

Mainstreaming genomics: Recontacting patients in a dynamic healthcare environment



Team

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Project webpage: <http://ex.ac.uk/mgc>

Current Definition

‘Duty to recontact’: Ethical responsibility and/or legal obligation to recontact former patients about new genetic information (Otten *et al* 2014).

1. Ethical responsibility/Legal obligation
2. Excludes ongoing treatment relationships
3. Excludes recontacting in research setting

Example

A new gene is discovered, which when mutated, is the explanation for a group of previously undiagnosed patients. Clinical genetics services in the UK have patients they have seen over the years who might have this diagnosis.

Recontacting increasingly pressing

Rapid accumulation of new genetic and genomic knowledge resulting in better diagnosis and treatment of some health conditions

- Intensified by use of whole exome or genome approaches in healthcare
- Greater integration of genomic knowledge into 'mainstream' medical practice

Issues

1. Patient

- Promote better healthcare and patient autonomy (new information = new possibilities)
- Consent to be recontacted can never be autonomous as patients do not know what type of information will be disclosed
- Negative impact (e.g., breach of confidentiality, anxiety), putative right not to know
- Familial implications

2. Relevance of new information

- Greater justification for recontacting former patients with clinically relevant information about a life-threatening disease than a small increased risk for a slowly progressive disease
- Personal utility

3. Responsibilities

Unclear

HCPs – what specialty/es?; clinical scientists; patients; patients
and HCPs; support groups; media...

4. Medicolegal aspects

Recontacting could prevent HCP from being held liable for negligence by former patients

Embracing recontact policy may make HCPs liable if they cannot meet expectations

5. Ethical Desirability vs. Practical feasibility

Recontacting barriers:

- Lack of infrastructure for tracking data of former patients (e.g. informatisation of databases)
- Lack of time and resources (staff, money) to perform recontacting
- Lack of up-to date patient addresses

Project Objectives

1. Explore and analyse ethical, legal and social issues relating to recontacting
2. Survey current clinical practices regarding recontacting, in light of new genetic information, within and between different medical specialties in the NHS
3. Investigate patients' expectations of genomic information management and expectations regarding responsibilities and mechanisms for recontacting
4. Investigate healthcare professional perspectives and expectations concerning recontacting in different medical specialties
5. Provide an empirical basis to support arriving at a professional consensus, which is currently lacking
6. **Engage with stakeholders to integrate the above findings and analyses to work toward a professional framework**

Methods

- Survey of recontacting practices 20/24 clinical genetics centres
- Currently administering the same survey in EU
- Interviews 30 HCPs and clinical scientists using vignettes
- Questionnaire 130 patients
- Interviews 41 patients

Plan

1. Project introduction
2. HCPs' perspective
3. Parents/Patient' perspective

Q&A

LUNCH BREAK

4. Concluding remarks and plenary discussion

2. HCPS' perspectives

Exploring genetic HCP's duty to recontact

- Research question:
 - When and how should clinical geneticists recontact a patient when new clinically relevant information becomes available?
- Hypotheses:
 - That the cultures and norms used to practice recontacting are socially structured
 - That cultures and norms will vary across professional subcultures

Healthcare Professional Interviews

- Total of 31 interviews with healthcare professionals
 - 7 Exeter
 - 12 Cardiff
 - 12 Southampton
- Professions:
 - 4 Genetic Counsellors
 - 11 Clinical Geneticists
 - 3 Genetic Registrars
 - 5 Laboratory Scientists
 - 9 Other HCPs (e.g., neurologists, oncologists, paediatricians, haematologists, cardiologists, endocrinologists, general practitioner)

