結合人工智慧以及生物資訊學進行基因體判讀

臺大醫院基因醫學部
陳沛隆醫師

Education

2003-2009 **Ph.D.**, Human Genetics and Molecular Biology Program, Institute of Genetic Medicine, Johns Hopkins School of Medicine

2000-2002 **Master of Medical Science**, Graduate Institute of Clinical Medicine, National Taiwan University (NTU)

1988-1995 **M.D.**, College of Medicine, NTU

Positions

2016- Associate professor and director, Graduate Institute of Medical Genomics and Proteomics, NTU

2009- Attending physician, Departments of Medical Genetics and Internal Medicine, NTUH, Taipei, Taiwan

2010-2013 Secretary general, the Endocrine Society of the Republic of China (Taiwan)

Research interests

Next-generation sequencing (NGS), genotyping technology, genomics, immunogenomics, pharmacogenomics, endocrinology, genetic mapping, bioinformatics

Selected Publications

Wu *et al*. (2019). Plectin mutations in progressive familial intrahepatic cholestasis. Hepatology. doi: 10.1002/hep.30841.

Yang *et al*. (2019). Distinctive genetic variation of long-segment Hirschsprung's disease in Taiwan. Neurogastroenterology & Motility. e13665. DOI: 10.1111/nmo.13665

Hsiung *et al*. (2018). Identification of a novel *LDLR* disease-causing variant using capture-based next-generation sequencing screening of familial hypercholesterolemia patients in Taiwan. Atherosclerosis. 277:440-447

Tung *et al*. (2018). Comprehensive human leukocyte antigen genotyping of patients with type 1 diabetes mellitus in Taiwan. Pediatric Diabetes, DOI: 10.1111/pedi.12645

Wu *et al*. (2018). ABO genotyping with next-generation sequencing to resolve heterogeneity in donors with serology discrepancies. Transfusion, 16.

Chen *et al*. (2016) First step towards precision medicine for antithyroid drug-induced agranulocytosis. The Lancet Diabetes & Endocrinology. 4:473.

Chen *et el*. (2015) Concurrent exome-targeted next-generation sequencing and single nucleotide polymorphism array to identify the causative genetic aberrations of isolated Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome. Human Reproduction. 30:1732-42.

Chen *et al*. (2015) Genetic determinants of antithyroid drug-induced agranulocytosis by human leukocyte antigen genotyping and genome-wide association study. Nature Communications. 6:7633.

Chen *et al*. (2013) Application of massively parallel sequencing to genetic diagnosis in multiplex families with idiopathic sensorineural hearing impairment. PLoS ONE 8: e57369.

Chen *et al*. (2009) Fine mapping on chromosome 10q22-23 implicates *neuregulin 3* (*NRG3*) in schizophrenia. The American Journal of Human Genetics 84:21-34.