Artificial intelligence (AI) could be a great thing in medicine. It could make healthcare safer and faster. It could make medicine more satisfying to practise and less unpleasant to receive. But we must test a hypothesis before we roll it out to the public. The huge datasets being collected to use AI in diagnostic radiography, for example, are indicative of the amount and quality of work needed to make reliable, safe tools. AI must be subjected to the same criteria we’d expect with other forms of evidence based medicine.

NHS 24 in Scotland has a symptom checker app. While there’s no published evaluation, it’s based on NHS 111 algorithms, has undergone user testing, and remains open to improvements (on raising a concern about the fact that a symptom of “fever” is managed without recourse to asking about other symptoms, I was told it would be promptly reviewed). Such positive feedback loops are welcome and should be normal.

Babylon, the healthcare company that offers private and NHS GP services, has created an “NHS 111 powered by Babylon” app. It’s being piloted in north London, as I’ve discussed before, and has been extended to offer a “new paediatric symptom checker for parents.” Babies are often terrifying. They become ill quickly. They get well quickly. One small thing—a rash, a temperature, a rapid heart rate—can tip the bayesian scales, requiring a blue light ambulance. The paediatric app version isn’t yet available on the NHS, when under 17s are told to use the NHS 111 phone line, but it is available through the Babylon NHS and private service.

Knowing the staggering lack of publicly available robust testing that had accompanied the adult symptom checker app, I thought that perhaps Babylon might have done better with its paediatric one. What’s Babylon’s evidence? I don’t know, for it replied with, “we won’t be responding to your enquiry.” The binary nature of the chatbot means history taking, in the medical sense, does not happen. (“Shut up, your patient is telling you the diagnosis.”) It has a series of yes/no questions and short multiple choices.

Who’s in charge of ensuring that this app is safe and fit for purpose? The Medicines and Healthcare Products Regulatory Agency (MHRA) has said it will ask Babylon to change the way it refers to the app as being “certified as a medical device with the MHRA.” It says that, for class I devices such as this app, the manufacturer must register with the agency and self certify that the device meets regulation requirements. The MHRA says that this process is purely administrative—it takes details of the types of devices manufactured, but it does not assess, certify, approve, or accredit devices as part of the CE (European Conformity) marking process.

Who else could act? The Care Quality Commission has inspected Babylon, but it made no mention of the reliability, or not, of the app that it uses to direct people to and from GP consultations. The GMC regulates individual doctors, not clinical devices.

We have many regulators but little proactivity, even for an app which—despite the small print warning us that it “does not constitute medical advice, diagnosis, or treatment”—is being used as the front door into NHS care. AI has great potential in healthcare. But this potential will not be realised, and harm may be caused, if we don’t accept the need for robust testing before it’s publicly launched and widely used.

We have no clear regulator, no clear trial process, and no clear accountability trail. What could go wrong?

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When the midwife visited the home of Sinthiya Rajatheepan the day after she gave birth, she found the baby boy lying in bed, pale and lethargic. Unfed for nearly 16 hours, he was hypoglycaemic. Although he was rushed to hospital, he developed cerebral palsy and severe disabilities.

No one at the hospital had explained to Rajatheepan, who had just turned 21, how to feed her baby properly and what to do in the event of poor feeding.

On the day of her discharge from hospital the baby had been crying all day. None of the midwives, however, had paid much attention to mother or baby. The ward had 26 beds but just two midwives and a support worker. On discharge, Rajatheepan, a Sri Lankan national who spoke very little English, was given a folder full of papers. She did not read them, nor did she understand the 20 minute discharge discussion with the midwife. When her husband and a family friend, who had arrived at the hospital after that discussion, expressed concern about the baby’s constant crying, they were reassured by the staff that it was normal for newborn babies to cry.

