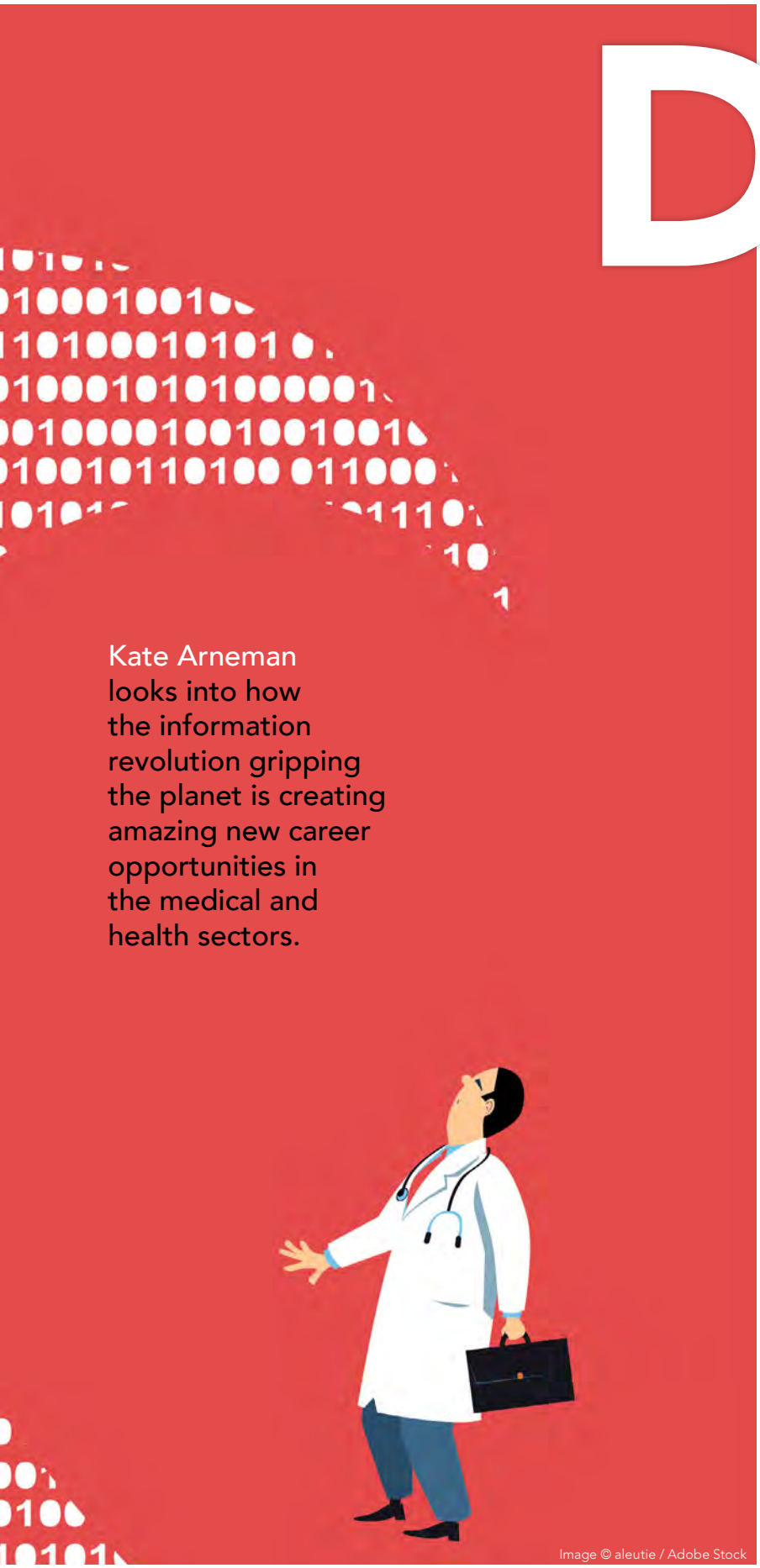
How big data is affecting your health!

Kate Arneman looks into how the information revolution gripping the planet is creating amazing new career opportunities in the medical and health sectors.



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DATA. IT SEEMS we just can't get enough of the stuff!

The global datasphere – a term for all data created, captured and replicated on our planet in any given year – is expanding exponentially. Experts predict it will reach 180 zettabytes (that's 180 trillion gigabytes) by 2025. Think of the difference in size between a Chupa Chups and a ten-pin bowling ball. In relative terms that's the difference between the amount of data generated in 2016 and the 2025 forecast.

It's only since 2000 that our digital existence has really taken off, with the ability to store data in the cloud made possible by wireless broadband and fast networks, and devices such as smartphones giving us instant access to information, social media and entertainment wherever we are.

