

PREDICTIVE MEDICINE AND DISCRIMINATION



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INTRODUCTION

On 26 February 2001, coinciding with the publication of the human genome sequence in *Nature* and *Science* journals, the Grífols Foundation brought together a panel of experts to discuss the bioethical issues raised by the advances in our understanding of the genome, namely the relationship between the benefits and potential dangers of predictive medicine.

In fact, the title of the seminar, “Predictive Medicine and Discrimination”, highlights one of the basic problems that can arise from knowledge of our genetic make-up, of gene mutations, and of the diseases and predisposition to disease these changes can cause.

Some of the bioethical aspects of predictive medicine were addressed last year during the “Genetics, Ethics and Politics” course organised by the Grífols Foundation and the International University Menéndez Pelayo.

In the seminar now being published, the discussion is based on speeches given by Dr. Xavier Estivill, who analysed the possibilities and limitations of predictive medicine, and by Dr. Mirentxu Corcoy, who from the legal standpoint, provided a point-by-point review of the benefits and perils that could originate from the use of predictive medicine, as well as the aspects now covered by legislation and those that should be regulated. These talks were complemented with contributions from several experts, which are presented in the appendix.

The basic points on which both the speakers and the experts agree mainly concern the fine balance we must strike among the various facets of the issue: the ability to detect genes for a certain pathology or that indicate predisposition or susceptibility, the need to obtain informed consent from the individuals undergoing genetic testing, the problems resulting from the invasion of privacy and confidentiality, the right not to know, the potential dangers that concealing information could entail for third parties, and the discrimination that may result from the proper or improper use of the results obtained.

The discussion led to a number of conclusions, primarily of a precautionary nature, that were derived from the idea that predictive medicine may not be the cure-all some are dreaming of and others are heralding. It was even expressed that a linear interpretation of genetic data, ignoring both gene

interactions and the influence of the epigenetic factors, is completely inadmissible.

While allowing that some data could have collateral discriminatory effects, the discussion pointed out the falseness of what has erroneously been called eugenics, the possibility that abnormal genes can be eradicated or children produced *à la carte*. In real terms, when applied individually, this is actually aimed at promoting health.

As a whole, the talks presented and the experts' comments can be considered a key contribution to the debate that must take place between social players in response to the advances that will unquestionably lead to radical changes within medicine in the future. This debate must also consider the possibilities of coverage of these new medical advances by healthcare systems and the fact that they will require a hierarchy of services that will not be simple, always right or even accepted at all times.

JOSEP EGOZCUE
Vice President of the
Grifols Foundation

ON THE ROAD TOWARD PREDICTIVE MEDICINE?

Xavier Estivill

Research on the genome of humans and other organisms has enabled us to identify the genes of the main hereditary diseases. In upcoming years we will obtain information on the genetic susceptibility of developing the most common diseases that affect us: asthma, cancer, diabetes, hypertension, psoriasis, schizophrenia, depression and anxiety, among others. This information will lead to further understanding and to the use of applications for the diagnosis, prevention, treatment (and cure) of diseases. Access to information on the risk of developing disease and the possibility to know more about an individual's character and personality involve trespassing the very depths of individual privacy. This new knowledge brings hope, while also creating suspicion and fear. Will we know how to properly use information on the genome sequence? Will predictive medicine be used to prevent, treat and cure disease or to discriminate against individuals with genomic weaknesses? For the time being, the lack of therapeutic measures for many diseases means that prediction is essentially a curse.

Genetic tests have a well-defined value in the clinical context. However, their use in nonclinical situations –for example, at the request of an insurance company or during the hiring process– pose an entirely different situation. Insurance companies would like to use genetic tests in the same way that they use data on medical or family history. Business owners want to be sure that their employees do not have risk factors or characteristics that could affect their capacity or ability in the workplace, endangering their safety or the safety of others. The technical aspects of genetic diagnosis are developing at a rapid pace and the related ethical and social aspects must be considered without delay. At present the accuracy of the predictive capacity of genetic testing is very difficult to establish and varies, depending on the type of disease and on the genetic test. In addition to the inherent problems of applying the information to the individuals directly involved, predictive tests also affect families and specific groups. Denial of employment or insurance on the basis of specific genetic traits could have very serious consequences and could affect families and groups of individuals already at a disadvantage with respect to the rest of society. Although there may be extremely radical initial attitudes toward this issue, considerable debate aimed at narrowing the distance between the various postures is needed. The debate becomes even more important when we consider that current health systems will experience progressive waves of restrictions on budgetary resources and on the services provided to users. The implementation of predictive medicine will require that society adapts its educational and healthcare structures to the new situation brought about by the scientific advances.

Human genome sequence: Why?

We must recognise that our knowledge of the cause of disease is still extremely limited. In many cases treatment is limited to arresting the symptoms or treating the complications of a specific disease. As physicians, we still do not have adequate solutions for most diseases, particularly with regard to combating disease at its origins.

Our current biological understanding of the human organism is reduced to less than 5,000 proteins of the hundreds of thousands (if we consider their variants) estimated to be involved in the functioning of the body. Moreover, we still do not know everything about how proteins interact with each other to perform the various functions of cells, tissues and organs. In the light of this lack of knowledge, it is not surprising that we do not have proper tools to treat psychiatric diseases, hypertension, diabetes, Parkinson's disease, Alzheimer's disease, asthma, cancer, inflammatory skin diseases, etc.

