Breast cancer risk prediction technology using genomic classifier

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Meet the Inventors

Hwang, Shelley

Department

Surgery (Dept. & CRU)

Publication(s)

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External Link(s)

• From the lab of Eun-Sil Shelley Hwang
• Newly Developed Gene Classifier Identifies Risk of Breast Pre-Cancer Progression (Duke Health, 2022)

Unmet Need

The earliest form of breast cancer, ductal carcinoma in situ (DCIS), is diagnosed in almost 50,000 people in the U.S. each year. DCIS is considered noninvasive, as abnormal cells have not spread out of the milk duct. However, the treatment for DCIS is often aggressive to prevent the possible spread of these abnormal cells. Women may undergo breast-conserving surgery, radiation, or a mastectomy. In anywhere between 50%–75% of cases, these invasive treatments are excessive and unnecessary, putting patients through an often painful and traumatic process. There is a need for a suite of genetic markers that can differentiate whether a person is high risk or low risk for breast cancer progression to avoid unnecessary treatment.

Technology

Duke inventors have developed a genomic classifier that distinguishes high risk from low-risk breast precancer. This is intended to be used by doctors diagnosing and staging breast cancer after DCIS has been detected. Specifically, Duke inventors have developed and validated a classifier that is able to predict the risk of overall recurrence and invasive progression of DCIS. This has been demonstrated through the identification of RNA expression patterns in DCIS tissue that correlate with later stage invasive breast cancer (IBC). Researchers identified a Random Forest classifier that was trained using 812 genes, which successfully predict either no later event or an ipsilateral breast event (iBE), which is a recurrent in situ or invasive carcinoma, after surgical treatment. The classifier predicts both recurrence and invasive progression. Researchers also found the high-risk subset was characterized by the activation of certain pathways including cell proliferation, immune response, and metabolism. The classifier was validated on a second dataset, indicating that it performed well across datasets with diverse race/ethnicities, geographical locations, median years of diagnosis, and time to recurrence.

Advantages

- Prevents overtreatment of DCIS
- Helps women avoid invasive and potentially painful and traumatic treatment if not necessary
- Predictive power across diverse datasets that is not observed in commercially available prognostic tests for DCIS