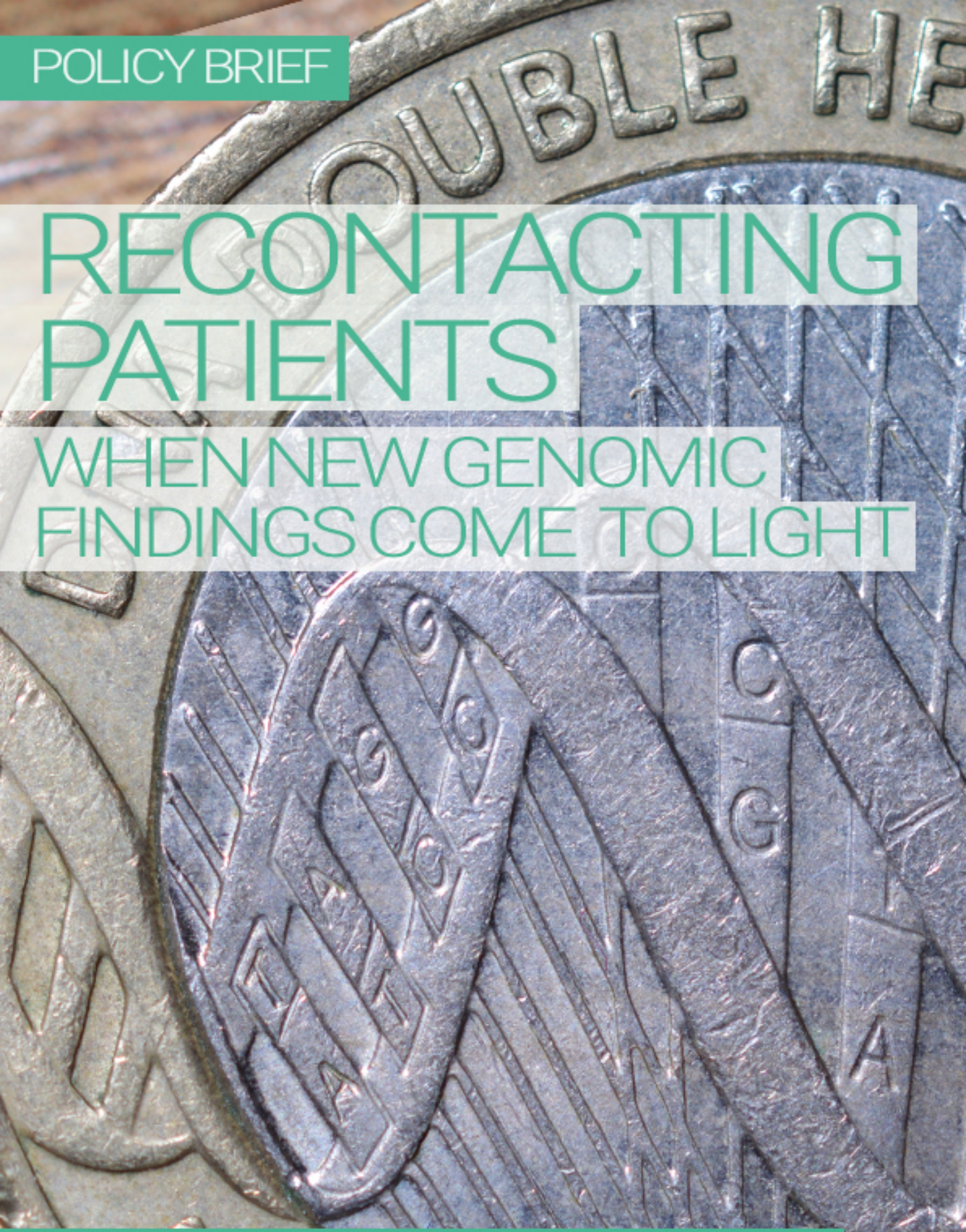
POLICY BRIEF

RECONTACTING PATIENTS

WHEN NEW GENOMIC FINDINGS COME TO LIGHT



University of Exeter

Cardiff University / Prifysgol Caerdydd

University of Southampton

Royal Devon and Exeter NHS Foundation Trust

BACKGROUND

Advances in genetic and genomic medicine, their reduced cost, and greater integration into medical practice – i.e. the mainstreaming of genomics – are generating **new information** about predispositions, diagnosis and treatment of some health conditions. This information is new because (a) more of a genome can be analysed routinely than previously possible – identifying new genetic predispositions, and (b) international collections of such data are providing more robust information about risks of disease than previously possible. This is happening **very rapidly**, projects such as the 100,000 Genomes Project have been set up to drive the 'mainstreaming agenda' with the goal of developing a genomic medicine service in the NHS to benefit patients. Detailed genomic information has the potential to allow more accurate diagnosies, improve preventative therapies or surveillance regimens, and tailor treatment to a patient's individual genetic makeup.

