GENOMICS AND BIOINFORMATICS

Principal Investigator

Professor Peiyong Jiang

Team

Bioinformatics

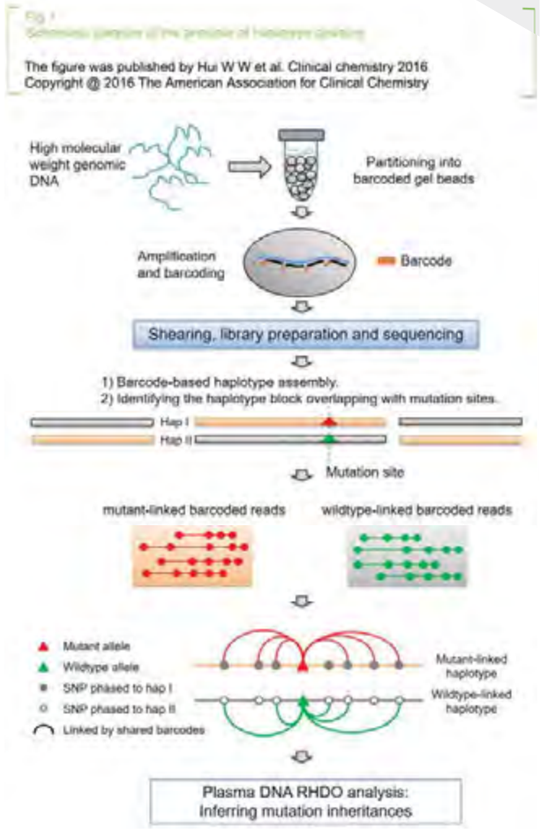
Research Progress Summary

In this year, Professor Peiyong Jiang and his team have made a number of achievements:

1) a haplotype-based bioinformatics tool was developed which can be universally applied to non-invasive prenatal testing (NIPT) for single gene diseases;
2) demonstrated that synergistic analysis of cell-free DNA sizes and counts could reduce the false positives and differentiate the origin of aberrations present in maternal plasma in NIPT;
3) illustrated that the methylation and size profiles of cell-free DNA could be used for predicting gestational age; and
4) a sophisticated bioinformatics tool was developed to improve the non-invasive detection of fetal *de novo* mutations in maternal plasma using the cell-free DNA properties.

Schematic diagram of the principle of haplotype phasing.

Long DNA molecules were partitioned into gel beads and amplified by unique 10xTM barcoded primers.



Recognitions

Grants and Consultancy

Full Name of PI	Project Title	Funding Source	Start Date (dd/mm/yyyy)	End Date (dd/mm/yyyy)	Amount (HK$)
Peiyong Jiang (co-PI)	Plasma DNA as a Platform Technology for Cancer Detection	Research Grants Council – Theme-based Research Scheme	01/06/2016	31/05/2020	44,444,000
Peiyong Jiang (co-I)	Research into Circulating Fetal Nucleic Acids	Research Grants Council – Theme-based Research Scheme	01/01/2016	31/12/2020	37,286,000

Publications

A. Journal Papers

1. Chan KC, Jiang, P, Sun K, Cheng YK, Tong, YK, Cheng SH, Wong AI, Hudecova I, Leung TY, Chiu RW. Second generation noninvasive fetal genome analysis reveals de novo mutations, single-base parental inheritance, and preferred DNA ends. *Proceedings of the National Academy of Sciences*. 2016; 113(50):E8159-68.
2. Hui WW, Jiang P, Tong YK, Lee WS, Cheng YK, New MI, Kadir RA, Chan KC, Leung TY, Lo YM. Universal haplotype-based noninvasive prenatal testing for single gene diseases. *Clinical Chemistry*. 2017; 63(2):513-24.
3. Jiang P, Lo YM. The long and short of circulating cell-free DNA and the ins and outs of molecular diagnostics. *Trends in Genetics*. 2016; 32(6):360-71.
4. Jiang P, Peng X, Su X, Sun K, Yu SC, Weng IW, Leung TY, Sun H, Chiu RW, Lo YM. FetalQuantSD: accurate quantification of fetal DNA fraction by shallow-depth sequencing of maternal plasma DNA. *npj Genomic Medicine*. 2016; 1:16013.
5. Jiang P, Tong YK, Sun K, Cheng SH, Leung TY, Chan KC, Chiu RW, Lo YM. Gestational age assessment by methylation and size profiling of maternal plasma DNA: a feasibility study. *Clinical Chemistry*. 2017; 63(2):606-8. (Letter to the editor)
6. Yu SC, Jiang P, Chan KC, Faas BH, Choy KW, Leung WC, Leung TY, Lo YM, Chiu RW. Combined count-and size-based analysis of maternal plasma DNA for noninvasive prenatal detection of fetal subchromosomal aberrations facilitates elucidation of the fetal and/or maternal origin of the aberrations. *Clinical Chemistry*. 2017; 63(2):495-502.
7. Wong FC, Sun K, Jiang P, Cheng YK, Chan KC, Leung TY, Chiu RW, Lo YM. Cell-free DNA in maternal plasma and serum: a comparison of quantity, quality and tissue origin using genomic and epigenomic approaches. *Clinical Biochemistry*. 2016; 49(18):1379-86.