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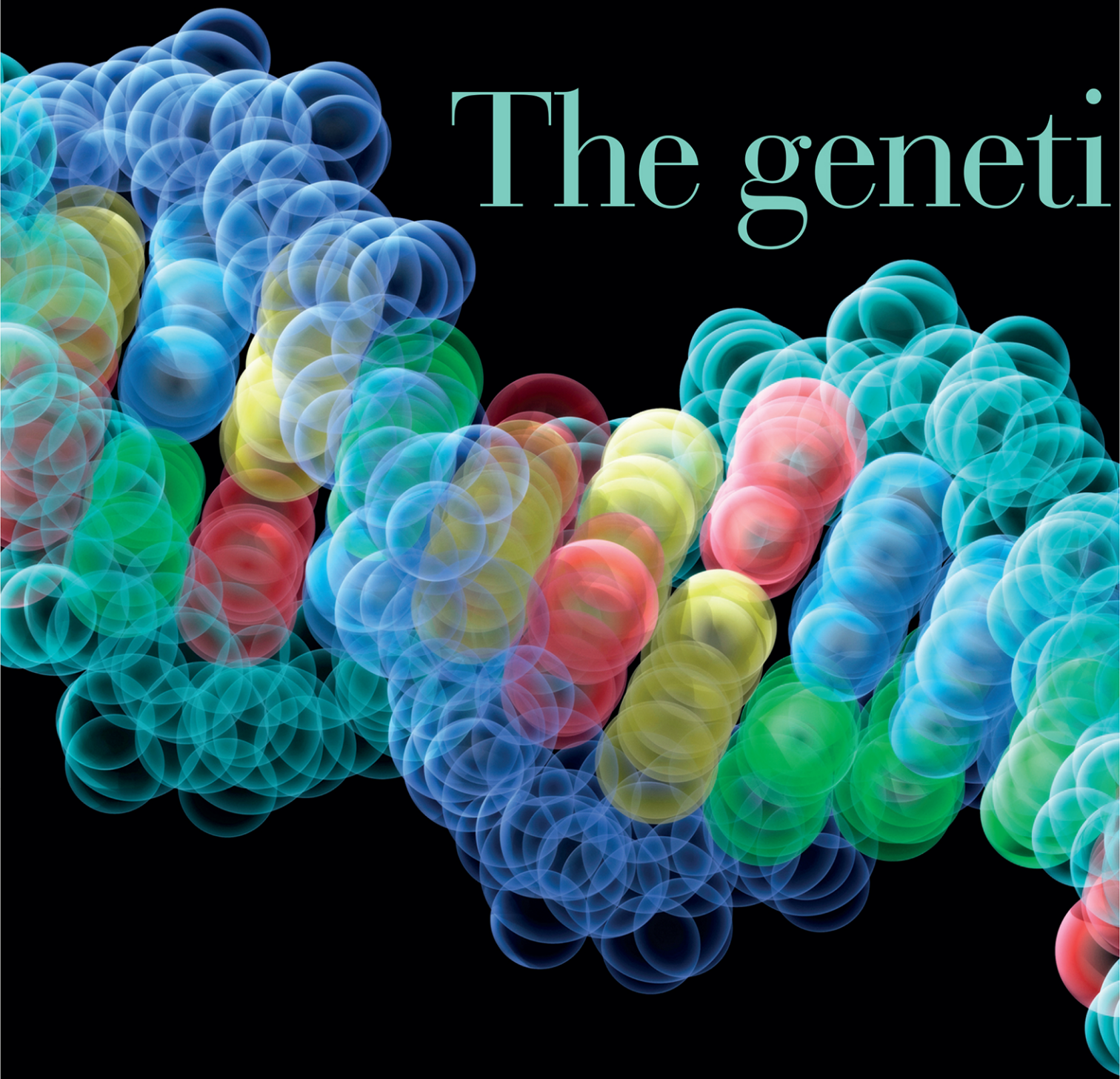

DNA can be examined to predict the likelihood of future illnesses
MARTIN MCCARTHY/GETTY

