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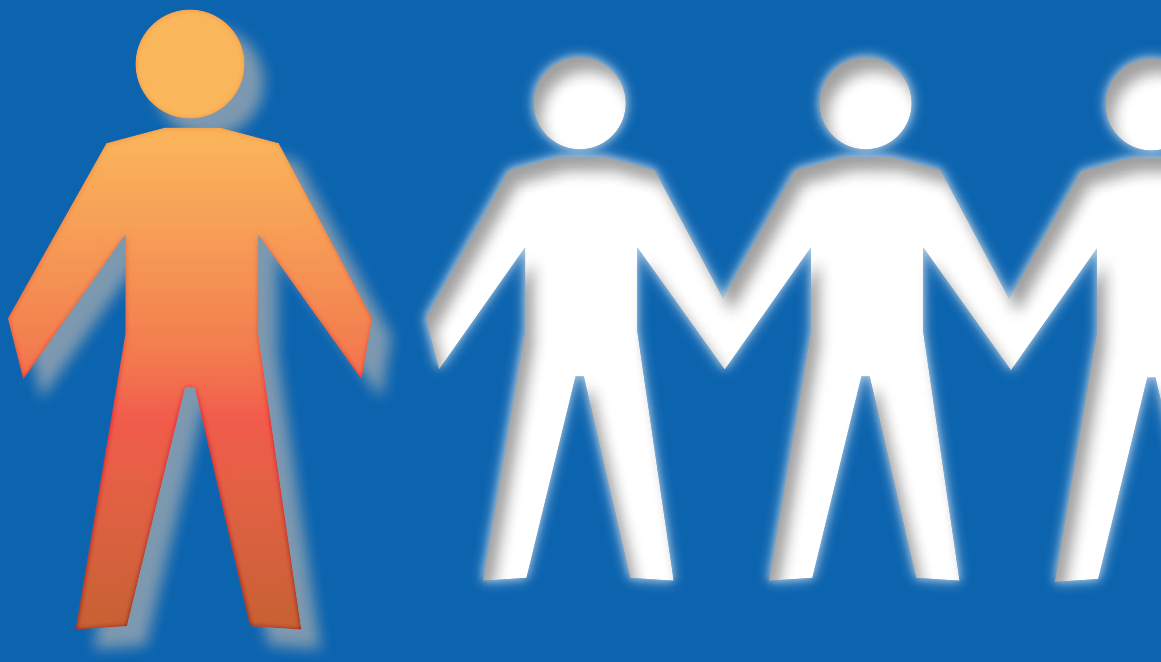
The personalised medicine technology landscape

Executive summary



As the National Health Service (NHS) marks its 70th anniversary it can boast a rich history of innovation. Over the years scientific and technological advances have transformed medical practice and ongoing innovation across biomedical, digital, computer science and engineering disciplines continue to offer novel approaches to improve patient care. More recently the convergence of technologies such as genomics and informatics is presenting significant opportunities to drive improvements through more personalised treatment and care of patients.

In recognition of these opportunities, NHS England has set out its vision for personalised medicine and how it intends to build on the work undertaken as part of the 100,000 Genomes Project. More broadly there is growing emphasis on the need for improvement in the effectiveness with which the NHS fosters innovation and delivers it to patients, in order to improve outcomes, to support the national economic interest and ultimately to ensure the long term sustainability of the NHS.

Executive summary

Objectives and approach

This review presents an independent evidence synthesis to inform the NHS as it seeks to develop its approach and policies to support the delivery of personalised medicine and realise its benefits for patients. To align with the timescales of the Five Year Forward View – a wide-ranging strategy for the NHS in England – this review predominantly focuses on the near-term opportunities and associated challenges, and briefly reflects on the longer term perspective on how developments in technology and knowledge could enable a whole system transformation and advance personalised medicine.

