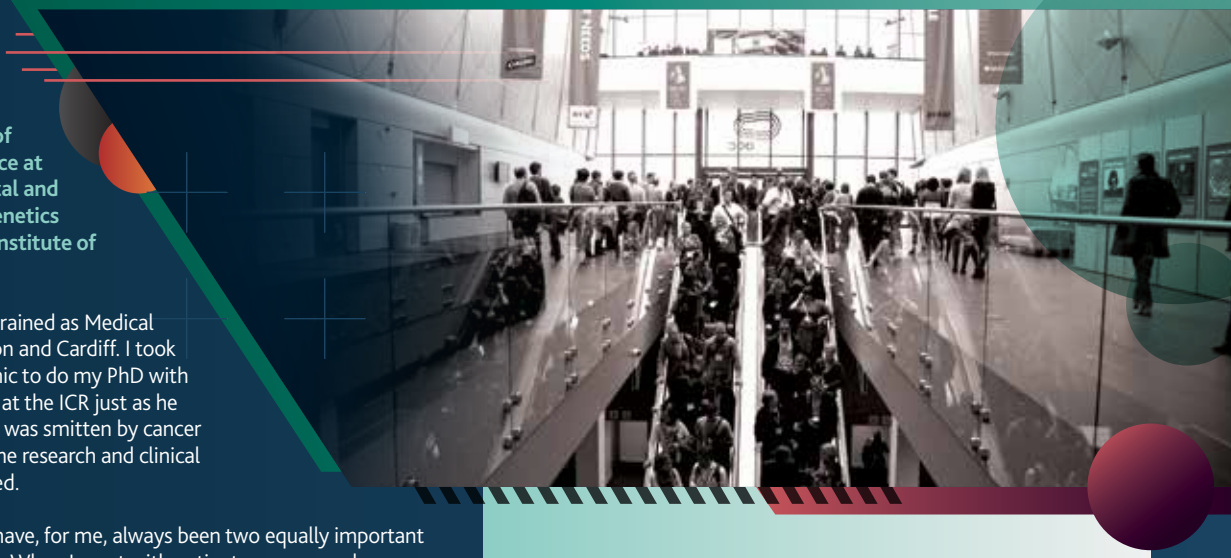
PROFESSOR NAZNEEN RAHMAN

Head of the Division of Genetics and Epidemiology
Institute of Cancer Research, UK

OVERCOMING CHALLENGES

In an enlightening talk, plenary speaker Professor Nazneen Rahman discussed genetic predisposition to cancer. Now, in an exclusive interview, she emphasises the equal importance she accords to research and clinical work, shares her thoughts on the necessity of making genome-related data accessible and reveals some of her team's most exciting studies underway in the quest to improve the clinical cancer landscape



How did you come to occupy the dual positions of Head of the Cancer Genetics Service at the Royal Marsden Hospital and Head of the Division of Genetics and Epidemiology at the Institute of Cancer Research (ICR)?

I qualified in Medicine and trained as Medical Geneticist at Oxford, London and Cardiff. I took some time out from the clinic to do my PhD with Mike Stratton when he was at the ICR just as he was discovering BRCA2 and was smitten by cancer genetics, particularly how the research and clinical aspects are so closely aligned.

Research and clinical work have, for me, always been two equally important parts of my professional life. When I meet with patients, my research knowledge helps me to provide information that is based on the best and most up-to-date scientific knowledge. When I am undertaking research, I always keep in mind the need to deliver impact for patient care, which greatly influences our research strategies.

Have you made any significant recent discoveries that you can mention through your two research foci, breast cancer genetics and childhood cancer genetics? Why did you choose these two themes initially?

We have recently identified two new genes that cause childhood cancers. We are currently completing those studies and hope to publish the results within the next few months.

It was chance that led me to these areas; when I started my PhD there were projects available in these areas that I started working on. In fact, though I wasn't aware of it at the time, there are a lot of links between them. One of the first cancer predisposition genes that I identified when I set up my own team, PALB2, causes both breast cancer and childhood cancer. We made the discoveries largely separately and ended up with two back-to-back papers in *Nature Genetics*.

At what stage are you in completing breast cancer exome analyses in 3,000 participants – the largest experiment of its kind in breast cancer?

