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Bringing medicines to *life*

Life Sciences R&D:

Harnessing the UK's genomics expertise to improve patient outcomes

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Contents

Executive Summary

Genomics is an interdisciplinary field of biology focussing on the genetic and/or epigenetic sequence information of organisms in order to understand the structure and function of these sequences and of downstream biological products and processes. Since the completion of the Human Genome Project in 2003, genomics has been transforming the lives of patients. By providing a more detailed understanding of the genetic causes of disease, it has helped better stratify patient populations, enabling the development of more personalised treatments and supporting earlier diagnosis and prevention. Sequencing the first human genome cost £4 billion and took 13 years. Today, it takes less than a day and costs below £1,000.[1]

Genomics now plays a critical role in understanding whether individuals have an underlying condition (complex or rare), whether they are at risk of developing a condition later in life, and whether they will be responsive or not to a particular treatment. Due to the significant application of genomics in medical research and healthcare, the global market for genomics has grown substantially over recent years and is estimated to grow by £10.75bn between 2021 and 2025.[2]

The UK has built up global leadership and expertise in genomic capability. This legacy is supported by considerable cross sector expertise, with the Bioindustry Association (BIA), Medicines Discovery Catapult (MDC) and Wellcome Sanger Institute identifying over 140 genomics companies in the UK,[3] generating an estimated total turnover of £2.4bn.[4] These genomic capabilities underpinned the UK's response to the pandemic, with the COVID-19 Genomics UK Consortium (COG UK) and a vast diagnostic and sequencing network, putting genomics at the heart of the UK's research and public health efforts.

Recognising the UK's strengths in genomics and the opportunities to unlock further benefits for patients across the country, the UK Government published a national strategy, Genome UK, which outlines how it will maintain and extend the UK's leadership position. Between 2022 and 2032, the four nations of the UK will work together with the NHS and life sciences sector to realise the potential of genomics for the benefit of patients and ensure that the genomics services thrive in each nation.

From a global pharmaceutical industry perspective, a virtuous cycle is needed, where genomic-enabled R&D drives the delivery of new genomic medicines, which in turn informs the next era of genomic R&D and genomic medicine, to maximise patient benefit. However, too few patients are currently benefiting from the UK's genomic expertise and assets.

To fulfil the ambition of Genome UK, the UK must drive improvements across the system to translate its R&D expertise and vast infrastructure into improved patient access to genomic medicine.

This report describes both the strengths and weaknesses of the UK's genomics offer, identifying how the sector can work together to attract further investment and deliver an enhanced offer for patients and their families.

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Key facts & figures

The global market for genomics has grown substantially over recent years and is estimated to grow by £10.75bn between 2021-2025. In the UK, genomics related activity has generated an estimated total turnover of £2.4bn over recent years.

As of 28th September 2022, the UK had conducted over 500 million Polymerase Chain Reaction (PCR) tests for SARS-CoV-2 (the virus causing COVID-19) and completed and uploaded over 2 million SARS-CoV-2 genome sequences to the international GISAID database, enabling the identification of current and new variants to inform the public health response to the pandemic.

The 100,000 Genomes Project has to date, led to a rare disease diagnosis for 25% of participants, identified a potential therapy or clinical trial for ~50% of cancer cases and created the National Genomic Research Library (NGRL), which to this day provides data for researchers to learn more about health and disease.

In September 2019, a consortium of government, charity and industry (Amgen, AstraZeneca, GlaxoSmithKline and Johnson & Johnson), came together to fund whole genome sequencing of 450,000 participants, with the aim of deepening the understanding of the interplay between genetic and environmental drivers of health and disease. Two years later, data on the first 200,000 participants was made available to researchers, representing the largest single release of whole genome sequencing data.

The UK's largest health research programme, Our Future Health, has leveraged investment of over £140 million from the life science industry, including 8 ABPI member companies, to evolve how disease is detected and potentially at-risk patients identified.

A study conducted by the International Quality Network for Pathology (IQN Path), European Cancer Patient Coalition (ECPC) and European Federation of Pharmaceutical Industries and Associations (EFPIA) found that access to biomarker testing in the UK needed to be improved, in line with best practice in Sweden and Germany, to reduce variability in access to genomic tests and speed-up the availability of genomic tests in conjunction with the introduction of new medicines.

The Archangel Newborn Screening Review found that the UK is lagging behind European counterparts, such as Italy, Iceland, and Poland, who screen up to four times as many conditions as the UK, where babies are only tested for 9 conditions. This means that babies in the UK are missing out on critical diagnostic testing and potential life-saving treatments.

Recommendations

Harnessing genomics for research and development

1. The Medical Research Council - UK Research and Innovation (MRC-UKRI) should partner with industry, academia, and charities to scope and deliver the UK Functional Genomics Initiative, ensuring plans build on the UK's existing expertise and infrastructure.
2. The UK government and NHS should fully deliver the commitments in the Data Saves Lives and Accelerating Genomic Medicine in the NHS strategies, to drive interoperability and connectivity between flagship research programmes and across genomic and health data assets.
3. Genomic medicine services across the four UK nations should work with the research sector to develop a UK-wide Genomic Research Collaborative (see figure 2) which:
    i. Supports healthcare systems across the four UK nations in offering the opportunity to participate in genomic research to every individual receiving a genomic test in the NHS.
    ii. Monitors and reports on participation in genomic research, promoting diversity and inclusion in recruitment and data collection.
    iii. Establishes a framework for genomic research involving clinical and laboratory resources, to ensure ethical and responsible conduct.
4. UKRI should work with industry partners and across research councils to establish an education and training programme for PhD students and post-doctorates to support upskilling in genomic-driven research approaches.

Using genomics to improve disease prevention and patient care

5. Genomic medicine services across the four UK nations should standardise processes, referral pathways and timelines for genomic testing, ensuring testing is delivered in clinically relevant timelines. For England, this should also include coordinated working with Community Diagnostic Centres on processes and flows for diagnostic genomic and pharmacogenomic testing.
6. Genomic medicine services across the four UK nations should publish annual reports on performance metrics and user feedback from healthcare professionals, industry, patients and their families. This should include a comprehensive overview of testing capabilities, turnaround times, and timelines for service improvement, to raise awareness of current and future capabilities.
7. The healthcare systems in all four UK nations should work with industry and regulators to enhance the current horizon scanning function for genomic advances and technologies, to ensure the genomic medicine services adopt new innovations, in line with global trends.
8. Genomic medicine services across the four UK nations should establish a clear process for adding new tests and increasing uptake of newborn screening, ensuring appropriate support is in place for families.
9. Genomic medicine services across the four UK nations should work with Royal Colleges and industry to disseminate and encourage uptake of genomics education materials and resources, including the NHS Genomics Education Programme and Royal College of GPs Genomics Toolkit.
10. The healthcare systems in all four UK nations should incorporate the needs of the genomics workforce in their long-term strategic workplace planning, ensuring there is a recruitment, retention and development strategy to grow the genomic medicine service workforce.

