

## **Recontact in clinical practice: a survey of clinical genetics services in the UK**

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### **ABSTRACT**

**Purpose:** To ascertain whether and how recontacting occurs in the UK.

**Method:** A web-based survey was administered online between October 2014 and July 2015. A link to the survey was circulated via an email invitation to the clinical leads of the UK's 23 clinical genetics services, with follow-up with senior clinical genetics staff.

**Results:** The vast majority of UK services reported that they recontact patients and their family members. However, recontacting generally occurs in an *ad hoc* fashion when an unplanned event causes clinicians to review a file (a 'trigger'). There are no standardised recontacting practices operating in the UK. More than half of the services were unsure whether formalised recontacting systems should be implemented. Some suggested greater patient involvement in the process of recontacting.

**Conclusion:** This research suggests that a thorough evaluation of the efficacy and sustainability of potential recontacting systems within the national health service would be necessary before deciding whether and how to implement such a service or to create guidelines on 'best practice' models.

**Key words:** recontact; next generation sequencing; ethics; clinical genetics; survey

## **INTRODUCTION**

### **The problem**

The increasing introduction of new genetic technologies in the investigation of patients is creating much new information. This raises important issues related to the communication of the potential health significance of new findings (e.g., new information about the natural history of a condition; surveillance or treatments available; improved diagnostic accuracy, such as a new test; or new information about previously uncertain test results, such as classification of a VUS - variant of unknown significance). This might mean that patients seen and tested in the past could now be offered more informative testing. As a result, questions may arise about whether healthcare professionals, such as clinical genetics specialists, have a responsibility or duty to recontact former patients. Innovations in genomic medicine can have significant implications for patients and families as regards health, reproductive decisions, lifestyle choices, employment, and psychosocial wellbeing. However, recontacting patients may also affect them negatively, potentially causing anxiety and concerns over health and economic activity, and being experienced as an intrusion into privacy [1]. While recontacting patients has been raised as a major issue in medical genetics, its importance will become even more widespread with the increasing integration of genomics in medicine [2]. Clarifying the issue of whether and how recontacting in clinical genetics should be implemented is of importance to the current information revolution in healthcare [3].

### **Policy/guidelines**

There is no professional consensus in clinical genetics about whether, and how, former patients should be recontacted when new genetic information relevant to them or their family members arises from the use of new technologies. A survey of regulations and

practices of genetic counselling in 38 European countries found that recontacting was amongst the least covered topics in both national legislation and applied practice guidelines [4].

The only guideline currently available is a statement originally published in 1999 by the American College of Medical Genetics [5]. This document highlights the logistical difficulties of locating and recontacting former patients, and identifies the primary care provider (the specialist tasked to provide continuing care, such as the GP in the UK NHS) as the principal responsible healthcare provider to alert their patients to the need for recontact if necessary. Genetic service providers would be responsible to give clinical updates to patients in the rarer cases in which they are offering continuing care. The statement also suggests that patients should be appropriately advised to update their primary care provider or the genetic service provider if relevant changes in their lives occur, such as pregnancy [5]. The 2008 revision of the statement recognises that with the uptake of next generation sequencing, testing laboratories may now be in a position to know about changes in interpretation of variants whose significance had previously been unknown (former VUS), or about reclassifications of previously classified variants – and should make an effort to contact relevant healthcare providers if new information changes the previous clinical interpretation of a sequence variant [6].

### **Systematic review of recontacting literature**

The only systematic review of recontacting literature [7] identified 61 articles published between 1991 and 2014 which explore ethical, legal, social, psychological, clinical, and practical issues. The review identified a divergence of expectations about who is responsible for keeping patients up-to-date with relevant information. While patients tend to assign this responsibility solely to healthcare professionals – especially genetic service providers –

healthcare professionals tend to think this responsibility should also be shared by patients [8-10].

The review also showed that although most patients want to be recontacted, not all do, because of the potential for strong emotional reactions to such recontact [11, 12].

