ORIGINAL ARTICLE

Imagining genomic medicine futures in primary care: General practitioners’ views on mainstreaming genomics in the National Health Service

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Abstract

Genomic medicine has captured the imaginations of policymakers and medical scientists keen to harness its health and economic potentials. In 2012, the UK government launched the 100,000 Genomes Project to sequence the genomes of British National Health Service (NHS) patients, laying the ground for mainstreaming genomic medicine in the NHS and developing the UK’s genomics industry. However, the recent research and reports from national bodies monitoring genomic medicine’s roll-out suggest both ethical and practical challenges for healthcare professionals. Against this backdrop, this paper, drawing on qualitative research interviews with general practitioners (GPs) and documentary analysis of policy, explores GPs’ views on mainstreaming genomic medicine in the NHS and implications for their practice. Analysing the NHS’s genomic medicine agenda as a ‘sociotechnical imaginary’, we demonstrate that whilst sociotechnical imaginaries are construed as collectively shared understandings of the future, official visions of...
INTRODUCTION

Genomic medicine and its associated complex technologies of whole-genome sequencing (WGS), molecular analysis, adaptive clinical trials and targeted treatments are perceived as offering wide-ranging transformative health-care potentials, including diagnosis and treatments of cancers, other rare or previously undiagnosable diseases (Erikainen & Chan, 2019; Hallowell, 1999; Kerr et al., 2019) such as Leopard syndrome (see Genomics England participants stories). Hence, in 2012, the UK government funded the development of the 100,000 Genomes Project (100KGP), through a collaboration with Genomics England, a private limited company owned by the Department of Health. The 100KGP aimed to transform clinical care and to lay the groundwork for the development of a national genomic medicine service. Recruiting National Health Service (NHS) patients with rare diseases, the 100KGP achieved its goal of sequencing 100,000 genomes of patients with rare diseases for clinical and research analysis by the end of 2018 (Genomics England, 2019; HSE, 2019a). The 100KGP is lauded a success by the NHS (DoHSC, 2018) and is now used as a template for mainstreaming genomics in the wider NHS, although the extent of this perceived success is debatable (Samuel & Farsides, 2017; van Beers et al., 2018) and not addressed in this paper.

These developments were preceded by significant sociological interest. Three decades earlier, Lipmann’s (1991) geneticisation thesis predicted a future approach to health heavily informed by genetic technologies. In this journal, Conrad and Gabe (1999) examined the future implications of the Human Genome Project (HGP), whilst Petersen (2005) examined biobanks’ role in securing genetic health. Other research examined media reporting of the HGP (Smart, 2003), family experiences and clinical implications of genetic testing (Atkinson et al., 2013; Featherstone et al., 2005) and professional roles and experiences in genetics (Beaudevin et al., 2019). Meanwhile, Weiner et al. (2017) interrogated Lipmann’s (1991) geneticisation theory’s predictions and theoretical relevance. More recently, the emerging research is examining the ethical (Ballard et al., 2020; Dheensa et al., 2016), clinical (Dheensa et al., 2018; Samuel et al., 2017) and policy (Samuel & Farsides, 2017) implications of the 100KGP.

However, less work examines the perspectives of the health-care professionals (HCPs) in relation to genomics, and where this exists, it has focussed on the elite and biomedical scientists working in this field (Beaudevin et al., 2019; van Lente, 1993).

Here, we examine key groups of clinical professionals currently situated on the periphery of genomic medicine but the key to the mobilisation of this programme. GPs are recognised to
act as gatekeepers (Rose et al., 2001), referring patients to genetic medicine services. Having interviewed GPs working on England’s South Coast, our analysis draws on Jasanoff and Kim’s (2009; 2015) concept of sociotechnical imaginaries, demonstrating tensions between policy-level visions of genomics and those of GPs being drawn into the genomic project.

Genomic medicine and primary care

Notwithstanding the recent developments which have made genomic interventions accessible (Harvey, 2011; Stark et al., 2019) and somewhat widely understood (Harvey, 2011; Slade, 2016) genomics is still considered a ‘new’ technology within primary care settings, with research examining the experiences and views of GPs still emergent (Caroll et al., 2016; Smit et al., 2019; Joshi et al. 2020). This is unsurprising, with genomics services to date being the preserve of secondary care specialists. However, GPs have increasingly become the first contact for patients wishing to access genetic services. They also now act as co-producers of genomic information by collaborating in or recruiting patients for genomic research (Sullivan et al., 2010). Despite playing such a critical role, a small number of studies have indicated GPs’ knowledge of genetic and genomic technologies remains ‘low’ (Smit et al., 2019), with research revealing a ‘lack of confidence’ to discuss genomics with their patients (Caroll et al., 2016; Smit et al., 2019). This has informed the publication of online training resources and materials by the Royal College of General Practitioners (RCGP), NHSE and Genomics England (NHSE 2019c; RCGP, 2019). Importantly, this approach utilises a deficit model of knowledge, in which training is developed for GPs and without their involvement. In contrast, we engaged with GPs to examine what matters to them (Sayer, 2011).

