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A 'joint venture' model of recontacting in clinical genomics: challenges for responsible implementation

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ABSTRACT

Advances in genomics often lead healthcare professionals (HCPs) to learn new information, e.g., about reinterpreted variants that could have clinical significance for patients seen previously. A question arises of whether HCPs should recontact these former patients. We present some findings interrogating the views of patients (or parents of patients) with a rare or undiagnosed condition about how such recontacting might be organised ethically and practically. Forty-one interviews were analysed thematically. Participants suggested a 'joint venture' model in which efforts to recontact are shared with HCPs. Some proposed an ICT-approach involving an electronic health record that automatically alerts them to potentially relevant updates. The need for rigorous privacy controls and transparency about who could access their data was emphasised. Importantly, these findings highlight that the lack of clarity about recontacting is a symptom of a wider problem: the lack of necessary infrastructure to pool genomic data responsibly, to aggregate it with other health data, and to enable patients/parents to receive updates. We hope that our findings will instigate a debate about the way responsibilities for recontacting under any joint venture model could be allocated, as well as the limitations and normative implications of using ICT as a solution to this intractable problem. As a first step to delineating responsibilities in the clinical setting, we suggest HCPs should routinely discuss recontacting with patients/parents, including the new information that should trigger a HCP to initiate recontact, as part of the consent process for genetic testing.

1. Introduction

Advances in genomics can lead healthcare professionals (HCPs) to realise that they hold clinically relevant information about patients seen in clinic previously. This information might pertain to a reinterpreted variant; a new test; or knowledge about treatment and surveillance. Do HCPs have a duty or obligation to recontact the former patients (or parents thereof)? What ought to be the nature of this duty? What should trigger action to fulfil the duty? Fig. 1 illustrates an example of one potential trigger.

There is little empirical evidence and no formal guidance about recontacting and no legally defined duty to recontact: a HCP has a legal *duty of care* to a patient, but might consider this to lapse once they discharge the patient from their service. Nonetheless, legal scholars have expressed concerns that deciding not to recontact could amount to negligence (Pelias, 1991, 1992; Hunter et al., 2001; Griffin et al., 2007; O'Connor, 2014). At the same time, recontacting could be perceived as a violation of privacy (Letendre and Godard, 2004). Legal arguments aside, Otten et al. (2014). have questioned whether there is an ethical duty to recontact and have presented several arguments in favour: beneficence, in that information has

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Meera has undiagnosed developmental delay and several signs and symptoms that are not explained by any particular diagnosis. Her parents have spent a long time trying to get a diagnosis to understand her prognosis. She has had many inconclusive genetic tests. Five years later, after discharging her from his service, a paediatrician is working with a geneticist using a large biorepository, and identifies a handful of people across the world who have similar signs and symptoms to Meera. They have received a molecular diagnosis of a particular rare condition. Remembering Meera and her parents, he considers this to be a trigger to recontact the family. He can now offer her a test, which might facilitate a diagnosis and better understanding of her condition.

Fig. 1. An example of recontacting.

potential clinical benefit and could reduce uncertainty (Sexton and Metcalfe, 2008; O'Connor, 2014), and enhancement of autonomy through offering information upon which the patient can act (Hunter et al., 2001; Hastings et al., 2012; Clayton et al., 2013). A counter-argument, however, is that patients cannot make an autonomous decision about whether they would want to be recontacted because they do not know what information there is to know. As well as a perceived invasion of privacy, being recontacted could cause distress and lead to discrimination (Andrews, 1992; Sharpe, 1999; Brown et al., 2006; Peshkin and Burke, 2007; Shirts and Parker, 2008; Pyeritz, 2011). For example, Chen et al. found that when HCPs recontacted patients/parents to say that their information would be used in a new research study, it caused confusion and sometimes even anger, e.g., where parents initially misunderstood and thought a diagnosis had been found (Chen et al., 2017). These concerns might be outweighed by the benefit of being alerted to a potential risk amenable to intervention (Hunter et al., 2001; Sexton and Metcalfe, 2008; Andrews, 1992; Sexton et al., 2008).

