



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



Sandi Dheensa, PhD ^{a, b, *}, Daniele Carrieri, PhD ^c, Susan Kelly, PhD ^c,
 Angus Clarke, DM, FRCP ^d, Shane Doheny, PhD ^d, Peter Turnpenny, FRCP, FRCPATH ^{c, e},
 Anneke Lucassen, Dphil(Oxon) FRCP ^{a, b, f}

^a Clinical Ethics and Law, Faculty of Medicine, University of Southampton, UK

^b ELSI Group, Department of Genetics, University Medical Center Groningen, University of Groningen, Groningen, The Netherlands

^c Egenis, University of Exeter, UK

^d Division of Cancer & Genetics, School of Medicine, Cardiff University, UK

^e Peninsular Genetics Service, Royal, Devon and Exeter Hospital, UK

^f Wessex Clinical Genetics Service, University Hospital Southampton NHS Foundation Trust, UK

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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.

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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

* Corresponding author. Clinical Ethics and Law, Room AB 203, MP 801, University of Southampton, Southampton General Hospital, South Academic Block, Tremona Road, Southampton, SO16 6YD, UK.

E-mail address: s.dheensa@soton.ac.uk (S. Dheensa).

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 on-line 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

Table 1
Participants' clinical profiles.

Identifier	Diagnosis	Recontacted in a clinical setting? (i.e. contacted with new information or a new test after being discharged)
1	Hereditary breast cancer (<i>BRCA2</i>)	
2	Hereditary breast cancer (<i>BRCA2</i>)	
3	Possibly hereditary breast cancer (tested negative for <i>BRCA1</i> and <i>BRCA2</i>)	
4	Possibly hereditary bowel cancer (tested negative for Lynch syndrome associated genes)	
5	Possibly hereditary breast cancer (tested negative for <i>BRCA1</i> and <i>BRCA2</i>)	
6	Carrier of <i>BRCA</i> gene and parent of child with hereditary breast cancer	Yes – recontacted for genetic test when one was unavailable before
7	Possibly hereditary breast cancer (tested negative for <i>BRCA1</i> and <i>BRCA2</i>)	
8	X-linked myotubular myopathy	Yes - recontacted for genetic test when one was unavailable before
9	Parent of children with possible mosaic trisomy 16	
10–11	Parents of children with undiagnosed microdeletion syndrome	
12	Parent of children with undiagnosed microdeletion syndrome	
13–14	Parents of child with chromosome 17 duplication	
15	Fragile X	
16	Fragile X	
17	Parent of child with undiagnosed chromosome disorder	
18	Familial hypercholesterolemia	
19	Familial hypercholesterolemia	
20	X-linked myotubular myopathy	Yes - recontacted for genetic test when one was unavailable before
21	Parent of deceased child with <i>Xq28</i> duplication	
22	Parent of child with brachytelephalangic chondrodysplasia punctata	
23	Possibly hereditary cardiomyopathy	
24–25	Parents of child with <i>PCDH19</i> epilepsy	
26–27	Parents of undiagnosed child	
28	Possibly hereditary hypertrophic cardiomyopathy (tested negative for main genes)	
29	Parent of child with <i>1q21.1</i> microdeletion	
30	Parent of undiagnosed child	
31	Parent of child with undiagnosed microdeletion syndrome	
32	Autosomal recessive Alport syndrome and parent of a child with Alport	Yes - recontacted for genetic test when one was unavailable before
33	Hereditary breast cancer (<i>BRCA1</i>)	
34	Possibly hereditary breast cancer (variant of unknown significance)	
35	Hereditary hypertrophic obstructive cardiomyopathy	
36	Possibly hereditary breast cancer (variant of unknown significance)	
37	Possibly hereditary ovarian cancer (variant of unknown significance)	
38	Possibly hereditary breast cancer (variant of unknown significance)	
39	Lynch syndrome	
40	Possibly hereditary arrhythmogenic right ventricular cardiomyopathy (ARVC)	
41	Possibly hereditary breast cancer (tested negative for <i>BRCA1</i> and <i>BRCA2</i>)	

reasoning, are (1) recontacting should not be driven solely by patients/parents; (2) recontacting ought to be a 'joint venture'; (3) improved technical infrastructure is needed to operationalise any joint venture; and (4) difficulties remain around defining and allocating responsibilities.

