Is There A Duty to Share Genetic Information?

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Abstract

A number of prominent bioethicists such as Mike Parker, Anneke Lucassen, and Bartha Maria Knoppers have called for the adoption of a system in which by default, genetic information is shared among family members. In this paper, I suggest that a main reason given in support of this call to share genetic information among family members is the idea that genetic information is essentially familial in nature. Upon examining this 'familial nature of genetics' argument, I show that most genetic information are only shared in a weaker way among family members and do not necessarily lead to the actual manifestation of particular diseases. The upshot is that the idea that genetic information is familial in nature does not provide a sufficient ground for why we should move towards a system in which by default, genetic information is shared among family members.

Is There A Duty to Share Genetic Information?

I.

As predictive genetic testing becomes prevalent, the following kinds of cases will arise more and more frequently in clinical genetic settings:

> **Anna, Betty and Carl Case:** Anna and Betty are sisters who do not get along and rarely speak with one another. Anna just learned that her 2-year-old son, Carl, has Duchenne's muscular dystrophy. Betty has recently married and is planning parenthood. Anna knows that Betty would consider preimplantation genetic diagnosis or not have children at all if Betty knew about Carl's condition. The genetic staff offered Anna to help communicate Carl's diagnosis to Betty so that Betty can make an informed decision about her reproductive choices. However, Anna declines this offer, reasoning that Betty should be personally responsible for her own health and reproductive conditions.

> **Fred, Garry and Heather Case:** On his 55th birthday, Fred was diagnosed with Huntington's disease. Fred informs the medical staff that he does not want his son, Garry, to know about this diagnosis, believing that Garry will blame him for bearing faulty genes. Garry himself has explicitly said that he does not want to know about any predisposition to any genetic disease. His wife, Heather, is however 9 weeks pregnant and would consider an abortion if the fetus were found to have the Huntington's disease gene mutation.

These cases raise at least the following ethical questions: Does Anna have a duty to disclose the fact that her son, Carl, has Duchenne's muscular dystrophy to her sister, Betty? Does the medical staff have a duty to inform Heather of the fact that her father-in-law, Fred, has Huntington's disease?

While some governmental bodies have argued that a confidentiality agreement with a patient should be paramount,[1] other government bodies have said that it may sometimes be justified to disclose genetic information to other family members, even if an individual desires confidentiality. For example, the U. K. General Medical Council guidelines on confidentiality, which are not specifically aimed at genetics, but which are the main guidelines to which practitioners in genetics would be accountable, states that confidentiality 'may' be breached when there is 'risk of death or serious harm.'[2, 3] Similarly, the American Society of Human Genetics Social Issues Subcommittee on Familial Disclosure permits professionals to breach confidentiality in exceptional cases.[4]

Some prominent bioethicists have gone further and argued that, by default, genetic information should be shared among family members. For example, Mike Parker and Anneke Lucassen proposed that in the case of genetic information, we should seriously consider moving away from a 'personal account model' towards a 'joint account model.'[5] As Parker and Lucassen explain, "Whereas on the personal account model the default position is an assumption of confidentiality, on the joint account model it is assumed that information should be available to all account holders unless there are good reasons to do otherwise." [5] Other bioethicists have made similar claims.[6, 7]

A main reason given in support of this call to share genetic information among family members is the idea that genetic information is essentially familial in nature. Call this the Genetic Information is Familial Thesis (GIFT). For example, Parker and Lucassen write, "Genetic information is, spontaneous mutations aside, essentially and unavoidably familial in nature."[5] Or, Bartha Maria Knoppers says that "the very nature of genetic information, as both individual and universal, now mandates its treatment as familial."[6]

On the surface, GIFT seems like a very plausible thesis. Certainly, those who believe in 'genetic exceptionalism,' according to which genetic information is unique in medicine and deserves special treatment, also typically point to GIFT in order to support genetic exceptionalism. For instance, Patricia Roche and George Annas cite the fact that "an individual's DNA can also reveal information about risks and traits that are shared with genetic relatives" as a reason for holding onto the view that separate rules and laws are needed for handling genetic information.[8] Also, even those who have argued against genetic exceptionalism seem to accept GIFT. For example, with respect to the point that genetic information is familial in nature, Tom Murray's response is to accept it but to note that other kinds of medical information such as one's cholesterol level or whether one has been exposed to an infectious disease are also familial in nature.[9] Given that GIFT appears to be widely accepted, basing the call to share genetic information among family members on GIFT seems to have certain initial plausibility.

