SOCIÉTÉ CHIMIQUE DE GENÈVE

Genomes, evolution, and human disorders

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A large number of human disorders and other phenotypic traits are caused by or associated with genomic alterations. The current goal of genetic medicine is to perform the matchmaking between genomic variability and phenotypic variability. The completion of the sequence of the human genome and that of other species provided unprecedented opportunities to determine the functional elements and functional variability of these genomes. The elucidation of the cause of monogenic disorders was a great success of the past decade and will certainly continue in the next years not only to provide precise diagnostic tools, but also to understand the molecular pathophysiology. The main challenge ahead, however, is the discovery of nucleotide variability that confers (positive or negative) susceptibility to complex, common phenotypes: For each genomic variant to differentiate between “neutral” *vs*. “functional” or “pathogenic” variation. The completion of recent genomewide association studies for numerous phenotypes made it clear that common variation in the genome only accounts for a small fraction of the genetic etiology of common, multifactorial diseases. The reading of individual genomes will provide an enormous challenge in the discovery of causative genomic variants that could be used in predictive medicine. Comparative genomic analysis between species and between individuals, knowledge of the polymorphic structure of the genomes of different human populations, introduction of new tools to assess gene function, transcriptome analysis, rapid, inexpensive, and accurate DNA re-sequencing of genomes, and assessment of the quantitative variability of gene expression, are all necessary requirements to meet this enormous challenge of genomic and epigenetic pathology. In addition, the remarkable similarity of functional genomic elements in mammalian and other species provides further opportunities of animal experimentation for disease allele identification. In turn, functional analysis of the genome and characterization of the functional variability are likely to provide new therapeutic opportunities.

Conférence présentée le

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La conférence est publique

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