To warn or not to warn – Do doctors ever have a moral obligation to warn their patient's family members about hereditary disease risks?

This paper addresses the question of whether doctors ever have a moral duty to warn a patient's family of possible risks of hereditary diseases. In answering this question the value of the information gained by genetic testing, and its ability to prevent or alleviate hereditary disease, must be considered. It will first look at the nature and background of genetic testing before going on to identify the relevant moral and technical issues, using legal cases as examples, discussing them in light of ethical principles and concluding that doctor – patient confidentiality should generally not be breached. Often there are alternative means for the patient's relatives to learn of the genetic predisposition or the knowledge hereto does not significantly alter the outcome. The information gained by testing one individual must remain the privilege of this individual and the respect for privacy and self-disclosure must be maintained. It will emphasise the inalienable need for counselling in genetic testing and discuss some of the conflicts of interests in this matter. A suggestion will be made that post-test counselling should be mandatory for as long as the physician deems appropriate. Finally, it will be argued that doctors have a duty to warn their patient's family if the patient has asked them to warn a family member whom they are not comfortable warning themselves or when genetic disorders are found in children, the parents of a child-patient must be advised.

Genetic testing

The genome of every person (except for identical twins) is unique[i] and genetic testing has now become a widely used tool for paternity issues, forensic matters, and is also used in a medical context. This paper will be limited to the latter.

The link between chromosomes and certain diseases was first described in 1902. But only the development of gene sequencing by scientists like A. Maxam, W. Gilbert and F. Sanger in the mid 1970s[ii] and the discovery of the polymerase chain reaction (PCR) in 1983, [iii] combined with the mapping of the human genome in 2003 (Human Genome Project 1990 – 2003)[iv], made it possible for genetic testing to be used as a dignostic tool for testing for genetic mutations in certain diseases.

Where some genetic abnormalities invariably lead to symptomatic individuals, as in Trisomy 21 (Downs Syndrom) or XXY syndrom, the link between mutations and disease is not as clear in other illnesses. Geneticists are exploring possible correlations between a large variety of maladies and genetic mutations, but with varying success. Mutations at given loci have repeatedly been observed in patients with cancer for instance, but these mutations have also been observed in asymptomatic individuals. Whereas some symptoms can be directly attributed to one particular locus, others are attributed to several loci. Genetic mutations "are not our destiny"[v] i.e. they are not the sole cause of complicated diseases like cancer, diabetes or heart disease.

As Dawn Allain, a genetic counsellor, cited in Health Link (website of the Medical College of Wisconsin) explains; "Our technology and knowledge is still limited. We can identify some, but not necessarily all, genes identified with a particular disease."[vi] Tests vary in sensitivity and can provide a false positive or false negative result. The interpretation of data is difficult, even for physicians, and requires specialized training. [vii] Psychologist G. Gigerenzer, director of the Max Planck Institute

for human development, Berlin points out that most people, even trained scientists and physicians, have difficulty in mentally juggling statistical data, arriving at false conclusions,.[viii] M Rugnetta et al. writes that "the results of genetic testing today are hardly reliable, yet they are already part of the medical marketplace.[ix]

Types of Genetic Testing

Firstly we must differentiate between genetic testing and genetic screening. The former refers to the testing of individuals whereas the later refers to mandatory testing of groups of a specific background. There are several different types of genetic testing available.

Carrier Identification may be used by couples considering having children, who know of recessive genetic disorders in their families. Cystic fibrosis, Tay-Sachs disease and sickel-cell anaemia are examples of these.

Prenatal Diagnosis is used to screen the foetus for congenital abnormalities leading to physical or mental abnormalities. Amniocentesis screening for Downs Syndrom is the most common of this type. Sex selection testing is possible also, but often illegal.

Newborn Screening is mandatory in many countries, including Australia, for diseases such as phenylketonuria or congenital hypothyroidism. It is seen as a preventative health measure as early intervetion can prevent the symptoms of these conditions and allow the individuals to influence the outcome of their genetic disorder.
Late-onset Disorders are often complex and involve an interaction of environmental factors and genetic mutations. Testing may indicate an increased susceptibility to diseases such as cancer or heart disease. Huntington's disease (HD) however is an example of a late-onset disease caused by a single gene mutation.[x] The outcome of HD can not be influenced by early diagnosis.

