Informational autonomy and the preservation of free choice in genetic testing: The Cyprus Case

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Summary of Thesis:

This thesis aims to explore informational autonomy by demonstrating the correlation between genetic testing and informational autonomy, focusing on the preservation of the free choice option with special reference to thalassemia in Cyprus. This exploration does not intend to deliver a groundbreaking and bullet-proof new concept of informational autonomy that should be used without exception in every application of genetic testing, but rather to constitute the bedrock for reinstating the Cyprus strategy on genetic testing for thalassemia trait: it is a specific case study.

This thesis makes a case for protecting informational autonomy, the ability for people to make their own responsible decisions, and argues that Cyprus strategy on thalassemia – as the empirical work demonstrates – is an example where the option of free choice is limited.

The Cyprus Thalassemia Programme is recognised as being unique in the world for its success in almost completely eliminating new cases of thalassemia within 15 years. Along with premarital testing, genetic screening is mandatory for a couple wishing to get married in a Christian Orthodox Church. In the case of premarital testing, the option for couples to know or not know if they are the thalassaemic trait carriers is not considered at all.

The challenge for Cyprus in the 21st Century is to advance the existing control programme while respecting freedom. By introducing pre-natal diagnosis along with premarital screening, Cyprus succeeded in eliminating the number of births with thalassemia, but increasing the number of abortions at the same time. People in Cyprus must have the option to know or not know if they are thalassemia trait carriers and should not be "forced" to make a premarital testing. Informational autonomy is a right that Cypriots are allowed to have. Human genetic information is ultimately not about genes; it is about people.

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CHAPTER 1 - INTRODUCTION

This thesis aims to explore informational autonomy, by demonstrating the correlation between genetic testing and informational autonomy, focusing on the preservation of the free choice option with special reference to thalassemia in Cyprus.

My thesis is divided in two parts. The first part which is mainly theoretical covers all the issues presented in the literature which are strongly connected with genetic testing. In order to understand and elaborate the case of genetic testing in Cyprus I consider it useful to analyze some of the issues surrounding genetic testing, such as informed consent or "the right to know", focusing on informational autonomy.

Therefore, as Cyprus strategy is based on genetic testing, the theoretical part constitutes the basis for the second part which presents Cyprus case concerning thalassaemia, an example of how several issues discussed in the theoretical part of my thesis, can be applied in a real life situation.

Specifically, the empirical study based on discussion groups investigates how and in what degree people in Cyprus are influenced nowadays from Cyprus strategy on thalassaemia and whether the establishment of premarital testing as the national public policy for thalassemia major affects their informational autonomy and the right to know or not to know genetic information.

The main objective of this thesis is to clarify some of the mystification surrounding informational autonomy by combining the theoretical with the empirical part and demonstrate a way of basing genetic testing on informational autonomy and more specifically on the preservation of the option of a free choice.

This exploration does not intend to deliver a groundbreaking and bullet – proof new concept of informational autonomy that should be used without exception in every application of genetic

testing, but rather to constitute the bedrock for reinstating the Cyprus strategy on genetic testing for thalassemia: it is a specific case study.

A stress on autonomy positively contributes to put the individual and ones rights, at the centre of the ethical reflection. At the same time, it simplifies the scope of ethical reflection because ethical analysis and decisions becomes a reserved domain of individuals in their singularity. It also presupposes ones ability to make decisions individually and the availability of all information which are necessary to choose.

Autonomy is derived from the Greek words autos (self) and nomos (law or rule). This term was originally a political term, but now extends to the personal level. Personal autonomy is "*self rule that is free from both controlling influence by others and from limitations, such as inadequate understanding, that is present in meaningful choice*"[1].

Therefore, acting voluntarily is the action of a will not controlled by another influence. Such an influence is usually construed to be that of another person or people. In regard to the maxim it could mean society, friends or family.

Informed consent can only occur through autonomy. The practice of acquiring informed consent is rooted in the post-World War II Nuremberg Trials. A new standard of ethical medical behavior was established at that time as well as the concept of voluntary informed consent.

A crucial component of informed consent is that the person signing it is competent or able to make a rational decision and meaningfully give consent. This situation gets more complicated when people are unable to understand what has been explained or are unable to make a decision. Despite the fact that it is necessary to present a procedure or a treatment, informing patients about the risks of treatment might scare them into refusing it when the risks of non-treatment are even greater.

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[1] Beauchamp, T.L., and J.F. Childress, *Principles of Biomedical Ethics*, Oxford: Oxford University Press, 2001, p.57–112.

However, the informed consent process is vital as it can be empowering to patients to understand that they play an important role in their own treatment and it may encourage future participants, when it comes to genetic research, to know their options well enough to make the best decisions for them. Generally speaking, the informed consent process shifts the responsibility to individuals, increasing at the same time their self-confidence and autonomy.

As the mysteries of the human genetic code are unravelled, people need protection so that breakthroughs are used to treat and heal, not to isolate and discriminate. Many people do not know how to protect their genetic information, nor do they even realize that they have a right to do so. Hence, this thesis makes a case for protecting informational autonomy, the ability for people to make their own responsible decisions and argues that Cyprus strategy on thalassemia - as the empirical work demonstrates - is an example where the option of free choice is limited.

The Cyprus case was chosen because of the Cypriot Government's prevention programme for thalassemia which combined health education and community involvement, genetic counselling but most important population genetic screening. This prevention programme has as an aim to limit the number of affected births.

The Cyprus Thalassemia Program is recognized as being unique in the world for its success in almost completely eliminating new cases of thalassemia within 15 years. Additionally, along with premarital testing, genetic screening is mandatory for a couple wishing to get married in a Christian Orthodox Church. Therefore, in the case of premarital testing the option for couples to know or not know the specific genetic information (if they are thalassaemic trait carriers), is not considered at all. However, if they do not wish to possess this information, then they can always choose a civil wedding instead – a case which in Cyprus is not preferred, as religion is very strong.

To convince readers that Cyprus policy on thalassemia needs to be revised the present thesis uses the following path, which consists of eight chapters including this introduction and conclusion. Although some issues are dealt with throughout several chapters, the thesis has attempted to follow

a logical and structured path for suggesting that the foundations of genetic testing - especially the "right to know or not know" genetic information - is neglected in Cyprus. Therefore, it is important for Cypriots not only to preserve their informational autonomy but to be let to make their own decisions.

Following this short introduction, some essential information is provided, in the first section of Chapter 2, so as to understand the nature of genetic testing and genetic screening. The next section addresses the clashing interests different stakeholders might have with respect to genetic information coming from genetic testing. This leads us to the question whether genetic information is different from other kind of information and if it can be considered as exceptional.

Since genetic testing and screening belong to an area which not many people are familiar with, and this is the reason why often people tend to mystify genetic information, a brief reference concerning the meaning of genetic information is provided. Chapter 2 ends with the conclusion that genetic information needs protection, the kind of protection which is based on trust. This trust has to be tested first, in order to be gained. This demands very strong foundations concerning people's understanding on what they are consenting for when it comes to genetic information.

Chapter 3 introduces the discussion which follows further in my thesis concerning informed consent, autonomy and the "right to know" as it provides an overview of several ethical theories concerning the acquisition of genetic information. This chapter presents the utilitarian approach focusing on the dichotomy of act and rule utilitarianism, Kantianism – specifically the categorical imperative and finally paternalism. Paternalism is used as a starting point to refer to Mill's liberalism and Onora O'Neill's interpretation of Kantian categorical imperative as principled autonomy.

Chapter 4 is dedicated to the concept of informed consent as a central component of medical ethics. The history of informed consent is related to the development and progress of medical

research. Atrocities during the Nazi era in Germany concerning human experiments without the knowledge or permission of patients ushered in an increased emphasis on informed consent.

As a starting point for this chapter the definition of informed consent is examined, as well as the debate around informed consent as a principle. Among others are presented: the meaning of "informed consent" as a term with reference to the procedures followed to obtain an informed consent, the "informed" and the "consent" part of the term, the two notions of the informed consent as they are presented in the Faden R. and Beauchamp T. book: *A History of Informed Consent,* and last but not least the limits, the risks and the considerations around the principle of informed consent.

After having informed consent introduced, the issue of the "right to know or not know" genetic information needs to be addressed. The rapid development of genetic testing and screening techniques which can provide an increasing amount of genetic information raises several conflicts in terms of "rights to know or not to know".

Following the analysis concerning informed consent, Chapter 5 analyzes the meaning of "the right to know" as a principle and the links between the "right to know", Kant's and Mill's theory. Moreover, the analysis extends in terms of who has "the right to know", mainly focusing on the invasion of genetic privacy and autonomy concerning reproductive choices.

The chapter supports the idea that each person individually should understand the option to choose between knowing and not knowing, examine whether he/she really needs this information, and most important whether he/she is ready to accept the information and take any responsibilities involved.

After exploring the main elements evolving from genetic testing in the aforementioned theoretical discussion, Chapter 6 introduces the author's empirical research, focusing on thalassemia and thalassemia trait. The case of testing for thalassemia trait in Cyprus constitutes an

example of how several issues discussed in the theoretical part of the thesis, can be applied in a real life situation.

The specific study took place in Cyprus between September 2003 and February 2004, and it was based on discussion groups. To date, no systematic study concerning Cypriots' perceptions on thalasssemia and thalassemia trait has been undertaken. The purpose of this study was to discover how and in what degree people in Cyprus are influenced by the decision of Cyprus Government to establish premarital testing as the national public policy for thalassemia major. Additionally, the study explored whether people believe that the existence of thalassemia and thalassemia trait in Cyprus affects them in any way as personalities, active and productive members of the society.

The first section of the chapter presents some background information on thalassemia minor and thalassemia major, the reason why Cyprus was chosen for this empirical study and how Cyprus case is differentiated in terms of genetic screening. Furthermore, some information is presented concerning the research procedure including the hypothesis, methodology and the design of the study, the criteria for selecting study participants, and its duration.

The second section of the chapter provides an in-depth examination of the findings. This part looks at some important issues deriving from the study such as how people define "thalassemia" and "thalassemia trait", how the word "serious" is described and what people understand by saying that "thalassemia or thalassemia trait is serious", what the word "stigma" means for the participants and whether Cypriots stigmatize thalassemia or thalassemia trait carriers.

Chapter 7 highlights the author's findings and proposals. It is stated that by introducing pre-natal diagnosis along with premarital screening Cyprus succeeded in eliminating the number of births with thalassemia. However, it is doubted whether the combination of premarital screening and prenatal testing, succeeded in making people more responsible about their decisions.

There are not really any criteria which may clearly indicate whether people became more responsible or irresponsible since 1970 (when Cyprus strategy on thalassemia was lounged).

Nevertheless, it can be assumed that premarital testing can be considered from lost of people as a routine examination (without necessarily be a means to increase someone's responsibility towards thalassemia).

The following part of the thesis introduces some recommendations concerning Cyprus strategy on thalassemia. Summing up the theoretical and the empirical part of this thesis, the author would like to suggest some further improvements which can help Cyprus to improve its screening programme and at the same time to allow people in Cyprus take their own decisions concerning their genetic information.

The closing chapter summarizes the argument of the thesis. Informational autonomy allows people to have the freedom to make choices among alternative sets of information, ideas, and opinions. This includes the freedom to decide what information someone wants to receive and possess. Human genetic information is ultimately, not about genes; it is about people.

The challenge for Cyprus in the 21st century is to advance the existing control programme while respecting freedom. By introducing pre-natal diagnosis along with premarital screening Cyprus succeeded in eliminating the number of births with thalassemia, but increasing the number of abortions at the same time. People in Cyprus must have the option to know or not know if they are thalassemia trait carriers and should not be "forced" to make a premarital testing. Informational autonomy is a right that Cypriots are allowed to have.

CHAPTER 2 - BACKGROUND INFORMATION

Recent scientific advances in the understanding of human genetics, particularly those achieved but the Human Genome Project, hold the hope of significant progress in the prevention, diagnosis and treatment of a disease. The identification of genetic information belonging to a specific individual is one of many new technologies with which advanced health care aimed to unlock the causes and factors in diseases and provide better treatments.

In the last decade there was a lot of debate about Human Genome Project (HGP) an international program that concentrates on constructing genetic and physical maps of the human genome. The genetic research, which is being held in the frame of HGP, is focusing utterly on mapping human genome and determining the nucleotide sequence of human genome in its whole.

Genetic research is a very broad term and despite the fact that it is in a very primary level it has to do with researching the gene and has various aims and applications.

Genetic research also includes the investigation of single gene diseases. Gene therapy is concentrating i.e. on immune deficiencies or cystic fibrosis and gives some very optimistic promises of a possible future cure of these diseases. It is also important the fact that during the last few years gene therapy has developed some new perspectives focusing on diseases that are influenced by multiple genes as well as environmental factors.

To identify and specify the genetic environmental factors, which seem to influence specific diseases may lead us to indicate any predispositions to a development of a disease. By discovering whether an individual, a biological relative or a population may have a genetic predisposition, e.g. in breast cancer, does not guarantee that they will definitely suffer from the specific disease, but makes it possible to identify and eventually eliminate those factors that cause a specific disease.

Human genetic research does not aim directly to offer a therapy for any genetic disease or any other disease which will be proved as the result of a gene disorder combined with other environmental factors. On the contrary, it investigates several issues that have to do with genes and their function and disorders. Genetic research does not consider therapy as its ultimate intention. Therapy is emerged as the result and its final achievement.

It is also vital to mention that because human genetic research has an experimental nature; it includes lots of risks along with the benefits. The clearly significant findings in the scientific level not only open new horizons of expanding knowledge but also they make obvious the need of protecting and at the same respecting the people who volunteer to be subjects in a research, or who are tested for a specific genetic disease etc.

Genetic testing is defined as the use of a scientific test to obtain information on some aspects of the genetic status of a person, indicative of a present or a future medical problem. The information obtained from such tests may sometimes be used to anticipate the onset of certain genetically determined diseases and to initiate appropriate early therapy or any other necessary action. Most tests can be done at any stage of life, either before birth (pre-implantation and prenatal diagnosis) or after birth (neonatal and adult screening and testing).

Genetic screening usually takes place when an individual or a group shows risk for a disease or trait. It can be defined as the identification of individuals possessing certain genotypes that are either associated with a specific disease or a predisposition, or which lead to a disease in their descendants. Genetic screening aims to an early recognition of a disorder either for intervention which may prevent the disease process or for future management of a disease when symptoms are anticipated.

Despite the fact that many of the common complex diseases such as cardiovascular disease or cancer are a product of genes combined with different environmental factors and genes have only a contributory rather than a causative role, there are several monogenetic diseases such as

Huntington's disease or cystic fibrosis in which genes have a major causative role. Therefore, in these cases genetic screening and genetic information are vital for detecting these inherited genome modifications.

There are different categories concerning genetic testing and genetic screening: [2]

(1) Diagnostic testing: the genetic information of an individual is probed for defects or variants in one or more genes to confirm or exclude a specific disease diagnosis. In the same way, carriers of genetic defects can be identified.

(2) Diagnostic Screening: Prenatal screening for major chromosomes and neural defects is done using serum tests combined with ultrasound imaging, neonatal screening for treatable diseases as hypothyroidism and carrier screening for monogenic diseases as cystic fibrosis.

(3) Predictive testing: tests in this category cover a broad range of disease and acquired conditions and can lead to the prediction of future health status of an individual. Two different groups of predictive tests can be distinguished, based on the nature of information resulting from them: a) Presymptomatic tests where the presence of defects in certain specific genes or gene products creates an almost 100% risk of developing a particular disease later in life. b) Predisposition tests which include tests for other disorders in which defects in a single major gene are considered to increase substantially lifetime risk of developing the disease.

As genetic research is evolved, other possibilities for testing and screening will become available, leading to an increase in the accuracy of the tests. This will result to more precise predictions of the risks of developing a particular disease and its likely progression.

Precisely because genetic testing can reveal information about more than one family member, the emotions caused by test results can create tension within families. Test results can also affect personal choices, such as marriage and childbearing. Additionally, as genetic testing deals with

[2] As these categories are defined in European Commission Report, *Ethical, legal and social aspects of genetic testing: research, development and clinical applications,* Brussels 2004

genetic information issues, this raises some concerns surrounding the privacy and confidentiality of genetic test results. This debate can be also applied in the case of genetic research where the participant may be asked to provide personal genetic information.

Persons being tested or participate in a research are not the only people with an interest in the test (or research) results. Family members and potential partners, employers, insurers, and the government all may desire information about a person's genetic endowment. In the following part of the chapter some of these clashing interests are briefly analyzed.

2.1 Interests of research participants

Research participants are the people who consented to provide some personal genetic information for a specific genetic research. Research participants may have or may have not different interests in the context of genetic research. If we accept that more scientific knowledge is beneficial, and then if a specific genetic research is carried out then this can be considered for the participants' best interest, provided that the participants are not exposed in high risks arising from the research. There are always the positive interests of the participants concerning the discovery of a new cure for a disease or an improved treatment, however there are some risks that can be concerned as negative interests and refer to the exploitation of genetic information.

Informational risks and protection of confidentiality are by far the most important issues in the context of genetic research. However, even the best protection safeguards cannot guarantee the prevention of any risk concerning confidentiality. Disclosure may inflict psychological harm, but the reason why research participants deserve protection from informational risks is that, once the genetic information has been disclosed, then this information can be used to cause some very serious consequences in various aspects of the participant's everyday life.

If for example any genetic information is disclosed to third parties (employers, insurance companies) then the participants is exposed to social risks including stigmatization, and this may have a possible impact on other members of the participant's family.

Informational risks are prevalent but various legal regulation try to ensure that genetic information are efficiently protected and if a possibility of information leakage occurs then, because of these regulations the results of such event are minimized.

2.2 Interests of the society

It is undoubted that science itself and many times science's results do not benefit the human kind. However, properly conducted scientific research (and that is the research which follows the appropriate guidelines) is beneficial to the society, and people are benefited from it as well. Additionally, even if we do not account the public health context and focus to a more individual level, it can be argued that genetic research and trust in scientific progress give people hope for the future not only in a personal level but also for future generations. This can also be a possible motivation for participating in a research in the first place – the hope of a cure, the possibility of findings which will provide a more efficient treatment for a disease etc.

However, in order to receive benefits from the society it is essential that people should contribute in the society as well. John Harris argued that "*minimal risk research is something that every reasonable and decent person who does not want to be a free-rider should participate in*".[3] Additionally, Onora O'Neill supported the idea that the use of health data "*is part of the obligation of any society to assist medical advance for future generations, repaying the debt to earlier*

[3] Harris, J., "Ethical genetic research on Human Subjects", *Jurimetrics Journal,* Vol. 40 (1999), p. 87

generations for the medical benefits they in turn had assisted".[4] Therefore, society in general has an interest in the participation of its members in genetic research.

It is only through genetic research that it can be determined how much and what type of information and support are required for the increasing numbers of people being offered genetic testing, and how these are most efficiently provided, to achieve good understanding of a test and its results. In this way, behaviors can be facilitated to reduce risk, without high levels of emotional distress.

It is also important that genetic research is a means to investigate the implications for the society itself, of uncovering any genomic contributions that there may be to traits and behaviors. On the other hand, it is important for society to define the appropriate and inappropriate uses of genetic research. A fruitful discussion between diverse parties based on an accurate and detailed understanding of the relevant science and ethical, legal and social factors will promote the formulation and the future application of effective policies.

2.3 Interests of the researchers

It is undeniable that researchers' only driving force is their desire to increase knowledge and progress science. Academic success and personal well being are two factors which influence researchers a lot. If in a country genetic research is rapidly developed then it is natural that economic incentives will play a crucial role.

However, freedom of science and freedom of enterprise is a basic right in the society. It is also important that even for these basic rights there are limits which regulate them. Concerning for example the scientific freedom, none should be forced to participate in a research.

[4] House of Lords, Committee of Science and Technology: http://www.publications.parliament.uk/pa/ld200001/ldselect/ldsctech/57/5709.htm

The trust that people have in a research is fundamental in order to ensure that research can be carried out. If people do not trust the researchers then this will be an obstacle for people to participate in a research. Therefore, it is necessary to facilitate some necessary safeguards for genetic research (i.e. informed consent) in order to encourage people to place their trust on researchers. As Onora O'Neill argues "*informed consent is always important, but it is not the basis of trust. On the contrary, it presupposes and expresses trust, which we must already place to assess the information we are given*".[5]

As the above part of the chapter aimed to present a sketch of different interests that clash concerning genetic research and consequently genetic information and briefly outline their major concerns and hopes, the following part provides a brief reference on genetic information and whether this information can be considered as exceptional and different from other kind of information.

To start with, we have to clarify what genetic information is. When we use the term genetic information we refer to personal genetic data. This kind of data is able to reveal information about a person's genetic inheritance, including gender, race, height, weight and many other features.

One approach takes for granted that there is something special about genetic data compared to other medical data and therefore this presupposes that genetic data should be treated in a different way than other medical data. Those who view genetic information as something special usually base this argument to (i) the predictability (ii) the inheritability (iii) the sensitivity of genetic information (iv) the fact that we share genetic information with other family members, (v) the fact that genetic information identifies us and that genetic information (vi) has been misused for discrimination and for eugenics purposes.

As technology continues to progress, DNA testing becomes cheaper, and can provide instantly genetic information. Consequently, we have to examine precisely whether these genetic

[5] O'Neill, O., *A question of trust: The BBC Reith Lecture 2002,* Cambridge: Cambridge University Press, 2002, p.87

information are more sensitive that the other medical data and demand any institutional and individual safeguards in order to protect privacy, and prevent discrimination.

There are many types of scientific tests that can give us genetic information. DNA tests are one kind of such tests that are more accurate and can reveal among others, through the analysis of an individual's DNA, any disorders or susceptibility to a specific genetic disease. Additionally, other biochemical tests of non-genetic substances (i.e. ordinary blood tests for cholesterol) are able to provide similar genetic information.

It has to be mentioned that there is a great debate around the issue of genetic information and whether it is sensitive in order to be treated individually from all the other medical data. Thomas Murray in his article concerning genetic information claims that we cannot support the argument that genetic exceptionalism is an issue for debate since as a term genetic information is not in any way different from other kinds of medical information and does not demands special protection.

He suggests that *"genetic exceptionalism is an overly dramatic view of the significance of genetic information in our lives...The more we repeat that genetic information is fundamentally unlike other kinds of medical information the more support we implicitly provide for genetic determinism for the notion that genetics exerts special power over our lives".*[6]

Additionally, Søren Holm agrees with the above idea claiming that "*the sooner we rid ourselves of the idea that there is nothing special about genetic information the sooner will be able to deal constructively with the large issues raised by the use of all kinds of health related information*"[7]. He argues – as Murray does – that it is all a matter of what people tend to think about genetic information. Specifically, Holm suggests that "*genetic essentialism is the idea that the essence, the nature of human being is defined by genes*" and although he admits that genetic

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[6] Murray, T.H., "Genetic Exceptionalism and 'Future Diaries': Is Genetic Information different from Other Medical Information?", *Genetic Secrets: Protecting Privacy and Confidentiality in the Genetic Era*, edited by Mark A. Rothstein, Yale University Press 1997, p. 60 - 73

[7] Holm, S., "There is nothing special about genetic information", *Genetic Information: Acquisition, Access and Control* edited by Thompson A. and Chadwick R., Kluwer Academic/Plenum Publishing, New York 1999, p. 97

essentialism plays an important role in the public perception, according to his opinion, "*there is no doubt that genetic essentialism is false*".

Additionally, Murray argues that there are other medical data that can provide if not the same, equally important information about individuals as genetic information. Among others he claims that genetic information cannot be categorized as the most sensitive information for individuals as its disclosure can be important for other people as well i.e. if one partner in a marriage has a sexually transmitted disease this information can be equally important for the other partner. Furthermore, we cannot claim that the disclosure of genetic information can be a basis for discrimination as even insurance companies have always used evidence of current or future disease risk and these evidences included also non-genetic information. Consequently according to Murray we need to ask: is it fair to discriminate on non-genetic information but unfair to discriminate on genetic ones?

Following the above way of thinking, we have to ask whether genetic information as a special kind of information is just an *idea*, a false perception, which happened to bother the public. And if it is just an idea, a false perception then somebody may argue that what is has to be done is to change people's ideas by opposing the right arguments-evidences.

If we take as a fact that human beings are not defined totally by their genes and there are other environmental factors that influence not only physically human beings but psychologically as well, by affecting their personality, then the idea of genetic essentialism seems to be wrong.

On the other hand, if we support the argument that genetic information is connected with the very personal and most private element of a person – DNA – and also it can practically influence the way of life, the personality, family members and in many cases ethnic groups, may suggest that genetic information is something special compared to other medical information and not just an idea that people happened to develop in their minds.

G. J. Annas, L. H. Glantz and P. A. Roche's in their article entitled: "Drafting the Genetic Privacy Act: Science, Policy, and Practical Considerations" support the idea that a genome metaphorically can be a *"coded probabilistic future diary because it describes an important part of a person's unique future and, as such, can affect and undermine an individual's view of his/her life's possibilities. Unlike ordinary diaries that are created by the writer, the information contained in one's DNA ...is in code and is largely unknown to the person"*[8].

Murray's answer to the metaphor of 'future diaries' is that *"our genes are a list of the obstacles we are likely to encounter and perhaps as a somewhat better prediction of how long we will have to do what matter to us, to be with people we love, and to accomplish tasks for ourselves...Our genes might be regarded metaphorically as the physical, but blank volume in which we will create our diary...the physical volume is not the content of the diary. The content we must write ourselves"*.

The following part of the chapter argues that despite the fact that some people believe that genes are not considered to be the most crucial factor which tends to define the whole human existence, genetic information (which in Murray's answer is consisted by the information about the condition of the volume and not the content) is very important for the *diary* itself.

Exactly, because the condition of the "volume" plays a vital role and although writing on a blank page according to Murray gives you the freedom to create your own content, the question opposed to Murray is: what happens if "the volume is damaged"? If for example one person has a severe genetic disease and therefore the "volume" is affected, then the content of the "volume" is not affected as well?

Generally, my argument is that personal genetic data are concerned to be very important in terms of privacy and confidentiality, and raise lots of ethical concerns that go beyond any medical or other data that is able to be collected from individuals.

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[8] Annas, G. J., Glantz L.H. and P. A. Roche, "Drafting the Genetic Privacy Act: Science, Policy, and Practical Considerations", *Journal of Law, Medicine and Ethics*, Vol. 23 (1995), *p. 360*

To start with, genetic information can constitute a proof of each person's uniqueness. Because of this uniqueness we are able to use the term "individual" meaning each autonomous person separately. It cannot be argued that people's uniqueness can be justified entirely because of their genes. However, it can be characterized as the sum of our unique genes along with our special characteristics and features of our personality. Those two elements: genes and personality are equally important and are not necessarily linked with each other i.e. the case of identical twins, although they are genetically identical they have different personalities.

Another important point is that genetic information *"acted upon by environmental factors, gives the backdrop to disease susceptibility, perhaps indicating a potentially increased (or decreased) risk of disease many years into the future"*[9]. This is similar to the 'future diary' argument although again it is vital to understand that genetic information indicates only a *potential* risk of a disease. This should not be confused in any point with genetic determinism. It cannot be argued that genetic information can foretell in any way, how people's life is going to be or how someone is going to spend the rest of his/her life. It is just a hint to what *may* appear and must be seen as a way to prepare individuals and enforce their will of living and encounter with anything that may occur.

In this way even in the case in which genetic information can influence other family members regarding their susceptibility to a disease, in order to prevent stigmatization and any possible exclusion socially or in any other way, genetic information must be protected and treated with the proper sensitivity according to each case. If we recognize that genetic information needs special treatment legally, ethically and socially, we are able in this way to anticipate any cases of discrimination.

Someone can argue that if we protect genetic information legally, ethically and socially we tend to impose to the public the idea that genetic information is very "dangerous" and "harmful"

[9] House of Lords, *Report on Human Genetic Databases,* March 2001

and in this way increase the possibility of discrimination making everybody too cautious and superstitious against genetic information. However, to this argument it can be opposed the fact very often the lack of knowledge concerning "what genetic information is", may influence the public. Thus it may be more effective instead of arguing that genetic information is not so sensitive and therefore must not be protected in any way, we should perhaps invest more in informing the public more about potential risks or benefits the disclosure of genetic information may has, in order for people to be well prepared and possess more accurate knowledge on the matter.

"If a genetic information...is no less accessible than a person's banking, educational or telephone records and as long as most of the population fears genetics and believes that genetic information poses a risk to their personal welfare, the law will protect the information as " private" notwithstanding any ethical, legal or economic reasoning about need"[10].

As the above points out, people's opinions about genetic information counts a lot, and consequently this raises the need for protection of this information. People deal with disclosure of genetic information in their own environment as well. They should be informed about the nature of genetic information in order to be prepared if a friend, a neighbor or even a colleague, decide to disclose genetic information without the possibility to be stigmatized.

It is important that since we recognize the sensitivity of genetic data to find ways to protect it in a theoretical level but also more practically, legally and socially. As Annas observes *"current rules for protecting the privacy of medical information cannot protect either genetic information or identifiable DNA samples stored in DNA databanks. A review of the legal and public policy rationales for protecting genetic privacy suggests that specific enforceable privacy rules for DNA databanks are needed"*[11].

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[10] Fink, S.F., "EEOC vs. BNSF: The Risk and Rewards of Genetic Exceptionalism", *Washburn Law Journal*, Vol. 42 (2003)

[11] Annas, G.J., "*Privacy rules for DNA databanks: protecting coded future diaries*", The Journal of the American Medical Association, Vol. 270, No. 19, November 1993

Therefore, if we want to cultivate - let us call it "genetic consciousness" (meaning to be conscious about the meaning of genetic information and understand the risks and the benefits of its disclosure) - we need to gain public's trust. As Onora O'Neil supports that *"only 'personal data' supplied by individuals for specific purposes are to be protected from disclosure and here again technology supposedly rides to the rescue, providing new standards of encryption and hence new possibilities for data protection"*[12].

To succeed in this protection of personal data sufficiently we need apart from *integrity, objectivity, accountability and honesty*[13] to provide a strong bedrock in order for the public to be convinced to place its informed consent and give any genetic information feeling secure. What is needed is not only to trust the government and scientists but also to trust each other. Is this easy?

This is a very difficult question to be answered, as trust has to be tested first, in order to be gained. This demands very strong foundations of understanding and realizing to what people are consenting for when it comes to genetic information. It is undoubted that genetic information is a very important element to be disclosed without informed consent.