On 13 April 2018 Judge Martin McKenna found the NHS trust negligent, concluding, “The reality is that no one had ever in fact given Mrs Rajatheepan a clear and understandable explanation of the importance of feeding, still less as to how she should respond if she had concerns. Because of the language barrier, Mrs Rajatheepan had been unable to communicate her concerns to hospital staff and when those concerns were communicated on the parent’s behalf by Mr Gunaratnam [a family friend] they were not acted upon.”

**ETICS MAN** Daniel Sokol

**Patience for patients**

Practitioners must resist the temptation to take shortcuts when dealing with people whose English is poor.

**ACUTE PERSPECTIVE** David Oliver

**Tube feeding—guidelines can take us only so far**

Like most geriatricians, I look after many patients with progressive vascular or Lewy body dementia, severe disabling stroke, near end stage Parkinson’s disease, and other progressive neurological conditions.

The natural history of patients with such conditions is that many run into problems with swallowing and lose cough strength. They’re at risk of aspiration pneumonia and of poor nutrition. They may find it hard to take key medicines, which can further worsen swallowing, alertness, or overall medical fitness.

In some families or cultures the idea of withdrawing intravenous fluids or artificial nutritional support, and focusing on palliative care, can require several sensitive and lengthy conversations, over days or weeks, for clinicians and family to arrive on the same page. It’s sometimes necessary to carry on with interventions while that joint understanding is reached. We’re often under pressure from families to use tube feeding and to continue intravenous fluids—or to let patients continue to take food and drink by mouth when it has become very risky.

There’s no shortage of literature and official guidance on when tube feeding or its withdrawal is most clinically appropriate or effective or most legally or ethically permissible.

In the disease trajectories I’ve described, patients are likely to become progressively disabled and have repeated infections and admissions, even if long term artificial nutrition and hydration are provided by percutaneous endoscopic gastrostomy (PEG) or radiologically guided gastrostomy (RIG). For instance, a systematic review of PEG feeding in patients with severe dementia found no improvement in survival and showed high complication rates. And these procedures carry a high risk of mortality in patients with severe disease and poor physical reserve.

The more temporary use of nasogastric feeding is not without complications, including aspiration, fluid overload, discomfort, and tubes requiring repeated reinsertion. I don’t know many doctors in my field who would want to subject patients in the last year of life to potentially burdensome, distressing, or invasive treatment such as tube feeding.

However, my colleagues and I have also been in situations where, even if PEG or RIG isn’t safe or desirable,
There can be no respect for patient autonomy if the patient cannot understand what we say

eyed the language barrier through gestures and sign language.

When I taught medical students, I would ask them to name the virtues of a good doctor. Out came “competence,” “fairness,” “honesty,” “kindness,” and “compassion.” I cannot recall, however, anyone saying “patience.” Yet this may be a key virtue when dealing with patients with poor English.

Life on the ward, or in the GP surgery, is so busy that the temptation for healthcare staff is to ignore, deliberately or subconsciously, the linguistic struggles of foreign patients. To acknowledge them would mean spending much longer on the task, whether to set up a language line, arrange for an interpreter in person, or even modify their own speech to increase comprehension. It can also be tedious or frustrating.

We all know that ignoring a patient’s language difficulty is wrong. There can be no respect for patient autonomy if the patient cannot understand what we say. At times, as in the sad case of Rajatheepan, patients may come to harm if they misunderstand instructions.

**Silence should be a trigger**

The hardest part is reminding ourselves not to take shortcuts when we encounter a language barrier, however tempting they may be. It is much easier to assume the patient understands, or to ignore a patient’s blank expression and other telltale signs of linguistic confusion. Silence should be a trigger to test comprehension.

Once doctors are aware of a possible language barrier, and the innate inclination to ignore it, they can then remind themselves of the neglected virtue of patience and ensure that patients understand their words of wisdom.