Issues surrounding Genetic Testing

Genetic testing may have several serious implications for the persons concerned. Whilst the benefits of **newborn screening** seem to be only positive - preventing the effects of potentially detrimental genetic disorders - **prenatal diagnosis** could lead parents to abort the pregnancy. It can allow parents to choose not to have a disabled child and to terminate a pregnancy, but it can also assure worried parents that their child is free of the genetic disorders tested for. At the very least it will allow them to prepare for what lies ahead. It is also possible to select offspring for gender, although illegal in many countries. **Carrier identification**, combined with genetic counselling, will allow prospective parents to make informed choices, appease their fears or allow them to consider alternatives to conventional pregnancy such as donor sperm or oocytes, or adoption. Usually, families who come for carrier identification are aware of a hereditary disease in their family, as would most other family members, henceforth, all members of the family can themselves make a choice about the need for testing or genetic counselling. There appears no need for a physician to breach the patient's confidentiality and inform other members of the family. Conflict could arise, if one partner of a couple considering having children comes for testing and does not want to inform the other partner of a negative outcome. **Testing for late onset disorders** usually also involves testing for a disease that is part of the family history. The correlation of the genetic mutation and the

onset of the disease however is, except in the case of Huntington's disease, much less clear. A positive result can indicate an increases susceptibility to diseases such as cancer, diabetis or heart disease, but is not a diagnostic tool. In the case of cancer, only 5-10 % of all cancers are hereditary.[xi]

As suggested, there may be moral and ethical issues in almost all forms of genetic testing. This paper however, shall primarily focus on the implications of testing for late onset disorders as this is where most ethical questions arrise and the arguments brought forth can also be applied to carrier testing.

Problems highlighted by the US - National Institute of Health regarding testing for late onset disorders are;

* *No effective interventions are yet available to improve the outcome of most inherited diseases.*
* *Negative (normal) test results might not rule out future occurrence of disease.*
* *Positive test results might not mean the disease will inevitably develop.*[xii]

In genetic testing there several ethical issues need to be considered. Firstly, the individual must give **informed consent.** Secondly, there are issues regarding the person's **right to privacy.** Thirdly, **confidentiality** must be maintained by limiting access to the information and fourthly, the patient's **autonomy** must be respected.

Informed consent

The definition of informed consent given by Ralph Warner, an American lawyer, is “an agreement to do something or to allow something to happen, made with complete knowledge of all relevant facts, such as the risks involved or any available alternatives. “[xiii] “Complete knowlege” entails that the person giving consent has understood all relevant facts. The signing of a “consent form” does not suffice, as it gives no indication whether the individual has understood the content of the form or the issues in question.

As in many areas of medical practice the subject of informed consent is difficult to judge for various reasons. Firstly, there is the problem of predicting emotional reactions to the outcome of a genetic test in advance. A person might initially feel that they would inform their family of an adverse outcome, but when faced with the actual results decide that they did not want to burden their relatives, after all.

Secondly, the patient's state of mind may prevent them from understanding the meaning of the words they are hearing. The emotional issues surrounding an adverse outcome will be explored further below. In light of the law suits described hereafter, it has been suggested that a kind of genetic 'Miranda' warning could be employed i.e. that patients should be advised that in case of particular genetic mutations the information would be disclosed to other interested parties, in order to clarify the situation.

To warn or not to warn – Do doctors ever have a moral obligation to warn their patient's family members about hereditary disease risks?

Right to privacy

The boundaries of what is considered private vary between cultures and individuals. Generally it refers to the individual's rights to reveal themselves and information pertaining to themselves, selectively.
Spearheaded by the September 11, 2001 destruction of the US World Trade Center, civil liberties and the public's right to privacy has consistently been eroded on a worldwide scale. Mandatory breach of the patient – doctor relationship, e. g. disclosing genetic information to other interested parties, might be seen as further eroding this right. Breach of the patient – doctor confidentiality can lead to intimate personal information to be accessed by interested parties such as insurance companies or employers. This in turn, could lead to discrimination on the basis of genetic profiles. The patient's right to privacy must be balanced however, with the family's right of having access to vital knowledge concerning themselves. A genetic counsellor's role would be to point this out to the patient.

