

Prevent Breast Cancer SNPs 3 Research Project



About Prevent Breast Cancer

Predict. Prevent. Protect. You could call it our mantra. As the only UK charity entirely dedicated to the prediction and prevention of breast cancer, we're committed to freeing the world from the disease altogether. Unlike many cancer charities, we're focused on preventing, rather than curing. Promoting early diagnosis, screening and lifestyle changes, we believe we can stop the problem before it starts. And being situated at the only breast cancer prevention centre in the UK, we're right at the front line in the fight against the disease.

We **predict**, we **prevent**, we **protect**.

We predict – by identifying who is at risk of breast cancer

We prevent – by offering preventative interventions, to **stop** breast cancer before it starts

We protect – our goal is to **shield future generations** from breast cancer

Prevent Breast Cancer seeks to create a breast cancer free future for the next generation. We conduct ground-breaking research into the prediction and prevention of breast cancer.

We are a registered with the Charity Commission in England with the Registered Charity Number 1109839.

About the SNPs 3 Project

Project Dates

This is a two-year pilot study running from December 2018 – December 2020.

The Need

Breast cancer is the most common cancer in the UK, accounting for 15% of all cancer diagnoses. Across the UK, 150 individuals are diagnosed with breast cancer every day; tragically 32 of them will lose their fight against the disease. Unfortunately, these figures are rising and if trends continue, we can expect a 12% increase in diagnoses in just 10 years.

We can estimate a woman's chances of developing breast cancer by studying traditional risk factors such as family history of breast cancer, the age at which periods start and finish, and diet and lifestyle. Additional information on the density of breast tissue from mammograms further improves prediction.

Our previous research has shown that the information from common variations in our DNA called Single Nucleotide Polymorphisms (SNPs)¹ can be highly predictive of breast cancer on their own, but also further improve predictions alongside mammographic density and other risk factors. In 2010, 18 of these high-risk SNPs were discovered, and a further 150 SNPs have since been identified.

Development of Previous Research

This project is a direct follow on from two previous studies funded by Prevent Breast Cancer which aim to revolutionise the breast cancer screening process: SNPs 1 and SNPs 2.

SNPs 1 confirmed that a number of small DNA variations can increase breast cancer risk and in total 94 of the variations were discovered. These are gene changes within the general female population and are not confined only to those with the high-risk genes BRCA1 and BRCA2. So, analysing a woman's genes for SNPs can help predict risk of breast cancer even if they have no family history and they do not carry one of the major, well known breast cancer genes.

This study recruited 10,000 women with a high risk of developing breast cancer who took part in Prevent Breast Cancer's screening study PROCAS. Our researchers analysed their genes from saliva samples, comparing them against 18 of the identified SNPs. The study found that the SNPs acted as a predictor of which women would develop the disease.

SNPs 2 built on this progress by using all 94 identified SNPs to analyse 400 of the 800 women in the original study who developed breast cancer and comparing this sample to 600 women who did not.

So far the results from SNPs 2 have been overwhelming, indicating that these gene fragments are highly predictive of breast cancer, which mean that women theoretically could receive a more personalised risk prediction for breast cancer when attending routine screenings. Additionally, when SNPs are used alongside other risk prediction factors, our researchers have found that over-diagnosis of breast cancers may be reduced as well.

¹ SNPs - A **Single Nucleotide Polymorphism, (SNP)**, is a gene fragment discrepancy occurring commonly within a population (e.g. 1%) in which a single molecule in the DNA differs between human beings.

Our **SNPs 3** study will take this a step further. It will assess the feasibility of offering such a test alongside a personalised breast cancer estimate at a screening appointment, and whether these results could be provided in a timely fashion.

Project Summary

This is a large scale feasibility to offer women attending the NHS Breast Cancer Screening Programme at Oldham Integrated Care Centre in Greater Manchester a 'risk score' for developing breast cancer based on the analysis of their DNA. Our research team will collect and analyse saliva samples of consenting women for relevant SNPs. Approximately 1500 women attending the centre for a routine breast cancer screening will be offered this additional SNPs test. We estimate 1000 women will opt to complete the test and receive a risk score.

The lead researcher is Gareth Evans, Professor of Medical Genetics and Cancer Epidemiology at the University of Manchester, and Consultant in Medical Genetics and Cancer Epidemiology, Central Manchester Hospitals NHS Foundation Trust and The Christie NHS Foundation Trust.

Aims

The team will test whether it is feasible to offer a personalised risk score using DNA SNPs, and whether the results can be provided in a timely fashion (within six weeks of screening). Without trialling this development in a real-life situation, such a programme is unlikely to be incorporated into standard medical care. This project will ultimately result in a smarter, streamlined screening programme within the NHS which we believe could be offered in as little as 5 years' time.

Research Impact

This vital pilot study has the potential to change the screening process for breast cancer if rolled out nationally. If women are aware of their risk score for developing breast cancer, based partly on their DNA SNPs score, they may find that they are of a high risk of developing the disease. With this knowledge, they would be able to make their own personal choices around changing their lifestyle, the frequency of their check-ups, or choosing to take preventative medication such as tamoxifen, which lowers the chances of developing the disease.

This targeted personalised approach to breast cancer risk could allow many more breast cancers to be picked up through screening at a very early stage and ultimately lead to a lower death rate from breast cancer. This is because it could help prevent many cancers from starting or from spreading.

Costs Breakdown

SNPs 3 will run for two years and the costs break down as follows:

<u>Item</u>	<u>Cost</u>
Personnel costs	£29,111
Materials and Consumables	£70,889
TOTAL	£100,000

The costs include funding a full-time administrator to help with recruitment, answer queries and collect DNA samples. We also require materials including 1000 oragene kits, 1000 onco-arrays from Illumina and the costs for DNA extraction and sample processing.

Please note this is part of a wider study funded by the National Institute of Health Research (NIHR). The NIHR is funding the calculation of a risk score for women based on breast density and a selection of other risk factors. Prevent Breast Cancer is funding the SNPs element of the study. Adding information from SNPs raises the number of high-risk women from 4% to 6% of the population.

Monitoring and Evaluation

Our researchers are required to provide 6-month progress reports for their research so that Prevent Breast Cancer can monitor the work. These progress reports will be shared with the any funders as a means of reporting. Additionally, any publications in peer-reviewed journals resulting from this research will also be shared with any funders.

Contact Details

If you have any questions about this project, please contact **Vicki Wilkinson, Charity Manager** by emailing vicki@preventbreastcancer.org.uk or calling 0161 291 4400.