

PROCESS AND SYSTEMS Embedding genomics across the NHS: a primary care perspective

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ABSTRACT

Primary care remains the point of access to the NHS as well as having key roles in care coordination and prescribing. Therefore, embedding of genomic medicine in the NHS relies on successful implementation into the primary care landscape. Primary care is currently facing considerable challenges, including increasing numbers of patients and consultations per GP, multiple health conditions and polypharmacy, all contributing to increasing workload within a resource-constrained system. Although genomic medicine has enormous potential to benefit patients, its successful implementation demands alignment with existing skills and working practices, development of underpinning informatics infrastructure, integration into care pathways with consideration of commissioning and leadership. Here, we set out current initiatives and future strategies to support primary care colleagues in the delivery of genomic medicine, covering issues of workforce development and education, primary care leadership, and data and digital considerations.

KEYWORDS: primary care, genomic medicine, implementation, education, workforce development

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Introduction

Rapid advances in, and falling costs of, genomic technologies over the past two decades have led to increasing hope in the enormous potential of genomic medicine to improve disease diagnosis and prevention, and targeted treatments for patients across healthcare settings. *Accelerating Genomic Medicine in the*

NHS¹ sets out a strategy for implementation within the UK NHS across four priority areas: embedding genomics in end-to-end clinical pathways from primary and community care through to specialist and tertiary care, delivering equitable genomic testing, developing data and informatics infrastructure, and interfacing closely with research and innovation to ensure rapid implementation.

Primary care remains the point of access to the NHS as well as having key roles in care coordination and prescribing. Thus, the embedding of genomic medicine in the NHS relies on its successful implementation into the primary care landscape. Primary care is facing considerable challenges, such as increasing numbers of patients and consultations per GP, multiple health conditions and polypharmacy, all of which contribute to increasing workloads within an already resource-constrained system. Although genomic medicine has enormous potential to benefit patients, its successful implementation requires alignment with existing skills and working practices, development of an underpinning informatics infrastructure, and integration into care pathways, with consideration of commissioning and leadership.² Here, we highlight current initiatives and future strategies to support primary care colleagues in the delivery of genomic medicine. We discuss issues of workforce development and education, primary care leadership, and data and digital considerations.

Genomics in primary care

As genomic medicine embeds within the NHS, there will be an increase in patients and families who might be eligible for, and benefit from, genomic testing to guide diagnosis and risk stratification, who will receive a diagnosis of a rare disease and, in the near future, who might benefit from genomic testing to guide prescribing. Patients might present with a significant family history or clinical features suggestive of an inherited condition, seeking advice regarding reproductive issues, with a genomic test result from themselves or a family member, or they might have a diagnosis of an inherited condition themselves, which influences the management of other clinical presentations (Table 1).

For patients with a family history of cancer or a known inherited cancer predisposition syndrome (eg BRCA1 or Lynch syndrome), primary care facilitates access to genomic testing for a familial variant and to surveillance and risk-reducing measures, and provides holistic care for other clinical management issues presenting within that context. Rare diseases are individually

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Table 1. Summary of presentation of genomics issues in primary care and related themes from Delphi-style survey seeking expert consensus

Issue	Themes identified
Presentation of genomics issues in primary care: patient led	<ul style="list-style-type: none"> Family history Antenatal query Requesting tests Suggestive or 'red flag' clinical features Existing diagnosis of an inherited condition General concerns, such as implications of pursuing genetic testing
Presentation of genomics issues in primary care: clinician led	<ul style="list-style-type: none"> Family history Ordering or facilitating onward referral for genetic and/or genomic tests Clinical management issues Signposting, such as to patient support groups Recording of test results and data in electronic health record
Required knowledge	<ul style="list-style-type: none"> Core genetic and genomic concepts Specified conditions Referral pathways Risk factors Clinical features and red flags Principles of undergoing a genetic and/or genomic test Role of genomics in common complex conditions, such cardiovascular disease and cancer Ethical and legal issues Communication and consultation issues How to access resources and information for professionals and patients
Required skills	<ul style="list-style-type: none"> Ability to take and utilise family history information Recognition of risk factors and red flags Specific communication and consultation skills Ability to understand and explain when genetic testing might or might not be indicated
Required attitudes	<ul style="list-style-type: none"> Use resources appropriately, including support groups Apply relevant ethical and legal issues Reproductive choices
Applied key skill areas that are core for GPs	<ul style="list-style-type: none"> Core communication, consultation and listening skills Keeping up-to-date Practice non-judgemental patient-centred consulting and skills in reflection Recognise limits of competency and seek advice appropriately