The shortest route to accessing information on all the proteins of the human body is to identify the DNA sequence, which contains the instructions for each and every human gene and its respective proteins. In fact, obtaining the genome sequence of humans and other organisms, the so-called "Periodic Table of Human Biology" was the main objective of the Human Genome Project. This gateway provides access to the entire body of information related to the functioning of our organism and creates a branch of medicine grounded on scientific knowledge of humans and human biology. All biomedical research conducted in the future will be based on the data in the genome sequence. The information in the human genome constitutes a scientific cornerstone that will allow further developments in the fields of human physiology and pathology.

Recent years have seen numerous advances in the study of the human genome, and we have obtained extraordinary results in the identification of genes related to a number of hereditary diseases (cystic fibrosis, Huntington's chorea, neurofibromatosis, polycystic kidney disease, muscular dystrophies, hereditary breast and colon cancer, mental retardation, Alzheimer's disease, hereditary deafness and blindness). These breakthroughs have opened the door to the molecular diagnosis of a considerable number of genetically-based diseases, allowing the confirmation of these diseases, as well as early, prenatal or preimplant diagnosis and the identification of carriers.

Human genome sequence: useful for what?

When we look at the consequences of our knowledge of the genome, it seems clear that in upcoming years we will progressively develop a better use of medications on the basis of genetic determining factors. Comprehensive genetic testing that allows simultaneous detection of an extensive number of genetic defects will most certainly take more time. In the next ten years we will be able to analyse an individual's susceptibility to develop a dozen common diseases, and we will undoubtedly be able to take steps to reduce the risk that the disease will manifest. Broad diagnostic testing for several pathologies will be possible. When that time arrives, it will be necessary to have well-developed legislation that avoids genetic discrimination and guarantees the privacy of the individual's genetic information.

The new medicine derived from our knowledge of the genome –known as Genomic Medicine– will have enormous repercussions on health. However, its impact will depend on our capacity to modify the attitudes of individuals with risk factors for developing specific diseases. This may involve changes in diet, job and lifestyle, but it also implies the availability of therapies that will make it possible to modify the consequences of genetic susceptibility, or that effectively act on the disease once it develops. Within twenty years, the armamentarium will include drugs based on our knowledge of genes for a considerable number of common pathologies, including diabetes, hypertension, psychiatric disease, asthma, etc. Application of our knowledge of the genome to the use of medication will be standard practice for managing all pathologies. Gene therapy will be fully developed and constitute a basic tool in our effort to conquer disease.

Human genome sequence: useful for whom?

What causes some individuals (and not others) to suffer from cardiovascular, respiratory, psychiatric and neurodegenerative diseases? For the most part, the answer is probably found in the variants existing in our genetic material. One out of every 1,000 nucleotides of our DNA is variable. Considering that human DNA is composed of 3,000 million nucleotides, we can expect that there are more than three million variable sites, with specific combinations for each person. These variants are mainly due to changes in the sequence of one or more nucleotides and are known as SNPs (single nucleotide

polymorphisms). Some SNPs occur relatively frequently and can be studied on a population basis. Some SNPs involve functional changes for a specific protein (variations in its levels or characteristics). Single nucleotide polymorphisms can be used as markers to identify the genetic factors predisposing an individual to common diseases (asthma, diabetes, hypertension, schizophrenia, depression, anxiety, thrombosis, cancer, autoimmune diseases, etc.). The use of SNPs in the study of these diseases will make it possible to identify genetic susceptibility factors and facilitate our understanding of the main biochemical pathways involved in their development.

Genetic susceptibility studies using SNPs do not involve detecting an existing disease, but rather identifying risk factors for the development of a disease. Genetic variants resulting in a higher risk for developing certain diseases have already been identified. A variant in the gene for protein C, which inhibits coagulation, appears in 5% of individuals and represents an important risk factor for developing thrombosis. There is a mitochondrial DNA mutation that increases an individual's susceptibility to deafness, particularly in patients treated with certain antibiotics. Certain mutations in the haemochromatosis gene (present in one of every 500 individuals) facilitate the development of cirrhosis of the liver. One of every 40 individuals carries a mutation of the connexin 26 gene that is responsible for congenital deafness. A variant in the cystic fibrosis gene causes infertility in males. Individuals with allele 4 of the APOE gene have a greater risk of developing Alzheimer's disease.

In upcoming years more information will become available on the genetic factors involved in psychiatric diseases (depression, anxiety, anorexia, bulimia and schizophrenia). Genomic studies of these pathologies will have an enormous medical and social impact, allowing pharmacological and cognitive therapies to be applied well before the diseases develop fully. Nevertheless, until we have detailed information on the implications of genetic factors in the development of disease, genetic studies must be used only for diagnostic rather than predictive purposes. Significant ethical issues are raised in this setting, and must be tackled as soon as possible. Despite the major role of genes in the genesis of common diseases, environmental factors are also key components in their development. These factors can be modified, with examples including diet in hypertension, allergens in asthma, sugar in diabetes, etc. An understanding of genetic susceptibility factors will facilitate the introduction

of changes in a person's lifestyle, but will also make it possible to develop specific treatments that allow genetic susceptibility to be modified or that act effectively on the pathology.

Genetic testing in Spain: insurance companies and employment

The information derived from knowledge of the human genome and the genomes of other organisms will result in sweeping changes in all medical specialties and will be a mainstay in the future diagnosis, prevention and treatment of disease. The new biomedical knowledge unquestionably affects the role played by medical genetics, a specialty that does not yet exist in Spain but is particularly relevant in developed countries. One of the main missions of medical genetics is genetic counselling, whereby an individual is informed about a disease, its mode of inheritance and the risks of developing it and/or transmitting it to one's offspring, and is offered solutions and support. This medical assistance must be provided solely by trained specialists in this field who can give patients up-to-date information on diseases having a genetic basis. Armed with better information on the genome and on genetic susceptibility factors, the geneticist will clearly play an increasingly important role in genetic counselling and in molecular biology studies related to disease.

