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EXECUTIVE SUMMARY

The NHS is famed as one of the top healthcare systems in the world, having provided quality healthcare free at the point of use for more than 60 years. Going forwards, the NHS must adapt to increasing challenges by exploiting opportunities provided by innovative technology.

Personalised medicine is about bringing the right treatment to the right patient at the right time. It is one of the key vehicles by which the healthcare system of tomorrow can achieve better outcomes for patients and financial efficiency.

By increasing the participatory role of patients and providing individually targeted health information and treatment, the role of personalised medicine supports objectives outlined in NHS England’s ‘Five Year Forward View’ (2014). However, despite the ‘Personalised Medicine Strategy’ initiated in 2015, there is still an urgent need to address the lack of preparation, awareness and engagement in this emerging field. With enormous potential benefits to be gained, the UK must not lag behind in its legal, technological and social infrastructure to prosper from future advancements.

A cautionary tale can be seen in the present example of electronic health records, where a lack of planning has led to challenges with interoperability (Houses of Parliament, 2016).

Today’s younger generation will be the first to truly witness, understand and experience the benefits of personalised medicine. Thus, we need to ensure that young people in particular are able to understand the need and reason for population-wide genetic data sharing and the implications for the future health of the UK.

“A move away from ‘a one size fits all’ approach...”

Sir Bruce Keogh

Personalised Medicine...uses emergent approaches in areas such as diagnostic tests, functional genomic technologies, molecular pathway, data analytics and real time monitoring of conditions to better manage patients’ health and to target therapies to achieve the best outcomes in the management of a patient’s disease or predisposition to disease.

NHS Personalised Medicine Board (2015)
This paper analyses the current state of affairs in personalised medicine, identifies gaps in NHS readiness for future developments, and explores public perception through quantitative primary data to identify challenges that must be solved in order to create a successful foundation.

Personalised medicine has the potential to revolutionise the NHS. With the transformation already beginning to take place, now is the time to act to ensure future tangible benefits.

**RECOMMENDATION 1:**
Ensure electronic medical records are compatible with personalised medicine. The technological strategy for personalised medicine needs to include the ability to link symptoms and physical traits with genomic information, and the capacity to store large quantities of genetic data.

**RECOMMENDATION 2:**
Redraft genetic data privacy rules to prepare the legal framework for personalised medicine, ensuring there are UK laws in place that safeguard genetic data confidentiality and privacy after leaving the European Union (EU). Our research suggests that the general public are not confident that the current agreement with insurers offers sufficient protection against misuse of personal genetic data. The UK leaving the EU provides the perfect opportunity to review and strengthen UK genetic privacy laws.

**RECOMMENDATION 3:**
Develop a Patient Charter on genetic data to educate the public about personalised medicine: the legal implications, ethics and potential benefits. The charter would provide a simple explanation of the applications of genetic data, the different levels of data sharing – including the benefits of sharing with industry – and the protection in place for patients. The Charter would enable patients to opt out of specific levels of data sharing.

**RECOMMENDATION 4:**
Develop targeted education programmes in schools, including the introduction of bioethics into the GCSE science curriculum, to increase awareness and understanding about personalised medicine and the recommended Patient Charter. Despite the fact young people today will be the first to interact significantly with personalised medicine, the majority report low understanding. This needs to be addressed to ensure they feel well informed and confident in their ability to make genomics-based treatment decisions in the future.

Our research suggests that the general public are not confident that the current agreement with insurers offers sufficient protection against misuse of personal genetic data. The UK leaving the EU provides the perfect opportunity to review and strengthen UK genetic privacy laws.
INTRODUCTION

The genetic basis of disease is an area of active research where much progress has been made, especially in the fields of monogenic disease and cancer, but there is still a considerable amount we do not know.

A recent example of the power of personalised medicine comes from the first diagnosis within the Genomics England 100,000 Genomes Project (see case study).