This evidence synthesis, informed through a process of desk-based research and analysis of public sources of information, grey literature, and peer-reviewed publications, along with interviews with relevant experts and stakeholders, sets out to:

- Review developments in biomedical and digital technologies that have been proposed to contribute to the personalisation of medicine
- Identify and describe specific examples that have a sufficiently well-developed evidence base for validity and utility such that they would be able to underpin the delivery of personalised medicine in the next three years
- Analyse how some of these approaches could be integrated most effectively within the NHS and highlight key considerations for action that NHS England could take to develop and deliver personalised medicine

The road to greater personalised medicine in the NHS

Genomic information is an important component of personalised medicine, helping to inform and refine the diagnosis, treatment and prevention of disease. A National Genomic Medicine Service, to be operationalised in 2018, will form the foundations upon which many other elements of personalised medicine can be built, including infrastructure and improving genomic knowledge, that may serve to galvanise wider developments for personalised care.

Whilst genomics is a key element of personalised medicine, it is not the only element. In total 25 different technology areas (including genomic based technologies) that will potentially have a significant impact either on patient outcomes or on health system implementation were reviewed.

These technologies can broadly be grouped into one of the following four categories:

- Technologies for greater molecular characterisation of individuals or disease
  e.g. genomics, metabolomics, proteomics
- Technologies for personalised therapeutic interventions
  e.g. stem cell therapy, genome editing/therapy, robotics
- Technologies for personalised disease and health monitoring
  e.g. consumer mHealth apps, digitally enabled wearables and sensors
- Underpinning and enabling technologies to transform the performance or capabilities of other technologies
  e.g. artificial intelligence and machine learning, microfluidics, nanomedicine

Seven areas were examined in greater depth due to (i) the near-term opportunities presented by the technologies to contribute to the greater personalisation of medicine, and/or (ii) their identification as key areas of strategic interest and importance to the health system. These areas were:

- Circulating tumour DNA (ctDNA) testing
- Pharmacogenomics
- Transcriptomics
- Pathogen genomics
- Regenerative medicine (specifically stem cell therapies and gene editing/gene therapies),
- Advanced image analysis for histopathology
- 3D printing

In analysing how the most promising applications of these technologies could be implemented the report sets out 53 key policy considerations for NHS England as it seeks to develop and deliver personalised medicine approaches that will contribute to the goals of the Five Year Forward View.

Realising the near term opportunities

Specific applications of each of the seven technology areas reviewed in greater depth offer near-term opportunities to realise the benefits of personalised medicine. Ensuring their potential can be fully realised into patient benefit is pivotal to raising awareness of the value of personalised medicine approaches in the short term and to accelerating the drive towards prevention and earlier disease detection in the medium to longer term.

The main themes emerging within each of the analysed areas are:

ctDNA testing

ctDNA testing is a form of genetic testing to analyse fragments of cell-free tumour DNA found in the bloodstream. The technology is having an impact on patients with non-small cell lung cancer (NSCLC) by increasing access to targeted therapies for those in whom solid tumour biopsy has failed. ctDNA testing can be used instead to inform treatment selection. It is likely that ctDNA testing could expand to other cancers within the next 1-3 years.

The health system will need to consider how to ensure that all eligible NSCLC patients can access the testing that is already available, and how current services can be supported and strengthened to deliver tests and expand as future uses of ctDNA testing become available.

Pharmacogenomics

Pharmacogenomics is the analysis of how genes affect an individual's response to drugs, with the aim of personalising therapy to maximise therapeutic benefit, and to avoid adverse drug reactions and undesirable side effects. Pharmacogenomic tests will be formally included among the genomic tests available as part of the National Genomic Medicine Service.

The appropriate uptake of pharmacogenomic testing can be supported through the incorporation of best evidence into UK guidelines, through training and development of the clinical workforce, appropriate clinical, laboratory and digital infrastructure, and through the collection of evidence of the impact of pharmacogenomic information on clinical decision making.

Transcriptomics

Transcriptomics is the study of RNA (ribonucleic acid) and how genes are expressed in a cell, tissue, or sample at a specific time point. There are a growing number of targeted gene expression tests emerging for early detection, prognosis, and therapy targeting – particularly for cancer.

The health system should prepare to respond to evidence around gene expression tests as and when it emerges, and consider how elements of existing laboratory genomics infrastructure could be used to support the timely implementation of transcriptomic testing when appropriate.