Introduction: The UK's genomics landscape

The COVID-19 pandemic has demonstrated the UK's world-leading genomic capabilities. In the early stages of the pandemic, the UK Government established a vast diagnostic network, with dedicated high-throughput facilities in the form of Lighthouse laboratories, set-up to conduct NHS Test and Trace COVID-19 PCR testing.[5] At the Alderley Park Lighthouse Laboratory alone, over 1,000 scientists were trained, who performed over 23 million COVID-19 PCR tests between March 2020 and March 2022, when the laboratory was ultimately demobilised.[6] Cumulatively as of 28th September 2022, the UK had conducted over 500 million PCR tests for the COVID-19 virus, SARS-CoV-2[7] and completed and uploaded over 2 million whole genome sequences for SARS-CoV-2 to the international database GISAID.[8]

The COVID-19 COG-UK also provided a network for SARS-CoV-2 genome sequencing and analysis in the first 18 months of the pandemic, helping to advance our scientific understanding of COVID-19 and informing policy decision-making. The consortium has since refocused on research, data linkage and analysis, and training, aiming to create new value from existing data by working with the UK Health Data Research Alliance and the Outbreak Data Analysis Partnership, to link SARS-CoV-2 genomic data to other datasets such as human genome data.[9] In a report evaluating COG-UK, RAND highlights that the data linkage undertaken was critical in informing and iterating the UK's clinical and research response to the pandemic and that its approach to sequencing and analysis influenced international practice.[10]

The response to COVID-19 underscored the UK's existing strengths in genomic capability that were evident before the pandemic and has left an important legacy on which to build.

In 2013, the 100,000 Genomes Project was set up to sequence and study the role genes play in cancer and rare diseases, jointly led by Genomics England and NHS England, and supported by the National Institute for Health and Care Research (NIHR), the MRC, the Wellcome Trust, Cancer Research UK (CRUK) and the Devolved Nations.[11] The initiative set out to sequence 100,000 genomes from around 85,000 NHS patients living with a rare disease or cancer, who consented to having their samples analysed for clinical and research use.

By December 2018, Genomics England and NHS England in partnership with Illumina and other key delivery partners, had successfully sequenced 100,000 whole genomes across the UK, making it the first country globally to conduct whole genome sequencing at scale in healthcare.[12] Patient organisations interviewed by the ABPI, report that project participants found it a very positive experience, with clear aims and benefits.

This project has led to a new rare disease diagnosis for 25% of participants,[13] identified a potential therapy or clinical trial for ~50% of cancer cases[14] and created the NGRL, which to this day provides data for researchers to learn more about health and disease.

Alongside the 100,000 Genomes Project, is the large-scale UK-wide research initiative, UK Biobank, which aims to improve our understanding of prevention, diagnosis and treatment of many serious and life-threatening conditions.[15]

UK Biobank offers a rich dataset on 500,000 people living in the UK, with genotyped blood samples and accompanying urine, blood and saliva samples from consented participants recruited between 2006 and 2010.

In September 2019, a consortium of government, charity and industry (Amgen, AstraZeneca, GlaxoSmithKline and Johnson & Johnson), came together to fund whole genome sequencing of 450,000 participants,[16] with the aim of deepening the understanding of the interplay between genetic and environmental drivers of health and disease. Two years later, data on the first 200,000 participants was made available to researchers, representing the largest single release of whole genome sequencing data. Data on the rest of the cohort is expected to be available in 2023.[17]

UK Biobank's impact over the last 20 years has been far reaching, with advances made in the understanding and treatment of sepsis, the link between diet and people's health, and the understanding of the clinical and genetic factors that affect the outcome and spread of COVID-19.[18] It has also supported medicine discovery and strengthened ties between industry and academia, through an Exome Sequencing Consortium with biopharmaceutical companies.[19]

With over 30,000 global registered users from over 100 countries producing over 6,000 scientific published papers to date, UK Biobank is a highly sought-after resource, driving ground-breaking research in health and disease, across the global research community.[18]

Beyond these bespoke and world-leading initiatives, the UK continues to advance the genomic medicine services across all four UK Nations.

In **England**, the NHS Genomic Medicine Service (GMS)[20] provides a single, national, coordinated approach to genomic testing and genomic research, supported by:

- A network of seven Genomic Laboratory Hubs (GLHs), which coordinate genomic testing within their regions for rare disease and cancer services.
- The National Genomic Informatics Service (NGIS), which is used to register, consent and refer patients, and enter and track sample test results.
- The National Genomic Test Directory (NGTD), which outlines the range of genomic tests available through the GMS as well as patient eligibility for testing.
- The NHS GMS Alliances, which oversee and coordinate the embedding of genomics into mainstream clinical care.
- The NHS Genomic Medicine Service Research Collaborative, which brings together NHS England, Genomics England, the NIHR and other partners to facilitate genomic research on a national scale.

Overseen by the Genomics Clinical Reference Group, the GMS aims to sequence 500,000 whole genomes by 2023/24, extending access to genomic testing and early detection to enable diagnosis and treatment for people with rare diseases and cancer in England.

In Wales, the All Wales Medical Genomics Service (AWMGS)[21] provides specialist genetic services to individuals and families with, or concerned about, rare genetic conditions. The service is made up of clinical genetics and laboratory genetics, with specialities covering haematology, neurology, cancer genetics, oncology, rare diseases, pre-natal, pathology and pharmacogenomics.[22] The Welsh Government and the Welsh Health Specialised Services Committee (WHSSC) agreed in April 2019 to expand the genomic testing available for patients and their families with rare diseases, with increased funding aimed at improving patient care in Wales.[23]

In Northern Ireland (NI), much progress has been made since the set-up of the NI Genomic Medicine Centre (NIGMC).[24] Since 2015, the NIGMC has been actively involved in the 100,000 Genomes Project, helping to establish infrastructure in NI to sequence genomes of patients with cancer and rare diseases in NI. This is accompanied by the Northern Ireland Regional Genetics Service,[25] which helps diagnose and manage conditions with a genetic basis. The service brings together specialists from prenatal, cancer and neurology genetics, to provide clinical diagnostic expertise, genetic counselling, and information to help patients and their families with treatment decision-making and disease management.