No shared view on a duty to recontact

- **Formal duty**
 - Well, I think you are in a form of contract with patients. So patients come to see you and establish a contract and which you are going to perform certain parts of that contract and that contract is to investigate them and to notify them of that investigation. [...] and I think that that contract does not terminate when you have finished the consultation and say “we haven’t found anything”. [...] (P15)
- **Critique of formal duty**
 - [...] I think you’d feel more responsible if you saw, “oh I saw someone last year and that seems quite recently, I should do that”. And then you think you know “I only saw you last year and you didn’t tell me about this”. Whereas if you’d seen them ten years before they probably wouldn’t expect you to remember them. And probably wouldn’t (P19)
- **Transfer responsibility to the patient**
 - Yeah I mean I just say ... so I’d say you know “we found this genetic variant erm it might be linked to your condition, it might not, we don’t know”. [...] it’s just I have to say “we can’t do anything else now erm we might have more information in the future. Get back in touch with us in a few years’ time”. And it’s very much yeah leaving the ball in their court and I don’t ... I don’t know if they will come back or not really. (P9)

No shared view on recontacting:

Personal, professional and organisational memory

- ... So it's a very random process. So I might only remember three or four cases. It's not consistent, that's another point. It's not consistent. So okay, fine, I have got a new gene panel for say, as I said, cardiomyopathy, [unclear-0:12:12] cardiomyopathy. I'm interested to write to ... but I may only remember a few. And er ... (P3)
- [...] There was a way within the department that you could recall cases or you'd just put the files down to be reviewed in a year or two years or eighteen months or something, and then they'd just be brought up for you, [...] All that sort of thing's lost. Secretaries don't really get files for you any more anyway. (P19)
- No. We have a database that we use [...] but we do not put results in this database, we put the letters. I think of course in the letter you will have the result of the patients, but I don't think there is an easier way to retrieve the data. [...] (P16)

Recontacting and clinical practice

- RES: But it still won't do what I want, which is, "Please give me a list of patients with left big toe syndrome". And don't forget, when you get a brand new, say, mutation as it says there, it may be a very muddy sort of phenotype. You know, "like boys with mental retardation ... who've got small ears and big testicles. ... Have this gene". Now, how am I going to pull back boys with mental retardation, small heads and big testicles? There is never going to be a database with that on it, is there?

INT: No. No.

RES: You know, all I can do is think about all the patients I've seen and try and think, now, did anybody fit that pattern? (P7)

Recontacting and laboratory practice

- you know I try to be very responsive to a clinician asking the question because they know their patients, they know those disorders so erm they do tend, that's where I see my role and the lab's role is to be responsive to that. [...] Erm but then within the laboratory you also have erm scientists who're specialists in certain different areas so scientific areas erm and I think they also have a role to be able to bring to the attention of the service, to me and of the clinical team to be able to say there is this new development, there is a new gene that's known as well. So I think we've got a responsibility, the responsibility is two way. (P11)

Recontacting and civil society support

- What I mean is, patients who are at risk of Lynch syndrome are referred to our service for genetic testing and we let them know about aspirin and the research studies that are ongoing as part of that process. We don't go out there and solicit patients who are at risk of Lynch syndrome and we don't go back to our records and pick out all the families with Lynch syndrome and let them know. And again, it's because we rely on... there are other ways for these families to find out - the Lynch Syndrome Association and other media sources. (RHCP6)
 - [...] I wouldn't be leaving it to a, you know as a support group, for example if it's a, you know ectodermal society or you know any other, some big society. But erm, I know that we can use them to contact patients if needed. You know if they are taking part in research and other things. But it is a clinical situation where there's a medication of a drug or anything like that, I have seen this patient before, then I think it should ...
- INT: It's your responsibility and you do that.
- RES: Yes, I should think yes. (P6)

Recontacting a patient: establishing grounds to recontact

- Relevant new information might be made available:
 - Erm and er then in April we had a significant uplift in funding for BRCA testing, so that we can implement NICE guidance that had come out in June 2013, erm which we hadn't been complying with previously. And so we had this uplift in funding in April, which meant that we could now test more people including these young women with triple negative breast cancer ... diagnosed under forty. [...] (P4)
- But does new information warrant a recontact?
 - If it's a VUS, then it would have had a lot of discussion, so I would have taken our meeting we have, our VUS meeting we have, which is a joint meeting with clinical and laboratory staff [...] then there would be well actually you know, does this patient need to be contacted, is this new information that would be really helpful to them. And if you have changed the classification, I think it is, my view would be that actually it's important that that person knows that, you know especially if you've classified it as a higher classification [...] (P24)