Confidentiality

This is an important part of privacy. The information disclosed by a patient or gained in treatment of a patient may not be disclosed to a third party. The patient – doctor relationship has long been regarded as a privileged relationship and many countries have penalties for its breach.

Any legislation, forcing physicians to disclose information gained from patients or in the course of treatment, could lead to mistrust towards the medical profession. It may result in patients becoming reluctant to seek medical help for fear of the information gained being used against them. A 'Miranda warning' would appear to contribute little to appease this fear. Quite often, the full implications of a duty to disclose this information would only become apparent to the patient much later then the initial test, leaving the patient feeling betrayed.

Autonomy

Our democratic system demands that we respect the individual's autonomy. That is, their right to make their own decisions. It also pertains to the right not to know ones own future if one so wishes. If physicians took the liberty of informing relatives of their patients or were forced to do so, this would clearly interfere with this right, if it is against the wishes of the patient.

Technical Issues

As mentioned earlier, one intrinsic problem with testing for late-onset disorders is that the link between the genetic mutation and complex diseases like cancer, diabetes and heart-disease are far from clear. One of the most common errors is statistical misinterpretation.

Interpreting the Information

Statistical information of often very difficult to understand. An article by Germany scientist and director of the Max Planck Institute for Human Development G. Giegerenza gives an example of

the German Health Ministry's plan to introduce mass mammography for women over the age of 50. The Berlin Medical Council was opposed to this idea and pointed out that the costs would not warrant the benefits and that it would put many women at risk of a false diagnosis. The Ministry labled this attitude close to irresponsible and claimed that the tests would save 25% of the women tested. The statistical information had been grossly misunderstood. The absolute benefit of mammogram screening is in fact only 0.1% for the total population, in this case. Over a period of 10 years, out of a group of 1000 women, 4 will die of breast cancer. With mammography, only 3 would die. Hence out of these 4 women, 25% will be 'saved' using mammography. Over the course of these ten years however, many women will receive a false positive result and will suffer anguish and needless fear.[xiv] The German authorities had misinterpreted the benefits. Australia does offer mass screening but campaigns never mentioned that the benefit is only to one in 1000 women. The potential benefits have been misrepresented, presumably due to poor understanding of the statistical information. A topical example is the massive surge of breast cancer screening of young women who had no family risk of breast cancer after popular singer Kylie Minogue announced she suffered the disease. Thousands of unnecessary biopsies and imaging procedures were carried out without a significant improvement of breast cancer detection.[xv] Researchers are still struggling to come to terms with in interpretation of their findings. Euhus at al points out that "the predictive value of a test (e.g., the BRCAPRO computer model or the subjective estimate of a risk counselor) is related not only to the sensitivity and specificity of the test but also to the prevalence of the condition of interest (in this case, BRCA gene mutations) being evaluated." and insists that "given the current state of the art, the value of an experienced genetic risk counselor who understands the limitations of the computer-based probability models cannot be overstated."[xvi]

The sensitivity of the tests

Tests like those used for BRCAPRO are considered highly accurate, but still miss 15% of mutation of BRCA1 and BRCA2[xvii], the gene considered responsible for an increases susceptibility to breast cancer, ovarian cancer and prostate cancer. In other words, out of out of 100 women who carrie mutation and have testing, only 85 will receive a positive result. 15 will go home thinking that they do not have this genetic mutation. This is called a false negative result. Similarly tests sometimes indicate a positive results when no mutation is present. Hence, upon receiving a positive result further testing should be carried out.

The value of the result

A negative test result for a mutation like the BRCA gene does not mean that the individual is not susceptible to this disease but only that their susceptibility is that of the general population. Furthermore, the test may simply not have worked or the mutation may be on another gene. Similarely, a positive test result is not a diagnosis of cancer. It merely indicated that regular screening tests for cancer may be warranted. If a positive result is not positive and a negative result is not negative it could be argued that the value of this type of test is very limited indeed but a closer examination of their value would go far beyond the scope of this paper. As genetic tests are expensive and it is important to have a good reason for doing them.

To warn or not to warn – Do doctors ever have a moral obligation to warn their patient's family members about hereditary disease risks?

