

Cracking the cancer code: How a revolution in DNA sequencing is transforming our understanding of cancer

Cancer is a disease where the cellular software code – the genome – is corrupted. The code can be corrupted by deletion of large segments of DNA, point mutation, amplification, or chemical (epigenetic) modification. Identifying the spectrum of genetic changes in a cancer cell and understanding how they interact has been a major focus of cancer research for over thirty years.

Until recently, only a handful of the potentially significant genetic changes in a cancer cell could be scanned at high resolution in an individual patient's tumour. In the last few years, highly parallel techniques have been developed that have allowed a larger proportion of the cancer genome to be analysed in real-time. In particular, second-generation DNA sequencing technology has increased DNA sequencing output enormously, allowing for the first time full genome sequence analysis of an individual cancer genome.

Information from cancer genomes is redefining how different cancers are categorized, pointing to unexpected relationships between cancers arising in different organs. Genetic information is increasingly used to facilitate the diagnosis and management of cancer patients, and identify unaffected individuals at risk of developing cancer.

My presentation will provide a brief overview of the critical hallmarks of a cancer cell and will then turn to the development of technologies that are providing an unprecedented insight into the cancer genome. I will describe our involvement in the International Cancer Genome Consortium, an ambitious project underway aimed at mapping the genomes of the 50 most common cancers. Lastly, I will show how genomic information can provide profound insights into the biology of human cancers and improve patient outcome.

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