COMMENT

Equity implications of patient-initiated recontact and follow-up in clinical genetics

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The practice of recontact in clinical genetics has both the ability to rectify longstanding inequities in genetic testing, and the potential to perpetuate these same inequities. Recently, there has been a robust conversation about equity issues in clinical genetics, such as those resulting from the lack of diversity in genomic datasets, with corresponding efforts being made to identify and advance solutions [1]. While the broader conversation around equity in clinical genetics has progressed rapidly, the same is not true for equity in recontacting practices.

Recontact—where contact between a healthcare provider and patient is reinitiated in the absence of an ongoing relationship—has long been a topic of discussion in clinical genetics due to the dynamic nature of our ability to test for genetic conditions and interpret the results. However, with the increasingly widespread use of sequencing technologies, debates around duties to *reinterpret* genetic data and *recontact* patients—where the latter concept typically incorporates the former—have recently come to the forefront. In particular, the increasing volume of variants of uncertain significance (VUS) has catalyzed careful consideration of recontacting practices, roles, and obligations. In contrast to the disclosure of other diagnostic test results, there is an expectation that the classification of these uncertain results will change over time and could meaningfully impact patients.

Given the potential for updated results to provide benefit to patients, recontact in clinical genetics is felt to be an ethically desirable goal [2]. Significant barriers, however, exist to operationalizing systematic reinterpretation and recontact, including feasibility, limited resources and infrastructure, changing contexts or consent for the recipients, and an overall lack of guidelines and consensus [3]. Although desirable, a comprehensive systematic review on the subject by Otten et al. ultimately fell short of identifying a legal or ethical duty to recontact in light of new genetic information [2]. Subsequently, some authors have argued that such an ethical duty may exist [4], however, this is not widely accepted or reflected in practice, largely due to the operational barriers discussed above.

Following from these early ethical and legal discussions, as well as empirical work exploring practices and stakeholder views on recontact, recommendations for recontact in clinical practice were published by the European Society for Human Genetics (ESHG) [5] and the American College of Medical Genetics (ACMG) [6]. Ultimately, to balance the likely benefit afforded to patients through recontact with system and resource limitations, both groups propose a model of shared responsibility. In such a model, three distinct groups—patient-facing healthcare providers (including genetics physicians, genetic counselors, referring providers), laboratory professionals, and patients themselves—all have a role to play.

It is our view, however, that the equity implications of a shared model of responsibility in recontact have been insufficiently discussed to date. On one hand, equity can be considered an important argument in favour of routine recontacting practices. It is well-established that variants of uncertain significance (VUS) are more common in non-European ethnic groups, due to their systematic under-representation in the datasets used to inform the analysis of these variants [4, 7]. Therefore, individuals in these under-represented groups are the most likely to benefit from the reclassification of VUS over time, as efforts are made to diversify our reference datasets [7]. This positions reinterpretation and recontact among other tools identified to help resolve inequities in genetic testing.

To achieve this goal, however, recontacting practices must be universal. Presently, clinicians and laboratories indicate that recontact is not systematic, but happens in an ad hoc manner [8, 9]. Recontact is triggered by scenarios such as the discovery of new information, detection of the same variant in a further patient, or upon request. Although responsibility for recontact is intended to be shared, it is our experience that these ad hoc instances of recontact are overwhelmingly patient-driven. In our publicly funded healthcare system, and at least at our institution, we in fact promote this: patients who receive VUS are often not booked for routine follow-up but are advised that they may reconnect with the service in the future to revisit uncertain results. This patient-driven experience is mirrored by empirical data from Australia demonstrating that most genetic services rely on patients or family to initiate recontact [10]. Ultimately, constraints on clinicians and laboratories due to workload and patient volumes, as well as lack of standardized practices or systems, cause the aspirational shared model of responsibility for recontact to fall apart and rely almost entirely on patients.

A paradigm reliant on patient-initiated recontact directly undermines the potential for routine recontacting practices to promote equity in genetic testing. Patients who most frequently and readily access medical care are those with social advantage, including those with higher levels of education and health literacy, those whose first language is consistent with the dominant

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language of the region, and members of majority ethnic groups. In fact, the recommendations from both the ESHG and ACMG caution against placing primary responsibility for recontact on patients for this reason [5, 6]. This is, unfortunately, consistent with our local experience of those patients who initiate recontact—they are overwhelmingly white, educated, English-speaking families, who are not representative of the total population of patients who access our service. We are therefore entirely missing patients from the underrepresented groups that we anticipate recontact and reinterpretation can benefit the most. Furthermore, the unique population seen in clinical genetics serves to compound these inequities when recontact is patient-driven. Patients with disabilities, including both intellectual and physical, are enriched in clinical genetics. These patients may have impaired abilities to initiate recontact compared to others, relating to the very reason they had testing in the first place.

The equity implications of patient-initiated interactions in genetics extend beyond recontact to include follow-up more broadly. In contrast to recontact, "follow-up" entails a new interaction between a healthcare provider and patient, where the care relationship remains ongoing. Patient-initiated follow-up is common practice in clinical genetics, where an offer is made for follow-up at the discretion of the patient, such as for additional post-test counselling. Although recontact is the point of current consideration in the genetics community, the two concepts overlap and share important equity implications when patient-initiated. Therefore, consideration of responsibility and practices in recontact can extend to follow-up in clinical genetics more broadly.

Moving forward, as policies and practices are updated, the equity implications of a shared model of responsibility in recontact (and follow-up) must be carefully considered. As it stands, the aspirational shared model is not shared equally: patients are driving recontact in clinical genetics. As a result, there is a disconnect between the potential for recontact—if systematic—to promote equity in genetic testing and the inconsistent patient-initiated product. Instead, the practice of recontact serves as a tool to reinforce existing inequities in genetic testing. In response, greater efforts must be given to implementing a systematic approach to recontact to ensure the practice is equitable.

At the level of genetics services and laboratories, consideration should be given to developing uniform recontacting policies, rather than employing an ad hoc approach. Systematic recontact may become increasingly feasible as technologies improve, but may also become more expected as the practice becomes more widespread. As feasibility improves, the equity implications of a failed shared model of responsibility lend additional support to arguments in favour of an ethical duty or responsibility to recontact in clinical genetics.

At the level of individual genetics healthcare providers, while consistent service-level policies take shape, efforts to educate and inform patients around reinterpretation and recontact should continue. Consideration could be given to personal practices where patients less likely to reinitiate recontact themselves or those most likely to benefit from follow-up of an uncertain result or challenging disclosure receive provider-initiated follow-up or recontact. This short-term solution is limited by its subjective nature, relying on provider judgment which is subject to its own biases. Consideration could also be given to how other clinicians, such as primary care providers, can take on greater responsibility. Provider-driven recontact is also limited by the current state of the healthcare workforce, with major shortages and many clinicians experiencing unprecedented levels of burnout. This leads to varying capacities of individual providers, and potential apprehension around taking on further responsibility, leading to further inequities for patients.

Finally, at the level of the health system, efforts should be made to advocate for resourcing genetics services in a manner that supports a more universal system of recontact. These efforts should be complemented by empirical and health technology assessment to ensure costs and efforts are proportionate to expected benefits [3, 5]. Enabling a universal system of recontact in clinical genetics will ensure that the benefit of reinterpretation and recontact can be afforded to all patients—rectifying current inequities in genetic testing, rather than perpetuating them.

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ADDITIONAL INFORMATION

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