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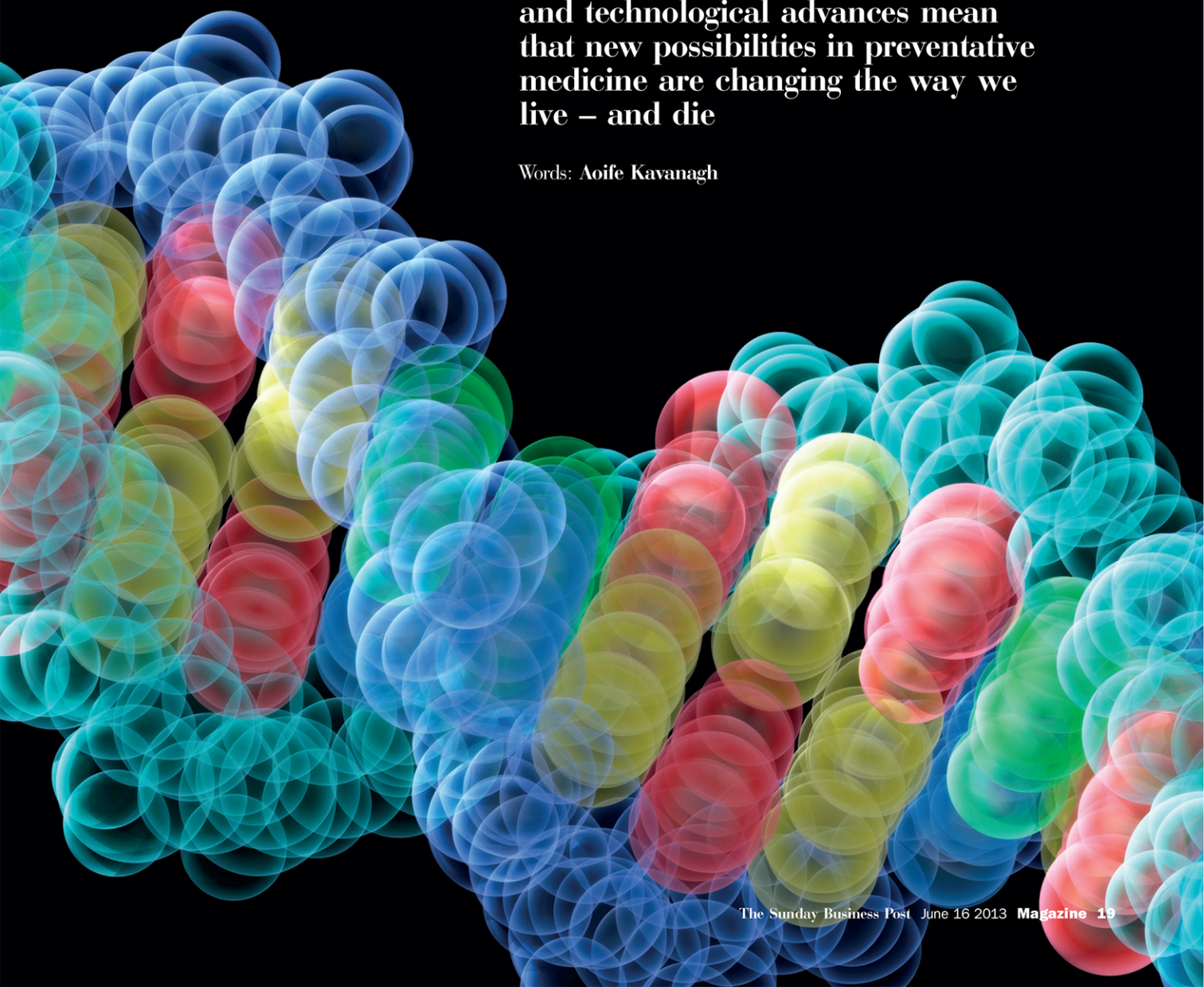
Humans have always been at the mercy of their genes, but medical and technological advances mean that new possibilities in preventative medicine are changing the way we live – and die

Words: **Aoife Kavanagh**



Professor Andrew Green, director and consultant geneticist, National Centre for Medical Genetics in Crumlin, Dublin MAURA HICKEY

Imagine a future where the extraordinary advances in medical genetics already realised in recent years are fully embraced. Where, from our very earliest days, each one of us will have access to a genetic file detailing the myriad medical conditions we are more, or less, likely to develop during our lives.

A world where parents who have been advised that their child carries genes putting them at risk of developing Alzheimer's disease will question whether they should allow their child to play rugby, where a blow to the head could exacerbate the problem.

Imagine a future where would-be parents, with no apparent problems conceiving, will choose IVF so they can screen for eggs that don't carry genes associated with particular genetic conditions. A future where a woman opts for preventative surgery because of what she views as an unacceptable risk of developing breast cancer.