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**Broken legs: the lodestar of mental health outcome measures**

Outcomes in mental health are getting a lot of attention. It’s a troublesome concept for us—how do we measure what we do? Measurement is central to quality improvement. In order to consider whether the work we are doing is having an impact, we need an indicator. Otherwise how will we know if anything is happening?

I find myself talking about broken legs when I’m explaining the challenge—though, to be fair, it’s been a while since I’ve seen one. The analogy is that a broken leg is either broken or it isn’t. Most people (including the patient) can agree whether it’s broken. The treatment is clear, the course of recovery is predictable, and it is obvious when no further treatment or care is needed.

I’m aware that this is a massive oversimplification. But contrast it with mental health problems. Where is the cut-off between misery and depression? Where does “normal” anxiety end and an anxiety disorder begin? How do you describe the challenges someone with a personality disorder faces?

The way we are trying to tackle this (and how we are being guided by NHS England) is similar to how other branches of medicine approach it; in using PROMs (patient reported outcome measures), PREMs (patient reported experience measures) and CROMs (clinician reported outcome measures). Some infrastructure is already present for us to be able to do some of this—we use the friends and family test, for example, which is a type of PREM. We even use a CROM—the health of the nation outcome scale (HoNOS)—as part of our arrangements with commissioners. This is used as part of the national “care cluster currency” in the NHS in England, and data must be submitted by all providers for adults to NHS Digital as part of the mental health dataset.

As yet, however, there is no universal national PROM, and one can see why. Given the diversity of problems we tackle in mental health, it is difficult to find a one-size-fits-all tool that can cover the whole population. And the need for agreement is pressing.

It’s no coincidence that the five year forward view for mental health saw specific investment go to areas that are relatively further ahead when it comes to evidencing what they do. Once our range of outcomes are agreed, we need to mobilise to make sure we measure them regularly and reliably, record them in a way that is accessible, and be able to make use of them meaningfully.

We’ve a lot to learn from broken legs that have gone before, but patients and carers deserve that we get this right.

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**Where is the cut-off between misery and depression?**

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**Broken legs:**

**Billy Boland**

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**BMJ OPINION**

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The 100 000 Genomes Project: bringing whole genome sequencing to the NHS

In partnership with NHS England, Genomics England’s ambitious plans to embed genomic medicine into routine patient care are well under way. Clare Turnbull and colleagues discuss its progress.

KEY MESSAGES
- The 100 000 Genomes Project has established delivery of whole genome sequencing in the NHS
- The project has driven transformation of local systems at participating centres, including tissue handling, collection of data, and result processing
- NHS England is establishing a genomic medicine service to deliver systematic access to genomic tests, including whole genome sequencing
- Whole genome sequencing can enable rapid diagnosis in children with rare disease
- Whole genome sequencing of tumour tissue can inform selection of treatments for cancer

Many disorders we encounter in clinical medicine have a genomic basis, from rare “single gene” disorders such as cystic fibrosis, to complex, polygenic disorders such as ischaemic heart disease, drug toxicity, and tumour evolution driven by serial somatic mutations.

Next generation technology has transformed the capacity, speed, and cost of genomic sequencing. This has provided important advances and new opportunities for the clinical application of genomics. However, radical expansion of genomic medicine requires new infrastructure, extended skills, workforce education, and diligent engagement with the public. The Genomics England 100 000 Genomes Project was initiated in 2013 to establish the use of whole genome sequencing in the NHS and drive change within services to adopt this technology.

Transforming genomics in UK
The UK has long been at the forefront of discovery in human genomics and is recognised for its world leading genetic research studies, such as UK Biobank and Deciphering Developmental Disorders. In parallel, the UK has evolved a mature network of NHS funded regional genetics laboratories and clinical genetics departments.