rare but collectively common, with a lifetime incidence of one in 17, with patients often experiencing a protracted 'diagnostic odyssey' before diagnosis. Primary care has a key role in identifying patients at risk of rare disease, facilitates access to care pathways and services while coordinating on-going care. It already performs risk assessment for multifactorial conditions, such as cancer, through utilisation of family history information; imminent is the application of genomic information (polygenic risk) within risk stratification to guide clinical management.³ Pharmacogenomics describes the use of genomic information to help predict medication effectiveness and likelihood of adverse drug reactions (ADRs), that is, 'the right dose of the right drug at the right time' (Fig 1).⁴ Incorporation of genomic information

into prescribing decisions is being piloted in UK primary care and has the potential to address challenges of polypharmacy and medicine optimisation, with anticipated NHS implementation in the medium term.⁵

As generalists, GPs are highly skilled in care coordination and utilising existing local resources, managing complexity, communication skills including the assessment and communication of risk, ensuring safe and quality prescribing within the primary care setting and promoting screening programmes and healthy lifestyle. These skills are echoed in the Royal College of General Practitioners (RCGP) *Curriculum Topic Guide for Genomic Medicine*, which includes the taking of family histories to identify families with, or at risk of, genetic conditions and familial clusters of common conditions, such

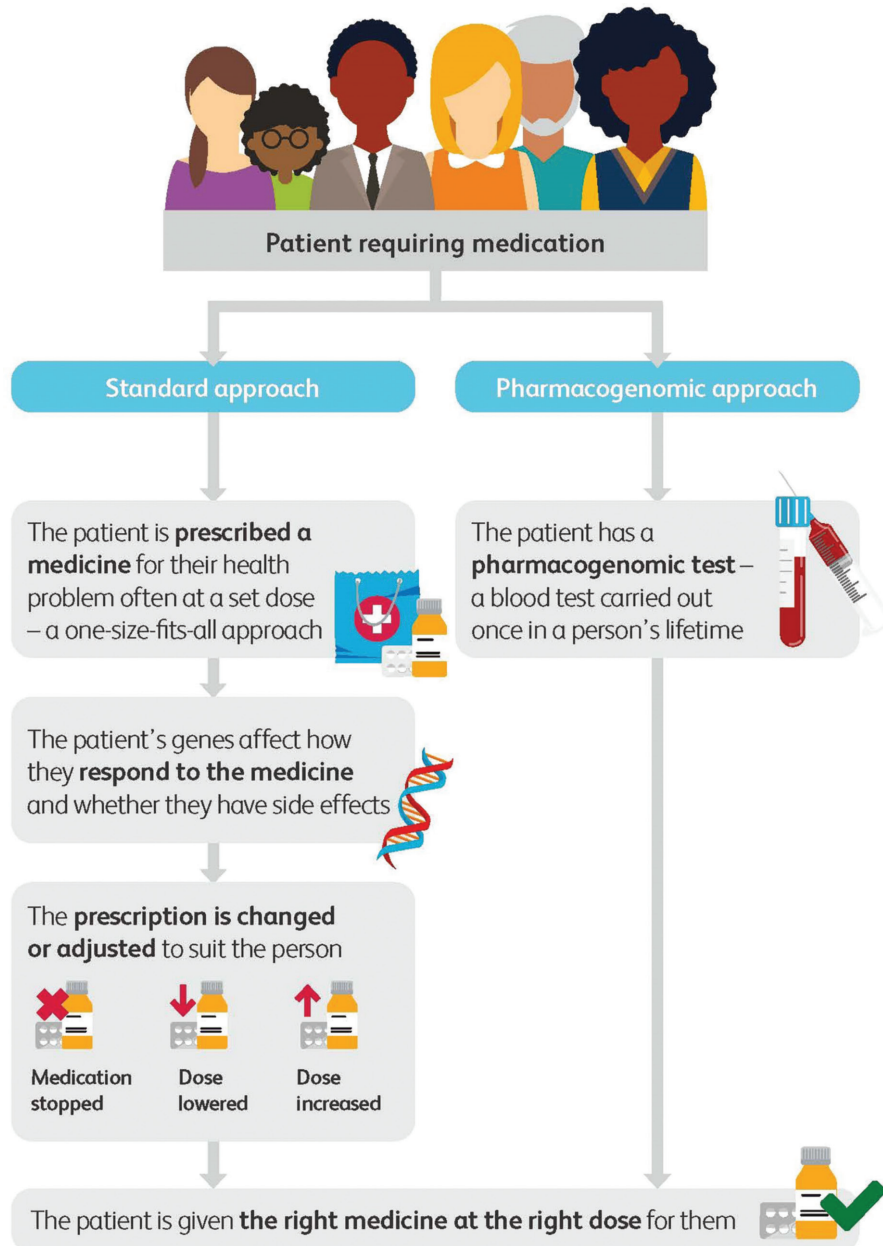


Fig 1. The aim of using pharmacogenomics is to help predict medication effectiveness and likelihood of adverse drug reactions, resulting in 'the right dose of the right drug at the right time'. Reproduced with permission from the RCP.⁴

as cancer, cardiovascular disease and diabetes. Thus, primary care has a key role in identifying those who would benefit from onward referral to services and testing.⁶

This combination of skills is unique to general practice; in combination with additional expertise in genomic medicine, it is also crucial to delivering care in the scenarios listed in the previous section. GPs also have an in-depth understanding of the challenges of overdiagnosis.

Workforce and educational perspectives

To address workforce and educational issues, it is crucial to understand the presentation of genomics issues within primary care and the nature, educational needs and behaviours of the primary care workforce (Fig 2).

GPs access information regarding genomics issues at the point of clinical presentation, for instance when patients present with concerns regarding family history, seeking education reactively or 'just-in-time' rather than proactively or 'just-in-case'.⁷ In addition, lack of point-of-care resources has been identified as a barrier to engagement with genomics issues in primary care.⁸ Primary care is now delivered electronically through the Electronic Prescription Service (EPS) and electronic health records (EHRs), which provide broad functionality for integrated point-of-care resources, such as clinical pathway templates, clinical alerts, pathology requesting and reporting, risk assessment tools and clinical decision support systems.