Amidst the zettabytes, all kinds of data about our health is flowing in. There's medical data generated when we're treated by health professionals and data from participants in health and medical research. And then there's health-related data collected as we go about our lives on everything from our activity levels to our mental health. This comes from the devices we carry (smartphones), wear (fitness trackers, patches) and, increasingly, swallow or have implanted in our bodies (see Incredible sensors, p12).

Trawling social media for treasure

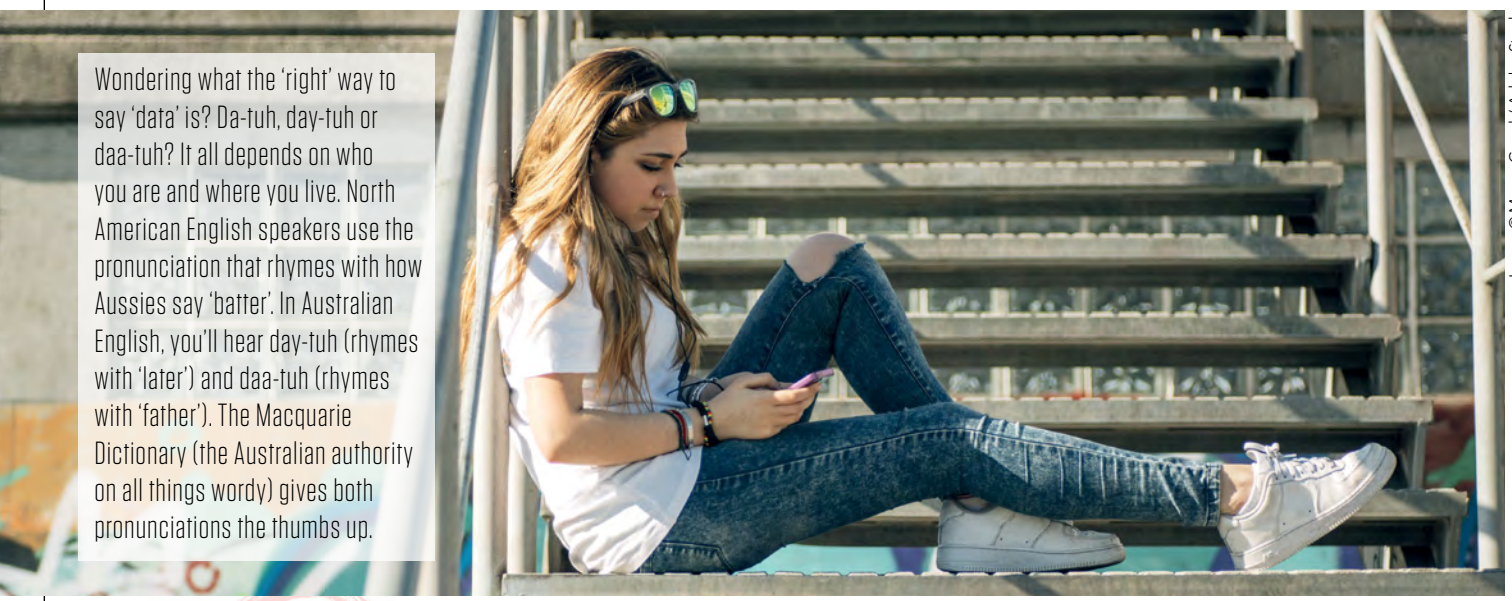
"They're addicted to their phones." "Social media is making teenagers more depressed and anxious." "It's destroying a generation!" Sound familiar? When we hear parents or media commentators talk about how young people use social media, the focus is almost exclusively on negative associations, like bullying, online predators and poorer social skills in 'the real world'. The true impact of growing up in a digital world is a little more complicated.

There's no denying there is a dark side to social media that can be harmful to the health of people with certain vulnerabilities. Online groups that promote eating disorders are just one example. But it's not all bad! Studies have found that for young people with mental illness, using moderated social networking sites has benefits such as increased self-esteem and more supportive relationships.

Social media can also play an important and positive role in managing chronic, or ongoing, health conditions like long-term pain. Online forums, Twitter, Facebook and Instagram are data goldmines for health informatics researchers like Dr Mark Merolli, Academic Director of Digital Health at Swinburne University.

"We know that there are more and more people turning to social media as a platform to manage their healthcare. But they are often quite rich sources of where people comment and log their health activity as well, in online support groups, et cetera," he explains.