Genetic analyses focus on two types of disease, namely monogenic diseases and multifactorial, or polygenic, diseases. The basic difference between the two categories resides in the greater importance of genetic factors in monogenic disease. In the case of monogenic diseases, individuals who have inherited the allele related to the disease will almost certainly develop the disease. Depending on the process, the disease will appear sooner or later and with greater or lesser severity, according to the mutation, the influence of environmental factors and the contribution of other genetic components. There are numerous monogenic diseases, but they occur infrequently as isolated cases. This means that genetic testing focuses on families or groups having a higher risk of developing these diseases (e.g., familial breast or colon cancer, rather than sporadic cases).

In multifactorial or polygenic diseases, the influence of the genetic alterations on the pathology is smaller than in monogenic diseases. In these cases, certain individuals have a greater risk of developing a given pathology,

but probably only under specific conditions. Whereas a considerable amount of molecular and clinical information has been gathered for monogenic diseases, we still lack data on the repercussions of genetic variants (most still unidentified) on the genesis of specific multifactorial diseases. On the other hand, due to the high prevalence of common diseases, it would be expected that the carrier status for genetic susceptibility variants would also imply a certain advantage in other situations. Comprehensive epidemiological studies are needed to determine the role of the genetic variants involved in multifactorial and polygenic diseases. Thus, the current debate on genetic testing and on the potential access of insurance firms and businesses to the results of these tests should focus mainly on monogenic diseases. In the case of multifactorial pathologies, we have still not reached the point where we can use genetic testing for predictive purposes.

When we look at genetic testing within the context of monogenic diseases, good medical practice in countries where this speciality is fully developed indicates that genetic testing must be prescribed and reported by geneticists (i.e., genetics experts with experience in this field of human biology and medicine). The fact that this medical speciality does not exist in Spain and the lack of specific training in this field in the university medicine career puts us at a disadvantage regarding the development and application of genetic testing. Under institutionally normalised conditions, the individual prescribing a genetic test should be a geneticist and not a company or insurance firm physician directly requesting this type of screening from a laboratory. It should be remembered that behind the results of a genetic test, there is a family composed of individuals who may or may not develop a specific disease. Each case and each disease must be handled individually within the clinical context of the family and the specific situation.

The healthcare systems of many European countries do not seem to be able to meet all the needs of their populations. There is enormous discord between pharmacy costs and the cost of specific patient care. This means that certain services are not adequately covered. In Spain there are no guidelines concerning the application of genetic diagnosis. For the greater part of the ten-year history of the genome revolution, there has been no effort on the part of our governments (state, autonomous community or local) to develop sufficient resources for the diagnostic applications most needed by the population or to co-ordinate the efforts of professionals. In all cases, the

investigators themselves have developed diagnostic tests for some of the main genetic diseases and have provided continuity with financing from their own research projects. For many diseases there are no laboratories offering necessary testing, and there are no logistics of any kind for implementation of such testing in Spain. In fact, in recent years the diagnostic efforts initiated by several investigators have been cut short by the change of direction required in their research or professional careers. In this chaotic context, with no medical specialty, no basic teaching for physicians in medical genetics and no co-ordination in the molecular diagnosis of genetic diseases, the prospects look extremely bleak. If this is the case for monogenic diseases, the situation is even more disturbing for polygenic or multifactorial diseases. We are certain to see clinical analytical laboratories spring up in the next few years to offer screening tests aimed at determining an individual's risk of developing certain diseases. We will be witness to a scenario characterised by medical ignorance regarding the influence of genetic factors in the genesis of disease. Moreover, individuals will suffer discrimination on the basis of ill-founded genetic risk factors.

The genome sequence will allow scientific advances scarcely imaginable even a few years ago. Socially, economically and scientifically developed societies will be the first to reap the benefits of this scientific advance. Limited Spanish investment in science and its products, the anachronism of healthcare and university institutions and the lack of foresight on the part of our politicians indicate that we will suffer the consequences of scientific advances rather than enjoy their benefits.

The future of predictive medicine can be summarised in a list of keywords, whose significance we must be able to define: confidentiality, reproduction, treatment, information, prediction, uncertainty, presymptomatic, susceptibility, age of onset, multifactorial, external factors, reliability, mutation, variant, disease, genetic defect, heredity, family, life insurance, health insurance, employment, safety, accuracy, prognosis, medical genetics, specialist, genetic risk, risk of disease, prevention, research, family history, benefit, risk group, medical history, genetic test, degree of risk, consent and many more. Will we be able to define the terms and set the stage for the consequences of our understanding of the genome, at both the individual and collective levels?

PREDICTIVE MEDICINE AND DISCRIMINATION

Mirentxu Corcoy

1. Social and legal prospects of predictive medicine

1. 1. Introduction

With regard to scientific advances within the field of medicine and specifically to genetic advances, I feel that the start of this new century is witnessing a problem similar to the one raised by technological breakthroughs in the second half of the 20th century. Since the Enlightenment, the modern state has been prominent in fostering scientific and technological progress. From the onset of industrial development until well into the 20th century, no limits whatsoever were placed on advances. The initial controversies arose in response to the adverse consequences of continued growth in industrial production, with these consequences already appearing in the 19th century in countries where the industrial revolution took place. Among the various problems arising, the one most directly related to the problems resulting from genetic advances is the debate on the consequences for natural resources posed by technological growth. In the last quarter of a century, this discussion has acquired a new dimension, with the benefits of economic development being openly questioned and warnings being issued about the problems it causes. An analogous situation is now arising with respect to the scientific advances implied in genetic breakthroughs. Society has begun to consider what limitations should be placed in this area, based on the problems posed by certain applications of our new knowledge.