Until recently, genomic technologies available in the clinic have enabled us to look for the “causative mutation” just one segment of a gene at a time, limiting both the speed and volume of clinical testing. Over the past decade, next generation sequencing has made it possible to sequence millions of fragments of DNA simultaneously. This step change in scale enables us to offer genetic testing to many more people and test one person for hundreds or thousands of genes at a time. Indeed, while the initial sequencing of the full human genome took more than 10 years and cost more than £2bn, an individual’s genome can now be sequenced in around a day at a cost of less than £700.

To harness the new possibilities availed by this technology shift, successive governmental strategy reports have emphasised the need for new approaches to delivering genomics services. These reports have called for centralised provision of whole genome sequencing and related (bio)informatics to improve cost effectiveness and adaptiveness, a national database of genomic information (and associated clinical data) that is accessible throughout the NHS, as well as expansion of the workforce and improved genetic literacy across the clinical workforce.

In 2012, the then prime minister, David Cameron, announced funding for whole genome sequencing of 100 000 genomes from patients in the English NHS to capitalise on the potential of this technology for patient benefit. Genomics England, owned by the Department of Health and Social Care, was set up to deliver the project working in partnership with NHS England. Rare disease and cancer were selected as the areas that had the most immediate potential for clinical benefit from whole genome analysis.

Rare diseases and the diagnostic odyssey
The project’s rare disease programme was established to initiate and embed use of whole genome sequencing within the NHS to identify genetic causes in people with rare inherited diseases (box 1). Clinicians and researchers nominated more than 200 recruitment categories (spanning over half of the roughly 7000 recognised rare diseases) that were deemed underserved by current clinical diagnostic testing or required further research to elucidate their genetic basis.

Historically, the “diagnostic odyssey” in a child with a rare disease could span several years. Children would have investigation of multiple organ systems by different specialists, and even after referral to clinical genetics, serial testing of different genes (often at different laboratories) could take years. Sequencing of the coding regions of all 20 000 genes by whole exome or genome sequencing eliminates reliance on the clinical hypothesis.
NEWS 2 AND SEPSIS

NEWS 2 is not validated in primary care

Inada-Kim and Nsutebu urge the health system to use the NEWS2 score in all settings (Personal View, 24 March). They say that “a score in one setting must mean the same in any other.” The problem is that it doesn’t.

Improved communication in health systems should be encouraged, but as Bernard Shaw (maybe) said about the UK and US, primary and secondary care will always be separated by the same language.

A NEWS2 score of 4 in an emergency medical setting does not mean the same as in a GP surgery, for the simple reason that serious illness is not as common.

Clinicians should observe patients and be alert to sickness, but we should acknowledge that the NEWS2 score is not validated in primary care, let alone as a screening tool. Until it is, we must accept that the NEWS2 score from primary care is spoken with a different dialect.

Alex Burns, GP, Truro
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ANTIDEPRESSANTS

Should we rethink treatment duration?

McCormack and Korownyk say that, “if 10 patients with moderate to severe depression take an antidepressant for two months, five will report being ‘better’ but in four of them the response will not be because of the drug” (Editorial, 24 March).

Yet NICE guidance says that a person in remission from an episode of depression should continue medication for at least six months.

The implication is that the four patients who coincidentally got better while on antidepressants will subsequently receive at least 24 months, in total, of unnecessary treatment, which is a major cost (in all senses) to them and to the NHS.

Should we rethink long courses of antidepressants? Stopping treatment after two months would prevent an unnecessarily long course in four of five patients, but risks relapse in the patient who genuinely has benefited. Is this a trade-off we should be considering or discussing with our patients?

Keith Hopcroft, GP, Basildon
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CERVICAL MYELOPATHY

Mobile phone sign

A key feature of degenerative cervical myelopathy (Easily Missed? 24 February) is clumsy hands, and all my recent patients have told me that they keep dropping their mobile phone. Not only is the phone badly damaged but it is also in a robust case that their children have bought for them to try to protect it.

Degenerative cervical myelopathy should be suspected in all patients who have recently started dropping their phone regularly to the point that their family have become concerned.