In Scotland, the Scottish Genomes Partnership (SGP) brought together collaborators across government, academia, industry, and the NHS to sequence over 2,500 genomes between 2016 and 2020, for research programmes exploring genetic determinants of health and disease, with a focus on cancer and rare diseases. In addition to contributing to the 100,000 Genomes Project, the SGP Cancer Programme sequenced pancreatic, oesophageal, ovarian and brain cancer samples and developed a novel and comprehensive genomic profiling platform for advanced cancers, which has since been successfully tested in clinical care settings.[26]

Building on the progress made to date in genomic medicine and research, shared commitments for UK-wide implementation over the next 3 years were published in March 2022.[27] These shared commitments will be followed by separate, detailed implementation plans for England, Scotland, Wales, and Northern Ireland.

Furthermore, in October 2022, NHS England published its first genomics strategy, Accelerating Genomic Medicine in the NHS, which sets out the ambitions for embedding genomics in the NHS. The strategy details actions across key priority areas, for the next 5 years.[76]

Public-private partnerships in genomics: UK case studies

The UK's strong position globally in genomics, is in part due to the cross-sector collaborations underway to advance basic scientific understanding and genomic-enabled clinical practice. The following case studies outline how pharmaceutical companies are working across the UK, with the NHS, academia and charities in genomics.

Box 1. Identifying novel drug targets to better treat cancer patients and overcome drug resistance[28]

In 2018, AstraZeneca and Cancer Research UK (CRUK) launched their Functional Genomics Centre. Delivering state-of-the-art functional genetic screens, cancer modelling and big data processing, the centre aims to accelerate the discovery of new cancer medicines. Based at the Milner Therapeutics Institute in Cambridge, the centre is developing novel CRISPR (Clustered Regularly Interspaced Short Palindromic Repeats) technologies to better understand the biology of cancer, creating biological models that may be more reflective of human disease and advancing computational approaches to better analyse big datasets. In 2022, CRUK and AstraZeneca completed their first collaborative CRISPR screen in radiation, to detect genes that increase radiotherapy sensitivity in lung cancer, identifying new potential drug targets.

Box 2. Advancing the understanding of human genetic signatures[29]

In 2020, The University of Oxford's Big Data Institute established a strategic collaboration with Janssen Biotech, one of the Janssen Pharmaceutical Companies of Johnson & Johnson, to accelerate the use of human genetic data to understand how genetic variations can affect the ways in which diseases develop. This collaboration brings together expertise across academia and industry to interpret and translate genetic insight into potential new treatments to improve patient health. Key areas of focus for the collaboration are inflammatory disorders, neuroscience, and population datasets.

Box 3. Identifying new therapeutic targets for osteoarthritis through genome-wide analyses of UK Biobank[30]

With 10 million people in the UK suffering from the condition and 40% of individuals over 70 years old affected, there is a pressing need to support the research and development of therapies for osteoarthritis. In 2019, a cross-sector research consortium set about identifying potential therapeutic targets for this condition. Leveraging resources and assets from the UK Biobank, the arcOGEN consortium and the United Kingdom Household Longitudinal Study, researchers performed a genome-wide meta-analysis for osteoarthritis across ~17.5 million single nucleotide variants in up to 455,221 individuals, identifying several potential novel therapeutic targets for osteoarthritis and highlighting where existing therapeutics could be repurposed as osteoarthritis treatments. This consortium involved GlaxoSmithKline, the Wellcome Sanger Institute, the European Molecular Biology Laboratory (EMBL) and research institutes from across the UK, Germany, Australia, USA, Canada and Greece.

Box 4. Introducing histology-independent therapies (HITs) to the NHS[31]

Histology-independent therapies (HITs), also known as tumour-agnostic therapies, are a class of medicines used to treat solid tumours expressing a particular genomic alteration, regardless of where in the body the cancer originated. For some patients, they offer a game changing cancer treatment and were one of the initial priority categories of focus for the Accelerated Access Collaborative (AAC),[32] a cross-sector partnership aimed at accelerating access to transformative health technologies, of which the ABPI is a partner. The AAC has worked to identify HITs that could be available for use in the coming years and supported NHS England & Improvement in focussing efforts on making these treatments and required diagnostic genomic testing available to patients. Bayer's Larotrectinib▼ was the first of its kind to be recommended for use in the Cancer Drugs Fund (CDF) by National Institute for Health and Care Excellence (NICE).[31] This treatment is indicated for adults and children with advanced neurotrophic tyrosine receptor kinase (NTRK) fusion-positive solid tumours, for which there are currently no satisfactory treatment options. Following the initial phased introduction, NTRK testing is now routinely commissioned and is on the National Genomic Testing Directory, taking advantage of the available genomic testing capacity across the 7 Genomic Laboratory Hubs in England, where it will have an impact as part of the Genomics Medicines Service (GMS).[33] Despite the disruption and challenges of the pandemic, the AAC's programme has successfully delivered several key achievements, including the implementation of a diagnostic genomic testing pathway for HITs, and work to support uptake is ongoing.

▼This medicinal product is subject to additional monitoring. This will allow quick identification of new safety information. Healthcare professionals are asked to report any suspected adverse reactions.

Box 5. Designing and implementing services across the Genomic Laboratory Hubs[34]

Non-Small Cell Lung Cancer (NSCLC) is the most common type of lung cancer, accounting for over 85% of cases.[35] To improve disease management in patients diagnosed with NSCLC in England, Amgen has partnered with the North-West and South-West Genomic Laboratory Hubs to design and implement a service optimisation project. The aim of this project is to support the optimisation of clinical pathways, which includes standardising access to and turnaround times for NSCLC diagnostic testing. The project will map the pathology pathway and develop guidelines and a "case for change" to improve adoption and uptake of genomic testing locally and nationally.

