Accessing Genetic Information

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As technology becomes available, we are more and more able to learn about ourselves through knowledge of our genetic make-up. The information determined can be useful in correlating the occurrence of disease and disorders to specific genes or gene mutations. Individuals with a family history of disease with a known genetic correlation (e.g., Huntington's chorea) may choose to determine whether they carry the allele for this illness in order to make life choices. Others may wish to determine if they are a carrier of an illness to see if, in combination with a heterozyogtic partner, they are likely to pass the disease to a child (as is possible with Cystic Fibrosis).

The question is, who should have access to the genetic information of others? When, if at all is this ethically appropriate or necessary? Through the analysis of case studies, this paper will address the ethics of the three following scenarios in the pursuit of the human genetic load: 1) accessing the genetic information of parents; 2) determining genetic make-up of offspring; 3) the occurrence and consequence of access to an individual's genetic information by employers and insurance companies. Genetic testing and resulting information is fundamentally embedded in the context of health care and therefore will be discussed in terms of medical ethics.

Selecting a meta-ethical theory: Before we may engage in an ethically meaningful discussion we must first select an appropriate meta-ethical theory. There are three main theories from which to choose: *Ethical non-cognitivism*: This theory is founded on the notion that we are all emotionally based beings who cannot come to an agreement or disagreement because differing opinions cannot be described as right or wrong (Kluge 1999a). Even when our logic is lacking, we may insist on our opinions because they 'feel right'. This cannot account for situations where our emotional reactions are at odds with what is ethically correct. This stance is impossible to use because ethical decisions are not simply emotional assertions.

Ethical Relativism: This theory states that while ethical statements are cognitively meaningful, they do not hold in any objective state because they depend on our point of view (Kluge 1999a). Since all our judgments would be relative, we would never be able to have a disagreement with people of different perspectives Discussions would simply result in circuitous crossed-monologues. Our goal in discussing ethics is to develop solid foundations for ethical decision making applicable to all people, not simply those from one perspective.

Ethic objectivism: According to this theory, right and wrong are objective phenomena (Kluge 1999a). This allows us to make ethical judgments within the parameters of the situation regardless of the perspectives and emotions involved. We will adopt this ethical theory in our discussion of the ethics of access to genetic information.

Selecting a normative ethical approach: Within the realm of ethical objectivism, there are several ethical approaches which can be used. These include: Feminist ethics, Virtue ethics, Religiously oriented ethics, Agapistic ethics, Teleological ethics, and Deontological ethics. While there are arguments for all of these approaches, there are fundamental flaws in the context of healthcare and genetics in all but one. The two most common of this list are Teleological and Deontological ethics.

Teleological Approach: This approach focuses on outcomes rather than the means to these outcomes. Utilitarianism falls into this category. This is based on the principle of utility which promotes actions resulting in the greatest amount of good for the greatest number of people (Kluge 1999a). There are two fundamental drawbacks to this method: 1) it does not define the nature of the good we are striving for, nor the bad we are trying to avoid; 2) it does not address the needs of the individual. The very nature of the subject at hand relates directly to individuals and the consequences to them. To discuss this using the principle of utility is to deny these individuals ethical consideration which is the complete opposite of what we are trying to achieve.

Pluralistic Deontological Approach: This approach is based on the contention that there are several basic principles from which all judgments

and rules of right and wrong must ultimately be derived (Kluge 1999). The following principles are widely accepted among pluralistic deontologists:
Principle of Autonomy and Respect for Persons: Everyone has a fundamental right to self-determination. This right is only limited by unjust infringement on the rights of others.
Principle of Impossibility: A right that cannot be fulfilled is ineffective as a right, and an obligation that cannot be met under the circumstances ceases to be effective as an obligation.
Principle of Fidelity or Best Action: Whoever has an obligation also has a duty to discharge that obligation in the best manner possible.
Principle of Equality and Justice: A right is effective to the degree that it preserves or promotes justice.
Principle of Beneficence: Everyone has a duty to maximize the good of others where the nature of the good is defined by the other persons themselves.
Principle of Non-Malfeasance: Everyone has a duty to minimize harm where the nature of the harm is defined by other persons themselves.