CASE STUDY: 100,000 Genomes Project (Genomics England, 2015)

Leslie Hedley (57) has a history of high blood pressure, which led to kidney failure and multiple failed kidney transplants. Whole-genome sequencing revealed that the kidney failure was caused by a genetic variation. Subsequent sequencing of Mr Hedley’s daughter revealed she had inherited the mutation; however his granddaughter had not (Genomics England, 2015). Fortunately, knowledge of the variation in Mr Hedley’s daughter meant that she could start taking blood pressure medication as a means of preventing future kidney failure. Not only is this a great outcome for Mr Hedley’s daughter on a personal level, but preventing the onset of kidney failure using cheap blood pressure medication has spared the NHS the cost of a future kidney failure patient. This type of intervention also has wider societal implications, including reducing the likelihood of productivity losses in the future. This example of disease interception demonstrates how access to genomic data, linked to health records and details of symptoms, is critical for improving our knowledge of complex diseases and developing new treatment strategies.

Globally, several direct-to-consumer genomic testing companies have emerged over the last decade. Although they have proven popular with consumers, many concerns have been raised about the analytical and clinical validity of these tests (Adams, 2013). A study conducted by the US Government Accountability Office in 2010 found that the companies were making misleading claims, that different companies were making contradictory risk predictions for the same condition in the same patient, and that good-quality expert advice was lacking (Kutz, 2010). However, a strong advantage of the increase of such tests is the accompanying increase in data available for research, which may contribute towards future breakthroughs.

The UK is already promoting itself as a world leader in the field of personalised medicine and genomics, inviting governments, healthcare providers and commercial companies to partner with the UK and learn from our expertise (Department for International Trade, 2016). Together with the creation of Genomic Medicine Centres and many other initiatives, this shows the UK’s commitment to accelerating the pace of research and the use of personalised medicine. Therefore, it is crucial we have the right policies and technical infrastructure in place now to ensure we continue at the cutting edge of personalised medicine – in planning, research and delivery.

2.1 VISION FOR THE FUTURE

Imagine a future where everyone’s genome is sequenced and forms the basis of their health record. If any genetic diseases or increased risk factors are detected, health management plans can be developed and individuals can be advised on lifestyle choices or preventative treatments, and they can be closely monitored for any early signs. For example, prophylactic surgery and chemoprevention have been shown to significantly reduce the risk of breast cancer for women who carry mutations in the BRCA1/BRCA2 genes (Hartman et al, 2001).
The potential of genomics extends further than one-time genome sequencing. For example, cancer cells have a mutated genome which can be compared with the individual's healthy genome in order to detect the driver mutations, which can indicate the most beneficial treatments. In addition, new technologies are showing promise for vaccinating patients against specific cancer cell mutations, causing the immune system to attack the cancer cells (Snyder, 2016, p.44).

Stem Cell Technology is a big enabler of personalised medicine. Cells can now be ‘reprogrammed’ into a different cell type; cells from a patient’s cheek swab can be reprogrammed into, for example, heart cells (Cullis, 2015, p.42). This means that drugs can be tested on cells genetically identical to those in a sick individual to test for adverse reactions or to establish dosage. Looking further into the future, this technology could be paired with 3D printing to grow new organs genetically identical to the original failing organ (Cullis, 2015, p.124).

Whilst recognising the significant potential, there is a risk of ascribing too much weight to genetics as a determinant of disease and overlooking the complex interactions with environmental factors and behaviour. It is important to remember that this is an area of active scientific research and the roles played by different genetic variants in both health and disease states are not yet fully understood. In the direct-to-consumer genetic testing industry especially, reports of increased risk for specific conditions are still often insignificant (Adams, 2013). There is a long way to go before all the benefits described above can be realised.
3 FINDINGS & IMPLICATIONS

3.1 CURRENT STATE OF AFFAIRS

Progress of personalised medicine is accelerating in the UK due to the work of Genomics England, a company owned by the Department of Health and launched in 2013 with the aim of creating a genomic medicine service for the NHS. It was set up to deliver the 100,000 genomes project, which aims to sequence 100,000 whole genomes from NHS patients, focusing on both rare diseases and cancer (Genomics England, 2016). This makes it the largest national sequencing project of its kind in the world. In October 2016, Genomics England reported it had sequenced 14,000 whole genomes. This work is key to the progress of personalised medicine, setting standards in data collection and analysis, genetic privacy and patient consent, and the commercial use of data.