We cannot oppose these advances, even if only for pragmatic reasons. Regardless of the stance we take, science will continue advancing and mankind must face the new challenges it brings. As genetics is introduced in the treatment of disease, a complete turnaround will occur in traditional medicine. This is undeniably positive; however, the experience we have gathered in establishing limits on industrialisation and the application of nuclear energy must be used to advantage in this case as well, before it is too late (and we hope it is not already too late). Consequently, scientific advances must be accompanied by a social and legal analysis of the consequences thereof for society. Sounding the alarm about the problems and seeking legal and social solutions to counteract the adverse consequences of medical advances is the first limit –and perhaps the only one– that must be established in relation to the application of genetics to humans.

We must not forget that we do not know as much as we think. A clear example of this is illustrated in the problems posed by the introduction of genetic techniques in agriculture and stockbreeding. The danger to both the

environment and to human health produced by genetic engineering as applied to agricultural and stockbreeding development has become evident in recent years and has required government intervention to limit and control these techniques.¹ It is clear that extreme care must be taken when these techniques are applied directly to human beings, whether at the time of conception (embryo screening, genetic treatment of the foetus, etc.) or later (genetic analyses, gene therapies, etc.).

The solutions we are seeking to the conflict between the harm and the benefits of using genetic techniques in medicine (whether in analysis or treatment) must include listening carefully to the opinions of the scientific community, as this is the only way to predict the consequences of genetics with any measure of success. Even these opinions are constrained, however, by the limitations in scientific knowledge regarding the negative results of genetic research and by the tendency of researchers in this area to deny that any related risk exists or that the risk is warranted in all cases on the basis of the inherent goodness of scientific research. For this reason, and because the problems posed often have ethical connotations, the scientific community is not the only one with a right to an opinion. The very nature of the problem means that every facet of society is entitled to express its opinion in this regard.² There are numerous difficulties to achieving any degree of consensus, as the scientific community is overly influenced by its inherent eagerness to acquire knowledge. Scientists pursue their research, despite the risks they themselves predict, stating that unless they assume risks, science would not advance.³ The

¹ On 15 October 1999, *The Lancet*, a highly prestigious medical journal, published an article describing an experiment conducted by the researchers Arpad Pusztai and Stanley Ewen, on the possible harmful effects of genetically modified (GM) potatoes. The article was published against the advice of the journal's scientific advisors, including John Pickett, an expert in plant chemistry.

² This is addressed by the *Convention on Human Rights and Biomedicine* (the Oviedo Convention) of 4 April 1997, ratified by the Instrument of 23 July 1999, in Art. 28, urging governments to encourage public debate on the fundamental issues raised by biology and medicine, and the subsequent decision by legal experts after hearing the scientific reports from an ethical and legal perspective, concerning which genetic engineering applications are considered admissible by society.

³ In general, scientific work (even when only informative) on genetics and biotechnology is based solely on the benefits produced by these sciences. The discussion has even focused on who will decipher the human genome first and on which teams have advanced further in improving the genetics of plants, animals, etc. To cite just one example, the various articles published by the journal of the International Society of Bioethics in its December 1998 issue contain no dissenting note to contrast with the scientific consensus that research and the application of genetic and biotechnological advances cannot and should not be stopped and that advances should be achieved in as little time as possible.

principle of scientific freedom leads scientists and researchers to venture forward and modernise their efforts in human research without accepting the need for limits, in fact often exaggerating the benefits offered by certain techniques.⁴ Researchers may actually conceal the failures of their genetic treatments as much as possible. Recently the deaths of several individuals from the use of gene therapies have been reported in the US. Moreover, severe malformations appearing in genetically modified animals have been swept under the carpet, including a case in which salmon engineered by scientists to grow quickly, actually were born with gigantic heads and practically no body.

Although the need for limits is patent, we must mention that scientific production and creation constitutes a basic right laid down in the Spanish Constitution, Art. 20. 1.b), and this framework includes advances in biogenetics, a science that, in order to fulfil the essential objective of preventing and eliminating diseases on which it is founded, should be promoted by the public authorities. By virtue of Art. 44. 2. of the Spanish Constitution, public authorities must promote science, and scientific and technological research to the benefit of the general interest. Nevertheless, despite the constitutional support of scientific research, we cannot deny the need for limits. This raises a serious practical problem regarding the control or limitation of genetic engineering applications, however, and such control can be attained with some measure of effectiveness only if there is world-wide agreement. Otherwise, any such constraint becomes impossible. This is precisely the situation we are currently facing. In the United States there is broad consensus that the application of genetic engineering in humans and in plants and animals should be widely allowed (in fact, in the US a living cell has been patented), whereas Europe considers that the human genome belongs to the world as a whole and therefore, must be protected but not patented.⁵ Nevertheless, the American position has made people in Europe speak up. It has been predicted that if the current attitude prevails, Europeans will find themselves importing genetic

⁴ For example, at the International Symposium on human implantation held in Valencia in March 1999 Robert Edwards, the scientist who achieved the first test-tube baby in 1978, stated that cloning can be used to “improve the possibility that women who want to have children become pregnant”, without discussing the problems derived from this effort.

