Pushing the boundaries to expand diagnostic applications of circulating DNA

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Blood plasma and other body fluids contain cell-free DNA, and in individuals with cancer this includes fragments of circulating tumour DNA (ctDNA) that originate from tumour cells. Analysis of cell-free DNA by appropriate genomic tools can provide information about the cancer status and its changes over time and in response to treatment. This has enabled the development of diagnostic applications based on cell-free DNA analysis for non-invasive molecular profiling and for detection of minimal residual disease, that are rapidly becoming part of the clinical standard of care. Large efforts are underway to develop, assess and establish the utility of cell-free DNA for earlier detection of cancer. Research in our group pursues new developments in sequencing technologies, and capitalises on discoveries about the molecular characteristics, epigenetic and fragmentation patterns of ctDNA. We apply our cutting-edge methods to cohorts of samples, carefully collected in collaboration with clinical investigators, to improve existing tools and develop new diagnostic strategies. Key projects include pre-symptomatic detection of cancer in populations at high-risk; improvement and implementation of tools for detection of residual disease; and intense monitoring of advanced cancer using simplified finger-prick self-sampling.