Recontacting a patient: anticipating the patient's response

- It depends. I'll go through the notes and my letters first, to see how I've contacted them in the past, because usually there's some record of how they'd like to be contacted. If I've already done testing there'll be some record of how they wanted to be contacted with that result. So that will give me a clue. [...] And usually it would be by letter or phone. (P24)
- Yes. So if I know them myself, and it had been fairly recently like within the last five years, easily be in my memory, I would probably write to them myself. But if I have had no contact or it's been many years or they were family that were seen by my registrar many years ago and I never saw them, I would try and go through their physician, be it their hospital practitioner or their GP [...] (P18)

Recontacting a patient: respecting the right not to know

- Yes, yeah. And sometimes people, because I think sometimes what people decide at one time changes, but the difficulty is you don't know that, and it may be just that they were really struggling that that time. They might have just been recently diagnosed with a cancer, and really it's all too much for them, whereas it could be five years on, they're in a very different place, and that's really difficult to, you can't, I can't just that because I don't know. It would be helpful if the notes are a bit more extensive and say why they don't want to be re-contacted. [...] (P24)

Recontacting

- Giving patients time to think
 - ... what I find is I don't tend to contact people by phone because I think when you could call someone they've got to think on their feet. ... And then whereas when you contact them by letter, they've made an active decision to phone you and they will have given some sort of thought to the letter. And therefore they may have prepared themselves, even only if it's a little bit psychologically, to find out. ... (P8)
- Contacting the patient's GP
 - They could put it, bin it and it may be that you'll have discussed with the consultant or your colleague, and you say "actually I'll drop them one more letter and say 'if we don't hear from you we'll understand we don't want to get in touch'" and perhaps copy that to the GP. Because at least then there's someone else there that knows. [...] (P8)
- Securing compliance
 - I've known some patients who have been pretty poor compliers, and therefore I have been quite blunt about the seriousness of something or other, in order to try to encourage them therefore to respond either to phone or to come to clinic by saying "look this is quite an important issue, not just for you but for other family members and therefore you do need to address it". (P5)

System barriers:

Discharging patients

- Because if I discharge somebody then it makes it much more difficult, I feel it makes it much more difficult for me if they have been discharged, because if I discharge them and then I contact them at a later date, they're actually not my patient anymore because they've been discharged. And so if I then, if that contact then led to them having an appointment, I would have to get a re-referral from their GP, which is very frustrating for me. So I'd prefer not, if I had my choice, I wouldn't discharge anybody. (P24)

Overcoming system barriers

- And we also don't have a standard, it's variable about when and if patients are discharged as well. And so if it's a family where I think that there may be, we may be coming back to in the future, I wouldn't discharge the patient, but there's not a uniform practice across the department. [...] (P24)
- For me a discharge is the current episode is completed. It doesn't mean that you won't have another episode or that there won't be a reason to see you later and so on, so I don't consider them dismissed if you like, just discharged, just temporarily discharged. (P25)

Discussion

- Recontacting a good thing
- No clear sense of duty to recontact
- Recontacting dependent on memory systems
- Recontacting different in different circumstances
- More consensus on modalities of recontact
- Recontacting and patient discharge

3. Patient/parents' perspectives

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Background

- New information can have significant implications for patients' and families' health, reproductive decisions, lifestyle choices, employment, and psychosocial wellbeing
- Existing empirical evidence indicates that not all patients value recontacting
- Anxiety; stigma; privacy; interest/right not to know

Sample

Interviews: 41 Questionnaires: 130

Patients and parents of patients:

- without a diagnosis
- recently offered a test for a condition or carrier risk
- with a rare condition
- with a Variant of Unknown Significance (VUS)
- who had been recontacted.

Recruited through NHS and online condition-specific social media/charities

Themes: ethical & psychosocial issues

Recontacting is
valued

Different types of
info should trigger
recontacting

Information has
complex
psychological
impact

Lines of
responsibility are
unclear

Is recontacting
justified in
resource-
constrained NHS?