The primary care workforce providing direct patient care is multidisciplinary and becoming increasingly diverse, comprising



Fig 2. Different levels of education and training within primary care.

GPs, practice nurses, advanced nurse practitioners, physicians associates, pharmacists, paramedics and physiotherapists with overlapping roles in patient care. They can also be considered in the context of career stages: pre-registration, post-registration in specialty training, post-accreditation (generalised workforce) and practitioners with additional expertise or roles. The preference for point-of-care learning, the diversity of roles and career stages and delivery of care electronically all support an IT systems-based profession-agnostic approach providing a wealth of point-of-care support and education.

Delphi and GP educational needs

In 2017, Health Education England's Genomics Education Programme, now NHS England's National Genomics Education Programme, conducted a Delphi-style survey to identify how genomics presents (or not) in primary care.

Primary care practitioners and relevant stakeholders with representation across patient groups, clinical genetics, clinical science, pharmacy and nursing were asked two open-ended questions:

Q1. Please list the ways in which patients with genomic issues are likely and unlikely to present to primary care.

Q2. Please list the knowledge, skills and behaviours required and NOT required by primary care practitioners in dealing with patients with genomic issues.

Then, over a series of consecutive rounds, participants were asked to refine their responses to the initial questions and reach consensus.

The outcomes of the survey were presented in two parts. The first listed how genomics could present in primary care and was split into patient-led and clinician-led queries. For instance, when looking at family history, a patient might bring concerns of a possible inherited condition within the family, where as a clinician might ask about a patient's family history to inform a clinical decision. The second part then listed the agreed knowledge, skills and behaviours required by practitioners to meet these queries.

These findings were utilised within a collaboration with the RCGP to underpin the RCGP *Curriculum Clinical Topic Guide: Genomic Medicine*, and in conjunction with an Educational Resource Gap Analysis to develop the RCGP Genomics Toolkit. The toolkit is a 'trusted and familiar resource' in a well-recognised format within primary care education (see below), which can be used as a quick reference, as on-going continued professional development or to support more formal education and training.