However, I shall argue that GIFT requires refinement, and that once refined, one sees that most genetic information are only shared in a weak way among family members and do not necessarily lead to the actual manifestation of particular diseases. The upshot

is that GIFT does not provide a sufficient ground for why we should move towards a system in which by default, genetic information is shared among family members.

It is worth saying at the outset that I am not making the stronger claim that there can be no valid reason why we should adopt a system in which by default genetic information is shared among family members. There may be other ways of grounding the duty to share our genetic information with family members. In this paper, I am only making the weaker claim that GIFT does not furnish us with such a reason.

II.

Without refinement, the impression one gets from the idea that genetic information is familial in nature, i.e., GIFT, is that there is a very high correlation between knowing that an individual has the genes for a particular genetic disease and being justified in believing/inferring that a relative will also have this particular disease. But GIFT requires refinement in the following ways:

First, when Parker and Lucassen say that genetic information is *spontaneous mutations aside* essentially familial in nature, this statement makes 'spontaneous mutations' seem as if they occur infrequently. However, depending on the genetic disease at issue, spontaneous mutations can in fact occur frequently. For example, in the case of Duchenne's muscular dystrophy, 1 out of 3 cases is the result of spontaneous mutation.[10] This means that the probability that this disease is 'familial in nature' is only 2/3 or 66%.

Secondly, it is helpful to distinguish between different kinds of genetic relations; some are stronger and some are weaker:

Stronger Genetic Relation: If two individuals, X and Y, are biologically related in

a certain way, and if all you know is that X has the genes, G, then you are

justified in believing to a high degree of probability, P, that Y also has G.

Weaker Genetic Relation: If two individuals, X and Y, are biologically related in

a certain way, and if all you know is that X has the genes, G, then you are

justified in believing to a medium-to-low degree of probability, P, that Y also has

G. (Note that I shall use actual probabilities later on).

An example of a Stronger Genetic Relation is the case of monozygotic twins.

Suppose Adam and Bob are monozygotic twins, and suppose all you know is that Adam

has genes for Hungtington's Disease. In virtue of the fact that monozygotic twins

typically have identical genetic makeup, you are justified in believing that there is a high

degree of probability that Bob also has the genes for Huntington's Disease. An example

of a Weaker Genetic Relation is the case of a mother and her son. Suppose all you know

is that Clara, a mother, has genes for Hungtington's Disease. Then, given the fact that

typically an offspring gets only ½ of his or her genes from the mother, you are justified in

believing that there is a medium-to-low degree of probability that David, her biological

son, also has the genes for Hungtington's Disease.

It should be obvious that Stronger Genetic Relations occur only in a handful of

cases. In addition to monozygotic twins, if human cloning became possible and were

practiced, then there would also be Stronger Genetic Relations in those cases. Other than that, most genetic relations would be Weaker Genetic Relations.

Thirdly and finally, some genes are highly penetrant while others are not, where penetrance is the proportion of individuals' carrying a particular variation of a gene that also expresses a particular trait, e.g., a particular disease. Let us call cases in which individuals have highly penetrant genes 'deterministic,' and cases in which individuals have low penetrant genes 'non-deterministic.'

Deterministic Case: If all you know is that an individual, X, has the genes for a disease, then you are justified in believing to a high degree of probability, Q, that X will develop the disease.

Non-Deterministic Case: If all you know is that an individual, X, has the genes for a disease, then you are justified in believing to a medium-to-low degree of probability, Q, that X will develop the disease.

An example of a Deterministic Case may be someone, Paul, who has the genes for Huntington's Disease. Given that Huntington's disease has a 95% penetrance, whereby 95% of those with the dominant allele for Huntington's Disease develop the disease, while 5% do not, you may be justified in believing to a high degree of probability that Paul will develop Huntington's Disease. An example of a Non-Deterministic Case may be someone, Peach, who has a BRCA1 mutation. Given the medium-to-low penetrance of

BRCA1 for breast cancer, (the penetrance of BRCA1 is about 49.9% (27.5% to 72.3%) [11] for someone with moderate risk, that is, someone with 1 first-degree relative or 2 second-degree relatives on the same side of the family with breast or ovarian cancer), you may be justified in believing to a medium-to-low degree of probability that Peach will develop breast cancer.