Mark's research looks at how people interact with health-related digital technology and what motivates them to do so. "I've always felt that if we had a better understanding of why and how people engage with technology, we might be able to better tailor digital



Wondering what the 'right' way to say 'data' is? Da-tuh, day-tuh or daa-tuh? It all depends on who you are and where you live. North American English speakers use the pronunciation that rhymes with how Aussies say 'batter'. In Australian English, you'll hear day-tuh (rhymes with 'later') and daa-tuh (rhymes with 'father'). The Macquarie Dictionary (the Australian authority on all things wordy) gives both pronunciations the thumbs up.

health solutions and interventions and technologies to people," he says. "And, if we can better tailor them, we can have a better chance at improving people's health outcomes because we're following a more evidence-based approach."

Incredible sensors

From wearables to ingestibles to implantables: the next wave of innovative data-generating devices is going to transform healthcare at every stage – from diagnosis to treatment, rehabilitation and self-management.

Imagine you're one of many Australians who has a gut disorder, like irritable bowel syndrome or undiagnosed colon cancer. At the moment, the only way for your doctor to find out for sure what's going on in your gut is through invasive tests and surgeries. But what about if, instead of the indignity of providing poo samples or having the interior of your colon closely examined, you simply had to swallow a capsule the size of a vitamin pill? Luckily for hypothetical you, researchers from Monash University have not only imagined but developed and tested an ingestible sensor that detects and measures gases in the stomach in real time, sending the data to a smartphone. Not only is this a lot less embarrassing and uncomfortable than the alternatives, it's a more accurate diagnostic tool. The capsule's inventors hope the device will be available to the public by 2020.

Another ingestible tech breakthrough is the first 'digital' pill containing a sensor the size of a grain of sand (see p5). This medication, used to treat schizophrenia and other serious mental illnesses, was approved for use by the US Food and Drug Administration in November 2017. When it dissolves in the stomach, a tiny sensor inside the pill is activated on contact with stomach acids. It then sends a signal to another sensor in an adhesive patch worn externally on the person's stomach, with the ID number of the pill. The wearable sensor detects whether the wearer is sitting, moving or standing when they take their medication, then sends this information to a smartphone app via Bluetooth. In response, the app asks the person to rate their mood. If they choose to, patients can make this data available to their doctors or caregivers via an online platform.

Also in the works: a biodegradable implantable pressure sensor for monitoring chronic lung disease and brain swelling. And then there's a 'cancer detection system' that consists of implantable nanotube sensors and a device worn on the wrist that sends infrared light to the sensors and analyses what is reflected back.

The genomic promise

While implantable sensors give us access to valuable data about how well our bodies are functioning, geneticists are getting better and faster at extracting and analysing the data that's naturally 'embedded' in every cell of our bodies. Each of us has a unique set of genetic information called a genome, which is needed to build our bodies and allow us to grow and develop. The instructions for this huge and complex project are known as our genetic code, which is contained in a long molecule called DNA (deoxyribonucleic acid).

Genes are small sections of DNA, containing instructions for particular characteristics, like your height and eye colour. Everyone's DNA has a combination of four substances: adenine (A), thymine (T), cytosine (C) and guanine (G), which are variations of a type of molecule called a nucleotide. What makes everyone unique is the order – or sequence – in which the four nucleotides are arranged, forming a one-off code. If you read that code out, saying one letter every second for 24 hours a day, it would take a hundred years to get to the end of the 3.2 billion nucleotides in one human being!

Given the size and complexity of what they were dealing with, scientists were pretty chuffed when in 2003 they completed the sequencing of the human genome for the first time – even if it had taken them more than a decade to do it and cost US$3 billion. Far more exciting than the achievement itself were the possibilities it offered in terms of understanding, treating and, ultimately, preventing diseases with a genetic basis.

In February 2018, Rady Children's Institute for Genomic Medicine in San Diego, US, set a new Guinness World Record by sequencing a whole genome in 19 hours. This Institute is known for providing ultra-rapid diagnosis – considered to be five days or less – of rare genetic conditions. A group of Australian researchers are pioneering a similarly speedy service, made possible by next generation sequencing (NGS) techniques

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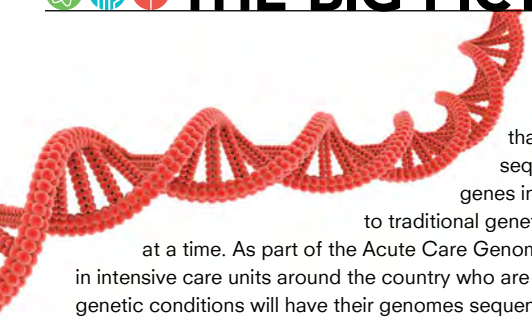


that allow the simultaneous sequencing of many or all genes in a genome, compared to traditional genetic testing of one gene at a time. As part of the Acute Care Genomics study, 250 babies in intensive care units around the country who are thought to have rare genetic conditions will have their genomes sequenced with the hope of identifying what is causing their symptoms.