Recontacting is valued

Improve knowledge, management or prevention, of a condition (or signs and symptoms) in a timely fashion

P2 Forewarned is forearmed isn't it, because then, if there's something preventative that you can do to, [it] doesn't rule you out from getting it but [...] your chances are better

P40 If the information is there I think it should be given to somebody. I don't think there's any point in holding back

Recontacting triggers

What counts as a trigger differed between different participants.

- **Some wanted tailored ('bespoke') information**

P23 [I would expect to be recontacted] if there's something significant that would be relevant to me and my particular condition. [...], but if it was just a case of, "We've found a new gene that causes cardiomyopathy, we are just letting you know", then no, I wouldn't expect that

Recontacting triggers

- **Some wanted even very general information or updates to say there's no new information**
- **Helps to manage uncertainty & 'slow pace' of clinical practice & research (recontacting vs. follow -up)**

P22 Even if it's: 'We haven't found anything'[...] I would just like to know, because it's just like a waiting, isn't it? You just think 'ooh I wonder if they have found anything or I wonder if anything has happened or how it's all going'

P31 I'd like to be kept informed like annually maybe [...] there is the element of support within information.

P12 We've learnt with genetics, "soon" can mean anything between 1-2 years

Information has complex psych impact

Double-edged sword

*P20 I wasn't expecting a hospital letter. And it did throw me. [...] I can't explain why because to me it was **brilliant**, I was really glad that things had moved on, but it was still **quite a shock**. I thought, maybe I'd get an answer now [...] I had a long chat with [consultant who recontacted respondent] on the phone then before I went to see him. And I was honest with him, I said, "You've completely floored me." It was just completely out of the blue, really, but in a good way*

Warrants immediate support

P12 The worse you can have, is "Oh I've got information, but I can't see you until two months"

Lines of responsibility are unclear

Responsibility has no time limit

P29 You can't put a time limit on it, because we're always going to be finding out something new about it, so no there shouldn't, well why would there be a time limit, it seems a silly thing to put a stop on it. It's like saying, 'we're going to stop the research now, it's, what we know is all we want to know, we don't want to know anymore'

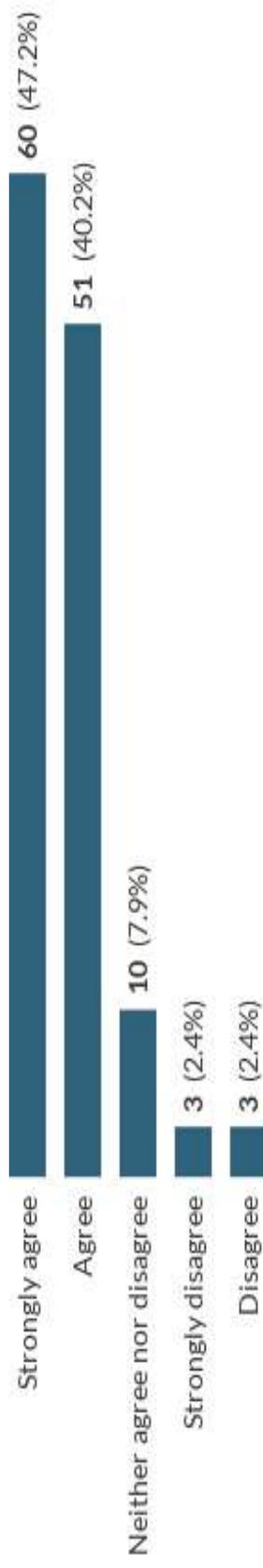
Patients & multiple HCPs have responsibilities

*P30 I think it's everyone's responsibility. But I think there needs to be some kind of mechanism there to **bring everyone back together** periodically, to go over that. [...] Professionals have access to tools that we don't, that give them information that we would never have access to. That's why it needs to be both*

