Evaluate Vantage

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# Congenica aims to turn data into information

Elizabeth Cairns

## Sequencing data is all well and good, but what does it mean?

Genomic sequencing technologies have revolutionised the diagnosis of disease and, via companion diagnostics, its treatment. These days the approach is close to routine, and this poses a problem in the shape of the vast amounts of data it generates. A UK-based start-up, Congenica, is trying to address this problem using its analysis software, Sapientia.

“The problem we solve is you get a sequence from a patient and you immediately have about a hundred gigabytes of data on your hands – absolutely no use to anyone,” says Congenica’s chief executive, David Atkins. There is certainly burgeoning demand for this kind of data interpretation service, but Sapientia is currently sold for research use only, and cannot be used to diagnose disease by itself.

With companies like Illumina and Thermo Fisher Scientific able to sequence a whole human genome for less than $1,000 – provided the customer spends around $10m on one of their machines first, of course – data is accumulating so fast doctors are struggling to interpret it.

### Pinpointing the differences

Congenica’s tech can sort through whole genome data and identify variants between two different datasets – a patient and a reference, for example, or a child and one of its parents. There can often be millions of variations, of course, because a whole genome encompasses non-coding “junk” sequences, so the next step is to find the important ones.

“We prioritise the variants, whether it’s five million as compared to a reference or several hundred compared within a family. We identify the clinically significant variants and we can annotate that using our own clinical team,” Mr Atkins says. The information is then fed back to the doctor as a report, to help with the diagnosis.

At this stage the Sapientia report is just one factor among many that a doctor uses to make a diagnosis. Congenica’s tech is not regulated as a diagnostic; in the US it is used in Clia-certified facilities as a form of lab-developed test, and in the UK the company is selling it in partnership with Genomics England under a health institution exemption.

“Generally we provide the results as research use only and then typically the variant is confirmed by Sanger sequencing,” Mr Atkins says. This is then used along with factors such as the patient’s symptoms and family history to allow a diagnosis and recommend a treatment regimen.

Congenica is initially focusing its efforts on rare diseases that strike children. “Between 3% and 6% of

newborns have a rare disease of some description," Mr Atkins says, adding that diagnosis of rare conditions takes an average of four to five years and can involve a number of invasive procedures "and obviously a huge cost on the healthcare system".

Therein lies Congenica's health economic argument. The company will not disclose the prices it charges for access to its software, which is sold either via a licence or on a per-use basis. Mr Atkins concedes that genomic sequencing is "quite an expensive process compared to doing PCR tests", but says that getting a diagnosis within a couple of weeks, rather than the four to five-year timeframe, saves a huge amount of money.

Karen Megy, a bioinformatician at the University of Cambridge, uses Sapientia as a tool to visualise data to aid her research. "It is nice to be able to see the data," she says. "Bioinformaticians tend to see it in big text files – so much data that you just can't use it."

If Sapientia were not available the only way to achieve comparable results, Dr Megy says, would be for her clinician colleagues to learn programming. "Most of them would not be able to do it."

**Competition**

This is in fact what Congenica considers its main competition: clinicians analysing genomic sequencing data using spreadsheets they have built themselves.

Convincing these early adopters to move away from their home brew methods is the primary competitive challenge, though Mr Atkins says it is happening quite quickly, particularly in the US. He claims that Sapientia can do in an hour what would typically take a clinical geneticist working off spreadsheets a month.

In terms of traditional competitors four or five major companies are offering something similar "but materially different" to Congenica's platform, he says. He cites Qiagen and Agilent Technologies as companies looking into the genomic data analysis space.

Looked at another way, competitors might be potential partners – or acquirers. Mr Atkins suggests that the big sequencing companies are hitting a ceiling in terms of how many of their machines they can place with customers, and might soon come looking for an easy way to improve the value of their technologies.

"I wouldn't be shocked if Illumina and BTG and all the others sell a comparable or stripped-down version of what we do at some point," Mr Atkins says, though he stops short of naming them as likely buyers of Congenica. Lab chains and CROs might also be interested in the tech, he says.

Despite having launched Sapientia in 2014 the company is still not self-sufficient, and is currently seeking another funding round – it has already raised around £11m ($14.5m). There is, at least, plenty of interest from investors in this space, Mr Atkins says.

A technology that is relatively cheap to develop, bolts on nicely to a fast-growing area of clinical and scientific research, does not require expensive clinical trials and can be sold with limited regulatory oversight? No wonder.