Keeping genomics in perspective

Reporting healthcare genomics in the real world — tips from *K.S. Jayaraman* on uncovering hype, tackling the issues and staying relevant.

A decade ago scientists unveiled a draft of the human genome sequence and then-US President Bill Clinton declared it would "revolutionise the diagnosis, prevention, and treatment of most, if not all, human diseases".

We may not be there yet but, after ten years and an explosive growth in research worldwide, genomics offers increasingly important stories for the scientific or medical journalist.

But reporting on genomics involves more than just writing about science and medicine. Issues like ethics, privacy and intellectual property can be equally challenging. So here are some tips on finding your way.

Genomics: a step up from genetics

A genome is the sum total of the genetic information encoded in an organism's DNA (or RNA in the case of viruses). Genomes include both genes — short lengths of DNA that code for specific proteins — and non-coding DNA sequences lying between genes.

While genetic research focuses on genes one at a time, genomics studies entire genomes and how the genes within them interact. It can cover all areas of biological research but this guide will focus on genomics for human health and medicine.

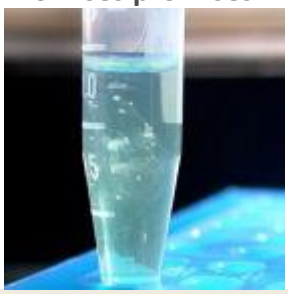
Even within one field, genomics can have many uses.

For example, you may be reporting on research that sequences a pathogen's genome to study how it causes human disease. Or genome-wide association studies may be investigating how natural variation in the human genome within and between populations affects the outcome and occurrence of disease.

Or maybe your story is about the young field of personalised medicine, which looks at an individual's genome to predict the risk of disease or to choose the most appropriate drugs for that person.

So you'll need a good grasp of what genomics is and does. The US National Human Genome Research Institute's factsheets about the science and ethics of genomics can be a useful starting point.

Promises promises



DNA in the lab: scientists can study entire genomes

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Genomics has the potential to improve health in developing countries. For the first time, researchers and doctors can look at entire genomes for signs of vulnerability to disease, or at the whole genome of pathogens to search for new drug and vaccine targets.

Certainly the advent of genomics kindled hopes of treatments for cancer, diabetes, Alzheimer's disease and heart disease. But there are many issues to resolve and this promise will likely take decades to materialise.

So don't oversell your story by raising false hope. And don't ignore the issues that must be overcome if research findings are eventually to become part of new treatments or techniques (such as diagnostic kits).

Ten years on from sequencing the draft human genome, the promised 'medical revolution' has not arrived even for developed countries (see a series of articles in *Nature* marking the tenth anniversary). And a recent poll of more than 1,000 life scientists found that most don't anticipate it materialising for decades.

Next time you report that scientists have discovered a range of genes associated with a particular disease, stop and ask what that really means.

Yes, they may contribute towards new tests and treatments. But ask researchers for estimates of when — and if — the research might be useful in the clinic. Then discuss with your editor how, and even whether, you should cover the story. The discovery might be news to scientists but is that who you're writing for? If not, the story may not yet be relevant to your audience.

Testing tests

Genome sequencing, the process that reveals the order of the nucleotides making up DNA, is becoming ever cheaper. Companies offering to sequence part, or even all, of your own genome have sprung up. They claim, among other things, that this kind of testing gives people an idea of their risk of developing certain diseases.

But answers are rarely clear cut. For example a genetic test for Huntington's chorea, a disease caused by a single genetic aberration, can give someone a 'no' answer or indicate a clear risk.

But other diseases, like heart disease or Alzheimer's disease, can involve many interacting genes. Genomic tests for these can only say whether you have a higher risk of developing the disease than the general population. What is someone to do if they discover you have a seven per cent above average risk of developing Alzheimer's disease?

Genetic-testing services are proliferating faster than regulators can catch up. In 1993, tests were available for about 100 diseases. By 2009, the number was almost 1,900 and even in countries like the United States the industry is largely unregulated. Can your country regulate such an industry?

In India there are at least a dozen labs offering genetic predisposition tests. But there is no agency to regulate the tests or accredit the labs. And, in the United States, the General Accounting Office concluded in July 2010 that some direct-to-consumer predictive genomic test results were "misleading and of little or no practical use".