[12] O'Neill, O., *A question of trust: The BBC Reith Lecture 2002,* Cambridge: Cambridge University Press, 2002, p.68
[13] ibid, p. 67

CHAPTER 3 – PHILOSOPHICAL BACKGROUND

Dealing with the problematic question of what makes a research project involving human subjects an ethical one several theories express a variety of approach. In this chapter we are sketching the basic principles of utilitarianism, referring to the application of utilitarianism in genetic research focusing on the dichotomy of act and rule utilitarianism. Additionally, Kantianism and specifically categorical imperative and finally paternalism are also analyzed. We would like to mention that we use paternalism as a starting point to refer to Mill's liberalism and Onora O'Neil's interpretation of Kantian categorical imperative as principled autonomy.

3.1 Utilitarianism

In general, utilitarianism is a theory based on the consequences of an action. For utilitarianism an action or a decision is ethically right or wrong if it maximizes happiness, and therefore minimizes any harm. In the case of a genetic research a utilitarian approach would approve the constitution of a research and will considered it morally obligatory to be held, if through the investigation and the retrieval of any genetic information, its outcome is going to be useful and hence will maximize health, to a majority of people.

The philosophical idea that underlines the above is the utilitarian argument stating that the general aim of humankind must be to maximize happiness. Definitely, this argument has many implications as it involves the hunting of happiness at any cost, focusing on the majority and putting aside the minority and the preferences of the individuals.

In what follows we will examine utilitarianism as an ethical theory, discuss its origins, the implications deriving from its arguments, its strengths and weaknesses, and finally in what way genetic research can take advantage of the utilitarian approach.

3.1.1 Origins

Utilitarianism is a method of moral thinking according to which in order to decide if an action or a decision is morally right or wrong we have to examine its consequences first. Jeremy Bentham (1748-1832) was the first one to apply this way of thinking in practical problems. For Bentham, applying the principle of utility was a matter of a simple procedure. If an action was maximizing happiness for a large group of individuals compared with the group of individuals that were unhappy then it was morally right.

A utilitarian approach rests on the premise that everyone wants to experience pleasure and avoid pain and in this way the morally right action to be taken is to proclaim the greatest good for the greatest number. *"By utility is meant that property in any object, whereby it tends to produce benefit, advantage, pleasure, good, or happiness (...) or (...) to prevent happening of mischief, pain, evil or unhappiness to the party whose interests are considered"*[14] .

In general, Hare points out correctly that the main constituents of utilitarian theory may be called consequentialism, welfarism and aggregationism[15] . According to consequentialism what determines the morality of an action is the consequence of the specific action.

Welfarism is also connected with the consequences, but refers to the impact those consequences have to people concerning their welfare. An action can be characterized right or wrong if its consequences tend to increase or not the welfare of people affected by these consequences. The fundamental point here is in which way welfare is defined. For each individual

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[14] Bentham, J., *An Introduction to the Principles of Morals and Legislation*, republished Oxford: Clarendon Press, 1907, "Chapter 1: Of the principle of utility"

[15] Hare, R.M., "A Utilitarian Approach", edited by Kuhse, H. and P. Singer, *Blackwell Companion to Philosophy: A Companion to Bioethics*, Blackwell, 2001

person welfare can take various meanings, because it is connected with the preferences every person has. Consequently, according to utilitarianism, if the consequences of a specific action satisfy our preferences and bring us welfare, then the specific action is morally right.

Hare mentions that when we talk about preferences we must always have in mind that we have *now-for-now* preferences (*a preference now for what should happen now)*, *then-for-then* preferences (*a preference at some later time for what should happen at that time*), and *now for then* preferences (*a preference now for what should happen at some later time*).

Accurately, Hare makes an interesting point as he distinguishes how both *then-for-then* preferences and *now for then* preferences can conflict with each other. It is possible that a *then-for-then* preference, i.e. in case someone has a gene that can cause a genetic disease in the future, may have the preference that a GP will inform family and employer. However, in the case which this really happens someone may change his mind and at that time may have the preference to keep it as a secret. Consequently, it is not possible following the utilitarian approach to satisfy both preferences; consequently *now-for-then preferences do not count.*

To satisfy individual's preferences and therefore increase welfare is very difficult to be succeeded and that is why utilitarianism approach suggests that we should choose the action which maximizes the welfare *of all in sum, or in aggregate.*

Aggregationism, suggests that we should choose the action which produces the most welfare for a very large group of people. Hence, if we have to choose between two actions our first criteria should be if their outcome will bring more welfare, and our second (criteria) to choose the action that can bring the maximum welfare for as many people as possible.

3.1.2 Implications - The application of utilitarianism in genetic research

Although in the bibliography there is a lot of debate against utilitarianism and its weaknesses as an ethical theory, we do not consider it useful for this specific thesis to comment on utilitarianism as a theory, but rather to outline its basic principles in order to proceed to an examination of its weaknesses and what utilitarianism has to offer to genetic research. It is true that although many authors analyse the theoretical frame of utilitarianism do not concentrate on how it can be applied -if this is possible- to our everyday life in general, and specifically in genetic research.

Utilitarianism has accepted a lot of criticism especially having to do with its consequentialism. An important point is the fact that, most of the consequences are not so obvious and therefore they cannot be predicted at the time we act. A genetic project that involves human beings may include some risks or have some consequences that even the scientists do not know or are able to predict a priori.

The use of genetic samples in a research project, their future use and the possibility of revealing any personal genetic information are factors that can have unpredictable consequences. Even if in some point consequences can be predicted in advance, it is not possible to cover the whole range of consequences that may occur during or after the research is finished.

It is true that when Bentham is analysing his utilitarian theory follows a kind of mathematical approach, meaning that the only thing we must do is to *identify and count the number of individuals affected by our actions, calculate for each person whether the action would increase or decrease his/her happiness, count up how many individuals would be made happy and how many made unhappy, and then act accordingly*[16].

[16] Bentham, J., *An Introduction to the Principles of Morals and Legislation*, republished Oxford: Clarendon Press, 1907

Obviously, the above mathematical approach cannot be applied in genetic research involving human DNA because on the one hand, human beings are involved and on the other hand, issues as respect to individualism, confidentiality and personal autonomy are at stake. The consequences for not having in mind all the aspects in their plurality can lead to discrimination, stigmatisation of individuals and populations and in the worst case the misused of human DNA.

As it can be assumed genetic research is not about people making choices and acting according to their preferences, but also it involves many other factors such as insurance companies, employers and governmental policies. Possible consequences are too many to be numerated not even to be predicted a priori and therefore according to utilitarianism it is difficult to characterize actions ethically right or wrong.

Moreover, in a utilitarian approach certain actions are able to be characterized as ethically right if they only increase welfare for a majority, sacrificing in this way the preferences of the minority. This is completely inapplicable in the case of genetic research since individuals are of great importance. If for example the majority of scientists involved in a genetic research wanted to allow other scientists to use the DNA samples collected for a specific research project, it is up to the participants to accept or not further use of their DNA after the project has finished. If a single participant does not prefer to allow his DNA sample to be used after the specific research is finished, his preference must be respected.

3.1.3 Rule and Act Utilitarianism

It can be argued that weaknesses mentioned above can be avoided if we follow *rule utilitarianism* instead of *act utilitarianism*. *Rule utilitarianism* mostly has to do with the

formulation by individuals or a society of rules which "*take into account what is most likely to be of benefit, all things considered and then sticking by the rules as if they were duties*"[17] .

Rule utilitarianism can avoid many of ambiguities of utilitarian approach because it does not concentrate on maximizing the welfare (*act utilitarianism*), but on the adoption of a general justified (*by utility*) moral rule that can be the guide to human actions. It is important that this moral rule is going to be justified through utility, meaning that it will have the least harmful consequences.

Hence, according to rule utilitarianism, if we say that deception and coercion must be prohibited in genetic research, we can therefore perceive that the informed consent will be based on the fact that the participant will never be deceived and receive all the valid information needed. In this way any consequences deriving from deception and coercion on behalf of the researcher can be diminished.

First of all rule utilitarianism can be more a *kind of rights and duties theory* and it could be easily considered as to be a *virtue theory*, because it presupposes the engagement on behalf of the scientist in this moral rule and consequently the fact that the scientist will never under any circumstances break this moral rule. Additionally, we must also have in mind the procedure which will be followed to formulate this rule. Rules still have to be accepted as a justifiable moral rule by a majority of a society or a country otherwise it cannot be considered as valid? Consequently, rule utilitarianism will still have to refer to the majority and not the minority, a fact that leads us to the weaknesses mentioned above.

Despite the fact that utilitarianism has lots of weak points, it can still be beneficial through some vital issues derived from utilitarian theory per se. In general, we should credit utilitarianism for the importance it gives to the consequences of an action. This indicates that we should first look at all possible consequences an action can have and then proceed to its application.

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[17] Campbell, A., G. Gillett, and G. Jones, *Medical Ethics*, Oxford: Oxford University Press, 2001, (3rd edition), p. 8

Although it is undeniable that those consequences can not be the only criteria for categorizing an action as ethically right or wrong, it is an important factor for undertaking an action and consequently a genetic research that has morally questionable consequences (at least those consequences that can be predicted a priori).

On the one hand, we can use utilitarianism in a sense that can give validity to a genetic research in general, by investigating and analysing any potential harmful consequences and in this way try to improve in any way the whole procedure. On the other hand, utilitarianism can indicate that it is important to diminish any unwanted consequences or if this is not plausible at least to clarify what the potentials of these consequences are. In this way we will be capable to put a basic framework – especially in genetic research- which can lead to better results.

3.2 Kantianism

According to Kantianism we should seek our boundary to moral law nowhere else than in the existence of pure reason inside us. To understand this we should first examine what pure reason means for Kant. It is nothing less but the natural source inside human beings which can provide human beings with the objective criteria to distinguish absolute truth and in this way act morally right, without any influences from the empirical world. Reason is autonomous because of the fact that it does not need any external assistance to recognize the truth, and therefore directs the will. This direction of the will is expressed in Kantianism as the motive of duty. Consequently, morality can be sought in actions which are performed solely by the motive of will. The supreme maxim/law which directs our actions is the *categorical imperative*.

3.2.1 Categorical Imperative

Categorical imperative is a criterion of judging which of the other maxims that govern our actions are morally acceptable or not. Nothing else such as the empirical world can provide us with the necessary foundation for our actions, but the categorical imperative.

Kant identifies the following categorical imperatives: (a) act in such a way that the maxim governing your action can be a universal law (b) act in such a way that you can at the same time *will* that the maxim governing your action can be a universal law of nature. The above for Kantianism are very basic and are required as moral laws which govern our life. Obviously, many times we fail to follow this moral laws and this does not presuppose that we are *irrational* beings, but that our preferences and consequently our will, tend not to be obedience to our Reason but to external influences.

According to Kant's categorical imperative a scientist must not use deception, coercion or in any way try to mislead a participant to a research project not because of any external purpose, but exactly because he is a scientists and he has a *duty* to act for the participant's best interest. He is not doing it because everybody believes it is rational to do so, or because everybody is doing the same, but because it is rational if he calls himself a scientist to act as such. Hence this leads us to a duty-based moral thinking and finds application to all cases that involve human beings in a research.

On the one hand, it is very crucial to mention the importance of categorical imperative for the principle of "informed consent". Following categorical imperative will increase the validity of genetic research, as the potential participant is going to know that a scientist will not ask him to do something that is going to be harmful for him and also will not use any mischievous methods for example psychological pressure to persuade him participate in a research.

In this way strong bedrock is created based on trust and belief between the participant and the researcher. This makes things easier (for the researcher and the participant) as time will be invested

in different aspects of the actual research and not on informing the potential participant in order for him/her to decide whether or not he/she will participate.

3.2.2 Criticism

On the other hand there are a lot of objections concerning categorical imperative. To know that a researcher according to categorical imperative is not going to coerce or deceive the participant because he is a scientist, can lead the potential participant to agree to participate in the research not because he has made an informed decision but mostly because he is convinced that the researcher is not going to display him/her in any danger. In this case the whole procedure of the informed consent is loosing its meaning.

Additionally, categorical imperative it is categorical and that means that it does not allow any exceptions. In some cases the researcher has to lie if asked about personal genetic information of a participant in a research. If the participant does not want his/her personal genetic information to be disclosed then the researcher is not allowed to reveal those information and therefore tell lies when asked even if other people or a whole population is in danger.

Therefore, according to the universal law "always telling the truth" (and that can be translated to a full disclosure of information), in the case when some people will be stigmatised by the revealing of any genetic information or results from the research, the researcher following categorical imperative has the duty to reveal them.

In addition, Kant refers to a *universal law*: for example always telling the truth. Consequently any other duties or obligations in a relation between the researcher and the participant are omitted. A very important obligation the researcher has is to appreciate that each participant has a unique

personality and therefore a unique comprehensive time[18], consequently it is impossible to talk about a universal law that can be applied in all cases.

In general Kant's categorical imperative cannot be applied practically in genetic research since it does not cover all of the issues, especially concerning the informed consent procedure. To obtain informed consent, a special interactive relationship between the researcher and the participant is needed. The application of a universal law is not enough to cover all the issues, and the will on behalf of the participants to be a subject in a genetic project does not make the project morally right. It takes more than *will* to validate a genetic project as morally right, and Kantian categorical imperative cannot provide the criteria needed[19].

3.3 Paternalism

Paternalism comes from the Latin word *pater*, meaning to act like a father, or to treat another person like a child. In modern philosophy and jurisprudence, it is to act for the good of another person without that person's consent, as parents do for children. As a theory it is very controversial because it is assumed that a person is not able to take the decision on his own and needs someone else to decide for him. The criteria for deciding whether a person is unable for decision-making vary. It can be argued i.e. that a scientist can decide to use or not a genetic sample in a research without the consent of the participant since he can understand better all the scientific information involved.

From the above there are many issues that need to be commented. It is obvious that we are talking about autonomy and whether people have the right to express their autonomy and in what degree. Are the people allowed to take (autonomous) decisions even if these decisions can harm other people? It is also important to define what we mean by harm. Harm can take various meanings depending on how people define harm. Is our own judgement able to take the decision

[18] This is analyzed further in Chapter 4

[19] For a pluralistic perspective of Kantian theory, see also the commenting on Kant's idea of human dignity in a following part of the chapter.

suitable for us or sometimes for example for our participation in a research we need the guidance from a scientist in order to take a decision? These among others are some questions raised in paternalism.

In general paternalism pays more attention to secure safety than liberty for one person. Paternalism considers very important to prevent people for taking a decision that can cause harm either to themselves or to others and puts personal liberty aside. Consequently, since the principle "not to harm" is situated on top of all other principles including liberty we should discuss in what cases the limitation of liberty and at the same time a paternalistic perspective can be justified.

The paternalistic argument is not that liberty is not important or a vital substance for personal autonomy. In the case of a genetic research – according to paternalism - the scientist can decide on behalf of the participant concerning the use of the participant's DNA sample because he (the scientist) can have a better perspective of all the scientific issues involved. In this way, the possibility for an individual to take a decision and consequently, to act in a way that either harms himself or somebody else, is minimized.

Discussing the criteria according to which someone is more "appropriate" than someone else to take a decision, we must mention that paternalism does not formulate specific criteria that each time must be fulfilled from the person that can be authorized to take the decision. For paternalism the only criteria is the degree of causing harm. The one that has to take the decision for somebody else is the one that has the fewest possibilities to cause harm with his decision. Naturally, this is practically very difficult because even in the case of a scientist, a decision can be judged as "harmful" by the participant but not for the scientist.

Although as a principle "not cause harm" is benevolent, in its means paternalism is coercive. It does not include (a) self-competent self-harm (i.e. in cases where a participant is aware that by participating in a specific genetic research there is a possibility of causing himself a severe health damage) and (b) risk of self-harm (since the procedure of informed consent is based on

understanding the risks involved in a genetic research and by accepting to participate in it, it means that someone decides to take all these risks)

3.3.1 Mill's notion of autonomy

In this point we must refer to the *harm principle*, and particularly Mill's notion of autonomy. For Liberalism limiting liberty can only be justified to prevent harm to other people, not to prevent self-harm. Coercion can only be justified if its aim is to prevent harm to others that do not consent to it, not to prevent harm to people that competently consent with the specific action.

In this point it is important to understand why to Mill the differentiation between self-harm and harm to others is important. Mill's naturalistic approach of autonomy allows him to treat personal decisions and therefore action as an expression of human nature. Hence this kind of decisions and actions represents not only the individual and the unique character of each human being but they also represent the expression of our individual desires and impulses.

For Mill the liberty to act as an individual human being is vital "*for each to cultivate his or her own individuality and character and so to contribute both to individual and to social well-being*"[20] . Consequently, Mill rejects Paternalism in the sense that Paternalism uses coercion to promote good (not do harm) against person's wishes. On the other hand, Mill's definition of individualism approves paternalism applied in a frame that has to do with preserving a wider version of freedom, for example when establishing laws that prohibit slavery.

In general, we should mention that for Mill an informed and competent decision and therefore an action is distinctively autonomous precisely because it is deriving from desires of a distinctive sort. I completely agree with Onora O'Neill who tracks the great importance of Mill's theory concerning autonomy because it underlines the significance of choosing, deciding and acting autonomously achieving an informed consent, without any paternalistic influences (in a sense that

[20] O'Neill, O., *Autonomy and Trust in Bioethics,* Cambridge University Press, 2002, p.32

coercion is used against our own wishes, desires and impulses). As Onora O'Neill argues *by insisting on the importance of informed consent we make it possible for individuals to choose autonomously, however that is constructed.*

Mill's notion concerning autonomous choices is obviously coming into contrast with paternalism. Kantian categorical imperative consists also an argument against Paternalism. Although we referred above to Categorical Imperative we consider it useful to expand it here a little further and justify its contrast with paternalism.

3.3.2 Kant's principled autonomy

As mentioned before, one of the versions of categorical imperative is *act in such a way that you can at the same time will that the maxim governing your action can be a universal law of nature.* Onora O'Neill gives a definition to categorical imperative which emphasises the role of autonomy and shows not only why Kantian definition of autonomy is completely against with any kind of coercion (and therefore paternalism), but also how Kantian principled (and not individual as many believe) autonomy *can identify substantial basic ethical requirements*[21] .

Onora O'Neill sees in categorical imperative a form of self-legislation. The key to understand what *self-legislation* means for Kant is not to concentrate on *self* but on how *legislation* is defined and therefore practically applied in everyday life. Kant's categorical imperative is not a version of self-expression, a way in which someone or some people can selfishly decide and impose the application of a law to everybody else and in this way this law can be a universal one. Kant does not talk about an individual who autonomously decides something and pass it as a universal law to everybody else.

[21] ibid. p.89

Principled autonomy is a *test that shows which principles of an action could be chosen by all, that is to say which* principles *are universable, or fit to be universal laws*[22]. People according to Onora O'Neill in their everyday life tend to act on principles that *meet or that flouts the constraints set by the principle of autonomy, but have Reasons to act only on those principles that meet those constraints*[23] . Analysing the above we should first explain further how people can judge that a principle is compatible with the constraints set by the principle of autonomy and also what we mean by *constraints*.

First of all, *the constraints set by the principle of autonomy* is the ability of a law to be applied as a universal one, and as Onora O'Neill accurately argues this presupposes that we are also going to take into account any reasonably foreseeable results of an action. Consequently, to establish a universal law we must first pass it through the test of principled autonomy. If it passes the test then it can be established as a universal law.

The test can be understood easier through an example. Let us assume that an agent decides to adopt a principle of publishing all the personal DNA information gathered from genetic research without the consent of the participants. Let us also assume that this is happening by using some means like coercion, misleading the owners of the DNA information etc. Assume that a group of people is committed to this principle. The consequence of this action will be that some people of this group will not be able to adopt this principle because their *capacity for action* will be diminished because some people will take advantage of them; perhaps they might feel that their individuality is undermined etc. Consequently, the test showed that the specific law cannot be applied as a universal law because of this internal contradiction.

Hence, from the above we can argue that categorical imperative posses a strong mechanism exactly because of its structure that rejects everything which is not compatible with principled

[22] ibid. p.84
[23] ibid.

autonomy. In this case paternalism is one of those laws that can not be applied as a universal because it lacks of compatibility with principled autonomy.

CHAPTER 4 – INFORMED CONSENT

Informed consent is a central component of medical ethics. The history of informed consent has its roots in the development and progress of medical research and dominated mostly the second half of the 20th century. Atrocities during the Nazi era in Germany concerning human experiments without the knowledge or permission of patients ushered in an increased emphasis on informed consent.

Specifically, during World War II, doctors in Nazi Germany were conducting research on prisoners in concentration camps. This research was done on involuntary participants who usually died as a result of the experiments. After the end of the war, many of these doctors were tried at the Nuremberg trials for their crimes. The international community was shocked by the revelations of

their research and the foundations of the physician-patient relationship began to suffer, as it was no longer clear what was "benefit" and what "harm".

The Nuremberg Code was created in 1948 and it was one of the earliest documents to address ethics in medical research. In the document the principle of consent is mentioned in the very first chapter as "absolutely essential" and that voluntary consent should be mandatory for any clinical research. The experiments conducted by Nazis were atrocious and needed to be condemned not because they were conducted without the consent of the people "participating" but because of the aim and the nature of theses experiments.

The Nuremberg Code is viewing informed consent as a proof of assurance and evidence that there has been no "force, fraud, deceit, over-reaching, or other ulterior for of constraint or coercion". That is why the Nuremberg Code refers to "voluntary consent" and not to "informed consent".[24] According to the Nuremberg Code voluntary consent meant that the participants were able to consent, were not being coerced into participating in the study and understood the risks and benefits involved. The Nuremberg Code does not analyse informed consent explicitly and it does not mention anything about information or autonomy.

The adoption of the Declaration of Helsinki in 1964 introduced again informed consent as the principle requirement of medical research agenda.[25] The Declaration stated that no non-therapeutic clinical research could be conducted without the fully informed and free consent of the human being concerned.

As it can be assumed from the above, the consent requirement was introduced in the field of medical research as a reaction to the misuse of medical science during the Second World War, and aimed to protect the life and health of research subjects and also to protect them from any coercion

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[24] Levine R., "Informed consent: Consent issues in Human Research", *Encyclopaedia of Bioethics,* Reich W. (ed), New Work, 1994, p. 1244

[25] World Medical Association Declaration of Helsinki, *"Ethical Principles for Medical Research Involving Human Subjects"*: http://www.wma.net/e/policy/b3.htm

or exploitation. However, the creation of these early codes of ethics demonstrates that the general intention was to regulate research which affects individuals' physical integrity.

As genetic research rapidly evolves it raises demands concerning informed consent as a principle, as well. This does not presuppose a modification of informed consent which discards historical experience, but rather a modification which recognises that in order to maintain the vitality of informed consent as a principle and meet new challenges, some changes are urgently needed.

Nowadays, informed consent procedures must be able to provide an improved protection not only for patients, as doctors are obliged to make sure that a patient understands the risks and benefits or any medical procedure, but for research subjects as well, as requiring informed consent may protect many marginal groups from being persuaded to participate in medical studies without understanding the risks involved.

The historical background of informed consent is not analyzed here in detail as the chapter aims to investigate the informed consent principle as it is used nowadays in medical research ethics and medical ethics in general.

As a starting point it is fundamentally important to examine the definition of *informed consent*. The vocabulary, which is used by geneticists, ethicists or generally scientists concerning informed consent, includes phrases as "the principle of informed consent", the "informed consent principles"[26], "the idea of informed consent"[27], "giving informed consent"[28], the procedure of informed consent etc. Therefore, it important for further analysis to clarify what informed consent is.

This chapter attempts to present the debate around informed consent. Among others are examined: the meaning of "informed consent" as a principle with reference to the procedures

[26] Williams, E.D., "Informed Consent in Genetic Research", *Croatian Medical Journal,* 42(4): 451-457, 2001
[27] Beauchamp, T.L., and J.F. Childress, *Principles of Biomedical Ethics*, Oxford: Oxford University Press, 2001 (6th edition), p.143
[28] Campbell, A., G. Gillett, and G. Jones, *Medical Ethics*, Oxford: Oxford University Press, 2001, p. 222

followed to obtain an informed consent, the "informed" and the "consent" part of the term, the two notions of the informed consent as they are presented in Faden and Beauchamp's book: *A History of Informed Consent* with emphasis on "autonomous consent".

4.1 The meaning of the informed consent as a term

"Informed consent is nothing strange. It is a familiar and ethically important aspect of everyday transactions. Shopping and borrowing a book from the library, taking one's clothes to the cleaners and buying a train ticket are ethically acceptable if, but only if, all parties to the transaction take part willingly in awareness of ways in which others' proposed action will bear on them".[29]

It is undoubted that informed consent is not only a part of our everyday life but also an important concept in medical ethics. It acknowledges the right of the patient or the participant (in the case of genetic research) to have control over his or her own body. Procedures should be conducted on a patient's body only with his or her permission. This is also applied in the case of genetic research as very often the participant is required to provide personal genetic information.

In both clinical and research settings, informed consent is specific: informed consent is a research subject's affirmative agreement to participate in a research. Specifically, informed consent refers to the capacity of the patient to grant permission for the healthcare provider to proceed with diagnostic tests and medical treatments or for the researcher to proceed with genetic research (for therapeutic or non therapeutic reasons). Written informed consent is not required for routine medical care, rather for certain tests and for more complex procedures (surgeries etc). Verbal informed consent should be a part of standard practice in healthcare.

Before being permitted to request the consent of potential participants, a researcher must draft a document stating the research hypothesis, the research methods, and any possible risks or benefits

[29] O'Neill, O., "Some limits of informed consent", *Journal of Medical Ethics,* 2003, 29: 4 – 7

to participants. This document, known as a research proposal, is then submitted to a review board or in many cases an ethics committee for independent scrutiny of its ethical and scientific merits. If the board accepts the document then the researcher may start recruiting research participants following the purposes of the specific research as described in the document

Informed consent can be used as a means for the healthcare provider to communicate with the patient concerning the patient's medical condition and medical treatment options. This communication is vital as it assists the patient in becoming an active participant in his or her care. Verbal or written informed consent provides also a record for the healthcare worker and the researcher as well, regarding patients' or participants' understanding of any treatment course or research procedures as these procedures were approved from the review board. It is also a means for communication between the researcher and the participant concerning specific research procedures, or any other issues involved in the research.

To start with, we should ask: is informed consent an agreement, or is it a signature to a consent form, is it a formal approval given to participate in a research, or perhaps a mutual decision-making between a researcher or a doctor and the participant?

Informed consent can take several meanings. It can mean the consent document, or it can indicate the whole informed consent process. However, it is primarily important to distinguish between informed consent in a moral and informed consent in a legal sense. Let us call these two notions of informed consent "autonomous consent" and "effective consent". [30]

In general, the components of the "autonomous consent" are (a) disclosure of information (b) comprehension (c) voluntariness (d) competence (e) consent. Therefore, an autonomous consent can be acquired from a competent person to whom any relevant information has been disclosed, and who after adequately understanding the information, acts voluntary without any coercion, giving

[30] These two notions are mentioned as sense1 and sense2 in Faden R., Beauchamp T., *A History of Informed Consent*, Oxford University Press 1994 and Beauchamp T., Childress J., *Principles of Biomedical Ethics*, Oxford University Press, 1994 (4th edition)

the final consent. In this sense, "a person must do more than express agreement or comply with consent".

Therefore, in order for the informed consent to be, an action of "substantial autonomy", an "autonomous authorization" defined as "an autonomous action by a subject or a patient that authorized a professional either to involve the subject in research or to initiate a medical plan for the patient" it should include "substantial understanding in substantial absence of control by others"[31].

"Effective consent" requires the fulfilment of the criteria set forth in law or in a guideline and therefore it covers only any regulatory obligations. As "effective consent" is a policy-oriented kind of informed consent it can be translated as i.e. an authorization from a patient or a subject which is "effective" meaning that it "has been obtained through procedures that satisfy the rules and requirements defining a specific institutional practice in health care or in research".[32]

However, there are cases where legislation does not refer to a clear definition of informed consent, but it may only contain references to informed consent features. This definitely does not clarify what an "autonomous consent" is.

"Over 70 studies performed in a variety of clinical settings indicated that legally and institutionally valid consents and refusals had frequently failed to reflect genuinely autonomous decision making, hence result in genuinely autonomous choices. Low socio-economic status, poor education, old age, lengthy hospital stay, stress, language barriers and misinterpretation of probabilistic data were found in these studies to be associated with such outcomes".[33]

There are many cases where, "autonomous consent" is compatible with "effective consent". However, obtaining "effective consent" does not presuppose that an "autonomous consent" was obtained as well, or vice versa. It is very important that the requirements for "effective consent"

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[31] All the abstracts are from the Beauchamp T., Childress J., *ibid.* p. 143

[32] ibid, p.280

[33] Epstein M., "Why Effective consent presupposes autonomous authorization: a counter-orthodox argument", *Journal of Medical Ethics,* 2006, 32: 342 - 345

should overlap those concerning "autonomous consent" in order to avoid situations where consent in a legal sense is something different than consent in a moral sense.

4.2 The informed consent process

In order to investigate what exactly effective informed consent is, it is important to look at the informed consent procedure. Informed consent for clinical trials and for genetic research in general, involves more than just reading and signing a piece of paper. It involves two essential and equally important parts: the informed consent document and the process.

The actual document provides a summary of what the patient or the participant is consenting for. This covers any purposes, treatment, procedures, potential risks and benefits etc. Signing the informed consent document designates the official consent on behalf of the participant to be involved in a specific study. The document is designed to assist the informed consent process, including discussion between the participant and the researcher.

The informed consent process has as its aim to provide the future participant with ongoing explanations which may help the participant decide whether to participate or continue participating in a research. The informed consent document alone cannot ensure that the participant fully understands what participation means. Therefore, it is important for the participant to have the opportunity to ask questions and raise concerns as the quality of explanations given by the researcher may or may not help the participant to reach a decision.

Consequently, it is important to stress out that informed consent is – and should be - an ongoing, interactive process, rather than a one-time information session.

Both words "informed" and "consent" each one separately and both of them together, signify the informed consent procedure as vital for medical ethics. The word "informed" emphasizes the

significance of this procedure. That is the reason why we will examine first what the word "informed" contributes in the whole meaning of the principle.