Box 6. Introducing new molecular techniques for cancer into routine clinical practice[36]

The NHS Greater Glasgow & Clyde (NHS GGC) molecular diagnostic pathway development Joint Working Project (JWP), co-funded by GGC, Novartis and Celgene (now part of Bristol Myers Squibb), employed two R&D scientists and purchased a next-generation sequencer, to bring new molecular techniques into routine clinical practice and evaluate the use of next-generation sequencing (NGS) to perform tumour-specific tests. The aims of the JWP were to support better diagnosis, encourage clinical trials and improve access to innovative treatments for patients. The JWP introduced somatic BRCA NGS to perform tumour-specific tests on tumour tissue (primarily breast and ovarian), allowing for the development of new quality and performance parameters to establish acceptable results. This led to successful implementation of the test into routine diagnostic services. The JWP also helped develop a Myeloid NGS Panel for Leukaemia, containing 28 targets developed for Acute Myeloid Leukaemia, increasing the chance of obtaining useful genetic information upon screening. As a result of these new techniques, around 18% of diagnoses, 20% of prognoses and 19% of patient management approaches were altered, paving the way also for a multiple cancer type test to come into service.

Realising the UK's potential in genomics by reducing variability in access to genomic testing

Reviewing availability and access to biomarker testing, in particular genomic testing, in the UK and internationally gives an insight into how patients are benefitting from precision medicine technology. Biomarker testing is a way to look for biological changes in genes, proteins and metabolomic profiles, which cause disease. Genomic biomarkers (or gene alterations) are detected using genomic testing, which is mostly used currently in cancers and rare and inherited conditions.[37]

In February 2021, the IQN Path, ECPC and EFPIA published findings of a consortium study looking at cancer biomarker testing in the UK and EU27.[38] The consortium aimed to identify barriers to biomarker testing and develop policy recommendations to ensure all eligible cancer patients had access to testing. **They found that access to high-quality cancer biomarker testing is inconsistent across Europe (EU27 + UK) and is driving inequalities in patient access to tailored treatments.**

Drawing on a wide range of secondary sources, surveys of over 1,500 patients and nearly 60 expert interviews, the research reviewed access to and quality of biomarker testing conducted by immunohistochemistry, single molecular testing (e.g. for BRCA, Epidermal Growth Factor Receptor (EGFR) and NTRK), NGS (targeted up to 50 genes or comprehensive) and liquid biopsies.

Regarding access to single- and multi-biomarker testing, the study found there was significant variation across Europe, with the UK scoring medium. This was due to variance in laboratory capabilities and time taken to introduce and conduct biomarker tests. Figure 1 summarises access to and quality of biomarker testing in the UK relative to EU comparator countries, in terms of access to single biomarker tests, multi-biomarker tests and biomarker test quality.

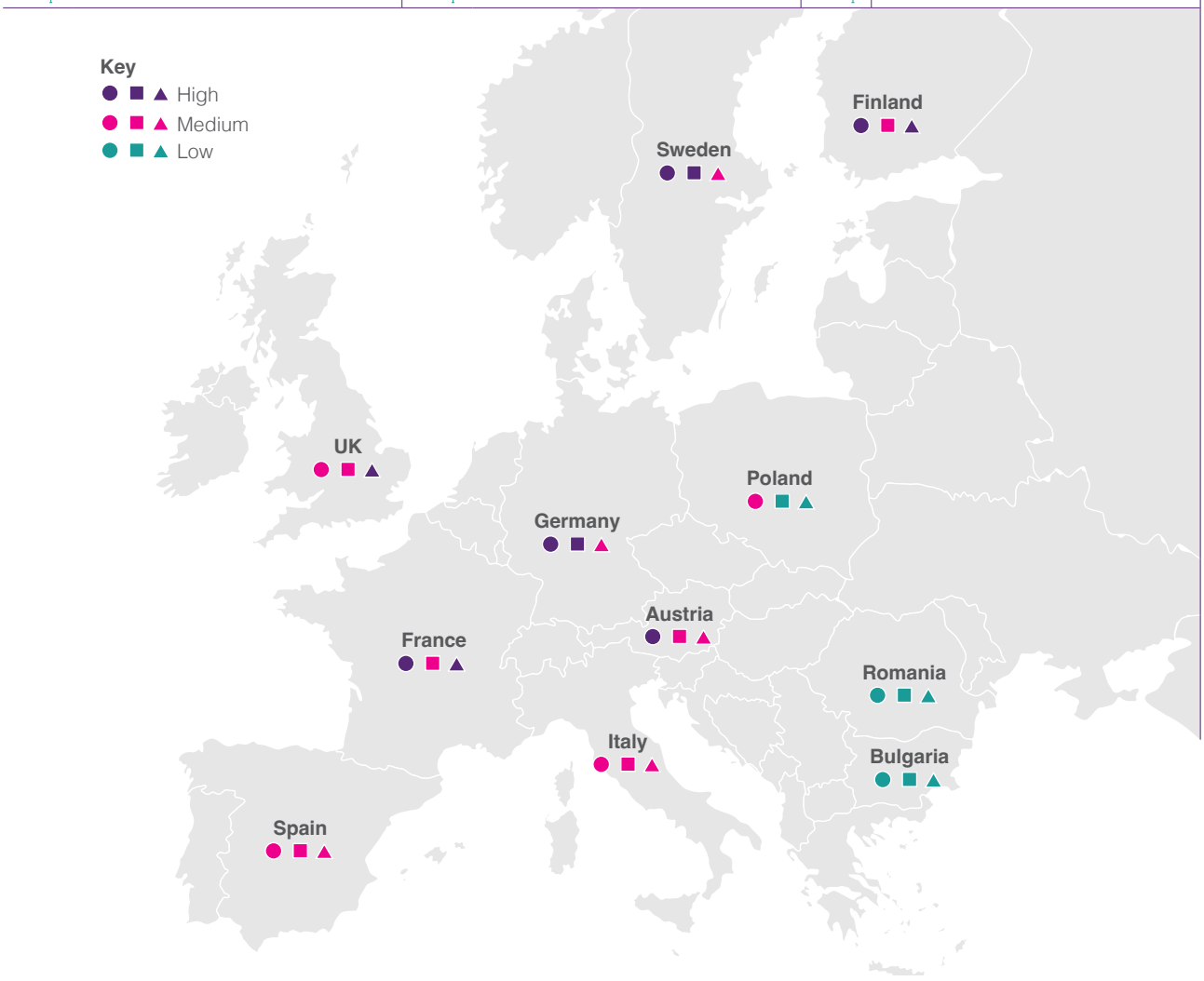
For example, test turnaround times for NGS multi-biomarker panels and EGFR, which is a commonly used biomarker for non-small cell lung cancer and colorectal cancer, were both slower in the UK than in the EU (average turnaround time for NGS multi-biomarker panel testing in UK 20 days and in EU 14 days; average turnaround time for EGFR testing in UK 16 days and in EU 11 days).