⁵ In the United States, experiments with human embryos are only limited by the fact that they cannot be performed with government financing, which poses a problem since all genetic research is being carried out by publicly traded laboratories that are out to make a quick profit.

advances from North America, creating a source of income for the US and an economic burden for the European Union.⁶ Once again, we affirm that society can only achieve some degree of effectiveness when there is legislative harmonisation at the global level.⁷ Along this line, the declarations of international organisations will only achieve importance when they are not binding for the various countries. The relative importance of these declarations is evident in the fact that although they generally⁸ lean toward the concept that the human genome is common human heritage, the European countries⁹ support this stance, whereas in the United States, proprietary positions predominate, as mentioned earlier.¹⁰

In the midst of this conflict, predictive medicine raises problems of an entirely different type, which can be summarised in two major blocks: one of a scientific nature and one of a sociological and legal nature. For some time, public officials have made it clear that regulations are needed. In particular, a decision handed down by the Council of Europe on 29 June 1990 states that, given the potential risks derived from the uncontrolled use of genetic analyses, “consequently, there are sound reasons to take appropriate measures to prevent unacceptable advances, particularly regarding predictive medicine”. Now we will go on to mention the diverse risks that could result from the uncontrolled use of predictive medicine.

⁶ The problems stem from the discussion on public or private financing of genetic research. Celera Genomics is fighting to prevent free scientific access to data on the human genome, as has been demanded by Bill Clinton and Tony Blair. Even when Celera and other private companies undertaking research on the human genome achieve this, direct experimentation with several therapeutically useful genes is being reserved only for strictly financial purposes. In other words, Celera Genomics and others have reserved the patent and marketing rights for certain genetic techniques and drugs derived from some human genes.

⁷ This problem, as expressly related to the possibility of patenting human material versus considering a common heritage of humanity, see Knoppers, B.F., Hirtle, F., Lormeau, S., *Bancos de materiales humanos derechos de propiedad intelectual y cuestiones relativas a la titularidad: nuevas tendencias en la literatura científica y posiciones en la normativa internacional (Part II)*, in *Rev. de Derecho y Genoma Humano*, no. 6, 1997, p. 67f.

⁸ In this regard, see the World Medical Association’s *Declaration on the Human Genome Project* in 1992 or the UNESCO’s *Proposed Declaration on the Human Genome and Human Rights*.

⁹ Both the Council of Europe and the various member-states (France, United Kingdom, Norway, Netherlands) reject the idea that any proprietary rights to the human body exist.

¹⁰ Limits are indeed set, particularly when the right to privacy is infringed or the person whose cells have been used has not given his or her consent; in general the idea that DNA is an asset is defended. The only point under discussion is who is the owner, an issue that is related to one’s right to privacy.

1.2. Problems of a scientific nature

From the scientific perspective, in both groups of cases the root of the problem resides in the lack of sufficient scientific knowledge on the subject. Predictive medicine, particularly the branch related to genetics, is still in an “infant stage”, since the current state of the art is only in an initial or incipient stage with respect to the predictions made.

The incipient state of gene therapy means that:

- a) The results of genetic tests are not entirely reliable. On the contrary, they can be erroneous or at the very least, uncertain or based on probabilities, a scenario that implies the possibility¹¹ of discriminatory situations or psychological effects with no real scientific basis. This is not only due to the incipient state of genetics, but also to the fact that external influences are at least as important (if not more) than the genetic makeup, producing what is known as a phenotype.¹² Apart from this, the search for more reliable results (at least for the time being) is conditioned by the need to genetically analyse as many individuals as possible. However, this comes into conflict with human dignity, particularly with the right to personal freedom and privacy.

Based on the current knowledge in the field of genetics, even with predictive analyses that provide relatively certain information, the actual development of a disease depends, in most cases, on both genetic factors and other multiple factors of a socio-cultural and environmental nature. Thus the development of a disease is only a probability, which becomes a fact on very few occasions. Only a limited number of genetic diseases are truly monogenic.¹³ We now know that most are multifactorial and

¹¹ One simple, explanatory example showing that we are dealing with uncertain, probability-based predictions is the following: a pair of alleles determining a characteristic may not be identical, in which case the dominant allele determines the manifestation of the characteristic, masking the recessive allele. Thus, an individual will be heterozygotic regarding this characteristic and in the worst case, will give rise to the fallacy that he or she has a disease or will develop disease because he or she is “at risk”.

¹² Cf. Marshall, K., *A brief history and science of genetic technology*, in *Advances in Genetic Information (A Guide for State Policy Makers)*, Lexington (The Council of State Governments), 1993, p. 2.

¹³ For example, haemophilia, cystic fibrosis, Huntington’s disease, some forms of Alzheimer’s disease, haemochromatosis, hypercholesterolemia, some types of cancer, etc. are currently defined as monogenic diseases. Nevertheless, the way in which these diseases develop (and the time at which they will do so) is not evident. Alzheimer’s disease, for instance, was first thought to be always monogenic, whereas now several types of this disease are known to exist.

result from one or more genes and from the interaction of these genes with environmental factors or socio-cultural factors over which the individual has little or no control, either because he or she is not aware of them or cannot avoid them. In these cases the genetic component just represents potential risk factor, however.¹⁴

In cases where there is some type of treatment or when measures that can impede or slow the development of the disease are known, in general, we still do not have precise, effective solutions. We can say that data on the conditions and advantages of pursuing a particular lifestyle –diet, exercise, etc. are practically random. Little is known about the type and measurement of the potentially positive effects of “contingent genes”; nor about whether or not the positive effects of these genes would be adversely affected by interventions to eliminate the influence of susceptibility.¹⁵