4.3 The "informed" part of informed consent

A basic element of informed consent is the idea that somebody i.e. a potential participant to a research, ought to be provided with all the available information that is *relevant* to the decision concerning participation, in order to ensure an adequate level of awareness. The problematic part with the above idea is what constitutes an adequate level of awareness? How much disclosure is needed? How plausible a genuinely informed consent is? Can we assume that it is possible for anyone to be informed?

4.3.1 Disclosure of information

"Genuine consent is not a matter of overwhelming patients with information, arrays of boxes to tick or propositions for signature. (...) Genuine consent is apparent where patients can control the amount of information they receive, and what they allow to be done".[34]

In order to examine how much disclosure is needed to perceive a genuine consent, we should first ask why we seek informed consent. The degree of disclosure depends on how this question is answered.

Someone can answer that we seek informed consent because it is very important to ensure that "*the patient or subject will receive information that is personally material – that is, the kind of description that will permit the subject or patient, on the basis of his or her personal values, desires, and beliefs, to act with substantial autonomy*".[35] Therefore, it can be assumed that if a

[34] O'Neill, O., "Some limits of informed consent", *Journal of Medical Ethics,* 2003, 29: 4 – 7

[35] Faden R., and T. Beauchamp, *A History of Informed Consent*, New York: Oxford University Press, 1994, p.307

person is ignorant concerning specific information about the research, then that person is not able to provide an autonomous informed consent.

A second answer can be that we seek informed consent because we want to ensure that individuals are aware of any potential risks which can be harmful for their safety. Thus, it can be argued that if a person remains ignorant of important and crucial circumstances and details about a research then for the sake of that person's own safety, his/her consent cannot be conceived as valid.

Last but not least a third answer could be that, if we seek informed consent precisely because its procedure respects individual autonomy then one way of recognising this autonomy in a decision- making procedure is to respect individual choices. The meaning of "choosing" can be translated into people's way of expressing their desires and needs. Respecting the autonomy of individuals justifies their right to know, or not to know, their right to remain in ignorance.[36] This ignorance however is not unlimited as according to Mill's understanding of liberty, the liberty to remain in ignorance can only be limited in order to prevent harm to others.[37]

Let us assume that a research participant does not wish to be informed about any relevant information concerning a specific research. According to the third answer, we can argue that the participant is still treated as having the ability to provide genuine informed consent, as this consent is compatible with free decision-making. Therefore, if a participant does not wish to receive as much information can be concerned as adequate by the specific consent approach, then he/she can still provide genuine informed consent.

On the other hand, if we consider the case where a normal conscious person wishes to donate his/her DNA to science then according to the first two answers in order to avoid violating individual autonomy this person should be stopped. On the other hand, the third answer would suggest that preventing this person from giving his DNA sample violates individual autonomy.

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[36] The right to know or not to know is analysed in Chapter 5

[37] Mill's theory is analysed further in Chapter 5. For Mill's notion of autonomy see also Chapter 3

The above leads us to another question: whether it is possible to be genuinely informed of all benefits and especially risks in genetic research.

4.3.2 Is it possible to be genuinely informed?

It is clearly important for people to be genuinely informed, as researchers or scientists must provide information to enable i.e. future participants to make informed decisions about participating or not in a specific research. That is why researchers need to negotiate a delicate balance in the procedure of providing any information.

There are many authors supporting the idea that it is not possible to foresee the range of applications to which the genetic material may be used – and therefore this cannot guarantee against its misuse – especially concerning future uses.

However, the fact that it is not possible to foresee all future uses of the genetic material does not presuppose that seeking a genuinely informed consent is impossible. Nevertheless, is it possible in both 'narrow' and 'broad' sense of informed consent to be genuinely informed? In order to answer this question we need to examine how 'narrow' and 'broad' senses of informed consent can be defined.

4.3.3 'Narrow' and 'broad' sense of informed consent

In its 'narrow' and 'broad' sense genuinely informed consent concerns the specification of the conditions under which the research is held and conducted. When referring to a 'narrow' sense of genuinely informed consent, we usually refer to a specific genetic research in which the use of genetic material is limited only for the purposes of it exclusively. It is also defined by the conductors of the research as to what will happen to the genetic material collected for the research. It is important to specify whether i.e. genetic samples will be destroyed afterwards or never be used

again. In this case a genuinely informed consent is achieved by the most accurate description of the conditions and the procedures that will be followed concerning the specific research.

However, we should ask whether this description is enough? If we say that this description is satisfactory then we should ask for who is it satisfactory? Do the researcher and potential participant - with very different levels of background knowledge - equally understand the information which is disclosed?

Somebody may argue that it is not important to examine what future participants think concerning the information provided by the scientist. An idea may be that by giving information the aim of informed consent is fulfilled as on the one hand the participant is "informed" since general information – description is provided and on the other hand the participant is supposed to give his consent based on the provision of this information. We should therefore ask if not seeking for the participant's opinion is compatible with the principle of informed consent and additionally, if the role and contribution of the researcher stops in the description of the research or it extends further than that. These questions are analyzed further below in the chapter.

The 'broad' sense of informed consent refers to the case where an unspecified range of conditions is involved, in which genetic samples are possibly going to be used during or after the specific research. Obviously, in this way the potential participant is informed about the wide range of possible future uses of his genetic material. In this broad sense, informed consent is more difficult to be given in the sense that future participants should consent to a more broad research. As mentioned above, even in the case of 'narrow' informed consent future participants may believe that the information provided is not enough therefore in the 'broad' informed consent this may occurs more often as participants may feel the need to ask for feedback and assurance that any personal genetic information will be dealt as agreed.

Broad consent entails that participants agree that their samples may be used for a variety of future studies which it may not be possible to specify in any detail at the time of consent. Even if

the participants are informed that i.e. their samples are going to be used after the end of a specific research, the researcher has to clarify any details concerning participant's rights over their samples especially concerning any future use. There were cases where participants demanded the removal of their sample years after they gave it. Recently, there was the case of Icelandic database where lots of people gave their samples and when the first results from the analysis were known, people expressed their wish for their samples to be removed from the database.

Concerning the Icelandic database it was stated that "because it would take great effort, time and money to gain consent from every individual", therefore a coding system was adopted which made data anonymous and which was not possible to be reconstructed later except with a decoding key[38]. The main argument for not seeking explicit informed consent in the Icelandic database was the unidentifiability of the genetic data. The Icelanders were reassured that the data were going to be unidentified and therefore anonymity could be secured.

According to the Recommendation of the European Council of Ministers "an individual shall not be regarded as "identifiable", if identification requires an unreasonable amount of time and manpower". Therefore, data in the HSD (Health Sector Database) in Iceland were regarded as unidentifiable because of the sophisticated coding techniques employed[39]. One of the main areas of controversy concerning the HSD was the matter or not acquiring informed consent of individuals to be included in the database. *The assumption was made that each and every individual would be part of the database unless they formally opted out of it.*[40] Consequently, Icelandic database removed from an informed consent based database, to a more blanket-consent database, because of the coding method which was believed to preserve privacy and anonymity and therefore, it could easily avoid any ethical implications.

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[38] Santosuosso, A., "The right to genetic disobedience: the Iceland case", in Mazzoni,C.M., *Ethics and law in biological research*, The Hague: Kluwer Law International, 2002, 163-172, p. 2

[39] Árnason, V.,"Coding and consent: moral challenges of the database project in Iceland", *Bioethics,* 18 (1), 2004,p.31

[40] Hayry, M., R.Chadwick, V. Arnason and G. Arnason, *The Ethics and Governance of Human Genetic Databases,* Cambridge University Press, 2007,p. 56

However, although the Icelandic Act recommended that Icelanders will have the opportunity to opt out of the database and will be informed continuously about their right to withdraw from the database, this was not practically possible. Precisely because of the coding method used in the database, a sample was not possible to be withdrawn from the database because of its unidentifiability[41]. Thus, Icelanders provided their samples to the database without consent and without practically be able to control the luck of their own samples[42] or any other future secondary uses of their data. Data are supposed to be unidentifiable and therefore no feedback was given[43].

Nevertheless, it is very interesting to look at the ELSAGEN survey carried out in 2002 in Iceland. People were asked if they thought that consent should be sought each time biological samples are collected for purposes of genetic research. Fifty-seven per cent agreed that consent ought to be obtained. This indicates that people in Iceland did not fully comprehend the purposes of data collection in HSD. Additionally, according to ELSAGEN survey, there were also concerns regarding the operation of HSD. *Just over 10% of the respondents had taken certain measures to ensure their personal privacy. The two most frequent measures were to opt out of the HSD, and to have one's telephone number removed from the general telephone directory.*[44]

Summing up, as it can be assumed from the above in both cases of 'narrow' and 'broad' informed consent, the quality and the quantity of information plays a crucial role for both the researcher and the participant. As information is supposed to be a prerequisite for people making decisions concerning participating or not in a genetic research, the Icelandic case shows that it is possible for the participants to lack important pieces of information, even if theoretically they were provided with some information.

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[41] _____, "Gene privacy for Icelanders", *New Scientist magazine,* April 2004, Vol.182, Issue 2442

[42] Andersen, B., E. Arnason, and S. Sigurdsson, "Kafkaesque ethics for post-modern vikings?" BMJ, Aug.1999: comment on the article: R.Chadwick, "The Icelandic database – do modern time need modern sagas?"

[43] Kaye, J., H. H. Helgason, A. Nõmper, T. Sild, and L. Wendel, "Population Genetic Databases: A Comparative Analysis Of The Law In Iceland, Sweden, Estonia And The UK", *Trames*, Issue1/2, 2004

[44] Hayry, M., R.Chadwick, V. Arnason and G. Arnason, *The Ethics and Governance of Human Genetic Databases,* Cambridge University Press, 2007, p.57

Why participants do not possess important information concerning the research they are participating? Is it because the quantity of information is not enough? Or is it the quality, or perhaps both? If we assume that we have two Projects who investigate the same thing but in Project 1, the researchers provide potential participants with 8 written pages in which they analyzed in detail the aims, the scope and the limits of the specific project. In Project 2 researchers summarized all the above information in 2 written pages. Which of the two projects can be argued to protect adequately participants' autonomy? The following part discusses the problematic of how much information is needed.

4.3.4 Dealing with the information

According to the Helsinki Declaration, when obtaining informed consent the information must be adequate, comprehensible and potential participants are to be informed generally about the purpose and design of the study, what it means to participate, voluntariness with regard to participation, and the option to withdraw.

However, what is mentioned in the Helsinki Declaration may raise various questions: According to what criteria information can be judged as adequate? Who is going to decide that it is comprehensible, and in what degree? Is the researcher able to tell when a participant adequately understood all the necessary information to give consent? It is undeniable that the communication between the participant and the researcher sometimes may be difficult most probably because of their different backgrounds, but can we argue that it is impossible?

As Diana Dutton argues that *"experts are more likely to suffer from what has been called specialized blindness: the tendency to define issues in the narrow and technical terms of their own specialty and to ignore related non-technical problems"*. [45]

This "specialized blindness" which is described above, can be easily characterized as natural as the researcher is trying to explain facts very well known for him, using a vocabulary with which he is familiar. The question is whether this "blindness" can be an obstacle for a robust communication and dialogue between the scientist and the future participant.

Since it is vital potential participants to use the information provided and take an active part in decision making, then extra attention should be paid concerning the quantity, quality but most important the formatting of information. There are people that tend to understand more easily written information compared to oral, or vice versa and also some people may require further technical details, whereas others do not. Additionally, there are studies which indicate that participants consent to participate without even read the information provided.[46]

It is true that medical research largely depends on the trust public has in researchers and research settings. On the other hand, participants tend to rely a lot in this trust and sometimes do not bother to read any information given by the scientist because they feel they can trust him. However, Onora O'Neill supports the idea that trust does not depend solely on the form of consent used in research settings as upon the way the participants are treated.

"Paper trails (...) are ideal from the point of view of administrative quality assurance and provide good defence against possible litigation. (...) But secure paper trails may not reassure or secure the trust of patients, donors or relatives who are asked to consent. (...) Those who give tissues may not be looking for a consent process in which they tick many boxes (...) They would

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[45] Dutton, D. B., "Medical Risks, Disclosure, and Liability: Slouching toward Informed Consent", *Science, Technology & Human Values*, Vol. 12, No. 3 / 4, Special issue on the technical and ethical aspects of risk communication (Summer – Autumn, 1987), p. 49

[46] Hoeyer K. "Science is really needed—that's all I know. Informed consent and the non-verbal practices of collecting blood for genetic research in northern Sweden", *New Genetics and Society* 2003, 22:229–44

probably prefer a process that provides real evidence that they can choose or refuse to give tissues for research". [47]

The potential participant has to deal with information whose context is medicine and is not easily understood. Furthermore, the informed consent procedure can be even harder when tailoring this information-giving process to the participant's needs and abilities. It is therefore absolutely essential not to undermine participant's trust for the benefit of one research.

It is unquestionable that it is impossible to ask from the scientist to forget that he is an expert and that he knows his profession perfectly well. However, even if the scientist manages to put himself in the position of ignorance – in which the future participant is – it is not guaranteed that the *blindness* will not appear again.

The debate over how to balance these sometimes conflicting issues continues, but two guidelines are emerging which may bridge the difficulties. First, obtaining informed consent guarantees that the right of individuals are always respected, and that there is no risk of enrolling people against their will. And second, as mentioned, the consent process can incorporate practices which are compatible with each person's abilities and necessities. Solutions which bridge any difficulties regarding communication between the researcher and the potential participant, offer ways to combine respect for individual autonomy with respect for individual capabilities.

However, do we presuppose that the information provided from the researcher contains all the adequate and comprehensible data necessary for decision making or it also encompasses information of minor importance not essential to making a rational decision which may confuse and disorient the future participant? If we assume that the researcher provides sufficient factual information how is this factual information given?

In order to answer the above questions we have to take as granted the fact that the quality of communication between the researcher and future participant is the key to a successful outcome.

[47] O'Neill, O., *Autonomy and Trust in Bioethics,* Cambridge University Press, 2002, p. 157 - 158

We count as a successful outcome a decision on behalf of the participant which supports his individual autonomy.

In theory to achieve an ideal fully informed consent, the researcher should provide information in a way which is compatible with the capabilities of each individual potential participant and taking also into consideration the individual *comprehension time,* meaning the time each person needs to comprehend a specific set of information. In this way, all future participants will have the opportunity to not only to comprehend easily the information but also be able take a decision which reflects to their values and beliefs.

However, very few of the researchers may seem "to have any appreciation of the need to assess potential subjects' understanding of what they have been told". [48] Therefore, researchers are not in a position to either elucidate any misunderstandings, or in many cases through the discussion reveal some information needed for the participant's decision to participate or not.

Nevertheless, as Angela Hall points out, the core objectives of communication process are constituted by three stages. If emphasis is given in these three stages, then communication problems can be overcome with a much better outcome. The stages are: a) information gathering b) relationship building and c) explanation and planning. The last stage is the most important as we can claim that b) and c) are in a way interwoven with each other since the whole participant-research relationship is built through the whole informed consent procedure.

The problem is how the above stages can be practically applied. Future participants in genetic research can be divided in two major groups. The first group includes those who belong to the general public who do not have an explicit knowledge and the second group includes those who belong to a more expert public which happens to be more familiar with any scientific terminology.

Specifically, people who have a continuing medical problem – or happen to have relatives and friends with a medical problem - tend to be experts as a result of their greater involvement in

[48] Hall, A., "The role of effective communication in obtaining informed consent", *Informed Consent in Medical Research* ed. Len Doyal and Jeffrey Tomas, BMJ Books 2001, p. 292

medical decision. People in this group gain their health literacy attempting to reduce their anxiety and fear concerning their medical situation. Consequently, people have different expertise and this can affect their comprehension of information.

Very often researchers in order to give emphasis on information they tend to compose very long consent forms. This can be counterproductive, however. Subjects, even when they can read the material easily, will often not be willing to read i.e. a 10-page consent form.

If the form is necessary to be long due to the complexity of the research, researchers have to make sure the process includes sufficient time to read the information, and include procedures that will ensure participants have actually read and understood the consent form. The greater the risks the research poses, the greater the ethical responsibility the researcher has to provide information in a way that will allow it to be understood.

However, not only the information provided should be able to satisfy both groups (general and more expert public) but also both groups should be able by the end of informed consent process to fully understand what the specific research project is about and be able to take an informed decision concerning participation.

At the end of the day, it is a matter of ensuring that informed consent does not manipulate, coerce or deceive participants but provides sufficient ground for voluntary choice. If the information with which the researcher and the potential participant deal is of consistently high quality then definitely the benefits are obvious. In genetic research – a research which can reveal information about an individual's susceptibility to disease and hence about the individual's future health – informed consent plays a crucial role[49] .In the case that the individual decides to consent, then this high quality information can guarantee better cooperation concerning the procedures of the specific research.

[49] Genetic information is analysed further in Chapter 5

4.4 The "consent" part of the informed consent

We cannot ignore the fact that "informed" and "consent" parts of informed consent are linked to each other. While the "informed" part addresses questions concerning the adequacy of the information provided and the competence to understand the information given, the "consent" part has to do mostly with the capacity for competence correlated with decision-making. Particularly important, is also the voluntariness characterizing this decision, as it indicates that the process of informed consent must be free of coercion or undue influence.

The aim is clearly not an informed consent for every detail but genuine consent for research as a whole. Genuine consent and control require accurate basic information, the availability of any additional information to ensure lack of deception and most important the opportunity to withdraw consent to ensure lack of coercion.

Given the complexity of biomedical research, it is obvious that people cannot gasp the meaning of all relevant information. Onora O'Neill condemns those overly complicated and detailed consent forms that have been introduced allegedly for purposes of protecting autonomy.[50] Consent as an element of genuine informed consent is construed as an authorization which has as a reference point a specific research and involves all the elements the participant is consenting for.

What a competent person is consenting for depends upon what was proposed for his consent by the researcher. Thus, how can we define "competence" in the context of informed consent?

4.4.1 Capacity for competence

Competence refers to the ability of potential participants to give informed consent compatible with their own values. It also involves the ability to comprehend the information which is presented by the researcher, to realize any possible consequences of a decision and eventually provide a free and informed consent. This ability of understanding the information may depend on the

[50] O'Neill O., "Autonomy and trust in Bioethics" ibid, p.157

circumstances surrounding the decision or the time in question. In addition, competence is not a static condition as it may be temporary or permanent. Most important, competence does not require potential participants to have the ability to make every kind of decisions. Generally, it does require that participants are competent to make an informed decision concerning participation in a specific research.

In the UK, as in many other European countries, the law regards everyone over the age of 18 as competent unless there are compelling reasons not to, e.g. if the person is unconscious or under the influence of drugs etc. Despite the fact that problems of competence among vulnerable groups are not discussed here - as this part of the chapter aims to a more general analysis of competence as a part of informed consent – it has to be mentioned that such problems exist especially where the environment limits opportunities for individual choices i.e. prisons "*tend to encourage the development or enhancement of coping skills that rely upon the use of manipulation and coercion. The more powerless and unable to influence their own circumstances people feel, the more likely they are to resort to increasingly desperate measures in order to feel as though they have some control over their lives. In the case of women in prison, this often results in women resorting to self-injurious behaviour*"[51]

As the above reference shows the environment of the prison affects a lot prisoners' psychology and therefore, this can influence their capacity to act and choose autonomously.

It is important at this stage to examine the difference between comprehension and competence. Comprehension signifies the understanding of any disclosed information during the informed consent procedure whereas competence signifies the ability to make independent decisions concerning participation.

An important element of comprehension is comprehension time. Specifically, comprehension time is a specific and unique chronological point in the informed consent procedure, in which each

[51] http://www.elizabethfry.ca/phase2/policies.htm

potential participant may comprehend the information given. It is unique because individuals tend to understand information make decisions in their own individual and personal time.

Competence presupposes comprehension as the informed consent principle requires potential participants to understand all the information provided in order to be able to take an autonomous decision. The researcher always has to remember that participants in research are "people, who do not like to appear stupid and say they do not understand an explanation that has been provided".[52]

Sometimes it is possible for participants to say that they understand an explanation when in fact they do not. Consequently, the obligation and the commitment to explain everything in a non-technical language are imposed on researchers, in order for the participant not only to comprehend the information, but also to become conscious of any relevant detail.

On the other hand, comprehension does not presuppose competence. There are cases where individuals may fully understand all the information concerning a specific research but for various reasons they are incapable to make an independent decision i.e. they are strongly influenced by relatives, by personal fears etc

Amnon Goldworth suggests that "*what is needed (...) is a way of dispelling irrational fears that serve as obstacles to autonomous choices*".[53] Comprehending the information provided from the researcher encompasses at the same time that future participants will have the opportunity through discussion to get over those fears in order to reach a decision. "Irrational fears" may be detected in all kinds of research but they exist especially in genetic research because of the delicate matters that are at stake i.e. personal information, information about other family members, genetic discrimination etc.

However, we have to ask what fears can be categorized as irrational? What is the connection between informed consent and these fears? Is it possible for the participant after understanding all

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[52] Munson R., *Intervention and Reflection: basic issues in Medical Ethics*, Wadsworth/ Thomson Learning 2000 (6th edition), p.476

[53] Goldworth, A., "Informed Consent in the Genetic Age", *Cambridge Quarterly of Healthcare Ethics, Vol. 8, Issue 3, July 1999, p. 393 - 400*

the information given and signed the informed consent form not to fear or worry about anything else concerning the research?

Regarding the rationality of fears I will not refer to specific philosophical theories concerning what rationality is, rather I will try to examine what we mean by "genetic fears". Are these fears sometimes rational or irrational? Can we argue that the participant in a genetic research always gives his/her informed consent free from any fears or worries?

It is undeniable genetic progress is very rapid in such a way that many people find it hard to follow. They hear about new genetic developments through their television or read about them in the newspaper. The majority do not have a personal contact with geneticists or genetic researchers. Many of the people as they are not familiar with new genetic technologies or genetic testing find it really hard to understand their application and implication in everyday life.

Consequently, it is natural for people to fear several things i.e. that if they participate in any genetic research this means that they may lose their privacy etc. Arnold Relman argue that "*it seems paradoxical that scientific research, in many ways one of the most questioning and sceptical of human activities, should be dependent on personal trust. But the fact is that without trust, the research enterprise could not function*"[54].

Although participating in a genetic research does not mean that your individual autonomy is in danger or that it is possible to discover your predisposition to a genetic disease, people are afraid that something unexpected may happen. As people do not feel familiar with genetic research in general, it is possible to develop some *fears* about any implications the participation in a genetic research may have. People do have genetic fears that exist in their minds and it is very difficult for these fears to be extinguished in one day as they are based on personal beliefs.

The informed consent process is an opportunity for the researcher to approach the future participant and through discussion attempt to dispel those fears explaining in detail any ambiguities

[54] Crease R., "The paradox of trust in science", *Physics world,* 2004

the participant has concerning the research itself, in order to bridge any obstacles leading to autonomous choices.

4.4.2 Voluntariness

Voluntariness requires that consent be obtained under conditions that are free from coercion, undue influence and unjustified pressures. Free and voluntary informed consent presupposes participants who: consent without manipulation, have freely agreed to participate in the study on the basis of well – understood information concerning the objectives of the research and are *theoretically* fully informed of all risks and possible benefits. (As it was mentioned before in this chapter it is very difficult to predict all possible risks and benefits). Consent must be obtained under conditions that do not involve explicit or implicit coercion that place limits on the freedom and voluntariness of participation.

It is interesting at this point to examine the Aristotelian view on voluntariness. Aristotle trying to identify the meaning of "voluntary" refers to any of the things in a man's own power which he does with knowledge. A man acts voluntary when he does not ignore (*μη αγνοών*). [55] It is undeniable that Aristotle proclaims acting in knowledge over acting in ignorance. But what happens if a man chooses – for personal reasons - to stay in ignorance, to act without any knowledge?

Despite the fact that Aristotle does not answer the above question directly, he seems to divide this kind of actions in two categories: actions due to anger or passion and actions because of choice. He seems to believe that if a person acts in ignorance doing something that is harmful to others, but his action is due to anger (or any other human passion), then this act is unjust but the doer is not because the harm is not due to vice.

Aristotle introduces in Nicomachean Ethics a more humanitarian approach to acts in ignorance. He seems to believe that actions due to passions are not synonyms to vice actions. This

[55] Aristotle, *Nicomachean Ethics (Book 5 [1135a]20)*

Aristotelian view is very interesting as many bioethicists nowadays tend to characterize acting in ignorance as ethical or unethical and this characterization refers at the same time to the person who is acting. Perhaps this kind of characterizations deserves a second thought. As Aristotle proposes, by differentiate the action, from the person who acts, we transfer the focus not to the act itself rather to the person who acts.

Additionally, Aristotle gives great emphasis on "choice". According to Aristotle, *if a man harms another by choice, he acts unjustly; and these are the acts of injustice which imply that the doer is an unjust man.*[56] He categorizes injustice actions as the actions that can cause harm. According to Aristotle whoever *chooses* to cause harm by acting in a specific way then he is an unjust man. However, there are some actions that may cause harm but are excusable. *For the mistakes which men make not only in ignorance but also from ignorance are excusable.*

It is interesting to look at the difference between mistakes *in ignorance* and *from ignorance*. To examine the above we should look at the ancient text to achieve a better approach. In ignorance is written as «αγνοούντες» which can be translated as those who ignore things and from ignorance is written as «δι' άγνοια» which can be translated as "because of their ignorance".

In the first case Aristotle seems to refer to the knowledge status of a person. Whoever ignores things is an "αγνοών». In the second case Aristotle seems to refer to the mental status of a person. A person who "is in ignorance" but because of his ignorance he cannot understand some important things. Aristotle seems to imply that in this case the person «αμαρτάνει» (literally 'making a sin', metaphorically 'is acting unjustly') because (δια) of his ignorance – because of his mental status which does not allow him to understand.

Despite the fact that Aristotle himself does not clarify what are the premises to distinguish a person who acts unjustly «δια άγνοιαν», this gives clearly the message that ignorance may lead to unjust acts. As mentioned above, Aristotle seems to concern this kind of actions as excusable but it

[56] ibid

is clearly important to take into consideration that these people need special treatment and definitely a greater effort to elaborate the information they receive.

Very often having the information that is relevant to the research - what Angela Hall calls "information gathering" - is not enough to entail a final and fully conscious decision about participation. Instead, subjects must adequately apprehend the relevant information and give their genuinely autonomous authorization freely. This constitutes a clear demonstration of a subjects' own will and determination to face any risks involved, without being coerced by any other reason or attitude. The decision is taken by the potential participant and in no other case is genuine unless it is voluntary and expresses his own ideas, values and beliefs.

Furthermore, it is important to give people the right to withdraw from their participation in a study at any point. This implies the need for researchers to ensure that they have participants' ongoing consent to be involved in a study and that the researchers recognise a possible desire on behalf of the participant to opt out of a study. This can be sometimes problematic in relation to some groups who might be reluctant to state they don't want to continue being involved with a project i.e. children. The same issue can apply in situations where people because of the power relation that exists between the researcher and the participant find it difficult to say no to something they have previously agreed to.

It is true that the researcher's position is more powerful in relation to the subjects' position. It is powerful in the sense that, the researcher is the information holder whereas the potential participant is the information receiver and the information seeker. Consequently, the participant may unconsciously develop notions such as it is an obligation to consent to the researcher's requests because otherwise he will disappoint the researcher. Moreover, people may think that since the researcher is a scientist he *will know better* and not suggest anything that can cause harm.

Milgram's experiment "Obedience to authority study" constitutes a proof of the above: "*The legal and philosophic aspects of obedience are of enormous import, but they say very little about*

how most people behave in concrete situations. I set up a simple experiment at Yale University to test how much pain an ordinary citizen would inflict on another person simply because he was ordered to by an experimental scientist. Stark authority was pitted against the subjects' (participants') strongest moral imperatives against hurting others, and, with the subjects' (participants') ears ringing with the screams of the victims, authority won more often than not. The extreme willingness of adults to go to almost any lengths on the command of an authority constitutes the chief finding of the study and the fact most urgently demanding explanation"[57].

Milgram concluded that when people are ordered to do something by someone they view in authority, most will obey even when doing so violates their consciences.

These problems of consent and voluntariness between the researcher - potential participant and the physician - patient, should be treated very carefully. Researchers have to evaluate these problems and take steps to eliminate them, by encouraging discussion, bridging the gap between the two parties. Consent must be obtained under conditions that do not involve explicit or implicit coercion and at the same time do not place limits on the freedom and voluntariness of participation.

Very often in the literature concerning informed consent the researcher and the participant are metaphorically characterized as "partners". Obviously, partnership is translated here in terms of mutual understanding and co-operation between the researcher and the participant. The term "partnership" comes from economics. In economics partnership is a popular and useful form of business organization. A partnership is an association of two or more persons formed to carry on a business for profit. It is interesting to examine whether we can draw a simile between economics and bioethics – specifically genetic research – concerning partnership.

In economics there are three forms of partnership: the general partnership, the limited liability partnership and the limited partnership. From these three forms of partnership, we would give emphasis to limited partnership as it is closer to partnership as it defined in genetic research.

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[57] Milgram, S., "The Perils of Obedience", *Harper's Magazine,* 1974, http://home.swbell.net/revscat/perilsOfObedience.html

A limited partnership is a partnership with one or more general partners and one or more limited partners. The general partners are by law given the authority to manage and control the partnership. The general partners are also fully responsible for all debts and liabilities of the limited partnership. The limited partners are not responsible for any of the debts and liabilities of the partnership unless the limited partners participate in the management of the partnership.

Having in mind the limited partnership it is interesting to examine the similarities with the partnership between the researcher and the participant. Let us assume that the researcher is the general partner who by law has the authority to manage and control the research. The participant is the limited partner. The researcher is fully responsible for a possible misuse of genetic information, or any violations of the consent agreement. Signing the informed consent form creates an engagement between the participant and the scientist, meaning that the scientist has to handle participant with respect and give feedback (or not) – depending on what it was agreed from both parties - concerning the progress of the research. This engagement is very fragile as it is founded on mutual trust, understanding and respect.

As in financial partnership the above three elements play an important role. The participant trusts the scientist and gives the informed consent, and if a parameter needs to change the two engaged parties needs to come in touch again with respect and understanding.

However, the question is whether in genetic research partnership, both parties (researcher – participant) carry the same amount of responsibilities. Having in mind the case of limited partnership can we argue that the participant as a limited partner is not responsible for any of the liabilities of the partnership? Does this place great weight of responsibility to the general partner – the researcher?

