

Precision medtech: why genomics & bioinformatics will matter to medtech companies

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Gene-based precision medicine has become central to the strategies of many pharmaceutical and diagnostics companies working to prevent and treat cancer using bioinformatics and genomics. Sagentia Innovation's Simon Forbes, Head of Genomics & Bioinformatics, and Rob Morgan, VP - Medical, share their thoughts on why gene-based precision medicine matters increasingly to companies in the wider medtech and medical device market, and the threats and opportunities it presents.

What do we mean by precision medicine?

Precision medicine (sometimes 'personalised' or 'genomic' medicine) is an emerging approach to preventing, treating and managing an individual's medical treatment by focusing on the genetic causes of disease rather than a patient's symptomatic consequences. It is based primarily on DNA sequencing technologies which are becoming ever cheaper and faster, while also considering the individual's symptoms, biochemical and imaging metrics as well as other environmental and lifestyle factors. By focusing on genetic causes of disease, precision medicine is designed to avoid potentially vague or misleading symptoms by determining the exact piece of DNA that is the disease source.



Why precision medicine matters to medtech companies

By focusing patient assessments on distinct disease causes, therapy can be tailored to the biological functions that are going wrong. \$billions are now aimed at designing new drugs to treat specific gene mutations causing a wide variety of cancer types. In addition to pharmaceutical design, therapy is being transformed as we better understand how tumour cells with specific genetic mutations can react better or worse to radio and chemo-therapies, driving huge new opportunities in targeted design.

Precision medicine outside oncology is catching up fast as we better understand the wider impact of genetics across human disease. There are fast growing opportunities for medtech companies to offer diagnostic solutions based on

genomics and design new interventional therapies better suited to more precisely defined diseases; we've started to call this 'Precision Medtech'. Advances in genomics also pose a threat to the current generation of medtech, either as better-specified disease activity becomes more druggable, or as therapeutic device requirements become more narrowly defined toward very specific functions.

As medicine increasingly focuses on genetics to stratify disease and therapy, medtech companies should be aware of the potential impact of genomics on their markets and plan for the opportunities which may emerge.