Most authors identified a clash between the ethical desirability and the practical difficulty of recontacting. The most common practical barriers to recontacting mentioned are lack of: infrastructures for tracking data of former patients (e.g., digitalisation of databases) [13]; time and resources (e.g., staff, money) to perform recontacting [8, 9, 14]; not having patients' current address details [15].

Suggestions to overcome these barriers include: implementing digital communication systems between laboratory, clinicians, and patients [14]; involving patients in the processes of recontacting [10, 16]; involving patient support or advocacy groups [8, 17].

### **Need for more empirical evidence**

The authors of the systematic review suggest that more empirical evidence is needed to advance the discussion over whether and how recontacting should be implemented [7].

There is limited empirical evidence concerning the perspectives of healthcare professionals and patients on recontacting or what is occurring in clinical practice. There is an urgent need for more research on the practical and ethical implications for healthcare systems of recontacting recommendations or guidelines. To begin to provide such empirical evidence, we have conducted a survey of clinical genetic services in the United Kingdom about their current recontacting practices.

## **METHODS**

### **The Survey**

The survey we conducted is part of an ongoing study which investigates the place of recontacting in current clinical practice in the NHS in the UK; the ethical, legal and social issues raised; and the expectations of patients and healthcare professionals concerning recontacting (Study webpage: <http://ex.ac.uk/mgc>). The main objective of the survey reported here was to ascertain whether and how recontacting occurs in the UK. The topic areas were identified from debates in the relevant literature. The questions were developed from relevant literature, the clinical experience of members of the research team, and a pilot survey on recontacting conducted by some members of the team in 2011 as part of the 'Development of a draft NHS Information Standard for Genetics' by the National Genetics Reference Laboratory in Manchester, England<sup>1</sup>. The questions were further refined by pilot testing the survey with clinical genetics service providers.

The web-based survey was designed using Bristol Online Survey (BOS) and it was administered between October 2014 and July 2015. A link to the survey was circulated via an email invitation to the clinical leads of the UK's 23 clinical genetics services, with a follow-up email to senior clinical genetics staff. The 'landing page' of the survey contained information about the study and the research team, and invited respondents to discuss the survey with colleagues in their service before completing it.

The survey included closed and open questions with expandable text boxes to elicit explanatory comments, and examples. This combination of closed and open questions allowed the research team to collect both quantitative and qualitative data. Free text

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<sup>1</sup> <http://www.ngrl.org.uk/Manchester/newsitem/project-develop-draft-nhs-information-standard-genetics-launched>

responses from the survey were analysed using thematic analysis [18]. The survey is appended as an online resource.

### **Research Ethics approval**

The study was approved by the University of Exeter's Social Sciences and International Studies Ethics Committee.

## **RESULTS**

### **Respondent characteristics**

Twenty of the twenty-three clinical genetics services in the UK<sup>2</sup> completed the survey and were from all four nations comprising the UK. Of the twenty respondents who completed the survey (one per service), nine were consultant geneticists, seven were consultant genetic counsellors and the remaining four were genetic counsellors.

### **Recontacting experiences and practices**

The vast majority of the services (19/20) reported having recontacted patients and relevant family members in light of significant new information. However, 16/20 services indicated that recontacting occurred on an occasional basis. Only three services reported that they routinely recontact former patients.

A variety of reasons for having recontacted patients were reported (see Table 1). The most common were: the availability of new tests or new results; new clinical guidance; and reclassification of a VUS. The table below (table 1) reports the main themes from respondents' answers with some quotations to illustrate each theme.

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<sup>2</sup> For a complete list of clinical genetics services in the UK visit: <http://www.bsgm.org.uk/information-education/genetics-centres/>. Please note that this list includes also laboratories.

[Table1]

Only seven services responded that they have developed recontacting procedures; three of these were the centres who responded that they recontact former patients routinely.

The procedures reported varied across these centres with no significant pattern. Common across these responses was the mention of a lack of a codified procedure for recontacting.