- b) The problem is aggravated when, with the current scientific knowledge, many of the predictable diseases have no effective treatment that can prevent or cure the disease. As a result, a person who knows he or she is likely to develop a specific disease but can do little or nothing about it obtains no benefits from predictive analysis. Rather, the effects of such predictions are harmful as they can lead to legitimate or illegitimate discrimination of the person in the workplace or when obtaining insurance, mortgages, etc. In fact (as we will see shortly), the situation can and does lead to embryo screening and eugenic abortion, making eugenics the only therapy. The prediction of a disease without any means to treat it can have psychological effects for the person and his or her family.¹⁶ An awareness of one’s predisposition to a genetic disease

¹⁴ Cf. Husbands, R., *Employment testing: An international comparison*, in *ILO: Workers Privacy. Part III: Testing in the Workplace*, Geneva 1993, p. 55, which points out that the genetic tests designed to detect polygenic disorders (interaction of multiple genes) are less predictive than direct examination of the specific, still undeveloped, diseases governed by a single gene and, therefore, are a poor element for detection of a disease and are even poorer predictors of development of a disease.

¹⁵ Cf. Roscam Abbing, H.D.C., *La información genética y los derechos de terceros. ¿Cómo encontrar el adecuado equilibrio*, in *Rev. de Derecho y Genoma Humano*, no. 2, 1995, p. 36-37.

¹⁶ Genetic analyses have the peculiarity that they reveal other real or potentially affected members within the family of the person analysed, transferring the psychological and social problems from one generation to the next. One paradigmatic example is Huntington’s disease, a fatal disorder that can be predicted but for which there is no treatment. The Canadian Collaborative Study of Predictive Testing has reported on the psychological consequences of the tests used to predict this disease, since tests on other family members are needed to obtain results having informative value.

can produce a psychological burden with severe psychosomatic consequences. Moreover, it can affect a person's self-image and self-esteem, and the way one plans and orients the future.¹⁷

Consequently, it is necessary to consider whether or not predictive analyses are pertinent in cases where the detection of a predictable, probable or certain disease is not coupled with a solution to the health problem. In any case, the costs and benefits of predictive analyses and the resulting consequences for the patient and society should be weighed. The lack of treatment options must be a decisive factor in the development of regulations on genetic testing.¹⁸

1.3. Problems of a sociological and legal nature

From the sociological and legal perspective, there are essentially two problems:

- a) Awareness of a potential health problem can produce severe anguish, psychological or psychosomatic diseases, and basic life changes (e.g., not having children, losing one's mate, being estranged by friends, etc.), particularly in cases where awareness of the potential disease is not accompanied by any solution to the problem. For example, we know what Alzheimer's disease is and the gene that causes it. However, we do not exactly how and why it develops, except in an occasional specific type, nor do we have a satisfactory treatment. Moreover, there is no certainty that the disease will develop even with the presence of causative factors.
- b) The awareness of predictable disease can lead to severe discrimination both from a labour-related and financial perspective (e.g., in relation to loans, mortgages, insurance premiums) and from an emotional and social point of view. Thus, whereas biologists and doctors must continue their research, the legal field must develop mechanisms that impede or reduce the adverse and discriminatory effects of predictive medicine.

¹⁷ On the issue of genetic tests conducted within the context of labour relations, see Wiese, G., *Implicaciones del conocimiento genético en las relaciones laborales*, in *El Derecho ante el Proyecto de Genoma Humano*, Fundación BBV, Universidad de Deusto, Vol. I, 1994, pp. 263-264.

¹⁸ Cf. Tabarrok, A., *Genetic testing: An economic and contractarian analysis*, in the *Journal of Health Economics*, vol. 13, 1994, p. 80.

1.4. Predictive medicine and respect for fundamental rights

In view of the relevant conflicts of interest (public and private, ethical and economic), we must consider how to weigh these conflicting interests against a deep respect for fundamental rights. The Convention on Human Rights and Biomedicine sustains that the interests and welfare of the human being shall prevail over the sole interest of society or science. In the same vein, a systematic analysis of the Spanish Constitution of 1978 clearly shows that fundamental individual rights such as life, liberty and privacy, prevail over other rights such as public health and research, which are considered to be principles governing the conduct of public officials.

2. Scope and limits of predictive medicine based on respect for the right to freedom –principle of autonomy–

2.1. Informed consent

As with any medical care or medical-surgical treatment, predictive medicine must adapt to the requirements set forth in Spain's General Healthcare Act, in particular, to the requirement of informed consent.¹⁹ There is broad consensus, both legal²⁰ and deontological,²¹ on the need for the patient's informed consent as an indispensable prerequisite for undertaking any medical treatment. Informed consent is not only required to perform surgery but also to proceed with any treatment, including analyses. The crucial importance given to consent is grounded in the subject's freedom of self-determination and personal autonomy and is considered a fundamental right in all modern constitutions. Consequently, health professionals must not decide by virtue of any "rules governing the medical profession" or any

¹⁹ Art. 10. 5 of Spain's General Healthcare Act states that consent is compulsory. The patient has the right to receive complete, continued, verbal and written information on everything related to his or her condition, including diagnosis, alternate treatment(s) and their risks, and prognoses, with such information provided in understandable language. In the event that the patient cannot or does not want to receive this information, the information must be provided to legally responsible family members or individuals.

²⁰ For instance, the new law on the right to health-related information, patient autonomy and clinical documentation of 21 December 2000, passed by the Catalan Parliament, also requires the informed consent presented in Sections 2 and 3, with Art. 22 requiring consent for "all healthcare measures" being particularly relevant.