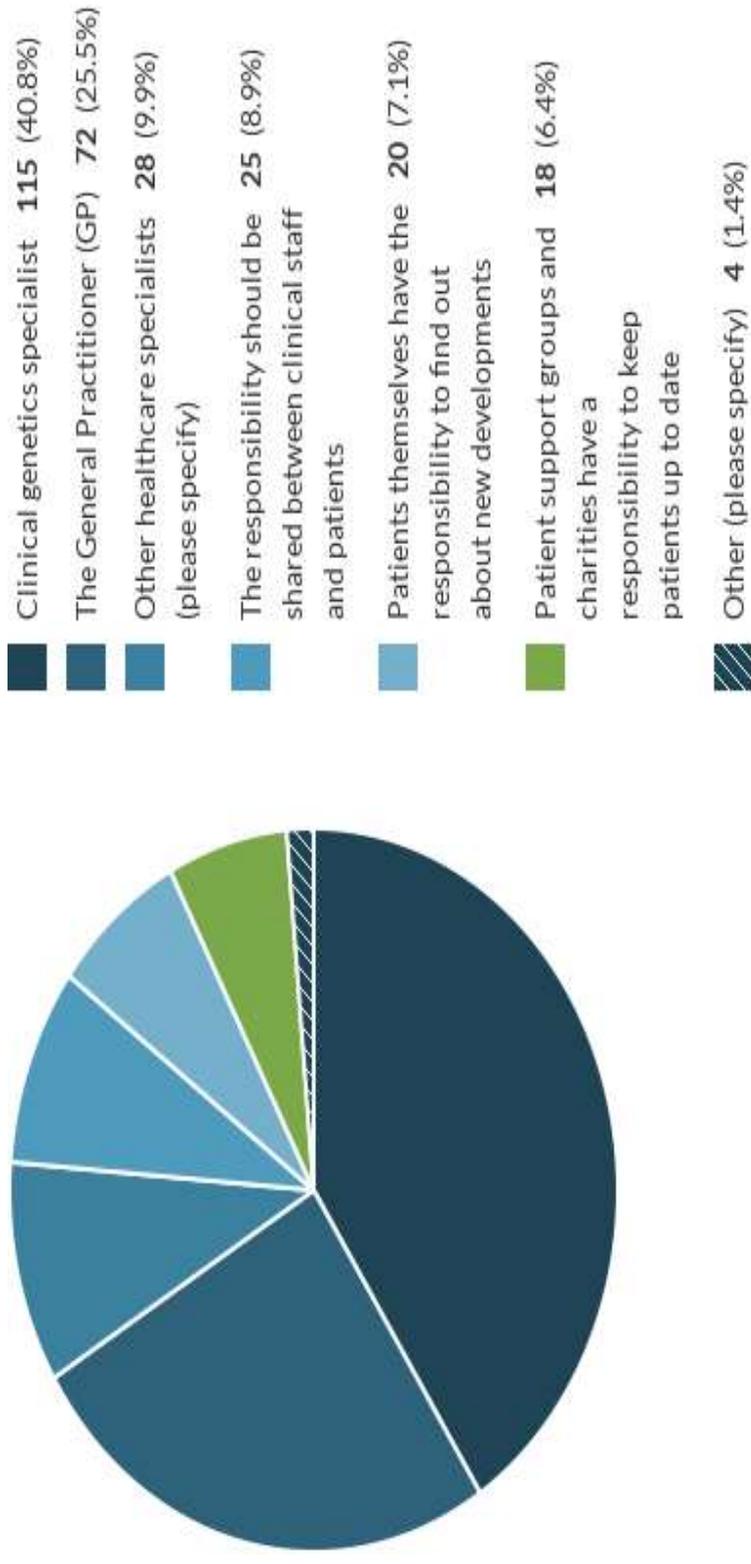
Lines of responsibility are unclear

P5 this is where I think where the sort of two-way responsibility comes. It is your responsibility to look after yourself but you sometimes need some help and support to do that and you know I suppose the NHS in whatever format it may be, should be the place that you could go to do that.

I expect clinical staff to keep me updated with relevant information about my health issue or condition



Who do you think should be responsible for updating former patients with new information?



It is my own responsibility to follow developments in genetic medicine relevant to my condition



Is recontacting justified in resource-constrained NHS?