"Time is absolutely precious when providing care to sick babies and children in intensive care units," says co-leader of the study, Associate Professor Zornitza Stark (below). "At the moment, turnaround times for genomic results can be up to six months, which is far too slow to help families and clinicians caring for children in intensive care," explains Zornitza, a geneticist with Murdoch Children's Research Institute. "We have already seen the benefits of early rare disease diagnosis locally, and will be extending testing across much of Australia, while providing clinical results in as little as five days."

Until very recently, most families of children with rare genetic conditions faced a long, exhausting and expensive road to diagnosis, full of hospital stays, invasive tests and uncertainties. The standard procedure has been to create a short list of genes that might be causing the problem and carry out traditional genetic testing on each gene, one at a time. Only then, if no answers had been found, would NGS testing be considered. An earlier study carried out by Zornitza and her team found that using NGS sooner rather than later could reduce the cost of each diagnosis by $9000, and save families years of waiting.

Although there is currently no treatment for most rare conditions, a diagnosis helps medical staff to deliver better care and in some cases prevent complications that can develop from the original condition.

Zornitza, who finished her medical degree two years before the Human Genome Project came to a close, says she had no way of knowing how far the field would advance during her career. "Not in a million years could I have imagined that today I would be sitting in the clinic ordering a patient's whole genome as a medical test," she says. "It's a very, very exciting area that is just going to explode. It's going to have multiple applications in medicine and there are just so many opportunities in this field."

Researchers like Zornitza work in large multidisciplinary teams to be able to deal with the complexity of the data they use."There are many opportunities at the moment for bioinformaticians in particular, laboratory scientists that are interested in genomics and genetic counsellors and also medical geneticists," she says. "It can suit a variety of interests ranging from how much you want to deal with data versus also dealing with patients and families."

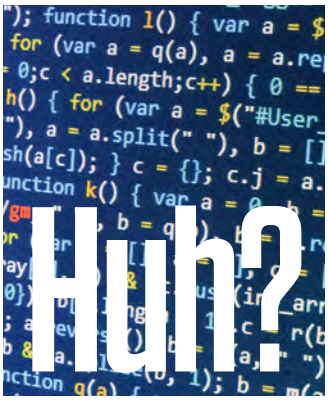
Case Study: The Empowered Patient

Health informatics researcher at Melbourne University Dr Mark Merolli began his career in health as a physiotherapist 12 years ago. The first iPhone was yet to be released and healthcare was almost exclusively offline.

He did have the occasional dedicated patient who would track their symptoms in a physical notebook and bring that to appointments in the clinic. "The odd savvy person might have had a print-out from Microsoft Excel," he recalls, laughing. "Fast forward a few years and I did find it quite fascinating that people would come in having googled stuff, having watched videos on YouTube or asked questions to their friends on Facebook."

In a relatively short space of time, he witnessed some big changes in how his patients interacted with him. "[Initially] patients would come in seeing you as the font of all knowledge and turning to you for professional advice wanting to know, 'What's wrong with me? How do I fix it?'," Mark says.

"There really was this shift in, A, the level of preparedness and information that people had before they came to the physio and, B, their expectations of what that relationship would be, in the sense that they were starting to want to take a more active role. The impact of that technology on patients impacted me - it was something I really took notice of."



Interested in a career that involves big data and health, and researching courses that can take you there? You'll need a few key definitions to make sense of the many technical terms you'll encounter.

BIG DATA: data that is collected or combined in such large quantities that it cannot be stored, analysed or used by traditional methods

BIOINFORMATICS: focuses on getting meaning out of the vast amounts of digital information produced by genetic sequencing and related technologies

BIOSTATISTICS: the application of statistics to scientific research in health-related fields, like medicine and public health

DATA MINING: a process used to explore already existing large data sets for patterns and relationships; uses tools drawn from statistics, machine learning and database systems

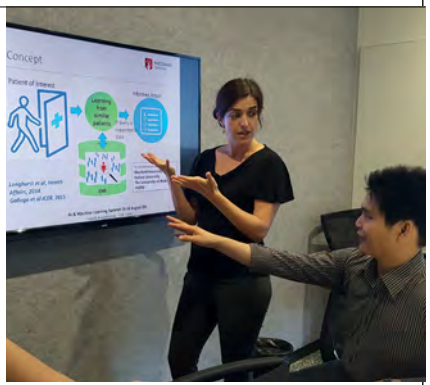
HEALTH INFORMATICS: covers the collection, analysis and movement of health information and data to support health care

Case Study: Patient Modelling

Associate Professor Blanca Gallego Luxan (at right) heads up the Health Analytics lab at Macquarie University's Australian Institute of Health Innovation. She's previously used her skills to model climate systems, the spread of disease in the community and more. But she's now focused on what makes hospitals tick. Her team uses data analysis to better understand patterns of patient safety in hospitals, and computational modelling to create predictive tools that support medical staff when making decisions about their patients.