In terms of responsibilities, for the participant the greatest responsibility is to take the decision and consent to participate in a research. Other secondary responsibilities entail participants to show up for any projects they agreed to participate. If the research involves providing the

researcher with information, the participants are expected to provide as accurate information as they can. Providing inaccurate information can distort the data being collected and perhaps lead to inaccurate conclusions. Additionally, if participants decide to drop out of a study, they should also let the researcher know about it as soon as possible. Also, if participants have any concerns about their condition, particularly if they think that they may be feeling ill as a result of participating in a research, then they should let the researcher know. This allows the research team to evaluate any unexpected risks and take steps to deal with them if necessary.

Both general and limited partners (researchers – participants) come from a different background and consequently communication can be difficult. They carry different weight of responsibility and it is not only through their fruitful and effective co-operation that the research can be conducted. The fruitful interaction between the two potential partners has as a first step the informed consent procedure. In this vital first step the cooperation and understanding among the partners is founded. It is also important that the researcher relies on participants' honesty, just as participants expect the researcher to be honest with them.

Summing up, it is important to remember that informed consent procedure is the key to respecting autonomy and that is because it can provide a kind of guarantee that no deception or coercion will be attempted on behalf of the researcher. The researcher should provide any relevant information to potential participants in a language and vocabulary level that the participant can understand, free of bias or pressure. It is very crucial to consider that participation in a research must begin as a voluntary action and remain voluntary.

4.5 Justifying informed consent

Despite the fact that we analysed the content of informed consent above, to achieve a more pluralistic perspective we have to examine how exactly it is ethically justified. Why are we using it and why it has such a great value and importance?

In order to validate the status of informed consent as a fundamental principle correlated with its use in research involving human subjects we divide its justification in two types of reasons: consequentialist and non-consequentialist.

Consequentialist reasons claim that the only basis for judging the morality of an act is by its consequences regarding all affected people. If we have to choose between two kinds of action then according to consequentialism it is morally right to select the action which has the best outcome. Choosing an action should not be done with self centred criteria, rather with criteria which serve the general human welfare.

Arguably, consequentialism will suggest that seeking informed consent is morally right as it has the best-expected consequences compared with any alternative solution applied. Specifically, in genetic research seeking informed consent from people concerning their willingness to participate in research not only proclaims their own adherence to the protocol, but also it gives a great value to the research itself. In this way it helps the participant to feel more secure and that his own will, opinion and values count a lot. In any other way, i.e. without informed consent it is possible that participant's confidence in research as an enterprise be undermined.

Additionally, informed consent can be considered as a moral right act as it manages to advance public trust in the research community. This is vital because it engages people in a way of thinking which benefits solidarity and a duty to participate, and obviously without those fundamental elements the research enterprise could not flourish.

However, despite the fact that when conducting a genetic research seeking informed consent is fundamentally important, there can be cases especially in epidemiology where research and experiments can be conducted without consent and save thousands of lives. For example, if we assume that Experiment 1 - if successful – can save thousands of lives, but the only way to be conducted is by using coercion to enrol several people, then coercion in this example would be morally right according to consequentialism, since it will have a significant benefit for lots of

people. It is true that when dealing with experimentation and research no-one is able to guarantee a positive outcome. Scientists refer to statistical possibilities, however if a research is never conducted then these possibilities would remain numbers and graphs.

Conducting a genetic research without informed consent can be legally impermissible. However, consequentialism supports that a specific action is impermissible when, and only when, this action leads to worse consequences than other available acts. Therefore, if a researcher decides to get DNA samples from people without their consent because he believes that he has a project which according to his own opinion it can be beneficial for many people in the future, then his action is morally right and therefore permissible.

Nevertheless the question is whether making an attempt to predict or to foresee consequences, and therefore basing our decision on this attempt, is a trustable method to make moral decisions.

Based on the above way of thinking, it can be argued that according to consequentialism the Tuskegee Syphilis Experiment was morally right. Between the years 1932 – 1972 the U.S. Public Health Service conducted an experiment on 399 black men in the late stages of syphilis.[58] The true nature of the experiments had to be kept from the subjects to ensure their cooperation. The study was meant to discover how syphilis affected blacks as opposed to whites. The hypothesis was that whites experienced more neurological complications from syphilis whereas blacks were more susceptible to cardiovascular damage. After forty years it was announced that "nothing learned will prevent, find, or cure a single case of infectious syphilis or assist the basic mission of controlling venereal disease in the United States". [59]

The subjects of the experiment were informed that they were being treated for "bad blood" and their doctors had no intention of curing them of syphilis at all. For the experiment, the data was

[58] Jones, J.H., "The Tuskegee Syphilis Experiment", *Bad Blood,* New York: Free Press, 1993
http://www.infoplease.com/ipa/A0762136.html
[59] ibid

to be collected from the autopsies of the men, and they were deliberately left to degenerate under syphilis which can cause tumors, heart disease, paralysis, insanity and death.

According to consequentialism, since syphilis is a serious disease and this experiment the year it started it was assumed that it may provide scientists with some information on how syphilis reacts on black people and therefore provide a better treatment, thus it was morally right not to disclose to the subjects the nature of the experiment.

On the other hand, non-consequentialist reasons requiring the use of informed consent are mostly founded in inherent capacities of human beings e.g. in the belief that people have the inherent capacity of self-determination and consequently in respect of this principle, subjects' decisions must be respected. Thus, nobody has the right to influence or mislead subjects concerning their decision to participate or not in a research.

Furthermore, as Kantianism – the theory opposed to consequentialism – supports, consequences have moral significance only if we examine the intention of the moral agent (the person who acts) and his/her moral principles which rule the action itself.

Kant argues that the idea of morality is a categorical imperative, and that is a rule that applies to all people because of our rational nature, at all times and circumstances. The most important formulation of the categorical imperative is: "So act as to treat humanity, whether in your own person or in that of any other, always as an end and never merely as a means".

This presupposes that all people have an equal moral worth, thus people should not be treated merely as means but as ends. For Kant it is not morally right to take advantage of people, even if there is the possibility to help a greater number of people. Even if an experiment requires the sacrifice of one single person to save fifty others, Kant will oppose to such an action as it treats a person as a means and fails to recognize the moral worth of that person.

Kant's vision of the perfectly moral society is one that is a "kingdom of ends," one in which there are no superiors or inferiors, and each person recognizes the equal and moral worth of every

other person. A person may choose to volunteer for a genetic research, but this is morally acceptable if and only if the person freely consents. Therefore, the principle of genetic research that a person must give free and informed consent follows from the Kantian idea that persons have a moral value and dignity and must be treated as ends and never as means, provided that the research itself does not impinge upon other's people autonomy.

The non-consequentialist rationale provides strong bedrock for the principle of informed consent, since it requires the researcher to protect subjects' informed consent irrespective of the expected consequences of doing so. In complement of the above, it is not enough for the researcher to only perceive an adequate risk/benefit profile of the potential participant, but also to aspire to the establishment of a genuine partnership between him and the participant and preserve it along all stages of the research.

4.6 Informed consent and autonomy

As this chapter examines the essence of informed consent consisting of the elements of informed consent along with several philosophical theories which attempt to ethically justify informed consent, the discussion leads us to analyse further a principle which strongly underpins informed consent: autonomy.

If we consider autonomy to be something intrinsic to human beings, then autonomy appears to be somewhat similar to dignity as both are founded in Kant's categorical imperative. To respect autonomy means to respect people and how they choose to live their own lives. The exercise of autonomy gives people the right to control their own body.

It is undoubted that we make choices everyday but what differentiated choices in medical research and in medicine generally, is that these choices has to do and have a direct impact on our own body. Autonomy requires that individuals be offered choices concerning significant decisions in their lives.

However, respecting one's choices does not automatically give rise to the obligation to promote these choices by informing i.e. future participants in a genetic research. Nevertheless, if consent in medical research and medicine is so important then it would be irrational not to act in a way which ensures it, especially since an ordinary person sometimes is unable to grasp the meaning of various epistemological terms without the help of the researcher.

Thus, in order to be a genuine product of autonomy, consent – an autonomous authorisation should be informed. Since the practice of medical research and medicine evolves rapidly and creates choices of the highest importance for most people, it must be required that these choices should be respected and promoted.

Conclusion

In the beginning of our analysis we made a distinction between "autonomous consent" and "effective consent". We concluded and we will conclude in later chapters that the requirements for "effective consent" should meet those for "autonomous consent". The elements of "autonomous consent" are based primarily on disclosure of information and consent.

As Graeme Laurie mentions "our opinion on how much information a person must receive before consenting, whether or not he really wants to receive this information, is a result of the balance struck between autonomy and paternalism".[60] On the one hand, those who feel that they have to protect people from their own actions tend to use more paternalistic arguments, whereas on the other end there are those who believe that people should be let alone to decide for themselves freely and autonomously.

It is undoubted that the responsibility to protect and inform research participants lays ultimately with the researcher and cannot be ignored or delegated. The principle of autonomy entitles that each person should be given the respect, time and opportunity to make his/her own

[60] Laurie,G.T., *Genetic privacy: A challenge to medico-legal norms*, Cambridge University Press, 2002, p.194

decisions. Potential participants must be given the information they will need to decide without any coercion or pressure to participate.

CHAPTER 5 – THE RIGHT TO KNOW

When researchers ask for consent, they are asking for voluntary agreement to take part in a research study. Informed consent means more than signing a consent form. It means that a participant knows about the benefits and risks of the study. A participant must know that he/she is free to take part or not, and that your decision will not affect your health care now or in the future. The research team should give the future participant all the facts needed to make personal choices.

In the case of data mining - the application of statistical methods to potentially quite diverse data sets, in order to accumulate data about real or hypothetical entities (such as a person, a group of persons, a commercial enterprise, or other entities or events) - a crucial role plays the provision of information - and when data mining concern genetic databases – the provision of genetic information. As the data pertaining to individuals may be specific to an identified person; may be anonymized by removing direct identifiers such as name, address, or social security number; or may be aggregated over geographic, demographic, or other variables, it is very important that these data are collected under a specific agreement or understanding as to how they will be used.

Therefore, the rapid development of data mining using statistical analysis, genetic testing and screening techniques which can provide an increasing amount of genetic information has raised several conflicts in terms of 'rights' to know or not to know.

Following the discussion on informed consent and as the last part of this thesis is my analysis concerning Cyprus strategy on thalassemia based on premarital testing[61], in this chapter I will argue that it is not a matter of adopting (or not) the idea of collecting genetic information - having the right to know or not to know – but rather of each person individually to understand the option to choose between knowing and not knowing, examine whether he/she really needs this information, and whether he/she is ready to accept them and take any responsibilities involved.

In medical literature the "right to know" as a principle is well defined by several authors and some very clear lines are drawn concerning the characteristics of what is defined as the principle of people's "right to know".

In the context of genetics, the right to know can be identified as a right to genetic information. Genetic information is arguably the most important and delicate information as it concerns DNA, considered by some to be the safe keeper of our own very personal and unique genetic

[61] See Chapter 6

characteristics[62]. Therefore, genetic information is highly valued and raises several issues concerning access to genetic information: either by individuals or any other third parties –relatives, future partner, insurance companies etc.

5.1 The basis of the right to know

5.1.1 Information and knowledge

Before proceeding to further analysis of the right to know, a distinction between information and knowledge must be made. In philosophy "knowledge" is taken to mean a belief that can be justified as true to an absolute certainty. Philosophers often define knowledge as a justified, true belief. However, there is a debate on how a belief can be justified as true. My purpose in this chapter is not to seek for an accurate definition[63] of what "knowledge" is, but rather to draw a distinction between genetic information and genetic knowledge.

Information is a collection of data[64] from which conclusions may be drawn. However, defining genetic information may be problematic as it has to be examined whether all medical information can be considered as genetic. Is genetic information fundamentally different from any other kind of medical information?

Many studies have shown the correlation between various disorders and some genetic components. Diabetes until recently was not considered to be genetically inherited; however in diabetes Type II (which is characterized by resistance of the body tissues to the action of insulin) patients whose mothers had diabetes are twice as likely to get the disease as those whose fathers had it. Therefore is it possible to consider medical information as genetic information as well?[65]

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[62] For further comment on genetic information see also Chapter 2

[63] For further references concerning the background around the debate of knowledge see also Hume, D., *The treatise of Human Nature*, Oxford University Press, 1978

[64] We can characterize as data the stream of raw which if organized constitute information.

[65] For the definition of genetic information see also Chapter 2

These questions increase the difficulty of constructing a definition of genetic information. Do we construct a definition having in mind any interests on behalf of third parties (health insurers or any other affected entities) since in the case of insurance companies the family history is considered to be an important issue? A broad definition of genetic information may cover genetic test results of individuals or family members, family history, data about genetic testing, inherited characteristics, or asymptomatic and presymptomatic conditions. Depending on the criteria, this definition can be narrowed based on what we value as highly important in order to be protected. In this chapter we refer to genetic information having in mind its broader definition as we believe it covers more sources from which genetic information can be extracted.

5.1.2 The desire to know

The question is: why do people seek genetic information? Is it because they want to feel secure about their lives? Is it because information can be linked with security and relief, and a sense that they can be in control of their lives and future? Is knowledge able to offer security, and if yes in what degree?

Security can be an important reason why people tend to seek genetic information. They want to feel secure and they consider the possession of any relevant information a means through they can conquer this security e.g. having the capacity to know a possible predisposition to a specific disease. The question is whether someone can be sure? Do people realize that someone can never be sure since sometimes even the most precise genetic tests can give wrong answers?

A. J. Ayer when examining the essence of knowledge, he refers to the difficulty of defining how someone can be sure. Although he does not refer to genetic knowledge but in knowledge in general, his argument on the right to be sure is very interesting: The sufficient conditions of knowing are that *what one is said to know be true, secondly that one be sure of it and thirdly that one should have the right to be sure. This right can be earned in various ways; but even if one*

could give a complete description of them it would be a mistake to try to built it into the definition of knowledge, just as it would be a mistake to try to incorporate our actual standards of goodness into a definition of good[66].

Applying Ayer's way of thinking linking the 'right to be sure' with the 'right to know' we can argue that firstly, we cannot talk of a 'right to be sure' rather than the need to feel sure. The feeling of security should not be considered a moral right but it can be a motive and it can explain why people want to preserve and exercise their right to know. Secondly, since everyone has different personal standards and different perceptions, genetic knowledge cannot give to all people the same degree of security. It can be argued that the majority of the people want to feel secure, and this can be promoted as a general idea in some campaigns to gain solidarity for a specific purpose e.g. persuade people to undergo screening for a specific predisposition or trait.

However, we should take into consideration that there are different perceptions of security and risk among people, social groups and populations. People's translations of Ayer's 'right to be sure' have to do with their own perceptions of risk. Seeking security, the right to know and the right to take uncoerced decision, to take risks or no risks, are some factors which are linked with each other.

In many cases people combine receiving information – knowledge, with developing a sense of 'security' concerning their health condition. They believe that in this way they can avoid the danger to face any unexpected circumstances and implications. In the cases which people consent to take genetic tests and choose knowledge instead of ignorance, they feel that by 'knowing' they can be prepared.

On the other hand, there is the possibility that someone may consider the actual test as a small risk compared to other risks he may face if not taking the test. Consequently, how someone

[66] Ayer, A.J., *Knowing: Essays in the Analysis of knowledge*, ed. By Michael Roth, Leon Galis, Random House, New York, 1970, p. 15

translates 'risk' and therefore acts has to do with lots of factors e.g. his background, beliefs, way of life etc.

The question is: can we characterize choosing knowledge over ignorance as something that can give people the sense of security. If people are allowed to exercise their right to know, wanting to know what to expect, genetic information can only provide an idea of a personal genetic condition and does not guarantee any future outcomes[67].

5.2 Kant's and Mill's theory and "the right to know"

Kant's belief that individuals are ends in themselves and should not be used merely as a means to an end is strongly connected with the ethics of what is right. Kantian approach focuses not on consequences, but rather on identifying what is right for the agent to do.

The individual agent has a duty to act as a rational and at the same time moral agent, as according to Kant the rational agent is by definition a moral agent. It is not so important to focus on the outcomes of a decision, but to examine whether the maxims of an individual choice can be universalized.

In a Kantian way of thinking along with the duty to yourself to take a moral, rational decision you have also a duty to other moral agents to take such a decision. It is not only a matter of deciding the morally and rationally right according to your judgment but universalizing your decision. According to Kant *to be human is to interest oneself in the fate of other men; inhumanity is to take no interest in what happens to them.*

Based on the above maxims and following the Kantian theory we can construct an argument for a duty to know. If this knowledge can help other people and can be of benefit for the whole then I have a duty to know any genetic information. *We should not only take satisfaction in the welfare*

[67] See also Chapter 6: the case of a couple who had two prenatal tests positive for Down syndrome and their child was born healthy

and happiness of others but this satisfaction should relate to the effectual actions that contribute to this welfare.

In addition, the above argument can be strengthened with Kant's theory of duties to oneself. According to Kant, *freedom (...) is the capacity which confers unlimited usefulness on all the others. Man has a general duty to himself, of so disposing himself that he may be capable of observing all moral duties, and hence that he should establish moral purity and principles in himself, and endeavor to act accordingly.* Therefore, if by acting in knowledge someone may have the ability to benefit some other people as well, then he should choose knowledge over ignorance in order for his action to be morally right. Acting in this way, *a person can indeed serve as a means for others (...), but in such a way that he does not cease to exist as a person and an end.*

However, Kant himself did not explicitly discuss the right to know or not to know, but his principles suggest the following: if someone chooses to act in ignorance (an action which serves solely personal satisfaction) instead of acting in knowledge (an action which may help another human being apart from the moral agent), then Kant's position will support acting in knowledge. *So act that by the maxim of your action you may present yourself as a universal legislator.*

Someone can generally argue that Kant's theory is generally promoting acting in knowledge and not in ignorance. In his reference to suicide he mentions the duty of self-preservation. He argues that we have the duty to preserve life in any way and must not let ourselves to *be deterred from living by any fate or misfortune, but should go on living.*

Therefore, in the case where a person is offered a genetic test in order to detect any genetic susceptibilities, according to Kantian theory, he should accept it, since following the duty of self-preservation, with the right treatment he may preserve his life and go on living. In the case which the person who takes the test is for example a smoker and the test shows a genetic susceptibility of lung cancer (and he finally dies from lung cancer), according to Kantian theory this is an offence a

culpa[68] but not *dolus*[69]. *He, who shortens his life by intemperance, is certainly to blame for his lack of foresight and his death can thus be imputed indirectly to himself; but not directly, for he did not intend to kill himself (...) it is the intention to destroy oneself that constitutes suicide.*

Despite the fact that Kant did not refer directly in reproductive choices, Kantian theory will have to support the idea of having a prenatal test to act in knowledge. If we consider the embryo as a human being, then according to Kantianism the mother has the duty to preserve life by not killing the embryo. Killing a human being is totally condemned by Kantianism as an immoral action. Kant would argue that from the minute someone has self-consciousness then he is considered as a moral agent. Therefore, if the embryo would be severely handicapped to the point where self-consciousness would be impossible or freedom to take control of their being then perhaps abortion can be justified.

Someone may argue that the embryo is not a human being but a child and therefore according to Kant not a moral agent. It is true that in Kantian theory children's agency is not the same thing as mature, rational adult agency but the only difference seems to be that unlike competent adults, children may be treated paternalistically so as to promote their welfare according to a best interest standard. Therefore Kant's problematic with children not being moral agents is their lack of ability concerning decision making process and this – in my opinion – does not subtract anything from their moral existence as human beings.

On the contrary Mill's liberalism introduces a position entirely different from Kantianism. Despite the fact that, in his essays he also did not refer directly to the 'right to know or not know', his references to harm principle and the notion of autonomy can be applied. Mill's harm principle established that individuals have the right to do as they please with their bodies provided that they injure no one else in the process. Therefore limiting liberty can only be justified only in the case to prevent harm to other people.

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[68] Due to fault
[69] Done with intent

Mill's position on liberty is strongly connected with his idea of autonomy. Based on his naturalistic approach to autonomy, any personal decisions and actions are considered to be expressions of human nature including any desires and impulses. Therefore, for Mill it is very important to act as an individual human being and person's wishes constitute a major priority. The individual is likely to be the best judge of his or her good. That is the reason why Mill's liberalism does not accept paternalism as it uses coercion to promote good. In general, for Mill an action is distinctively autonomous because it is deriving from desires of a distinctive sort.

Mill's theory then seems to support the idea of the right not to know. If a person expresses his desire not to know any genetic information then his will should be respected unless this choice will harm others. Since the desire not to know cannot cause any injury or harm to any other person then according to liberalism this desire can be justified.

Mill's respect for a person's autonomy requires that we let the person make decisions for and by him, no matter how foolish we find these decisions to be – as long as they do not inflict unnecessary and avoidable harm upon innocent third parties or interfere with the same rights of third parties to decide for them. Following the same way of thinking, the right to know can be also accepted by Mill's liberalism insofar as it represents the personal desires of the individual. In the case of a smoker who continues smoking without having any test (act in ignorance) to know his/her personal risk, Mill's liberalism will have to support such an action. However, by continue smoking and eventually die from lung cancer can cause psychological harm to others (family, friends), and such an action will be against Mill's principle of harm.

5.3 The right to know and personal decisions

The 'right to know' means that there is a "duty" to communicate about all public health risks and consideration of the principles of prevention, precaution and environmental justice[70].

The quotation suggests that the "right to know" presupposes a "duty" on the grounds of principles of prevention, precaution and justice. Therefore according to the above phrase public authorities or generally people in a position may have a duty to convey information. Additionally, there is also the question whether the right to know presupposes a duty to know. To what extend individuals have the right to know and at the same time are they obliged to know? What is the connection between the 'right to know' and confidentiality, personal autonomy, data protection legislation, and most importantly responsibility?

'Responsibility' is an element which is very often underestimated when referring to the 'right to know' however, it is considered to contribute a lot to the principle itself. Nuffield report in *Genetic screening and ethical Issues,* has commented on the responsibility concerning genetic information:

The question of responsibility has at least two dimensions in this area. The first is the responsibility of the individual to pass on relevant information to other family members, and the second is the responsibility of the other family members to receive the information. [71]

In the latest supplement the council adds the following information: "*The Council recognised that the results of screening might have serious implications for members of a family. When genetic screening revealed information that might have implications for the relatives or the person being screened, the Report recommended that health professionals should explain why the information should be communicated to other family members. (...) Where a screened individual did not wish to inform relatives of a genetic risk or to give permission for test results to be used by them, the*

[70] Lambert, T.W., C. L. Soskolne, V. Bergum, J. Howell, and J.B. Dosserot, "Ethical percpectives for Public and Environmental Health: Fostering Autonomy and the Right to know", *Environmental Health Perspectives,* Vol. 111, No.2 Feb 2003

[71] December 1993

Council accepted that under exception circumstances it may be appropriate to disclose genetic results 'without consent' to benefit family members".[72]

The Nuffield Council believes that the primary responsibility does not lie with the professionals but with the individual and the way he/she will handle the information. This is extremely important, as it assumes that it is not enough to want to perceive the information: the moment someone receives the information, at the same time he has the responsibility for it. When a doctor possesses information and he mistreats it or makes a choice between disclose it or not, then he possesses the whole responsibility for his action. In addition, if an individual receives information then he/she has the responsibility to take any necessary decisions (take further tests, change his lifestyle etc) and how and to whom to pass or not the information. These issues are analyzed further below.

If we follow a strictly narrow definition of genetic information which focuses mainly on DNA sequence it is possible to argue that genetic information itself is neutral; it cannot cause harm or benefit. On the other hand, if we use a broader concept of genetic information including any information coming from diagnosis then there can be a significant harm or benefit depending on how each person receives this information. However, its use or the passing of genetic information to people can be very controversial. Genetic information as a kind of information is like a piece of gel. It can be shaped, having the ability to take various forms and shapes (graphs, data tables, micro arrays etc), depending on whom and for what purpose is using the information.

The problem is that genetic information alone cannot do all the above. Therefore, it is with the intervention of an agent (professionals, individuals etc) that genetic information can take shapes. People who seek genetic knowledge must have in mind that genetic information is not 'good' or 'bad' but can be given a significance that can have good or bad effects.

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[72] Annual Report by the Nuffield Council on Bioethics, 2006: http://www.nuffieldbioethics.org/go/publications/publication_429.html

Precisely because genetic information can be examined from various perspectives and each person has their own way to encounter with information, it is difficult to reach general characterizations about its 'goodness' or 'badness'. The information per se e.g. "someone has a mutation that puts him at a higher lever of risk to develop a particular form of cancer" cannot be considered as necessarily 'bad' or 'good' information but depends on the person who receives the information.

On the contrary, knowing that you are at a higher risk than others to have cancer cannot be as good news as someone who is not in a risk at all however, e.g. to a doctor who has seen several cases of patients with cancer (most of the time severe), the information that a person is in a higher risk may not be characterized as 'bad'. There are many types of cancer e.g. breast cancer[73] that can be detected and cured in an early stage. In addition, there were many cases in the media where people went through a disease, and came out stronger as personalities, re-evaluating in this way their lives[74].

5.3.1 Who has the right to know?

The concept of the 'right to know' is normally understood as the right to have the right to access your own personal genetic information – information that can be possessed by a Public Hospital, the Government etc. The 'right to know' is then the right, which allows people access their personal genetic information and also the freedom to know what information has been collected and how it is been used.

[73] The BRCA 1 & 2, are genes mutations that were discovered to have a link to an increased risk for breast and other cancers. About 10% of breast cancer cases are directly due to inherited mutations in these genes.
[74] Frank A.W., *The wounded Storyteller: body, illness and ethics,* Chicago Press, 1995

The 'right to know' must be seen as a positive right as it gives the freedom to people to know their personal information (and to perceive it in any possible way) if they wish to know. This right can be exercised when people want, or need to know their own genetic information.

Primarily, when we are talking about who has the right to know, we are talking about the most immediate agent of this information, which is the individual per se. People seek for their genetic information in order to be aware of any important aspects of their health, including any predispositions or disorders that can affect them in the near or distant future.

However, the question arises as to whether people other than the individual can legitimately have a right to information about them. First, there is also a group of people, which is genetically linked with each of us. Relatives in the cases of hereditary diseases or traits may be affected and therefore any genetic information about us may be of great interest for them. For example, if a woman diagnosed with breast cancer subsequently tests positive for a known disease causing mutation of the BRCA1 or BRCA2 gene, then her first-degree relatives have a 50% likelihood of carrying the same mutation. They therefore have a significantly increased cancer risk.

Along with relatives, potential partners may claim their right to know, for example to know whether there is any chance to give birth to a child with genetic predisposition. It is clear that a future partner has the right to decide whether he/she wants to have a child with us. Reproductive partners may have the right to know but also the right to choose another partner. On the other hand, it is undoubted that the individual has also the right not to disclose the information to his/her partner if he/she feels that in this way the privacy is protected.

Some people may argue that there is a duty to disclose such information to future partners in order to take more informed decision. However, I strongly support the idea of personal autonomy in a sense that the final decision and responsibility of disclosing or not the information lies with the individual who possesses it. Therefore, in any case it is the individual that has to decide and take any responsibilities involved.

Lastly, there is a group of people whose right to know any genetic information about other people is most controversial. This group includes people with whom we have contracts, economic agreements e.g. insurance companies, employees, banks etc and also society.

Some strong arguments exist which support the idea that genetic information should not be treated the same as any other kind of information[75]. In addition, many genetic tests can only indicate if an individual has a predisposition to develop a medical condition not a certainty that eventually they will. If insurance companies demand disclosure of these test results then, some people may not take these tests at all, fearing of a possible discrimination by insurance companies or employers.

On the other hand, insurance companies may argue that if genetic disorders are reported then, the cost of medical treatment or a premature death can be calculated and therefore better financial aid can be provided. However, this can lead to many negative implications as insurance companies may eventually deny life or health policies to those who are genetically predisposed to specific diseases.

As mentioned above I will argue that the main consideration in resolving these conflicts of interest is that the major keeper of this knowledge should be the individual *per se* since he is the source of the information and *the primary responsibility for communicating genetic information to a family member or other third party lies with the individual*[76].

The problem whether this information should be disclosed to any third parties is clearly a matter of the individual to consent. Passing information to third parties raises issues having to do with the obligation or not on behalf of the individual to disclose the information, as the individual has to outweigh any potential harm and benefits as well. In order to address these issues it is necessary to examine the concept of genetic privacy.

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[75] For the importance of genetic information see below
[76] Nuffield Report

5.3.2 Privacy and Genetic Privacy

To start with, I would like to make a brief reference to the meaning of privacy, since it can be frequently confused with confidentiality. Privacy is a very broad term and can be characterized as *something which is to be respected, which may be forgone, lost, forfeited or invaded* involving the right to be free from invasion, the right to preserve autonomy, or to be left alone.

The intention of this chapter is not to analyze the value of privacy[77] and the meaning of preserving privacy in general among the society, rather to focus on genetic privacy defined as a right to control personal genetic information and the ability to choose: if, how and by whom this personal information should be collected and used. Confidentiality is only one of many means to protect personal information, and it cannot in any case take the same meaning as privacy.

In the literature it is argued that the issue of genetic privacy is not new as it raises the same concerns as the old problem of "privacy". Despite the fact that both privacy and genetic privacy share the same essence since in both cases personal information constitute their basis, genetic privacy is slightly differentiated from privacy itself because of its delicate character as it is linked with genetic information.

5.4 Genetic privacy - limits - implications

When analyzing privacy in general, the main concern focuses on its limitations and on the right to exercise privacy[78]even if this means the right to control the information to prevent others from accessing it. In the case of genetic privacy the main issue remains the same: its limits and the exercise of genetic privacy, with the only difference that in the case of genetic privacy we are not dealing with any kind of information - but possibly with the most important one: genetic information. As discussed above, the right to genetic information is argued to be a prima facie right

[77] For privacy in general see also Chapter 3
[78] For the problematic of "privacy" in general see Rossler, B., *The value of Privacy*, Polity Press, 2005, p. 67-71

exactly because of its uniqueness and importance not only to the one who possess this information but at the same time to his environment: family – relatives, future partners and the society in general.

Genetic privacy is facing a lot of challenges and threats, not only because of its linkage with genetic information but also because it may be violated in many ways, with or without the consent of individuals. As McCloskey argues *privacy may be violated not only by the intrusion of a stranger, but by compelling or persuading a person to direct too much attention to his own feelings and to attach too much importance to their analysis*[79]. In genetics this can take place during genetic counselling. The counsellor should remain neutral without persuading the person to take a specific decision – using any means e.g. direct too much attention to his own feelings – but rather, to present the facts in order to help the individual to have an informed choice.