²¹ All recently approved medical codes of ethics contain a reference to informed consent.

supposed “superior judgment”. The patient is entitled to decide on the risks he or she is willing to run in pursuit of health.²²

At present, once consent is assumed as a legitimate requirement for any medical intervention, an essential component for validity thereof is stressed: information. Engisch affirmed that medical intervention is only legitimate if consent is given; in the case that an operation is to be performed, consent requires that the meaning of the information be clear in order to be effective, and the patient must give his or her consent. As part of these ideas, the obligation to inform appears only as a hypothetical “obligation” –in the Kantian sense– or precisely as a “burden” that the physician must assume if he or she wants to undertake the intervention.

When it involves a diagnosis, predictive medicine also requires informed consent, with the information in this case comprising not only the nature of the analyses to be performed, but also the intended objectives, consequences and potential treatments if a positive result is obtained.²³ This aspect is particularly important in cases where the decision to undergo certain tests can be conditioned by the fact that there are various treatment options if the results are positive. As mentioned earlier when describing the negative aspects of predictive medicine, a key issue for setting regulatory limits for the performance of genetic analyses is whether or not there is any treatment in the event that the analytical results point to the probability that an individual will develop a certain disease. The information must be simple and appropriate for the patient’s cultural and intellectual level.²⁴

Even when General Healthcare Act does not require consent because the patient is a minor or incompetent, this fact does not preclude the possibility that the staff provide information and listen to minors or incompetent

²² The Austrian penal code penalises unauthorised treatments in § 110 as a crime against freedom, protecting the patient’s right to free self-determination. In Spain the fact that there is no specific law punishing medical intervention without consent does not preclude the possibility that such conduct is punishable as an offence of coercion.

²³ In Art. 2.2., the Catalan Parliament Law of 21 December 2000 introduces a useful interpretative criterion for understanding what the information should contain, stating that “information... must be given in an understandable manner that is adequate to the patients’ needs and requirements in order to assist them in autonomous decision-making”.

²⁴ Cf. Jorge Barreiro, *Derecho a la información y el consentimiento informado*, in “La declaración de los usuarios de los servicios sanitarios”, *IV Congreso de Derecho y Salud*, Ed. Gobierno Vasco, Vitoria 1996, p. 151.

individuals to the extent possible, that is, at their level of understanding and discernment. To some degree, these patients can provide consent personally and not only through those having legal authority or guardianship. The condition of minor or incompetent is grounded on one's capacity to manage oneself and one's assets, hence in each case the person's capacity to decide on a specific medical intervention must be considered.²⁵

As to the predictive analyses and gene therapies carried out on embryos or foetuses, informed consent must obviously be obtained from the donors of the embryos and, if applicable, from the person or persons who will subsequently assume charge of the future child. In any regard, in these cases the staff must be guided by the principle of the "greater welfare" of the future being.

2.2. Right to informed self-determination. Right "not to know"

The principle of autonomy determines that consent also includes control over the data that are obtained. The control of any data, apart from the exceptions laid down by law, pertains to the owner.²⁶ The individual's right to decide about the use of his or her medical data, particularly genetic data, implies the right to gain access to the data, to control their existence and accuracy and to authorise disclosure.²⁷

An essential consequence of informed self-determination is the right "not to know". If the owner of genetic data is entitled not to inform third parties of the information concerning him or her, he or she is obviously entitled to remain ignorant of the data, since this right is simply an exercise of the owner's right to freedom. As we mentioned at the start, awareness of the probability of a future disease can affect a person's personality by causing psychological alterations or by changing his or her present and future

²⁵ Cf. Jorge Barreiro, *La relevancia jurídico-penal del consentimiento del paciente*, CPC (16) 1982, p. 23, before the Protection Act for Minors came into effect, based on a certain capacity of autonomy of minors in Art. 162.1 of the Civil Code after the May 1981 reform, which excluded from the legal representation granted to parents over emancipated children "acts related to personality or other rights that the child can perform for him or herself in accordance with the law and the level of maturity".

²⁶ Lucas Murillo, *El derecho de autodeterminación informativa*, Madrid 1990, pp. 15ff.

²⁷ Art. 13 of the Catalan Parliament Law of 21 December 2000 is particularly significant and innovative in this regard, as it stipulates that the patient is entitled to access his or her clinical history. Moreover, by virtue of Art. 14, he or she is entitled to active, diligent custody of clinical histories by the healthcare centres.

behaviour patterns. Thus, it is obvious that a person is entitled not to know, precisely in pursuit of safeguarding his or her health, understood globally as physical and psychological welfare.²⁸ In principle, it is evident that an individual who goes to a specialist for genetic analyses wants to know about his or her condition. The “right not to know” becomes particularly important in the case of genetic analyses imposed on a person for reasons related to the public interest.

2.3. Limits to informed consent requirements

In a social and democratic state, no right enjoys absolute protection, not even the right to life. Consequently, the requirements derived from the protection of fundamental rights of third parties represent a limit on the principle of autonomy at all times. If we consider that the guiding principle in the resolution of the conflict of interest raised by predictive medicine is respect for fundamental rights, then the limits on the requirement for consent (and if applicable, confidentiality) must be founded on the concurrence of the right to freedom pertaining to an individual who refuses to submit to certain analyses versus other rights pertaining to individuals who could be concerned by this refusal. In order for the right to freedom to wane, in principle the opposing right must involve a threat to the life or health of others. Regardless of existing legislation, therefore, the interpretation of this legislation should be undertaken with this principle in mind, limiting the scope of legislative restrictions to the requirement for informed consent.²⁹

Public health should not be considered an absolute that allows limitation of the right to freedom. Instead, it should be regarded as the health of individuals as a whole. Infringement of the inviolability of fundamental rights on the basis of a duty of general solidarity must be exceptional and clearly warranted.³⁰

²⁸ Corcoy Bidasolo, F., *Libertad de terapia versus consentimiento*, in *Bioética, Derecho y Sociedad* (coord. M. Casado), Madrid 1998, p. 112f.

