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# Study finds genetic predisposition to certain conditions in ethnic groups

## A better understanding of the risk of getting a disease can boost preventive care efforts

**Zhaki Abdullah**

A new study covering about 10,000 people in Singapore has identified genetic variations which predispose people of different ethnicities here to certain medical conditions and adverse drug responses.

The upshot is that a better understanding of the genetic risk of getting a disease can enhance the preventive care strategy under the Republic's Healthier SG initiative that will be rolled out later in 2023.

The study found that mutations leading to familial hypercholesterolemia – a genetic condition in which high cholesterol levels are passed down in families – were more common among Chinese people.

The mutations were found in 1.05 per cent of Chinese participants, compared with 0.15 per cent of Indians and 0.25 per cent of Malays.

And while there were no differences in cancer predisposition across the races overall, genetic variants in some genes that predispose carriers to hereditary breast and ovarian cancer were seen in one in 110 Malays, one in 160 Indians and one in 160 Chinese.

The study, published in scientific journal Nature Communications in November 2022, built on another study published in Nature Genetics in January 2023, where researchers from the Genome Institute of Singapore mapped a genomic reference database of almost 10,000 Singaporeans.

Participants comprised ethnic Chinese, Malays and Indians with a median age of 47. Of these participants, 57.3 per cent were female.

This database revealed millions of genetic variants previously undetected in studies of Western populations, said Precision Health Research, Singapore (Precise). It coordinates efforts under the country's 10-year National Precision Medicine strategy.

The study published in Nature Communications also identified genetic variations that can predict adverse reactions to treatments such as anti-epileptic medications and chemotherapy.

It reported that 26.8 per cent of participants carried a genetic variant that raises the risk of life-threatening side effects of at least one medication.

Knowing if someone carries such a genetic variant can help reduce side effects when these specific drugs are prescribed, Precise said.

The study, which aims to provide early insights for SG100k, a project to map the DNA and the complete set of genes of 100,000 Singaporeans, was led by researchers from Precise and institutions such as the Agency for Science Technology and Research and the Duke-NUS Medical School.

The study is needed because while Asians make up 60 per cent of the world's population, they are underrepresented in current genomic studies, said Precise executive director Patrick Tan, adding that this is especially so for South-east Asians.

EARLY DETECTION HELPS

**If we can identify individuals who are genetically predisposed to develop certain diseases, then there is actually a lot that can be done to prevent them.**

**ASSISTANT PROFESSOR LIM WENG KHONG**, the study's lead principal investigator. He is from the cancer and stem cell biology programme at Duke-NUS Medical School.

Precise noted that this under-representation increases the risk of misdiagnosis or mistreatment by healthcare professionals unfamiliar with managing genetic disorders in non-European populations.

While earlier studies pointed to the prevalence of certain diseases among different races, it was not obvious whether this was due to factors such as diet, lifestyle or socio-economic status, the study's lead principal investigator Lim Weng Khong said.

"Now we can say with a higher degree of confidence that these are driven by genetics," added Assistant Professor Lim, who is from the cancer and stem cell biology programme at Duke-NUS Medical School.

He noted that data from the study suggests that some 3.4 per cent of Singapore's population are at risk of a genetic disease.

While this figure may not seem very high, at a national level, this could translate to "a very large number", he said.

"If we can identify individuals who are genetically predisposed to develop certain diseases, then there is actually a lot that can be done to prevent them," he noted.

Dr Saumya Shekhar Jamuar, one of the study's principal investigators, said an improved understanding of genetic diseases in the population will benefit couples planning to have children.

"For instance, they can opt for screening to identify any potential risks of passing severe genetic diseases to their offspring and take proactive measures to mitigate those risks before starting a family," added the genetics services senior consultant at KK Women's and Children's Hospital.

One participant, 41-year-old engineer Lim Swee Guan, discovered he had familial hypercholesterolemia after undergoing genetic testing in 2019. The condition makes him eight times more likely to have a stroke or heart attack.

Mr Lim was then prescribed statins, which lower cholesterol, and was advised to make lifestyle changes, such as cutting back on fried and fatty foods.

His two older children, aged 15 and 13, were found to have the condition and have been prescribed statins. His younger children, aged six and nine, are also highly likely to have the condition and are awaiting the results of further tests.

"I am thankful that my high cholesterol was detected early and I was able to take action proactively," he said.

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