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Precision medicine aims to make better diagnoses and improve patient management by understanding more about the human condition.

Much attention is given to the potential for designing better medicines based on an increased knowledge of the pathology of conditions. The other side is improved diagnostics to better direct patient management. The quantity and complexity of diagnostic information reflects the complexity of a patient's genetics, environment and lifestyle. Genomic technology is a huge source of information and has the potential for identifying new conditions, prognostic indicators, or patients who will respond better to treatments. The increased detail by which we understand patients will result in increased demand for information management. Technologies, such as digital health and artificial intelligence, can play roles in supporting the use of these complex data for diagnoses, prognosis and decisions on the best treatments.

The future of precision medicine rests on how we integrate these technologies, and the knowledge and skills to use them, into clinical practice. This means ongoing research to understand what information is important and whether associated changes in patient management work. And the development of new training schemes in genomics, computer science, informatics and data science, for example.

IMAGE: ISTOCK

Dr Matt Griffiths

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The future of precision medicine holds much promise – further development of current approaches and more specific treatment for particular disease. But also a revolution in diagnosis may come – with the increasing availability and affordability of "omics" a person may have a unique molecular profile for their disease created – and unique treatment provided. With so many having their ancestry reported via home DNA test kits, how long until these data are analysed into health prediction services? A test to predict your likelihood of developing cancer, diabetes and Alzheimer's, among others, could be around the corner.

A genome can now be sequenced in a day. The volume of data is extraordinary, and there is much that we cannot reliably interpret yet – but it provides a glimpse into an exciting future. Whilst we have a huge amount of information about how to deliver personalised treatment, how can we utilise the knowledge of risk to reduce incidence? Should precision medicine focus on prevention, rather than cure? What if there is no treatment or cure, do you want to know that you are at risk of developing a disease? There are several ethical considerations around findings of unknown significance.

Precision medicine has brought tremendous advances in patient outcomes by delivering better treatments – perhaps the future holds, not better treatments for disease, but better health without disease.

Brendan O'Sullivan

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How about... haematoxylin and eosin (H&E) staining? We tend to see H&E as the fundamental but limited basis for cellular pathology; most precision medicine relies on detailed genomics and/or proteomics to therapeutically classify tumours in ways that our current understanding of morphology alone cannot. But we also know the morphological hallmarks of certain targetable variants – dMMR/BRAF mutant medullary CRC; ALK-translocated signet-ring cell NSCLC. These phenotypes are not sufficient for precision medicine; molecular tests for these targets are more accurate. But could artificial intelligence overcome our own limitations?

Quantitative image analysis for classical IHC/fluorescence markers is garnering interest, but many commercial and academic groups are showing that H&E, subject to classical image analysis or machine/deep-learning algorithms, can yield prognostic and even predictive information not apparent to the human eye. What if you could determine HER2 expression level or BRAF mutation status using the very H&E slide from which you're making your diagnosis? Same-day diagnosis and complete molecular classification? Yes please! Implementation of such assays is likely to begin in the near future. These applications will only work with the largest and most accurately labelled datasets, and healthcare scientists should be delivering these.