3.1. Recontacting should not be driven solely by patients/parents

Almost all participants thought that recontacting ought to happen for 'significant' information (that which could lead to, or shed light on, the meaning of a diagnosis, or which could change treatment). However, as the quotations below emphasise, they did not think patients/parents could always be expected to instigate recontact. They would not know where to look for, or how to interpret, information that might help them spot significant changes, and even if they did look for it, there was a dearth of good quality information available;

P5: "[Patients are] not going to be able to find it on their own. So somebody has to be responsible for making that happen".

P18: "[HCPs] know what they're looking at, whereas I could read something online and misinterpret it. And if you're the worrying type, you could start to worry about things."

Availability and searchability of information aside, participants argued that patients/parents would not always have the confidence to ask their HCPs whether potentially relevant information they had seen was indeed relevant and they would not always know who to contact and how. Indeed, several participants were, "confused about who is responsible ... for care or screening" (P39). Some had even sought updates after being invited to do so, but could not get through to anyone who could help them. Participants also pointed out that patients/parents might find seeking information distressing and that it could lead them to "dwell" on their condition;

P33: "It would be nicer if it wasn't the patient that had to do that sort of thing. It keeps you in that cancer mentality and I don't feel that's part of my life anymore."

By contrast, a few participants argued that everybody ought to take responsibility over their own health and that there are insufficient resources to force reluctant patients/parents to engage with seeking information;

P41: "Government-information has to be accessible, but it's not someone else's responsibility to feed it to me. I'm an individual

being and it's my responsibility to use the resources around me. They need to disseminate it and I need to read it [and] act on it".

Underpinning all of these issues was a concern that recontacting, if left entirely to patients/parents, could result in harm, such as them not acting upon information that was relevant to them. Participants perceived this as a harm that could be avoided through HCPs' input.

3.2. *Recontacting ought to be a 'joint venture'*

Leading on from the finding that participants did not think patients/parents ought to be the main drivers of recontacting, some participants thought HCPs had a "duty to contact ... if they find something out" [P22], in part because working for the UK National Health Service (NHS) gave them unique access to data, as well as the knowledge of what information would merit recontact;

P37: "The NHS is probably in pole position for collating this information. I don't know who could be responsible in the way the NHS is."

P30: "You [parent] are probably a good investigator and [can] look into the bits that are specific [to you], but professionals have access to tools that we don't, that give them information that we would never have access to."

For the most part, participants did not argue that the health service should be solely responsible for recontacting, but that recontacting should be a "joint venture" (P37) between patients/parents and professionals. These participants argued for this approach not because, as existing literature suggests, it would be more protective of autonomy and choice about whether to receive information (rather, participants were keen to receive information) but because this model was the only one perceived as practically feasible given the resource constraints in the health service. In fact, many participants thought it would be difficult for the health service to assign any resources to recontacting, because of the pressure on HCPs to see newly referred patients. Some thought NHS staff ought to continue prioritising care for the unwell and public health, and were resigned to thinking it impossible for them to also offer recontact services;

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."

Interestingly, some participants considered the health service's resource-constraints to be an inadequate justification for jettisoning its responsibility to keep patients/parents informed;

P30: "Professionals now, they're given more and more responsibility, more area of people to cover, still the amount of time to do that, and a reduced budget. I understand why [they don't recontact], but it doesn't make it any easier for us to accept".

3.3. *Improved technical infrastructure is needed to operationalise any joint venture*

Participants discussed the limited capacity for recontacting

given the currently available infrastructure:

P5: "It almost comes before the question about should you contact people ... you need a mechanism to highlight the information first before that can happen."

P26: "We go to appointments and we're asked things about [child]. It's all in his notes [but] because he's got five stacks of notes, you think, 'No one ever reads those.' If even the notes can't get sorted, and they [i.e. the different HCPs involved in his care] are asking [us] the same questions, how is someone going to remember in six months' time to phone us up or send a letter, unless there's a foolproof system in place and it says, 'This child has got this', and there's something that prompts them [to contact us]?"