Whilst a small body of evidence suggests that GPs view the potential for patients’ genomic data to improve decision-making and health-care outcomes, many see genomic medicine as in its infancy (Harvey, 2011; Sullivan et al., 2010; Vassy et al., 2013). Although GPs are supportive of genomic medicine as one of the many useful tools for understanding elements of their patients’ health (Smit et al., 2019), they also voice concerns about potential psychological impacts of the uncertainty or worries caused by genomic testing (Van El et al., 2013). For community health-care services already under pressure and with limited resources, concerns exist that these technologies will increase the numbers of ‘worried well,’ increasing GPs’ workloads (Manolio, 2017; Moscarello et al., 2019). GPs also expressed concerns that genomic testing alone was insufficient to explain their patients’ health, without wider socio-environmental factors of health and disease (Smit et al., 2019). Genomic medicine is, thus, challenging for GPs with their culture of evidence-based practice (Trinidad et al., 2015), privileging evidence of immediate benefits to patients (Vassy et al., 2013). This reflects the lack of evidence of direct benefits of genomic medicine to patients in the primary and community care services (Berberich et al., 2018; Vassy et al., 2013). Beyond these, there has been little substantive research (Cox & Starzomski, 2004; Will et al., 2010) exploring the perspectives of HCPs working on the periphery of genomic technologies.

GENOMIC MAINSTREAMING AS A
SOCIOTECHNICAL IMAGINARY

This paper examines how visions of genomic futures in the NHS are conceived and received by GPs by engaging Jasanoff and Kim’s (2009) concept of sociotechnical imaginaries: ‘collectively,
institutionally stabilised and publicly performed visions of desirable futures, animated by shared understandings of forms of social life and social order attainable through, and supportive of, advances in science and technology (Jasanoff, 2015; 4). Collective imaginaries are reflected in social life and ‘in the design and fulfilment of nation-specific’ goals and ambitions (p120).

This approach echoes work on the sociology of expectations (Brown & Michael, 2003; Dimond & Stephens, 2018; Hedgecoe & Martin, 2003; van Lente & Rip, 1998), which presents expectations as a means of structuring anticipatory discourse and practices, and as a mechanism for designing futures. This body of work highlights the need for a critical investigation of biotechnological innovations and evoked ‘expectations’, defined as ‘real-time representations of future technological situations and capabilities’ (Borup et al., 2006). Often, by evoking hope and transformation, expectations define and contour professional and public sensitivities to promised technological futures and the structure of emerging technological fields (van Lente et al., 2013). Expectations guide innovations by influencing agendas, legitimising the mobilisation (van Lente et al., 2013) and allocation of resources across macro (national policy), meso (institutional capacity building) and micro (practice and implementation) levels (Borup et al., 2006). Therefore, expectations are by nature performative and shape the course of innovations.

The performative (Butler, 1993; Goffman, 1959) character of expectations has implications for studying sociotechnical imaginaries. Building novel technological futures is habitually based on publicly performed and expressed ‘positive visions of social progress’ and desirable futures (Jasanoff, 2015: 4). This includes repeated optimistic pronouncements about the promise of new technologies, mobilising resources and instituting civic professional roles to bring visions into being (Dimond & Stephen, 2018). Rooted within the concept of sociotechnical imaginary is cognisance that futures emanate from imaginations in social, political and scientific spheres. As such, ‘technoscientific imaginaries’ are simultaneously also ‘social imaginaries’, ‘encoding collective visions of the good society’ (Jasanoff & Kim, 2009, p. 123). Jasonoff (2015) explains how these ‘collectively held and performed visions of desirable futures’ are also ‘durable, capable of being performed; yet they are also temporally situated and culturally particular’ (p. 19).

The capability to perform sociotechnical imaginaries is apposite. We argue mainstreaming genomic medicine in the NHS is a reiteration and enacting of a particular UK sociotechnical imaginary relating to expectations of healthy futures. Other STS scholars (Dimond & Stephens, 2018; Jasanoff, 2005; Nelkin, 1995; Nerlich et al., 2002; Petersen, 2001) have identified how notions of good technoscience, seeking to alleviate the suffering of citizens in media, policy and political discourse works to legitimise state policy and power. Recently, a promissory discourse (Borup et al., 2006; van Lente, 1993) relating to the 100KGP has emphasised genomics’ capacity to transform health care and provide hope for cancer and rare disease patients (Samuel & Farsides, 2017). Wrapping the genomic medicine imaginary in a promissory discourse (Poli, 2010) facilitates ‘collectively held visions’ and building coalitions for enacting genomic futures.

However, emphasising hope and transformation negates the messiness and uncertainty (Kerr, 2004; McGoey, 2009) as genomic testing is an emerging, complicated and uncertain science (Kerr, 2004). Whilst uncertainty is an inherent part of medical diagnostic practice, and some uncertainty is useful for facilitating autonomy (Newson et al., 2016), complexities associated with vague information and outcomes, and probabilities of risks may have psychological impacts (Newson et al., 2016). This is because genomics can facilitate what Poli (2010) calls an ‘anticipatory system’, in which testing purports to enable people to make health decisions in the present to mitigate against the possibility of ill health occurring in the future (Poli, 2010).
Additionally, as Groves et al. (2016) observe, evoking hope and desirable futures often obscures taken-for-granted questions about the political contestations underlying such imaginaries. Critical evaluations of assumptions underpinning imaginaries are important contributions to conducting an ‘upstream’ analysis of innovations (Groves et al., 2016). This allows for diverse voices, including public and professional interests (Groves et al., 2016) and ambivalence (Ehrich et al., 2011) to sociotechnical innovations. Understanding desirable futures requires attention to shared, often quiet, fears of policy and practical implications of emerging technologies (Jasonoff, 2015), since imaginaries are characteristic of particular definitions of social priorities, mostly presented as ‘taking care of the future’ (Stilgoe et al., 2013). However, political agendas, thus, neglect other social concerns (van Lente, 1993). Hence, deliberations about genomic futures must be unavoidably inclusive of a diverse range of societal viewpoints.