"You have to look, not just at the mathematics, but also at the complexity of the human behaviour, human computer interaction; you need to understand the medical setting. That's why it's so multidisciplinary, this research," Blanca says.

In her view, that's one of the perks of the job, "You work with all types of people. I've worked with doctors, with nurses, with psychologists, with physicists, with mathematicians, with engineers, with computer scientists."



What, exactly, is big data?

There's no universally agreed upon definition for big data. But generally, it refers to data that is so vast and so disorganised that traditional methods of storing and managing it just won't cut it. (Sorry Excel, but this is way out of your league!) Ninety per cent of all digital data is unstructured and data scientists are in high demand to make sense of these massive messy data sets.

Drawing on techniques from computer science, mathematics and statistics, they're using what they find to answer curly questions and solve all sorts of problems.

The Centre for Big Data Research in Health at UNSW has a definition for health data science. It's "the science and art of generating data-driven solutions through comprehension of complex real-world health problems, employing critical thinking and analytics to derive knowledge from (big) data."

Data science can tackle real-world problems at the level of the healthcare system, such as hospital overcrowding, all the way down to the level of the individual, when it comes to preventing disease, for example.

Case Study: What's in a name?

In 2015, Melbourne-based couple Kate and Tim McMahon were given the life-altering news by clinical geneticist Dr Zornitza Stark that their three-year-old daughter, Olivia (right), had a genetic condition called Kleefstra Syndrome.

Children with the Kleefstra Syndrome experience seizures and heart problems and have delayed physical development and intellectual disabilities along with a range of other symptoms. There are only around 400 known cases of Kleefstra Syndrome worldwide.

"I'm sure for a lot of parents it would be the worst moment of their lives," Kate says. "But for us it was such a celebratory moment, obviously with a lot of mixed feelings, but…it had been three years of uncertainty."

The diagnosis was made possible by next generation genome sequencing techniques, which identified a mutation (change) in a gene associated with Kleefstra Syndrome. Most cases of the disease are caused by a gene deletion, which means the gene is missing from the person's genome. This deletion can be picked up relatively early in the diagnostic process using traditional genetic tests. But mutations can be far trickier to pinpoint, and it takes longer to do so.

"[It's as] if you were looking at a book and there was a stray full stop in the middle of a sentence. And that full stop just happened to be in the gene which would result in Kleefstra Syndrome," is how Kate describes it. Understanding the genetics behind her daughter's condition made a difference on many levels.

"It helped me a lot with acceptance to know that this is who Olivia is. It's just as if she had red hair or she was really tall – Kleefstra is literally part of her DNA and that's who we need to support and recognise," says Kate. "We want to help her as much as possible but that's who she is."

Before the diagnosis, when people would ask what was wrong with Olivia, Kate would jokingly say she had 'Olivia Syndrome'. It was life-changing to discover that there were other families going through similar challenges, including two in Victoria. "Through our own networks and Facebook (there's an amazing Kleefstra parents Facebook group) we've been able to learn a lot about what our future might look like with Olivia," Kate says. "That's been really empowering because even though not all of it is positive at least we're better prepared. We know what we might be able to expect and we can make future plans based on that."

One of the biggest decisions for the McMahons following the diagnosis was to have another child. "We were able to get tested ourselves. That meant that we knew that Olivia's syndrome, which is Kleefstra Syndrome, was just a random lottery 'win' for her. And it wasn't something that we've passed on to her. So that meant that we could consider expanding our family naturally." And that's exactly what they did. Olivia now has a little brother called William.

"It's been amazing for Olivia too to have that sibling experience because she's learning so much from him every single day. They've got such a beautiful relationship and it's a real joy as a parent to see that." The couple invited Dr Stark to William's baptism as a guest of honour. "We made note, in front of the congregation, that without her he wouldn't have been there. Everyone gave her a round of applause," Kate says, laughing. "That was a really special moment for us to acknowledge publicly the impact that she'd had directly on our lives. And the proof was in our arms, with William."

For more on Olivia's story:
youtube.com/watch?v=-tFWaJKSYyY