Breast cancer's deadliness is influenced by genes

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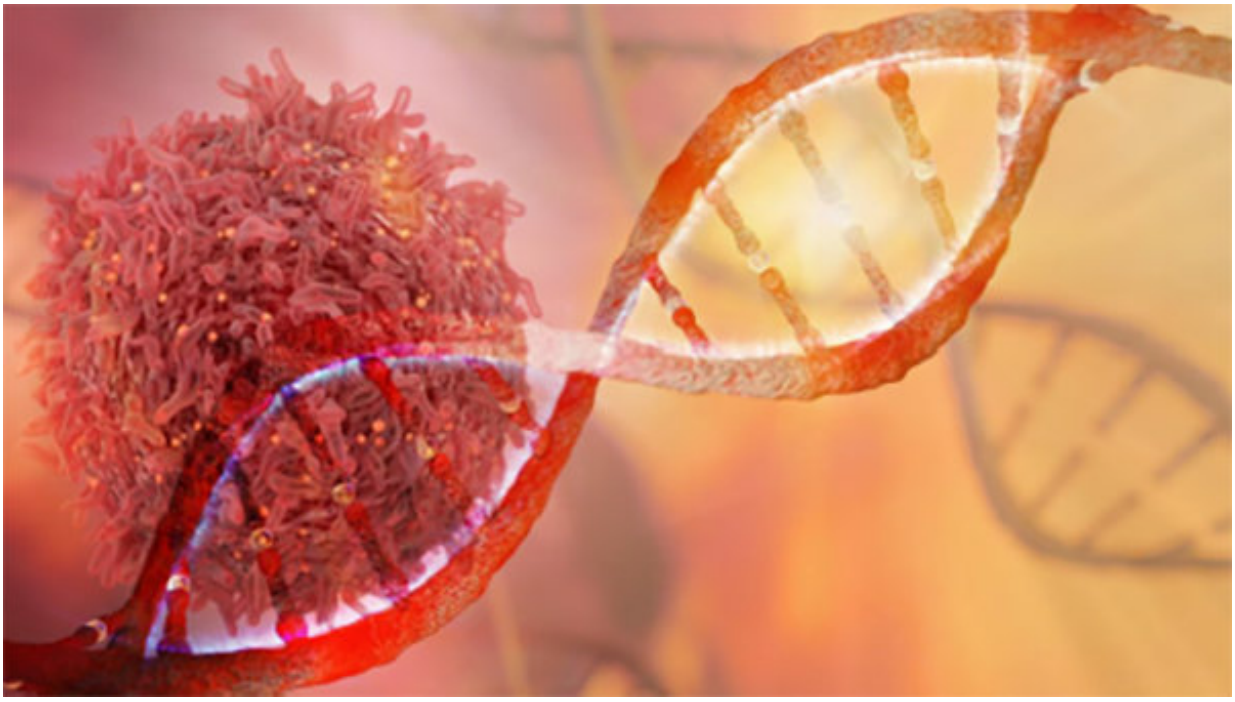

Genes can tell which breast cancers are deadly. Credit: Agency for Science, Technology and Research (A*STAR), Singapore

An international team of scientists have confirmed that both rare and common mutations of our genes are linked to the development of breast cancer, including tumour characteristics and corresponding deadliness. Interestingly, they discovered that the rare mutations were linked to more aggressive tumours, while the common mutations were associated

with less aggressive tumour characteristics.

The study was led by Dr. Li Jingmei, Senior Research Scientist at the Agency for Science, Technology and Research's (A*STAR) Genome Institute of Singapore (GIS), together with a team at Karolinska Institutet in Stockholm, Sweden. The results were published in the peer-reviewed journal *Cancer Research* on 1 November 2018.

In 2013, breast cancer's link to genetics was thrown into the limelight when celebrity actress Angelina Jolie made headlines with her decision to undergo a preventive double mastectomy. Her reason: an 87% risk of breast cancer due to what she believed were "faulty genes" – BRCA gene mutations inherited from her mother's side of the family. Fortunately, very few women in the general population carry the BRCA gene mutation – approximately one in 400 women. However the question remains as to why women who are not BRCA carriers are still at risk of contracting breast cancer.

Dr. Li's study noted that both rare and common genetic mutations are associated with different types of breast cancer. She also compared the effects of both types of genetic mutation on cancer tumours' characteristics, survival and modes of detection. Her study discovered that women with rare genetic mutations, compared to those with common genetic mutations, are more likely to develop more aggressive cancer tumours and succumb to the disease. They are also less likely to benefit from routine mammography screening as they tend to develop cancer during the screening interval.

Dr. Li said, "Angelina Jolie's decision to undergo double mastectomy has greatly increased the public's awareness of genetic testing. However, not all genetic mutations are equally deadly. By identifying the type of genetic mutation, we can better predict what kind of breast cancer the patient will develop."

GIS' Executive Director, Professor Ng Huck Hui said, "Most research studies focus solely on identifying the genetic mutations that lead to cancer. GIS took a step further and examined how these genetic mutations, both rare and common types, affect the disease progression."

A successful long-standing collaboration exists between Sweden and Singapore in the fight against breast cancer. "This study is yet another example of how clinical and genetics expertise from different parts of the world are brought together. A deeper understanding of how inherited genetic variants affect breast cancer biology could have profound implications for clinical management and future screening practices," said Professor Kamila Czene, Professor in Cancer Epidemiology and Deputy Chair at the Department of Medical Epidemiology and Biostatistics at Karolinska Institutet.

More information: Jingmei Li et al. Differential Burden of Rare and Common Variants on Tumor Characteristics, Survival, and Mode of Detection in Breast Cancer, *Cancer Research* (2018). DOI: 10.1158/0008-5472.CAN-18-1018

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