We are a few weeks away from completing the exome sequencing, which includes 2,000 individuals with breast cancer and 1,000 controls and we are performing analyses of the data.

THE THREE MAIN AREAS OF THE MCG:

- Development of a test that rapidly and cost-effectively sequences cancer predisposition genes at high throughput and large volume
- Development of processes that analyse and interpret the sequencing data to provide clinical information to guide the management of patients and their relatives
- Development of a flexible, practical, robust patient-centred system that allows increased capacity and equity of access to genetic testing

We will be updating on progress of the project next year, hopefully with some positive findings!

Could you also offer a glimpse into another of your avenues of investigation, the Mainstreaming Cancer Genetics programme, which seeks to translate germline genetic testing of cancer genes into routine cancer patient care?

The Mainstreaming Cancer Genetics programme (MCG) aspires to develop the necessary lab, analytical and clinical infrastructure that will allow cancer predisposition gene testing to be available to anyone that could benefit.

Next-generation sequencing and genome-wide association studies have already transformed health and medical research. How do you foresee these technologies shaping clinical practice in the future?

These technologies deliver new insights into cancer predisposition mechanisms that give an explanation for why individuals develop cancer, can be used to optimise cancer treatment and can be used to identify individuals before they get cancer so that screening and preventative measures can be implemented.

In the future I anticipate that this type of information will be routinely integrated into clinical trials and clinical practice. There are challenges, particularly in relation to correctly interpreting and integrating the information, and it will be important that the data generated is shared and accessible for research.

Is it becoming increasingly important for medical researchers to work in closer collaboration with computer scientists, mathematicians and

IN PROFILE: PROFESSOR NAZNEEN RAHMAN

PROFESSOR NAZNEEN RAHMAN is both Head of the Division of Genetics and Epidemiology at The Institute of Cancer Research and Head of the Cancer Genetics Clinical Unit at The Royal Marsden NHS Foundation Trust.

Having qualified in medicine from the University of Oxford in 1991, Rahman undertook her general medical training in Oxford and London and, in 1999, completed a PhD in Molecular Genetics. Rahman leads two research groups seeking to identify genes that predispose to breast and/or childhood cancers. Rahman's research involves thousands of UK families and which has been successful in identifying new genes and genetic mechanisms responsible for cancer.

TALKING ABOUT CPGS

PROFESSOR RAHMAN gave a keynote lecture on cancer predisposition genes (CPGs). In addition to discussing her team's current search for predisposition genes in families affected by multiple cases of breast and ovarian cancer, Rahman discussed the potential benefits CPGs could bring to the future treatment of breast cancer, such as improved risk prediction and diagnosis as well as bespoke treatments.

THE MAINSTREAMING CANCER GENETICS PROGRAMME

USING NOVEL TECHNOLOGIES to develop a single test able to more rapidly and inexpensively analyse all genetic information relevant to cancer, the Programme's ultimate goal is to incorporate genetic texting into routine cancer patient care.

It is led by the Institute of Cancer Research in partnership with the Royal Marsden NHS Foundation Trust and is funded by the Wellcome Trust. It will run for three years – 2013-16.

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other physical scientists? Could you describe your multidisciplinary collaborations?

Our research as well as our clinical practice involves computer scientists, mathematicians and data processors. Estimating risks and analysing genetic data has always required that, but the needs are ever greater and increasingly relevant to other branches of cancer as handling big data becomes an everyday requirement.

Away from science, you are an accomplished jazz singer, with a following on SoundCloud. Do you find having an alternative presence beyond the health and medical sphere helps to break down boundaries and perceptions of scientists?

Until very recently my musical and scientific lives were completely separate. I am waiting to see what, if any, the (accidental) integration of my alter-egos has so check back with me in a year or so!

What are the main messages you hoped to convey in your plenary speech?

I hope that people will come away with a better understanding of the clinical and scientific value of researching cancer predisposition genes, optimism about the high promise of future clinical impact of the research and realism about the complexities and potential pitfalls of clinical implementation if injudiciously undertaken.

Do you believe conference attendance is still an integral part of a researcher's life, or is online contact becoming the preferred choice for many?

I think both have, and will continue to have, important value. I think it is excellent that more conferences offer opportunity for talks to be seen online so that more people can benefit from them.

When I am undertaking research, I always keep in mind the need to deliver impact for patient care, which greatly influences our research strategies