3.2 CHALLENGES FOR PERSONALISED MEDICINE

3.2.1 Data Collection and Analysis

In order for personalised medicine to be integrated into everyday healthcare, it is important that the right information is available in the right format to allow analysis. The NHS is ‘considerably behind other industries in terms of its use of data analytics’, and potential efficiency savings upwards of £16 billion a year could be unlocked if the NHS employed data analytics more successfully (EMC-Volterra Partners, 2014). The potential saving is largely attributed to better quality healthcare through personalised medicine. This would be achieved by integrating datasets to enable all patient information to be analysed, leading to an accurate diagnosis and a personalised treatment plan: getting the right treatment to the right patient at the right time. This leads to efficiencies by reducing money wasted on ineffective treatments, reducing re-hospitalisation costs due to severe side effects caused by non-effective treatments, and reducing costs associated with disease progression. In cancer treatment, the first drug given to patients is ineffective in 75% of cases (Spear et al., 2001). To improve data collection and analysis, Genomics England is combining genomic sequence data with medical records to allow the causes, diagnosis and treatment of disease to be investigated (Genomics England, 2016). Funding has also been contributed to Oxford University’s Big Data Healthcare Centre, which plans to analyse NHS patient records and data from DNA sequencing and clinical trials in an effort to improve the detection and treatment of a range of conditions.

Genomic information needs to be correlated with detailed and accurate phenotypic information to advance research. A phenotype is the observable physical or biochemical expression of a specific trait, such as raised blood pressure, which can be found in patient records (Duke University, 2014). Standardisation in the recording of phenotypes will support bigger datasets that include all NHS patients, as opposed to isolated datasets from different NHS trusts all using their own terminology. Ultimately, there should be a standardised worldwide phenotype dictionary used for all electronic health records.

3.2.2 Privacy

As personalised medicine grows, more people will be sharing their genetic data. It is vital to protect this sensitive information to ensure it will not be used to discriminate in healthcare, employment or social scenarios. A survey of 117 16–30 year olds showed that public willingness to share genomic data will depend on who has access to the data and what it will be used for. The group gave a response of 4.2 out of 5 on willingness to share (0 not willing, 5 completely willing) when asked if they would share their data with life sciences companies to increase the amount of research undertaken. Interestingly, when informed about the potential threat from insurers accessing genetic data, and data protection in place, respondents’ willingness to share data with life sciences firms actually fell (to 3.8/5). It is possible that respondents had not fully appreciated the risks of sharing their genetic data when answering the first question. However, it appears that even the mention of protection against insurer access to data stifles public willingness to share data.
Misunderstandings over the consent model and patient privacy were contributing factors to the National Data Guardian’s (2016) recommendation to end the care.data programme. Following the closure, the National Data Guardian made recommendations for better safeguards in the sharing of patient data. This included a method for testing compliance against the standards, and clarity as to how people’s health and care information would be used and in what circumstances they can opt out.

Our survey indicates that the public want legal reassurance that their data will be protected. The Equality Act 2010 bans employers from requesting or accessing genetic information as part of pre-employment checks, and the UK is currently bound by the EU Lisbon Treaty (Article 21), which bans any discrimination based on genetics, although it is yet to be seen what protections will be put in place once the UK leaves the EU. In addition, a voluntary moratorium between the UK government and insurers limits insurers’ access to genetic information, meaning that people who take a genetic test and find out they are predisposed to a condition are not required to disclose the results when purchasing cover. Ellen Thomas (2016), Clinical Lead for NHS Genomic Medicine, is confident that the moratorium will be renewed when it ends in 2019. However, just over half of the participants in our survey thought the current moratorium arrangement with insurers did not represent sufficient protection.