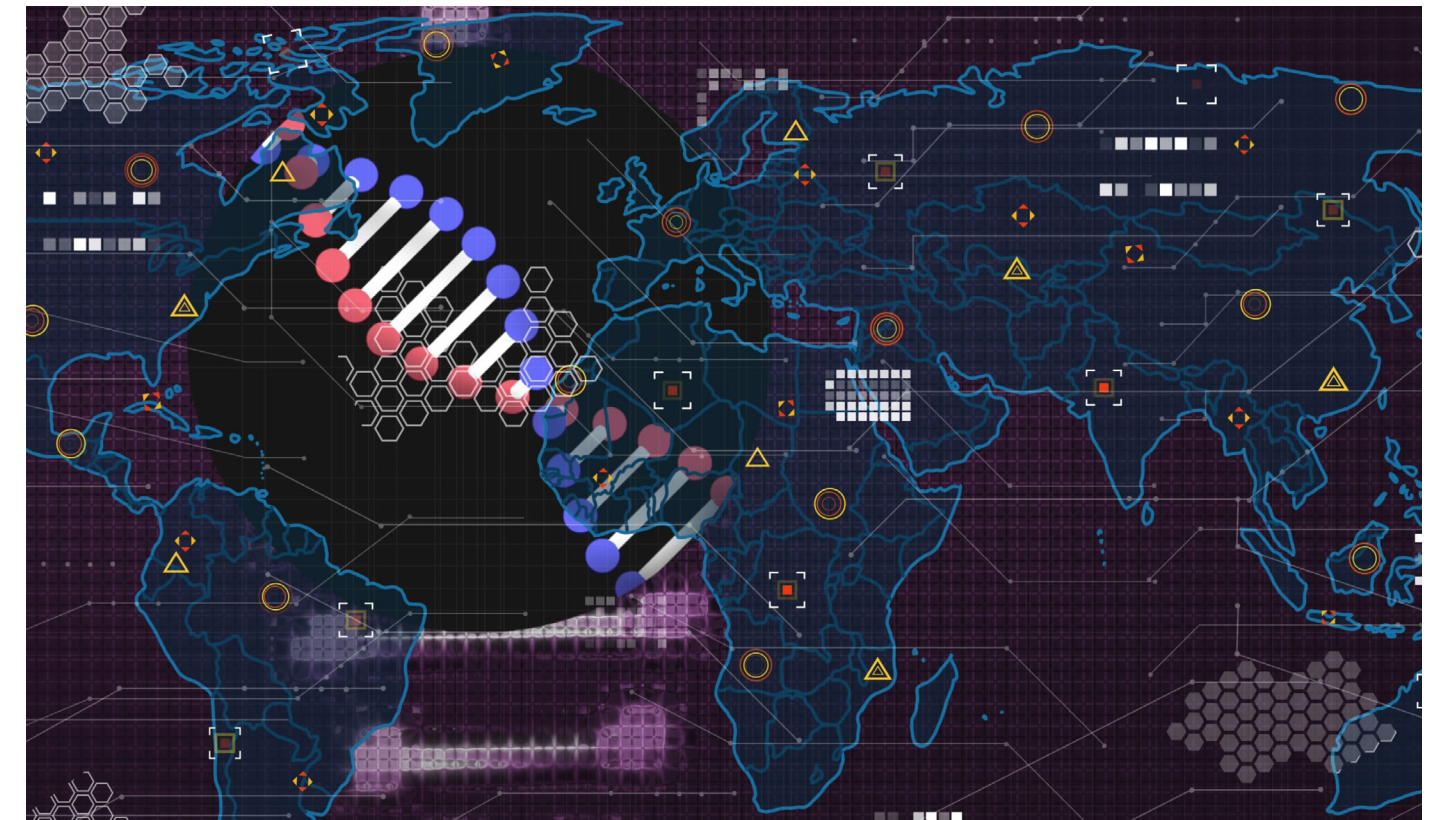
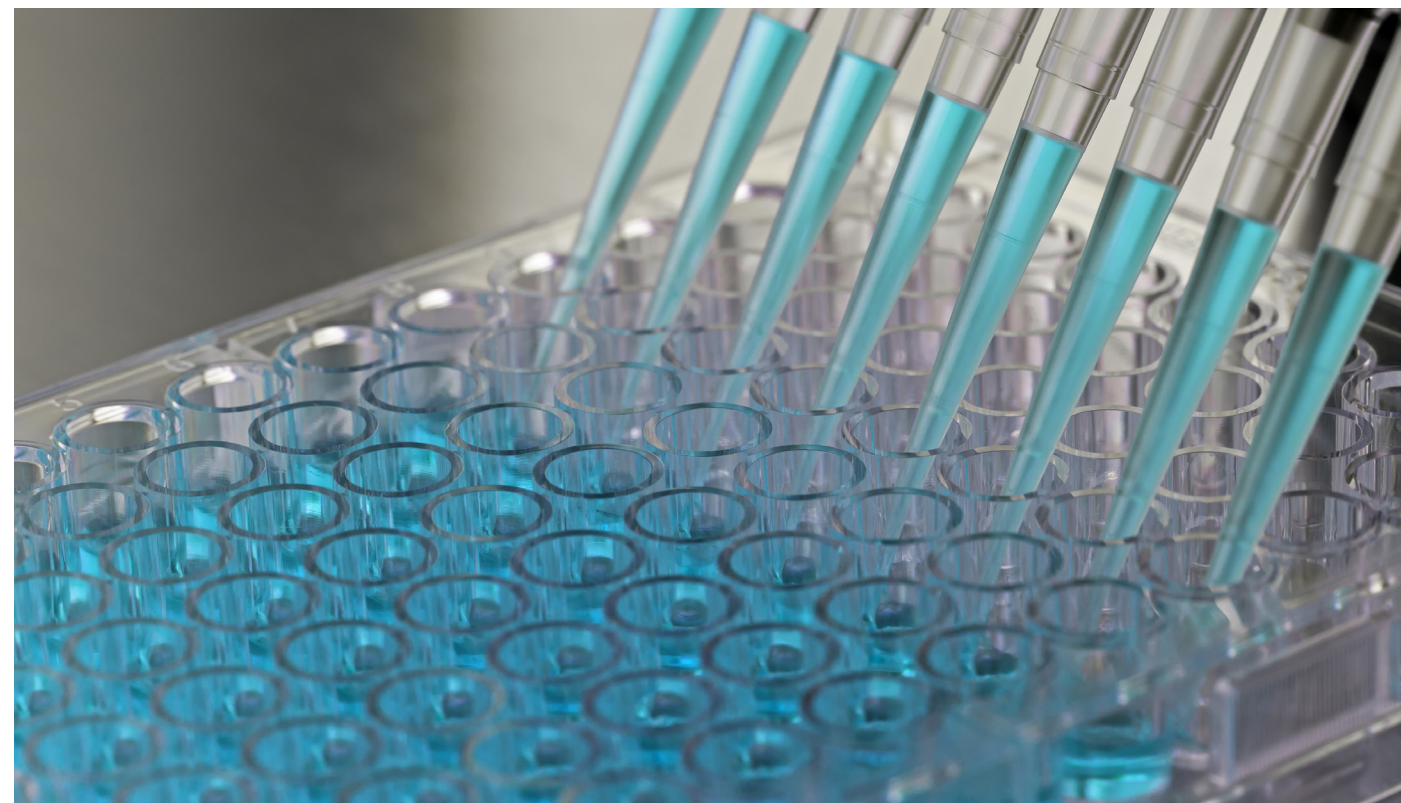
How bioinformatics and genomics are being used in precision medicine