Emotional issues - the burden of knowledge

The correlation between a late-onset disorder like Huntington's disease (HD) and a genetic defect is very clear - a mutation on the 4th chromosome. This illness affects approximately 3-7 people in 100,000 of European ancestry. The incidence in other populations such as African or Asian being somewhat lower.[xviii] In Huntington's disease an ancestor suffering the disorder is usually know within the family. The child of a parent with Huntington's disease has a 50/50 chance of contracting the disease. Here, genetic testing can be seen as a diagnostic method. In what way, if at all, do individuals who carry the genetic mutations benefit from early diagnosis? At present there is no cure for HD or a means of slowing its progress.[xix] There may be some advantage in early diagnosis in that a patient with early symptoms might be spared the burden of false diagnosis or treatment of early symptoms but one burden is without a doubt, that the patient has a greater vested interested in a positive outcome i.e. a negative result of the test, as a positive result means inadvertently a bleak future. Here, the test is a dignostic tool and rules out all hope of a cure. The psychological impact of this knowledge is difficult to predict and requires close monitoring and, one would assume, intense counselling.

In other illnesses tested for, the situation is quite different. In Australia for example, around 70 of 100 deaths are attributed to coronary heart disease and cancer,[xx] but as mentioned earlier, the causative factors for these disorders are mult-factorial and genetic mutations give at best an indication of a predisposition to the condition. Some doctors and patients might claim that upon knowing that a particular genetic mutation is present the condition suggested by the mutation can be more closely monitored for. In answer to this, on might suggest that if the absence of this gene does not mean the individual will not suffer the condition there lies a danger that the individual or their medical practitioner might then dismiss a perceived abnormality as insignificant in light of a negative test result.

However strongly a person might feel beforehand that they will be able to cope with an adverse outcome, having the actual knowledge that a genetic mutation suspected of causing an increased susceptibility to a potentially fatal disease is present, affects different people in different ways. There appear to have been no studies carried out that indicate in what way the lives of the individuals were changed after testing. For some patients, knowing that their lives may be shortened might persivably be as devastating as a diagnosis of the disease. It appears that adequate post-test counselling should be mandatory and followed up after a period of several months, allowing the patient to process the information.

Legal Implications

Offit at al report four legal cases in the United States contending the physician's “duty to warn” a patient's family members of inherited health risks. All three cases weighed the patient's right to privacy against the physician's duty to warn. In the case *Tarasoff v the Regents of the University of California,* where a person was killed by a psychiatric patient who had announced his intention to the psychotherapist and had not been forewarned by the psychotherapist, the Supreme Court ruled that a physician must warn where the patient poses a “serious and imminent threat” to that party.[xxi] In genetic diseases however, the patient is not the agent of the harm to the third party, nor is there any iminent harm.[xxii]

In a later case, *Pate v Threlkel*, Ms Heidi Pate claimed that her cancer could have been prevented had her mother's physician warned her of the risk of thyroid cancer. Although in 1994, the district court held that the physician had no legal duty to warn a patient's family[xxiii], the judgement was overturned in 1995 by the Supreme court, which held that the physician had a duty to warn the patient to advise their family of hereditable disease risks.[xxiv] This could be seen as a judgement in line with the American culture of victimizing the consumer of a service or of goods, absolving them of their responsibilities. The question arises whether Heidi Pate did not have ample opportunity to inform herself about the nature of her mother's illness and of possible risks to her own health without there being a need for her mother's physician to point this out to her. Furthermore, the information the author of this paper was able to access did not include information as to whether Heidi Pate did in fact have a genetic mutation or whether she contracted cancer as part of the normal susceptibility of a population to this form of cancer. It seems unlikely, that Heidi Pate had not visited a doctor in the three years between her mother's diagnosis and her own. A standard anamnesis includes the question whether there are any known disorders in the family - such as heart disease, cancer or diabetis. Hereby, her own doctor who, unlike Dr. Threlkel, did have a patient – doctor contract with Ms Pate should have alerted her to the increased risk factor.

This also holds true for the third case mentioned in the paper *Safer v Estate of Pack*. The plaintiff in this case claimed that her cancer could have been prevented had the late Dr. Pack informed her of the increased risk to adenomatous polyposis. During the course of the trail it became aparent that she had in fact been screened for the ailment as a child. The court ruled that physicians must take "reasonable steps" to guarantee that immediate family members are warned, failing to provide what such 'reasonable steps' would be defined as. The judgment appears somewhat hypothetical however, as it showed that informing the patient had sufficed for the daughter to be screened. That this did nothing to prevent the illness, showed that other measures would have had to be taken, but notifying the child could not have helped to improve the outcome.