3.3 THE IMPORTANCE OF COLLABORATION

The challenge of privacy needs to be addressed in order for research to advance. Publicly-funded research in both hospitals and universities is an important source of new scientific knowledge. However, to accelerate the benefits from personalised medicine, the volume of research should be maximised through collaboration. Industry has the resources, motivation and expertise to uncover new science and bring that science to market in the form of new treatments. Furthermore, industry represents 48% of the funding for health research in the UK (Figure 1).

Genomics England recognises the need for collaboration and is aiming to accelerate the UK genomics industry by developing methods of charging industry players for access to NHS genomic data. This will ensure that the costs of maintaining the data are shared with companies and that the UK taxpayer benefits, should they successfully develop treatments, diagnostic tests or other services through its use.
As more sequence data becomes available and healthcare data is more accessible in digital records, it is important that the public understand the implications of sharing their data and that appropriate consent processes are ready to support continued collaboration and data-sharing with industry. Education becomes a vital part of this picture.

3.4 EDUCATION: IMPLICATIONS, ETHICS & BENEFITS

According to research commissioned by the Wellcome Trust, the public is thought to suffer ‘context collapse’ when thinking about private firms accessing health data (Ipsos MORI, 2016). There are two traditional data sharing contexts. In the social contract context, patients have an ‘open, vulnerable mind-set’, where they are comfortable actively sharing data that will contribute to their own care, while passively contributing data that will help society (e.g. through service optimisation). When private companies enter the patient data equation, the commercial transaction context of data sharing is activated. In this context, data is valuable – and private firms should not get valuable patient data at zero cost and turn it into profit. When faced with the collision of these contexts, patients revert to familiar prejudices, such as ‘the NHS should deliver my healthcare’ and ‘private companies are untrustworthy’ (Ipsos MORI, 2016). When asked about the risk and reward of various examples of data sharing, the public found genetic data to be the most risky and ‘the most potentially valuable’.

However, respondents to a quantitative survey largely supported commercial access to health data for research purposes, especially if there was a risk that the research might not otherwise be undertaken (Ipsos MORI, 2016). Reconciling public desire for progress with its aversion to private firms profiting from public genetic data will require a mechanism to ensure the general public benefits from any private research outcomes. Potential examples include discounted access to treatments for NHS Trusts that have supported data gathering and maintenance, or incentives for private firms to share some of their analytical tools or processes with other researchers.

3.4.1 Patient Charter

A Patient Charter is a tool that could help patients unpick this complex collision of contexts. The top driver of acceptability of data sharing is public benefit (Ipsos MORI, 2016). A charter could clearly explain how private organisations accessing patient data will contribute to the public good (through paying to access the data, or through another profit-sharing arrangement). A significant amount of work on the concept of patient charters has been conducted by the charity Genetic Alliance, which brings together patient groups to understand what factors need to be considered to protect and enrich the patient’s experience. Particularly relevant is the charity’s 2015 Patient Charter (Genetic Alliance, 2015), which outlines 15 recommendations for consideration before genome sequencing is widely offered throughout the NHS (including the crucial role of genetic counsellors). A charter to support personalised medicine policy should represent a promise to patients about what will and won’t happen should they consent to sharing their data.

