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ABSTRACTS

HGM2008 plenary abstracts: genomics and the future of medicine

001: From disease genes to biology, function and therapy

Veronica van Heyningen

MRC Human Genetics Unit, Crewe Road, Edinburgh, EH4 2XU, UK

The availability of multiple eukaryotic genomic sequences has revolutionized our ability to dissect the mechanisms of disease. It has become almost commonplace to identify genes underlying single gene anomalies; and recently genome-wide association studies have made great strides in pinpointing genomic regions that contain variants predisposing to common multifactorial diseases. Gene identification provides a major entré into the biological networks implicated in different disease states and therefore an essential springboard to the design of rational management regimes and therapies. The genome revolution has also brought a number of other advances. New sequencing and array technologies combined with molecular cytogenetics allow us to identify a broader spectrum of disease-associated mutations. Documented regulatory variation and mutations provide novel insight into how genes work and how they interact. Information on detailed genomic architecture and chromatin organization make possible the systematic exploration of copy number variation. Comparative and evolutionary studies allow us to use increasingly sophisticated model systems to understand human disease and phenotype modulation. The advent of induced pluripotential stem cells will permit direct analysis of multiple differentiated cell types from individuals and may ultimately lead to the development of acceptable routes to stem cell therapies.

002: Genetics as the basic science of medicine: but what of its practice?

Aravinda Chakravarti

McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University, Baltimore MD 21205, USA

There has likely been no better time to be a geneticist studying the human species. Long derided as hopelessly inadequate for genetic investigations, owing to its long generation time and limited offspring size, the human is now the premier object of study. This is not simply an anthropomorphic reaction: the wide diversity and ecological range of humans coupled with an uncanny ability to self-report and self-examine is the reason. Moreover, we no longer do genetics only 'through breeding' but do so 'through sequence'. Consequently, human genetics, particularly at the molecular level and particularly with a medical focus has gained in prominence. Recent studies from my laboratory, in traits as diverse as Hirsch sprung disease and sudden cardiac death, and as only one of many examples from many laboratories, emphasize that genetics is likely to predominate as the basic science of medicine since it provides the most direct route to understanding pathophysiology in an unbiased manner. The challenge is now to broadly apply what we have already learnt over the last 50 years and what we are learning now for the first time. This last task is more difficult than our community lets on.

003: Synthetic biology in pursuit of low-cost, effective, anti-malarial drugs

Jay Keasling

Center for Synthetic Biology, University of California, Berkeley, 5885 Hollis St. MC3224, Berkeley, CA 94720-3224, USA

Synthetic biology is the design and construction of new biological entities such as enzymes, genetic circuits, and cells or the redesign of existing biological systems. Synthetic biology builds on the advances in molecular, cell, and systems biology and seeks to transform biology in the same way that synthesis transformed chemistry and integrated circuit design transformed computing. The element that distinguishes synthetic biology from traditional molecular and cellular biology is the focus on the design and construction of core components (parts of enzymes, genetic circuits, metabolic pathways, etc.) that can be modeled, understood, and tuned to meet specific performance criteria, and the assembly of these smaller parts and devices into larger integrated systems that solve specific problems. Just as engineers now design integrated circuits based on the known physical properties of materials and then fabricate functioning circuits and entire processors (with relatively high reliability), synthetic biologists will soon design and build engineered biological systems. We are using synthetic biology to create inexpensive, effective, anti-malarial drugs. Currently, malaria infects 300–500 million people and causes 1–2 million deaths each year, primarily children in Africa and Asia. One of the principal obstacles to addressing this global health

threat is a lack of effective, affordable drugs. The chloroquine-based drugs that were used widely in the past have lost effectiveness because the *Plasmodium* parasite which causes malaria has become resistant to them. The faster-acting, more effective artemisinin-based drugs—as currently produced from plant sources—are too expensive for large-scale use in the countries where they are needed most. The development of this technology will eventually reduce the cost of artemisinin-based combination therapies significantly below their current price. To reduce the cost of these drugs and make them more widely available, we have used synthetic biology to engineer microorganisms to produce artemisinin from renewable resources. I will describe the process by which we engineered production of this important drug and the prospects for translating this research to people most in need of the drug.