Previous Science and Technology Studies’ (STS) research has explored the roles of researchers and technicians (van Lente, 1993), policymakers (de laat, 2000), business (Martin, 1999) and developments in pharmacogenetics (Hedgecoe & Martin, 2003). Recently, Dimond and Stephens (2018) examined contestations in the development of mitochondrial donation as sociotechnical imaginary engaging patient groups and policymakers. However, little attention has focused on examining the variance or coherence of visions of genomic futures between policy and HCPs who will be gatekeepers to public access to mainstream genomic medicine services.

The NHS genomics agenda has emerged from the ‘top’ (Government policy level) (Davies, 2016; DoH & DfB, 2020), with genomic medicine futures construed as transformative and a necessary investment to safeguard the nation’s medical and economic futures (Davies, 2016). Public involvement in the 100KGP, and perceived success (DoHSC, 2018) and related research have been presented as signalling uncontested public and professional support. Arguably, the genomic medicine agenda is in keeping with Weick et al.’s (2005) idea of sensemaking, in which they contend that knowledge, including resultant policy programmes, can be partial and intricately tied to those in power who may use it to legitimise policy actions. So far, genomic medicine has included the elite and powerful who set the agenda, terms of engagement and the constitution of the ‘genomic public’ (Samuel & Farsides, 2018). We argue it is beneficial to gather knowledge by disrupting the status quo (Weick et al., 2005, with space made for ‘multiple knowledge and marginalised voices’ (Schultze & Stabell, 2004: 560). Our sociotechnical imaginaries approach gives voice to HCPs whose views on genomic medicine futures in the NHS are, as yet, overlooked. This demands a consideration of the socio-material, everyday clinical realities of such genomic imaginaries (Grove-White et al., 2000) and interrogating the allure (van Lente et al., 2013) and implications of the ‘worlds’ (Groves et al., 2016) coalescing around the sociotechnical imaginaries (Macnaghten & Szerszynski, 2013), including the intersections of global, corporate and national political interests, professional roles and public demands for improved health care. We also consider the symbolic and material actions that facilitate genomic futures and briefly examine medico-political interests and assumptions underlying claims about desirable futures.

We explore the role of visions of technoscience in health-care futures (Dimond & Stephen, 2018). Our contribution to this subject is twofold. Conceptually, we extend Jasanoff and Kim’s (2009) analysis by underscoring the role of sociotechnical imaginaries within more micro-social processes at the intersections of policy and practice. Empirically, we have carried out micro-sociotechnical imaginary analysis by bringing the views of GPs into dialogue with policy positions to demonstrate complexities associated with integrating technologies in the ongoing development of health-care services.
METHODOLOGY

We conducted in-depth semi-structured interviews with GPs focusing on perceptions and experiences of ethical preparedness for genomic medicine's introduction, to obtain an in-depth contextual and nuanced understanding of GPs’ experiences and views of the NHS genomics agenda in the context of everyday clinical practice. Participants were GPs working in diverse settings including inner city and suburban areas, with GPs at various career stages including newly qualified (three), partners (GPs who part-own the practice business) (eight), experienced practising GPs – non-partners (seven), retired or about to retire (three) and some in composite roles as GP clinical teaching fellows and practitioners (two), totalling 23 participants.

The interview guide focussed on the participants’ career biography up to current roles, and a discussion of the health challenges for their patient populations. The interview then explored participants’ experiences with technological innovations, and how these related to their everyday practice, progressing to explore views of genomic medicine and the potential impacts participants identified mainstreaming genomic medicine could bring to their patients and clinical practice. The aim was to engage with GPs in a critical and open dialogue providing space to reflect on their everyday practice.

In parallel, a detailed documentary analysis (Bowen, 2009; Dixon-Woods & Bosk, 2010) was undertaken of publicly available policy documents relating to the mainstreaming of genomics. Considering the Human Genomics Strategy Group (2012), the Chief Medical Officer of England’s (2016) report, the Life Sciences Industrial Strategy (2020) (Bell, 2020) and editorial material on NHSE and Genomics England websites provided an alternative ‘official’ account of how genomic futures are imagined, presented and enacted (Jasanoff, 2015; Jasanoff & Kim, 2013).

Participants were self-selecting, recruited with the help of a local Clinical Research Network (CRN). The CRN circulated study information to regional GPs requesting those interested to contact the CRN, whose details were forwarded to SM to arrange an interview. Interviews were between May 2019 and December 2020, as part of larger multi-method research exploring the views and experiences of UK HCPs, the findings of which are reported elsewhere. The research was approved by the University of Sussex Research Governance Sponsorship committee (049/MWA) and the Health Research Authority (HRA) (19/HRA/1701).