Two centres reported that they inform the patients' general practitioner (GP) or paediatrician as appropriate; while two others reported that they inform the patient directly (via letter or telephone call). The three remaining centres mentioned that they use clinical databases which are held by NHS services (when undergoing testing, patients are consented about being on the database and about whether they wish to be recontacted). However, one of these three centres added that they were experiencing problems with their clinical databases – as a result of having moved to electronic notes – and that they were using external databases as well.

### **Recontacting 'Triggers'**

We asked about the type of information that would be sufficiently relevant to trigger recontacting (giving the example of relevance being based on clinical actionability of the information, or on its analytic validity). The majority of the services (14/20) indicated that one of the most important elements would be clinical actionability- i.e., information that has an impact on the clinical management of the patient and their family. One service added that in practice they would recontact only if new information relates to a small group or an individual patient, because of the workload of recontacting large numbers of patients. Other common answers included the publication of new or revised guidelines or new laboratory reports.

### **Use of clinical databases**

The majority of the responding centres (18/20) use existing clinical databases, rather than bespoke recontacting systems for recontacting purposes. Of these, nine answered that they use the databases mainly to identify patients; the other nine used them to review notes and/or to flag patients for recontacting. The clinical databases respondents referred to are databases held by the UK NHS. They are mainly used for clinical purposes, but sometimes they can be used to identify patients for research. Their current format is mixed (some centres use electronic versions, others paper-based versions).

### **Recording of patient preferences**

The majority of services (12/20) indicated they do not routinely ask patients about their recontacting preferences as part of the procedure for obtaining informed consent for genetic testing. Of these, six services responded that they record patients' recontacting preferences systematically, six that they do so only occasionally, while the remaining eight responded that they do not record patients' preferences at all.

Consent forms for genetic testing vary across the services in the UK. Six services said they used consent forms that give the option to patients to express their recontacting preferences. However, as one of these respondents added, consent forms are not always used in clinical practice.

[Table2]

Seven services stated that the main reasons for not recording patients' preferences – or for not mentioning the possibility of recontact to patients at all – were lack of resources available to offer a recontacting service and concern about raising unrealistic expectations

in patients. Four services mentioned their 'open door policy' – whereby clinicians encourage the patient to get back in touch with the genetics department from time to time to check if there are advances or to update clinicians about important changes in the family.

When asked hypothetically if there were reasons to recontact patients even when patients had indicated they would not want to be recontacted, a majority of services (14/20) responded that they would. In line with previous answers about the type of information that would trigger recontacting, the main reason provided was new information that may have clinical implications for patients and family members. The GP was explicitly mentioned by 4 respondents as a (first) point of contact to involve in this process. One respondent argued that patients' decisions not to be recontacted could not necessarily anticipate the specific information subsequently available, and might therefore be an insufficiently informed refusal.

Of the six services stating that they would respect patients' preference not to be recontacted, one added that if new information was likely to have significant implications for a family member, they would inform the patient's GP; while two mentioned the possibility of seeking external advice (e.g., from the Medical Defence Union, or the Genethics Club<sup>3</sup>). One respondent also made the point that although patients should not be recontacted if they clearly expressed this preference, in practice very few patients would be definite about their preferences.

### **Implementation of recontacting systems**

A slim majority of services (11/20) were unsure about whether routine recontacting systems

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<sup>3</sup> The UK Genethics Club is a national forum for the discussion, by health professionals, of practical ethical problems encountered in the working lives of clinical genetics departments in the United Kingdom. <http://www.ndph.ox.ac.uk/research/ethox-centre/ethics-support/genethics-club>

should be implemented. Of the remaining, five services indicated that they should be implemented, while four responded that they should not be.

The main arguments in favour of implementing systems for recontacting revolved around the ideas that this would improve the quality of care received by patients; it would increase patients' autonomy; and it may reduce the potential for litigation. We report below some illustrative quotations:

*“New information may help reduce the risk of disease/mortality to other family members- e.g. where a mutation is identified - screening and treatment may be offered to those at risk”.*

*“Technology and knowledge are increasing rapidly and what was known or possible even a short time ago may well be different and allow individuals more choice or better risk assessment or treatment”*

One respondent also made the point that clinical genetics would be the only medical speciality in a position to offer recontacting. Another respondent highlighted that having recontacting mechanisms in place would facilitate the process.