Consent is an important part of the personalised medicine debate. The recently published Caldicott Review suggests that consent may be given singularly or in parts (National Data Guardian, 2016). The notion of consent in parts may be beneficial to personalised medicine policy to ensure patients are able to share data up to the level they feel comfortable, rather than a clear-cut binary decision. For example, patients may choose to opt out of:

- any personal information being used for purposes beyond their own care
- any personal information being used in support of research by academics and commercial organisations
3.4.2 Public knowledge, awareness and opinion of personalised medicine

A survey by the Wellcome Trust confirms many of the themes we found in our research. Although 86% of respondents said they were aware of genetic tests that predict the likelihood of developing genetically-influenced diseases, only 12% claimed a good understanding of what a genome was (Wellcome Trust, 2016). This highlights the significant mismatch between the level of general information shared across the media and the amount the public truly understand. Further to this, our youth survey indicated that the more information people have around genomics, personalised medicine and potential benefits, the more willing they are to share their personal health data, thus highlighting the importance of education as a way of driving engagement.

Another crucial question concerns how to best communicate important educational content on personalised medicine, especially to younger generations. Firstly, it will be important to incorporate the key biological concepts of personalised medicine, alongside bioethics and consent, into the GCSE Science curriculum. Secondly, there will need to be a national educational campaign to support introduction of the Patient Charter. In our survey, healthcare professionals came in first as the most popular channel of communication. Content (videos and infographics) shared through social media followed a close second, with traditional channels (pamphlet and TV advertising) lagging behind. The implications for the NHS are twofold: first, healthcare professionals must be confident in explaining the Patient Charter and the risks and benefits of any levels of consent. Next, the NHS should focus on an accessible and engaging social media education campaign that increases the public’s understanding of personalised medicine and genetics.

Imagine the NHS were to have a short, simple explanation of data sharing practices, after which they asked you to consent to various levels of data sharing for your own genetic and health data. How would you like to receive this explanation?

Figure 2. Preferred channels of education. YHP survey, 2016.
The era of personalised medicine has begun. Researchers are understanding ever more about the human genome, leading to breakthroughs in treatments and diagnostics. The more we discover the closer we get to the innovative personalised treatments of the future. This does not have to be decades in the future: our research has shown that changes can and should be made now to accelerate the translation of personalised medicine from scientific research to the application of innovative diagnostics and treatments for NHS patients.

RECOMMENDATION 1.
Ensure electronic medical records are compatible with personalised medicine.

In order for personalised medicine to be integrated into everyday healthcare, there needs to be a well-functioning, nationwide electronic health record system with the capacity to store large amounts of data, with interoperability across analytical systems. NHS health records should adopt a standardised dictionary to enable symptoms to be searchable and easily tied to genomic information. Researchers and the NHS should collaborate to develop a common standardised format to create high quality genomic data that can be used in research and diagnostics.

RECOMMENDATION 2.
Redraft genetic data privacy rules to prepare the legal framework for personalised medicine, ensuring there are UK laws in place that safeguard genetic confidentiality and privacy after leaving the EU.

The government should review genetic data privacy rules, including how genetic information should be used and who will have access. Regardless of the level of legal protection provided in real terms, the public will only engage if they believe they are protected. More than half of the young people we surveyed felt that the government’s voluntary moratorium with UK insurers on the use of genetic test results did not represent sufficient protection. We also found that the public’s willingness to share their data depended on who would have access and what the data would be used for. In particular, knowledge that insurance companies could have access to data led to a decrease in willingness to share.

In order for personalised medicine to become a reality, the government needs to ensure that data will not be used to discriminate in healthcare, employment or social scenarios. Alongside this, they must ensure that the public understand how they are protected and appreciate the full implications of genetic data sharing.

“By failing to prepare, you are preparing to fail.”

Benjamin Franklin
RECOMMENDATION 3.
Develop a Patient Charter on genetic data to educate the public about personalised medicine: the legal implications, ethics and potential benefits.

