



Biomarkers in breast cancer prevention pathways

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A wide range of emerging technologies are showing considerable potential for improving healthcare through enabling personalised approaches at different stages of care and disease prevention pathways.

Currently, health promotion and screening are the predominant vehicles for breast cancer prevention and, in the main, are targeted at large sub-sets of the population. This means they do not take into account detailed biological characterisation of individuals or provide tailored prevention pathways.

We know that the development of breast cancer is influenced by many different factors, which are likely to vary between individuals, and that breast cancer is not one disease but has many sub-types with different outcomes. Can we create prevention pathways that take these factors into consideration?

In this series of briefings we provide some perspectives on particular technology areas to stimulate discussion on the vision for the future. These perspectives have been developed together with experts in these fields with the aim of stimulating discussion about the 20 year horizon.

There is some way to go in gathering the scientific knowledge and technical capabilities sufficient to optimising the impact of these technologies. Nevertheless, it is important to reflect on their potential in order to visualise how prevention pathways could differ in the future and how health systems will need to adapt to move towards more personalised prevention pathways.

We'd like to thank Dr. Esther Lips, Dr. Pedro Estrela, Professor Brian Caulfield, and Professor Jonas Almeida for their work on these think pieces.

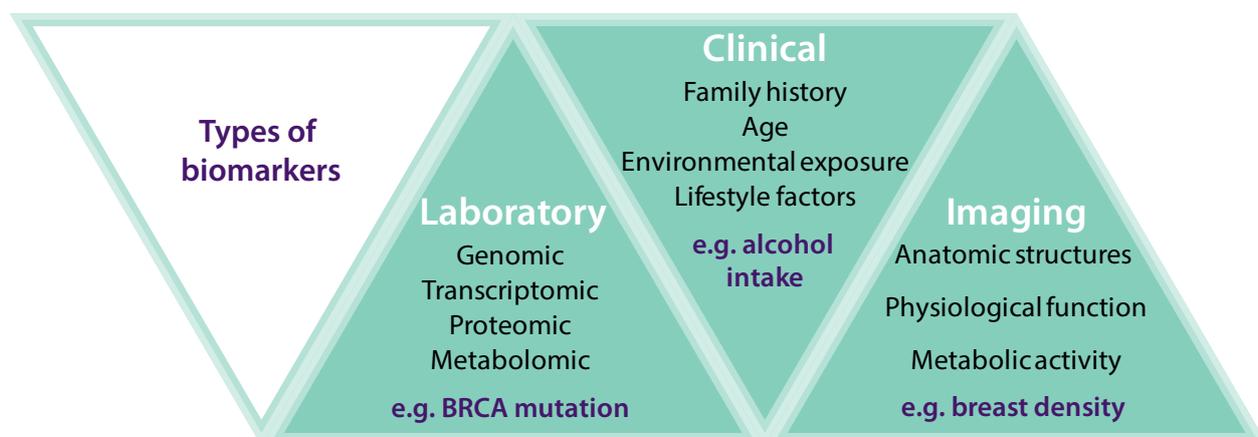
What is a biomarker?

A biomarker is anything that can be measured in an individual to predict incidence or outcome of a disease. They can consist of simple measures such as clinical features (e.g. age) to more complex factors such as biochemical or imaging markers.

What are they useful for?

Biomarkers have many uses in healthcare including identifying those at increased risk of disease, screening, diagnosis and guiding management and treatment of patients. To be useful in a clinical context, biomarkers need to be measurable with a high degree of accuracy and reproducibility, and must be clinically meaningful. Different biomarkers or combinations of biomarkers are relevant in different contexts.

Biomarkers are an important mechanism through which to stratify individuals to offer particular preventative options. They are already in use within current breast cancer prevention pathways. For example, age is one marker considered when offering routine mammography screening while genes such as *BRCA1/2* are used as markers to identify high risk individuals and inform decisions on more specific prevention pathways such as enhanced screening, chemoprophylaxis and mastectomy.



Emerging developments

Knowledge of biomarkers in relation to breast cancer is increasing, with research endeavours such as B-CAST examining various markers that may be used in assessing an individual's risk of developing the disease. These could be important in identifying the specific sub-type of breast cancer they may develop.

Other efforts include the PRECISION project, in which researchers are defining which ductal carcinoma *in situ* (DCIS); a lesion that sometimes develops into breast cancer will need treatment and those that will not, based on a deep characterization of various molecular and imaging markers.

Parallel developments are being made in the characterisation of novel and existing biomarkers. For example, there are currently attempts to identify blood-borne tumour markers which could be used for early disease detection. This includes techniques to isolate and analyse circulating tumour DNA, proteomic or metabolic signatures and volatile organic compounds in the breath that may be indicative of disease development.

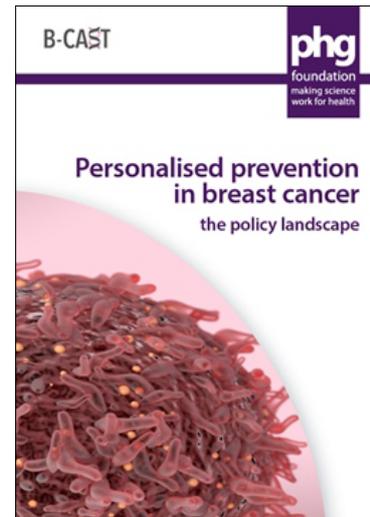
Meanwhile, improving progress in the range, utility and accuracy of biosensors and their incorporation into wearable and digital tools, could be used for precise collection of data on biomarkers which are more difficult to measure, such as lifestyle and environmental factors. Better knowledge of these factors could improve risk assessment processes and contribute to more refined preventative pathways.

Points for reflection

The future of personalised prevention for breast cancer will be very different in the light of further development and knowledge on biomarkers. How will this knowledge be used in prevention?

- Do we see a future in which women undergo yearly blood tests to identify and examine circulating tumour DNA rather than participating in mammographic screening?
- What are likely to be the different preventative pathways developed for different sub-types of breast cancer?
- Which biomarkers are likely to be incorporated into preventative pathways over the next 20 years?

A vision for the future starts with an understanding of the present. We have undertaken an analysis of current approaches to breast cancer prevention, and the discourse around personalised breast cancer prevention, with focus on primary and secondary prevention programmes.



About our work for B-CAST

As part of a European Commission (EC) funded research project, Breast Cancer Stratification (B-CAST), PHG Foundation is leading work on examining the potential for developing personalised prevention for breast cancer within national health systems. Building a better understanding of the influence of different risk factors on specific subtypes of cancer can, ultimately, help clinicians target treatments and prevention strategies to deliver improved health outcomes for patients.

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