In countries such as Sweden and Germany, there is better coordination across networks, greater awareness of testing availability and processes, clearer and well-established referral pathways for patients, prompt and uniform testing, and rapid introduction of testing alongside new medicines; all of which put them ahead of the UK in terms of ability to provide timely and accurate biomarker testing to patients.

Overall, the study concluded that access to biomarker testing in the UK has much room for improvement, particularly in terms of reducing variability in access to genomic tests and speeding-up the availability of these tests in conjunction with the introduction of new medicines. The establishment of the NHS GMS, alongside government policies and funding, provide the infrastructure and framework to deliver precision medicine, however lessons can be learnt from countries such as Sweden and Germany, to ensure the UK delivers timely and accurate testing for patients.

Figure 1. Access to and quality of biomarker testing in the UK relative to EU comparator countries[38]

○ Access to single biomarker tests		□ Access to multi biomarker tests		△ Biomarker test quality	
High	Sweden	High	Sweden	High	Finland
	Finland		Germany		France
	France	Medium	France		UK
	Germany		Finland	Medium	Germany
	Austria		Austria		Austria
Medium	Spain		Spain		Spain
	Italy		Italy		Sweden
	UK		UK		Italy
	Poland	Low	Romania	Low	Romania
Low	Romania		Bulgaria		Bulgaria
	Bulgaria		Poland		Poland



Source: EFPIA, 2021. Unlocking the potential of precision medicine in Europe. Available online here.
This figure does not include all countries listed in the EFPIA report

Harnessing genomics for research and development

Research and healthcare delivery are in a virtuous cycle, with research in genomics helping us to better understand health and disease, evolve genomic technologies and develop new medicines to treat patients. Genomic testing and sequencing across healthcare systems in turn generates data to inform the next cycle of research and development.

From a global pharmaceutical industry perspective, the UK must continue to improve this virtuous cycle to ensure success in advancing its genomic capabilities and delivering precision medicine for UK patients. Ensuring genomic data and clinical advances feed the discovery, development and launch of new medicines and closing the gap between laboratory bench discovery and bedside application are essential. As such, there are two key aspects the UK needs to focus on over the coming years to unlock the full potential of its genomic assets for research and development:

i. Using genomics to understand health and disease and discover new medicines.

ii. Using genomics to develop new medicines and treatments in clinical trials.

Understanding health and disease and discovering new medicines

Functional genomics is a rapidly evolving field, investigating the link between the information contained in our genome and its effect on health and disease, which is accelerating innovations in the discovery of new medicines and technologies.[39]

Novel gene editing technologies used for functional genomics studies, such as Nobel prize winning technology CRISPR,[40] enable researchers to simulate human disease states by silencing or activating genes in specific cell lines or animal models. These models serve as informative preclinical models before progressing into clinical trials, helping to improve the success rate of new medicines in clinical trials. Real-world genomic data, captured through diagnostic and pharmacogenomic testing, is also informing the drug targets of tomorrow, identifying novel targets that contribute to disease and variation in response to treatments.

With centres of excellence such as the Wellcome Sanger Institute[41] and public-private partnerships, such as the Open Targets initiative,[42] the UK supports the bringing together of pharmaceutical companies and academics, to harness genomics to understand how drivers of health and disease and identify novel drug targets.

As described earlier in this report, other initiatives such as the 100,000 Genome Project,[11] UK Biobank[15] and Genomics England's NGRL,[43] also provide large genomic datasets to researchers world-wide, helping to improve the scientific community's understanding of health and disease and identify novel drug targets (see case study in Box 3, page 9).

Coordination across these initiatives could be improved however, with a need to strengthen the linkage across genomic and clinical datasets, to understand how genomic alterations drive patient outcomes.

Furthermore, industry involvement in flagship programmes and access to datasets can be challenging, particularly for those who lack sufficient resource and capacity to navigate the complex data landscape and secure exclusive participation in flagship programmes.[44]

To address the challenge around data linkage, the government's data strategy, Data Saves Lives was published in June 2022.[45] The Department for Business, Energy and Industrial Strategy (BEIS), and the Office for Life Sciences (OLS) commits to continue collaborating with UK Biobank, Our Future Health, the Health Data Research UK Hubs and the Innovate UK digital pathology, imaging and artificial intelligence centres, to improve interoperability and create safe environments to access and analyse genomic datasets. The ABPI also welcomes the commitments in NHS England's strategy to develop an interoperable informatic data and digital infrastructure for genomic data and expand the use of NHS generated genomic data to support approved research.[76]

On linkage across research programmes, the Genome UK shared commitments for UK-wide implementation,[27] published in March 2022, lays out ways in which England and the Devolved Nations will coordinate via the Genome UK Implementation Coordination Group and its Research Working Group, to identify opportunities to bring together genomic research programmes and research infrastructure, to better harness genomic and clinical data for research. NHS England also announced in their first genomics strategy, that during 2023/24, the NHS will establish Genomic Networks of Excellence, to bring together system partners to deliver genomic research from bench to bedside. This is welcomed by the ABPI, in particular the plan to include industry in this initiative.

In addition, the former Health Secretary speaking at the ABPI Annual Conference in 2021,[46] announced £37 million for genomics projects and for data-driven initiatives delivered through Genome UK, and set out how new support for the UK Functional Genomics Initiative will drive ground-breaking new approaches to improve our understanding of how genetic changes cause disease.

The ABPI welcomes the government's commitment to transforming the UK's genomic data landscape and enhancing the UK's genomics offer for research and development.

To ensure these commitments translate into a globally competitive offer, the MRC-UKRI should ensure the UK Functional Genomics Initiative builds on existing infrastructure and best practice. To inform this, the MRC-UKRI should work with industry, academia, and charities to review the UK's current functional genomics ecosystem relative to global trends, to identify opportunities for scale and join-up. This can help ensure public funding is used to create a globally competitive offer that leverages further industry investment.

To drive greater connectivity across genomic and health data assets, the UK Government should fully deliver the Data Saves Lives and Genome UK strategies, ensuring implementation plans drive interoperability and connectivity between flagship research programmes and across genomic and health data assets.