More pressing priorities

P34 With obesity, diabetes et cetera, do I think it's realistic that there's going to be additional funding for this? I don't think that's going to happen. Other things are more impacting and could do with more energy and research. In an ideal world, yes of course. In reality, I think it's unlikely

e.g., prevention

P5 - Healthcare is not just there at the point of crisis, it's the point of where you are in dire need and you need a lifesaving operation or it should look after you or it should be there, be available to you throughout being ill, preventing you being ill, recovering, and helping you live a healthy life beyond any illness

Is recontacting justified in resource- constrained NHS?

Compromises: realistic view of what is possible

P5- I probably don't expect it because I think I've experienced how inundated healthcare professionals are in the day to day treatment of patients. [..]But I do think it would be helpful and beneficial if it could happen. But then it would probably have to happen on a general basis rather than on a very personal basis

Discussion

Recontacting is
valued

Different types of
info triggers
recontact

Information has
complex
psychological
impact

Lines of
responsibility are
unclear

Is recontacting
justified in
resource-
constrained NHS?

Discussion

Recontacting should be planned and tailored

- Potential psychological complexity of receiving new information
- RNTK

Need to clarify difference between recontacting and follow-up

Recontacting is
valued

Different types of
info triggers
recontact



Discussion

Joint venture between patients and multiple HCPs useful because

- Perception that it's patients' responsibility to manage their own health
- it's a sustainable way to enact recontacting in resource-constrained NHS
- but needs to be enabled by some form of coordination (see EJHG letter) - recontacting open process in which both patients and HCPs have the possibility to make contact

Lines of responsibility are unclear

Is recontacting justified in resource-constrained NHS?

4. Concluding remarks

Many questions remain

- What impact might change of practice, or development of guidelines based on professional consensus, have on a legal duty to recontact?
- Whose professional role(s) would it be to recontact?
- What does 'holding' information mean in terms of responsibility or duty?
- In what situations would recontacting be relevant (e.g., only when new information has tangible impact on patient management)?
- How often, and over what span of time, would genetic services be expected to conduct repeat laboratory analyses or reinterpretations on any samples or results that included VUSs? In other words, for how long might a duty or responsibility to recontact last?
- Which methodologies and infrastructures, if any, might be useful as recontacting systems?

Our recommendation

In line with a model of shared responsibility (as forwarded by some interviewees), recontacting & issues that might trigger it should be discussed and documented routinely in the consent process for testing, or whenever client data are collected.

The client should be offered the possibility of sharing responsibility for recontacting

The discussion

- The clinical team holds their records and provides the best information available at the time
- But the client is welcomed to contact the team
 - when a potentially relevant family event occurs, such as a death or birth, or a child reaching reproductive age
 - at regular intervals (if agreed by both parties depending on the specific condition).
- Future contact may also trigger clinicians to review the client's file to check whether any new information is relevant to them.
- If the client prefers not to share responsibility for recontacting, discussion could still help to clarify preferences & balance of responsibility between patients & clinicians

Benefits of discussion

- Reduce a potential clash of expectations between client and clinicians about responsibility regarding recontacting.
- Promote client autonomy – the client can decide whether to have more or less control over whether recontacting occurs.
- Respect confidentiality and a right ‘not to know’, as well as giving clients and their families some control over recontacting in sensitive family situations.
- Reduce some practical barriers – when clients agree to share responsibility for recontact with clinicians.
- Promote more standardised ways of triggering a file review and potential recontacting event.

Recontact as a symptom of a wider problem?

- Lack of reliable and compatible infrastructure to record, retrieve and share data within the health system
- Clients might not remember to recontact, and clinicians trigger recontacts on an ad-hoc basis
- Limitations to identifying triggers to recontact: need a “common framework of harmonized approaches to enable the responsible, voluntary, and secure sharing of genomic and clinical data” and a way of getting this information to clinicians
- And getting the information to clients – could they be alerted to information via an electronic health record?
- Need infrastructure to use and share genomic data responsibly

Next steps

- Meeting with ESHG PPC committee
- Engage a professional conversation about the broader issues
- Meeting with policymakers from Dept of Health etc in May