

PRESIDENT'S TECHNOLOGY AWARD 2018

Professor Stuart Cook

Tanoto Foundation Professor of Cardiovascular Medicine
Professor & Director, Cardiovascular & Metabolic Disorders Signature Research
Programme (CVMD SRP), Duke-NUS Medical School (Duke-NUS)
Director of Cardiac MRI & Senior Consultant, Dept of Cardiology, National Heart
Centre Singapore (NHCS)
Director, National Heart Research Institute Singapore (NHRIS)
Professor, Clinical and Molecular Cardiology, Imperial College

“For his outstanding research in human genetics and cardiovascular disease, which have impacted the field of precision medicine, and resulted in the development of tools for genetic sequencing now used in clinical practice worldwide”

Professor Stuart Cook is a leading cardiovascular research expert with extensive work in human genetics, heart muscle disease and cardiac imaging. Throughout his career, he has explored both the clinical and academic facets of the cardiovascular field in prestigious institutions such as Harvard University and Imperial College London. In 2012 and 2017, he was conferred the Singapore Translational Research (STaR) Investigator Award by the National Medical Research Council (NMRC), and appointed Professor at Duke-NUS and senior consultant at the National Heart Centre Singapore (NHCS). Professor Cook was also awarded the American Heart Association Council on Genomic and Precision Medicine (GPM) Medal of Honour in 2017, an international award which recognises scientists who are world leaders in the field of genomic and precision medicine, and whose work has transformed the field.

As director of the Cardiovascular & Metabolic Disorders (CVMD) Signature Research Programme at Duke-NUS and the NHRIS, Professor Cook heads a cross-disciplinary research team that seeks to identify new genes and pathways for heart disease, for better therapeutic targets. His work in Singapore is particularly important, as Asians have a different genetic makeup from Caucasian populations, the latter of which is more commonly the subject of research focus overseas. It is important to understand the Asian genetic makeup as this impacts the prevalence and course of several cardiovascular disorders.

In his capacity as clinician mentor at Duke-NUS, Professor Cook also supports and mentors budding clinician-scientists who wish to embark on both clinical practice and research work, helping to grow the critical mass of clinician-scientists in Singapore who act as a translational bridge between upstream research and downstream clinical practice.

About one in 250 people worldwide suffers from a heart condition called dilated cardiomyopathy, where the heart muscle becomes weakened, stretched and unable to pump blood efficiently, potentially leading to abnormal heart rhythms, heart failure and even sudden death. In 2012, Professor Cook and his collaborators discovered that mutations in titin, the biggest human gene, cause heart muscle weakness in Caucasian populations. Professor Cook went on to lead a genetic study in Singapore to find out whether mutations in the same gene are responsible for the illness in Asians as well. The titin mutation was found to be responsible for up to a quarter of cases in Singaporean-Asian populations.

This discovery catalysed the development of a sequencing assay which, in collaboration with genomics company Illumina, Professor Cook and his group translated into a commercially-available, next-generation test kit, with customized software to aid in the interpretation of genetic results. Marketed as the TruSight Cardio Sequencing Kit, the assay is used to screen many thousands of patients each year, for various gene mutations linked to inherited heart conditions, in laboratories around the world.

Efforts to discover the genetic causes of inherited heart conditions — much like the search for a genetic diagnosis in other diseases — have been stymied by the limitations of tests focused on single genes, or just a handful of targets. TruSight Cardio, however has a high depth and uniformity of coverage for its price point – the assay tests for 174 genes known to be associated with 17 cardiac conditions, including cardiomyopathies, arrhythmias, and aortopathies, covering all causal variants in these genes with a demonstrated link to inherited cardiac conditions. These genes were selected by researchers at the NHCS and Imperial College of London, and include those known to be associated with hereditary heart disease, as well as emerging genes found in academic literature in the field.

For his outstanding research in human genetics and cardiovascular disease, which have impacted the field of precision medicine, and resulted in the development of tools for genetic sequencing now used in clinical practice worldwide, Professor Cook is awarded the 2018 President's Technology Award.