The genetic information coming from a genetic test does not have implications only for the proband[80] but also for blood and future relatives (partners). Reproductive decisions depend on genetic information, and in many cases people who are currently healthy may be informed of a future illness because of some genetic predisposition.

Additionally, an extremely important aspect is that DNA analysis can be carried out in every stage of an individual's life. Both the foetus and a dead body are eligible to provide scientists with DNA. This underlines the significance and the delicacy of the genetic information, as the provision of DNA is not always the issue at stake as its use is also significant. In every stage of life - including the cases in which the individual himself/herself cannot control the information (in cases of death and sickness, children, embryos) privacy to genetic information including access and possible uses must be secured.

[79] McCloskey, H.J.,"The political ideal of privacy", *The Philosophical Quarterly*,Vol.21, no.85,1971, *p. 304*
[80] The individual who is been tested

As Graeme Laurie argues: a*n absolute right to privacy is not...advocated...Rather, we look for a prima facie respect which should be accorded unless due cause for disclosure can be shown*[81]. If we support an absolute right to privacy then we give absolute power to individuals or groups - communities (with blanket or group consent) to define and decide the lack of their own genetic information. However, Laurie's argument does not stand in favour of the absolute of genetic privacy, instead he supports *respect to genetic information unless due cause for disclosure can be shown.* Therefore, if the individual can provide justification for disclosing the information then disclosure is approved. Laurie suggests respect to the information and at the same time respect to individual and the personal justification that can be provided.

Therefore, on the one hand it is essential to understand that individuals have the right to know, decide about the genetic information and sometimes to choose not to give consent. On the other hand, there may be some cases and various circumstances where the need for disclosure of genetic information can be shown.

Disclosing genetic information is a decision which has several implications. There are many factors that should be taken into consideration e.g. the nature of a possible genetic disease (severe or not), the question whether disclosure can benefit the third parties or the public, the question of how the individual might react if exposed to unwarranted information[82].

Epidemiological studies can provide an example where public health can outweigh personal interests, as it can be argued that there is a duty to participate in research where consent to provide genetic information is urgently needed.

The question is what happens when someone does not follow the duty to participate but chooses not to. If his/her genetic information is really vital e.g. for a research, the possession of

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[81] Laurie, G.T., "In defense of ignorance: Genetic Information and the Right Not to Know", *European Journal of Health Law*, Vol. 6, 1999, p.129

[82] Graeme Laurie gives some more factors concerning the decision of the disclosure of genetic information.

his/her genetic data without his/her consent constitute loss or invasion of privacy? In what cases – if any – can such loss or invasion can be justified?

5.4.1 Loss and invasion of privacy

At this point it has to be clarified what is the difference between loss and invasion of privacy. It is essential to stress the fact that different societies judge different things to be or involve losses and invasions of privacy, depending on their beliefs etc[83]. Thus, it is difficult to present the exact definition of loss and invasion of privacy as it is applied in every society. However, there are some basic factors, which can be easily identified.

When we are talking about loss of genetic privacy we are referring to the loss of genetic informational ignorance. This is the situation where the individual either *comes to know the new personal information or ...comes to know that there is something to know that is considered quite urgent for her or him to know*[84].

It is the case where individuals do not possess any genetic information but the doctor instead of disclosing the exact information to him/her, he discloses the possibility of personal genetic information that may be of interest or of great importance for the individual to know. If the individual had already expressed the interest to know this information then this is not the case. On the other hand, if the individual without expressing any interest to know or not to know any personal genetic information receives the statement from any professional that there is something that is considered as urgent to know, then he loses his ignorance.

The above point is crucial as it leads us to think about genetic counselling and its significance, especially concerning the role of genetic counsellor in respect to the value of the genetic privacy and autonomy of the individual. The psychological pressure is not easily identified.

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[83] See also Chapter 6.

[84] Husted, J., "Autonomy and the right not to know", *The right to know and the right not to know,* ed. by Ruth Chadwick, Mairi Levitt and Darren Shickle, Avebury, Aldershot 1997, p.56

On the one hand, the consent-based argument is considered to be an important aspect of genetic privacy and on the other hand, in order to achieve an informed and competent consent, a discussion between the counsellor and the individual is vital, since the genetic counsellor has the duty to discuss about how the individual feels concerning a specific genetic issue and at the same time to remain neutral[85].

The cases of loss and invasion of privacy have as a common parameter the fact that someone else possesses private-personal genetic information. However, the difference between loss and invasion is that the case of privacy involves forcing a person's will. The right to privacy in the latter case can be understood as involving a right to confidentiality, by ignoring the individual's desire not to disclose any personal genetic information or his/her right not to know any genetic information, we can talk about an invasion of genetic privacy.

An important decision someone must take is whether he wants to know e.g. if he is at risk for a genetic disease. This is a personal decision and has to be taken without any interference and most important without forcing knowledge on people. However, someone may argue that there is a duty to be genetically informed even in situations where one cannot medically benefit from the knowledge. Following the Kantian theory one has a duty to know, fulfilling in this way his obligation to others[86].

Lastly, it must be mentioned that there are cases where someone can experience at the same time: loss and invasion of genetic privacy. In this particular case people who possess the right to know personal genetic information: e.g. the government or hospitals, in some cases (e.g. for epidemiological purposes) are entitled to collect and use personal genetic information without the consent of the individual. In this case, those institutes, professionals, or people who have the right to know this information should: a) be fully consciously about their actions, having in mind the loss and invasion of genetic privacy and b) respect the right to genetic privacy in terms of not passing

[85] See also Chapter 4
[86] See also Chapter 5

these information to those who lack the right to know the specific genetic information[87]. In this way genetic privacy is respected and treated as a basic ethical value.

When people were asked about genetic information during a research conducted by IFSA[88] in Australia (2002) concerning genetic testing and life insurance perception, one of the answers was that: "*credit card details is just money. Your genes-it is yourself, can be used against you in too much ways-job, insurance, marriage*"[89].

Therefore, it can be assumed that people concern genetic privacy as something extremely important and should be respected as such. Despite the fact that the concept of genetic privacy is newly introduced as we currently gained the means to gather and manipulate the genetic information, it is undoubted that one of its foundations is the concept of autonomy.

5.5 Genetic autonomy

The correlation between privacy and autonomy has many parameters. There are several arguments, which denote either that privacy is protected by autonomy and that *the success of autonomy and consent-based argument... has to be seen as something of an ethical panacea for the dilemmas in medicine*[90] and we need and value privacy precisely because in this way we exercise our autonomy[91], or that *privacy respects autonomy in* the sense that *the exercise of autonomy is depended upon the control of the 'access' of others*[92] to any personal genetic information.

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[87] For the right to know or not to know see above
[88] Investment and financial services association ltd
[89] www.ifsa.com.au
[90] Laurie,G.T., *Genetic privacy: A challenge to medico-legal norms*, Cambridge University Press, 2002, p.183
[91] Beate Rossler ibid. p. 72,
[92] ibid. p.73

Most of the authors agree that autonomy consists as the basis on which privacy is founded and therefore it is strongly linked with genetic privacy per se[93]. Therefore, the purpose is not to examine the value of autonomy in general and its relationship with genetic privacy, rather to support the idea that *personal autonomy ...is the ideal that individuals should have the power to make uncoerced choices and bring about what they have chosen*[94].

The idea of the autonomous person as a 'moral chooser'[95] is very important. It reflects the need for a sufficient provision of information in order for individuals to make significant choices. In this context, autonomy is seen as the right to choose. An autonomous person can make autonomous choices. If we want to achieve an autonomous choice then we have to respect people's right to know or not to know any genetic information. The preservation of genetic ignorance and the choice not to know can be easily reversed by the geneticist with just a single question: '*Do you want to know whether you are at risk?* By asking he *has already made the essence of information known*[96].

How can the idea of a moral chooser making autonomous choices be compatible with the right to know or not to know? The above question is examined in the small case study below concerning reproductive choices and prenatal testing.

5.6 A case study

The following story belongs to a young mother: "*My mother was very ill with breast cancer and I became seized with a desperate desire to have a baby. To my amazement I conceived at the first attempt and told my mother on her birthday, when I was ten weeks' pregnant. A week later, I*

[93] See also Chapter 3 and the reference on principled autonomy. Onora O'Neill's conception of autonomy promotes a conception of autonomy based on the Kantian notion of categorical imperative. As Onora argues principled autonomy is a test of which principles can be universable.

[94] Clayton, M., "Individual autonomy and genetic choice", *A companion to genetics,* ed. by Justine Burley and John Harris, Blackwell 2002, p. 195

[95] For the term 'moral chooser' see also Greame Laurie, ibid. p. 127

[96] Wertz, D.C., and J.C. Fletcher, "Privacy and Disclosure in Medical Genetics Examined in an Ethic of Care", *Bioethics*, Vol.5, 212, 1991, p. 221

paid for a detailed nuchal scan[97]*. We opted for this precautionary measure because my husband's sister had been born with severe health and problems and hadn't survived infanthood. The scan operator switched on her monitor and within seconds had located the embryo with its tiny fingers and toes already visible. But there was something hesitant in her manner that chilled my heart. Tears were flowing down my cheeks long before she said there were 'some anomalies'. I was referred to Cambridge's Rosie Maternity Hospital the same day (...). A consultant gave me another scan and pointed out the foetus's flat profile. He said there was a more than 50% chance our baby had a severe disorder. I already knew I did not wish to continue the pregnancy. Giving birth is precarious enough without the odds stacked against you. I don't think parents seek to eradicate 'imperfect' babies, but most of us desperately wish to maximise the chance of our children being alive to live a full and happy life. (...) The foetus had a chromosomal disorder called Patau Syndrome*[98]*, which affects all the major organs and always proves fatal. The consultant recommended a termination and three days later I arrived at the hospital for one of the saddest days of my life.* (...)[99]

Analysis

Concerning the prenatal tests and the right to know Graeme Laurie supports the idea that the *availability of prenatal test for a condition for which there is no cure or treatment only allows for a more 'informed' abortion decision.* Therefore he argues that if people are advised to take a prenatal test for a specific condition which does not have any cure – just medical treatment – then most of the times, having the knowledge about the medical condition they consent to abortion. Someone

[97] The Nuchal scan (or the nuchal translucency scan) is a special ultrasound scan used to measure the nuchal fold at the back of the baby's neck. It is thought that babies with a particularly thick nuchal pad at the back of the neck are at a higher risk of suffering from heart defects, Down's syndrome or some other chromosomal problem. Once the risk factor is calculated using this scan, you will be offered a subsequent amniocentesis or other invasive test to confirm, or rule out, these suspicions.

[98] Patau's syndrome occurs when the baby has an extra copy of chromosome 13. Most Patau's syndrome babies do not survive more than a few days. The majority of pregnancies with this chromosomal abnormality result in a miscarriage. The odds of a live birth having Patau's syndrome is 1 in 12,000. Babies with Patau syndrome experience many complications. They are mentally challenged and have multiple physical abnormalities such as malformed feet, hands and facial features. They may also be deaf and have difficulties seeing and smelling. This is usually a result of incomplete brain development.

[99] The true story belongs to Rowan Pelling from Cambridgeshire, published in magazine *Glamour, No. 45, December 2004, p.221*

may argue that Laurie's argument can be the explanation of the increase concerning the number of abortions in Cyprus.

Thalassemia does not have a genetic cure (marrow transplantation is not considered as cure as it can be applied only in a very young age). Consequently, as statistical studies show, the majority of the couples with thalassemia trait – if they find out after prenatal testing that they are going to have a child with thalassemia – almost in all cases they choose abortion. Thus, the question is: should doctors offer prenatal test to these couples? In my view, as I argued before, based on the right to know or not to know, if the couples want to take the prenatal test they should be allowed to do so, having in mind all possible implication the test may have for their lives.

Graeme Laurie also argues that *a comprehensive package of pre-natal testing might seem attractive in facilitating reproductive choices, but it can also be used as a means of encouraging the abortion of 'undesirable' foetuses. A right not to know would place the onus on the state to justify such testing*[100]. It is undoubted that prenatal testing can offer the opportunity for couples or mothers to take more informed reproductive choices. However, sometimes promoting prenatal testing may not spring from the best interest for the individual and their choices. For example in Cyprus, prenatal testing for thalassemia was also promoted for financial reasons. Because of the abortions the expenses for medicine (Desferal treatment) and blood transfusions were diminished, and nowadays fewer doctors are needed to work in Thalassemia Department. Therefore Cyprus strategy by encouraging prenatal testing at the same time it encouraged abortions – a strategy which facilitated not only reproductive choices but financial purposes as well.

However, my opinion is that concerning reproductive choices people should not only be informed that they have the option of prenatal testing but also that they have the right not to know. Someone may argue that people are aware of their right not to know, but in my view if we want to claim that we offer a pluralistic and neutral genetic counseling then we should present only options

[100] Graeme Laurie, ibid. p. 123 and 128

which promote the option and the ability of the right to know, but at the same time remind people that there is also the option to exercise their right not to know.

In the above case the woman is consenting to the prenatal test because of a previous medical history in the family. She took her decision of not keeping the baby, when she had the second scan and the counselor informed her that the chances were more than 50% for a child with a severe disorder.

However, there are two interesting issues in her story: (a) the fact that she adopts a rather utilitarian-consequentialist approach, stating that most of the people *desperately wish to maximize the chance of* living a full and happy life and also (b) that she finalized her decision as *the consultant recommended a termination*, showing that she was influenced by the consultant's recommendation.

According to her way of thinking, she took her decision of not having the baby because her baby could not live a full and happy life on account of the genetic anomaly. By taking the prenatal test she exercised her right to know and eventually she made her mind and chose abortion.

It is true that the second scan for Rowan's baby with Patau syndrome – a condition without a cure – proved efficient in the sense that it allowed Rowan to take a *more informed abortion decision* as she had the right to know and she exercised her right. It is known that between 80 - 90 per cent of babies with Patau syndrome, do not survive infancy and in those that do survive learning disability is present. However, some children survive into their teens and seem to fare better than might be expected based on reports from those who die in the prenatal period. Reports of adults with Patau syndrome are rare.

The question is: when we adopt the idea that people have the right to know (not allowing them not to know) then at the same time, do we support full knowledge even if people do not ask for it? In Rowan's case, she asked for the information and therefore the consultant gave her all the

information she had to know, in order to take an autonomous fully informed decision. However, what about counselor's recommendation of a termination of pregnancy?

This can be considered as a case of loss of genetic privacy. Is this information included in what Rowan needed to know to exercise her right to know and therefore take the 'appropriate' decision? Precisely because we know little about what kind of information perspective parents themselves consider relevant to their situation, providing personal recommendations or suggestions can be a loss of genetic privacy as Rowan did not ask the consultant for a personal opinion.

If we suppose the right to know as a right for not interference, a right to be left alone to have autonomous choices then a prenatal test or scan should preserve this right. Therefore, it can be argued that the consultant - respecting Rowan's right for an autonomous choice - should not make any recommendations, expressing personal ideas unless she/he is asked to do so, since as Rowan argues: *birth is precarious enough without the odds stacked against you.*

If we want to give an answer whether people should have the right to know and the right not to know as well, then we should not approach the whole issue choosing which is right and which is wrong. Based on the right to preserve genetic autonomy we cannot deny people the right not to know. Making a genetic decision is a very sensitive matter, a path, which can be either walked alone or not, meaning that decisions can be taken either with or without the help from the professionals. If the aim is to make responsible and autonomous choices, then the question is if we do need as moral agents, the information to maximize the good (*live a full and happy life*).

The difference between making all the genetic information available to people in order to take autonomous and responsible choices and forcing knowledge to them without their consent is really vital to preserve genetic privacy. As mentioned at the beginning of the chapter genetic privacy is linked with genetic information which can play a serious role in taking decisions that may reflect on peoples way of thinking, living and lead them to modify their self-picture for the rest

of their lives. Information concerning a possible genetic disease can even cause them several psychological problems and generally make them to re-evaluate lots of things in their lives.

Therefore when we are talking about genetic decisions, we are referred to decisions that can affect someone's life as a whole. It is not a matter of adopting or not the idea of collecting any genetic information available because the right to know entitles you to do so. It is a matter of whether someone really needs and wants this information, whether he is ready to accept them, to take the responsibility - a responsibility that does not involves only individual decisions, but in many cases, relatives and potential partners.

Rowan *became seized with a desperate desire to have a baby* because of her mother's breast cancer. In the case that her mother was healthy Rowan may not want a baby so desperately. Her mother's cancer makes her think of having a baby in the first place. Later, she decided to take the first scan because of her husband's sister. Her husband was not a blood relative for her but because of the unborn foetus it was important to know the genetic information about his sister. This information led her to take her decision and have an abortion, describing the day of the abortion as *one of the saddest days of her life.*

It is beyond doubt that genetic information concerning our relatives can be important and in some cases it can be crucial, as in Rowan's case. Even if someone knows or does not know anything about the genetic record of his/her relatives, or finds out that it is different from what he/she imagined, it can easily affect his/her attitude towards life and in some cases towards personal environment.

In many cases the right to know or the right not to know in genetics is linked with the procedure of taking reproductive choices. I support the idea that such choices are formulated through a procedure in which the right to know or not to know can make great difference, and that is precisely why I chose the case of Rowan. Exercising her right to know she had two prenatal tests. She did not take any decisions concerning abortion until after the second test and after visiting a

consultant. The consultant thinking that Rowan's right to know allows her (the consultant) to express her own idea, she recommended abortion.

Summarizing, someone may argue that there are two notions of genetic autonomy: (a) claim that autonomy implies a duty to keep oneself genetically informed, even in situations where one cannot medically benefit from knowledge (b) claim that the principle of autonomy supports a right to genetic ignorance. Therefore what exactly genetic autonomy entails you to do?

Conclusion

My view – based to the analysis in this chapter – is that truth stands somewhere in the middle. Auto (self) –nomy (nomos = law) as a term was first used from Greek Historians as an essential definition of the polis, and meant the ability of polis to be self-ruled. In addition the term is mostly strongly associated with Kant as well, for whom it meant the ability to give the moral law to each person individually. Genetic autonomy is strongly connected with the above meanings.

I support the idea that when we refer to genetic autonomy we place the moral responsibility for any decisions or actions to the individual *per se*. The crucial point is not whether we must support the right – or the duty - to know or not to know any genetic information, but rather to exercise our genetic autonomy primarily by understanding that we do have a choice between knowing and not knowing. Genetic autonomy does not stand in favour neither of the right to know nor not to know, but in favour of the ability of people to take responsible decisions which reflect their own morality.

In this chapter I tried to analyse the idea of the right to know or not to know, its basis as a theory, followed by the analysis of genetic privacy and genetic autonomy which as principles represent its ethical foundations.

CHAPTER 6 – EMPIRICAL RESEARCH

This chapter introduces my empirical research, which focuses on thalassemia and thalassemia trait. The specific study took place in Cyprus between September 2003 and February 2004. The study was based in discussion groups. To date, no systematic study concerning Cypriots' perceptions on thalasssemia and thalassemia trait has been undertaken. The purpose of this study was to discover how and in what degree people in Cyprus regard thalassemia as part of their individual life and whether they feel that they are influenced or affected by it in any way as personalities, active and productive members of the society.

The chapter is constructed as follows:

Section I (a)analyses some important background information on Thalassemia trait (thalassemia minor) and thalassemia major, (b) explains the reason why Cyprus was chosen for my empirical research, (c) introduces Genetic Screening in Cyprus, (d) evaluates the question whether genetic screening in Cyprus can be characterized as "ethical eugenics" and closes with details about (c) the study - hypothesis, methodology – the reason behind chosing qualitative research, design of the Study, criteria for selecting study participants, duration of the study, how data were analyzed.

Section II presents the findings of this study as follows:

(a) Defining thalassemia trait and thalassemia major

(b) Defining "serious"

(c) Understanding how "serious" thalassemia/ thalassemia trait is

(d) Stigma - stigmatization

6.1 Section I

(a) Background information on Thalassemia trait[101] (thalassemia minor) and thalassemia major - why Cyprus was chosen for my empirical work

Thalassemia comes from the Greek word "thalassa", meaning "sea", and it was given this name because it mostly affects people living in the Mediterranean region, including Italy, Greece, Cyprus, Turkey and Syria. It also affects people in the Middle and Far East.

[101] In Cyprus most of the times the word "stigma" is used instead of "thalassemia trait"

The original occurrence of haemoglobin disorders in areas where malaria is found indicated a possible connection and we now know that the healthy carrier- state (also known as trait) appears to offer some protection in early childhood against malaria. Thalassemia trait is a mild anemia. All anemias are connected with a deficiency of hemoglobin in the red blood cells.

Thalassemia trait or thalassaemia minor occurs when a person inherits one thalassaemia gene from one parent and a normal haemoglobin gene from the other parent. Someone who has thalassaemia trait can pass it to his children. Apart from the advantage thalassemia trait carriers have in respect to malaria, there is a hypothesis that it may also provide some protection against heart attacks, otherwise known as a myocardial infarction[102].

Thalassaemia major occurs when a person inherits two thalassaemia genes, one from each parent. A baby born with thalassaemia will appear normal at birth, but towards the end of the first year appetite and energy diminish. The skin becomes pale and growth is slower than normal. Anemia is found and treatment with iron fails, so that blood transfusions are required and are generally continued throughout life. Although treatment is available, it is not entirely satisfactory because life span is reduced.[103]

Thalassaemia does not cause mental retardation, but it is considered to be a severe genetic disease. Treatment may extend the life into early adulthood but it is very costly as it requires blood transfusions every 2 – 4 weeks and administering Desferal (known as iron chelation therapy) by continuous infusion for 10 – 12 hours daily using a pump, to remove excess iron from the body.

If *one parent has thalassaemia trait and the other parent has the normal type of haemoglobin*, there is 50% chance with each pregnancy that the baby will be born with thalassaemia trait and 50% chance to will be born healthy. If *both parents are thalassaemia trait carriers*, then they are at risk for having children with thalassaemia major: there is 25% chance

[102] Crowely J.P, Sheth S., Capone R.J. and Shilling R.F., "A paucity of thalassemia trait in Italian men with myocardial infarction", *Acta Haematologica,* 1987, 78 (4): 249 - 51

[103] See also appendices for diagrammatical presentation

with each pregnancy that the baby will have thalassaemia major, 25% that the baby will be born healthy and 50% that the baby will be a thalassemia trait carrier. If *one parent is thalassemia trait carrier and the other has thalassemia disorder* then there is 50% to give birth to a child with thalassemia disorder and 50% for the child to be a thalassemia trait carrier.

In the case which *both parents have thalassemia disorder* then every child will be thalassemic. If *one parent has thalassemia disorder and the other has the normal type of haemoglobin* then every child will be a thalassemia trait carrier. This is extremely important as it proves that a person with thalassemia disorder is able to make a family and give birth to a child without necessarily inheriting the disorder.[104]

(b) The Cyprus case - Why Cyprus was chosen

A British doctor, Alan Faudry, during a campaign in the 40's to eliminate malaria from Cyprus, recognized thalassaemia as a disease. He calculated that the frequency of the non-physiological gene went up no higher than 17%. [105] This was considered a success, given the means available just after the Second World War. Further studies and calculations were made by other researchers, both Cypriots and foreigners, which confirmed Faudry's results.

In those early years, if a thalassaemic child received no treatment or if the treatment was inadequate, the thalassaemic child - who was exceptionally pale - hardly ever reached the age of eighteen months. He was too ill to eat, and was restless and nervous. If he had no treatment at all, he died in the first years of his life from heart failure or infection. If he received some treatment and was given blood transfusions, as was the policy in the 60's, so that the haemoglobin level could be maintained at 7-9 g/dl, the picture did not change.

Death was merely put off for few years, and the siderosis which resulted from the blood transfusions, together with the increased absorption of iron, contributed to the faster onset of

[104] See Appendix I for graphical presentation
[105] Faudry, A.L. "Erythroblastic anaemia of childhood (Cooley's anaemia) in Cyprus", *The Lancet* (1944) I : 171-176

haemosiderosis and its negative consequences such as heart problems etc. The Cypriot population evidently faced a severe problem in the years 1950-1960.

In one of his articles Dr. Michael Angastiniotis - one of the main initiators of the program of thalassaemia control in Cyprus - records the details of the disease as follows:

1 out of 7 Cypriots was a carrier of ß-thalassaemia

1 out of 49 couples, both the husband and wife, were heterozygotes

1 out of 158 new born babies was expected to be homozygote

The prevalence of homozygotes in the population was 1 out of 1000, whereas there were already 600 affected individuals in a population of 600,000 [106]

As Dr. Panayotis Ioannou mentions, during the 50's and 60's parents who had already given birth to one or more children with thalassaemia had no choice but to watch them silently die, secluded from the hospitals and the world - while the parents prayed God for mercy and forgiveness. [107]

During the years 1950-1970 many families had a child suffering from thalassaemia. Even in bigger towns and villages the thalassaemic child was a secret well kept in the family from fear of stigmatization. Fear, ignorance of the medical facts and compassion for the diseased child led the families to seclude themselves from the rest of society and see their children's dramas through without daring ask for help and support either from the other families or from the state.

The Cypriot state, in its turn, was entirely helpless and powerless to support its own citizens. This was mainly due to the fact that previous experience of a similar situation was lacking, consequently no medical, social, legal or other infrastructure existed which could have provided

[106] Angastiniotis, M., Kyriakidou, S. and Hadjiminas, M. (1986) How thalassaemia was controlled in Cyprus, *World Health Forum,* 7 : 291

[107] Ioannou P., "Thalassemia Prevention in Cyprus", *The Ethics of Genetic Screening,* ed. Ruth Chadwick et al., p. 58

support to the families who had been afflicted by the disease. Gradually Cypriot society began to realize the disastrous consequences that this disease had for the population.

At this stage, information and knowledge about the disease began to reach Cyprus from other – mostly Mediterranean - countries which were facing the same problem and Cypriot parents themselves started to seek treatment for their children. Some of them were brave enough to try to find blood donors or to buy blood. Parents - who discovered that others had been through the same pain and suffering - formed a pressure group, and established the Pancyprian Anaemia Association. Since support from the scientific community was almost non – existent at the time parents felt that by establishing this association was the only way to support their selves.

The first meeting took place on May 14 1973, at the Pallouriotissa Primary school with around 30 parents. An organization was established then under the name "Association of Parents with children with Mediterranean Anaemia", with its headquarters in Nicosia. Later in March 1974, as it was found the problems with thalassemia affected the whole of Cyprus, it was decided at an extraordinary general meeting to rename the association Pancyprian Thalassemia Association, so that the problems could be dealt with on a national level.[108]

With the introduction of a drug called Desferal and the availability of blood transfusions, many patients began to survive on into their child – bearing years. This was an economic challenge for the government of Cyprus.

The existing thalassemia patients were consuming more than 50% of the available blood supplies, while more than 20% of the total drugs budget of the Ministry of Health was used for the purchase of Desferal. Furthermore, with an expected birth rate of 60 – 70 new patients per year, the number of patients could double in about ten years, thus stretching the limited blood supplies and

[108] http://www.thalassemia.org.cy/

other resources to the limit and compromising the quality of care not only for the existing thalassemia patients but for other patients groups as well. [109]

In Cyprus, before 1970, 17% of infant mortality was due to thalassemia and the disease was recognized to be a significant health problem. Many children born with thalassemia died undiagnosed at a young age (<1). The volume of blood required and the cost of desferrioxamine (DESFERAL) began to approach unmanageable levels. Increased survival rates were predicted to cause 300 – 400% increase in blood requirements and a 600 – 700% rise in the cost of treatment of the population within 50 years[110].

In 1978 the Cyprus government made available free diagnostic testing to couples for carrier testing. [111] Three years later the Church of Cyprus began to require couples be tested for carrier status before marriage. The birth of new thalassemic patients has dropped to 0 to 2 per year since 1985. [112]

The Cypriot Government realized that the only solution was to limit the number of affected births by introducing a prevention programme: this programme involved health education and community involvement, genetic counselling but most important population genetic screening.

Cyprus' 1972 Thalassemia Program is recognized as being unique in the world for its success in almost completely eliminating new cases of thalassemia within 15 years. The availability of prenatal diagnosis in Cyprus - especially CVC (Chorionic villus sampling) which involves sampling the mother's placenta and testing for abnormalities - solved several problems connected with the free choice of a marriage partner without the "burden" of giving birth to a child with thalassaemia. Couples carrying thalassemia gene with the help of pre-natal diagnosis can decide if they want to have a child with thalassaemia, or otherwise choose the "option" of abortion.

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[109] Ibid
[110] Angastiniotis, M., S. Kyriakidou, and M. Hadjiminas, "How thalassaemia was controlled in Cyprus", *World Health Forum,* Vol. 7, 1986
[111] ibid p. 59
[112] Ibid p. 61 - 62

Five years after the emergence of prenatal diagnosis, thalassemia births fell from 15% to 2% of expected. Three vital factors contributed to this reduction: avoidance of marriage if both partners were carriers of thalassemia trait, avoidance of pregnancy, and prenatal diagnosis followed most of the times by abortion. Prenatal diagnosis became the main reproductive choice.

Along with prenatal testing, the Greek Orthodox Church introduced premarital screening. The Orthodox Church in Cyprus requires screening for this genetic disease where couples wish to marry in the Church, but there is no requirement for them to make the results known and they are free to marry even if they know that they are both carriers of the gene. With the Church's cooperation, the number of people screened grew significantly from 1,785 in 1977 to 18,202 in 1983[113].

With premarital testing, genetic screening is mandatory for a couple wishing to get married in a Christian Orthodox Church. Therefore, in the case of premarital testing the issue of the right of couples to know or not know the specific genetic information (if they are thalassaemic trait carriers), is not considered at all. However, if they do not wish to possess this information then they can always choose a civil wedding instead.

To sum up, high public acceptance of new technologies combined with religious endorsement not only challenged longstanding social norms on abortion but also endorsed abortion as the ideal socioeconomic solution to the major problem of thalassemia. In addition, the dramatic fall in affected births with thalassemia in Cyprus has promoted the idea that a 'successful' heamoglobinopathy screening programme can be measured in terms of the reduction in the number of the affected births.

