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Public Lecture

Non-invasive prenatal testing using fetal DNA in maternal plasma: from dream to reality

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Since 1997, it has been known that during pregnancy a fetus will release its DNA into the plasma of its pregnant mother. Hence, fetal DNA represents a mean of 15% of the DNA that is present in maternal plasma. This phenomenon has been rapidly translated into a number of non-invasive prenatal tests. Thus, fetal sex and RhD blood group status can be robustly detected from maternal plasma. Over the last 5 years, much of the efforts in the field have focused on the detection of fetal chromosomal aneuploidies, such as Down syndrome, through the use of massively parallel sequencing of maternal plasma DNA. The robustness of this approach has been validated in many large-scale clinical studies. Clinical applications of this technology for fetal chromosomal aneuploidy detection have started since the autumn of 2011 and have extended to over 15 countries around the world. Recently, genome-wide molecular karyotyping of the fetus and even fetal whole genome sequencing have been achieved through maternal plasma DNA sequencing. Hence, a new era of non-invasive prenatal testing has arrived. It is thus timely for ethical, social and legal implications of this new technology to be discussed amongst all stakeholders in the field. Ultimately, it is hoped that such developments would make prenatal testing safer for the fetuses and less stressful for pregnant women and their families.

Biography

Professor Lo is the Director of the Li Ka Shing Institute of Health Sciences and Chair of the Department of Chemical Pathology of The Chinese University of Hong Kong. In 1997, Professor Lo and his co-workers reported the presence of cell-free fetal DNA in the plasma of pregnant women. This discovery challenged the conventional wisdom regarding the role of the placenta as a barrier between the fetal and maternal circulations. The finding of circulating fetal DNA in maternal blood has also opened up new possibilities for non-invasive prenatal diagnosis. Over the past 15 years, Professor Lo has elucidated the fundamental biological characteristics regarding circulating fetal DNA, including its concentrations, gestational variations, length distributions, and clearance patterns. Professor Lo has demonstrated the use of such fetal-derived molecules for the prenatal diagnosis of sex-linked diseases, blood group genotyping, and a variety of monogenic disorders. To develop a non-invasive test for Down syndrome, Professor Lo and his team developed an approach based on molecular counting and showed that massive parallel sequencing is an efficient method for detecting fetal chromosomal aneuploidies. In 2011, he and his team published the first large-scale validation of this sequencing-based technology for Down syndrome detection, with confirmations by numerous groups since then. This technology was quickly introduced into clinical practice. Recently, Professor Lo has pushed the envelope of the field by demonstrating that a genome-wide genetic map of the fetus can be deduced by deep sequencing of the pregnant mother's plasma. Professor Lo's work has created a paradigm shift in prenatal diagnosis, making such testing safer for the fetus and less stressful for the pregnant mother.

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