Few empirical studies exist about recontacting: those reviewed by Otten et al. (2014). did not explore the topic specifically. Recently, we conducted the first study to survey current recontacting practices in UK clinical genetics services (Carrieri et al., 2016), which revealed that genetic HCPs recontact patients/parents in an *ad hoc* way and would recontact them with information of potential clinical significance, even if they had said they did not want to be recontacted. Our follow up paper exploring the views of a sample of UK genetic HCPs showed that they perceive several practical barriers to recontacting: there is insufficient money, staff, and infrastructure, and no good way to track patients and relevant information over time (Carrieri et al., 2017a). These findings support those from previous research (Ali-Khan et al., 2009; Murray et al., 2011; Mulla, 2015). Arguably, however, HCPs might struggle to define their responsibilities, and the limits thereof, even with limitless resources.

Uncertainty thus remains about what form recontacting should take: should it be entirely patient-led, whereby the patient/parent routinely checks for updates? Or should it be a HCP-led model, whereby HCPs routinely recontact former patients? If so, which HCPs should recontact? Managing genetic information might be new territory for non-specialist HCPs—would they know how (best) to act on new information? Some HCPs in our study (Carrieri et al., 2017a) argued that decision-making about whether recontacting happens, and efforts to make it happen, should be shared between HCPs and patients/parents. For example, HCPs could invite patients/parents to contact them at regular intervals, which would trigger a check for updates. To an extent, this option could circumvent limitations of resources and could give patients/parents some choice about learning new information and thus respect their preferences. Other HCPs argued that patients/parents would not always have the understanding, organisational skills, or time to recontact and that their decisions not to request updates would sometimes be uninformed, so placing responsibility on them would be practically and ethically problematic. It is unclear what patients'/parents' views are about recontacting. This paper explores their views, specifically about the way recontacting might be organised in an ethically sound and practical way.

2. Methods

2.1. Sampling and recruitment

We purposefully sampled participants from four regional genetics services serving a combined population of ~8 million. Local collaborators at each site sent out study information and interested parties contacted the researchers directly. We did not ask collaborators to record the number of patients they contacted, so we are unaware of our response rate. We also posted information on online condition-specific support groups. Participants were 41 patients or parents thereof. Conditions were self-reported: 18 had a condition that was rare (e.g. myotubular myopathy) or undiagnosed; 11 had a suspected hereditary cancer or cardiac condition for which the genetic basis had not been found (e.g. BRCA1/2-negative breast cancer, or a variant of uncertain significance); and 12 had a diagnosis that was clearer (e.g., hereditary breast cancer or Fragile X). All were potentially 'eligible' for being recontacted—either for a test, a variant reclassification, or because a newly identified risk-reducing intervention was available. Four had been recontacted by the genetics service, who offered the patient a test where one was previously unavailable. Table 1 contains more detail.

2.2. Data collection and analysis

We designed an interview schedule (based on our research questions and empirical and conceptual literature) comprising non-leading questions that were general and open-ended. We arranged a suitable time, date, and, for face-to-face interviews, location. This was somewhere the participant could speak with us undisturbed to keep discussions as confidential as possible. We piloted the interview schedule in our earlier interviews, after which we reformulated potentially directive and complicated questions. Dheensa, Carrieri, and Doheny conducted all interviews and had regular team meetings and data analysis sessions to ensure that there was consistency across our approaches. Analysis was thematic and was underpinned by aspects of grounded theory methodology, such as constant comparison (Corbin and Strauss, 2015). Data collection and analysis were iterative, thus we were able to identify new and important questions and areas of ambiguity or tentativeness in the analysis that were then explored in subsequent interviews. The wider team oversaw, discussed, and compared analyses to enhance reliability and rigour. We ceased data collection once we approached saturation of the emerging themes.

3. Results

The four main themes touch upon the overlapping questions of to whom the responsibilities for recontacting belong and the challenges of operationalising different responsibilities, e.g., via ICT infrastructure. The themes, which mirror the participants' lines of

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