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NEWS

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'*The future of genomic medicine*'—satellite symposium at the American Society of Human Genetics meeting, 22 October 2007, San Diego, CA, USA

The medical and health applications of genome sciences and technologies were a highlight of the programme for the last annual meeting of the American Society of Human Genetics (23–27 October 2007) held in San Diego, CA, USA. The importance of genomic medicine was elegantly presented at a satellite meeting held on 22 October 2007. This meeting was appropriately entitled '*The future of genomic medicine*' and was attended by several delegates. It was a joint initiative by Scripps Genomic Medicine, a joint collaboration between Scripps Health and the Scripps Research Institute, and the J. Craig Venter Institute, founded by J. Craig Venter, representing its two divisions—The Institute for Genome Research (TIGR) and The Center for the Advancement of Genomics (TCAG). The conference faculty included several key researchers led by Dr. Eric J. Topol, an eminent clinical cardiologist and the Professor of Translational Genomics and the Director of the Scripps Translational Science Institute. He is also a practising senior cardiologist at the Scripps Clinic Division of Cardiovascular Diseases in La Jolla, California.

The guest faculty included several eminent invited speakers who delivered high quality lectures on a broad range of topics. Professor Eric Topol started deliberations by highlighting gaps and fallacies that exist in the current clinical practice. He cited examples for which applications of genomics would offer a reliable and effective approach. The importance of human genome variation in the form of SNPs and copy number variations (CNVs) was reflected in several presentations (Feuk et al. 2006; Redon et al. 2006). Professor Leena Peltonen from Helsinki in Finland presented the scope of complex traits mapping in managing common complex medical diseases such as diabetes mellitus, cancer and heart disease. The symposium covered advances and clinical applications of proteomics (Yates et al. 2005), metabolomics (Duarte et al. 2007), and cancer pharmacogenomics (Cheok and Evans 2006). Reports on genome wide studies in immune diseases (The Wellcome Trust Case Control Consortium 2007), autism (The autism genome project consortium 2007), cancer (Sjöblom et al. 2006) and cardiovascular disorders (The Wellcome Trust Case Control Consortium 2007) were stimulating for enthusiastic young researchers. Each session was concluded with a panel discussion involving speakers and moderators.

The symposium finished with a splendid talk by Dr. Samuel Levy, Senior Scientist at the J. Craig Venter Institute in Rockville, Maryland. He presented a full account of the sequencing of the diploid genome of Dr. J. Craig Venter who is renowned for his pioneering work on sequencing the Human Genome (Levy et al. 2007). This is probably the first step towards the goal of individualized genomic medicine achieved through the acquisition of diploid genome sequences at high accuracy and low cost. Dr. J. Craig Venter volunteered to have his complete diploid genome sequenced using Sanger sequencing technology. This revealed 0.5–1.0% sequence difference between chromosome copies. When inspecting contributing sequence reads and comparing the HuRef assembly to the NCBI version 36 human genome, it was possible to describe 4.1 million DNA variants, encompassing 12.3 Mb. Whilst 22% of these variants were *insertion/deletion events* (indels) the remaining 78% being SNP; they constitute the vast majority (74%) of all variant bases. This suggests an

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important role for non-SNP variants in defined diploid structure. Heterozygous variants occur in the untranslated and protein coding exons of 44% of all genes (10,208/23,224) suggesting a significant portion of the transcriptome is impacted by differential states of the diploid genome. This has led to some interesting stipulations made on Dr. Craig Venter's genetic predisposition for medical diseases and association with his behavioural characteristics.

Rapid commercialization of the personalized medicine—a dangerous slippery slope!

Any major international scientific and professional meeting could be an attractive occasion for a commercial profit-making incorporation to stage a mini-symposium sponsored by another linked commercial group, usually in the form of a delicious dinner and free supply of drinks. This was exactly the situation at one of the evening mini-seminars hosted by Affymetrix on behalf of Navigenics at the last American Society of Human Genetics meeting held in San Diego, California, USA.

This mini-symposium had an interesting title—'*Navigenics and the era of personalized medicine-the science, policy and ethics of personalized genetics*'. The seminar was well attended by several delegates and the panel included notable scientists, certified genetic counsellors, and clinical scientists engaged in the state of the art applied genome research in medicine and health. The keynote address was delivered by Dr. Dietrich Stephan, Director and Senior Investigator of the Neurogenomics division of Navigenics, who also happened to be the co-founder of Navigenics. The main purpose of his presentation was to introduce a high-throughput diagnostic kit designed to sequence 500,000 base pairs, enough to cover practically all known single nucleotide polymorphisms (SNPs).

It was envisaged that this would enable the prediction of an individual's risks for common medical diseases, notably common cancers, coronary artery disease, stroke, diabetes mellitus, dementia and probably many more! This was obviously very alarming and sparked off heated discussion on its purpose and perceived medical and health benefits. Furthermore the basis for risk calculations was unclear as the plan was to include data from several research reports that would have been produced using varying types of cohorts under differing conditions, often biased in the reporting methods. What was even more disturbing was a total lack of medical input in the decision making for an individual to embark on this kind of risk assessment based on crucial and sensitive genetic information? The plan had only made provision for some input by a certified genetic counsellor to assist in the interpretation of risks and advising on medical and health intervention. Not surprisingly several practising clinicians (including the author) felt very uncomfortable and raised concerns on this commercial approach that was apparently pre-mature lacking sufficient evidence and without any logical ethical justification.

This report highlights the danger of setting a precedence that could potentially undermine the importance and enormous scope of genomic medicine in the future clinical medicine. Dr. Eric Topol and his colleagues have also raised similar concerns and warned on the negative impact of commercialization of genomic medicine (Topol et al. 2007). This requires a concerted effort of all clinical scientists, researchers and regulatory organizations to ensure carefully introducing the each advancement supported by reliably collected evidence and taking into account all possible ethical, legal and social implications.

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