(c) Genetic Screening: The Cyprus case

The parameters of genetic screening

[113] Angastiniotis M., "Cyprus: Thalassemia Programme", *The Lancet* (1990) 336 (8723): 1119-1120

Genetic screening can be defined as a test focusing on the early detection of a hereditary disease, the predisposition to a specific genetic disease or to determine whether a person may be a carrier of a predisposition, which may result in a hereditary disease in offspring.

Precisely because genetic screening is addressed to asymptomatic individuals, populations or sub-populations and not patients who simply ask for professional advice, the implications and the ethical dilemmas deriving are particularly important. The nature of a genetic disorder and the risks, which are involved, do not concern only the individual who consents to genetic screening but also in many cases other family members, therefore we are dealing with *issues of consent, confidentiality, and data protection*[114].

In Cyprus premarital screening has the aim of making people think about the responsibility of giving birth to a thalassemic child, to detect if two potential parents (a couple) have thalassaemia trait, thus providing the couple with the opportunity to decide a priori whether they want to proceed and get married.

As someone can easily ascertain from the above, the need for efficient and generally acceptable safeguards, standards and procedures concerning informed consent, genetic counseling, confidentiality, stigmatization and discrimination in society (including employment and insurance) is really important.

In this part of the chapter my objective is not to comment on the efficiency of genetic screening programmes or on any advantages or disadvantages their application among the society may have, but rather to proceed to an analysis of the existing general principles and guidelines of genetic screening as these principles are defined from worldwide organizations (WHO, Nuffield Council on Bioethics, Council of Europe). Additionally, it is also very important at this stage to address possible question related to the degree in which principles and guidelines are applied in

[114] Nuffield Council on Bioethics, *Genetic Screening and Ethical Issues*

Cyprus and whether the new era of post-prevention policy requires revised guidelines concerning the control of Hemoglobin Disorders.

WHO published two important guidelines linked with thalassemia: "Guidelines for Genetic screening" (1988) and "Guidelines for the control of Hemoglobin disorders" (1989). It is very interesting the fact that both guidelines have some substantial similarities, as they both promote some basic parameters concerning genetic screening and its application.

In the "Guideline for the control of Haemoglobin disorders" it is mentioned that *to date, the best model for such services are the disease-orientated "thalassaemia control programmes" organised in some countries of the Mediterranean area where thalassaemia is particularly common. These embody the WHO concept of a control programme for hereditary disorder, i.e., a comprehensive strategy combining the best possible patient care, with prevention by community information, carrier screening and counselling and the offer of prenatal diagnosis". They have proved to be effective, acceptable and highly cost-beneficial".*

It is obvious from the above that WHO considers as criteria for a successful prevention programme not only carrier screening and counselling but also an important element seems to be the fact that a programmes should be *highly cost-beneficial.* This is extremely important in order to understand why WHO considers Cyprus strategy on thalassaemia a successful one as it fulfils all the above criteria.

Specifically, WHO considers that *in Cyprus there is a long-standing public and professional education programme and both the population and health professionals understand the implications of thalassaemia. Single carriers are given their result by letter, and rarely require specialist counselling as they can discuss problems with their own doctor. Individual counselling is offered for unusual carrier states and at risk couples.*

Therefore, it is obvious that WHO seems to think that not only community information is efficient in Cyprus but also counselling is offered in a satisfactory level for *unusual carrier states*

and at risk couples. Cyprus strategy programme is pointed out by WHO guidelines as an ideal programme which is not only socially but also economically beneficial.

Additionally, both WHO reports stress the value of: voluntariness in genetic screening, informed consent procedures concerning the purpose and the outcomes of the genetic screening, the preservation of the confidentiality, the right to full information, the disclosure of the results to third parties (insurance companies, employers, schools) only with the individual's consent and last but not least the offer of genetic counselling.

Concerning genetic counselling with carrier couples three ethical principles are introduced: a) the autonomy of the individual or couple b) the right to full information and c) the highest standard of confidentiality. It is also stated that in order to preserve autonomy, people *must be fully informed, and counselling must be "non- directive".*

Additionally, in the Guidelines for hemoglobin disorders (1989) it is suggested that *screening in High Schools may ultimately be an ideal strategy* for hemoglobin disorders. Specifically, it is mentioned that *it requires a well-informed population, enthusiastic teachers, and a developed infrastructure. It is offered in Latium (Italy) and Montreal (Canada) in the* near future may become the policy of choice in Cyprus. Moreover, it is mentioned that *premarital testing is unlikely to suit communities in which identification of a genetic risk prior to marriage can lead to stigma especially for women.*

As Cyprus is considered as a country where *couples themselves without any outside pressure would decide what is best for them*[115], the above guidelines raise various questions concerning Cyprus case. Is Cyprus strategy on thalassemia respectful to the principle of free choice? Does this strategy facilitate the ability for *couples to decide what is best for them*? How possible a non – directive genetic counselling is in Cyprus as Cyprus's policy on thalassemia aimed at the reduction

[115] Hadjiminas, M., "The Cyprus experience – Screening to combat a serious genetic disease" *Council of Europe on Bioethics,* Council of Europe Press, Strasbourg 1994, p. 26 - 48

of the number of births with thalassaemia major? The issue is whether a counsellor who works for the Government is the ideal person to advise and offer the proper guidance to a carrier couple?

It is true that for a period of time screening in High Schools was established in Cyprus, but unfortunately this attempt was abandoned as it was replaced by premarital screening and prenatal testing. The aim of Cyprus prevention programme was to *secure a high level of care and improved prognosis for thalassaemia patients, and to bring about a significant reduction in the number of new cases*[116] , as in this way the Government *by diminishing the frequency of new cases* could keep *future costs down*[117] .

Consequently, it is essential to ask if it is possible to focus on the reduction of patients with thalassaemia and at the same time promote autonomous decisions concerning keeping or not an embryo with thalassaemia after prenatal screening? Is it possible for a policy based on a mathematical thinking (reduction of the number of thalassemics = lower costs) to focus at the same time on an "*improved prognosis*" for patients and a fully informed non directive counselling?

It also interesting to examine the Additional Protocol to the Convention on Human Rights and Biomedicine concerning Genetic Testing for Health Purposes by the Council of Europe as it was discussed on Strasbourg on May 2008. The rapid developments in the field of human health have prompted the Council of Europe to consider the ethical and legal aspects of applications of genetics, particularly genetic testing, and to draw up legal rules to protect fundamental human rights with regard to these applications.

Specifically, Council of Europe with the article 19 of the above protocol states that *a health screening programme involving the use of genetic tests may only be implemented if it has been approved by the competent body. This approval may only be given after independent evaluation of*

[116] Angastiniotis, M., S. Kyriakidou, and M. Hadjiminas, "How thalassaemia was controlled in Cyprus", *World Health Forum,* Vol. 7, 1986, p. 297
[117] ibid.

its ethical acceptability and fulfillment of the following specific conditions: (...) *the programme provides measures to adequately inform the population or section of population concerned of the existence, purposes and means of accessing the screening programme as well as the voluntary nature of participation in it.*[118]

On the one hand, it is undoubted that Council of Europe recognize the significance of a health screening programme and the benefits which derive from it, but on the other hand it is crucial the fact that it clearly states that its voluntary nature is a significant component. The question is whether this component exists in Cyprus screening programme for thalassaemia. Since premarital screening is mandatory people do not have the ability to choose to screen or not to screen and thus, the screening programme does not have a "voluntary nature".

In the same article it is mentioned that *the purpose of proposing a genetic test as part of a screening programme for health purposes is to allow the members of the population or section of population concerned to make appropriate personal choices concerning their health or in relation to procreation, on the basis of the results of the proposed test.* We can assume from the above that the Council of Europe concerns that the aim of genetic testing is to allow people *make appropriate personal choices.* By forcing people to be tested we violate the very essence of the test which is genuinely voluntary.

The above is also linked with stigmatization derived from genetic screening. In Chapter II, article 4 of the above protocol it is also stated that *problems of stigmatisation may indeed arise with regard to those taking part in such a screening programme. (...)Screening programmes of this type are aimed at detecting or excluding, by means of a genetic test, the presence of certain genetic characteristics linked to a disease. The perception of the disease in question and the interpretation that could be made of the purpose of the screening could result in the individuals concerned being stigmatised.*

[118] http://www.coe.int/press

Therefore, it is important to stress out that in genetic screening we should not only focus to the danger of violating people's will but also to the fact that the nature of a screening programme which does not preserve people's right to choose may lead to unwanted consequences and increase stigmatization among the society.

This imposes new questions for Cyprus strategy as the specific programme does not seem to follow the guidelines provided by the above protocol. Does Cyprus need a new strategy on thalassemia in order to follow European guidelines on genetic screening? Does Cyprus need to revise the existing screening programme in order to reinforce "the voluntary nature" of the programme and avoid discrimination among people? Premarital screening seems to be a burden for the voluntariness of Cyprus strategy on thalassemia and therefore it is important to examine its significance for Cyprus society.

Cyprus and premarital screening

To begin with, it is very important to stress that premarital screening and prenatal testing especially in Cyprus must not be seen as a public service, something that the Government - provider should offer as a routine. *Technology may become routine but this does not necessarily mean the ethical and social issues have disappeared or been resolved*[119]. Following the philosophy of WHO in genetic screening[120] it is important to offer along with premarital screening genetic counselling as well.

In Cyprus, genetic counselling has to be engaged with these ethical and social issues of couples during the procedure of premarital screening. We should not analyze here the role of genetic counselling in general; however it is interesting to ask whether the role of genetic counsellor is differentiated in Cyprus when dealing with thalassemia trait carriers.

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[119] Levitt, M., "Let the consumer decide? The regulation of commercial genetic testing", *Journal of Medical Ethics, 2001, 27, p.398 - 403*

[120] For WHO guidelines see above

Despite the fact that thalassemia occurs in many countries apart from Cyprus, it is important to note that every society is unique and has to be treated as such. In Cyprus, a genetic counselor has to deal with all these *ethical and social issues* of each couple. Therefore, through discussion the counsellor does not necessarily have to help the couple to get over these issues, rather to help them understand what they really want, what is best for them.

In general, the role of the genetic counsellor is extremely important as he has the ability to help the individuals use the information and genetic technologies in a way which is primarily non-directive, supports and facilitates the counselee's autonomy. In the case of premarital screening genetic counsellors are committed to help the couple *discover what course of action, upon reflection, is best for them*[121]. Additionally, genetic counselling has to be available for the couple not only during premarital testing but also in prenatal testing. The couple should also be informed by the obstetrician, or the genetic counsellor about the prenatal test, its procedure and the possibility of a misdiagnosis.

I believe it is important to analyze further the issue of misdiagnosis as not only it is something that most of the times, is not stated as a possibility to the couple, but also it is possible that a genetic test is expected that it would be 100% accurate. In a recent research, 25% of the participants expected that a test for carrier status *would detect 100% of affected individuals*[122] , thus it may be the case that most couples do not take into consideration the possibility of a misdiagnosis as they trust its accuracy. Even, if it is mentioned to the couple, it may not make any difference as the percentage of a misdiagnosis is very small.

A current research[123] reviewed the accuracy of prenatal diagnosis for the thalassemia and sickle cell disorders performed for UK residents since the service began in 1974. Prenatal diagnosis has been performed in 3254 pregnancies. The number of homozygotes diagnosed was 808 and 25

[121] Erik Parens, Adrienne Asch, ibid.
[122] Mary Levitt, ibid
[123] Old, J., M.Petrou, L.Varnavides, M.Layton, and B.Modell, "Accuracy of prenatal diagnosis for haemoglobin disorders in the UK: 25 year' experience", *Prenatal diagnosis,* 2000, p. 986 - 991

diagnostics errors have been recorded. The authors stated that each prenatal diagnosis is confirmed at birth therefore many errors may have gone undetected if the fetus was aborted. Additionally, cases where a baby was diagnosed unaffected but eventually the baby was affected they were known only if they were reported in the UK thalassemia patient register. It is estimated that in the period of nine years 4 of 107 births with thalassemia were due to misdiagnosis.

It is very interesting the case of twins at risk for thalassemia were diagnosed as identical and unaffected as the laboratory suspected that both samples came from the same fetus. The obstetrician reviewed the case, but decided against repeat sampling. Non-identical twins were born, one with thalassemia. Nevertheless, the authors state that *diagnostic accuracy has improved with each advance in diagnostic technique and increasing experience has led to the adoption of precautionary measures to minimize the risk of diagnostic error.* This is fundamental as it ensures more accurate results however it does not presuppose that couples should not have the right to be informed about misdiagnosis.

It is vital to let "*the couple to discover*" what they want, why and how ready they are for what they want. Having a child with thalassaemia, a disease which people in their majority categorize as a "serious" one, presupposes commitment on behalf of the parents to their informed decision to keep (or not to keep in the other case) the baby after the prenatal screening. The goal is to *help parents think harder and better about what having a child with a disability really means for their lives together*[124].

It is important that parents should first think about how having a child with thalassaemia will affect their lives. Since this is an extremely difficult decision it would be vital for the couple not only to meet other parents who have a thalassaemic child but also to talk with other thalassaemic people. As Nuffield Council on Bioethics supports, *the availability of prenatal screening and diagnosis, together with the termination of seriously affected pregnancies, both reflect and*

[124] ibid

reinforce the negative attitudes of our society towards those with disabilities[125]. Consequently, a thalassemic person may also have to deal with some sort of discrimination from the society i.e. in working environment etc, and therefore parents of a thalassemic child should be psychologically ready to deal with such attitudes.

"The doctor asked us if we are going to have the child. I answered yes. And he asked; Do you know what this costs, do you know what this means, and do you know anyone who is disabled? He went on and on...in a nervous blabber. I tried to explain that we know disabled people and that we had made our decision. Then our obstetrician came (...) He said: We will try and do all we can for you and for the rest of your pregnancy. He supported us and respected our decision".[126]

The above story belongs to a pregnant woman who discovered after the prenatal test that she was going to give birth to a disabled child. It is important the fact that she knew other disabled people and consider that they were leaving a normal life. This affected her decision to keep her baby. Therefore, as it can be assumed it is crucial for future parents with thalassaemia trait not only to receive genetic counselling but also to be encouraged to visit and talk with other parents and thalassaemic people.

Cyprus has succeeded in diminishing the number of thalassaemic patients, but someone can claim that this is not enough. The priority should not only be the provision of an excellent treatment, put also the promotion of research on a cure. Specifically, in Sardinia, a voluntary screening program is applied and according to Renzo Galanello, a professor of Pediatrics at the University of Cagliari, the number of babies with thalassaemia has increased in the past two years *most likely because parents see improvements in treatment*[127]

If on the one hand, there is an excellent provision of treatment and at the same time a further concentration on genetic therapy for thalassemia, not only Cyprus can upgrade the existing health

[125] Nuffield Council on Bioethics, *Genetic Screening and Ethical Issues*
[126] Bjarnason D.S., *Is life worth living if you have a disability?* Conference on Preimplantation Genetic diagnosis and Embryo selection, Iceland, May 2004
[127] Guterman L.,"Ethical Eugenics?" , *The Chronicle,* 2nd May, 2003: http://www.chronicle.com

care system but it would be possible to encourage couples who may choose to give birth to a thalassaemic child.

Summing up, it is essential that the provision of genetic counselling in Cyprus along with premarital screening and prenatal testing should primarily and most significantly enhance autonomous decisions - decisions which should be translated as informed ones, giving in this way couples more control of their lives. Angus Clarke supports the view that, if our strategy is to prevent a specific genetic disorder then *we immediately abandon the non-directive nature of genetic counselling in favor of a genetic public policy. It is impossible to maintain a sincerely non-directive approach to counselling about a genetic disorder whilst simultaneously aiming to prevent that disorder*[128].

(d) "Cyprus case: Ethical eugenics?"

The word *eugenics* etymologically derives from the Greek word *eu* (meaning *good* or *well*) and the word *-genēs* (*born*), and as a term was firstly introduced by Sir Francis Galton in 1883. In a historical and broader sense, eugenics can be a study of "improving human genetic qualities." This can be translated as the attempt to select or change or improve or discard a genotype or phenotype of an offspring in order to improve the line. It is sometimes broadly applied to describe any human action whose goal is to improve the gene pool.

It is true that the very word makes people in the Western world cringe. It conjures up images of Nazi atrocities back in the World War II. Despite the fact that eugenics is associated with cruelty and racism we will examine how eugenics is applied in Cyprus and specifically concerning Cyprus strategy for thalassemia.

Cyprus strategy is focusing on a mandatory program which aims to eliminate thalassemia. Before getting married, people must get tested to find out if they have the gene that causes thalassemia. If

[128] Clarke A., "Is non-directive genetic counselling possible?", *Lancet Vol.338, Issue 8773, 1991*

carriers of the gene decide to marry and have children anyway, the women can undergo prenatal testing to see if their fetus is doomed to the disease. If parents decide to have an abortion then Cyprus government will pay for the abortion.

Having in mind the above, someone could argue that since Cyprus program allows reproductive choices after the genetic screening and that refers to the fact that couples can still decide whether to marry and have babies with thalassemia, then Cyprus strategy on thalassemia is not eugenics.

However, the term eugenics itself is about improving the gene pool and it is true that a mandatory programme which screens people for thalassemia it primarily aims to keep the "bad thalassemia genes" out of Cyprus gene pool. When the programme was launched, Cyprus was in a very bad financial condition. Therefore, it was natural for Cyprus Government to think eugenics as the right solution to keep costs down.

If eugenics was not used back in 1970 then investing money on patients with thalassemia may had some negative impacts in other aspects of Cyprus society in general. Thalassemia is treatable, but procedure is very costly and obviously in 1970 threatened to bankrupt the entire health care system. Therefore, with limited amount to spend and with no genetic technology on thalassemia, it can be argued that perhaps Cyprus strategy was ethical eugenics.

The question is ethical to whom? Do we have to sacrifice thalassemics for the prosperity of Cyprus' society? Do we sacrifice the society and stand in favor of thalassemic people letting thalassemic children be born and live? Cyprus government decided. Cyprus adopted a strategy which gradually eliminated the number of thalassemic people but at the same time it forced people to screen in order to get married in an Orthodox Church, but what about genetic autonomy and "the right to know or not to know"?

True "choice" depends on options and a full objective disclosure of information pertaining to the options available. Individual autonomy is the bedrock of the society and healthcare ethics. It is

important to realize that we need to avoid falling into the trap that because the technology exists it should be utilized (i.e. we can, therefore we should). As Martin Heidegger noted *technology is a human activity* representing the distillation of our collective wisdom and intelligence with the freely chosen applications of this wisdom a reflection of our intrinsic values.[129]

Thus the key element in genetic screening is not only counseling and information given at the time of diagnosis, but also to let people decide whether they want to be screened. People in Cyprus should receive counseling prior to actual screening so that parents are aware of what choices they may have to make in the event of abnormal screening results. To truly permit choice the counseling made available must be non-directive and value neutral. Even if a child is diagnosed with thalassemia he or she can live among the society, work and enjoy life as everyone else does.

Reasonable concerns may arise that a publicly funded program of detection and counseling occurring at a time of funding limitations will be driven by a cost-containment emphasis that may weigh the scales of choice in one direction. Diseases as thalassemia are indeed chronic and have high associated hospitalization costs but this cannot be the reason for "forcing" people to screen or choosing abortion or to deny thalassemic people the right to live.

(e) The study – Aims and Objectives

Hypothesis

In the last few years since Cyprus succeeded in the reduction of the affected births with thalassemia the first hypothesis of this study is that young people age 18 – 27+ do not have experience of thalassaemia, in their everyday life and so are not aware of it. They may hear the term "thalassaemia" from parents and other older relatives but not from their own friends.

[129] Heidegger M., *The question concerning technology In: Basic writings,* New York, Harper & Row, 1977, p. 288

On the other hand, I hypothesize that middle aged people, who have had apparently a more intimate experience of thalassaemia, will consider thalassemia to be a serious genetic disease. Their attitude can be traced back to when Cyprus was a poor country with very few doctors (1950 – 70). At that time to have a genetic disease or to be unhealthy was a social stigma. For a woman this would cause difficulties in finding a partner and being accepted among the society.

Why choosing qualitative research

Qualitative research study things in their natural settings, attempting to make sense of or interpret phenomena in terms of the meanings people bring to them. [130] Qualitative research is ideally suited to illuminating the context of social behaviors and the processes the underlie them. It is important the fact that quantitative research generally tells people what is happening, while qualitative research can illuminate why it is happening. This was the reason why qualitative research was chosen for this study.

Qualitative research focuses on words, action and records whereas quantitative approach seeks for their mathematical significance. All the studies[131] on thalassaemia in Cyprus so far used quantitative research seeking for meaningful evidence of the extent of thalassaemia among Cyprus society. Thus, I decided to use qualitative research as our aim was not to find numbers and percentages rather to investigate people's words and actions and at the same time capture language and behavior. I did not want to examine e.g. how many people approve Cyprus strategy on thalassaemia but rather what people had to say about it, how people felt about it and how they believed it affects their life.

Therefore, qualitative research was chosen searching for the patterns of meaning which emerge from the data as they are presented in the participants' own words.

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[130] Denzin, Norman K. and Yvonna S. Lincoln, eds., *Handbook of Qualitative Research,* Thousand Oaks, CA: Sage1994,

[131] Examples of quantitative studies: Angastiniotis, M., "Cyprus: Thalassemia Programme", *The Lancet* (1990) 336 (8723), Angastiniotis, M., S. Kyriakidou, and M. Hadjiminas, "How thalassaemia was controlled in Cyprus", *World Health Forum,* Vol. 7, 1986 see also bibliography

Method of Data Collection

Since my basic and initial intention was to explore how the participants experience thalassemia and how do they feel about it, I chose discussion groups as a method to analyse the topic in depth.

By choosing discussion groups, I aimed to a quickly and reliable method to trace common impressions and perceptions. In addition, I believe that focus groups as a method is an efficient way to get much range and depth of information, as I wanted participants to express themselves. To me the interaction among the participants as part of the method was really valuable as I wanted to observe what participants they say to each other, how they interact within the group, what vocabulary they use.

"The idea behind the focus group method is that group processes can help people to explore and clarify their views in ways that would be less easily accessible in a one to one interview. Group discussion is particularly appropriate when the interviewer has a series of open ended questions and wishes to encourage research participants to explore the issues of importance to them, in their own vocabulary, generating their own questions and pursuing their own priorities. Everyday forms of communication may tell us as much, if not more, about what people know or experience. In this sense focus groups reach the parts that other methods cannot reach, revealing dimensions of understanding that often remain untapped by more conventional data collection techniques". [132]

Some basic questions were the guidelines for the discussion[133]. The original questionnaire included twenty seven questions which did not focus solely on thalassaemia. The questionnaire tried to investigate issues like genetic information, DNA, biobanks etc. The rationale behind the questionnaire was to construct a questionnaire which will help the participants express their ideas on many controversial genetic matters and help them expand their feelings and beliefs in the

[132] Kitzinger,J., "Qualitative research: Introducing focus groups", *British Medical Journal,* 1995, 311: p. 299 - 302
[133] For the questionnaire see Appendix II

discussion group. The discussions in each meeting were recorded, and were translated from Greek to English by the researcher in a later stage of the study.

Criteria for selecting study participants

It was essential to the design of the study that those who might volunteer to participate met only one criterion: participants should be a minimum of 18 years old. It was essential for the study that all the participants had graduated from High School –as the Cyprus programme for thalassemia was partly based on the induction of thalassemia in the High school curriculum, so practically the majority of the participants were taught about thalassemia and thalassemia trait in High School.

People were divided into groups by age, thus I had five discussion groups: Group 1 (18 – 20), Group 2 (21 – 30), Group 3 (31 – 40), Group 4 (41 – 50) and Group 5 (51 +). Additionally, it was important to examine whether there was a clear distinction between perceptions of thalassemia especially between groups 1, 2 and groups 4 and 5. People in groups 4 and 5 were in their 20s and 30s when the thalassemia programme was launched and thus according to my hypothesis they may have a different approach in the issues at stake. Concerning the sex of the participants I tried to keep a balance between male and female participants. In groups 3, 4 and 5 the participants were couples and thus the number of male and female was equal. In group 1 the majority of the participants who volunteered were women. The study did not take into account the participant's background or health status however the fact that some of the volunteers were thalassemia trait carriers was beneficial.

For the first two groups I posted some announcements concerning my study in the University of Cyprus trying to attract people who finished High School and wanted to participate in such study. As in Cyprus "thalassamia" and "thalassaemia trait" is included in High school curriculum I wanted to attract people who graduated from High school and were taught about the disease.

In the announcement, I clearly stated that participants will not receive any reward for their participation and they will not receive any future benefits from the study as well. In order to find

people for the rest of the groups I posted some announcements in public places like Hospitals, in some public buildings such as Ministry of Economy and Minitstry of Education - buildings that people from all social and financial levels visit very often. On the announcements I had my telephone number and participants contact me themselves showing interest for my study.

Since I wanted to include various age groups in my research people were divided into discussion groups with two or three people in each group. A copy of the questionnaire was given to each member of the group but questions were not answered individually. Participants had the opportunity to study the questionnaire silently and then the group was asked to discuss each question. As the questions were connected often the participants referred back to previous questions and made additional comments. The researcher did not answer any of the questions even if the participants asked for further explanations. If an explanation was necessary, then terms or phrases were explained without giving indications of possible answers.

As mentioned before, the original questionnaire included twenty seven questions.[134] This resulted in a vast amount of data. All discussions were recorded and translated in English but because of the amount of information I decided that for the purposes of this thesis I should focus on the set of the questions having to do with thalassaemia and thalassaemia trait. My aim was to present an efficient and in depth analysis of the collected data and investigate how and to what degree people regard thalassemia as part of their individual life and if they feel that they are influenced or affected in any way in their everyday lives.

Duration of the study

In the time period between September 2003 and February 2004, all discussion groups took place in Nicosia, Cyprus. I had the meetings for Group 1 and the majority of the meetings for Group 2 in the University of Cyprus as all the people from these groups were studying there. I arranged meetings for Group 3, 4 and 5 where it was most convenient for the participants (e.g.

[134] See appendixes for the questionnaire

friend's houses etc). I wanted participants to feel comfortable enough to speak their own mind and express their ideas. It was really helpful the fact that in most discussion groups people knew each other therefore they felt relaxed. I had 46 meetings with 2 or 3 participants in each.

Analysis of data

All discussions were recorded as mentioned before; therefore at the first stage of my analysis I replayed the tapes and did the transcription.

Since I had a large amount of data in front of me I tried to find patterns of questions having in mind what do people talk about most. After going through all the data I noticed that the questions concerning thalassaemia were the most controversial ones as many people felt really strong about "thalassaimia" and "thalassaimia trait". Therefore, for the purposes of this thesis I decided to analyze data which derived from the questions having to do with thalassaimia.

At the second stage of my analysis, I translated all the answers connected with thalassaemia from Greek to English. This was really difficult for me as in many cases it was extremely hard for me to find the exact translation or the exact word in English that would depict the meaning in Greek.

During the third stage of my analysis I made a list of all the dominated themes for which people talked the most and tried to see how themes relate to each other. Those themes are analyzed below.

6.2 Section II – Analysis and Discussion

(a) Defining thalassemia trait and thalassemia major

This part presents the participants' definitions and the level of understanding of the medical definition of thalassemia and thalassemia trait.

The majority of the participants found it hard to describe thalassaemia trait, perhaps because of the fact that as a term it is very similar to thalassaemia and they are both inheritable. Consequently, the majority seemed a bit confused. In most of the discussion groups the answer "it is something you inherit" was very common. People of all ages, mentioned the heredity factor first.

"If I have the stigma it means that I got it from my parents..."

"Stigma means that in my genetic code I have a specific characteristic that one of my parents has and if this is combined with another person's characteristic then my children will have a problem"

"Stigma I think is because one of your parents has thalassemia..."

"It is something that you have it when you are born, your parents inherit this to you..."

"What is the difference between stigma and thalassemia? I think it is the same thing...you get from because of your parents..."

"I do not know exactly what stigma is...but I know that you do not get it during your life...you have to be born with it" (Group 1)

"If one of your parents has the illness and the other one is healthy then they inherit it to you..."

"Stigma happens when one of your parents has the problem..."

"Stigma is not a disease. If I have it and my husband has it as well then my baby will have stigma as well"

"Stigma means that one parent has the healthy part and the other parent the sick one"

"I think stigma and thalassemia have many things in comment because sometimes we say stigma of thalassemia so I think perhaps it is the same thing...You must have sick parents to have stigma"

"When I have stigma means that I am carrying this from my parents"

(Group 2)

"When I have stigma it means that I have some characteristics, something like the footprint of thalassemia which is not developed into the disease itself"

"Stigma is not a disease. To have stigma one of your parents must have it as well..."

"Stigma has to do with genes because is something that is inherited from your family..."

"If I have stigma it means that I have anemia but not the disease. I am not ill but I am carrying the sick gene"

"Stigma does not mean that somebody is sick but it that because of his family genes it has the potentiality to transmit this disease to his children"

(Group 3)

"Stigma is a disease which can be inherited..."

"Perhaps stigma is related in a way with thalassemia. They definitely have some things in common..."

"Stigma is like a blood disease having to do with your genes"

"If my wife has stigma then there is the possibility to have a thalassemic child"

"If your genes are anemic then you may have stigma"

"You do not get stigma as a disease...It is something different...you have it in your blood"

(Group 4)

"Stigma is something that exist inside you...in your genes"

"If somebody has stigma he does not have to worry because this can be dangerous if it is combined with another person that has stigma as well..."

"The person who has stigma does not face any dangers but his children may have many difficulties as they may be born with thalassemia"

"It is very difficult to define stigma as it is connected with thalassemia...Stigma is like the sign that you are carrying the sick gene"

"If someone has stigma is because he inherit it from one of his parents"

(Group 5)

It must be mentioned that in Groups 1 and 2 there was another element which, mostly girls (78% including both groups) tend to connect with thalassaemia trait: "marriage". They felt that someone must have in mind thalassemia trait when it comes to the selection of a partner. Even though the issue of marriage and its connection with thalassemia and thalassemia trait is analysed further below, I quote here some of the participant's references to marriage:

"If I have stigma then the Church does not permit me to get married...because if me and my partner have stigma then our child may have it as well"

"When somebody has stigma then when he is going to decide to get married he must be careful in order to choose the right partner if he wants to have healthy children"

"My cousin has stigma but thank God she did not fell in love with someone who has it as well so they did not have any more problems..."