SM led the data analysis utilising an approach based on Charmaz’ (2006) constructivist grounded theory analytical approach. This approach amounts to what Richardson and Kramer (2006) call an ‘abductive approach’, involving a constant interplay between interview and documentary data, for the comparative element key to the sociotechnical approach (Jasanoff & Kim, 2013; Jasanoff et al., 2007), with broad themes of ‘impacts of genomics’, ‘ethical implications’ and ‘understandings of the genomic agenda’, guiding the initial reading of the data. SM read and reread transcripts ‘abductively’ to become intimately familiar with the data, then undertook a detailed coding of key data characteristics relevant to the research question. Coded data were searched and collated into themes. Similar data were grouped together and coded quotes were read. Coded data were further analysed to refine the scope and focus of themes. Themes and data were scrutinised and compared, to pinpoint similarities, differences and refine themes (Glaser & Strauss, 1967). This facilitates the identification of synergies and divergencies within the data (Bowen, 2009). Finally, the data extracts were weaved into narratives in relation to the wider literature.

Wider theoretical frameworks guiding analysis were drawn from the aforementioned literature on the sociology of expectations and sociotechnical imaginaries (see the previous discussion).
The wide-ranging number of participants and the consistent issues raised by participants in individual interviews gave us confidence in the validity of our findings.

**FINDINGS**

The findings are structured using three main themes emerging from interview data and mirror topics in the policy documents examined: ‘genomics as a technology in formation’, ‘deprioritisation of primary care needs’ and ‘risk and uncertainty in genomic medicine’.

**Genomics: a technology in formation?**

Jasanoff and Kim’s (2009) concept of a sociotechnical imaginary emphasises the relationships between envisioned futures and current practice, and their mutual shaping in a process of co-production (Jasanoff, 2004). Analysis of policy documents revealed a vision of genomic medicine in the NHS in which genomic technology is ready for wider NHS roll-out. This is evident in the Chief Medical Officer’s report (2016):

“Genomics is not tomorrow. It’s here today. I believe genomic services should be available to more patients whilst being a cost-effective service in the NHS... Now we need to welcome the genomic era and deliver the genomic dream”

(Davies 2016:1).

The unmistakeably positive discourse here is not unusual in political policy pronouncements (Jasanoff & Kim, 2009), being symbolic of ‘publicly performed visions of the future’ (Jasanoff, 2015). Used as such, this discourse is significant in positioning genomics as an available technology within the NHS, simultaneously locating it as a ‘dream’ and ‘acknowledging that the genomic era’ is yet to come. Its realisation requires health-care staff to both ‘welcome’ and ‘deliver’ this ‘dream’, which must also be ‘available’ and ‘cost-effective’. In addition, it has the performative power of making future imaginaries both desirable and achievable by evoking hope, rousing public expectations and emphasising the salience and imminence of genomic medicine. The use of specific concepts alluding to ‘widely available’ and ‘cost-effective’ health care make intangible futures tangible and vivid. Hence, this discourse legitimises the political attention (Grove et al., 2016; Stilgoe et al., 2013) and policy actions (Dimond & Stephen, 2018) required to realise the genomic future. In practice, these statements can clash with the realities of everyday health-care practice. This was evident in the majority of our GP accounts. Reflecting on this planned roll-out, many of them specifically, and in contrast to the above, perceived genomics as an *emerging* technology:

“I think genomic medicine is still in its infancy, poorly understood, not widely used. I think it is an emerging tool”

(GP9).

“This (genomics) is all-new, and its actual utilisation will be a long way down the road.”

(GP7)
Perhaps pointing to complexities in the tensions between hoped-for futures and everyday clinical practice, GPs felt the translation of genomics into clinical practice was in its ‘infancy’ with any clinical benefits ‘a long way down the road’ in a distant future (Hernes & Schultz, 2020). GPs’ categorisation of genomics as an emerging technology was also in part due to their recognition of a significant knowledge deficit among the ‘average GP’ in practice:

“But the average doctor does not really know what genomics medicine means for practice. It is some fancy stuff happening in a lab somewhere. But what does it mean for the average doctor seeing someone in a clinic, I do not think that is really trickled down. I think the average genome, genetics study, still seems like an ivory tower to the average GP,…”

(GP1)

Strikingly, most GPs acknowledged lacking awareness of the clinical relevance of genomics, the 100KGP, and the NHS plans to roll out genomics. GP1’s use of the ‘ivory tower’ analogy is emblematic of GPs’ felt distance from genomic medicine and its application to their clinical practice (Sullivan et al., 2010). The quote also illustrates the policy sphere’s selective approach to engaging with HCPs. Working closely with the ‘lab’ and the ‘ivory tower’ is seen as having little relevance to them and their colleagues, positioned in contrast as ‘the average GP’ and ‘average doctor’, who appear to feel side-lined, whilst simultaneously failing to reach the patient ‘someone in a clinic’. This contrasts with how positive narratives of genomics are situated (Jasonoff, 2015) and performatively frame genomics as a necessary technology, hence, grounding policy action (Dimond & Stephen, 2018). Decades of claims about the clinical benefits genomics presents to the NHS and patients (Collins et al., 2003; Conrad & Gabe, 1999) are yet unrecognised by many primary care practitioners (Vassy et al., 2013). References to a ‘trickle down’ suggests a perception that it takes time for information to reach practising professionals.

Concerns about the relevance of genomics for clinical practice was also linked to capacity-building and frontline infra-structure, as highlighted by this participant, a clinical teaching fellow and partner GP with expertise in genomic medicine.