For our purpose, Deterministic Cases are the exceptions rather than the norm. With a few relatively rare disorders such as Tay-Sachs, Huntington's Disease, PKU, Duchene muscular dystrophy, Cystic fibrosis, and so on, a single gene alternation does not in itself predict disease. In fact, even in these single gene disorders, gene–environment and gene–gene interactions can still influence whether a disease will be expressed.[12]

III.

To see how these refinements affect the debate regarding whether a patient's confidentiality should be paramount or whether we should by default share our genetic information with our family members, let us first generalize the discussions above.

As a start, we should consider the likelihood that a genetic disease under consideration is the result of a spontaneous mutation. If the genetic disease is not due to spontaneous mutation, we can next identify at least four variants of the thesis that genetic information is familial by combining the Stronger and Weaker Genetic Relations with Deterministic and Non-Deterministic Cases.

Stronger Deterministic GIFT: If two individuals, X and Y, are biologically related in a certain way, and if all you know is that X has a disease as a result of having certain genes, then you are justified in believing to a high degree of probability (high P x high Q) that Y will develop the disease. (Recall that P is the degree of probability that one individual will have certain genes, if another individual has them; and Q is the degree of probability that an individual will develop a particular disease if the individual has certain genes).

Stronger Non-Deterministic GIFT: If two individuals, X and Y, are biologically related in a certain way, if all you know is that X has a disease as a result of having certain genes, then you are justified in believing to a medium degree of probability (high P x medium-to-low Q) that Y will also develop the disease.

Weaker Deterministic GIFT: If two individuals, X and Y, are biologically related in a certain way, if all you know is that X has a disease as a result of having certain genes, then you are justified in believing to a medium degree of probability (medium-to-low P x high Q) that Y will also develop the disease.

Weaker Non-Deterministic GIFT: If two individuals, X and Y, are biologically related in a certain way, if all you know is that X has a disease as a result of having certain genes, then you are justified in believing to a medium-to-low degree of probability (medium-to-low P x medium-to-low Q) that Y will also develop the disease.

As one can see, the pressure to share genetic information with other family members is the strongest when Stronger Deterministic GIFT is operative. For example, if in virtue of knowing that John has a certain disease as a result of having certain genes you are justified in believing to a high degree of probability that Jack will also have the same disease, then it seems that you would have a stronger obligation to let Jack know about this risk. On the other hand, if weaker versions of GIFT are operative, then the pressure to share genetic information with other family members will be lower. For example, if in virtue of knowing that Paula has a certain disease as a result of having certain genes you are only justified in believing to a medium-to-low degree of probability that Pamela will also have the same disease, it seems that you would have a weaker obligation to let Pamela know about this risk.

Lest it lead to misunderstanding, let me note that the fact that there is a 'medium-to-low' degree of probability that an individual will have a particular disease does not thereby mean that this is only a 'medium-to-low' level of moral significance. A 'medium-to-low' degree of probability that an individual will have a particular disease may still be quite a morally significant matter. As I shall shortly discuss, the crucial issue for us is whether such a level of probability is sufficient to override confidentiality.

It should be clear that most clinical cases will be cases in which some weaker versions of GIFT are operative. This means that in most cases, there will be less pressure to share genetic information with other family members. To illustrate this, let us consider again the cases at the outset, which are representative of the kinds of cases that clinicians are likely to face in clinical genetic settings.[13] Owing to space, I shall describe the

Anna, Betty and Carl Case in more detail, and summarize the finding of the Fred, Gary and Heather Case.

Anna, Betty and Carl Case

Recall that Anna has a 2-year-old son, Carl, who has Duchenne's muscular dystrophy, and we are trying to decide whether Anna has a duty to inform her sister Betty who is thinking about having a child of her own. Duchenne's muscular dystrophy is one of the more penetrant genetic diseases. So this case falls under Weaker Deterministic GIFT.