As illustrated, they argued that having a mechanism in place that would enable recontacting preceded the question about whether HCPs had, or could act on, any duty to recontact. Indeed, several participants suggested that the operationalisation of a 'joint venture' model of recontacting required a vastly improved ICT-system.

While participants' ideas about this system were not well-defined, a main feature was that they should have access to an electronic health record (EHR) that could automatically alert them when there was new information available, such as a reclassified variant in a genome database;

P29: "Working in IT, I can't see any reason there can't be some sort of database when they find some new bit of research and this database is updated, from the medical [world] to people. So that [database] can be updated, sends you an email [or] letter, however you want, and you can read it and go back to your consultant if you want to discuss."

Crucially, some also argued that the health service needed better infrastructure for communication between HCPs, as well as between HCPs and patients/parents, and that this was needed for providing joined-up care for multi-system disorders, and not just for recontacting;

P26: "You might have somebody who looks after hearts or looks after lungs, but the body all works in sequence together. They all have to talk."

Some participants mentioned previous expensive ICT failures in the NHS, and suggested that a system had not been set up because it would be "a massive cultural change ... [which] by the time they've started to implement, is already out of date" [P26]. Others pointed out that improving infrastructure might not be difficult, because the data already existed: it would simply involve better integration and curation and for HCPs to use the data more efficiently;

P30: "The [health service has] a lot of centralised database sharing, so I can't see that it would be too difficult to obtain and return information that's relevant to you ... Various different people and users can have different levels of access ... [It] wouldn't be difficult to set up searching criteria. Databases are fast, so it would only take seconds to come back with information."

They argued that such infrastructure would enable patients/parents to get some information without waiting a "long period to get a genetics appointment" (P21), that it would be "money well spent" [P24] and that it could ultimately save the health service

money as it could reduce unneeded referrals [P31], record investigations already done [P20], improve efficiency and communication between HCPs and patients/parents, and improve rates of diagnosis and screening. This could in turn reduce hospital stays [P24] and reduce litigation against the health service [P26]. They perceived the potential benefits and cost-savings as particularly great in the genomic setting because;

P31: “It’s both parents, extended family. So it could save the NHS thousands. It costs lots of money [for the NHS] to input the data to start with, but I can see exponentially it would be better in the long run”.

From participants’ perspectives, increased efficiency could enable even those patients/parents whose hospital trusts were understaffed to be recontacted, thus enhancing fairness in treatment across the country. They contended that investing in such infrastructure would help to build trust in the health service because patients/parents would be reassured that it was using the best possible tools for diagnosis [P41]. By enabling ongoing contact and recontact, infrastructure could also make it more possible for patients/parents to receive support from the health service. P31, who had seen the genetics service once when her (now adult) son was first noticed to have autism, highlighted a desire for such support;

P31: “Nobody’s ever put any input into it; it’s just, ‘go away and get on with it’. They give you this, ‘he has this wrong, but we don’t know what it means, but we don’t want to see you again’. And that’s quite frightening. There is the element of support within information.”

Importantly, some participants expressed concern about who would have access to their data and stressed the need for transparency and rigorously-controlled access to protect their privacy;

P23: “I wouldn’t want my particular medical details going to just anybody”.

P26: “I’d like to know who’s been looking at my records, because you hear and see of people who work in the NHS and they’re looking at other people’s records which aren’t relevant to them. The trust thing, that when you give information – and it’s quite personal, your health information – you want to choose who you share it with, unless obviously something you’ve got is a risk to the rest of society. [It’s important] that you can control that data.”

Participants thought that seeing who had accessed their records, and seeing that only ‘relevant’ people could do so, would enhance transparency and build trustworthiness. However, in the above quotation, P26 points to a bigger question about how control over data privacy balances with, or potentially hinders, the ‘openness’ of data required for care and investigation. Another bigger question was whether and how any updates about *research* studies could and should be integrated into records used in *clinic*, with some participants thinking EHRs could integrate research findings via the process described.

