



Pathology Biology Section - 2012

G94 The Coming “Omics” Revolution and Forensic Pathology

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After attending this presentation, attendees will gain an appreciation for the quickening of genomics, transcriptomics, proteomics, and metabolomics research that will likely revolutionize clinical medicine.

This presentation will impact the forensic science community by increasing recognition and raising awareness of why the “omics” revolution is closer than generally appreciated and what this may mean for forensic pathologists.

The medical community has been hearing about the importance of clinical genetic testing for some the last couple of decades, but the promised tsunami has not materialized. Nonetheless, advances have progressed. The convergence of cheaper technologies, the ability to data mine the information, and the greater understandings that have been achieved set the stage for transformative or disruptive new approaches to medicine. Specifically, the intersection of the informatics, robotics, microfluidics, and mass spectrometry has enabled the creation of vast data sets from single laboratories that are permitting ever deeper comprehension of integrative biologic systems. The human genome project began in 1990 and was completed in 2003 at a projected cost of \$3 Billion dollars. Today there are several commercial companies founded on the anticipation of whole genome sequencing for \$1,000.00 and the Archon Genomics X \$10M Prize has been recently offered for the first team to sequence 100 genomes in 10 days. Clinical efforts perhaps need only focus on the set of genes that comprise only a small part of the whole genome. Off the shelf gene chips can now analyze all 25,000 human genes. Clinical genetics laboratories are moving from single target assays to multiplex systems and high-throughput systems interpreted using massive parallel processing. In addition to DNA gene targets, the global expression of those genes through the messenger RNA in the transcriptome is now routinely tested and is more informative about what is going on in the cells because it is a measure of the gene regulation. Microarray chips have become cheaper, more ubiquitous and more consistent and robust. They are used in clinical labs now for leukemia classifications. The RNA is translated into proteins. The proteome is affected by post-translational processes, not seen in gene or expression arrays. Because proteins are present in higher concentrations, they offer some advantages to testing. Eventually, the proteins, as the work horses of the cell, result in metabolic functions which can be assessed through metabolomics methods. Through recognition of complex patterns, recognizable disease patterns emerge. Pathology, the study of disease, stands to be transformed by such interrogations. This has been recognized by the College of American Pathologists (CAP) and launched a multiyear campaign to transform the specialty and adapt to the new realities. The reintegration of clinical and anatomic pathology is a central concept of this effort. The CAP has created a Transformation Program Office which “facilitates, coordinates, and integrates Transformation-related activities across the College.” As Osler famously stated “as goes pathology, goes medicine” and “personalized medicine” is a goal that is becoming possible and even, perhaps, affordable. Accordingly, pathology residents are increasingly being exposed to these newer diagnostic technologies. If the health of a live individual can be so thoroughly described through omics testing, then surely it will have an impact on the diagnostics of those who succumb to such maladies. Yet there has been little talk of such these in forensic pathology. Genetic channelopathy testing and pharmacogenomics have garnered some discussion. Should the possibility of a molecular autopsy be considered in our armamentarium? Could a blood sample distinguish between an MVA caused by an MI or an MI cause by a MVA? Should we continue to cling to a procedure perfected in the 18th century as the main diagnostic tool?

Omics, Transformation, Genomics