Among arguments against the implementation of recontacting systems, lack of resources was again mentioned and linked to equitable provision of services within the NHS. Another common argument was that patients should be encouraged to be more responsible for their health and share the responsibility with clinicians to keep up to date about relevant medical information.

Legal implications were also mentioned as an argument against the implementation of recontacting systems. Specifically, the concern was that introducing such systems would create a standard practice and that failure to recontact could then be seen as negligent.

Concern was also expressed by one service that recontacting might cause anxiety for patients and family members; while another service mentioned that recontacting would be more difficult to implement as genetic testing is increasingly ordered by mainstream medical specialities.

Finally, one service said that the time frame for recontacting responsibilities would need clarification; for example, for how many years from the genetic test would such a responsibility apply?

## **DISCUSSION**

This was the first study in the UK to explore current recontacting practices in clinical genetic services.

### **Recontacting occurs, but on an *ad hoc* basis**

The vast majority of UK services reported that they do recontact patients and family members, confirming the significance of recontacting for good quality of care in clinical practice. The majority of respondents indicated that 'clinical actionability' of the new information is the main reason to recontact. Respondents' answers also suggest that recontacting is becoming more important as clinical whole genome approaches deliver many more genetic variants for interpretation.

Recontacting tends to occur in an *ad hoc* fashion when an event 'triggers' clinicians to review a file, rather than systematically as part of routine clinical practice.

### **Procedures vary greatly across the UK**

Our findings suggest that there are no standardised recontacting practices operating in the UK. This diversity may be the result of historical and resources allocation differences. Overall the fact that recontacting does occur, but not in a standardised fashion, reflects the tension identified in prior research between the desirability of recontacting and the current lack of mechanisms and resources to offer it more systematically, or at all, resulting in unequal recontacting service provision across the country [8, 19]. Only a few centres that recontact patients have developed systematic recontacting procedures, but these procedures vary.

We also found considerable diversity of practice in the use of clinical genetic databases for recontacting purposes (e.g., to retrieve patients' contact details, to review notes, to flag patients for recontacting), and in how and whether patients' recontacting preferences are recorded.

### **Patients' preferences and professionals' responsibility**

Our findings suggest a tension between respecting patients' preferences to not be recontacted (i.e., their right not to know) and the responsibility or duty of care of healthcare professionals towards patients and family members. Although very few clinical genetics services reported that they record patients' recontacting preferences systematically, in response to an hypothetical question many services responded that there were circumstances in which they would recontact patients and family members even if the patient had indicated they would not want them to do so. These circumstances were related to the emergence of new clinically actionable information that could have an impact on the patient or family members' health management. Respondents' views appear to be in line

with the European Society of Human Genetics (ESHG) recommendations on the use of whole-genome sequencing in healthcare [20], in particular with the recommendation that:

*'Patients' claims to a right not to know do not automatically over-ride professional responsibilities when the patient's own health or that of his or her close relatives are at stake' (pg.583).*

### **Implementation of recontacting**

Finally, although the majority of respondents indicated that recontacting does occur in their genetic centres, albeit in a non-systematic way, more than half were unsure about whether recontacting systems should be implemented. This finding suggests that a thorough evaluation of the desirability, efficacy, equitability, and sustainability of potential recontacting systems in the national health service would be necessary before deciding whether and how to implement such a service, or before suggesting guidelines. Agnosticism toward the implementation of recontacting systems was accompanied for some with the suggestion of greater patient involvement in the process of recontacting. This idea was supported as promoting patient autonomy while circumventing resource and infrastructural barriers that may prevent healthcare professionals from offering efficient recontacting services. Some framed this idea as being less paternalistic than a clinician-driven implementation model in which the responsibility for keeping patients updated is placed solely on healthcare professionals. However, it is important to point out that a clinician-driven implementation model would not be paternalistic if patients choose it. Further, shifting responsibility for recontacting from clinicians to patients may be seen as promoting

patient choice but may not necessarily eliminate a potential duty<sup>4</sup> clinicians could have to recontact patients.