3.4. Difficulties remain around defining and allocating responsibilities

Participants thought that, under a ‘joint venture’ model, there would remain difficulties defining and allocating responsibility. For example, P29 was unsure at what point a HCPs’ duty to recontact

patients/parents was discharged;

P29: “90% of me says, once you’ve been told something, like go and get your flu jab, if you don’t follow it up, if you get flu and die it’s your fault. But 10% of me says the NHS has a duty of care ... but you can keep going back to the funding, there’s only so much they can do. Once it’s been sent out and they’ve been told there’s new information there, that’s almost enough. But that becomes difficult. I’m thinking aloud.”

Potential inaction by patients/parents was another issue. P34 for example pointed out that “*big and very certain*” public health messages “*are ignored by most people*”, raising questions about how the “*NHS marshals its extremely limited resources*”. That is, she questioned whether any resources spent on recontacting would indeed be “*well-spent*”, as P24 claimed.

A few others thought that compared with the public health setting, the availability of specific information to specific patients in the genetic setting would both augment HCPs’ duty and make the dissemination of information more worthwhile, and that targeting messages might make inaction less likely. Yet, participants also gave examples of where they had not acted on information HCPs had given them—e.g., because they did not take advice “*religiously*” [P21] or because they felt contacting HCPs would be “*silly ... [because they felt] perfectly well, [which did not warrant] making a fuss*” [P39]. Given that there were so many perceived difficulties that could thwart any resulting benefit from a recontact—from practical issues (e.g., parents/patients not checking their EHR), to psychosocial issues (e.g. coping by avoiding information), to limited comprehension (e.g., misunderstanding the information), a conceptual question remained, and underpinned participants’ thinking, about whether a HCPs’ duty would be discharged once the EHR was updated, and how far HCPs should go to ensure the patient comes to clinic. Practical barriers might be more easily addressed, but others, such as patients/parents not thinking the information important or relevant, could be less easy to overcome.

4. Discussion

This study has explored patients/parents’ views about how recontacting might be organised ethically and practically in clinical genetic practice. A key finding was that most participants considered recontacting important and desirable, but that resource constraints in the UK’s health service would make it difficult or impossible for HCPs to recontact patients/parents. Thus, they thought any model of recontacting would have to be a joint venture, whereby patients/parents and HCPs share the efforts involved. To an extent, our findings echo those from a study by [Townsend et al. \(2012\)](#), in which participants (HCPs, parents of children who had undergone genetic testing, and members of the public) thought that patients/parents were responsible for contacting HCPs to check for genomic developments. While HCPs thought so for reasons to do with the practicality of recontacting, members of the public and parents’ reasons were about having a choice over whether to learn about new developments. In our study, the majority of participants were in favour of receiving new information, so the desire for choice in this regard did not materialise. Importantly, however, they maintained that the joint venture model would be in line with the idea that individuals ought to take responsibility for maintaining and promoting their own, or their children’s, health. Our findings reflect a wider movement in the western developed economies of increasing patient involvement in healthcare and, more broadly, the move towards ‘shared decision-making’ in the UK health service ([Coulter and Collins, 2011](#)). The findings also align with our recent recommendation advocating a

model of shared responsibility, with a suggestion that HCPs should routinely discuss recontacting with patients/parents, including issues that might trigger it, in the context of consent for genetic testing (Carriero et al., 2017b). Joint ventures between HCPs and patients have been proposed in other contexts of clinical genetics, for example by Wouters et al. (2016) regarding whether HCPs or patients are responsible for disseminating genetic information to at risk relatives and by Mackley et al. (2017) regarding disclosure of secondary findings.