“You need to have the infrastructure in place. So, for example, for red blood cells diseases, they describe what is called the diagnostic odyssey, where patients have to spend a long time through the system before they come up with a diagnosis....the reorganisation of genomic services and how information is going to flow, you know, from the clinician to the genomic medicine centres to the laboratories, and then back from the laboratories, back to the referring clinician... With genomics, the infrastructure is going to have to change, as more patients are going to be exposed to concepts of genomics because of relevancy to their condition.... And GPs need to be trained for this to work”

(GP10).

A lack of infrastructure, as information flows and patient pathways through services, means that policy-level genomic imaginaries do not reflect the realities in clinical practice, including NHS capacity to meet demands ‘as more patients are exposed’ to genomic medicine. This infrastructure gap is contrary to the deeply ingrained pathways and cultures of working between primary and secondary care.
See here the CMO’s quote outlining the training needs whilst also signalling the enormous work needed across the NHS for service delivery:

“...we need a new genomic paradigm to be integrated into all training curricula and specialty training of all clinicians, not just doctors. Adopting genomic technologies into routine practice will require changes in the design, operation and workforce of healthcare organisations…”

(Davies 2016,6)

In addition, the CMO recognises that the delivery of genomics ‘will require changes in the design’, and ‘operation’ in care organisation and delivery. The quote highlights a mismatch in visions (Jasonoff & Kim, 2015) of genomic futures. For GPs, infrastructure and capacity should be in place before mainstreaming, whilst at the policy-level mainstreaming, infrastructure and capacity building, can occur concurrently. The difference in perceptions may be temporal but is perhaps an illustration of how the policy sphere’s visions of genomic futures may be removed from everyday frontline realities.

Reflecting on the NHS plans to roll out genomics, GP7, a GP partner in a suburban practice was not alone in feeling that the current political focus on genomics was akin to a fad.

“Things like that are quite sexy areas of medicine, aren’t they? with the kind of gene mapping or genome mapping, but will it last?”

(GP7)

The term ‘sexy’ in this context refers to something ‘alluring’ or ‘fashionable’ - obfuscating its implications and limitations: ‘sexy’ suggests genomics is attractive in the current political context, fitting with the wider discourses of desirable futures, offering hope to patients. However, this view is attributed to (politicised) others, whilst these GPs see the NHS genomics agenda as a ‘temporally situated and culturally particular’ technology (Jasanoff, 2015) which they do not have to engage with.

GPs’ accounts should not be seen as in opposition to genomic medicine in the NHS per se; rather, they appear sceptical of the scale and speed of its introduction, its relevance for clinical practice, their ability to fit the agenda within the current organisation and delivery of care, particularly amidst constant unrealised promises of NHS reorganisation. Whilst analysis of policy documents reveals the symbolic material and the performativity of future-orientated practices (Van Lente, 1993; Dimond and Stephens 2018) in the enacting of genomic futures, for these GPs, genomics is nascent, with the work for its clinical application at a distant future.

**Genomic medicine and the deprioritisation of primary care needs**

Having suggested a possible tension between policy and practice-based accounts of genomics, it is interesting to consider to what extent genomics seen as a priority for GPs’ clinical practice. Stilgoe et al. (2013) and Van Lente et al. (2013) suggest that sociotechnical imaginaries characteristically redefine social priorities, presented as ‘taking care of the future’, displacing other current concerns from the policy agenda. The government strategy, ‘Genome UK – the future of health care (2020)’, declares genomic technologies a priority offering opportunities for transforming health care by improving diagnosis and identifying treatments:
“We will incorporate the latest genomics advances into routine healthcare to improve the diagnosis, stratification and treatment of illness. We will be the first national healthcare system in the world to offer whole genome sequencing (WGS) as part of routine care...”

(Genome UK- the future of healthcare 2020; Pg6)

This nuanced positioning of the patient, health care and UK global socioeconomic benefits denotes the political significance of the genomic medicine agenda. This positive discourse of genomics medicine facilitates the performative capacity of the agenda to ground resource mobilisation and allocation (Groves et al., 2016) to genomic medicine and its relevance for immediate implementation, as previously alluded to. The performativity of these discourses is also evident in their ability to perform future-oriented narratives by validating and facilitating state power to prioritise genomics over other health care needs.

However, for GPs, genomics is not a priority, contrasting starkly to their everyday practice and patient needs. When asked about a hypothetical budget, most participants felt genomic medicine would not address challenges they encounter in everyday clinical practice:

“I wouldn’t value genomics at the moment. If I had a restricted budget, it wouldn’t be top of the list. For me, it’s a bit of a pet project. Patients end up in A&E (Accident &Emergency department), rather than saying, ‘What’s wrong with you?’, it’s, ‘Why are you here?’ We have a lot of pressing social care issues that genomics may not solve.”

(GP9)

Describing genomics as a ‘pet project’ encapsulates these GPs’ beliefs that NHS investment should be addressing long-standing health service challenges including public health problems that mean ‘patients to end up in A&E’. For the GPs, these challenges lead to pressures in services such as A&E, where staff no longer focus on caring but wishing patients went elsewhere: ‘why are you here’. Therefore, genomics displaces pressing primary care needs from the local policy agenda (Groves et al., 2016; Van Lente, 1993). GPs are not opposed to genomic medicine per se, but display ambivalence (Ehrich et al., 2011) in the context of pressing patient needs in everyday clinical practice.