This charter should be based on the proposed genetic data privacy policy and led by the NHS with the support of specialist organisations and charities. The purpose of the charter is to simplify the complex environment of patient data. The charter should:

• explain the range of applications of genetic data, from improving healthcare delivery through to supporting medical research in the private sector, and the benefits of these applications to society
• explain how patients are protected if they choose to share their data
• enable patients to opt out of specific levels of data sharing

RECOMMENDATION 4.
Develop targeted education programmes in schools, including the introduction of bioethics into the GCSE Science curriculum, to increase awareness and understanding about personalised medicine and the recommended Patient Charter.

Ultimately it will be these generations, going forwards, that will have the first significant interactions with population-wide genetic data sharing. The launch of the Patient Charter should be supported by a social media campaign to educate the public, especially young people, on the power of their genetic data to deliver benefits to the NHS and to society.

Universities and hospitals should be encouraged to educate medical students and staff on the practicalities, ethics and implications of personalised medicine, to ensure the NHS is prepared for when new personalised treatment options come to market, and that physicians can confidently explain the benefits and risks to patients.
The NHS envisions a future with personalised medicine at its core. With a strategy already in place to drive this transformation, detailed planning and preparation to ensure success must begin now. As the Youth Health Parliament, we believe that an imperative part of this is to ensure the public is educated and engaged on this important topic.

Ultimately, today’s younger generation will be the first to significantly interact with population-wide genetic data sharing and reap the benefits of personalised medicine. Therefore, young people must understand how much their data can contribute to the creation of new treatments, even if part of the research needs to be done through the commercial sector. This is why a wider education campaign is critical to accompany the launch of the proposed Patient Charter. An informed public may not necessarily be more likely to consent to data sharing with commercial organisations, but at least decisions will be guided by fact, rather than misconceptions due to a lack of understanding.

Personalised medicine is already an innovative field with huge potential to lead the future direction of our healthcare. As the Youth Health Parliament, we want to build on current momentum and ensure that the essential groundwork is put in place now so that the UK can truly reap the rewards and maximise tangible benefits.


Genomics England, 2015. First patients diagnosed through the 100,000 Genomes Project. [online] Available at: https://www.genomicsengland.co.uk/first-patients-diagnosed-through-the-100000-genomes-project/ [Accessed: 05/09/2016].

Genomics England, 2016. 100,000 Genomes Project. [online] Available at: https://www.genomicsengland.co.uk/the-100000-genomes-project/ [Accessed: 05/09/2016].


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Personalised medicine is an approach to healthcare that involves analysis of a patient’s DNA sequence in combination with other highly specific test results and measurements.

Personalised medicine has the potential to:

- Improve patient outcomes by intercepting diseases earlier and improving the efficacy of the first treatment.
- Reduce healthcare costs.

The youth generation will be the first to truly benefit from personalised medicine. 85% of young people found it quite likely or very probable that they would benefit from personalised medicine.

Recommendation 1:
Ensure electronic medical records are compatible with personalised medicine – including the ability to link symptoms and physical traits with genomic information, and the capacity to store large quantities of genetic data.

Recommendation 2:
The legal framework must be ready – patients need to feel absolutely confident they are protected from any potential discrimination that may arise as a consequence of the results of their DNA sequence analysis. The UK leaving the EU provides the perfect opportunity to review and strengthen UK genetic privacy laws.

Recommendation 3:
Develop a Patient Charter on genetic data to educate the public about personalised medicine: the legal implications, ethics, potential benefits and possible risks.

A Patient Charter would allow patients to make an educated decision about who can use their valuable genetic information:
- Share it with their clinician
- Share it with clinicians and researchers
- Not share genetic data

Recommendation 4:
Education is key, especially amongst the younger generations who will be first to benefit. Personalised medicine should be introduced in the school curriculum, medical students need to learn how to incorporate genetic information into care and the public need to understand the potential benefits and risks.

The NHS envisions a future with personalised medicine at its core.

With a strategy already in place to drive this transformation, detailed planning and preparation to ensure success must begin now. As the Youth Health Parliament, we believe an imperative part of this is to ensure that the public is educated and engaged on this important topic.