This ethical approach has proven to be most useful and effective when discussing issues relating to personal health. Using these principles, we will come to ethical decisions regarding the access to an individual's genetic information.

Genetic access within a family - seeking parental information: Case #1: Mr. X, a forty-five year old man with a family history of colon cancer, participated in a colon cancer research study at Y University. This phase of study was aimed at determining whether there was a possible link between Z gene and an aggressive form of colon cancer. The preliminary results suggested that the Z gene was associated with a 10% increased risk of colon cancer. Mr. X did not learn whether he carried the gene and passed away from an untimely yet unrelated illness.

Shortly after Mr. X's death, his son, Mr. W. approached Y University and asked to learn whether his father carried the Z gene. The son was engaged to be married and wanted to find out all of the information he could before making a commitment to his fiancée. The University stated that the studies were preliminary and even if his father did have the gene, the risk

factor would be even less for him because there was no guarantee he had inherited the gene from his father.

Though the son claimed to understand the relative significance of his father's test results, he persisted and became increasingly stressed and irate at the University for refusing to provide the results to him.

This case brings up several ethical issues. The first deserving our attention is the issue of informed consent. Informed consent refers to the complete and appropriate disclosure to a subject or patient by the medical professional of the details and outcomes of a medical procedure or decision. To give informed consent allows the individual to decide what happens to his/her body thus maintaining his/her autonomy. When Mr. X volunteered to participate in this study, there was no indication that he assumed that anyone else would find out if he possessed the gene linked to colon cancer. Article 8.1 of the Draft Code of Ethical Conduct for Research Involving Humans states that in human genetic research, the genetics researcher must report the results to that individual. Article 8.2 goes on to say that the researchers and the Research Ethics Board (REB) presiding over the study must ensure that the results and records are protected from access by third parties unless consent is given by the participant. If we assume that the researchers at Y University adhered to this code, the onus was on Mr. X to divulge or consent to the release of any information to a third party. Mr. X did neither of these things. To disclose personal information about Mr. X to anyone (including family members) would be violating his autonomy and thus is unethical.

One may argue that Mr. X has no autonomy because he is deceased. It is true that when someone has participated in research and dies after, there are no legal obstacles to disclosing personal information because the deceased a) have no legal rights, and b) are not considered 'research subjects' under existing federal regulations (NHGRI).
Ethically, a dead being is no longer a person therefore we have no duty to them. However, individuals volunteer to participate in clinical studies with the assumption that their confidentiality does not come with an expiry date. If we allow for information to be released post-mortem, this may discourage participation in future research projects, particularly those which involve potentially socially stigmatized subjects.

The Principle of Beneficence on behalf of the researcher is also brought into question by this case. This principle states that everyone has a duty to maximize the good as defined by the individual receiving said good. In healthcare this may translate into an obligation on the part of the genetic researcher to disclose genetic information about a subject to his family so that they may take appropriate actions to prevent harm to themselves. It would seem that this principle is in violation of the Principle of Autonomy. According to the President's Commission for the study of Ethical Problems in Medicine and Biomedical and Behavioural Research, confidentiality could be overridden in certain cases where blood relatives were at risk of serious harm. In this case, the increased risk to Mr. X, if he did in fact carry the gene, was stated at approximately 10%. Because Mr. W only carries half of his father's genome which itself, may or may not carry the cancer-causing gene, the actual increased risk for Mr. W is marginal to non-existent. Because there is no significant medical threat to Mr. W in either case, the researchers were not violating the Principle of Beneficence and thus Mr. X's autonomy and confidentiality remain.

The Principle of Non-Malfeasance can be discussed similarly with the same result. In this case, the maximum possible risk to Mr. W is not such that inevitable harm will result if he is not made aware of his condition (or lack thereof). Again, Y University has no obligation do disclose the information to Mr. W.