How GPs access information

With the wider adoption and use of genomic technologies, primary care clinicians are increasingly exposed to genomic scenarios; with this, there is a need for accurate and timely information so that clinicians can best support their patients. Building on the knowledge that GPs access information 'just in time', a recent survey of UK GPs presented four common primary care genomic scenarios to understand how information was accessed in the context of a primary care consultation.⁷ This confirmed that GPs access genetic information when they perceive it to be relevant, 'just in time', rather than proactively 'just in case', confirming previous findings.⁸ These resources should be internet based and found in trusted and familiar places, such as primary care-focussed online education resources. They should ideally be concise and primary care specific so that they can be accessed and interpreted quickly even during or between consultations. Local pathways, available through intranet systems or in referral templates, were also a valuable resource. The rare disease scenario highlighted the roles of internet search engines and disease charity web pages, revealing how important it is that search engine 'top-hits' contain accurate and relevant information.⁷

Genotes

In response to the above evidence of need, the National Genomics Education Programme has developed Genomic Notes for clinicians (GeNotes),⁹ an online resource designed to provide concise and practical information at point of care with the opportunity for the learner to self-direct their own deeper learning journey if they wish (Fig 3).

The resource exists as two tiers: Tier 1, 'In the clinic', contains articles centred around a clinical scenario at the point of patient care and framed around the presentation, or testing, stage; and results stage. All tier 1 articles are aligned to NHS England's National Genomic Test Directory and organised into clinical specialties. The tier 1 scenarios are then underpinned by the tier 2 'Knowledge Hub', an extensive library of extended learning that arguments the clinical scenarios in a variety of delivery methods, including animations, infographics and films. Primary Care GeNotes was launched earlier in 2023 and contains clinical scenarios covering a range of genomic presentations, including cancer, rare diseases and more common conditions, such as familial hypercholesterolemia, reflecting the shift of genomics into mainstream care.

Integration into clinical pathways

The integration of genomics into routine clinical pathways within the NHS is happening at scale and pace. With this rapid expansion comes a pressing need to upskill healthcare professionals

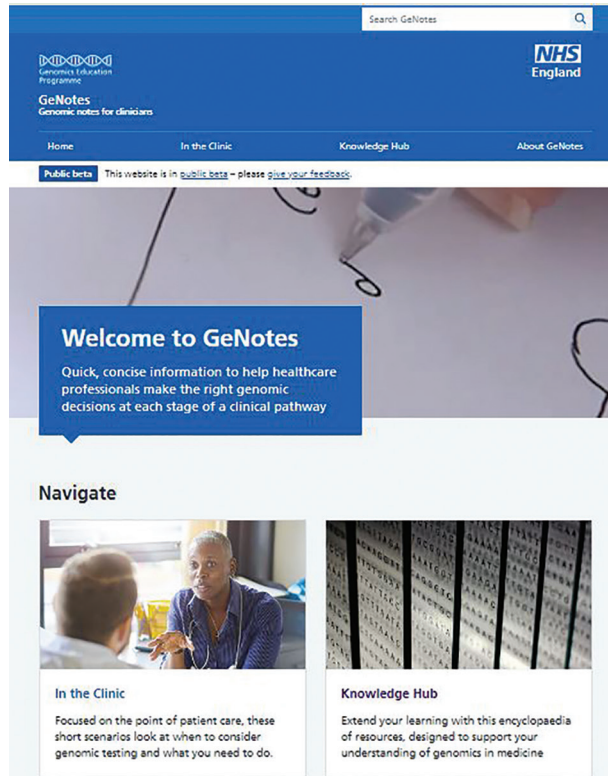


Fig 3. Genomic Notes for clinicians (GeNotes) is an online resource designed to provide concise and practical information at point of care and the opportunity for further learning.

involved in the delivery of any stage of an end-to-end clinical pathway. The National Genomics Education Programme has developed the Clinical Pathway Initiative (CPI) as a method to identify the workforce education and training needs within these pathways to provide a national consistent approach. Mainstream clinical pathways, either where genomics is used as a diagnostic tool or is aiding treatment or management, are broken down into component steps, or episodes of clinical activity, and the competencies required by healthcare professionals to deliver each step are identified and aligned to the educational resources required to meet the competencies. Crucially, the CPI is workforce-agnostic to take into account the variation in delivery of pathways (see Table 2 for a simplified example using familial hypercholesterolaemia), both regionally and locally. Therefore, the competencies that are identified are high level, allowing the CPI to be taken and applied in different settings irrespective of which element of the workforce is delivering that particular step in that locality.¹⁰