As part of the Genome UK commitments to support industrial growth in the UK and facilitate entrepreneurship and innovation for projects and companies of all sizes, BEIS and OLS should work with the life sciences sector to establish principles and standards for industry involvement and access, to maximise diversity of industry participation in current and future research programmes and support industry access to genomic data assets.

Recommendations:

1. The MRC-UKRI should partner with industry, academia, and charities to scope and deliver the UK Functional Genomics Initiative, ensuring plans build on the UK's existing expertise and infrastructure.
2. The UK government and NHS should fully deliver the commitments in the Data Saves Lives and Accelerating Genomic Medicine in the NHS strategies, to drive interoperability and connectivity between flagship research programmes and across genomic and health data assets.

Developing new medicines and technologies in clinical trials

The NHS GMS in England and its Devolved Nations equivalents provide critical infrastructure to bridge the gap between laboratory bench discovery and bedside application.

Earlier in this report, we highlight the importance of linkage across genomic and wider clinical datasets with research programmes, which ensures that data on genomic variants in health and disease can inform the discovery of novel medicines and treatments.

In addition to this, genomic data when linked with electronic health records and wider clinical datasets can help the development of novel medicines and treatments, by identifying the clinical consequence of particular genomic alteration(s) and matching patients with new targeted medicines being developed in clinical trials (based on their genomic profiles).

Genome UK acknowledges the huge potential genomic data offers in making it easier for patients to be enrolled in clinical trials tailored to their biomarkers, across all disease areas. The NIHR Bioresource[47] and the NGRL[43] play a crucial role in facilitating opportunities for patients to participate in genomic clinical research and capturing NHS GMS data in one place, respectively. However, the means of recruiting a clinical trial participant in the UK, with a defined genomic profile, is complex, with multiple tools and resources across the UK used to identify eligible research participants.

Alongside the NIHR Bioresource and the NGRL, the UK also has Be Part of Research,[48] the Experimental Cancer Trial Finder (ECTF),[49] the Clinical Practice Research Datalink (CPRD),[50] and the Find Recruit and Follow up service.[51] With no single front door and lack of clarity on the overlap between the underpinning datasets of these services, navigating the system and recruiting research participants is incredibly challenging and complicated.

Furthermore, patient organisations interviewed by the ABPI, have reported the consent process for research participation as being a very time-consuming and off-putting process, and flag the need for a more standardised approach, that enables patients to be contacted about research opportunities as part of the diagnostic and clinical genomic testing process.

The ABPI welcomes the NHS GMS Research Collaborative and the commitment to establish NHS Genomics Networks of Excellence,[76] which both aim to bring system partners together to facilitate genomic research across the NHS. How the collaborative and networks will support and streamline genomic clinical trial delivery, is however unclear.

Diversity relative to the UK population is also missing, with under-served groups poorly represented in datasets. This leads to disproportionate representation of certain groups in flagship research programmes and clinical trials, perpetuating the inequality of access to cutting-edge genomic treatments across the UK population and preventing research outputs from being as “real-world” relevant and useable as possible.

The ABPI welcomes Genomics England’s Diverse Data initiative,[52] for which the ABPI is a community partner, and Genes and Health,[53] which are focussed on improving patient outcomes within genomic medicine by engaging diverse communities in research programmes and providing researchers with the tools needed to be more inclusive in their research design and data curation.

With industry seeing a year-on-year decline in share of patient recruitment in the UK,[54] the implementation of Genome UK and NHS England’s genomics strategy needs to urgently deliver on its commitments to make participation in genomic-enabled clinical trials easier and more inclusive for patients.

Recommendations:

3. Genomic medicine services across the four UK nations should work with the research sector to develop a UK-wide Genomic Research Collaborative which:
   i. Supports healthcare systems across the four UK nations in offering the opportunity to participate in genomic research to every individual receiving a genomic test in the NHS.
   ii. Monitors and reports on participation in genomic research, promoting diversity and inclusion in recruitment and data collection.
   iii. Establishes a framework for genomic research involving clinical and laboratory resources, to ensure ethical and responsible conduct.

Figure 2. A proposed national approach for genomic-enabled research and development

Source: The ABPI

Growing the genomics research workforce

Addressing the current and future skills needs in research is essential to grow the UK's genomics workforce and deliver the benefits of precision medicine to patients. Training in fields such as computational sciences, bioinformatics and data science is required to equip the UK workforce with the right skills to maximise the potential of using and interpreting biological, genomic and clinical data for research purposes.

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The increased use of data-driven approaches in biomedical research and growing demand for a skilled workforce, puts bioinformaticians, statisticians and data analysts in high demand across the life sciences sector. The ABPI's recent biopharmaceutical industry skills report identified high priority skills gaps across these same computational roles.[55] The continuing rapid growth of genomic technologies and the UK's ambition to implement a genomics-based healthcare system, is resulting in an ever-increasing demand for support in genomic data generation, analysis, and interpretation, which is placing a strain on the current workforce. These skills gaps must be addressed to scale-up genomic approaches and technologies across the life sciences and health sectors.

To enhance its genomics offer and ensure the system moves from a data-rich to an information-rich environment, the UK needs to develop and incentivise participation in training programmes across the educational pathway and talent pipeline, from secondary school curricula through to PhD and post-doctoral programmes. This will ensure the future workforce has a solid foundation in genomics, with good awareness and understanding of genomic approaches and technologies, matched with strong analytical capabilities.

Pharmaceutical companies are important partners in workforce upskilling and development, with AstraZeneca's announcement on funding 55 additional PhD studentships with the University of Cambridge[56] just one recent example of how companies are working with academic funders and institutes to support research students to develop translational skills, such as those needed in genomics.

The ABPI welcomes the government's funding announcement of £25 billion for UKRI, who have committed to support world-class people, careers, ideas and innovation in their new strategy for 2022-2027.[57] Building on the success of schemes such as the Emerging Skills Project for manufacturing and engineering, UKRI should work with industry partners and across research councils, to establish an education and training programme for PhD students and post-doctorates which supports upskilling in translational, genomic research approaches.[58]

Recommendations:

4. UKRI should work with industry partners and across research councils, to establish an education and training programme for PhD students and post-doctorates which supports upskilling in translational, genomic research approaches.

Using genomics to improve disease prevention and patient care

Until recently, genomics has mainly been applied in the context of specific conditions, such as rare and hereditary diseases. With an increasing understanding of the genetic drivers of health and disease, healthcare systems have begun using genomics more routinely in care pathways in other conditions and in the context of disease prevention and early detection.