Genetic access within a family - seeking information about our offspring: Genetic tests for offspring refer to prenatal screening, newborn genetic testing, and childhood genetic testing. Do we have the right to know our child's genetic status? For the purposes of this essay we consider both screening and diagnostic tests as methods of accessing information.

Prenatal screening: Governing aims are to reduce the incidence of congenital abnormalities for which no treatment is available, and to produce information of use in the pre- and post-natal treatment or management of disorders (Robinson 1998). Methods of determining a diagnosis included amniocentesis, chorionic villus sampling, and fetal blood sampling. Currently pre-natal treatment prevails via therapeutic abortion. There are a

number of issues to consider when looking at the ethics of pre-natal screening.

The autonomy of both the mother and the foetus both need addressing. A pregnant mother or couple ought not to be given carte blanche to make decisions where the principle beneficiary or victim is an unconsulted foetus. The Canadian Medical Association recommends that a human foetus becomes a person when "the foetal nervous system has developed to the point where it has the basic capacity for sapient cognitive awareness"(CMA 1991). This is reached at approximately twenty weeks gestation (Kluge 1999a). From this point on, the foetus is considered an incompetent individual. Its parents would naturally take on the proxy role in which case they are obligated to do what is best for their unborn child. Prior to twenty weeks, although the status of the foetus is a serious ethical consideration, as a non-person, its rights are not equal to those of the mother. Her right to autonomy and self determination take precedent.

The mother's autonomy apart from the foetus must also be considered. Pre-natal screening conveys a recommendation to pregnant women that accepting the test is the responsible course of action and that a foetus identified as seriously affected should be aborted. Failure to comply with the standard screening program may be seen as irresponsible. Parents who have an affected child may be blamed for something they could have prevented (Clarke 1998). This sort of pressure could lead to informed consent under duress denying the mother an environment where a carefully weighed and considered decision could be made. This is a violation of her autonomy.

Prenatal screening tests also present issues of equality and justice. Cases have been documented where insurance companies have refused to pay for children with illnesses that could have been 'prevented' (through abortion) with prenatal screening (Billings et. al. 1992). There may also be a negative impact on individuals of society who have a condition that prenatal screening is designed to prevent. Overall, refusing to terminate the development of a seriously genetically affected child puts that child and her family at a disadvantage beyond the usual limitations of the illness. Ultimately, more questions than answers arise when addressing these

concerns. Do parents then have a duty to a) undergo prenatal screening and b) abort a child who displays serious abnormalities?

A third consideration brings us down to the nitty-gritty reality when dealing with issues surround health care delivery - allocation of resources. The cost of avoiding a birth with Down's syndrome through serum screening and termination is $56 000 (1992). The same study reports that the average cost of lifetime care for a Down's sufferer is $280 000 (1992) (Robinson 1998). From a utilitarian point of view, we have an obligation to terminate pregnancies where aborting the feotus with serious congenital abnormalities is cheaper than the cost of treating the illness after the children are born. This is regardless of whether or not their lives might be seen as worthwhile and whether or not they would endure physical suffering.

From the deontological perspective, approaching patient treatment from a purely financial perspective denies the patient's personhood. This brings us back to the status of the foetus. Using the twenty week gestation mark of personhood, we could simply state that all abnormal pregnancies prior to twenty weeks should be aborted, yet all those detected after should be brought to term. This of course denies the gradient of severity seen in many abnormalities and also the consideration of quality of life for the infant after birth. Clearly we need to derive a more sophisticated ethical method in order to confidently and rationally come to an ethical decision making mechanism in this situation.

Newborn genetic testing: Currently in Canada, two compulsory screening programs are at work in our hospitals: The Guthrie test for phenylketonuria (PKU) and that for congenital hypothyroidism. In both of these illnesses, early diagnosis permits effective treatment of the condition. Because the benefits of screening infants for these disorders are so great, many such programs have been implemented without an explanation or information given to the parents. Parents are often unaware for what conditions their infants are being screened.

