J Mol Genet Med 2015, 9:3
http://dx.doi.org/10.4172/1747-0862.C1.010

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Impact of multiplexed genotyping and histology techniques for cancer biomarker discovery

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Our laboratory has performed clinical genotyping for over the past 5 years in a large academic hospital setting in order to provide a rational direction for directing treatment to our cancer patients. We have tested ~9,000 cancer patients clinically using our first generation platform and have acquired data on how this has expanded clinical utility through accrual in targeted therapy trials. Opportunities have been provided for both common malignancies such as breast cancer and rare malignancies such as bile duct cancer. Over the last year, we have converted testing to a next generation sequencing platform. Early benefits include the ability to identify a much more diverse mutational profile. One area of particular excitement is the insight that has been provided regarding tumor heterogeneity and how that may impact response to targeted therapy. While NGS is a tremendous clinical testing tool, a number of examples will be given on how traditional slide-based techniques can be valuable in putting these results into perspective.

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Gut microbiota-based biomarkers development

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Hampered for a long time by the limitations of culture-based methods, the analysis of gut microbiota has entered the era of next-generation sequencing and big data. It now provides an extensive overlook of the various microbes inhabiting the gut. Yet, the process that leads to biomarker discovery remains a challenge from the first steps on. Disregarding these steps may compromise downstream analysis at great cost. Looking back at 10 years of international pioneer efforts from MetaHIT and HMP, many practical lessons can be drawn for designing studies, efficient sample collection, shipment and storage, DNA extraction and sequencing and finally data analysis. Following simple precepts, a signature that combines microbial markers to address a clinical question can be obtained. Even if such test may be desirable and scientifically valid, it may not be adapted for clinical use, as turn-around time and costs for NGS analyses remain high. Transfer on more suitable platforms such as qPCR, is feasible which paves the way for realistic gut microbiota diagnostic tests. Enterome's development of a gut microbiota-based diagnostic of Non-Alcoholic Steato Hepatitis (NASH), a metabolic disorder, is an illustration of the different aspects of such development.

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