"I know that to have stigma may cause you some troubles especially if your partner has it as well...but again it depends...I mean if you are careful and you ask first then you will not find someone with stigma"

"No...I do not think that stigma is something serious...I know that sometimes people may think it means something but I think it is important in terms of choosing your partner..."

"I do not have stigma so I do not know exactly what it is...but Helen (a friend) *has it and she told me that she must not find someone who has it as well"*

"The Church concerns stigma something important that is why you have to give permission to get married but I believe that if someone has stigma and falls in love with someone who has it as well then they have to think really well before getting married"

Very few people, coming mostly from Group 4 gave a definition of thalassaemia trait using medical terms. Medical terms were also used to define thalassemia trait from a percentage (30%) coming from all groups. For example participants mention that:

"Stigma is the possession of a healthy and a sick gene in the genetic code. If this sick gene is combined with my partner's sick gene <in the case he has also stigma> then our children will suffer from thalassaemia" [135]

Additionally, there was a small percentage coming from Group 3 that used the terms "genes", "genetic code", and "chromosomes" in definition of thalassaemia trait. This is really interesting and encouraging as it clearly shows that not only they understand what thalassemia trait is but also the fact that they feel familiar with medical terms and feel comfortable enough to use them.

It has to be made clear that seeking accurate answers about thalassemia trait was not the target of my study. On the contrary, my aim was to valuate participant's level of understanding concerning thalassaemia trait as a medical term. For me it was also important that participants, who

[135] See also answers above

were thalassemia trait carriers, seemed aware of what thalassemia trait (as well as thalassemia), and specifically mentioned that:

"I know that stigma is nothing important...it is not like you have a serious disease or something..."

"Stigma does not affect my life...I am not ill...I just have something like anaemia"

"I know that most of the people connect stigma with thalassemia and I do not know why...ok it is connected in a way...but honestly stigma is not something serious, you do not put it in the same category as thalassemia"

The word "serious" was frequently used almost by the majority of the participants especially when referring to thalassaemia as a disease: a disease, which according to most of them *is very serious*. Almost nobody used the phrase "genetic disease". However, in the case of thalassaemia trait there were some people who mentioned that it has to do with genes.

Defining the meaning of "thalassemia" some people pointed out that thalassemia is a severe anaemia and if someone has thalassemia he/she must visit the Hospital regularly for blood transfusions. The majority of the participants (95%) gave a description for "thalassemia" without using medical terms but using expressions which indicate that they understand the term:

"I think thalassemia is a serious disease and if you have it then you have to change your blood regularly – have a transfusion...or something like that"

"Thalassemia is something with the blood...our body is not able to renew our blood, because as we know the blood has to be renewed in every healthy human being, and that is why thalassemics have to go every six months ...not sure...to the Hospital to renew their blood"

"Thalassemia is something which exists in the countries around the Mediterranean...I think your blood is really thin and you have to put in you some more in order to become thick...However you do not die"

"Simply...when you have thalassemia you must do blood transfusion and you can get other diseases easily. It is very serious and you have to get treatment for it"

"I think the blood transfusion is the last stage of the disease. At the beginning I think you have to take pills"

"Thlassemia is a serious blood disease...and it is very important to know that you have this disease"

"It is a blood disease. A serious disease...there is something going on with the iron in your blood. Your organs are not able to remove the iron so the doctors have to do it using technology"

"It is a disease you inherit. It affects many organs from our body, the liver, the heart, the bones and if you do not get the right treatment you can die...and sometimes really young"

To sum up, I realised that that most of the participants understood in very basic lines what "thalassemia" is and while attempting to give their definitions about it they mentioned some specific key – words as: "blood, disease, serious, Hospital".

Nevertheless, there was a minority of 5% mostly coming from Group 2 which appeared to be confused about the meaning of "thalassaemia", mentioning among others that

"I think that when you have thalassaemia the number of your white blood cells is low"

"Thalassemia is when there is something wrong with your white blood cells"

"Thalassemia affects your immune system and especially your white cells"

"I think in a way doctors have to make your white cells stronger when you have this disease"

However, thinking about why people tend to mention "white cells" specifically I noticed that participants who belonged in this 5% used in their phrases the word "blood" which is directly linked with thalassemia as a haemoglobin disorder. If we assume that people mentioned white cells because they connect them with the immune system of human body which is weakened in the case of a genetic disease, then we may argue that their definitions are not far from the actual meaning of "thalassemia".

It is undoubtedly true that in some cases ignorance of the disease itself can affect someone's attitude towards thalassaemic people. However, based on the discussions of the study, I believe that all the participants were aware of what "thalassemia" really is, even if they did not mention the exact meaning or they felt a bit confused about it.

Summing up this part concerning the definitions of "thalassemia" and "thalassemia trait", it is important to point out that my study indicated that lay people have a satisfactory understanding of what "thalassemia" and "thalassemia trait" are. People in their majority may not use medical terms but using their own expressions are able to describe and give their own definitions.

It can be argued that the above was something to be expected as in Cyprus the estimated carrier frequency of thalassemia trait is 1 to 7[136]. Therefore as thalassemia trait is a trait that is commonly found among Cypriots most of the participants had a relative or a friend with thalassemia trait.

However, if we take as granted the fact that people possess the basic knowledge concerning thalassemia and thalassemia trait, then it will be interesting to examine whether this knowledge signifies social acceptance for thalassemics as well. As mentioned above, most of the participants used the word "serious" to define thalassemia disorder. As a starting point it is crucial to assess how serious people consider thalassemia as a disease and if their perceptions concerning the severity of thalassemia affect the way they treat or accept thalassemics.

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[136] This frequency seems to be higher than other ethnic groups (Asians, Chinese, Afro – Caribbeans, White British). See also Anionwu, E., and K. Atkin, *The politics of sickle cell and thalassaemia*, Open University Press, 2001, p. 12

(b) Defining "serious"

Another issue which is strongly linked with the definitions of the participants on "thalassemia" and "thalassemia trait" is the use of the word "serious". What do people mean by claiming that thalassemia is *serious*? Do all people understand the same when they listen to the phrase: "thalassemia is a *serious* disease"? In the medical world the word can take many meanings and is used in different ways by doctors and scientists. With what other meanings or ideas is the word "serious" linked? Do people think that a disease is serious only when it can cause death or other disabilities? This part presents the analysis of the use of the word "serious" among the participants.

Serious as a term can be characterized as problematic. To start with, I should mention that observing the minority of thalassaemia trait carriers which volunteered for my study I noticed that when they were answering questions about thalassemia trait they were giving answers like:

"What I have is nothing serious; it is not like I am ill and I do not understand why we have to discuss about it. I do not feel that my condition is something special...I know that perhaps sometimes this can be a problem with thalassemia but everybody knows that stigma is something totally different..."

"Stigma is not a disease - something serious...it is just anemia. I do not have any symptoms...ok perhaps sometimes I get tired easily but still I do not thing is because of stigma...I do not feel different. And if somebody really loves me and wants to be my friend will consider the same..."

"I believe that stigma is not a serious condition, and definitely it is not a disease, I know it is connected with thalassemia but still is something very different...ok stigma is not like you have a flu either because is something that you have it for your whole life, but again is not something which causes suffering. At least I never felt anything like that...I know stigma can cause future problems to my children, but not to me..."

On the one hand, they had the knowledge that thalassaemia trait does not have an impact on their health that thalassemia trait is not "something serious". On the other hand, they felt that they have to stress it out and reassure everybody that they are healthy and therefore there is no reason to worry about it.

Consequently, by claiming that what they have "is nothing serious", they are categorizing people into two groups: the first one includes those who do not have something serious (in this case: thalassaemia trait carriers) and the second one includes those who do "have something serious" (thalassaemic people). According to Botkin there are four criteria for people to decide whether a disease can be categorized as "serious": *effectiveness of treatment, impact on the child and family, age of onset, and likelihood that someone with the mutation will actually develop the disorder in question*[137].

In Cyprus case the first two criteria have an application. On the one hand, thalassaemia major nowadays is considered to be a severe physical handicap. There is no treatment apart from bone marrow transplantation in an early age, if a compatible donor can be found. Therefore, most of the people believe that because there is no cure for thalassaemia and since the recommended treatment involves lifelong regular blood transfusions (usually administrated every two to five weeks) thalassaemia is considered to be something "serious" whereas thalassaemia trait is not.

Someone may argue that in Cyprus, thalassaemia must not be considered as a "serious" disease since treatment is provided, which allows people with thalassaemia to live a normal life: they can have a job and make families, give birth to children etc. However, as Dorothy Wertz and Bartha Maria Knoppers found in one of their surveys, even professionals have different perceptions of what counts as a "serious" disease.

Specifically, it was stated that the word "serious" appear close to the word "genetic" in many state laws, *as criteria for availability or support of genetic services, including prenatal diagnosis*

[137] Wertz, D., and B.M. Knoppers, *"'Serious' Genetic Disorders: Can or Should they be defined?"*, American Journal of Medical Genetics, Vol.108, Issue 1, 2002, p.29 - 35

and abortion[138]. Practitioners were surveyed in order to see how they defined "serious", and were asked to list genetic conditions and rate them as serious and not serious. The results showed that there was not sufficient consensus for policy – making purposes not even for the most frequently listed disorders. There was an overlap between the categories serious and not serious which may *suggests the effects of economic, cultural, and social environment. For example, social ostracism could make a hand abnormality serious, and difficulty finding a mate could make it a genetic lethal disorder.*

We should therefore ask why participants in the specific study in Cyprus refer to thalassaemia as a serious disease. Is it serious because thalassemics have to receive medical treatment for their condition for the rest of their lives? If this is the case then people should feel the same for medical conditions as Hypothyroidism where somebody has to take thyroxin pills for a lifetime. Do they believe that thalassemia is serious because there is no genetic cure just medical treatment? Do they believe that thalassemia can be categorized as equally serious with cancer, heart abnormalities and Down syndrome?

Cyprus control programme for thalassemia is well structured and many Cypriot doctors refer to this programme as a good example for all countries in which thalassaemia occurs. Consequently, in a country like Cyprus where community involvement (Anti – Anaemia society: a parents' and patients' association, public education etc) was combined with public health policy, someone would expect that people would have developed an equivalent perception considering "thalassemia" as a "not so serious" disease.

All of the participants agreed that cancer, thalassemia, heart abnormalities and Down syndrome can be categorized as the most serious diseases which appear in Cyprus. Nobody could think of, any other disease only two people from Group 4 said that they believed that also diabetes can be a serious disease as well. When I asked why they believed that these four diseases

[138] ibid

(thalassemia, cancer, heart abnormalities, Down syndrome) were the most serious diseases almost all of the answers mentioned the fact that there is no cure and that the person who has these diseases is sick for a lifetime. (However, very few people also mentioned that cancer can be curable in some cases):

"If you have one of these diseases, then you are sick for a lifetime...They are very difficult diseases to cope with"

"Someone may die from these diseases and you know that you will have to receive regular medical treatment...You are actually visiting the hospital regularly and this may affect your life..."

"I think these diseases are serious because if someone has one of these then he knows that he will be forever sick. Especially in the case of Down syndrome...you always depend on others"

"I agree that these diseases are serious, I think you will never feel healthy... but I want to mention that when I saw how my grandmother suffered from diabetes – she even had depression because of it – I realise that this disease is very serious as well as you have to have injections and you cannot even have an operation...But of course I would not want to have a baby with Down syndrome- this is even more terrible than diabetes"

The majority of the participants 70% believed that cancer is the most common serious disease in Cyprus. The rest of the participants believed that heart abnormalities were the most common disease and there were also 3 participants (2 from Group 1 and 1 from Group 3) who mentioned that thalassemia was the most common serious disease in Cyprus. 80% of the participants named Down syndrome and 20% thalassemia as a serious disease which appears rarely in Cyprus. The participants who argued that thalassemia is the disease which appears rarely added that this is because of the prenatal testing and that most of children with thalassemia are aborted.

According to recent studies[139] in Cyprus there are almost 10 – 12 cases with Down syndrome and 0 -2 case with thalassemia per year. Cancer and heart abnormalities tend to appear more often than thalassemia and Down syndrome. However, after introducing premarital screening and prenatal testing for thalassemia in Cyprus, the frequency of cases with Down syndrome is higher than thalassemia.

Therefore, it is important that the majority of the participants felt that thalassemia occurs more frequent than Down syndrome in Cyprus. This indicates that they do not believe that thalassemia as a disease has eliminated in Cyprus and that it still exists among the society. This raises the question that if participants believe that thalassemia is a serious disease which still appears among the society then do they also believe thalassemics are socially accepted?

Trying to give some answers to the above issue we should examine further the *hidden persuader*: social control[140]. It is undeniable that despite education, information campaigns, and genetic counseling concerning thalassaemia, social environment and some notions existing in this environment seems to influence people in Cyprus.

The social acceptance and the social interaction between the members of the society are elements of great importance. Cypriots feel the need to show and prove to their social environment that they are healthy and they are normal even if they just have thalassemia trait. In my study there were participants (mostly females) who were healthy (and also not thalassaemia trait carriers) who perfectly understood that if someone has thalassemia trait has nothing to do with the disease per se (and therefore cannot be considered as something serious), and they were still expressing some worries concerning finding a partner with thalassemia trait:

[139] http://www.emro.who.int/publications/EMRO%20PUB-TPS-GEN-PRT2-CHPT2-2.4.HTM
[140] Hoedemaekers, R., and H. ten Have, "Geneticization: The Cyprus paradigm", *Journal of Medicine and Philosophy 1998, Vol. 23, no. 3, p. 280*

"I know that stigma is not a disease and it is just something like a missing thing from your blood. Nothing to worry about...The only thing about it is that if you have stigma you have to be careful when you marry someone and you both have stigma"[141]

"It is not that I am afraid or something to relate to a person who has stigma....Not at all...I have two friends with stigma and they look healthy....but I know that if you have stigma you have to know it, so when you find a partner, to ask him about stigma...and think carefully about having a family"

"I do not have stigma so I do not have to think about anything...but if I my future partner has stigma I will start thinking about it. You should always be careful when it comes to these matters"

"Thank God I do not have stigma or thalassemia, otherwise I do not know how I would react about different things...Of course stigma is not like thalassemia, you could say that is something totally different but again it makes you think, and be more careful if you marry someone with stigma"

Anionwu E. and Atkin K. trying to explain the uniqueness of Cyprus case concerning the extremely high up-take screening services presented two reasons: *In Cyprus there is a high up-take screening services for thalassemia. The widespread experiences of caring for a person with thalassemia and the accompanying financial and emotional stresses are often associated with the success of the thalassemia screening programme in Cyprus (...) Cypriot communities in Britain have a similarly high take-up of screening services (...) The reasons for this are complex and relatively unexplored* [142].

However, the fact that Cypriots – compared to other communities - do consider "thalassemia" as something "serious" may indicate an additional reason for this high up-take screening services.

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[141] The correlation of the *hidden persuader* the institution of marriage in Cyprus is analysed further in (d) "stigma – stigmatisation" part

[142] Anionwu, E., and K. Atkin, *The politics of sickle cell and thalassaemia*, Open University Press, 2001, p. 56

Unfortunately, there are not any studies which investigate the degree of considering thalassemia as something "serious" in other communities; if someone however considers a disease "serious" then naturally he may feel the need to be screened for it.

(c) Understanding how "serious" thalassemia/ thalassemia trait is

Almost all people who participated in the research understood that thalassaemia trait is not something serious as it is not a disease. In Groups 4 and 5 people had more specific views on the matter as they stated that "stigma is nothing, since someone will never develop thalassaemia".

On the other hand, in Groups 1 and 2 (especially in Group 1) participants often mentioned "you have to be careful if you have stigma (thalassemia trait)". This attitude can be easily linked with the perception of marriage. Having thalassemia trait does not have an impact on the person who has it, but as genetic information it can be of a great value if someone wants to have a family.

In Cyprus marriage and family are of great importance for people. Most Cypriots believe that family and marriage is something sacred. A successful and happy marriage is something extremely significant as especially for a woman, giving birth to a healthy child is very important for her social status.

If a family in Cyprus has women who are at the age for marriage[143], parents may advise them not to inform their partners (at least not from the beginning of the relationship) that they have stigma, because maybe himself or his parents think that it is something important and consequently, a relationship may not end up in a successful wedding. The other person will eventually know about it when the relationship becomes serious.

Clearly, the above way of thinking is not applied in the society as a whole, but it still exists in Cyprus (especially in small villages etc) and sometimes may force some people who are already in an engagement to separate. The number of these couples which eventually break up is not very big

[143] In Cyprus people tend to get married young, usually at the age of 20 - 26

but the phenomenon still occurs. Unfortunately, as mentioned before there are not any surveys to show how many couples eventually separate after knowing that both partners are thalassemia trait carriers. I got this information talking to some nurses who worked in the Department of Thalassemia in Cyprus. As they have a regular contact with people who go there for premarital screening they tend to have personal experiences with couples who eventually decide not to get married.

In Cyprus – as someone may notice from the answers of the participants - people seem to understand that thalassaemia trait is not a disease and a serious health disorder. The issue is that most of thalassemia trait carriers do not want to make it so obvious to the rest of the people that they have the specific trait. They are worried about the reaction they are going to face.

In addition, apart from the case where young people deliberately do not reveal in an early stage that they have thalassaemia trait, there is also the group of people which are really worried about it and tend to ask the other person in an early stage of a relationship about thalassaemia trait just to avoid the possibility of giving birth to a child with thalassaemia.

If a thalassemia trait carrier wishes to eliminate the risk of having a child with thalassemia may consider it right to let the other person know about it before even involving in a relationship. The question is what happens if both partners discover that they are thalassaemia trait carriers? Do they separate just because there is 25% possibility in every pregnancy for a child with thalassaemia?

Most of the couples in Cyprus are already engaged when they discover that they both have thalassemia trait usually after screening in order to get the premarital screening certificate and therefore get married in an Orthodox Church. Consequently, even if both partners realize that they are thalassemia trait carriers, very often they proceed to marriage thinking that they can always

have the option of a pre-natal genetic diagnosis and therefore, terminating the pregnancy in the case of a child with thalassaemia. Very few couples decide to separate after premarital testing[144].

When the participants were discussing how they would have reacted in such a case (both partners are thalassemia trait carriers), in Group 2 (18 – 20) there were some young men – thalassemia trait carriers - who answered that they are definitely asking their girlfriends about thalassemia trait because they consider it a serious matter, whereas young women in the same Group mentioned that this is not so serious that it can be an issue in a relationship.

In addition, in the same Group most of the participants said that they will proceed to have a family since they believe that thalassaemic people live a normal life and socialize without any problems with other people: "Since there is the opportunity for a medical treatment for these people I would not consider the possibility of breaking up because of that".

Some other participants claimed that thalassaemia is a serious disease and despite the existence of the appropriate medical treatment for thalassemics, they consider it "pointless to give birth to a child with thalassaemia". Specifically, they believe that the whole treatment can be a torture for the child during his life and thus as parents will feel really miserable.

From the above, it can be argued that people give extra emphasis to the fact that they are going to be miserable as parents if they bring into the world a child with thalassaemia. This means that they value equally high both their own and the child's happiness. As Anionwu and Atkin mention, the provision of genetic services is interwoven by two major themes: *a discourse looks to prevent disease in order to maximize human satisfaction and well – being* and the other discourse which emphasize *the importance of the active human agency, exercising control over one's own life*[145].

[144] As mentioned above these couples are coming mostly from villages in the rural area of Cyprus
[145] Anionwu E., ibid., p. 48

In Group 2, 50% of the participants had the opinion that concerning the question whether or not you should still have a family if both partners are thalassaemia trait carriers, they felt that question is strongly connected with abortion:

"It is a matter of whether you want to have an abortion or not and this is basically the problematic part of the question"

"You have to ask before if your partner approves abortion and then if you are both in favour of abortion - if the child has thalassaemia - then proceed and get married"

"I do not know really how I would have reacted...I know that I would still have my doubts about getting married...I mean having an abortion is really a big thing especially for the woman"

"I think in that case I would definitely choose to adopt a child. I know cases that couples decided to separate...I do not even stand to the idea of having an abortion so even if my partner was in favour of abortion I do not think I would even try for a baby and play with possibilities"

"I think that even if my child was born with thalassemia, I would definitely not decide to have an abortion since a thalassemic person is able to leave as a normal person, and talk, and laugh – he needs just a treatment – so...but I think my partner has to agree on that as well"

"I think it is all about your beliefs...Either you accept right from the start that there is the possibility to have an abortion or you decide to separate and find another partner...But I know this is a very hard decision...Honestly I do not know how I would reacted in such a case"

Additionally, there were many participants in this Group who believed that the possibility for a child with thalassaemia when both parents are thalassemia trait carriers is 50%. Therefore, it can be argued that this misleading information can definitely influence their way of thinking and acting. Some other participants mentioned the issue of "love":

"Depends how much you love the other person and consequently in what degree you are able to support each other, because when you decide to give birth to a child with thalassaemia you have to know that you are going to face lots of problems"

"I believe that nowadays is very difficult to find somebody to love you, so if you manage to find someone and he happens to be a thalassemia trait as well then you do not break your relationship just like that...you discuss your option and decide what to do"

"I do not see any reason to break up a relationship. If you really love the other person then you stay together...of course I know that there are some people who are not strong enough to face these difficulties so it is better if you decide to separate...However perhaps this means that the other person does not love you enough...or he does not want to get involve any further..."

Moreover, it is important to note that young women with thalassemia trait from the same Group did not think that it is very suitable to ask the other person about thalassemia trait, as sometimes it is uncomfortable and you can never know the reaction of your partner:

"You know it is not the easiest thing to do...You have to be very serious with the other person to ask something like that...What if he things that stigma is something serious?...I have a two years relationship and I did not dare up to now to ask my boyfriend about that...but he did not tell me anything either...I think if we decide that we are seeing this relationship serious then I will have to say something..."

"I like to be clear from the start...I am not ashamed because I just have an anaemia...lots of people have different sorts of anaemia and get pills. I believe that the right person for me would be someone who really understands me and will not have a problem with stigma"

"I do not think is something I should say something or discuss it with my boyfriend...eventually he will know but I do not believe it is necessary for me to say something about it"

In Group 3 many of the participants characterized thalassaemia a serious disease which requires extra attention from potential parents. They believed that since it is a matter of 25% in each pregnancy for a couple to have a child with thalassaemia, someone can diminish the possibility by asking future partners if they have the trait as well. Therefore, they supported the idea of a couple being fully informed about the issue in order to avoid any future surprises.

"I have stigma but it is a very bad thing to have...I asked my boyfriend if his parents have stigma but he did not know...If I knew I was a thalassemia trait carrier when we started dated I would have asked him, but I learn about it accidentally when I had my blood tested for an operation...so I guess now is too late"

"I believe that I do not have the right to say to the other person that I want to separate just because we both have stigma. I guess if I had stigma I would asked myself first if I want to have an abortion and ask my partner if he still wanted children. It better to discuss everything than to take decisions the last minute"

"When I met my husband I asked him about stigma. Thank God he did not have it. It is a difficult situation...Really it is better to know...I do not know what I would do...I know couples that went forward and got married..."

"I believe since it has to do with possibilities if you are going to have a child with thalassemia, then it is good to discuss your own possibilities as well and learn as many information you can concerning this situation"

Most of the people in this Group supported the idea that they will probably avoid having a family if they knew that their partner had thalassemia trait as well. However, if they were already in an engagement they claimed that they could always have the option of the abortion. Women with thalassemia trait from this Group seemed to be really conscious about the dangers, which lie in the decision to have children with someone who has thalassemia trait as well, as they stated that for

them it is a priority to ask someone if he is a thalassemia trait carrier before involving in a relationship.

In Group 3, 4 and 5 the majority were already married. Most of them believed that thalassaemia is a serious disease, and they were very concerned about whether it is "*right to have a family if both partners are thalassemia trait carriers*". Their reaction can be concerned reasonable in the sense that most of them were not single and consequently, they were not dealing with the issue of finding a partner or perhaps because most of them had daughters and they were extremely worried about them getting married and having a family – a concern which possibly influences their approach on the matter.

Overall, the majority of the participants from all Groups claimed that they do feel familiar with thalassemia. They believed that thalassemics are physically able to be active members of the society. Generally, all the participants seemed to recognise that thalassaemia used to be more spread among the society but because to an appropriate medical treatment thalassaemic people can live as ordinary as possible.

Most of the participants had colleagues who are thalassaemics and pointed out that "they (thalassemics) are pretty normal and most of them have also families and children". From the participants' responses it is undoubted that thalassemics are concerned to be *physically able to be active members of the society*, but are they practically members of this society? Are they treated equally, without being stigmatized? This question leads us to the next part of the chapter which examines the issue of stigma (thalassemia trait), thalassemia and stigmatization among the society in Cyprus.

(c) Stigma – Stigmatisation

In Cyprus - as mentioned previously - the word "stigma" is commonly used instead of the term "thalassemia trait". The "word" stigma is used in sentences like "He has stigma", or "I am

having a test for stigma". The use of the word "Stigma" in Cyprus is unique as in other countries in which thalassaemia occurs, the word "stigma" does not have the meaning of thalassaemia trait.

The question is whether in Cyprus "stigma" does not only refer to thalassemia trait, but also it encloses the meaning of "stigmatisation" as well. By using the phrase "This person has stigma" do Cypriots unconsciously stigmatise thalassaemia trait carriers? Do thalassemia trait carriers feel the need to "defend" themselves, by reassuring their environment (friends, relatives, partners etc) that what they have *is nothing serious*, or even deliberately hide their condition to avoid any further questions and explanations.

To investigate these questions further the participants were asked to define the word "stigma" and discuss about what they mean when they say that someone has "stigma".

The response to this question was very interesting: 70% of the participants stated that they do not believe that by using the term "stigma" thalassemia trait carriers are stigmatised. They believed that stigmatising these people was a phenomenon which belongs to the past since nowadays everybody knows that "stigma" is not a disease, nor something to worry about. Specifically, some of the responses were:

"Stigma is nothing but a slang term. Its use has been more or less like a habit to people, it does not mean anything, you do not stigmatise someone when you use it"

"Perhaps in the old days if someone had thalassemia trait was stigmatised and that is why we use the word stigma for thalassemia trait today. It does not have the meaning of being stigmatised"

"If somebody nowadays says that: <I have the stigma of the Mediterranean anemia> then it means that: <I have to be careful with whom I am going to get married because then I will have children with stigma>"

"I believe that the phrase <I have stigma> is used as a common code to communicate and understand the same thing. Personally, I do not use the phrase because I do not want to categorize people"

"It is the case that, in the old days stigma was something important and that is why we use the same word today. At the beginning, they stigmatized people who had stigma and today we use this word, but we do not mean something important"

"I believe that the word "stigma" is wrong. Somebody who is not educated may consider stigma as something really bad...Perhaps we should find another word for it"

"Of course when we use the word stigma we tend to stigmatize people....We do that...But because of the prenatal testing people do not pay much attention to the use of the word"

"I have to admit that there is a certain degree of superstition against stigma. I have met families which they broke up weddings and engagements because somebody had stigma. But nowadays, young people do not count stigma as something important"

"I do not think that stigma is used in a negative way nowadays...On the contrary, it is the word thalassemia that it is used as a disadvantage for someone"

This is very encouraging as it shows that people not only understand what thalassaemia trait is, but also they consider it as something acceptable without stigmatising and excluding from the society people with thalassaemia trait.

Nevertheless, there were 20% of the participants who admitted that sometimes by saying that someone has stigma they tend to "feel that there is something negative with the other person" and that "he is not totally healthy":

"I believe that subconsciously we tend to stigmatize people by using this word. I am personally doing this thing. I consider stigma as something negative for the other person something which cannot change. I see it as a disadvantage"

"I think the moment we use this word it takes a negative dimension about a specific person. Either you feel sorry about him or you panic"

"Of course someone with stigma is stigmatized. People tend to observe him and comment about him"

Thalassaemia trait carriers were stigmatized in Cyprus society 50 – 60 years ago when not only there was lack of treatment for thalassemic people and therefore thalassemics died really young but also the majority of Cyprus population were not well educated. In my opinion education played a vital role for people to familiarize with thalassemia trait and thalassemia. Cyprus prevention programme for thalassemia gave great emphasis on educating people by introducing thalassemia and thalassemia trait in school's curriculum. Additionally, it should be also noted that in Cyprus there is the Anti – Anaemia society, which supports thalassaemia patients and promotes research around thalassaemia and its web page in the Internet is easily accessible, explaining a lot about thalassaemia (www.thalassaemia.org.cy).

Definitely, the small percentage (20%) of the participants who felt that thalassemia trait carriers are stigmatized does not represent Cyprus society as a whole. However, it indicates the existence of the idea – though to a small extent - among the society that "stigma" is something "negative" for someone to have.

My hypothesis was that this way of thinking would probably appeared among participants in Group 3, 4 and 5 but I was surprised to discover that participants coming from Group 2 and most surprisingly from Group 1 felt that sometimes "stigma" was something negative for a person to have. As Cyprus strategy (1970) on thalassemia and thalassemia trait was partly based in the induction of thalassemia at High School's curriculum, I was expecting that people who were taught about thalassemia and thalassemia trait at school will not feel that having thalassemia trait is something negative.

In addition, participants with thalassaemia trait suggested that: *"doctors should find another term for thalassemia trait and stop using the word stigma"*, because as they stated sometimes they do mind saying that they have *stigma*: *"Stigma means like I have something serious, but I do not. Why we decided to use this phrase anyway?"*[146]. Specifically, one girl (a thalassemia trait carrier) from Group 2 (20 – 30) said: *"what I am supposed to do? Hold a sign saying "I have stigma do not approach? I do not like this word, I know that what I have is not something serious but I would prefer another word for it"*.

It is undoubted from the above that for a thalassemia trait carrier, the word "stigma" can be a problematic one. Naturally, because the word "stigma" is linked with disgrace or infamy thalassemia trait carriers may feel that by using this word for thalassemia trait they feel as if thalassemia trait is something you must be ashamed of.

To sum up, it is important to mention that the majority of the participants seem to have the knowledge to identify the word "stigma" as nothing more than a term – a different way to name

[146] See also above for responses considering thalassemia trait as something negative

thalassemia trait. However, despite the fact that they did not connect thalassemia trait with thalassemia disorder and that they really believed that a thalassemia trait carrier does not have a disease, a minority appeared to think that sometimes having thalassemia trait can be something negative for a person.