Rare disease care pathway mapping

Rare genetic diseases are broad in their nature and presentation. However, many share common characteristics: a complicated and protracted path to diagnosis; frequently multisystemic chronic disease with fragmented and poorly coordinated care; and an absence of disease-specific treatments and a good understanding of disease course and prognosis.¹¹ These challenges are reflected in the four priorities of the UK Rare Disease Framework.¹² One

of the four priorities, improving care coordination, is also a subject of a body of current research to define what coordinated care is and to suggest models for how to improve rare disease coordination.^{13,14}

For many rare genetic diseases, effective care coordination disappears after transition from paediatric to adult services. For most rare conditions, except those with a single organ-dominant phenotype, care is transferred to primary care, a role that the GP is often not explicitly aware is theirs, they often do not have capacity to take on and are not equipped with suitable resources to support them in their role.

A recent UK regional project mapped the care pathway for two rare genetic diseases, achondroplasia and neurofibromatosis type 1 (NF1), with the intention to improve the support for clinicians and patients in primary care. These pathways focussed on key touch points in primary care, such as transition from paediatric to adult services and at times of reproductive decision making. They emphasised key red flags with details of what to do if they occur; and concentrated on how care differs for a patient with the condition compared with routine care for a patient with a similar presentation but without the condition (eg more extensive investigation and referral of all patients with NF1 found to have hypertension). This enabled the pathways to be concise, highly relevant, actionable and suitable as a 'just in time' resource.

The approach to mapping the pathway and the format used to capture the pathway were deliberately designed so that it can be adopted and applied to other rare diseases. The suggested series of steps are:

- > Capture existing pathway based on literature, published guidance and existing local pathways or guidance with the input at this early stage of a single local disease expert.
- > Share draft pathway with key stakeholders, including patients' representatives and advocates, primary care clinicians and the breadth of hospital specialists involved in care. Accompanying the pathway is a survey, which is sent to capture stakeholders' views and identify areas within the pathway that are not clear or without consensus of agreement.
- > Arrange a pathway-mapping meeting (online was found to be suitable) concentrating on areas without consensus. This is also an opportunity to identify existing resources or suggested resources to support the pathway.
- > A final pathway is captured and shared in the same concise format as other pathways, highlighting key management issues, including 'red flags', to ensure familiarity and ease of use in primary care.

The pathway can then be disseminated to primary care by a range of methods and embedded into the primary care EHR as a template that can be actioned at each patient review, such as the annual learning disability review if appropriate.

Developing leadership and additional expertise

There is broad acceptance that clinical champions are crucial to effective healthcare-related implementation and to the sustainability of clinical innovation. Two GPs with a Special Interest (GPwSI) in Genetics roles, established by the Department of Health in 2004 in response to the Genetics White Paper *Our future: our inheritance*, continue to provide championing and leadership at the national level via working groups and committees, including National Genomics Education Programme, RCGP and Academy of Medical Royal Colleges

Table 2. An example of identifying learning needs and resources in a Clinical Pathway Initiative project: familial hypercholesterolaemia

Steps	Identify at-risk individuals	Assess individual genetic risk	Assess family history	Explain test and consent	Arrange genomic testing
Identify learning needs	1. Understand what FH is and recognise its associated clinical features 2. Recognise clinical signs that could indicate FH	1. Understand what FH is and recognise its associated clinical features 2. Be able to assess individual risk of FH	1. Be able to explain genomic testing in accessible language 2. Understand implications of positive and negative results for the family 3. Understand what variants of uncertain significance (VUS) are	1. Be able to explain genomic testing in accessible language 2. Understand implications of positive and negative results for the family 3. Understand what variants of uncertain significance (VUS) are	1. Understand the processes needed to send off a genomic test 2. Understand the GMSA/GLH structure
Identify resources	GeNotes Knowledge Hub article on PH	GeNotes Knowledge Hub article on PH Genetic risk assessment tool and allied learning	GeNotes Knowledge Hub article on PH Family history tool and supporting resources Taking and Drawing a Genetic Family History course Autosomal dominant inheritance animation	Talking Genomics: Tips and Tools for Communicating with Patients course Let's Talk About Genetic Testing film series Discussing Diagnostic Germline Tests course	Local/regional guidelines

All named resources are available at www.genomicseducation.hee.nhs.uk on a profession-agnostic basis. FH = familial hypercholesterolaemia; GLH = Genomic Laboratory Hub; GMSA = Genomic Medicine Service Alliance.