Discussion

An intrinsic value in any democratic society is respect for the individual. Apart from a right to privacy, this also includes their autonomy. Although some members of these societies, like police officers, appear to act paternalistically they however merely enforce the rules society has placed upon itself. Whenever specialized knowledge is required, a quality of paternalistic behaviour is intrinsic to the nature of a professional's position. In an effort to minimize harm and maximize good, an architect is expected to act paternalistically if they believe that materials chosen by the builder are not suitable for the purpose or that the building requires additional supports. A physician when prescribing medication or ordering tests, must act paternalistically, also, which however does not mean that he may not consider the patient's wishes. It however, goes beyond the responsibility of an architect or a physician to be responsible for building materials or drugs that are generally in use. Their sphere of influence is limited to their clients, who, as autonomous individuals can choose to go elsewhere. How then or why, do we propose to hold a physician liable for matters that are beyond their sphere of influence? The patient-doctor relationship requires the physician to maintain confidentiality towards his client and individuals seeking a physician's advise should reasonably be able to expect this to be maintained. In aforementioned case of Tarsoff v the Regents of the

University of California dealt with a psychotherapist failing to warn a third party of his patient's intention to kill them. In this case however, it could be argued that the physician's duty toward his patient required them to warn the third party. As argued afore, a physician's relationship with their patient requires a form of paternalism. A paternalistic duty is to limit choices for the other's good. It could not have been in the patient's best interest if they were allowed to kill the third party as this would a) have results which could be termed negative, such as loss of liberties by a jail sentence b) it would also decrease the happiness of many to increase the happiness of one. Hence, paternalistic duty, would have had to include whatever was necessary to prevent the patient killing his victim. And although seen as a precedent-setting case for other cases involving the breach of confidentiality, in this case poorly compares to matters involved in genetic testing.

The privilege of doctor – patient confidentiality has been respected by many generations. It enables individuals to seek help without fear of recrimination. In the case of a very young patient, the guardian has the duty to act paternalistically and in order to fulfill this duty the guardian must be privileged to all relevant information pertaining to the patient. Paternalistic duty entails the corresponding right of the individual to have choices made in their best interest. It can however not be in the patient's best interest to disclose information to third parties without explicit consent. Even in the case of a child or a patient who is not of sound mind, the right to privacy must be respected.

Whatever genetic disposition, the individual has no influence on the genetic makeup of another who has already been born. Any persons alive today can themselves be tested. Those in contact with the proband may be made aware of increased indication for testing but could also prefer not to know. It is more likely that the patient has knowledge of their relative's preference and likely reaction then the doctor. More distant relatives, whether by geography or genetically, can be privy to the information but they also have the option of themselves being tested.

A study conducted by K. Kohut at al. Indicated that in general patients who had been counselled and screened for HNPCC (hereditary non-polyposis colorectal cancer) generally understood the implications for their family and felt that it was their duty to inform relatives of the increased risk.[xxv] There were some instances where patients chose not to inform relatives. While reasons such as the relative being elderly or very young appear valid, others, such as 'living at a distance' may require some counselling to overcome. Those who identified relatives whom they would not be able to tell also specified reasons. It appears however, that if a patient had personal difficulty the doctor or genetic counsellor has a duty to assist the patient by informing 'difficult' relatives.

Conclusion

The rights of the patient should always be respected allowing them to decide who to make privy to this information. Doctors must be allowed to maintain patient – doctor confidentiality unless acting paternalistically for a patient not of sound mind or sufficient maturity. The counsellor or physician's duty also includes making the patient aware of the rights of other members of the family and their duties towards them. When asked by a patient to provide assistance in warning relatives about hereditary disease, doctors have a duty to assist and an obligation to warn the patient's family members about hereditary disease risks.