Actress Angelina Jolie's description of her double mastectomy operation as "like a scene out of a science-fiction movie" aptly depicts the extraordinary world of medical genetics. Genetic research has opened the door to preventative medicine with endless potential and possibilities. The actress's proactive approach to tackling the problems in her genetic profile is proof of that. Her move to write publicly about that decision has made the rest of the world think about where genetic medicine might be taking us.

In Ireland, genetic medicine and testing is still largely focused on diseases with a clear genetic link, such as hereditary cancers, Huntington's disease or cystic fibrosis. For more than a decade, clinicians at the National Centre for Medical Genetics in Dublin have been providing the kind of genetic testing Jolie recently underwent. Advances in genetic medicine have been massively important for the kind of rare diseases that staff at the centre deal with on a daily basis, but when it comes to "highly complex conditions", such as heart disease and diabetes, then genetic testing can only go so far.

"For many common conditions like diabetes, MS and high blood pressure, there is a genetic component, but that is not the whole story," says Professor Andrew Green, medical director at the centre, which is based at Our Lady's Hospital for Sick Children in Crumlin.

The eureka moment that was the completion of the Human Genome Project more than a decade ago was only the beginning of a flood of new information about our genetic make-up, of which experts are still struggling to make sense.

Genetic medicine has its very own 'nurture versus nature' battle, which means that when it comes to many illnesses, environmental factors play a role in how a particular gene affects a particular individual and what the risks are for them.

In other words, just because you carry a gene linked to a particular condition doesn't mean you will develop that condition. To further complicate the picture, whether somebody develops a particular illness also depends on how various genes interact with each other.

"We all have about 20,000 genetic variations that potentially might be a problem, but we have no idea yet how to interpret them. It may take ten or 15 years of study of people who have particular gene variations before we can go back and say, 'Well we found this gene variation and this is what happened to that patient'," Green says.

That's not to pour cold water on how much has been learned about our genetic make-up and how that knowledge can be put to work. If you want to discover how susceptible you might be to a particular illness, then there are ways of finding out more.

Companies specialising in genetic testing have already opened for business in Ireland and Irish customers are availing of tests that map out their risk levels of developing common illnesses like heart disease or cancers.

EasyDNA opened in Ireland more than three years ago. The company offers a 'genetic predisposition test' to its Irish customers. For €295 easyDNA will analyse the customer's blood sample and provide them with a report detailing their level of risk – low, medium or high – of developing one or more of 25 medical conditions. The conditions include a variety of cancers, heart disease, diabetes and age-related illnesses.

Demand for the service has grown in the past 12 months, and the company reported a 'visual jump' in requests for information in the wake of Jolie's piece in the New York Times detailing her decision.

To date, most Irish customers who re-

quest the test do so because a close relative is suffering or may have died from an illness with a strong genetic link. That pattern is changing though, as more people opt for the test, regardless of whether there is evidence of a genetic condition in their family.

"People really want to protect themselves and take control of their health; there is a big push on that now with people watching what they eat, going to the gym and this is just one step further where it is a test tailored completely to you," says Martina Sullivan, international operations manager with easyDNA.

So how does a test like this work, and how accurate are the results?

Customers are given a home sample kit and are required to provide a blood sample, which is sent to a laboratory in Britain for testing. The procedure reviews a number of genetic markers by testing for a total of 70 'SNPs' or single nucleotide polymorphisms, which can indicate varying levels of risk of developing a particular illness or illnesses.

After 12-19 working days, customers receive a test report which lists 25 medical conditions and indicates their level of risk of developing any one of those illnesses. Included in the 25 conditions are MS, heart disease, breast, prostate and skin cancer, diabetes, Alzheimer's and various forms of arthritis.

The company stands over the accuracy of its reports, pointing out that some of its rivals only test for 25-30 SNPs, whereas testing for 70 apparently gives a better reading.

As Sullivan explains, however, you need to be prepared for the results. "I had the test done and found I am at very, very high risk of lung cancer. I was sure cancer would come up; I had it on both sides of my family so I was expecting it," she says. "Smoking is a big no-no for me and I try to avoid polluted areas, so there are things you can do to set your mind at ease."

Despite the alarming news, Sullivan says she felt empowered rather than paralysed by the results, though she accepts that, "if you have a tendency to be very worried when you find out about these things, then maybe this is not the test for you".