Anthony N Williams, consultant occupational physician, Temple Ewell
Cite this as: BMJ 2018;361:k1713

Arm and leg reflexes

Davies et al suggest that arm reflexes are exaggerated in degenerative cervical myelopathy (Easily Missed? 24 February). I find that they tend to be diminished (lower motor neurone lesion), whereas leg reflexes alone tend to be exaggerated (upper motor neurone lesion).

Patients who present with neck symptoms should have a full neurological assessment of both the arms and the legs. Patients with exaggerated leg reflexes should have cervical MRI to look for cord compression before any forms of physical treatment to the neck are considered to avoid further compression.

Andrew Rowe, chiropractor, Abingdon
Cite this as: BMJ 2018;361:k1714

Author’s reply

Our patients with degenerative cervical myelopathy do report dropping their mobile phones, but we have not assessed their phone cases. We would be delighted to work with Williams in assessing the clinical relevance of his observation, starting with the experience of the myelopathy.org community and that of our patients.

We agree that patients with neck complaints should have a full neurological examination and that exaggerated leg reflexes should prompt cervical MRI to look for cord compression before physical treatment to the neck is considered. Examination findings are variable, but the high incidence of arm hyper-reflexia is published. One study found the incidence of arm hyperreflexia was around double that of hyporeflexia.

Benjamin Davies, neurosurgeon (specialist registrar), Cambridge
Cite this as: BMJ 2018;361:k1718
Doctors need to give up professional protectionism

Rather than resisting, doctors need to welcome the host of new clinicians

The need for the medical profession to protect its role was, until recently, deemed essential. The notion that others might take on some tasks traditionally thought of as being “doctors’ roles” was regarded as an existential risk to the medical profession.

Medical societies, colleges, unions, and regulators approached proposals of clinical practice by other health professionals with deep suspicion. Concerns were often couched hubristically in terms of patient safety. But the prime motive was a fear that the profession would be diminished.

If there were a shortage of work for doctors to do, then such attitudes would be a rational response. But these protectionist attitudes persisted long after the workload per doctor had become excessive. They have continued even when doctors’ workloads have become impossible. New clinician roles have not arisen out of some grand plot to do doctors down but from the need to deal with the workload crisis.

A junior doctor is a pluripotential clinician, a creature of learning and skill who is not yet fully differentiated into whatever their career may deliver. Conversely the new roles of nurse specialists (for heart failure, epilepsy, Parkinson’s, and other conditions) and nurse practitioners (emergency and critical care practitioners, and advanced clinical practitioners) are differentiated and settled.

Doctors provide continuity for services and patients, while nurse specialists and practitioners are an invaluable addition to the urgent and emergency care workforce. Medical associate professionals, critical care practitioners, and surgical and anaesthesia practitioners deliver procedure based expertise. Not one of these practitioners has made a doctor redundant, nor diminished one single doctor or the profession as a whole. Over the past 10 years the number of doctors on the GMC specialist register has risen from 60,000 to 90,000. Medical unemployment is a conjecture.

Experience devalued

Recognition of the value of these roles is, belatedly, a welcome reaffirmation of the benefit of experience. Since the introduction of Modernising Medical Careers in 2005, experience has been devalued in medical roles, albeit unintentionally. It has been sidelined by assessment, appraisal, and reflection. In contrast, most appointments to the new clinician roles are predicated on experience and encourage long term careers. The stable workforce that arises advantages patients and doctors.

Sadly, doctors have done little to support these new groups, which have prospered despite the medical profession’s indifference and antipathy. Notable exceptions are the establishment of a Faculty of Physician Associates by the Royal College of Physicians (London) and the credentialing of advanced care practitioners by the Royal College of Emergency Medicine.