“There are too many other things that have to come first on the list of priorities, like social care, especially for the elderly. I do not think we should be trying to deliver this service now, it’s politically convenient, but the NHS has more important things to start delivering first.”

(GP20)

GPs’ views about care are inevitably infused with concerns about pressing patient priorities, including challenges facing ‘social care, especially for the elderly’, weighing these more highly than technology’s perceived cost and inability to offer solutions (Caroll et al., 2016). Their response aligns with what Will (2019;71) calls ‘a shrug’, ‘a strategic retreat from engagement’. This retreat from engagement should be seen as an active performance by GPs as they negotiated what they believed to be uncertain, unclear and at times, conflicting, policy directives on introducing technologies such as genomics in their practice.
Meanwhile, officially genomics is presented as offering solutions to clinical challenges GPs may confront in their caseloads and as having the potential to improve life and health care through better diagnosis and treatment.

“We will help people live longer, healthier lives by using new genomic technologies to routinely identify the genetic determinants of rare diseases, infectious diseases and cancer... and... provide personalised treatments to illness...”

(Genome UK- the future of healthcare 2020; pg. 16)

Policy and political narratives of genomic medicine as a transformative technology have the performative power (Jasanoff, 2015) to usher the genomic medicine imaginaries into being by rousing public desire and support by promising a future of ‘longer and healthier lives’, and among others providing ‘personalised treatments to illness’. Given their work with a predominantly ageing population within a financially constrained service, many GPs viewed policy statements such as these as problematic. Five participants were concerned that these genomic technologies focused on living longer, with one stating:

“There is only so much money in the pot, and at the moment our bigger problems probably are dementia. That is what we are dealing with day-to-day. Another part of me, and I probably shouldn’t say this. We have created this problem of elderly frail population by keeping them all alive from all their heart attacks and diabetes that would have finished people off in their 50s, 60s, 70s, so we have now got this massive burden. So is there any way genomic testing could help with that? I don’t know, but! It would help them live even longer I suppose....”

(GP7)

Participants were concerned about the financial implications of genomic medicine suggesting that there is ‘only so much money in the pot’ which should be directed mostly towards addressing ‘bigger problems’ that they ‘deal with day to day’ such as ‘dementia’. The GPs believed that life-prolonging technological innovations, important as they are, have ‘created this problem of an elderly frail population’ underserved by primary and social care. For the GPs, genomics would contribute to the pressures facing primary and social care by helping people ‘live longer’. The GPs’ accounts demonstrate the conflicts between expectations in policy pronouncements and actions, and the realities in everyday (Groves et al., 2016) clinical practice. For the GPs, the policy landscape appeared removed from their everyday clinical realities, focused on extending lives and investing in expensive technologies rather than responding to health inequalities by improving service provisions, with genomic medicine scarcely relevant to solving everyday challenges facing primary care. However, the same GPs are being told that genomic medicine offers practice and cost benefits to primary care.

“Genomic technologies are more than just another innovation: ....They offer tangible benefits across the spectrum of patient care including GP surgeries, mainstreaming clinical specialities and highly specialist units. They can provide far greater insights into disease risk, thus supporting preventive action, and can help to ensure that patients receive the most effective treatment sooner. They are the very essence of an innovation that can add value and reduce cost.”

(Human Genomic Strategy Group 2012;16-17)
Whilst these GPs saw genomics as an emergent technology, arguably, for the NHS, genomic medicine is more than just a promissory technology (van Lente, 1993), and offers real material benefits for all patients in primary care now and in the future including information on disease risk and improving patient diagnosis and treatment, where potential benefits of reducing costs justify the need for investing and mainstreaming genomic medicine in the NHS. However, this significant investment, absent a lack of a compelling case in clinical application, led two participants to suggest that current visions of genomic medicine allow the policy sphere to ignore the pressing needs of services supporting and caring for patients, with one stating:

“I think they envisage some personalised medicine utopia, which may not come to pass, so I think the dream is to take somebody who’s, potentially may not biologically behave like every other patient with a certain condition and to realise why it is that this person with Crohn’s Disease or whatever, does or does not respond to this treatment, or does or does not do better than another patient.”

(GP20)

The participants’ views about genomics are summed up in the suggestion that genomics is an unattainable utopia. The statement highlights the tensions between what constitutes ‘real’ pressing health care needs and what GP9 referred to as a ‘pet project’ of genomic medicine. By bringing policy and participants’ accounts into dialogue, two divergent and conflicting visions emerge. Participants felt the genomics agenda displaces the ‘real’ pressing issues facing primary care from the policy agenda, whilst claims continue at the policy level that genomics offers answers to some urgent challenges facing primary care. Participants’ accounts highlight the complex interactions between hope (van Lente, 1993), ambivalence (Ehrich et al., 2011) and sociotechnical imaginaries. Whilst the policy sphere may project a positive future where genomics frees society from present and future health ills, it appears that for the participants, attaining the hoped-for future requires attending to current challenges facing their patients. Their accounts point to the ethical and political significance of imaginaries and how GPs’ experiences of existing health inequalities shape their responses to promised futures.