Our data provides some insight into genetic service providers' worries about legal consequences of implementing recontacting systems. Specifically, some respondents expressed the concern that introducing recontacting systems would create a practice standard and that failure to recontact could then be seen as negligent.

It is important to point out that – although in the UK there currently is no statute law and professional guidance regarding recontacting, and although there have been no known litigation cases – the fact that some centres do recontact might be seen to create a duty to do so. If recontacting becomes standard practice, then patients who are not recontacted, and are thus not able to avail themselves of interventions that might benefit them, could claim that a reasonable healthcare professional should provide this service and that the reasonable patient could expect it. Moreover, although there are no statute law or cases about recontact in genetics, there have been cases in North America relevant to recontact in other areas of medicine [16, 17]. It is difficult to assess whether the concern towards potential medicolegal consequences influence healthcare professionals' practices. We are exploring these issues in interviews we are currently conducting with healthcare professionals potentially involved in recontacting (clinical geneticists and other mainstream specialities) in the UK.

## **LIMITATIONS**

There are some limitations to this study. Although we asked respondents to discuss the

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<sup>4</sup> We follow the definition of 'duty to recontact' given in the first systematic review of this issue [7]. Such duty is defined as the ethical and/or legal obligation to recontact former patients in light of new genetic information.

questions with their centre's clinical team before completing the survey, we cannot know whether they did. As genomics enters mainstream practice, it will be important to seek wider representations of professional views and experiences. To address these limitations, we are conducting further research to investigate the views of healthcare professionals potentially involved in recontacting (including specialties other than clinical genetics), patients, and other stakeholders such as patient support groups.

This survey was administered in the UK and reports the views and concerns of genetic service providers working in this country. The medicolegal aspects of recontacting and healthcare professionals' views on the issue are likely to be subtly different in other countries, especially where legal systems are different (e.g. roman law as opposed to common law based systems). It will be important to conduct a wider analysis to support recommendations or practice guidelines, if any, in this increasingly complex area of clinical practice.

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#### **DISCLOSURE**

The authors declare no conflict of interest.