Demographic change is the biggest challenge we face in healthcare. The over 85s are increasing by around 2000 a month, of whom 25% are moderately to severely frail. Care of this cohort requires frailty nurse specialists, clinical pharmacists, occupational therapists, and physiotherapists. Enabling these professionals to start (and, perhaps more importantly, stop) medication, request plain radiographs, and determine safe discharge is the only way we can hope to meet the care needs of our grandparents, parents, and, in time, ourselves.

Changing boundaries of practice

One perfectly legitimate concern is the issue of scope of practice. This again has its roots in a notion of medical omnipotence and unbounded expertise that wasn’t credible even when the sum total of medical knowledge could be contained within a few large books on a very small shelf. All doctors have a clinical remit of varying radius and, in general, depth has supplanted breadth, of knowledge and of practice.

As the radius of practice has decreased, so more gaps have appeared in the fabric of healthcare. These gaps represent patients’ needs and employment opportunities. In filling these gaps—whether with more doctors or with new types of clinician—the boundaries of practice must be clearly defined.

It remains the responsibility of the regulatory authorities to police and enforce these boundaries of practice. Regulation is therefore an important issue; medical associate professionals need a regulatory home. The GMC has proposed to undertake this role in response to a 2017 government consultation. Now is the time for those of us who provide the GMC with its income to endorse, encourage, and expect our fellow practitioners to be overseen by our regulator.

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to select which genes to test and has enabled diagnoses of many previously unsolved cases.  

**Whole genome sequencing in cancer**

Whole genome sequencing of cancer tissue can provide information on cancer aetiology, prognosis, and potential therapeutic responsiveness (box 2). Procurement of tumour DNA of sufficient quantity, quality, and purity has often limited clinical and research tumour sequencing to date. The 100 000 genomes cancer project has collected a broad range of early stage and advanced solid tumours from diagnostic biopsy and surgical resection samples, as well as haematological malignancies. In current clinical testing, multiple standalone tests are used to capture the set of genomic biomarkers examined for a given tumour type, but the falling cost makes whole genome sequencing potentially attractive as a single all-encompassing test.

**An individual’s genome can now be sequenced in around a day at a cost of less than £700**

**How is the project being delivered?**

Thirteen centres across England with established expertise in molecular genetics, clinical genetics, molecular pathology, and molecular oncology were established by NHS England as NHS genomic medicine centres. These hub hospitals link to more than 90 local recruiting hospitals, providing substantial national coverage.

Genomics England has developed platforms and automated pipelines for processing, calling, quality checking, storing, presenting, annotating, and prioritising the variants identified at sequencing. In rare disease, the family set of genomes is analysed as a group, with four to five million variants identified in each individual. Algorithms incorporating variant frequency, familial inheritance, variant impact, and gene-phenotype association are applied to sort the identified variants into four groups according to the likelihood that they are causative (tiers 1-3 and untiered). Each genomic medicine centre has established multidisciplinary meetings to bring together laboratory, clinical genetics, and medical subspecialty expertise, variably organised at local, regional, and national level.

In the whole genome analysis for cancer, established knowledge bases are used to assess the potential diagnostic, predictive, or prognostic value of the identified somatic variants. Variants are highlighted to indicate suitability for NICE approved targeted drugs as well as eligibility for genomically stratified UK clinical trials. A full analysis of tumour structural and copy number variation is also presented, as well as other findings such as pan-genomic signatures and mutational burden. Tumour sequencing boards have been set up at each genomic medicine centre to bring together laboratory scientists, oncology clinicians, national and local clinical teams, and some patients.