‘Can of worms?’ Risk and uncertainty in genomic medicine

Erikainen and Chan (2019) argue that additionally to clinical benefits, genomic medicine is attractive in neoliberal contexts, where individual responsibilities are central to improving public health. This claim is supported in policy documents where the wider benefits of genomics are outlined:

“As the quantity and quality of available genomic information increases, further such links will be identified and communicated, via different public health channels, to help people to make more informed lifestyle choices”

(The Human Genomics Strategy Group, 2012; 35).

Clearly, enacting genomic futures shifts genomics from a mere diagnostic tool to what Poli (2010) calls an ‘anticipatory system’ aiding individual decisions now in anticipation of future health events. However, the hope that the policy sphere expresses negates the complexities that accompany this practice of medicine. Combined with increased emphasis on personal responsibility for health, some
felt genomic medicine could have profound consequences for the doctor-patient relationship and the management of expectations.

“The problem is the expectation people have of medicine. When people want an investigation, they are just driven straight to the test. There is this expectation that the blood test, the scan will answer everything. I think that people have this idea somehow that I had a full test, that they can now have the full picture of their health. They don’t really understand what those blood tests are showing or the limitations of testing without a clinical pointer.”

(GP20)

Most GP participants viewed the genomics policy discourse as a promissory technology (van Lente, 1993) that too easily locates genomic medicine as a catch-all solution. Interviewees sensed that expectations of genomic medicine render invisible its inherent complexities (Borup et al., 2006), including voluminous information and complex results, which may have scant utility to the patient’s health decisions. Yet, in official discourse, this promise is rendered straightforwardly.

“Central to this is ensuring that people understand the health consequences of their lifestyle and behaviour choices so that they can make informed decisions about their own and their family’s health, wellbeing and care. Our growing understanding of gene-environment interactions, driven by genomic technology, can make a significant contribution to this.”

(Human Genomics Strategy Group, 2012; 31)

This declared confidence in genomic medicine’s benefits reflects a detachment from the uncertainties (Van Lente, 1993) perceived by GPs and how their concerns impact the delivery of primary care services.

“It is what happens with any technology, isn’t it? If you make it, you have to use it, so genomics will not tell us anything much but fits well with the hyper-individualised society we live in, in which everything is about the individual, and this technology will aid that.”

(GP18)

Arguably, for these participants, mainstreaming genomics goes beyond the performative character of sociotechnical imaginaries (Jasonoff & Kim, 2009) and habitually publicly performed expressions of desirable futures. They see policymakers capturing the public’s imagination concerning possibilities whilst ignoring the GPs’ cautionary imaginary. In the policy sphere, genomics aids a shift to individual responsibility for one’s health through earlier disease detection and informed lifestyle choices (NHSE, 2016: 12). Among GPs, this logic ignores the uncertain and ambiguous nature of genomic testing and its impacts on the public and services. As GP18 further states:

“Genomic tests will tell you that you have a likelihood of developing Alzheimer’s as a risk but not you will get Alzheimer’s disease next year, nobody knows that it is just your gene pool might sort of predispose you to get it at some point. So, it is a lot of complicated messages that will just make them anxious”

(GP18).
Whilst policy-level discourse presents genomics as a tool to help patients attain diagnostic certainty, the participants were concerned about the indeterminate and uncertain nature of genomic testing and the patient’s ability to understand risk and susceptibility information (Kerr, 2000).

Participants’ concern about the anticipatory (Poli, 2010) and broad-reaching nature of genomic testing emerged in discussions around uncertainty and communicating risk to patients:

“...it gives me some anxiety that it is just more testing, more medicalising, earlier labelling of people and you know, that’s not really what we try and do in primary care, telling people they’re ill before they are actually ill”

(GP11)

Possibly due to their sense of a genomic knowledge deficit, GP11 was anxious about the potential challenges genomics presents when interpreting results, which can increase workload with more anxious patients seeking GP consultations. Consider Clarke et al.’s (2010) theorisation of the biomedicalisation of risk, where technological innovations such as genomics make health risks increasingly amenable to medical interventions, redefining patienthood from experiencing symptoms to the probability of developing a condition. GP9, concerned about the widening use of genomic technology, likened it to the metaphorical ‘can of worms’.

“Genomics can open a can of worms, for example, a guy comes in, says, "Look, doc, I’m 50, both my parents had dementia, I’m thinking do I retire at 50 and have ten good years travelling, so I’ll be demented when I’m 60. Or do I work till I’m 65 and have a retirement, a traditional retirement because dementia isn’t going to get me?" If you can help answer that question with appropriate counselling support, that might be good. I suppose the problem would be if the test says yes, you’re going to get dementia by the time you’re 60, that’s a big thing to take on board if you’re the patient.”

(GP9).

Participants imagine genomic medicine may uncover diseases lacking current treatments, exemplifying genomic medicine’s anticipatory nature (Poli, 2010) of, and expressed concern about, their patients’ ability to cope with the test results. This demonstrates the temporal and uncertain nature of genomic testing (Kerr, 2004; Brown et al. 2011).

Whilst the policy sphere imagines a ‘utopia’ where an assured and responsible public are informed of health risks and actively manage their health, GPs envision a ‘dystopia’, where genomic medicine’s increased biomedicalisation of everyday life overwhelms primary care with an anxious and uncertain public, focusing on very practical workforce concerns. Paradoxically, it is the uncertainty in diagnosis and treatments that genomics aims to resolve that most worries some GPs. As McGoey (2009) argues, uncertainty is generative and performative, generating a need for solutions to the opacity it propagates, resulting in the consolidation of influence by those who highlight the uncertainty in the first place.