Based on our previous discussions, we should consider how likely Betty's son (if Betty had a daughter, the daughter would only be a carrier and would not manifest this disease) will have Duchenne's muscular dystrophy by taking into account the following: First, as we have mentioned, 1 out of 3 cases of Duchenne's muscular dystrophy is a de novo mutation. So there is only a 2/3 chance that Carl inherited this disease from one of his parents. Secondly, to keep things simple, we can assume that Anna's husband did not have this disease, because symptoms for this disease usually appear in childhood, so if the husband had this disease, presumably Anna's sister, Betty, would have learned about this at some point. Thirdly, if Anna were a carrier, it is also possible that she acquired this disease through a spontaneous mutation. So, there is only a 2/3 chance that Anna inherited this disease from one of her parents. Fourth, if one of Anna's and Betty's parents was a carrier (we can assume that it is the mother for the same reason we assumed that Anna is the carrier), there is only a 1/2 chance that Betty is also a carrier. Finally, Duchenne's muscular dystrophy is an X-linked recessive disease. So, should Betty become pregnant, there is only a 1/4 chance that she would have a son who will

develop this disease. So a rough estimate of the probability that Betty will have a son who will develop this disease given that Carl has it is

2/3 [Anna's son inherited this disease from Anna] * 2/3 [Anna inherited this disease from her mother] * 1/2 [Betty is a carrier] * 1/4 [Betty will have a son with this disease] = 1 / 18 or 5.5%.

Fred, Garry and Heather Case

Using similar reasoning, a rough estimate of the probability that Heather's offspring will have the Huntington's Disease is

1/2 [Garry has inherited this disease from his father] * 1/2 [Heather's offspring has inherited this disease from Garry] * 95/100 [penetrance of Huntington's Disease] = 19/80 or 24%.

IV.

What can we learn from the analysis above? Admittedly, 24%, which is the higher probability of the two cases, is not an insignificant figure. But this is still a lot lower than the unrefined GIFT which gives the impression of a very high correlation between knowing that an individual has the genes for a particular genetic disease and being justified in believing/inferring that a relative will also have this particular disease. Also, given that de novo mutations of Hungtington's Disease are rare, it is likely that in this case there already exists a known family history of this disease. If so, this could possibly

render the issue of whether one should share genetic information with other family members in such a case moot.

In addition, we can see that the probability of harm in some cases of Weaker Deterministic GIFT can be as low as 5.5%, as for example in the Anna, Betty and Carl Case.

Moreover, it is worth mentioning that the kinds of harm at issue are not ones of causing harm, but rather ones of allowing harm to take place. We can all agree that with respect to causing harm, the threshold of risk of harm should be very low. Indeed, if an action of mine has even a small chance of causing you harm, then certainly I should take steps to refrain from taking such an action; or, if such an action were unavoidable, then certainly I am obligated to inform you of it. But, arguably, the situation is not symmetrical with respect to allowing harm. Indeed, those government bodies that allow confidentiality to be overridden permit this only in "exceptional circumstances," namely, if there is a risk of death or serious harm to a third party. In the US, Tarasoff case has been used as a precedent for a clinician's duty to warn in cases of immediate and serious threat.[14]

In fact, in non-genetic settings with comparable levels of risk and where an individual is not just allowing harm but could also cause harm, clinicians and government bodies have not tried to establish a duty to warn family members. For example, clinicians have not been required to inform family members that one of their family members is an alcoholic and may endanger them, e.g., by setting their house on fire. Nor have clinicians been required to acquire the names of sexual partners to whom an

individual may have transmitted a chlamydia infection so that they can warn these individuals of the danger to their partners.

Be that as it may, for our purpose, let us ask whether a 5.5% or even a 24% risk of harm is 'serious,' 'imminent,' and 'likely'? It does not seem so. It is hard to see how these probabilities constitute 'imminent' threats. Given that most clinical cases will be like these cases in that some weaker versions of GIFT will be operative, and given that the probabilities in most of these weaker versions of GIFT are not 'imminent,' it seems that most of these cases would not be exceptional enough to override a patient's confidentiality. If this is right, the idea that genetic information is familial in nature does not seem to provide a sufficient ground for why we should move towards a system in which, by default, we would share our genetic information with our relatives.

V.

Prominent bioethicists such as Mike Parker, Anneke Lucassen, and Bartha Maria Knoppers have called for the adoption of a system in which by default, genetic information is shared among family members. In this paper, I proposed that a main reason given in support of this call is the idea that genetic information is essentially familial in nature (GIFT). Upon inspecting GIFT, I showed that most genetic information are only shared in a weaker way among family members and do not necessarily lead to the actual manifestation of particular diseases. If I am right, the idea that genetic information is familial in nature does not provide a sufficient ground for why we should move towards a system in which by default, genetic information would be

shared among family members. If we should move towards such a system, it would have

to be on other grounds.

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