**Box 1 | Rare disease and genomics**

- Rare diseases are defined as those affecting <1 in 2000 people. Over 7000 rare disease entities have been described, estimated to affect a total of around three million people (1 in 17) in the UK
- About 75% of these diseases manifest before the age of 5 years. They are typically life shortening and confer serious disability
- Most are due to single gene defects (so called monogenic or mendelian diseases)
- A robust genetic diagnosis in rare disease can be critical to management. The specific genetic diagnosis enables the clinician to apply the therapies and interventions most likely to be effective, give a best estimate of prognosis, predict additional features, and pre-empt complications
- Genetic diagnosis can also provide information to the family about likelihood of recurrence in subsequent pregnancies as well as options for pre-implantation or prenatal genetic diagnosis

**Box 2 | Cancer genomics and molecular oncology**

- Cancer is a disease of disordered genomes: acquisition of serial genomic mutations results in progressive escape from the mechanisms that regulate cell division, leading to tumourigenesis, invasiveness, and metastasis.  
- Paired sequencing, subtracting the normal genome (eg, of the blood) from the tumour genome enables identification of the acquired mutations in the tumour, from small mutations in genes (base substitutions and deletions/insertions) to larger structural variants (translocations, large deletions, or duplications resulting in amplification).  
- Signatures (complex mutational patterns) can also be extracted from analysis across the whole genome. Trials are evaluating performance of these signatures to predict drug response and tumour behaviour  
- Technologies are evolving rapidly for genomic analysis to detect minuscule levels of cell-free circulating tumour DNA (cfDNA) in the bloodstream before the tumour becomes clinically or radiologically obvious. Current clinical evaluation is largely focused on early detection of tumour recurrence, but there is substantial interest in using the technology for primary screening for cancer.
pathologists, and germline cancer geneticists to advance molecularly driven patient management.

Accessing genome data for research
Individual identifiable data are available only to registered clinical users working within the NHS genomics medicine centres. De-identified, individual clinical and genomic data for research use are held within the Genomics England research environment. Access to the 100000 Genomes Project data is controlled through robust authentication systems and an “airlock” mechanism ensuring that only summary level data can be removed. Academic researchers can access the research environment through membership of one of the 42 Genomics England clinical interpretation partnership domains or through specific collaborations with approved industry partners.

Routinely collected national datasets, including Cancer Registry datasets and Hospital Episode Statistics, are regularly merged to the genomic data (at individual level). The linked longitudinal life course datasets are currently immature but will eventually facilitate analyses for associations of genomic factors with longer term outcomes.

Challenges, progress, and evolution
As of April 2018, more than 70 000 participants have been recruited to the project, more than 55 000 whole genomes sequenced, and more than 10 000 whole genome analysis reports have been returned to the NHS. Various challenges have had to be overcome on the way.

Tissue for cancer genome sequencing
Formalin fixation and paraffin embedding (FFPE) has been used for more than 100 years to prepare tumour tissue for microscopy. Formalin causes severe degradation of DNA, affecting the fidelity of the genomic readout (especially for large structural variant calls in whole genome sequencing). Fresh tumour tissue provides much higher quality results but processing, transporting, and storing it across diverse settings has been a sizeable challenge, requiring new practices such as vacuum packing, tissue refrigeration, and use of novel coolants and transport media.

Standardisation of submitted clinical data
The automated genomic analyses for cancer and rare disease require input of standardised clinical data, for which Genomics England has developed data models using internationally established nomenclature systems, such as the Human Phenotype Ontology. Obtaining complete clinical data in a consistent format has been challenging because of diverse electronic medical record systems, together with competing demands for local informatic resources. Substantial investment in local informatics and collaborative approaches across trusts working with NHS Digital, NHS England, and the Farr Institute have driven novel solutions.

Consistency in interpretation of genomic variants
Computational prediction tools, variant databases (commercial and academically maintained), and functional validation assays are all improving. However, determining whether a genomic variant is benign or pathogenic can be complex and interpretations of a specific variant are often inconsistent. The automated genomic analyses for cancer and rare disease require input of standardised clinical data, for which Genomics England has developed data models using internationally established nomenclature systems, such as the Human Phenotype Ontology. Obtaining complete clinical data in a consistent format has been challenging because of diverse electronic medical record systems, together with competing demands for local informatic resources. Substantial investment in local informatics and collaborative approaches across trusts working with NHS Digital, NHS England, and the Farr Institute have driven novel solutions.