Implementing genomics in medical practice to deliver precision medicine

The launch of the 100,000 Genomes Project,[11] set the UK on a path to genomic-enabled precision medicine. Since then, the NHS GMS in England has made much progress in establishing genomic testing across the NHS. Seven GLHs and seven GMS Alliances have been set-up, the NGTD launched and data from research-consented patients having whole genome sequencing through the NHS GMS has been made available by Genomics England in the NGRL. The Devolved Nations have been on a similar journey, making genomic testing available to patients with cancer and rare and inherited conditions across the UK.

This infrastructure has put the UK in a strong position to implement genomics at scale across the nation. Hindering this progress, are the many challenges faced by industry, patients, and their families, when engaging with the services.

As described earlier in this report, the major challenge in the UK is the variable operating environment across the genomic medicine services.

Lack of consistency in sample requirements, laboursome methods for requesting a test (often paper-based), limited capacity and capability (particularly in cancer and pathology), poor digital infrastructure to link genetic test results to electronic health records and ambiguity around the process for updating the testing directory, are just some of the technical and operational challenges described.

These deficiencies, seen more prominently in some areas of the country than others, are leading to high test failure rates and long turnaround times across services, with test results failing to align with clinical decision-making timelines. This perturbs healthcare professionals from referring patients for testing and delays the start of an appropriate genomic treatment, ultimately depriving patients of critical diagnostic and clinical care.

It is clear, that in order to establish a more uniform service offering across the UK, focus is urgently needed on harmonising and standardising approaches to genomic testing across the UK. This would also help tackle health inequalities.

Coupled with this, the performance and capabilities of these services need to be more transparent, with robust assessment, monitoring, and reporting processes. This is essential for service improvement and for industry and healthcare professionals to navigate the service offer and deliver care confidently, in line with treatment guidelines.

Pharmacogenomics, which looks at how a person's genes affect the way they respond to treatments, has a key role to play in influencing treatment guidelines also. A recent report from the Royal College of Physicians and British Pharmacological Society on personalised prescribing highlighted the role that pharmacogenomic testing was starting to play in the NHS in guiding safe, effective treatment decisions, for example for certain chemotherapy treatment in colorectal and breast cancer.[59]

The report also highlighted however, that wider implementation of pharmacogenomic testing was being hampered by several factors, including poor availability of tests outside of specialist settings, the need to train healthcare professionals, and a lack of information, amongst others. The report recommended addressing these factors and implementing pharmacogenomics skills, capabilities, education, and support across both primary and secondary care settings.

Furthermore, the needs of patients and their families at the heart of this, are not being met, with communication severely lacking. Through interviews with the ABPI, some patient organisations reported similar challenges to industry, in terms of long turnaround times and variability and fragmentation in the service offer across the UK, which creates a postcode lottery in access to high-quality care. They also stressed that the genomic test result is often viewed as the end of the road, whereas the diagnosis often marks the start of a long journey for patients and their families, particularly for those with rare and inherited conditions.

Given the reform underway within the NHS in England, with the introduction of Integrated Care Systems (ICSs) and 40 new Community Diagnostic Centres (CDCs),[60] now is the time to create a seamless pipeline for genomic testing, integrating the NHS GMS into clinical pathways and patient care. The ABPI welcomes NHS England's commitments to delivering equitable genomic testing for improved prediciton, prevention, diagnosis and precision medicine.[76] Complemented with engagement across the Devolved Nations, to support best practice sharing and coordinate service delivery, the UK will be equipped to implement genomics at scale across the nation.

Recommendations:

5. Genomic medicine services across the four UK nations should standardise processes, referral pathways and timelines for genomic testing, ensuring testing is delivered in clinically relevant timelines. For England, this should also include coordinated working with Community Diagnostic Centres on processes and flows for diagnostic genomic and pharmacogenomic testing.
6. Genomic medicine services across the four nations should publish annual reports on performance metrics and user feedback from healthcare professionals, industry, patients and their families. This should include a comprehensive overview of testing capabilities, turnaround times, and timelines for service improvement, to raise awareness of current and future capabilities.
7. The healthcare systems in all four UK nations should work with industry and regulators to enhance the current horizon scanning function for genomic advances and technologies, to ensure the genomic medicine services adopt new innovations, in line with global trends.

Establishing prevention and early detection capabilities

A paradigm shift in the healthcare system is underway, with a move towards prevention and early detection. As genetics contributes to health and disease, the ability to detect genetic variants and mutations early can help in predicting an individual's risk for disease and preventing disease progression. There are several national prevention and early detection initiatives underway, which are driving forward the UK's capabilities in prevention and early detection.

Our Future Health is a flagship research programme, which aims to bring together public, charity and private sectors to "help people live healthier lives for longer through the discovery and testing of more effective approaches to prevention, earlier detection and treatment of diseases".[61] Established with £79 million of seed-funding from UKRI, the programme has already leveraged over £140 million in industry funding,[62] indicating the life sciences sector's interest and commitment to this agenda.

Genomics England's Newborn Genomes Programme, is another ambitious research programme, aimed at accelerating diagnosis and access to treatments for rare genetic conditions.[63] The programme will sequence up to 200,000 babies' genomes and build on the principles of the newborn screening programmes across the UK,[64,65,66,67] which currently include a newborn blood spot test for rare and inherited conditions including sickle cell disease and cystic fibrosis.

In 2020, the NHS announced it was piloting a revolutionary blood test that detects more than 50 cancers.[68] **The Galleri blood test**, developed by healthcare company GRAIL, is being piloted in 165,000 patients. This includes 25,000 people with cancer symptoms who will be offered diagnostic testing post-referral, and 140,000 participants aged 50 to 79 who have no symptoms who will have annual blood tests for three years to check for any molecular changes.

The ABPI welcomes these initiatives and outlines here some of the practical and ethical issues that must be addressed as these programmes progress and look to scale across the NHS.

Firstly, the research outputs from flagship programmes such as Our Future Health, stand to benefit the global research community, and as such, partners from across the life sciences industry should have the opportunity to get involved and utilise the data generated in their R&D programmes. However, due to the need to be a funding partner on the programme to get access to the data, companies (particularly SMEs) that are unable to financially contribute are locked-out of the programme.

