FOCAL POINT

PRODUCED IN PARTNERSHIP WITH THE UNIVERSITY OF TOKYO

CROWD-SOURCED HEALTHCARE

In the age of genomic big data, THE WORLDS OF MEDICINE AND IT are rapidly colliding.

In 2016, data-savvy doctors in Japan took just 10 minutes to identify why a patient was responding slowly to treatment for what they had believed was acute myeloid leukaemia.

By comparing the genetic profile of the patient's cancer to data from 20 million clinical cancer studies, the doctors at The University of Tokyo's Institute of Medical Science (IMSUT) showed she also had an exceedingly rare secondary form of blood cancer, requiring a different treatment regimen. They immediately changed tack.

Oncologists have long predicted that genomic data could one day transform cancer treatment. But even as genome sequencing technology prices have plunged, oncologists could realistically only personally have insight into a small number of specific genes, maybe 100 at most, according to Professor Satoru Miyano, director of the Human Genome Center at IMSUT, a leader in genomic medicine and rare oncology cases.

Even keeping on top of that many genes is a heavy burden, requiring the doctor to trawl databases and keep up with the scholarly literature, Miyano says. Making use of the human genome's tens of thousands of genes? "It's beyond one medical doctor's ability."

The rise of machine learning and artificial intelligence (AI) is changing that requirement and providing new hope for people with rare diseases. For the last few years, supercomputer-driven AI has become essential to IMSUT's haematological diagnoses, providing ever-more powerful insights based on largely US data, such as that which helped their patient in 2016.

However, the picture isn't yet complete. The patient has since relapsed, and without a clear explanation, Miyano

© KTSDESIGN/SCIENCE PHOTO LIBRARY/Getty

Satoru Miyano is the director of **HUMAN GENOME CENTER** at The Institute of Medical Science, The University of Tokyo.

"To understand such conundrums at a deeper level we need Japanese big data," says Miyano. "Overseas cancer data is helpful to make good, but not exact, diagnoses."

believes something specific to her genetic make-up might play a role. A person's genetic ancestry can affect responses to genetically targeted therapies. This is one area where missing pieces of the genomic medicine puzzle becomes evident.

"To understand such conundrums at a deeper level we need Japanese big data," says Miyano. "Overseas cancer data is helpful to make good, but not exact, diagnoses." For rare diseases, they need thousands of Japanese samples and for drug discovery insights, maybe as many as 10 million genomes.

Local genomic big data is limited in Japan and in many other parts of the world. A 2016 study of 35 million samples from genome-wide association studies, reported in *Nature*, found that 81 per cent were of European ancestry. The study's authors pointed out that patients of Asian and African ancestry are more likely to receive ambiguous or even incorrect test results from exome sequencing.

Private companies are rapidly helping to fill gaps in this data by providing direct-to-consumer genomic health information, genomic data to researchers, and developing powerful AI advances. Miyano says governments need to carefully consider how to encourage this activity. "I think regulation should be minimal, so that the technologies from IT companies can be best employed within medical and healthcare policies." In Japan, he says some genomic data could have a clinical impact especially quickly, due to Japan's relatively homogeneous gene pool. The key is gathering enough data.

Privacy laws being put in place in Japan will require delicately procured consent from individuals to use their data for research. It is here that consumer companies may have the most to offer, providing an element of engagement which allows them to compile large and rich datasets via voluntary contributions.

Illustrations of how this might work are already operating in the US. One of the most well-known examples is consumer genetic-testing and data firm 23andMe. Companies like this gather the data of millions of curious customers by providing information on everything from your probable caffeine metabolism to risk of late-onset Alzheimer's disease. Increasingly, such information is becoming of interest to big data researchers. Some 30 per cent of 23andMe's two million users have agreed to contribute their data to research beyond their personal genetic profile. These people have now helped researchers publish more than 80 papers, some filling out voluntary questionnaires, in addition to genetic data and samples.

Initially part-funded by Google, 23andMe moved from regulatory shut-down between 2013 and 2015, to, in April 2017, becoming the first FDA-approved company to distribute direct-to-consumer genetic health risk tests in the US. This shot it into billion-dollar market-value territory, where it joins the ranks of another half-dozen genetic data companies and myriad smaller players.

Eleonore Pauwels, director of the Anticipatory Intelligence Lab at the Smithsonian Institution's Wilson Centre, says that it's natural that private-sector IT companies will become intertwined with medicine. "Biology and genetics and genetic sequences are just data, basically. If it's just data, you need to curate it, store it, protect it, sell it."

She's been closely watching the rise of US companies and also the very rapid entry of China into the genome 'race'. In March 2016, the Chinese government announced a huge US$9 billion, 15-year investment in precision medicine—and Pauwels has noted that it has been aggressively supporting and buying genomic data gathering entities.

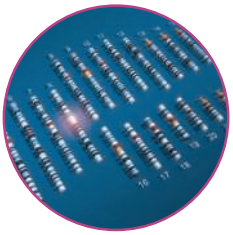
Certainly, says Pauwels, in the US the government has allowed private companies to take the lead in building the public's interest in their "genomic topography". While data players are mostly from the private sector she says, there's "huge investment, important investment, coming from the government's National Institutes of Health".

Miyano says Japan is watching US regulatory bodies closely to see how they will deal with this merging of interests. But, he says, increasingly close collaborations between IT and medicine are inevitable. "These are the kinds of collaborations and agreements that will create the foundations of the future of medicine, especially genomic medicine." ■

JAPAN: AN EXAMPLE OF HOW THE IT INDUSTRY IS INTERTWINED WITH GENOMIC MEDICINE'S CAPACITY

A surge in public funding for genomics in the Japanese government's research budget of 2000 saw Hitachi announce its intention to expand its sequencing facilities.

In 2014, Yahoo! JAPAN and DeNA both released direct-to-consumer genetic testing kits. These have been used by tens of thousands of Japanese people and offer potential data to researchers.



A record-breaking exascale supercomputer is being built by Fujitsu for Japan's largest research institute. It will be able to solve currently unmanageable genomic questions.

© GORODENKOFF/GETTY © CARLOS CLARIVAN/SCIENCE PHOTO LIBRARY © JAMES KING-HOLMES/SCIENCE PHOTO LIBRARY