

Venue: PYRAMID 2  
21<sup>st</sup> August 2007  
1415-1515 hr

## **Plenary Lecture 1**

### **PL1. Biology and diagnostic applications of plasma nucleic acids**

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It is now well established that cell-free DNA and RNA of human origin can be found in the plasma of human subjects. This field started initially with the detection of tumour-derived DNA and RNA molecules in the plasma of cancer patients. Thus, tumour-associated genetic mutations, oncogene amplifications, epigenetic alterations and viral sequences have been detected in the circulation of cancer patients. Our group has focused on the measurement of cell-free Epstein-Barr virus (EBV) DNA in the plasma of nasopharyngeal carcinoma (NPC) patients. Plasma EBV DNA levels has been shown to be a sensitive marker for NPC and to be a powerful prognosticator.

In 1997, inspired by the detection of tumour-derived nucleic acids, we have discovered the presence of cell-free fetal DNA in maternal plasma. This result has now been translated into a number of noninvasive prenatal diagnostic tests, e.g., for fetal gender determination for sex-linked diseases and fetal RhD genotyping. More recent developments in this field include the discovery of fetal RNA and fetal epigenetic markers in maternal plasma. These advances have culminated in the demonstration that circulating fetal DNA/RNA could be used for the direct detection of fetal trisomy 21 and 18.

Finally, there are many other species of circulating nucleic acids, including those derived from a transplanted or a traumatized organ. It is expected that cell-free nucleic acid analysis may play an increasingly important role in future molecular diagnostics.

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