The illnesses covered by these kinds of pre-disposition tests all have a genetic component, but they are also affected by other factors that have nothing to do with our genetic make-up. Lifestyle, whether we exercise regularly and eat well, can also have a huge impact on whether or not we develop certain illnesses. Just because your results tell you that you are at high risk of developing a certain disease or illness doesn't mean that you ever will.

Nor does it mean, unfortunately, that when your risk of developing a particular illness is marked as 'low' that you will never develop that condition.

Given the uncertainty inherent in these genetic tests, Sullivan is anxious to point out that easyDNA recommends that anybody taking the test should discuss the process with a medical professional both before and after testing.

However, there is nothing to prevent anyone from having a genetic predisposition test, regardless of whether or not they are fully prepared to accept the results. It brings the principle of assuming control of your health and your future to a whole new level – but is it too much information, and are the services in place in Ireland to deal with a population that may soon be much more knowledgeable than ever before about their genetic make-up?

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Ireland has the worst record in Europe when it comes to staffing levels for genetic medicine. That is the stark reality presented by Professor Andrew Green, who struggles on a daily basis to meet the growing demands for services at the National Centre for Medical Genetics.

Green is one of just four full-time consultant medical geneticists in the country. Likewise, there are just six publicly-funded genetic counsellors (or more accurately, the whole-time equivalent of 5.9) to serve the needs of the entire population. There are also two whole-time equivalent charity-funded counsellors.

Based on comparable staffing levels in Britain, Ireland should have 32 full-time genetic counsellors.

Inevitably these dismal staffing levels mean long waiting lists and delays for patients. There are 1,500 people waiting to be seen by the four consultants based in Crumlin on Dublin's southside, and it takes at least 12 months to be seen routinely.

Genetic counsellors are seen as critical in terms of patient care. If a patient discovers they are predisposed to a particular illness or condition, it raises all sorts of questions and anxieties: should they inform all the members of their family; if they are parents, what does it mean for their children, or their plans for a family if they're not; is surgery an option, and so on.

Under-funded services also mean, however, that we are missing out on opportunities to put to good use all of the knowledge that research into genetic medicine has uncovered over the past 20 years.

"Doctors who are currently practising would not have had the education to deal with all the genetic information that is available. They haven't been trained to do it, so investment is needed not just for testing but for training," says Green.

In Britain and certain Scandinavian countries, where genetic medicine is increasingly a part of standard medical care, it is resulting in more accurate diagnosis of illnesses and much more accurate treatment for those conditions.

Professor David McConnell, professor of genetics at the Smurfit Institute of Genetics in Trinity College Dublin, has been absorbed by the potential of genetic medicine for more than 25 years, but is quick to point out the gaping divide between what we can now discover about our genetic make-up, and how the Irish health service is responding. "The knowledge and the technology has arrived, but the services are not there to back it up," he says.

McConnell recently added his voice to a campaign against the planned destruction, for data protection reasons, of 1,400 so-called 'heel prick tests' or Guthrie cards, dating back to 1984.

These tests – routine blood samples taken from new-borns between 1984 and 2002 – are used to screen for a small number of conditions with strong genetic links. However, they contain a wealth of information about a section of the Irish population which, if analysed, could potentially reveal much about our genetic predisposition to thousands of illnesses.

Campaigning by the Irish Heart Foundation and others resulted in a stay being

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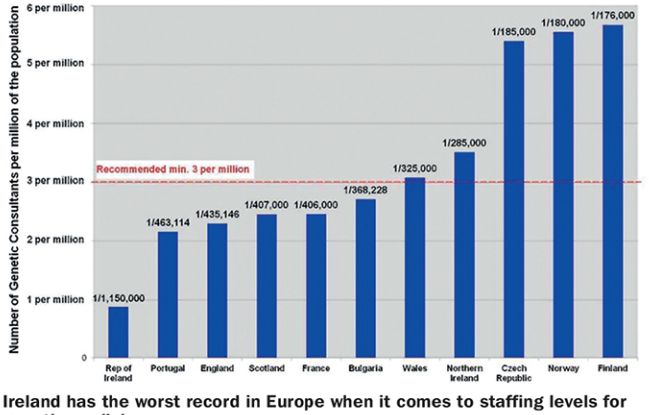
Angelina Jolie's description of her double mastectomy as 'like a scene out of a science fiction movie' aptly depicts the extraordinary world of medical genetics

Angelina Jolie pictured last week: the actress described her recent double mastectomy as 'like something out of a science fiction movie' GETTY



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