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Interview – Data key as Illumina touts sequencing for all

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There has been much talk recently of “moonshot” programmes aimed at curing cancer. But, while a lot of the buzz has focused around potential new drug treatments, there is another important piece of the puzzle: genome sequencing to allow therapy to be tailored to each patient.

At the forefront of efforts to move sequencing from a research tool into widespread clinical use is Illumina. The company has come a long way since Roche tried and failed to buy it for $6.7bn in 2012. The group now has annual sales of $2.2bn, and market cap of nearly $25bn.

But it has only just scratched the surface, claims Alex Dickinson, its senior vice-president of strategic imperatives. However, making the leap into everyday clinical use is still some way off.

The company is about to enter a new era, with its long-time chief executive, Jay Flatley, to step down in July and be replaced by Francis deSouza. Mr deSouza seems on the face of it an unusual choice, having worked for the computer software specialist Symantec before becoming president of Illumina in 2013.

But with an increasing focus on the data analytics side of genome sequencing his appointment makes sense, says Mr Dickinson. “The data interpretation pieces have become things that we need to get right for sequencing to be used in a healthcare environment. Obviously he brings a lot [of experience] in that area.”

And with “muted near-term demand” for sequencing instruments, according to Leerink analysts, expanding further into the data management side of the business seems prudent.

$1,000 genome

The basic cost of sequencing a genome has come down dramatically – from $3bn when the human genome project was carried out in the 1990s and early 2000s to as little as $1,000 per genome today. This is the price that many believe will allow sequencing to make the jump into clinical use.

But it is not the only cost to consider: there is also the expense of processing the raw data. “A sequencer generates around 100 gigabytes of data; let’s call it 20 high-definition movies,” Mr Dickinson says. “As the data come off it’s a bit like the pieces of a jigsaw puzzle, and they have to be assembled. That was a big challenge from the point of view of computation.”

This was "a big concern" a few years ago, but the cost has also fallen quickly, says Mr Dickinson. “The cloud, Moore’s law [that computing power doubles every two years, bringing down costs] and the general evolution of computation is making that problem go away. Say sequencing costs around $1,000, the data assembly part costs tens of dollars in the cloud, so it’s not a big problem.”

However, there is still one remaining issue: the cost of interpreting the raw data for doctors, a key consideration if clinical sequencing is going to take off. “Once that jigsaw puzzle is assembled, it’s still just a big pile of genetic information,” explains Mr Dickinson. “Typically, clinical geneticists look at that information and come up with a report that then goes to a doctor.”

These handcrafted reports not only cost $3,000-5,000, but there are currently few clinical geneticists to carry them out.

Illumina is “trying to take what was a traditionally human function and work out how to automate it”, Mr Dickinson says, helped by the emerging area of machine learning.

The company does not want to replace clinical geneticists entirely, he adds. “The next jump for software [is to] make sure the run-of-the-mill translations happen automatically, and you only need a clinical geneticist for the rarer ones.”

As to how long it might take until this becomes reality, he replies: “I think we’re going to see very rapid changes in the next few years.” Illumina recently partnered with Genomics England on the 100,000 Genomes

project, which should help it refine its interpretation software for everyday clinical use, he says.

Tech crossover

It is clear that software developments are important in bringing sequencing to the masses, explaining why Illumina's sole acquisition last year, that of GenoLogics, was focused on IT.

But wider technological considerations are also important in realising Mr Dickinson's amibitious vision for the future – that every baby is sequenced at birth. This would allow the immediate diagnosis of any genetic diseases and provide information on whether the individual is predisposed to conditions such as cancer or diabetes. Steps could then be taken to prevent the development of these disorders.

To do this effectively more research is needed into the mutations that cause or contribute to certain diseases, which requires linking up all the available data on a patient. Estonia is leading here, according to Mr Dickinson. The country has "amazing cloud-based IT infrastructure" that stores everything in one database, from a person's school record and driver's licence to their electronic health record.

This means that genetic information can be more easily correlated with disease than in countries where electronic medical record provision is more fragmented, such as the US and, to a lesser extent, the UK.

But, even with efforts such as these, making this vision a reality seems unlikely in the near future. It would require a vast leap in the understanding of which mutations cause which diseases and how they interact. And, even if it is proven to reduce costs in the long run, there would need to be a dramatic shift away from the current healthcare model of treating disease, to preventing it.

And this is without even thinking about the ethical considerations of data-sharing.

For now, at least, this application of sequencing looks set to remain a research tool. The more immediate use is in cancer, where sequencing can aid the selection of chemotherapy.

Technology is becoming increasingly important in genome sequencing and medtech in general, illustrated by interest in the sector from traditional tech companies including IBM, Google and Apple.

Rather than being worried about the increased competition, Mr Dickinson believes that this crossover is good. "The greatest steps forward happen when branches of science, technology and the arts come together unexpectedly. I think we're at an extraordinary time."

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