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Target: **EJHG**; type of paper: [Letter](#)

## **Recontacting in clinical genetics and genomic medicine? We need to talk about it.**

### **The problem**

An editorial in the AJMG (Levenson 2015) which accompanied the first systematic review conducted on the topic (Otten, Plantinga et al. 2014) suggests that advances in next generation sequencing technologies and the amount of genomic information they provide, are leading clinicians to consider recontacting former patients. This might be to see, for example, whether more sensitive testing could now provide a diagnosis or prediction which had not previously been possible, to review the evidence for particular genetic findings playing a part in the patient's disease, or to revise the interpretation of a genetic variant that had been given previously. As the Otten et al. systematic review shows, there is a perceived need for guidance in this area. However it is not clear whether a standardised protocol is necessary and whether a duty to recontact exists.

### **The project**

We conducted a survey of clinical genetics services (Carrieri et al. 2016) followed by interviews with healthcare professionals and clinical scientists in the UK (*EJHG paper on HCPs' interviews*) to obtain empirical evidence of current recontacting practices. This showed that recontacting does happen in the UK health system, but that there are no standardised procedures between regional clinical genetics services and no standard way of recording patient preferences about recontacting or of triggering a file review and potential recontacting event. Our findings converge with work on this area, which points to a tension between the ethical desirability of recontacting in some situations, but the difficulty with its feasibility in practice, such as lack of the necessary infrastructures, time and resources (Otten,

Plantinga et al. 2014). The issue of limited resources is very important, but is compounded with a struggle to define the responsibilities for recontact. For example we found unclear expectations between clinical scientists in the laboratory and clinicians in relation to who should keep up to date with reclassification of variants (EJHG paper). We are also aware that there can be a clash of expectations about responsibility regarding recontacting between patients and healthcare professionals. A study found that clinicians assigned more responsibility for maintaining contact with healthcare providers to patients than patients assigned to themselves (Fitzpatrick, Han et al. 1999).

### **Discussing recontacting with patients**

As a first step to encourage a debate, we would like to suggest that recontacting, including issues that might trigger it, should be discussed routinely between patients and clinicians in the context of consent for testing or whenever patient data is collected and recorded. This discussion would help to clarify expectations for clinicians and patients in recontacting, in the UK and elsewhere.

As part of this discussion the patient should be informed that the clinical genetics team holds their records and provides the best information available *at the time*, but that the patient is welcomed to contact the team: 1) when a potentially relevant family event occurs, such as a death or birth, or a child reaching reproductive age (Hirschhorn, Fleisher et al. 1999), and 2) at regular intervals (if agreed by both parties depending on the specific condition).

If the patient agrees, the future contact may also trigger clinicians to look at the patient's file to check whether any new information is relevant to them. This idea is in line with a model of *shared responsibility*<sup>1</sup> for recontact with patients, mentioned by some clinicians we

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<sup>1</sup> The idea of shared responsibility does not only stem from our empirical research. We are aware that other authors have recently argued for a shared responsibility in relation to disclosure of genetic information to

interviewed (EJHG paper). It also follows suggestions in previous studies (Pelias 1991, Almqvist, Adam et al. 1997; Godard, Kaariainen et al. 2003) that involving patients in the process of recontacting could be a solution to practical barriers, as well as ostensibly giving patients more autonomy and control.

If the patient does not agree – e.g., if they or the clinician feel they do not have the capacity/willingness/time to contact the team and/or to be kept up to date regarding their condition, the discussion would still help to clarify patient preferences, and the responsibility between patients and clinicians.

The patient would always be able to notify a change of preference to the clinician (this could be in itself a trigger for clinicians to review files).

This discussion and patient preferences should be recorded. Having and recording this discussion with patients could:

1. Reduce a potential clash of expectations between patient and clinicians about responsibility regarding recontacting
2. Promote patient autonomy – the patient can decide to have more control over whether recontacting occurs
3. Respect confidentiality and a potential right not to know, also giving patients and their families some control over recontacting in sensitive family situations
4. Reduce some practical barriers – in case patients agree to share responsibility for recontact with clinicians

5. Promote more standard ways of triggering a file review and potential recontacting event

### **Final considerations**

There are some issues related to our suggestion: e.g. What happens if the patient who agreed to recontact the service fails to do so? When is the most appropriate time to have the recontacting discussion with patients?

We believe that consultation with stakeholders, including patients, clinical scientist, and clinicians represents an important first step. We are engaging in this work in the 'Recontacting in Mainstreaming Genetics' research project in the UK (<http://ex.ac.uk/mgc>). Our intention with this letter is to initiate debate, and we welcome responses.

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