

## Singapore scientists uncover genetic landscape of distinct breast tumors



Senetic landscape of fibroepithelial tumours, a distinct breast tumor group. Credit: SingHealth Duke-NUS Academic Medical Centre

A team from the SingHealth Duke-NUS Academic Medical Centre, comprising scientists and clinicians from the National Cancer Centre Singapore (NCCS), Singapore General Hospital (SGH) and Duke-NUS Graduate Medical School, has uncovered the genetic landscape of a distinct breast tumour group called fibroepithelial tumours.

Their study identified the culprit genes behind the formation and progression of these tumours, potentially improving the diagnostic accuracy of <u>breast cancer</u> and enhancing clinical intervention for patients with such tumours. The study's findings have been published in the prestigious journal *Nature Genetics*.



Fibroepithelial tumours are a distinct breast <u>tumour</u> group which includes two tumour types - fibroadenomas and phyllodes tumours. Fibroadenomas are the most common benign breast tumours in women of reproductive age, with thousands of women in Singapore and millions worldwide estimated to be diagnosed with fibroadenomas every year. Only last year, the team identified novel MED12 mutations in the majority of fibroadenomas, a finding that was also reported in *Nature Genetics* and has since attracted attention and interest worldwide. Now, they have charted the genetic landscape for fibroadenomas as well as phyllodes tumours, another subtype of fibroepithelial tumours that have both benign and cancerous forms, the latter accounting for about two per cent of breast cancers in Singapore.

"Breast cancer is the leading cancer affecting women in Singapore. By shedding light on the genetic landscape of fibroepithelial breast tumours, we can delve deeper into how they are formed, their possible progression into cancer, and determine how to manage them," said Professor Teh Bin Tean, Deputy Director (Research), NCCS, a co-principal investigator (PI) of the study.

Currently, distinguishing fibroadenomas from phyllodes tumours can be challenging to clinicians. The tumours can also progress from benign into malignant forms, and recur even after surgical removal. A deeper understanding into their formation and progression is thus important for the accurate diagnoses and treatment of such tumours, and for breast cancer care.

The team studied 100 fibroepithelial tumours and uncovered the genes responsible for their formation and progression. Annex A illustrates the findings of their study.

## **Impact on patients**



Understanding the genetic landscape of fibroepithelial breast tumours allows doctors to improve the diagnostic accuracy of breast tumour assessment.

"In pathology, we see challenging cases where fibroadenomas and phyllodes tumours are not clearly distinguishable. The discovery of their causative genes brings hope that we can eventually test for the presence of culprit genes to identify these breast tumour types accurately. Patients can then receive appropriate clinical intervention in a timely manner," said Professor Tan Puay Hoon, Head and Senior Consultant, Department of Pathology, SGH and co-PI of the study.

Professor Steve Rozen from Duke-NUS' Cancer and Stem Cell Biology Programme and co-PI of the study further explained, "If gene mutations in a patient's breast tumour suggest it may progress from a benign to cancerous form, doctors can then advise patients to have it completely surgically removed to prevent cancer occurrence."

The findings also provide candidate therapeutic targets for fibroepithelial tumours, paving the way for alternative treatment options. Currently, there is no effective therapy for phyllodes tumours apart from surgery.

"This discovery unleashes tremendous potential for the treatment of fibroepithelial breast tumours. Other than surgery, drugs targeting the tumours' gene pathway could help to treat them or prevent recurrence, which is not uncommon among patients with these tumours," said Prof Patrick Tan from Duke-NUS' Cancer and Stem Cell Biology Programme and co-PI of the study.

Recently, Professors Teh Bin Tean, Patrick Tan and Steve Rozen were conferred the 2015 President's Science Award (PSA) for their outstanding translational research work in Asian <u>cancer</u> genomics, which



includes the team's study on <u>breast</u> tumours. The PSA is one of the highest honours bestowed on exceptional researchers in Singapore for their outstanding contributions to research.

## Provided by SingHealth Duke-NUS Academic Medical Centre

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