Diagnostic rate
The initial overall diagnostic rate in the rare disease programme from preliminary central review of tiers 1 and 2 small variants is 22%, with diagnostic rates higher in certain disease categories such as intellectual disability. This diagnostic rate will increase after feedback from detailed local clinical review and analyses of additional variant types such as copy number variants.

New NHS genomics medicine service
The 100 000 Genomes Project has catalysed evolution of informatics infrastructure, development of data pipelines, expansion of workforce capacity, development of skills in sequencing technology, and new professional networks and ways of working. Health Education England has also developed genomics training for the wider NHS workforce (box 3). The project is nearly finished, but in late 2018 whole genome sequencing will become part of an NHS England commissioned national genomics medicine service for rare inherited disease and cancer. The service will provide centralised accredited whole genome sequencing, with the results returned to a network of genomic laboratory hubs, where other genomic tests will also be done. NHS England will publish a national genomic test directory, linked to a national system for ordering genetic tests. This will support more systematic access to genomic testing across the country, with capture of clinical outcomes to enable ongoing evaluation.

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David Delvin

GMC rebel and bestselling author

David George Delvin (b 1939; q 1962; MRCS Eng, DObst RCOG, DCH Eng, Cert FPA, MRCGP, Instruc Doctor's Certif, Dip Ven Soc Apoth Lond, MFFP), died from cutaneous T cell lymphoma on 9 March 2018.

Every doctor writing for the media today owes a great debt to David Delvin—one of the few doctors who has been both the subject of a complaint to the GMC and a council member. As a young doctor in the 1960s, he developed a passion for writing, but the deeply conservative GMC was only too ready to rule that any doctor exposing themselves to the limelight of publicity was self advertising. Doctors were justifiably afraid about either talking to journalists or writing for newspapers or magazines.

GMC complaint

But Delvin reflected the values for which the “swinging sixties” are now remembered. The revolt of the age was symbolised by student uprisings, the civil rights movement, the anti-Vietnam war movement, women’s liberation, and the advent of the contraceptive pill. Nothing was sacred any more.

Delvin was adamant that doctors should not be compelled to write under pseudonyms, but was understandably alarmed when the GMC informd him, in 1974, that a complaint had been laid against him, alleging self advertising. Doctors were justifiably afraid about either talking to journalists or writing for newspapers or magazines.

He combined general practice, sexual health, and family planning and journalism with a heavy emphasis on sex

“Medical students had no teaching at all about it, except one lecture on the cap, and one on abortion, infanticide, and rape.” Sexual medicine attracted him, he said, because “you could put things right.”

After working with the Family Planning Association and the Institute of Psychosexual Medicine, Delvin became a founding fellow of the Faculty of Family Planning of the Royal College of Obstetricians and Gynaecologists. His 34 books included The Book of Love, which sold more than a million copies and became a family planning textbook. Translated into more than 10 languages, as well as braille, it won the American Medical Writers’ Association’s best book award.

Delvin was also bewildered to be awarded a medal (Medaille de la Ville de Paris, echelon argent) by French president Jacques Chirac. A former chair of the MJA, Delvin also appeared on more than 930 TV programmes.

He counted among his career highlights his tenure as the columnist Dr Jekyll in World Medicine, edited by Michael O’Donnell, one of his staunchest allies in his stand against the GMC. As the broadcaster and fellow MJA committee member Geoff Watts reported, Dr Jekyll took bawdy delight in demonstrating that the magazine’s highly qualified and intellectually gifted readership had a taste for salacious knockabout humour.

In 1988 Delvin married fellow writer and TV presenter Christine Webber, whom he had met at Anglia TV. They collaborated on many books and journalistic and broadcasting projects. He leaves three children and seven grandchildren.

John Illman, London

Cite this as: BMJ 2018;361:k1263