Pathogen genomics

Pathogen genomics examines the genome sequences of pathogens to enable more targeted management and control of infectious diseases. The utility of pathogen genomics in resolving challenging outbreaks within the health system has been demonstrated, and there is a firm evidence base for using whole genome sequencing for the management of tuberculosis.

The health system will need to determine how pathogen sequencing can be incorporated into infection control efforts when appropriate, especially for hospital based investigations or those falling outside the public health function remit of Public Health England (PHE), and how to access these services, for example by utilising existing sequencing provision.

Regenerative medicine

Regenerative medicine (stem cell therapies, gene editing and gene therapies) are treatments which seek to replace, repair or regenerate the body's cells, tissues and organs. A number of the regenerative medicine treatments offer potentially curative or long-term treatments for chronic diseases, and new opportunities for personalised cancer therapeutics using the patient's own immune cells.

Near-term and longitudinal planning – e.g. infrastructure development, workforce training, continued reassessment of regulatory structures and adapted methods for reimbursement – are all key to ensuring health system readiness for implementing these therapies as their number and range expand in the coming years.

Advanced image analysis

Currently most histopathology – the examination of tissue sections or blood samples on a glass slide – is carried out manually by scientists and doctors analysing slides under a microscope. Digital pathology processes capture slide images in a digital format so they can be stored, viewed, and analysed using a computer. This could facilitate advanced image analysis for histopathology

Histopathology has been highlighted as one of the areas that could be transformed by artificial intelligence (AI) technologies. However the digitisation of pathology workflows is the vital first step to harness the potential of computational approaches including artificial intelligence and machine learning for histopathology image analysis. Standardisation and multi-centre data collection will be crucial to advancing AI technologies for histopathology.

3D imaging and printing

3D imaging and printing is a manufacturing process used to create customisable objects by depositing or binding successive layers of material. 3D printed objects are facilitating the personalisation of medicine through the development of patient-specific anatomical models for surgical planning and the customisation of devices and implants for individual patients.

3D printing is a multi-use technology, but currently its implementation is fragmented and tends to be localised, and confined to specific clinical departments, or individual clinicians with knowledge of the technology. An NHS-wide strategy to support implementation of 3D printing is required to fully realise the benefits of this technology across the whole of the health system.

Strengthening the foundations for whole system transformation

In addition to technology specific policy considerations, there are cross-cutting aspects to the delivery of personalised medicine. These include top-down support for the implementation of new technologies such as:

- Harmonisation of methodologies and standards for data generation, capture, and analytics
- Engagement across the workforce around the benefits of personalised medicine approaches
- Mechanisms for sharing expertise
- Approaches for managing small groups of patients as personalisation results in more refined categorisation of disease

One of the most pressing cross-cutting requirements is the need for improved informatics infrastructure to collect, store, manage, share, integrate and analyse patient data. This is because many of the reviewed technologies can generate considerable volumes of data (e.g. genomics, metabolomics, wearables), or they may fundamentally rely on underpinning digital infrastructure to operate (e.g. genomics, medical imaging, artificial intelligence) as well as the digitisation of health records.

Whilst digitisation has been an ongoing aspiration of the health system, it has been challenging to implement. Without the underpinning informatics hardware and software solutions, progress towards greater personalisation will be stalled. However, if harnessed effectively, the data amassing from biomedical and digital technologies can provide better context to an individual's health. In turn, the effective flow of this patient information can enable greater coordination across the health system and greater personalisation of care. In a fast evolving digital-age it will be crucial that the health system's informatics solutions are sufficiently agile and flexible to respond to the evolving capabilities of biomedical and digital health technologies.

Conclusions

Personalised medicine holds enormous potential to transform healthcare in England and improve patient outcomes. Key to maintaining momentum towards greater personalisation in the long term are the near term opportunities set out in this report. The benefits for patients and the health system, including more precise diagnosis and prognosis, more targeted and personalised interventions, better understanding and prediction of individual disease risk, could together support more efficient and effective use of health system resources. These elements will be essential for delivering on the ambitions of the Five Year Forward View.

Each technology presents its own specific challenges, but with the increasing convergence of these technologies, successful utilisation will depend on a synergistic and coordinated approach to implementation. As the single biggest integrated healthcare system in the world the NHS is uniquely poised to achieve this.

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