Supplementary information is available at the *Genetics in Medicine* website

## REFERENCES

1. Hunter, A.G.W., et al., *Ethical, Legal, and Practical Concerns about Recontacting Patients to Inform them of New Information: the Case in Medical Genetics* in *Genetic Testing. Care, Consent and Liability*, N.F. Sharpe and R.F. Carter, Editors. 2006, John Wiley & Sons, Inc.
2. Burton, H., T. Cole, and P. Fardon, *Genomics in Medicine. Delivering Genomics Through Clinical Practice*, in *Report of the Joint Committee on Medical Genetics*. 2012.
3. Pyeritz, R.E., *The Coming Explosion in Genetic Testing - Is There a Duty to Recontact?* *The New England Journal of Medicine*, 2011. **365**(15): p. 1367-1369.
4. Rantanen, E., et al., *Regulations and Practices of Genetic Counselling in 38 European Countries: the Perspective of National Representative* *European Journal of Human Genetics*, 2008. **16**: p. 1208-1216.
5. Hirschhorn, K., et al., *Duty to Re-contact. Policy Statement: Social Ethical and Legal Issues* *Committee of the American College of Medical Geneticists*. *Genetics in Medicine*, 1999. **1**: p. 171-172.
6. Richards, C.S., et al., *ACMG recommendations for standards for interpretation and reporting of sequence variations: Revisions 2007*. *Genetics in Medicine*, 2008. **10**(4): p. 294-300.
7. Otten, E., et al., *Is there a duty to recontact in light of new genetic technologies? A systematic review of the literature*. *Genetics in Medicine*, 2014.
8. Fitzpatrick, J.L., et al., *The Duty to Recontact: Attitudes of Service Providers*. *American Journal of Human Genetics*, 1999. **64**: p. 852-860.
9. Dean, J.C.S., D.R. Fitzpatrick, and P.A. Farndon, *Genetic Register in Clinical Practice: a Survey of UK Clinical Geneticists*. *Journal of Medical Genetics*, 2000. **37**: p. 636-640.
10. Kausmeyer, D.T., et al., *A survey of patients' experiences with the cancer genetic counseling process: recommendations for cancer genetics programs*. *Journal of genetic counseling*, 2006. **15**(6): p. 409-431.
11. Bernard, L.E., et al., *Duty to Re-Contact: a Study of Families at Risk for Fragile X*. *Journal of Genetic Counseling*, 1999. **8**(1): p. 3-15.
12. Sexton, A.C., et al., *Impact of a Genetic Diagnosis of a Mitochondrial Disorder 5-17 Years After the Death of an Affected Child*. *Journal of Genetic Counseling*, 2008. **17**: p. 261-273.
13. Knoppers, B.M., *Duty to Recontact: a Legal Harbinger?* *American Journal of Medical Genetics*, 2001. **103**: p. 277.
14. Sharpe, N.F., *The Duty to Recontact: Benefit and Harm*. *American Journal of Human Genetics*, 1999. **65**: p. 1201-1204.
15. Murray, M.L., et al., *Follow-up of carriers of BRCA1 and BRCA2 variants of unknown significance: variant reclassification and surgical decisions*. *Genetics in Medicine*, 2011. **13**(12): p. 998-1005.
16. Pelias, M.Z., *Duty to Disclose in Medical Genetics: a Legal Perspective*. *American Journal of Medical Genetics*, 1991. **39**: p. 347-354.
17. Hunter, A.G.W., et al., *Ethical, Legal, and Practical Concerns About Recontacting Patients to Inform Them of New Information*. *American Journal of Medical Genetics*, 2001. **103**: p. 265-276.
18. Braun, V. and V. Clarke, *Using Thematic Analysis in Psychology*. *Qualitative Research in Psychology*, 2006. **3**: p. 77-101.
19. Burton, H., C. Alberg, and A. Stewart, *Mainstreaming Genetics: A Comparative Review of Clinical Services for Inherited Cardiovascular Conditions in the UK*. *Public Health Genomics*, 2010. **13**: p. 235-245.
20. van El, C.G., et al., *Whole-genome sequencing in health care*. *European Journal of Human Genetics*, 2013. **21**: p. S1-S5.

## **Table legend**

Table 1: most common reasons for recontacting patients and relevant family members  
(grouped by themes)

Table 2: number of centres who ask and record patient recontacting preferences

**Table 1.** Most common reasons for recontacting patients and relevant family members

(grouped by themes)

<b>Theme</b>	<b>Quotations</b>
<b>Availability of new genetic tests or new results</b>	<i>"If a new (or improved) testing technique becomes available, for example review of patients who had limited BRCA testing, when full analysis became available some were recontacted and asked if they wanted a full screen"</i>
<b>Family follow-up</b>	<i>"A new family member has been referred after a long period of time, prompting a review of the file and recontacting the family"</i>  <i>"If a result is issued on a deceased individual"</i>
<b>Reclassification of VOUS</b>	<i>"A specific result is re-classified as more (or less) pathogenic and a clinician may decide to recontact a patient(s) found to have this mutation to update them on this new information. This is more ad hoc than systematic at the present time"</i>
<b>New clinical guidance</b>	<i>"New recommendations for gene carriers"</i>
<b>Reproductive relevance</b>	<i>"Follow up for teenager for condition of reproductive relevance"</i>

**Table 2.** Number of centres who ask and record patient recontacting preferences

<b>Asking patient recontacting preferences</b>	<b>Number</b>
Centres who routinely ask patients whether they would like to be recontacted as part of the procedure for obtaining informed consent	8
Centres who do not routinely ask patients whether they would like to be recontacted as part of the procedure for obtaining informed consent	12
<b>Recording patients recontacting preferences</b>	<b>Number</b>
Centres who record patient recontacting preferences – systematically	6
Centres who record patient recontacting preferences – occasionally	6
Centres who do not record patients recontacting preferences	8