Why genetics services should contact at-risk relatives directly

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Finding out you have a genetic mutation is not an insular experience: this difficult revelation nearly always has implications for relatives as well. Often, good clinical interventions are available that could safeguard relatives' future health and perhaps even save their life. However, it's sometimes difficult to make sure this important information gets passed throughout families efficiently, sensitively and accurately.

For Caroline Rich, who was diagnosed with breast cancer at 28, the processes of family communication were unsuccessful (1). Caroline only learned that there was a BRCA1 mutation in her family after her cancer had developed. Although several members of her family knew about it, no-one had thought to tell her. There wasn't a feud or any explicit refusal to disclose, but the fact that her grandmother and two cousins carried the mutation never made it through.

Within clinical genetics services, there are two broad models for the dissemination of information throughout families. The first and most common approach is 'family contact', which requires a family member, usually the proband who has the detected mutation, to approach their at-risk relatives with information about their condition, and to tell them about contacting the genetic services for testing. The second method is for genetics services to approach relatives directly, using contact details provided by the patient.

In an article published last year (2) we argue that for many genetic disorders, clinical genetics services have a duty to attempt to use direct contact, rather than merely relying on family-mediated communication. Direct contact is one way by which Caroline could have been given the information she needed. Whilst it may not be suitable to use direct contact to warn relatives for every genetic condition, we believe that it is suitable and appropriate when there is a clear clinical intervention available to reduce risk. However, direct contact does not mean that patients are left out of the picture. In fact, our model depends upon working in close partnership with patients to ensure that serious harm can be avoided. It also has several advantages.

First, direct contact removes the burden on the patient of having to approach relatives to explain often complex concepts of genetic information and treatment. Some patients find this experience very difficult, perhaps experiencing feelings of guilt and blame. Others adopt an over-zealous approach and might end up causing harm to relatives in the process.

Second, direct contact also ensures that the information passed on to relatives is accurate and efficient. Several recent studies have shown that relying on the patient leads to a defective transfer of information, as 'Chinese whispers' filter throughout the family.

Third, a recent Australian study has shown that direct contact almost doubles the number of relatives who present for counselling, which both reduces the health burden and improves the cost efficiency of the service. Fourthly, direct contact should reduce the problem of relatives feeling unduly influenced or coerced into being tested by their family members. Any letters sent to a patient's relatives will emphasise the voluntary nature of testing, and that the recipient will be in control of the process at all times.

Finally, there is good evidence that patients and relatives endorse direct contact. Relatives who are contacted do not tend to express concern, and none in the above studies complained about a breach of privacy. Although some relatives may experience initial anxiety or shock, overwhelmingly they are glad to have heard about their risk status. This evidence counteracts concerns that relatives will be harmed through receiving unsolicited information 'out of the blue'. Any risk can be further mitigated by involving patients; asking them to informally approach relatives before they are contacted by the genetics service, or of course if a patient has concerns about how a particular relative may respond to direct contact, the process can be stopped at that point.

Direct contact does of course raise a number of issues. It is likely to be expensive to set up, but, it is important not to confuse this practical and logistical problem with discussion about potential ethical concerns. It could be argued that it isn't a genetics service's responsibility to contact relatives, but the patient's. We agree with this in principal and don't advocate that clinical services should take on an ethical or legal responsibility for disclosure to relatives. However we believe that genetics services do have a duty to get involved in the process, in partnership with the patient, and to become more proactive than they are currently. Finally, it has been stated that direct contact may breach a relative's right 'not to know' about their status. But as some genetic conditions have effective interventions available, we argue there is an ethical imperative to provide a choice about testing. Clearly no one should be 'forced' to know their mutation status, but we believe that for diseases with clinically proven treatment options, the right to remain in complete ignorance of risk is greatly weakened.

Many of the ethical concerns can be mitigated through careful structuring of contact with relatives. For example, concerns about discrimination can be reduced by providing only general information in contact letters. And worries about breaching privacy can be addressed through careful data management, such as destroying contact details of those relatives who don't respond.

It is a reality of genetics that information goes beyond individuals to the wider family. Direct contact offers a supplementary process that empowers patients but does not overly burden them. It involves genetics services and families working in partnership to warn at-risk family members, giving them the best chance of an accurate diagnosis and potentially a chance to avoid the health consequences of a genetic disease.

SOURCES & REFERENCES

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