It may be argued that the obvious benefits of the test to the infant override any need for parental consent. Although parents would be irresponsible not to have the tests completed, to allow the hospital to decide in which instances parents have a choice and when they do not would set a

dangerous precedent. This sort of attitude violates the fiduciary agreement necessary between doctor and patient. The paternalistic doctor patient relationship assumes that doctor always knows best thus denying the patient's autonomy in the decision making process. Hospitals that screen newborns without seeking consent from the child's proxy (the parents) are not only acting unethically, but also illegally in that nonconsensual physical interference is assault and battery.

Whether or not genetic screening is to the advantage of the child, consent is always necessary either from the parent or proxy except under emergency conditions. This brings up the question, what do we do when parents refuse to allow this screening? In such a situation, the physician should proceed on the assumption that the child's sensible experience and qualitative perceptions are like that of an adult. It is then appropriate for the physician to take into account the child's subjective expressions and balance them against the objective standard of what a reasonable person would decide when considering the proxy's decision. If the physician finds that the proxy decision maker has introduced his/her non-standard values in making the decision, the physician must challenge the decision. The courts may ultimately decide the course of action (Kluge 1999b).

In the case of newborn screening, the benefits are so great at so little cost that it is extremely likely that if a parent refused consent, the courts would order it regardless. The point of informed consent remains important however, because to pick and choose which situations the fiduciary agreement applies and which situations it does not, may lead to unethical decision making in the future.

Childhood genetic testing: There are many issues that arise when considering childhood genetic testing depending on the nature of the test being discussed. We must first ask ourselves, do parents have a right to access the results of their child's genetic tests or even to have them tested in the first place?

Some parents have the "my child, my business!" attitude with respect to this question. This attitude is not appropriate however. A parent does not have complete access to their child's genetic information simply by virtue that they are the child's parent. In some cases, this would be in violation of

the child's autonomy. However, there are circumstances where parents do have this right. As proxy decision makers for an incompetent child, it is in the best interests of the child that her parents have access to the results of diagnostic tests/screenings to ensure that properly informed medical decisions are made.

This brings us to the issue of competency of the child. In the past, children were assumed to be incompetent simply because they were children. This is in violation of section 15 of the Charter of Rights and Freedoms which prohibits discrimination on the basis of age. If a child is deemed competent, they are under no obligation to reveal their medical status to their parents and their physicians have a duty to preserve the child's autonomy through keeping all medical information confidential. There are two other sorts of genetic tests which lie outside the immediate medical context where this may be relevant: late onset diseases with possibilities for treatment and late onset where no presymptomatic treatment is available.

If a child is at risk of developing a late onset disorder and immediate treatment is available, the ethical role of the parents is the same as when immediate medical diagnostic tests are being completed. To test an incompetent child for a late-onset disease where no presymptomatic treatment is available removes the child's future right as an autonomous adult to make their own testing decisions. Also, the confidentiality that would be automatic for an adult undergoing testing would be removed. Both of these are in violation of the child's autonomy and are therefore unethical. If a competent child takes the lead in requesting this sort of genetic test, loss of the child's future autonomy is not an issue and testing can proceed as normal.

Genetic access by employers or insurance companies: Case study #2: The ABC Genetics Testing Corporation (ABC) has developed a test to identify two gene mutations associated with breast cancer. They are BRCA1 and BRCA2 found on the long end of chromosome 17 and chromosome 13 respectively (Stanford 1998). Although the figures are in some dispute, it appears that the possibility of contracting breast cancer before the age of sixty increases by 70-85% in women carrying both of these mutations.

ABC already markets the gene test to the public. MicroHard, a leading computer software manufacturer employs over 10 000 women nationwide

between the ages of 25 and 60. MicroHard proposes to have every female employee take the ABC gene test at its expense. MicroHard hopes that the use of the test will enable its female employees to have a better understanding of breast cancer and to take appropriate responses, including more frequent physical examinations and more aggressive treatment of any indications of potential cancer. MicroHard also believes that if environment contributes to the onset of cancer, female employees can modify their behaviour to reduce environmental risks, such as modifying their diet and avoiding sources of radon. While MicroHard is concerned about the physical well-being of its employees, it frankly admits that if it could lower the incidence of breast cancer, it would lower its medical costs. MicroHard is a self-insurer meaning that it pays its employees' medical expenses and does not use a third party insurer.