If we assume that stigmatizing means to distinguish a person from the whole (as someone who has something to be ashamed of) then in this sense, when people believe that thalassemia trait can be something negative for a person at the same time they stigmatize him, they exclude him/her – in a sense – from the rest of the people who happen not to have the specific trait. As it can be supposed from the discussion in the present study, thalassemia trait carriers sometimes feel this stigmatization – this exclusion and perhaps that is why they believe that another word for thalassemia trait instead of "stigma" should be used.

CHAPTER 7 – PRESERVING THE OPTION OF FREE CHOICE

Pre-natal diagnosis in Cyprus and especially CVC (Chorionic villus sampling) which involves sampling the mother's placenta and testing for abnormalities solved several problems connected with the free choice of a marriage partner without having to think about the possibility of giving birth to a child with thalassaemia. Couples with thalassaemia trait nowadays with the help of pre-natal diagnosis can decide if they want to have a child with thalassaemia, or otherwise choose abortion.

The participants were asked to comment in the discussion groups on whether they consider pre-natal diagnosis as a safeguard to thalassaemia and whether they believe that it preserves their right of a free choice. To start with, it is important to mention that as prenatal diagnosis is strongly linked with the issue of the abortion some of the participants referred specifically to abortion and expressed some strong opinions about it.

Most of the people who referred to abortion belonged to Group 2. They stated that whether two people get married if they are thalassaemia trait carriers depends mainly on how they feel about abortion. If someone is in favour of abortion then pre-natal diagnosis can be a solution and it can provide the "safeguard" of avoiding children with thalassaemia. On the other hand, if someone is against abortion, then pre-natal diagnosis automatically loses its "*deus ex machina*" sense.

It was very interesting in the case of a couple who drew a similarity with their own case. They mentioned that they are against abortion and that the pre-natal diagnosis for their fourth child showed that the child would have Down syndrome. They did the test twice to make sure and the results were the same. "We were going to keep the baby anyway. Perhaps we did the pre-natal diagnosis just to know the predictable difficulties we were about to face". The baby was born healthy.

Thus, as they pointed out, if someone is against abortion then pre-natal diagnosis is just the means to let you be prepared. In the above couple the woman was a thalassemia trait carrier but the

husband was not, but they both stated that even if both had stigma they would definitely not choose abortion.

Norman Ford referring to Cyprus case mentions: *In countries where there is a known high risk of thalassemia (...) there has been a successful switch from prenatal testing to premarital screening. The Greek Orthodox Church in Cyprus requires screening for this common recessive genetic disease if couples wish to marry in the Church, but there is no requirement for them to make the results known and they are free to marry even if they know they are both carries of the gene (...). This enables engaged couples to make their own informed and morally responsible decision about marriage and parenthood*[147] .

It is undoubted that, this *switch from prenatal testing to premarital screening* aimed to sensitize people about thalassemia and make them think carefully about the responsibilities of giving birth to a thalassemic child.

The question is: can we argue that pre-natal diagnosis in Cyprus case resulted in responsible decisions concerning thalassaemia? According to the expressivist argument *prenatal tests to select against disabling traits express a harmful attitude about and send a hurtful message to people who live with those same traits*[148]. As it can be assumed from the part on "stigma – stigmatization", this harmful attitude may already be a reality in Cyprus.

In order to examine this issue further it is interesting to look at some answers coming from Group 1 stating that "what is the point of examining thalassaemia trait anyway since someone can have an abortion if he/she chooses to do so. There is 25% chance of giving birth to a child with thalassaemia. If someone is so unlucky, then she will have an abortion".

It is true that premarital screening solved many problems in Cyprus as thalassemia births fell - during the first year of Cyprus prevention programme - from 15% to 2% of expected. However,

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[147] Ford, N., *The prenatal person: ethics from conception to birth,* Blackwell 2002, p. 126

[148] Parens, E., and A. Asch,, "Disability rights critique of parental genetic testing: reflections and recommendations", *Mental Retardation and developmental disabilities research reviews* 9: 40 – 47 (2003)

judging from people's responses the issue is whether this switch to premarital screening did fulfil its aim. Premarital testing does not seem to be a means for further consideration in order for the couple to realize the responsibility of giving birth to a child with thalassemia. This is not necessary as in many cases couples simply select abortion in case of a thalassemic child.

As it can be assumed from the above, another reason why people do not reveal to their partners that they are thalassemia trait carriers may be the fact that they believe it is pointless since they can always have the option of the abortion. However, people react differently towards abortion, therefore choosing not to disclose this information may not always be the right way of acting.

Summing up, it should be stated that by introducing pre-natal diagnosis along with premarital screening Cyprus succeeded in eliminating the number of births with thalassemia. However, it is doubted whether the combination of premarital screening and prenatal testing, succeeded in making people more responsible about their decisions. There are not really any criteria which may clearly indicate whether people became more responsible or irresponsible since 1970 (when Cyprus strategy on thalassemia was lounged). Nevertheless, judging by the answers of many of the participants in this study, it can be assumed that premarital testing can be considered from lost of people as a routine examination (without necessarily be a means to increase someone's responsibility towards thalassemia).

Thus, from this part of the study emerges the question of what is the need of premarital screening, since nowadays we have available an improved set of genetic technologies which can provide us with the necessary genetic information in order to take the appropriate decisions. Does premarital testing serves the same needs as it used to do back in 1970 where genetic technologies were not so developed?

CHAPTER 8 – RECOMMENDATIONS

It can be concluded that Cyprus strategy on thalassaemia has been effective in decreasing the birth prevalence of thalassaemia in Cyprus and therefore a large amount of medical expenses has been reduced. However, summing up the theoretical and the empirical part of this thesis I would like to suggest the following improvements which can help Cyprus to improve its screening programme and at the same time to allow people in Cyprus take their own decisions concerning their genetic information:

- The offer of prenatal screening must be voluntary for all couples. Orthodox Church in Cyprus should allow people to get marry and not to be screened for thalassaemia trait if they do not want to.

- Cyprus Government should ensure close contact with the families of affected children for personal help and support and better public information. People in Cyprus and especially young couples should be informed how affected people can work, live and enjoy life as members of the society. It is important to have in mind that life span of thalassaemic people now has been expanded and that they do not face the danger to die young as it was happening back in the eighties.

- Current Cyprus strategy should be changed or revised in order to follow European guidelines on genetic screening, which stress out the importance of voluntariness. People should be let to decide whether they want or not to possess this kind of information concerning thalassaemia trait and should not be forced to know it anyway.

- A new fresh approach on thalassaemia will eliminate any stigmatization currently existing among the society concerning male or female carriers. The word "stigma" should not be used in order to "exclude" or to differentiate people from the society as thalassaimia trait carriers.

- In conclusion, screening programme in Cyprus should not be oriented in maintaining the 0% in b-thalassaemia births but in preserving peoples' free choice in genetic testing. Informational autonomy should be valuated as something important and Cyprus government should make any efforts needed to protect and ensure this autonomy.

CHAPTER 9 - CONCLUSION

In the beginning of the thesis genetic information was identified in the context of genetic testing. Informational autonomy is the freedom and the right for people to have power over their own personal information. As such, informational autonomy is not a new feature. Protecting the privacy of personal information is a legally protected right - not only in Cyprus but internationally as well - and it emerged as a need partly because of the computerization of personal information, and the increased reliance public and private organizations placed on the collection, use, storage and exchange of personal information. The feature that distinguishes informational autonomy concerning personal information from informational autonomy concerning personal genetic information is the sensitivity of genetic information compared to other kind of information.

The traditional concept of informational autonomy was developed to protect the collection and use of personal information in both the public and private sectors. Any kind of information may only be collected with the consent of the individual. This traditional concept of informational autonomy entails that the individual must be fully informed of the purpose of the collected information and only the information necessary to accomplish this purpose may be collected and it may only be stored until that purpose is fulfilled.

In genetic research it is very difficult to describe a priori any implications the specific research may have. Genetic samples may be collected initially for a specific purpose but precisely because of their sensitive nature, the research may have more implications that can be possibly predicted even by the researcher himself. The results of a genetic research may contain an interest not only for the provider of the sample but also for family members, insurance companies or even employers. The nature of genetic information poses certain ethical questions and challenges, both for individuals and families, as well as for those persons and institutions that handle this information, such as medical practitioners, scientific researchers, hospitals etc.

For this reason, a central idea for this thesis is that genetic information is fundamentally different from other forms of personal health information and that it requires special regimes to regulate its collection, use and disclosure.

This thesis argues that each person's genetic information is unique since it can also reveal information about and therefore has implications for, that person's blood relatives. This is what gives to genetic information a special status.

Demonstrating that an individual is a carrier of a mutated allele for cystic fibrosis means that one of that person's biological parents is also a carrier, and that his or her siblings may be affected or may also be carriers. As sensitive health information, an instinctive reaction towards genetic information is to provide a high level of privacy protection.

On the one hand, precisely because genetic information is familial in nature, it can often provide great relief to those who receive the data. It is relatively rare that individuals learn of a risk through genetic testing that they did not already anticipate. On the other hand, some family members may wish to assert a "right not to know" the results of a test taken by a family member concerning a serious genetic disorder, such as Huntington's disease. Many people choose to organize their lives without the shadow of such information.

Information generated by genetic testing can be very precise, indicating whether a particular allele or mutation is or is not present. However, very often genetic information tends to be about possibilities rather than certainties, because only a proportion of those people with a particular disease-related mutation will go on to develop the disorder.

Its predictive nature makes it very interesting especially in situations where information about a person's future, even though it can be imprecise, it can lead into very important decision making by the individual or by others, such as employers, insurance companies or public health authorities. On the other hand, genetic information has the potential to empower people to make better choices about health and medical care for themselves and their families.

For the above reasons this thesis stand in favor of informational genetic autonomy. This autonomy allows people to have the freedom to make choices among alternative sets of information, ideas, and opinions. This includes the freedom to decide what information someone wants to receive and process. Additionally, informational autonomy as an aspect of individual liberty necessitates that everyone has the right to express his/her own beliefs and opinions and that everyone should give their consent voluntary and free of undue influence or coercion. Human genetic information is ultimately, not about genes; it is about people.

When the Cyprus strategy on thalassemia was launched back in 1970 it had as a target to control the number of people with thalassemia major. The challenge for Cyprus in the 21st century is to advance the existing control programme. By introducing pre-natal diagnosis along with premarital screening Cyprus succeeded in eliminating the number of births with thalassemia, but increased the number of abortions at the same time.

As this thesis argued it is doubted whether the combination of premarital screening and prenatal testing, succeeded in making people more responsible about their decisions, asking at the same time some serious questions concerning the option for people to manage their own genetic information.

Since nowadays there is available an improved set of genetic technologies which manages to extend the life span of thalassemics (Desferral treatment) and provide more options (PIGD) then we should ask whether premarital testing serves the same needs as it used to do back in 1970. People in Cyprus must have the option to know or not know if they are thalassemia trait carriers and should not be "forced" to do a premarital testing. Informational autonomy is a right that Cypriots are allowed to have.

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APPENDICES

APPENDIX I: FIGURES

Figure 1. Probabilities when one parent (i.e. father) is thalassemia trait carrier

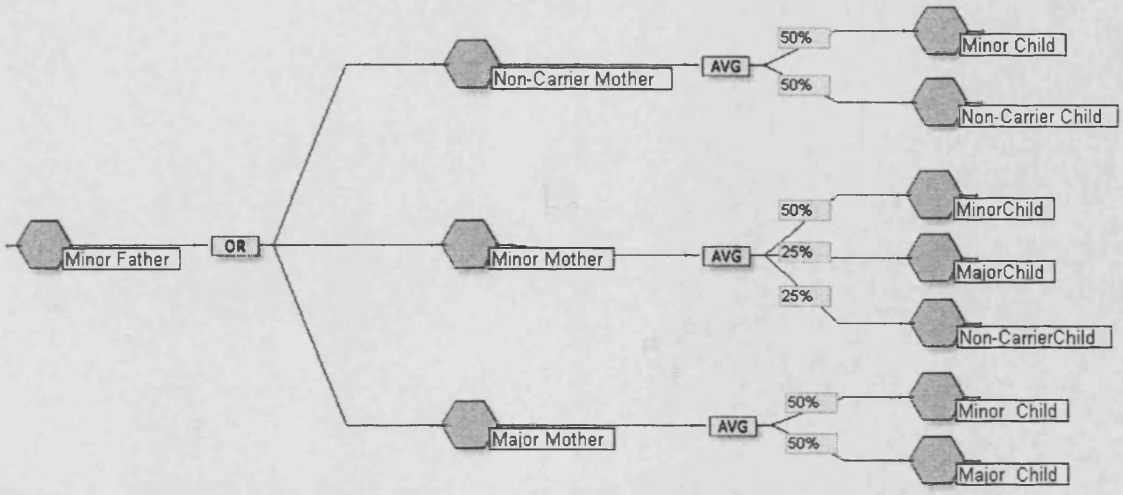


Figure 2. Probabilities when one parent (i.e. father) is thalassemic major

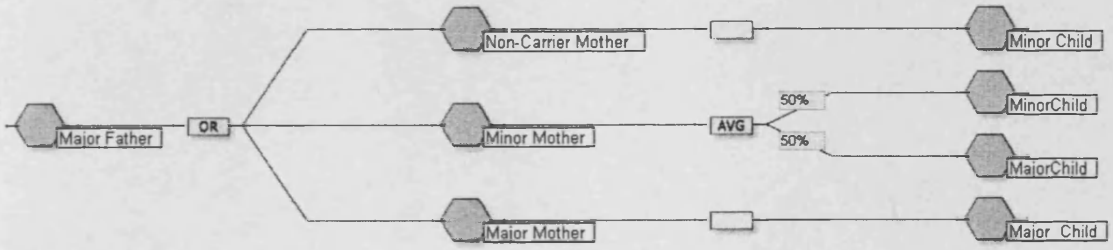
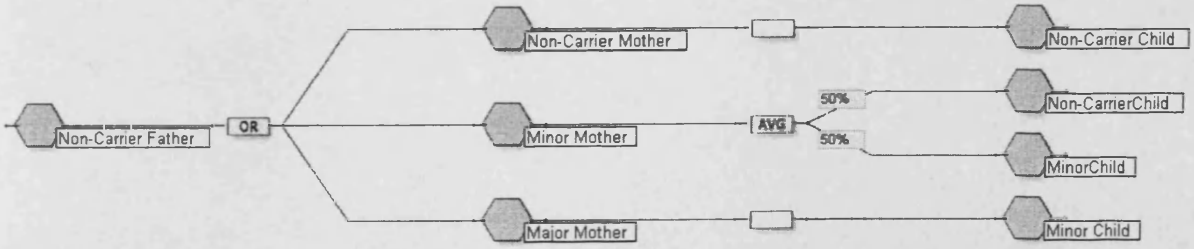


Figure 3. Probabilities when one parent (i.e. father) is non-carrier



APPENDIX II: QUESTIONAIRE

(a) The original version of the questionnaire as it was given in Greek language

1. Ακούγοντας τη λέξη γενετική τί σου έρχεται στο μυαλό;

2. Κατά πόσο συμφωνείτε ή διαφωνείτε με την ακόλουθη φράση: Οι νέες γενετικές ανακαλύψεις θα βοηθήσουν στην θεραπεία πολλών ασθενειών.

3. Κατά πόσο συμφωνείτε ή διαφωνείτε με την ακόλουθη φράση: Η έρευνα στην ανθρώπινη γενετική παρεμβαίνει στη φύση και είναι συνεπώς ανήθικη. Πώς καταλαβαίνετε τον όρο «παρεμβαίνει στη φύση». Θεωρείτε π.χ. ότι τα γενετικά μεταλλαγμένα προϊόντα αποτελούν παρεμβολές σε μια φυσική διαδικασία, ή ακόμα η κλωνοποίηση στα ζώα ή στον άνθρωπο; Πιστεύετε ότι υπάρχει γενετική έρευνα που θα μπορούσε να κατηγοριοποιηθεί ως ανήθικη και αντίστοιχα ηθική;

4. Κατά πόσο συμφωνείτε ή διαφωνείτε με την ακόλουθη φράση: Η γενετική πληροφορία μπορεί να χρησιμοποιηθεί από γονείς για να αποφασίσουν εάν θέλουν να αποκτήσουν παιδιά που πάσχουν από γενετικές ασθένειες. Ποιες ασθένειες νομίζετε ότι θα μπορούσαν να κατηγοριοποιηθούν ως σοβαρές γενετικές ασθένειες τις οποίες οι γονείς θα μπορούσαν ίσως να θεωρήσουν ανεπιθύμητες;

5. Κατά πόσο συμφωνείτε ή διαφωνείτε με την ακόλουθη φράση: Πρέπει πάντα να εξασφαλίζεται η συγκατάθεση πριν τη χρησιμοποίηση οποιοδήποτε δείγματος αίματος ή οποιοδήποτε δείγματος DNA σε κάποια γενετική έρευνα.

6. Ένας οργανισμός χρηματοδοτείται από την Κυβέρνηση και επενδύει μεγάλα οικονομικά ποσά καθώς και αρκετό χρόνο για την ανακάλυψη κάποιου καινούριου τρόπου χρησιμοποίησης μιας ανθρώπινης γενετικής πληροφορίας. Πιστεύετε ότι η συγκεκριμένη γενετική πληροφορία πρέπει να βρίσκεται αποκλειστικά στη διάθεση της Κυβέρνησης και συνεπώς να είναι διαθέσιμη δωρεάν και σε όποιον ενδιαφέρεται ή να είναι στη διάθεση κάποιου ιδιωτικού οργανισμού και αυτός να έχει τα αποκλειστικά δικαιώματα; Υπάρχει πιστεύετε διαφορά μεταξύ μιας Κυβερνητικής έρευνας και μιας ιδιωτικής έρευνας;

7. Ακούγοντας τη φράση ανθρώπινη γενετική πληροφορία τί σου έρχεται στο μυαλό;

8. Σε ποιους από τους παρακάτω τρόπους πιστεύεται πως θα μπορούσε να χρησιμοποιηθεί η ανθρώπινη γενετική πληροφορία και γιατί: α) Στην επιλογή από τους γονείς των φυσικών και πνευματικών ικανοτήτων των παιδιών τους. Πιστεύετε σ' αυτό το δικαίωμα των γονιών; β) Στην έρευνα για την πρόοδο χημείας και βιολογίας γ) Στο διακανονισμό ασφαλιστικών ποσών δ) Στην ανάπτυξη τεχνολογιών για τον ακριβή εντοπισμό τυχόν γονιδιακών ανωμαλιών ε) Στη ανάπτυξη

τεχνολογιών για τον ακριβή εντοπισμό τυχόν γονιδιακών ανωμαλιών επόμενων γενεών στ) Στη μελέτη της εξέλιξης και της προόδου του ανθρώπινου γένους

9. Διενεργείτε μια γενετική εξέταση είτε καθαρά για δικούς σας προσωπικούς λόγους είτε στα πλαίσια μιας γενετικής έρευνας. Πιστεύετε ότι μια ασφαλιστική εταιρεία θα πρέπει να έχει το δικαίωμα να ζητά τα αποτελέσματα αυτής της εξέτασης για να οριστούν π.χ. το ποσό των ασφαλίστρων που θα πληρωθούν < π.χ. αν ο ασφαλιζόμενος στο μέλλον πρόκειται να αντιμετωπίσει μια σοβαρή ασθένεια τότε θα πρέπει να πληρώνει ίσως πιο πολλά ασφάλιστρα από κάποιον που πρόκειται να είναι υγιής >

10.Αν ένας γιατρός εξετάσει ένα ζευγάρι το οποίο προτίθεται να δημιουργήσει οικογένεια και ανακαλύψει ότι σύμφωνα με το ιατρικό τους ιστορικό πολύ πιθανόν τα παιδιά τους να κληρονομήσουν μια γενετική ασθένεια ή μια ασθένεια που θα τους δίνει περιορισμένο χρόνο ζωής νομίζετε πως ο γιατρός α) θα πρέπει να τους αποκαλύψει αυτή την πληροφορία β) να κάνει διαθέσιμη την πληροφορία και σε άλλους επιστήμονες κλπ

11.Αν ενώ έχετε επιλέξει το/την μέλλοντα σύζυγο σας ανακαλύψετε με μια γενετική εξέταση πως και οι δύο έχετε το στίγμα και συνεπώς τα παιδιά σας πολύ πιθανόν να έχουν θαλασσαιμία θα προχωρούσατε στη δημιουργία οικογένειας;

12.Κατά πόσο συμφωνείτε ή διαφωνείτε: "Η δημιουργία μια τράπεζας δειγμάτων DNA θα ευνοούσε την έρευνα γύρω από τις γενετικές ασθένειες που εμφανίζονται στην Κυπριακή κοινωνία".

13.Αν σας ζητούσαν να δώσετε αίμα για τη δημιουργία της πιο πάνω τράπεζας θα δεχόσασταν ή όχι και γιατί;

14.Πιστεύετε ότι δίνοντας ένα δείγμα DNA σε μια τράπεζα δειγμάτων απειλείται με οποιοδήποτε τρόπο ο θεσμός της οικογένειας ή οτιδήποτε έχει να κάνει με την προσωπικότητα ή τη μοναδικότητά σας ως ανθρώπινη ύπαρξη; Τί σημαίνει για σας η φράση «ιδιαιτερότητα του κάθε ανθρώπου»; Νομίζετε ότι αυτή απειλείται ή παραβιάζεται με οποιοδήποτε τρόπο;

15.Κατά πόσο συμφωνείτε ή διαφωνείτε με την ιδέα για δημιουργία εκ μέρους της Αστυνομίας μιας τράπεζας δειγμάτων DNA για ανθρώπους που καταδικάζονται για διάφορα σοβαρά αδικήματα. Θα συμφωνούσατε αν ο κατάλογος των αδικημάτων συμπεριελάμβανε τα παρακάτω αδικήματα: α) φόνο β) σεξουαλικά αδικήματα γ) Ληστεία δ) Οδήγηση κάτω από την επήρεια αλκοόλ ε) Απάτη

16.Βάλτε τα παρακάτω με τη σειρά συχνότητας εμφάνισης στην Κυπριακή κοινωνία: α) Καρκίνοι β) Θαλασσαιμία γ) Καρδιακά νοσήματα δ) Σύνδρομο Down

17.Πώς πιστεύετε ότι χρησιμοποιείται το δείγμα αίματος που παρέχετε σε νοσοκομεία ή Κλινικές για γενικές αναλύσεις; Πιστεύετε ότι αχρηστεύεται και πετάγεται μετά την ανάλυση; Αν όχι πώς νομίζετε ότι χρησιμοποιείται;

18.Πιστεύετε ότι ο προσωπικός σας γιατρός θα σας ρωτούσε πριν παρέχει οποιεσδήποτε προσωπικές σας γενετικές πληροφορίες σε έρευνες ή άλλους επιστήμονες;

19.Πώς θα αντιμετωπίζατε το γεγονός εάν μαθαίνατε ότι ο προσωπικός σας γιατρός έδωσε το ιατρικό σας ιστορικό για να χρησιμοποιηθεί σε μια γενετική έρευνα ή για τη δημιουργίας μιας τράπεζας ιατρικών δεδομένων;

20.Πόσο οικείοι αισθάνεστε με τον όρο: τράπεζα γενετικών πληροφοριών; Τί νομίζετε ότι είναι;

21.Τί επιπτώσεις (αρνητικές ή θετικές) νομίζετε ότι μπορεί να έχει η ανακάλυψη ενός γονιδίου συγκεκριμένα στον Κυπριακό λαό το οποίο να ευνοεί την εμφάνιση κάποιας συγκεκριμένης γενετικής ασθένειας;

22.Τί νομίζετε ότι είναι η θαλασσαιμία;

23.Τί νομίζετε ότι σημαίνει η φράση: "έχω το στίγμα" ;

24.*Στίγμα: 1. το σημάδι που αφήνει στο δέρμα έγκαυμα ή τραύμα ή που προέρχεται από παθολογική αίτια 2. κηλίδα συγκεκριμένου χρώματος 3. ο λεκές 4. οτιδήποτε προκαλεί ντροπή ή κοινωνική καταδίκη <σημειώστε ότι ετυμολογικά προέρχεται από το ρ. στίζω, στίγ-jω: 1. προκαλώ στίγματα σε (κάτι) 2. δηλώνω γραπτώς τα σημεία στίξεως σε (κείμενο).>* Πιστεύετε ότι η έννοια στίγμα στην πιο πάνω φράση μπορεί να ταυτιστεί με κάποια από τις πιο πάνω έννοιες;

25.Έχετε στην οικογένεια σας θαλασσαιμικό; Πιστεύεται ότι η θαλασσαιμία εμφανίζεται σε μεγάλο βαθμό στην Κύπρο; Νομίζετε ότι αποτελεί πια μέρος της καθημερινής ζωής και ότι ο Κύπριος νιώθει οικεία πλέον με τη θαλασσαιμία ως αρρώστια;

26.Πιστεύετε ότι οι εργοδότες θα πρέπει να δικαιούνται να έχουν πρόσβαση στο ιατρικό ιστορικό του εργοδοτουμένου και να ενημερώνονται για τα αποτελέσματα τυχόν γενετικών εξετάσεων που διενεργούν οι εργοδοτούμενοι;

27.Αν καλούσασταν να δώσετε ένα δείγμα αίματος για μια γενετική έρευνα η οποία θα αρχίσει κάποια χρόνια μετά που θα δώσετε το δείγμα σας και για κάποια γενετική έρευνα η οποία πρόκειται να αρχίσει αμέσως μετά την παροχή των δειγμάτων, ποια θα προτιμούσατε και γιατί;

(b) Translation of the questionnaire in English language

1. When you listen to the word "genetics" what springs to your mind?

2. In what extend do you agree or disagree with the following phrase "New genetic developments will bring cure for many diseases"?

3. In what extend do you agree or disagree with the following phrase "Human genetic research violates nature and therefore is unethical"? How can you understand the phrase *violates nature*? Do you consider that e.g. the genetically modified products, animal or human cloning, constitute a violation in a natural process? Do you believe that there is a specific genetic research that can be characterized as ethical or unethical?

4. If you were asked to give your blood sample for a genetic research which is going to be contacted years after you give your sample and for a genetic research which is going to start instantly. Which one will you prefer and why?

5. In what extend do you agree or disagree: "Permission always be sought before any blood samples or tissues are used in a genetic research"?

6. When you listen to the phrase "human genetic information" what springs to your mind?

7. In what extend do you agree or disagree: "Genetic information may be used by parents to decide if they want to bring into the world children with disabilities"? Which diseases in your opinion potential parents may categorize as very serious?

8. An organization is funded by government and invests large amounts of money and time to a project concerning a new way to use human genetic information. Do you believe that the information must be publicly owned and available to anyone who wants to use it without any charges? Do you believe that there is any substantial difference between a genetic research conducted by the Government and a genetic research conducted by a Private Organization?

9. In which of the following ways do you believe that genetic information may be used and why: (a) parents choosing physical and mental characteristics of their children. Do you believe that parents should have this right? (b) Research for the progress of biology and chemistry (c) For setting the level of insurance premium (d) Developing techniques to correct defective genes for individuals (e) Research concerning evolution, ancestry and population

10.You are taking a genetic test either for personal reasons or because you are participating in a genetic research. Do you believe that an insurance company should have the right to ask for the results of this test i.e. to set the level of the insurance premium?

11.If a GP examines a couple and discovers that according to their medical history there is a great possibility for their children to inherit a genetic disease or a disease which does not allow them to live very long. In your opinion do you believe that the doctor should a) disclose this information to the couple b) disclose this information to any other scientists or organizations.

12.Do you believe that you GP will get your permission first before giving any of your personal genetic information to other doctors or scientists to use it in a genetic research?

13.Do you believe that the employers should have the right to access the medical history and be informed for the results of any genetic tests their employees are taking or took in the past?

14.How familiar do you feel with the term: *human genetic information bank*? What do you think this bank is for?

15.In what extend do you agree or disagree: "The construction of a databank which will include samples form all the Cypriots will help genetic research to examine genetic diseases that tend to appear among Cypriots".

16.If you were asked to donate blood for the construction of the above databank, will you accept? Explain why.

17.Do you believe that by giving your DNA sample to a biobank, "family" is influenced or affected in any way? How about your personality or individuality?

18.In what extend do you agree or disagree with the construction on behalf of the Police of a Databank which will include DNA samples of people convicted for the following crimes: (a) Murder (b) Burglary (c) Drinking-Driving (d) Fraud (e) Shop lifters? Comment on the above list and suggest any other crimes you believe should be included in the list.

19.Do you believe that the blood samples you give for general medical examinations can reveal any personal genetic information? How do you believe these samples are used after the analysis?

20.How will you react if you realize that your GP gave your medical history and some of your personal genetic information to be used in a genetic research without taking your permission first? If you find out that he did so in order to help in the construction of a databank that will contribute in finding cures for many diseases, how your reaction will be?

21.What in your opinion will be the impact if scientists discover the existence of a particular gene in Cypriots, which proves to be the reason of the appearance of "stigma" in many Cypriots?

22.Put the following medical conditions in order starting from the one you believe it appears more among Cypriots: a) Cancers b) Thalassaemia c) Heart diseases d) Down Syndrome

23.What do you think *thalassemia* is?

24.What do you think the phrase "I have *stigma*" exactly means?

25.*Στίγμα: 1. το σημάδι που αφήνει στο δέρμα έγκαυμα ή τραύμα ή που προέρχεται από παθολογική αίτια 2. κηλίδα συγκεκριμένου χρώματος 3. ο λεκές 4. οτιδήποτε προκαλεί ντροπή ή κοινωνική καταδίκη <σημειώστε ότι ετυμολογικά προέρχεται από το ρ. στίζω, στίγ-jω: 1. προκαλώ στίγματα σε (κάτι) 2. δηλώνω γραπτώς τα σημεία στίξεως σε (κείμενο).>* Do you believe that the word "stigma" in the phrase "I have *stigma*" can take any of the above meanings? **The Greek part is the explanation from a Greek vocabulary concerning the word stigma. Specifically the explanations are: 1. a mark or injury on human skin which is caused due to fire or any other pathological reason 2. stain of a specific color 3. dirtiness 4. what causes shame or social conviction.**

26. Do you have in you family someone that has thalassaemia? In what extend do you believe thalassaemia tends to appear among Cypriots? Do you think we could argue that thalassaemia is nowadays part of our everyday life?

27. Let us assume that you already met your future husband and you are planning to marry him, but you discover after a genetic test that you both have "stigma" and consequently your children are possibly going to suffer from thalassaemia, how do you react? Are you going to follow your plans concerning family?