Participants suggested that a way the joint venture model could be operationalised was via an EHR that centralises their information from across different services and automatically alerts them when there is new information available, e.g., when a genome database is updated and a variant is reclassified. Extrapolating from our participants' descriptions, the work involved in the process of recontacting would then be shared in that professionals (lab scientists, researchers, and possibly HCPs) would update genome databases, patients/parents would check their EHR and respond to the resultant alert, and HCPs would discuss the new information with the patient/parent to assess its relevance. This finding highlights a key issue, which is that the lack of clarity about recontacting is a symptom of a wider problem: the deficiency of necessary infrastructure to (i) pool and curate high-quality, harmonised genomic data, (ii) to link this data to other quality health data, (iii) to make this data available to HCPs involved in care, and (iv) to enable patients/parents to receive updates—and importantly to do so in a responsible and trustworthy way, that respects privacy and confidentiality. Currently, the absence of a global variant-calling database would be a barrier to implementing the optimal version of a system described by our participants, although groups such as the Global Alliance for Genomic Health are working to improve these processes (Siu et al., 2016). The American College of Medical Genetics and Genomics (ACMG) recently released a position statement advocating the “extensive sharing of laboratory and clinical data”, on the basis that sharing can improve care, research, and web-based decision support tools, and can offer financial benefit, such as reducing the duplication of unpublished but resolved pharmaceutical research. They argue that data sharing can be compatible with protection of privacy and that sharing systems should be secure and “provide transparency in the documentation of data sharing transactions” (ACMG Board of Directors (2017)). Our findings emphasise the importance of this: participants underscored the importance of privacy, as well as transparency and control over who could access data and for what purpose. We note the argument of Prainsack (2017) here, that the term ‘sharing’ is best avoided in the context of genomic data because it is too broad and has inappropriately moralistic undertones—we have used it here, albeit with reservations, because it has gained traction in the debates.

Further research and thinking is required around the enormous challenge of translating aggregate genomic data into alerts in individual EHRs. As well as the technical problem of how best to integrate different ICT infrastructures, challenges would arise for laboratory scientists, researchers, and possibly HCPs interpreting the data and deciding to which ‘types’ of patients the information would be relevant. They would then need to decide how to inform patients/parents (for example, in what format to provide the information and whether decision aids would be helpful) to enable patients/parents to make meaningful choices. Increasing the availability of up-to-date and secure genomics databases and EHRs is only part of the challenge.

More broadly, the use of EHRs raises ethical issues. While claims have been made that they can empower patients (Giardina et al., 2014), they also pose a risk to privacy—members of a patient's family or even employers or insurers could access records without

the patient's consent or knowledge (Wynia and Dunn, 2010). The implications of this could be amplified where the EHR includes genetic information, because this information is often thought of as particularly sensitive, as it can be predictive and relevant to family members. For this reason, clinical genetics notes are usually separate from a patient's medical record. Some patients/parents might also perceive the receipt of an alert as being in violation of their right, or interest, not to know the information, particularly in jurisdictions where the law specifically protects this right (German Genetic Diagnosis Act). Given these issues, we would agree with Shoenbill et al. (2014), who think HCPs should discuss the potential integration of genetic information into a EHR in the consent process and in any post-test counseling with patients/parents. We also argue that patients/parents should have the option of opting-out of receiving alerts about new information, as well as the opportunity to opt back in easily at any time, or after a certain period.

An overarching issue about any ICT-based solution is the risk of introducing inequity: disadvantage to the less ICT-savvy and/or those whose lives render them—for social, economic, or other reasons—less able to engage effectively with the process of recontacting. HCPs would end up seeing and reinterpreting information for patients/parents who do respond to automatic updates, leaving less time for actively engaging with the less responsive, and potentially needier, patients. This reflects Tudor Hart's inverse care law (Tudor, 1971): those who need healthcare less will use it more, while those whose needs are greater will not engage as effectively, and in turn, their needs will be relatively neglected. Any solution that puts some responsibility to act on patients/parents might lead to inequity, but one that uses ICT might make the divide greater.

5. Conclusion

This paper has shown support for a joint venture model of recontacting. Questions regarding this model, whether operationalised via ICT or otherwise, need to be addressed urgently, and of course the proposed ‘solutions’ offered here may generate ethical as well as practical difficulties. One consideration is what information a HCP should contact a patient/parent with in the first instance: should they send a general notice that there are new developments or information that is more specific? The more unwieldy questions include, when is the HCPs' duty discharged within this shared model? How would one know when the practice of recontacting has been sub-optimal and what possible forms of redress (e.g. claims of negligence) are available? How can recontacting be implemented without exacerbating pre-existing health inequalities? We need further debate about the limitations and normative implications of using ICT infrastructure as a solution to this intractable problem. This paper is intended to instigate such debate.