Precision medicine usually combines genetic assessments of a patient with multiple other medical measurements for an individualised high-resolution picture of their disease to enable focused and targeted therapies precisely for each patient.

Beginning 20 years ago with limited single gene assessments in cancer (for instance, BRCA/EGFR in breast/lung cancers), DNA technology is now solidly embedded in the medical process with large gene panels and even whole-genome evaluations increasingly deployed for high-sensitivity and high-resolution disease detection. This has led to a huge fast-evolving range of new DNA diagnostic products. After much consideration, the **FDA** has approved cancer gene tests provided by **Roche Foundation Medicine** and **Guardant Health**, and these companies are working towards getting reimbursement through US molecular diagnostic programmes, Medicare and private insurers. Even more revolutionary, the **UK NHS** is piloting genetic tests from **GRAIL** to detect presymptomatic cancer to improve morbidity (& mortality) by potentially testing everybody in the country when they reach a certain age.

These tests are valuable in cancer because key DNA mutations are well known as targets for pharmaceutical intervention, with huge successes seen when a patient with one of these mutations is offered the appropriate mutation-directed drug. Excitement is now building to take these procedures and apply them beyond oncology and aim toward other big diseases such as diabetes, cardiovascular, obesity, arthritis, etc. Now we are understanding much better how everybody's genetics can define their response to a wide range of reasonably simple treatments, these techniques can be applied across the entire spectrum of medicine. For instance, several common genetic polymorphisms can substantially raise or lower a patient's pain relief from not just surgical anaesthetics, but everyday drugs like Paracetamol.

The impact of genetics is growing fast in general healthcare provision. [Genomics England \(UK\)](#) has been spearheading genomic medicine, focusing on DNA sequencing technology as central to patient care in cancer treatment as well as investigation of rare, often monogenic diseases. Italy is



poised to introduce a new genomic Citizen Test with [Dante Genomics](#), allowing every citizen a full-genome assessment as a standard solution in public healthcare; the **Abu Dhabi Executive Office** is offering a similar healthcare service as part of its staff benefits package. In the US, the FDA is increasingly regulating a wide range of gene-based molecular diagnostics, increasingly approving new tests as more precise for patient therapy than existing techniques. Of course, the greatest population scale-up is to be seen in China, which is now taking a very strong stance to encourage genomic technologies, collecting millions of patients' genetic data to try to achieve global leadership in genomic biotech.

Currently, cost is still an issue for genomic assessment with laboratory sequencing and bioinformatic evaluations costing in the order of \$1000 per individual in high-throughput labs. However, new technologies, for instance at [Ultima Genomics](#), are aiming to achieve full-genome assessment for only \$100, suggesting genetic technologies are about to become much more accessible and create a major impact on everyone's lives. In this situation, the bottleneck moves to

interpretation. Since our understanding of how each gene works is still developing, it is essential for interpretation reports and individual recommendations to be regularly updated to ensure good patient care and to avoid outdated recommendations which may mislead treatment. Significant opportunities exist for companies willing to watch for new science and react accordingly in their recommendations.

Beyond precision medicine, a new industry is spinning up around precision wellness. Initially evaluating genes involved in fat, carbohydrate and vitamin metabolism, stress, exercise, and mental health; a growing range of small companies are recommending lifestyle modifications to customers looking to optimise their state of health. Watching these successes, larger organisations are now entering the market, with [23&me](#) using millions of individuals' broad genomic data for pharmaceutical research (with **GSK**), and medical telehealth (in the US with **Lemonaid**). Similarly, medical insurer **BUPA** has deployed its [SmartDNA](#) product to early adopters in the UK interested in optimising healthy lifestyles for disease avoidance.

In our next insight piece, we'll be talking more about precision medicine in cancer care – register your interest to have it emailed to you when it is issued: info@sagentiainnovation.com



Get in touch to talk to Rob,
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your genomics developments:
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