But others say such tests let consumers take control of their own health. So get independent expert opinion on a new test or service before reporting it. Don't let the excitement of genomics turn you into the company's uncritical PR tool.

And if you are reporting on genetic testing for chronic diseases such as heart disease, remember that genes are only part of the story. They determine a predisposition to the disease but environmental factors, such as diet, often have an enormous impact.

Beyond sequencing

Now that whole genome sequencing is relatively inexpensive and fast, there seem to be press releases every week announcing that the 'codes' of pathogens and their vectors have been 'cracked'.

But sequences themselves are no longer guaranteed news — look instead for useful results that come from the sequence. How will Indian scientists use the *Mycobacterium tuberculosis* genome to develop new drugs, for example?

Ask researchers what the next steps are. What work needs to be put in to make this genomic data useful? What are those potential uses? Have they produced a genetic sequence (the order of all the nucleic acids of DNA) or a genetic map (a representation of genetic 'landmarks' such as genes)? You might find that a new sequence isn't actually newsworthy once you have the answers to these questions.

And if a researcher tells you it will take three years to identify the genetic basis of a disease from a sequence, check back three years later. This will help ensure that your reporting isn't simply a long list of new sequences.

Real life stories

Is a genomic divide going to grow between developed and developing countries?

UN Development Programme

There are many interesting and important tales to tell about genomics' implications for our societies. For example, will a 'genomic divide' develop between rich and poor countries and between richer and poorer communities within countries?

Can pharmaceutical companies — both Northern and Southern — be persuaded to use genomic techniques to find treatments and improve the diagnosis of neglected diseases? Or will health R&D remain skewed towards the needs of richer countries?

And should biotechnology companies be allowed to patent genes? Some think that removing patents from genes would open up R&D to poorer countries and researchers working to improve health in developing nations. Others think it would stall vital R&D as profit-seeking companies move into other areas. Try to stay on top of these debates.

Ask researchers whether they intend to help poor people. Will technologies arising from the research be affordable? Have the scientists thought about the intellectual property implications? Do they intend to develop the technology themselves or license it to a company who could price it out of reach?

If your country is investing in genomics, ask whether it is affordable — or is it at the cost of more traditional (perhaps more effective) approaches to public health?

Thailand has justified investment in genomics by pointing out that one-third of its population carry genes for the inherited blood disease thalassaemia. Thai health officials have welcomed genomics as a tool to diagnose and treat the disease.

India is setting up a National Institute of Biomedical Genomics, in Kolkata, with the aim of improving public health. But not everyone in India thinks genomics should be a priority. Many believe that basic preventative healthcare and primary care should take precedence.

Even if genetic testing may be justified in specific communities, genetic counselling and any necessary treatment arises from the diagnosis could be prohibitively expensive.

And that's the crux of the matter. Has your country the resources to use the fruits of genomic medicine? You could ask for the views of nongovernmental organisations working in communities where samples are collected for genomic study.

Don't forget the ethics

Also ask whether your country has the ethical infrastructure to deal with the myriad implications of genomic research.

Your country may make the ideal testing ground for genomics. International researchers may want to carry out a genome wide association study, for example looking at genetic markers associated with diabetes in different ethnic groups. To do so, they'll need samples from hundreds or thousands of people.

Does your country have privacy laws to cope with genomic information?
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Are proper informed consent procedures in place? Can privacy be guaranteed? Might genetic information be used by employers or insurers to discriminate against people?

What happens to genetic information given for research purposes? Ask the researchers and your authorities whether research has been cleared. Is confidentiality guaranteed for people contributing to such DNA databases in your country?

And will participants be told the results of the study? Might results be distressing if there is no treatment or lifestyle change to offer participants?

But despite all the provisos, remember the potential. Genomics is here to stay and as genome sequencing becomes even cheaper, the wealth of genomic data will continue to increase.

So, if you are starting your journalistic career now, you have plenty of time to understand genomics. And you are guaranteed many stories to report in this exciting field.

In the words of Francis Collins, director of the National Human Genome Research Institute, "the best is yet to come".

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