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References

- ACMG Board of Directors, 2017. Laboratory and clinical genomic data sharing is crucial to improving genetic health care: a position statement of the American College of Medical Genetics and Genomics. *Genet. Med.* <http://dx.doi.org/10.1038/gim.2016.196>.
- Ali-Khan, S.E., Daar, A.S., Shuman, C., Ray, P.N., Scherer, S.W., 2009. Whole genome scanning: resolving clinical diagnosis and management amidst complex data. *Pediatr. Res.* 66, 357–363.
- Andrews, L.B., 1992. Torts and the double helix: malpractice liability for failure to warn of genetic risks. *Houst Law Rev.* 29, 149–184.
- Brown, K.L., Moglia, D.M., Grumet, S., 2006. Genetic counseling for breast cancer

- risk: general concepts, challenging themes and future directions. *Breast Dis.* 27, 69–96.
- Carrieri, D., Lucassen, A.M., Clarke, A.J., et al., 2016. Recontact in clinical practice: a survey of clinical genetics services in the United Kingdom. *Genet. Med.* 18 (9), 876–881.
- Carrieri, D., Dheensa, S., Doheny, S., Clarke, A., Turnpenny, P., Lucassen, A., Kelly, S., 2017. Recontacting in clinical practice: an investigation of the views of healthcare professionals and clinical scientists in the United Kingdom. *Eur. J. Hum. Genet.* <http://dx.doi.org/10.1038/ejhg.2016.188>.
- Carrieri, D., Dheensa, S., Doheny, S., et al., 2017. Recontacting in clinical genetics and genomic medicine? We need to talk about it. *Eur. J. Hum. Genet.* <http://dx.doi.org/10.1038/ejhg.2017.8>.
- Chen, S.C., Berkman, B.E., Hull, S.C., 2017. Recontacting participants for expanded uses of existing samples and data: a case study. *Genet. Med.* <http://dx.doi.org/10.1038/gim.2016.207>.
- Clayton, E.W., Haga, S., Kuzler, P., Bane, E., Shutske, K., Burke, W., 2013. Managing incidental genomic findings: legal obligations of clinicians. *Genet. Med.* 15, 624–629.
- Corbin, J., Strauss, A., 2015. *Basics of Qualitative Research*, third ed. SAGE Publications, Thousand Oaks, USA.
- Coulter, A., Collins, A., 2011. *Making Shared Decision-making a Reality. No Decision about Me, without Me*. King's Fund, London. Available at: https://www.kingsfund.org.uk/sites/files/kf/Making-shared-decision-making-a-reality-paper-Angela-Coulter-Alf-Collins-July-2011_0.pdf. Accessed 7 February 2017.
- Giardina, T.D., Menon, S., Parrish, D.E., Sittig, D.F., Singh, H., 2014. Patient access to medical records and healthcare outcomes: a systematic review. *J. Am. Med. Inf. Assoc.* 21 (4), 737–741.
- Griffin, C.A., Axilbund, J.E., Codori, A.M., et al., 2007. Patient preferences regarding recontact by cancer genetics clinicians. *Fam. Cancer* 6, 265–273.
- Hastings, R., de Wert, G., Fowler, B., et al., 2012. The changing landscape of genetic testing and its impact on clinical and laboratory services and research in Europe. *Eur. J. Hum. Genet.* 20, 911–916.
- Hunter, A.G., Sharpe, N., Mullen, M., Meschino, W.S., 2001. Ethical, legal, and practical concerns about recontacting patients to inform them of new information: the case in medical genetics. *Am. J. Med. Genet.* 103, 265–276.
- Letendre, M., Godard, B., 2004. Expanding the physician's duty of care: a duty to recontact? *Med. Law* 23, 531–539.
- Mackley, M.P., Fletcher, B., Parker, M., Watkins, H., Ormondroyd, E., 2017. Stakeholder views on secondary findings in whole-genome and whole-exome sequencing: a systematic review of quantitative and qualitative studies. *Genet. Med.* <http://dx.doi.org/10.1038/gim.2016.109>.