The first ethical question that springs to mind is how the autonomy of the employees would be preserved if these tests were to take place. The answer is, it would not. To seek this sort of information from an individual without her competent informed consent would be a direct violation of her privacy and confidentiality. In this situation, the competency of the employee may be compromised due to fears of being ostracized by management or even losing her job if she does not consent to the test.

Before we discuss further ethical implications here, it would be helpful to determine MicroHard's true motive behind this venture. It could be argued that MicroHard truly wants the best for its female employees and is just looking out for their health and well-being, irrespective of financial issues. If the tests will serve only to 'inform' the employee and allow her to use the information to make lifestyle changes so as to minimize the risk of developing breast cancer, then to ensure their employee's autonomy MicroHard could do two things.

1. Pay for the tests but maintain the confidentiality of the employees by never seeking the results themselves. This would allow the women to use the information if they so chose.
2. Forget the test altogether and spend the money informing all employees (men and women) of cancer-preventing lifestyle choices, the importance of self examination and frequent check-ups. This

route would avoid the ethical issues of consenting to the test and those of the company having access to this type of knowledge. MicroHard did not select either of these routes which leads us to believe that Micro Hard's true motive is to minimize cost. Certainly, helping their at-risk employees avoid cancer would achieve this goal. But wouldn't they save even more money if they reduced their number of at-risk employees? To fire someone simply based simply on their genotype is genetic discrimination and is certainly a violation of the Principle of Equality and Justice.

Genetic discrimination refers to discrimination directed against an individual or family based solely on an apparent or perceived genetic variation from the 'normal' human genotype. If MicroHard decided to use the genetic information of its employees when making hiring/promoting/firing decisions it would be guilty of this ethical infraction. If this did occur, Micro Hard would certainly not be the first company/institution to do so.

In such a case, a man was denied a job with the government (U.S.) because he was a carrier of Gaucher Disease (Billings et. al. 1992). Insurance companies in the US are also guilty of this discrimination (see Appendix A for reported cases of genetic discrimination). In many of these cases, decisions regarding allocations of health, life, and automobile insurance were based solely on a diagnostic label without regard to the severity of the condition for each individual. In these and other cases, having a particular genotype is equated with the presence of a severe illness and the lack of effective treatments. This demonstrates a lack of understanding of the concepts of incomplete genetic penetrance, variable expressivity, and genetic heterogeneity. In many cases, the worst possible scenario seems to be the standard used for policy decisions regarding at risk individuals. An individual may suffer severe consequences as a result of this inaccurate and unfair simplification of genetic conditions (Billings et. al. 1992).

Because MicroHard is in effect, both an employer and an insurer, they have twice the interest in the genetic health of their employees. While both companies and private insurers focus on increased production at lower cost, to attain these goals at the cost of the basic rights of their employees is

ethically questionable and could lead to yet another stratification in our social structure: a class of individuals unable to obtain employment or insurance.

Access to Genetic information - Conclusions: There is no doubt that the new technologies in medical genetics offer many advantages and improvements to the way we address our present and future health. As individuals, we have the ability to gain knowledge about our genetic health and the power to act with this information in any way we choose. Our case studies have shown that we, as individuals or companies, do not ethically have free access to genetic information of others. Even as parents, our only power over our children's genetic information lies in our roles as their proxy decision makers.

Although individually we may do what we please with the knowledge of our own genetic status, legally we still have no power over who else has access to our information. Currently in Canada, there are no laws regulating the use of genetic information, and there are no enforceable mechanisms regulating who may see these results. We may assume that our records are confidential, and ethically they should be; however, we must accept that this cannot be guaranteed. In the US this is a particular concern since insurance companies and some employers routinely access medical records where this information would lie. Ethics aside, until laws and infrastructure are in place, we must be careful in considering whether or not to make our genetic status known, even to ourselves.

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