

BDR SEMINAR in Kobe

"CDB SEMINAR" and "QBiC SEMINAR" have been renamed "BDR SEMINAR".

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Tuesday, November 6, 2018

13:30-15:00, 7F Seminar Room, DB Building A

How can we use genomics to prevent serious diseases?

Summary

Within the past decade, we have witnessed the burst of genome-wide association studies (GWAS) to map genetic determinants in complex disorders; the rapid development of DNA sequencing technologies with a drop of several orders of magnitude in costs; and the progress in transcriptome and epigenetic studies, culminating in single-cell methods. Medicine is approaching a new phase where the application of this knowledge might benefit early diagnostics and disease prevention. However, enthusiasm and realism do not always appear to meet. One such wishful but less realistic direction involves the projected advent of personalized medicine based on GWAS markers. Despite the discovery of thousands of genetic marker associations, even their combined sensitivity and specificity remain far too low for population-based screening for most if not all complex disorders, such as coronary artery disease. Reasons for this lack of predictive power include, e.g., the slow and mostly unpredictable inflammatory processes that are prominent in many pathogenetic mechanisms and contribute to a random component in addition to genetics and the environment; the unpredictable rate of degenerative processes; and stochasticity of neoplastic processes. In contrast to such genetic associations, that I call "static" gene tests, I will emphasize the development of "dynamic" gene tests that monitor pathogenic processes in the body by, e.g., sequencing targeted loci in free circulating DNA in the search for typical cancer mutations, or looking for signs of tissue damage by altered DNA methylation profiles in circulating DNA. Such "dynamic gene tests", being highly sensitive, specific, and low-cost, can help to detect signs of serious diseases earlier than conventional biochemical assays. Such tests can be applied to population screening repeatedly at regular intervals to diagnose disease processes at their earliest, and may thus allow higher hopes for curative therapy.



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