As more evidence is accrued, an increasingly important question arises about **if and when patients seen in the past should be recontacted to update them on these changes in technology and interpretation.** Where new genetic/genomic information could have significant implications for the health of patients and their relatives, their reproductive and lifestyle choices, employment, and psychosocial wellbeing, there is a clear benefit to recontacting them. However, how strong does new evidence have to be, how likely a diagnosis will be achieved, or how well a predisposition can be treated are important factors in any recontact policy. There is currently no professional consensus about recontacting in clinical practice, and very little guidance or empirical evidence about healthcare professionals' and patients' views on this issue. This brief provides policy recommendations based on the findings of the first UK research project to investigate these issues.

KEY FINDINGS

Patients view recontacting as desirable and as a sign of good quality of care, leading to improved health outcomes.

Recontacting does happen in the NHS and healthcare professionals report that this is mostly when new clinically "actionable" information becomes available; however there are no standardized practices or systems in place.

Recontacting thus raises the broader issue of equitability of current healthcare service provision in that there is no systematic way to ensure that all patients who may benefit from new information are identified and recontacted.

Healthcare professionals consider that the recontacting of patients is both ethical and legal, but some are concerned they may be held legally liable whether they do or do not recontact patients.

Both healthcare professionals and patients express concerns about the feasibility of routine recontacting within the current resource constraints of the NHS, and about a lack of clarity over roles and responsibilities. This lack of clarity is further complicated by the fact that genetic testing is increasingly offered by specialties that do not necessarily have an ongoing relationship with a patient.

Healthcare professionals see the need for a professional consensus and guidance about whether and in what circumstances to recontact, and whose responsibility this should be.

Healthcare professionals regard multi-speciality collaborations as one of the most effective ways to reduce misunderstandings about who to recontact. To date, decisions made by genetic healthcare professionals about whether – and how – to recontact patients have involved such multi-speciality collaboration.

POLICY RECOMMENDATIONS

Recontacting is relevant to major policy developments within the NHS – such as mainstreaming of genetic medicine, data sharing, public health, and e-health records – and crucial to its sustainability and performance. Considering future recontact practices provides an excellent opportunity to focus on long term strategies (as opposed to the prevalent 'short-termism' in the NHS) for the delivery of an efficient and sustainable health service. Appropriate recontacting practices will likely assist in increasing patients' trust in the NHS, which is key to the delivery of these pressing policy issues.

The four Departments of Health across the UK Nations should jointly:

Establish a professional working group to reach a consensus about the circumstances under which patients should be recontacted, the lines of responsibility that may ensue, and the pathways required.

Ensure that any resulting **recommendations are adequately resourced** so as not to compromise excellent, equitable clinical care due to the redirecting of existing funds and resources.

Involve healthcare professionals from different specialities in order to build upon the existing model of Genomic Medicine Centres to establish and improve the much needed multi-specialty collaboration required for the effective implementation of genomic medicine.

The need for a more effective multi-specialty collaboration highlighted by healthcare professionals in the current study is in line with international standards and guidelines for the interpretation of sequence variants – specifically that "clinical laboratories are encouraged to establish collaborations with clinicians". This guidance was also endorsed in the UK by the Association for Clinical Genetic Science in 2016, but extensive multi-specialty collaboration is still in its infancy.

The recent update to the NHS England Five Year Forward View emphasises the importance of harnessing technology and innovation, citing Genomic Medicine Centres (GMC) as a successful example. The GMCs are a multi-disciplinary infrastructure, bringing together clinicians, scientists, nurses, administrators and the academic health science network to deliver the 100,000 Genomes Project. Currently the teams are relatively small and rely on the wider healthcare community to propagate the message of genomics, but represent a good model structure for the implementation of genomic medicine into the NHS if adequately funded and resourced. The length of time between providing a sample and receiving a result means that the GMCs are already dealing with some of the logistical challenges presented by recontacting. Genomic Multi-Disciplinary Team meetings are being established to interpret the results and again this structure can be harnessed for uses beyond the project.

The huge research effort committed to variant interpretation and new gene discovery embedded in the 100,000 Genomes Project also means that the GMCs will need to develop pathways to update participants about any changes to their results in the future. However, the future of the 100,000 Genomes project beyond 2019 is still uncertain. We recommend leveraging existing GMCs set up by ensuring the financial viability and continuity of these centres, expanding them to areas not currently covered, and ensuring they remain integrated with regional genetic services.

EXAMPLE CASE

A new gene is discovered which, when mutated, provides a possible explanation for a group of previously undiagnosed patients. Each genetic service probably has about 50 patients they have seen over the years who might have this diagnosis. Whilst health professionals will bear this new diagnostic possibility in mind with any new referral, they do not have the time/funding/IT support to track back through historical patients who might have this diagnosis.

CONTACT

Professor Susan Kelly
Egenis, University of Exeter, UK
01392 725139
s.e.kelly@exeter.ac.uk
recontact-genomics@exeter.ac.uk

Project outputs including the evidence upon which this policy brief is based available here:

http://bit.ly/PPSGenomics

Produced by Public Policy|Southampton
www.southampton.ac.uk/publicpolicy