the presentation, the audience will be able to discuss the impact of molecular technologies on diagnosis and research of gastrointestinal/liver tumours, list molecular biomarkers for several tumour types and describe an approach to use molecular tools for research.

Neoplasms of the pancreatobiliary tract: is there still a place for conventional morphology in the era of molecular diagnostics?

M Reid

In recent years there have been numerous advances in our knowledge and understanding of the molecular alterations that underlie the development and advancement of cystic and solid tumours of the pancreas and biliary tract. These have significant diagnostic and prognostic implications. Although promising, many are still purely experimental or prohibitive in cost. Pathologists must therefore rely on morphologic clues as indicators of these alterations. This talk will explore recent molecular advances in pancreatobiliary pathology and their morphologic correlates, particularly in small biopsies, and with limited resources.

Main Workshop

Genomics or genetics? Common roots, diverse branches, shared opportunities

K Marshall

Genomics and genetics are multi-disciplinary branches of contemporary biological science; both are rooted in a common concept – the gene. Genomics, a framework that has come of age in the 21st Century, studies structural and functional aspects of the genome, the entire set of genes comprising an organism or cell. Genetics, on the other hand, emerged at the very beginning of the 20th Century, and investigates how the inter-generational inheritance of characteristics is associated with specific genes. In keeping with their common conceptual roots, these related disciplines share overlapping theoretical perspectives, methodological approaches and technological platforms. Not surprisingly, then, it can be challenging to draw the appropriate distinctions between these two gene-based sciences. Therefore, this introductory presentation employs recently published studies to make salient commonalities and contrasts between genomics and genetics. However, notwithstanding any differences between them, the presentation emphasizes how research utilizing these frameworks, either individually or collectively, is currently generating a vast amount of useable information, encompassing all areas of modern biomedicine. Moreover, it touches on how such research is helping to deliver on the promise of more personalized human healthcare, based on insights derived from these complementary sciences of the gene.

Plenary Lecture:
Genomics in clinical medicine: hope, hype and challenges

F Prendergast

In principle, genomics pervades all aspects of clinical medicine, from disease prevention to diagnostics and therapeutics. However, it is critical to realize that the universe of genomics in medicine perforce includes the inevitable impact of epigenomics as an equal and indeed vital modifier of genomic activity. In pondering the relevance to the current and future or potential applications to clinical practice, especially for personalized medicine, one must appreciate what the available technologies can and cannot do, the very real challenges facing the field for interpretation of genomic data in terms of the phenotype, and the very real ethical issues that may be the most difficult issues that need yet to be resolved.

Pharmacogenomics – current and future clinical applications

G Morse

Pharmacogenomics is the component of 'genomics' that includes the influence of genomic characteristics on clinical pharmacology and drug actions. This often is divided into genomic influences on pharmacokinetics (drug absorption, distribution, metabolism, elimination) or pharmacodynamics (drug response by cells/tissues/organs). Since pharmacokinetics often is related to pharmacodynamic responses, a triad emerges in which pharmacogenomics, pharmacokinetics and pharmacodynamics are inter-related and magnified when individuals require combination regimens and drug interactions occur. Post-human genome project, pharmacogenomics has been identified as the area of genomics that may impact the greatest number of individuals. Drug development research now incorporates pharmacogenomic testing during Phase I protocols, with continued inclusion of genotyping through to Phase IV studies. The subsequent evolution of proteomics, metabolomics and epigenetics have added complexity to protocols, yet, with the application of bioinformatics, has also identified important new aspects of pharmacogenomics in the pursuit of precision medicine and individualized medication regimens that may enhance treatment outcomes while minimizing drug toxicity.

About the *West Indian Medical Journal*

- In publication since 1951
- The only peer-reviewed medical journal in the English-speaking Caribbean
- Fully electronic (www.mona.uwi.edu/fms/wimj)
- The reference journal of diseases and medical research in the Caribbean
- Contains more Caribbean issues than any other journal
- Contents include original scientific research, reviews, brief communications, commentaries, viewpoints, proceedings of clincopathological conferences, continuing medical education sections, dental forum, case reports, and letters
- Published by the Faculties of Medical Sciences, The University of the West Indies, Mona, Jamaica, St Augustine, Trinidad and Tobago, and Cave Hill, Barbados
- Published in English, with abstracts of scientific research articles in both English and Spanish

For further information, visit or contact us at:

Faculty of Medical Sciences, The University of the West Indies,
Mona, Kingston 7, Jamaica, West Indies

www.mona.uwi.edu/fms/wimj • www.mona.uwi.edu/wimjopen
wimj@uwimona.edu.jm • wimjopen@uwimona.edu.jm

Telephone: +1 (876) 927-1214
Fax: +1 (876) 927-1846