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Probing gene link to heart disease

Study to focus on dilated cardiomyopathy in Asians

By FENG ZENGKUN

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.

It can lead to abnormal heart rhythms, heart failure and even sudden death.

Now, a group of researchers in Singapore has started a genetic study to find out whether the mutation of a gene called titin – linked to the disease in Caucasians – could be responsible for the illness in Asians as well.

The scientists also plan to examine about 60 other genes that have been associated with the ailment, and will sequence the patients' DNA to find out if there are other possible genetic causes.

If successful, the findings could be used to identify people with a higher risk of developing dilated cardiomyopathy, so they can keep an eye on their health. Patients could also get more personalised medicine to combat it.

Prof Stuart Cook, who discovered the titin mutation in Caucasian patients, is leading the team here in the Asian research.

While there is currently no cure for the disease, drugs can slow its progress. Patients who no longer respond to medication can opt for heart transplants or mechanical heart-assist devices, which are artificial heart pumps.

The team from the National Heart Research Institute Singapore has so far recruited about 180 patients. The institute, the National Heart Centre Singapore's (NHCS) research arm, works with the Duke-NUS Graduate Medical School.

The scientists plan to get more people into the study, including patients as well as healthy people to act as a control group.

Professor Stuart Cook, the Tanoto Foundation professor in cardiovascular medicine at the SingHealth Duke-NUS Academic Medical Centre, who is leading the study, discovered titin's link to the disease among Caucasians.

In his other role as a professor of clinical and molecular cardiology at Imperial College London, he led a multinational study, including researchers from Britain, the United States, Germany and Australia, to look into the disease's genetic causes.

After examining more than 5,000 patients and healthy people, the team found that the titin mutation is responsible for about a quarter of cases. Other scientists had found genes that accounted for about another 25 per cent.

Prof Cook said the research, which was published in scientific journal Science Translational Medicine last month, showed that the position of the mutation along the titin gene's DNA sequence also mattered.

The nearer the mutation is to the end of the sequence, the worse the patient's condition becomes.

"If you have the mutated gene, there is a 50 per cent chance you will pass it to your child," he said, adding that patients should test their children for the mutation.

Healthy people who want to volunteer for the local study can call the NHCS Biobank on 9159-7029, Mondays to Fridays from 8.30am to 5.30pm, except on public holidays.

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