(AoMRC). A survey of how GPs access information about genomics noted geographical clustering of awareness and response rates in areas where a GPwSI in genetics currently exists.⁷

There is a gradual increase in the number of GPs and primary care pharmacists with additional expertise in genomic medicine in clinical service delivery, leadership, academia, service development and commissioning roles. National initiatives support the growth and development of these roles, such as a national Primary Care Special Interest Group (SIG) in Genomics, established in response to the need to harness the expertise and realise the investment in GPs enrolling in the MSc in Genomic Medicine, which reports to the National Workforce Skills Development Unit, and the development of frameworks for primary care genomics advisors and GPs with extended roles (GPwERs) in genomic medicine. The latter framework is under development in collaboration and aligning with the RCGP GPwER Framework.

Data and informatics considerations

Genomic technologies generate huge amounts of data, which poses a challenge for storage, interpretation, and availability across geographies and care settings.

Storing a single human genome code requires 200 GB, and the interrogation of genomic information and variant interpretation requires significant computational power and human expertise. Currently in the NHS, whole-genome sequencing (WGS) interpretation involves defining the patient's phenotype and examining specific genes associated with the phenotype dictated in the National Genomic Test Directory.¹⁵ For scenarios in which one wishes to be gene agnostic, such as the rapid sequencing

of the genomes of unwell children, a series of pragmatic steps is utilised in the pipeline to enable rapid interpretation.¹⁶ As WGS is adopted more widely, the capacity to interpret genomes at scale poses a challenge for existing resources, particularly the number of expert bioinformaticians available to interpret the data.

Given these computational and interpretative challenges, creating pipelines that rely less on human interpretation is attractive, with artificial intelligence (AI) approaches, both supervised and unsupervised, a promising advance.^{17,18}

A requirement for these informatic solutions to genomic interpretation is ensuring that clinical information is captured in a format interpretable for analysis. Structured clinical vocabularies, such as Systematized Nomenclature of Medicine Clinical Terms (SNOMED-CT) used in primary care, and Human Phenotype Ontology (HPO), which both provide standardised phenotypical terms and are widely used in clinical genetics, are key steps in this.

To reduce duplication of effort and optimise sharing of information, linked EHR systems that enable seamless sharing of information are crucial. However, in primary care, there is a need for the structured clinical vocabulary to evolve to ensure that we capture some of the challenges and ambiguity of genomic results and be able to draw pedigrees and family structures that can be revisited, revised and searchable. For example, capturing specific genetic variants will be important if we want the informatic pipeline to automatically flag if a situation changes, for example a variant of uncertain significance being reclassified as pathogenic. The EHR system should work such that this information is there when needed but unobtrusive for most day-to-day work. Optimising the coding of clinical features will also augment the use of informatic tools

that can provide diagnostic suggestions or investigative pathways, a recognised solution for the challenge of rare disease diagnosis,¹² and the focus of recent and ongoing work.¹⁹

A particularly relevant area that involves the interface of primary care informatics and genomics is pharmacogenomics. How pharmacogenomic information, which can provide highly relevant and timely information for the GP making a prescribing decision, is integrated into the EHR, and Prescribing Decision Support Systems will be critical for its utility.^{20,21} There are examples of different implementation models²¹ and a pilot programme of pharmacogenomic testing in UK primary care with a significant emphasis on how results are presented in the EHR.²²

The promise of precision medicine is built upon being able to link the 'Big data' of genomics and clinical data found in the EHR, recognise patterns and translate these into helpful and usable information.¹⁸ A prerequisite for such advances is that the informatic systems in place can integrate with one another, and that data sharing across the health system is both seamless and secure, and trusted by clinicians and patients.

Moving forward

Primary care will continue to support implementation of genomic medicine in the NHS by deploying core and in-depth skills in risk assessment, diagnosis, facilitating onward referral when appropriate, complex care and communication.²³ End-to-end clinical pathways with integrated point-of-care information and supporting informatics are core to NHS strategy and successful implementation and are fundamental to maximising access of patients to genomic medicine; these factors are crucially interdependent and cannot be addressed in isolation. We have summarised the current approach, which addresses and integrates workforce development and education, patient access and informatics. As genomic medicine continues to embed, extending into polygenic risk and pharmacogenomics, this approach will be crucial in ensuring that implementation of end-to-end clinical pathways truly incorporates primary care, thereby maximising patient access and outcomes. ■

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