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A new model for genetic testing in cancer patients

A research programme that will lay the foundations for anyone with cancer to have access to genetic testing has been launched with £2.7 million funding from the Wellcome Trust.

Led by a team at The Institute of Cancer Research, London, in collaboration with The Royal Marsden, the Wellcome Trust Centre for Human Genetics and Illumina, the programme will implement a new patient pathway for cancer gene testing so that more genes can be tested in more people.

Mutations in some genes, known as cancer predisposition genes, greatly increase the likelihood that a person will get cancer. There are nearly 100 known cancer predisposition genes, but testing for them in the UK is currently very restricted.

Recent advances in methods for reading the genetic code, known as sequencing, mean that looking for gene mutations is now faster and more affordable than ever before. There is now an opportunity to transform cancer gene testing and to improve the health outcomes of many people with cancer and their families.

Professor Nazneen Rahman, lead investigator of the programme and Head of Genetics at the Institute of Cancer Research (ICR) and the Cancer Genetics Clinical Unit at The Royal Marsden, explains: "It is very important to know if a mutation in a person's genetic blueprint has caused their cancer. It allows more personalised treatment, so for example such people are often at risk of getting another cancer and may choose to have more comprehensive surgery, or may need different medicines, or extra monitoring.

"It also improves the information available for relatives about their own cancer risks. Sometimes a relative is found to also have an increased risk of cancer and screening or preventative measures can be employed. Just as frequently, testing provides the reassuring news that a relative is not at increased risk of cancer and does not need interventions."

Importantly, identifying cancer predisposition gene mutations provides an opportunity to prevent cancers. For example, about 1,000 women in the UK develop ovarian cancer each year because they have a gene mutation. If this was known before they got cancer, many may have chosen to have their ovaries removed by keyhole surgery after completing their families.

"Identifying people with cancer due to gene mutations and offering testing to their relatives is a very effective way of helping people at highest risk of cancer before they develop the disease," added Professor Rahman.

The Mainstreaming Cancer Genetics programme aims to develop the laboratory, analytical, interpretative and NHS clinical capabilities required to make cancer predisposition gene testing information routinely available in the clinic.

A new test has been developed in collaboration with Illumina that takes advantage of new sequencing methods. The test, known as the TruSight™ Cancer panel, can analyse 97 cancer predisposition genes within a few weeks for a few hundred pounds. It will be ready for use in the clinic in 2014.

However, generating the gene information is only the first-step. The data needs to be correctly analysed in order to identify any mutations. Then the effects of the mutations need to be correctly interpreted so doctors can make the appropriate clinical recommendations.

“Analysing genetic data and understanding how mutations actually affect a person is hugely complex. A real strength of this programme is that the analysts are working directly with the people in the lab generating the data and the doctors that need to use it,” said Professor Peter Donnelly, Head of the Wellcome Trust Centre for Human Genetics at the University of Oxford, who are working with the ICR scientists to develop the methods required to analyse the genetic data.

The programme also proposes a new model whereby the genetic testing can be done as a routine part of cancer treatment. Currently all cancer predisposition gene testing is done by geneticists, doctors who are specialised in diagnosing genetic conditions. In the new model it is proposed that testing in cancer patients will also be done by oncologists, doctors who are specialised in treating cancer. Testing in people without cancer would continue to be done only by geneticists.

“Many people with cancer are keen to have a gene test as soon as possible to help doctors plan the best treatment for them. The complex decision-making that people without cancer have to consider before having a test often doesn’t apply. We need a more flexible system that makes the testing process simpler when appropriate. This will allow many more people to benefit from gene testing,” said Professor Rahman.

The new model is being piloted, in the first instance, in women with breast or ovarian cancer at The Royal Marsden NHS Foundation Trust.

“Knowing whether or not a patient has a gene mutation is an important part of making personalised treatment plans. We want to be able to get the genetic information we need, when we need it. Patients are also increasingly aware of the value of genetic testing and more and more patients are requesting testing. This programme will help make genetic testing quicker and simpler and The Royal Marsden is excited to be leading on its clinical implementation,” said Professor Martin Gore, Medical Director of the Royal Marsden NHS Foundation Trust.

By the end of the three-year programme the team aim to have developed a ‘toolkit’ that will cover the full process involved in testing any cancer predisposition gene. This could then be used throughout the NHS, so that anyone with cancer can benefit.

Ted Bianco, Acting Director of the Wellcome Trust, said: “There is much expectation about the promise of new technologies in genetics contributing to a sea-change in medicine and this programme is a significant step on the road to making that a reality. With the outstanding team of clinical and academic scientists assembled to deliver the project, we are optimistic that the research will eventually make a real difference to people with cancer.”

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Notes to Editors

About the Wellcome Trust

The Wellcome Trust is a global charitable foundation dedicated to achieving extraordinary improvements in human and animal health. It supports the brightest minds in biomedical research and the medical humanities. The Trust's breadth of support includes public engagement, education and the application of research to improve health. It is independent of both political and commercial interests.

www.wellcome.ac.uk

About the Institute for Cancer Research

[The Institute of Cancer Research](http://www.icr.ac.uk), London, is one of the world's most influential cancer research institutes.

Scientists and clinicians at The Institute of Cancer Research (ICR) are working every day to make a real impact on cancer patients' lives. Through its unique partnership with The Royal Marsden NHS Foundation Trust and 'bench-to-bedside' approach, the ICR is able to create and deliver results in a way that other institutions cannot. Together the two organisations are rated in the top four cancer centres globally.

The ICR has an outstanding record of achievement dating back more than 100 years. It provided the first convincing evidence that DNA damage is the basic cause of cancer, laying the foundation for the now universally accepted idea that cancer is a genetic disease. Today it leads the world at isolating cancer-related genes and discovering new targeted drugs for personalised cancer treatment.

As a college of the University of London, the ICR provides postgraduate higher education of international distinction. It has charitable status and relies on support from partner organisations, charities and the general public.

The ICR's mission is to make the discoveries that defeat cancer.

www.icr.ac.uk

About The Royal Marsden

[The Royal Marsden](http://www.royalmarsden.nhs.uk) opened its doors in 1851 as the world's first hospital dedicated to cancer diagnosis, treatment, research and education.

Today, together with its academic partner, The Institute of Cancer Research (ICR), it is the largest and most comprehensive cancer centre in Europe treating over 50,000 patients every year. It is a centre of excellence with an international reputation for groundbreaking research and pioneering the very latest in cancer treatments and technologies. The Royal Marsden also provides community services in the London boroughs of Sutton and Merton and in June 2010, along with the ICR, the Trust launched a new academic partnership with Mount Vernon Cancer Centre in Middlesex.

Since 2004, the hospital's charity, The Royal Marsden Cancer Charity, has helped raise over £100 million to build theatres, diagnostic centres, and drug development units.

Prince William became President of The Royal Marsden in 2007, following a long royal connection with the hospital.

www.royalmarsden.nhs.uk