Secondly, with around 2.8 million people in the UK diagnosed with a rare genetic condition, the UK should be moving faster to implement and scale genetic newborn screening. The Archangel Newborn Screening Review[69] found that the UK is lagging behind its European counterparts, such as Italy, Iceland, and Poland, who screen up to four times as many conditions as the UK, where babies are only tested for 9 conditions. This means that babies in the UK are missing out on critical diagnostic testing and potential life-saving treatments.

To help address these challenges, a public dialogue is needed to raise awareness of the importance of programmes such as these and inform the public and expecting parents of the benefits of newborn screening.

The healthcare systems in all four UK nations should establish a clear process for adding new tests to newborn blood spot testing and increasing uptake of newborn screening, ensuring appropriate support is in place for families.

For the Newborn Genomes Programme, Genomics England have carried out extensive engagement with members of the public, expectant and new parents, and other stakeholders to inform the development and scope of the programme, with further engagement planned if the programme is successful. This is an exemplar for engagement and co-production with patient and public communities, that should be modelled in future research programmes.

The ABPI welcomes the commitments of the UK Rare Disease Framework[70] and England's 2022 action plan for implementing the framework,[71] which includes plans to improve how decisions are made on newborn screening for rare diseases, pilot new approaches for patients with undiagnosed conditions and support rapid access to drugs for patients with rare diseases in the NHS.

Recommendations:

8. Genomic medicine services across the four UK nations should establish a clear process for adding new tests to newborn blood spot testing and increasing uptake of newborn screening, ensuring appropriate support is in place for families.

Growing the genomics healthcare workforce

The UK Government's commitment to scale genomic medicine and preventive healthcare across the NHS, brings with it the need for transformation of the healthcare workforce, with implications for many professions. With the increasing use of genomics in prevention, early detection, diagnostics and routine care, there is an increasing need for healthcare professionals to have up-to-date genomics knowledge and for staff to be equipped with the skills and confidence needed to utilise genomic services and interpret and discuss the findings of genomic testing with patients and their families.

The ABPI welcomes the commitments in Genome UK to identify and plug skills shortages in the NHS workforce and recommends governments across the four UK nations to work with industry to address these. As industry has a wealth of expertise in the research, development and use of genomic medicines and treatments, industry is well equipped to help upskill clinicians and develop education material. To harness this expertise, a partnership has been established between the ABPI and Health Education England (HEE)'s Genomics Education Programme (GEP).[72]

The GEP aims to upskill healthcare workers across professions and specialties, to adopt and utilise genomic medicine for diagnosis and management of patients. The programme has four pillars:

1. Identifying workforce needs
2. Building and joining networks across the country
3. Helping educate and develop the workforce
4. Increasing awareness of genomics across healthcare

To date, the GEP has funded nearly 2,500 NHS staff to undertake the Master's in Genomics Medicine programme,[73] delivered through 7 higher education institutes, developing GeNotes,[74] an online 'just in time' educational tool to assist clinicians in decision-making, and creating an online education hub[75] of teaching resources, videos, podcasts, and bitesize modules.

The GEP's educational resources have been viewed over 650,000 times, which is encouraging, however, healthcare professionals across the NHS need more training and support if the UK is to succeed in scaling-up genomic medicine across the NHS, particularly given the current staffing shortages and workforce crisis.

For those already working in the genomics service, technical and operational upskilling in areas such as referrals, sample preparation, and processing, is needed to improve testing accessibility, quality, and consistency across the national service. Pathologists are key to delivering a genomics service, as those who conduct the laboratory testing, and hence must be involved in service improvement and policy development at local, regional, and national levels.

For those not currently engaged with the genomics service, a focus is needed on raising awareness, recruiting into the speciality and addressing skills gaps. To increase uptake of genomic medicines and diagnostics, non-specialists across primary and secondary care need to be trained in genomics and made aware of the services that exist across the system, informing them how best to use the available service.

Additionally, to scale the UK's genomic medicine service, investment in the genomic counsellor workforce will be needed, to support patients and their families engaging with the service to navigate healthcare decision-making.

Given the rapidly evolving nature of genomics, educational resources need to be kept up to date, to reflect changes in the field and introduction of new technologies and medicines. Through the ABPI-HEE GEP partnership agreement, pharmaceutical companies have already helped develop new material for inclusion in GeNotes. Many companies are also working directly with GLHs and other providers to help expand capabilities; this however can result in duplication of efforts and add additional strain on service providers.

As a sector that sits at the forefront of innovation, the life sciences industry is well equipped to support in upskilling of the healthcare workforce in genomic advances and is keen to work collaboratively with genomic medicine providers across the UK to do so.

The ABPI welcomes NHS England's commitments to upskill the workforce through the establishment of a new Genomic Training Academy and a future training and development model in partnership with industry. We remain committed to working collaboratively with NHS England, HEE and the Devolved Nations to help raise awareness, support training and coordinate the pharmaceutical industry's engagement.

Recommendations:

9. Genomic medicine services across the four UK nations should work with Royal Colleges and industry to disseminate and encourage uptake of genomics education materials and resources, including the NHS Genomics Education Programme and Royal College of GPs Genomics Toolkit.
10. The healthcare systems in all four UK nations should incorporate the needs of the genomics workforce into their long-term strategic workplace planning, ensuring there is a recruitment, retention and development strategy to grow the genomic medicine service workforce.

Conclusion

The UK has built up global leadership and expertise in genomic capability, with flagship programmes initiated, centres of excellence established and genomic services rolled-out across the NHS. With a national strategy in place, committed to building upon these strengths to advance our genomics offer, the UK is well positioned to become a world-leader in genomics.

The road ahead looks rocky however, with an NHS in crisis bringing into question the healthcare system's capacity to improve and expand its genomic medicine service, and skills and workforce shortages threatening our ability to fully harness our genomic assets to improve patient outcomes.

Currently too few patients benefit from the UK's advancements in genomics, stressing the need to get the basics right, with consistent, timely and reliable genomic testing in place, with appropriate support for patients and their families. Addressing these challenges, along with driving interoperability and connectivity across our scientific and data assets, will transform the UK's genomics offer, into one that is truly globally competitive.

From a global pharmaceutical industry perspective, the health and wealth of the nation can be greatly improved by supporting growth in the life sciences sector - and genomics is one of the key drivers of that. Looking ahead to the next phase of implementation for Genome UK, government and system partners must continue to work with industry to ensure ambition is translated into action, for the benefit of patients, the NHS, and the economy.

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