i Wikipedia. http://en.wikipedia.org/wiki/Human_Genome_Project. retrieved 01/06/2008
ii Wikipedia. http://en.wikipedia.org/wiki/DNA_sequencing retrieved 01/06/2008
iii Fuller R. (2007) The history of genetic testing. *Ezine articles* http://ezinearticles.com/?The-History-of-Genetic-Testing&id=438498&opt=print retrieved 02/06/2008
iv Wikipedia http://en.wikipedia.org/wiki/Human_Genome_Project retrieved 01/06/2008
v Rugnetta M, Russel J, Mureno J. (2008) Genetic non-discrimination: Policy considerations in the age of genetic medicine. http://www.americanprogress.org/issues/2008/04/genetic_nondiscrimination.html
vi Genetic testing offers information and dilemmas (2002) Medical College of Wisconsin URL: http://healthlink.mcw.edu/article/972930878.html retrieved 01/06/2008
vii Berkeley Lab. What is genetic testing? http://www.lbl.gov/Education/ELSI/Frames/genetic-testing-f.html retrieved 01/06/2008
viii Gigerenzer G. (2004) Playing with numbers – illusions of certainty. *Max Planck Research* 1/2004
ix Rugnetta M, Russel J, Mureno J. (2008) Genetic non-discrimination: Policy considerations in the age of genetic medicine. http://www.americanprogress.org/issues/2008/04/genetic_nondiscrimination.html
x Berkeley Lab. What is genetic testing? http://www.lbl.gov/Education/ELSI/Frames/genetic-testing-f.html retrieved 01/06/2008
xi Genetic testing offers information and dilemmas. (2002) Medical College of Wisconsin URL: http://healthlink.mcw.edu/article/972930878.html retrieved 01/06/2008
xii Genetic testing offers information and dilemmas. (2002) Medical College of Wisconsin URL: http://healthlink.mcw.edu/article/972930878.html retrieved 01/06/2008
xiii Warner R. Glossary. *NOLO*,http://www.nolo.com/definition.cfm/Term/12E86DD0-1F7A-411E-B6DB9A9F6C7C07EF/alpha/I/ retrieved 01/06/2008
xivGigerenzer G. (2004). Playing with numbers – illusions of certainty. *Max Planck Research* 1/2004
xv CBC News. 2008. Kylie Minouge's breast cancer leads to screening spike in young women: study. http://www.cbc.ca/health/story/2008/06/04/minogue-cancer.html retrieved 13/06/2008
xvi Euhus EM, Esserman L, Ganz PA, Mills GB, Tomlinson G. (2002) RESPONSE: Re: Pretest Prediction of BRCA1 or BRCA2 Mutation by Risk Counselors and the Computer Model BRCAPRO. *Journal of the National Cancer Institute*, Vol. 94, No. 20, 1583-1584
xviiBerri DA, Iversen ES, Gudbjartsson DF, Hiller EH, Garber JE, Peshkin BN, Lerman C, Watson P, Lynch HT, Hilsenbeck SG, Rubinstein WS, Huges KS, Parmigiani G.(2007) John Hopkins University Server http://astor.som.jhmi.edu/brcapro/301validation.doc retrieved 13/06/2008
xviiieMedTV. Huntington's disease statistics. http://nervous-system.emedtv.com/huntington's-disease/huntington's-disease-statistics.html retrieved 01/06/2008
xix eMedTV. Huntington's disease statistics.http://nervous-system.emedtv.com/huntington's-disease/treatment-for-huntington's-disease.html retrieved 01/06/2008
xx Charles Sturt University. NSW HSC online. http://www.hsc.csu.edu.au/pdhpe/core1/improving/3-2/3-2-3/3.2.3_injury.html retrieved 01/06/2008
xxi Offit K, Groeger E, Turner S, Wasworth EA, Weiser MA. The “duty to warn” a patient's family members about hereditary diease risks. *The Journal of the American Medical Association* (2005); 292 (12): 1469-1473
xxiiSulmasy D. Duty to Warn About Hereditary Disease Risks. *The Journal of the American Medical Association* (2005); 293 (6): 676
xxiiiFlorida. District Court of Appeal. First District. Pate v Threlkel. Wests South Report. (1994) Aug 1;640:183-6
xxivOffit K, Groeger E, Turner S, Wasworth EA, Weiser MA. The “duty to warn” a patient's family members about hereditary diease risks. *The Journal of the American Medical Association* (2005); 292 (12): 1469-1473
xxvKohut K, Manno M, Gallinger S, Esplen MJ. Should helathcare providers have a duty to warn family members of individuals with an HNPCC-causing mutation? A survey of patients from the Ontario Familial colon Cancer Registry. *J Med Genet* 2007; 44; 404-407