- Mulla, E., 2015. Severe lack of investment in electronic infrastructure in the NHS. *BMJ* 351, h4170.
- Murray, M.L., Cerrato, F., Bennett, R.L., Jarvik, G.P., 2011. Follow-up of carriers of BRCA1 and BRCA2 variants of unknown significance: variant reclassification and surgical decisions. *Genet. Med.* 13, 998–1005.
- Otten, E., Plantinga, M., Birnie, E., et al., 2014 Dec 11. Is there a duty to recontact in light of new genetic technologies? A systematic review of the literature. *Genet. Med.* 17 (8), 668–678.
- O'Connor, M.R., 2014. *Patient and Genetics Health Care Providers Attitudes Regarding Recontact* [Thesis]. University of Pittsburgh, Pittsburgh, PA.
- Pelias, M.Z., 1991. Duty to disclose in medical genetics: a legal perspective. *Am. J. Med. Genet.* 39, 347–354.
- Pelias, M.Z., 1992. The duty to disclose to relatives in medical genetics – response. *Am. J. Med. Genet.* 42, 759–776.
- Peshkin, B.N., Burke, W., 2007. Bioethics of genetic testing for hereditary breast cancer. In: Isaacs, C., Rebbeck, T.R. (Eds.), *Hereditary Breast Cancer*. Informa Healthcare USA, New York, pp. 35–49.
- Prainsack, B., 2017. Why we should stop talking about data sharing. Available at: <http://dnadigest.org/why-we-should-stop-talking-about-data-sharing/>. Accessed 7 February 2017.
- Pyeritz, R.E., 2011. The coming explosion in genetic testing—is there a duty to recontact? *N. Engl. J. Med.* 365, 1367–1369.
- Sexton, A.C., Metcalfe, S.A., 2008. Disclosing genetic research results after death of pediatric patients. *JAMA* 300, 1693–1695.
- Sexton, A.C., Sahhar, M., Thorburn, D.R., Metcalfe, S.A., 2008. Impact of a genetic diagnosis of a mitochondrial disorder 5–17 years after the death of an affected child. *J. Genet. Couns.* 17, 261–273.
- Sharpe, N.F., 1999. The duty to recontact: benefit and harm. *Am. J. Hum. Genet.* 65, 1201–1204.
- Shirts, B.H., Parker, L.S., 2008. Changing interpretations, stable genes: responsibilities of patients, professionals, and policy makers in the clinical interpretation of complex genetic information. *Genet. Med.* 10, 778–783.
- Shoenbill, K., Fost, N., Tachinardi, U., Mendonca, E.A., 2014. Genetic data and electronic health records: a discussion of ethical, logistical and technological considerations. *J. Am. Med. Inf. Assoc.* 21 (1), 171–180.
- Siu, L.L., Lawler, M., Haussler, D., et al., 2016. Facilitating a culture of responsible and effective sharing of cancer genome data. *Nat. Med.* 22 (5), 464–471.
- The 2009 German Genetic Diagnosis Act (GenDG) Section 8 Para 3–4.**
- Townsend, A., Adam, S., Birch, P.H., Lohn, Z., Rousseau, F., Friedman, J.M., 2012. “I want to know what's in Pandora's box”: comparing stakeholder perspectives on incidental findings in clinical whole genomic sequencing. *Am. J. Med. Genet. Part A* 158 (10), 2519–2525.
- Tudor, Hart J., 1971. The inverse care law. *Lancet* 297 (7696), 405–412.
- Wouters, R.H., Bijlsma, R.M., Ausems, M.G., Delden, J.J., Voest, E.E., Bredenoord, A.L., 2016. Am I my Family's Keeper? Disclosure dilemmas in next-generation sequencing. *Hum. Mut.* 37 (12), 1257–1262.
- Wynia, M., Dunn, K., 2010. Dreams and nightmares: practical and ethical issues for patients and physicians using personal health records. *J. Law Med. Ethics* 38, 64–73.