CONCLUSION

Set against the background of mainstreaming genomic medicine within the NHS, this paper illuminates how GPs view the NHS genomic medicine agenda in the context of their everyday clinical practice. The paper’s theoretical grounding contributes to the sociological discourse on ‘expectations’
(Borup et al., 2006; Brown, 2000; van Lente & Rip, 1998) and genomic medicine (Dimond & Stephens, 2018; Hedegoe & Martin, 2003). We claim that the genomic medicine agenda is intimately tied to ‘sociotechnical imaginaries’ (Jasanoff & Kim, 2009) including policy-level perspectives of genomic medicine framed as a positive discourse emphasising its transformational possibilities and potential benefits to public health. By speaking to GPs, this paper identifies their parallel account, which diverges from and challenges the official vision. These parallel visions reveal incompatibilities between national policy and local practitioners’ visions of genomic medicine. Engaging Jasanoff and Kim’s concept we analyse how at a policy level, technoscientific visions of genomic medicine futures and social order are embedded in, constitutive of, and produced by the discourse of hope, transformation and empowerment. This contrasts to visions of genomic medicine held by medical professionals responsible for everyday care delivery, which forefront uncertainty and complexity, challenge priorities and resources and question timeliness and uncertainty of impacts. The GPs interviewed here believe genomics has scant relevance for their everyday work. Whilst sociotechnical imaginaries are based on collective and shared understandings of the future (Jasanoff & Kim, 2009), our analysis shows that there are no shared understandings, and neither is the NHS genomics vision collective. Therefore, using the sociotechnical imaginaries as a conceptual tool requires delineating what is meant by ‘shared’ and ‘collective’. Rather than referring to a broad social and public consensus, we highlight how imaginaries can be held, advanced and enacted by a selected powerful few individuals or groups and supported by a section of society that may see potential benefits in the proposed imagined futures for these technologies. We must acknowledge the marginalised voices vital to the implementation of these transformational programmes who may resist and whose views may be ignored.

Policymakers optimistically present genomics as a transformative technology of national economic significance, ready for clinical application. For GPs, aware of the lack of evidence on the relevance of genomics for their practice, lack of resources and mindful of its complexity, genomics is an imaginary whose benefits are poorly understood and distant to the pressing health and social care needs of their patients, many of which risk being ignored or displaced. Hence, GPs suggest a cautious approach predicated on a critical analysis of wider socio-ethical and practical implications on clinical practice and society. This coalescence of views demonstrates the value of examining imaginaries at micro levels and, understanding how ‘utopias’ and ‘dystopias’ co-exist in parallel sociotechnical imaginaries (Jasanoff, 2015). Speaking with GPs revealed how HCPs can construct and cling to fears associated with what they are told to accept as ‘utopias’.

Analysis of public statements demonstrates how the genomic medicine agenda is firmly embedded in policy and corporate imaginations as desirable, attainable and tangible (Jasanoff & Kim, 2015) futures. Consequently, the primary care imaginary, suggesting a cautious and slow introduction, is politically inconvenient and, therefore, prone to marginalisation. However, rather than seeing a contest between parallel opposing sides, we can draw on GPs’ accounts to identify key strategy features for enacting workable NHS genomic futures. Substantively, our findings suggest that socio-imaginaries may be re-imagined by attending to the complex relationship between hope, imagined futures and everyday clinical practice. Whilst approaches to sociotechnical imaginaries emphasise the power of discourse in shaping technological innovations (Jasanoff, 2015), it is critical to attend to what matters to people (Sayer, 2011), in this case, the uncertainties and potential impacts technological innovations have on resolving or exacerbating inequalities. If the policy sphere ignores these concerns as ‘merely an incidental, subjective accompaniment to what happens’ (Sayer, 2011: 2) it risks alienating those needed to implement medical technologies within clinical practice.
Finally, as Jasanoff (2005) observes, in modern society’s processes, questions of governance and epistemological debates about achieving desirable futures occur in diverse, often unseen, spaces. Reconciling divergent sociotechnical imaginaries of genomic medicine futures requires providing space for discussion between policymakers and those charged with delivering services. We need research to bring together, recalibrate and seek realistic connections and spaces that bridge ‘work as imagined’ by policymakers and ‘work as done’ in everyday health-care delivery. Further research is needed on the impact of initiatives to address knowledge deficit, capacity and infrastructure building in primary care, to facilitate nuanced understandings of genomic medicine in primary care, and to open up channels of communication between those responsible for developing services and those charged with delivering them.

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AUTHOR CONTRIBUTIONS
Shadreck Mwale: Conceptualization (lead); Data curation (lead); Formal analysis (lead); Investigation (lead); Project administration (lead); Writing—original draft (lead). Bobbie Farsides: Funding acquisition (lead); Project methodology (lead) Supervision of data collection, analysis & reporting (lead); Writing—original draft (supporting); Writing—review & editing (supporting).

DATA AVAILABILITY STATEMENT
This is a qualitative study and therefore the data generated is not suitable for sharing beyond that contained within the paper. Further information can be obtained from the corresponding author.

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