²⁹ Cf. Romeo Casabona, *El médico y el derecho penal*, 1981, p. 289, which states that in medical measures which are not intended to cure (e.g., some cases of predictive medicine), the consent will permit intervention only when it actually takes effect. “*El médico...*”, p. 366f., understands that the physician can act justifiably in these cases due to the need.

³⁰ Hence, the Organic Law on Special Measures relating to Public Health, of 14 April 1986, designed to regulate compulsory treatments related to the existence of danger for the population’s health should be applied only on rare occasions.

A different problem altogether arises when genetic analyses are performed in order to save the patient's life.³¹ The problem in these cases originates in the discussion about the person's right to dispose of his life at will. Those who defend that a person has not the right to dispose of his life do so from religious perspectives or on the basis of assuming a social component in the legal value of life itself. If our initial premise is that a person can dispose of his life at will, the situation is very clear inasmuch as the refusal to perform genetic therapy would be simply another case of rejecting legitimate treatment. However, based on the opposite premise, intervention by medical staff without consent would be illicit even when its purpose were to save the patient's life.

Another problematic issue that, to some extent at least, also represents a limitation on informed consent, or rather the information, affects the scope of this information. Specifically, the question lies in knowing whether or not inconclusive information should be given, that is, information which, by its nature, would not enable the patient to make independent decisions because the actual meaning and scope of the information is unknown. This doubt becomes particularly significant in the case of genetic analyses since, as we mentioned earlier, all or many of the factors that might determine whether a predisposed individual actually develops a disease are unknown.

3. Scope and limits of predictive medicine based on respect of the right to privacy

3.1. Right to privacy. Confidentiality and legal protection of personal data

Privacy as a fundamental right of citizens is laid down in Art. 20. 1.d) of the Spanish Constitution and is further developed in two basic organic laws, Organic Law 1/1982 relating to the protection of privacy and self-image and the Organic Law (LOPD) and Royal Decree 994/1999 (both modified by Organic Law 15/1999 relating to the protection of data of a personal nature.³²

³¹ Cf. Romeo Casabona, *El médico...*, p. 366f., understands that the physician may act justifiably in these cases due to need, even if consent has not been given. Jorge Barreiro, *CPC* (16) 1982, p. 21f., is of the same opinion, even consent could be presumed to exist in these cases.

³² These laws modify the LORTAD (Organic Law relating to the regulation of automated data processing), expanding the protection to any kind of personal data and not just computer data, thereby extending protection to samples of DNA or any kind of biological material, provided they contain identifiable personal data. The regulations establish the safety measures for automated files containing data of a personal nature.

Penal protection, even as it existed in the limited, anarchic way described in the 1973 Penal Code, becomes more important and systematised in the 1995 Penal Code which includes a new title designated “Crimes against privacy, the right to self-image and the inviolability of the home”. These provisions contain stricter penalties for the disclosure, revelation or dissemination of data of a personal nature that reveal aspects related to health, racial origin or sexual life, increasing the sentence when computer data are involved.³³

The concept of “privacy” has countless meanings and connotations. In order to further define the concept of privacy, specifically “genetic privacy”, we must mention four preliminary presuppositions:

- a) In general, the consent of the individual affected by the disclosed data precludes the violation of privacy. Nevertheless, as we will see below 3.2., the special nature of “genetic information” means that not even the person to whom the data pertains has free access to the information. From a legal standpoint, the data pertaining to the person may be kept secret if he or she wishes to safeguard them from public knowledge.³⁴ In terms of what information is secret, and for which data disclosure would be illicit, this is understood to be any circumstance that the owner considers relevant to protect his or her privacy.³⁵ A ruling handed down by the Supreme Court on 21 May 1993 defines secret information from a legal perspective as follows: “the knowledge of certain data about a specific object by a limited number of individuals and which, for various reasons, should not be expanded beyond this circle, with the desire of the owner of the information considered as relevant”. In relation to the question being raised here, this definition of secrecy highlights the subjective aspect of the owner’s preference to reserve knowledge of the data, which are objectively “secret” since they are known to a limited number of individuals. Consequently, in the case of

³³ Conduct that violates privacy is even more serious in the case of computer data because of the extent of the damage. This aspect is particularly important in the case of genetic analyses because, as we will see, the information is obtained by using the computer to cross-reference the genetic data of a single person obtained with various means, as well as the data of various individuals.

³⁴ Cf. Queralt Jiménez, *Criminal Law. Special Part*, 3rd ed. Barcelona 1996, p. 193, “existence of reserved data or facts of a person which, due to their inherent nature, do not form a part of what is usually known by the general public”.

³⁵ Cf. Lenckner, in Schönke/Schröder, 25th ed. § 203/5, understand that secrecy, for criminal purposes, may even be an opinion that is defended.

genetic analyses, the fact that certain personal data are known by a limited number of staff does not preclude it from being classified as secret. This explains why, regarding criminal protection of privacy in the case of professional secrecy, only the disclosure of confidential data is penalised, inasmuch as these data have been disclosed to other staff members involved in the treatment. In the specific case of the analyses, it is evident that they are carried out so that they can be used by the specialist who requested them, hence communication between the analyst and the specialist would never be legally disapproved conduct.

The problem lies in determining where to draw the line regarding who has access to these data. I feel that the general criteria must be minimal, both with respect to the circle of individuals and to the data to which each individual has access. In determining who has access and what data should be disclosed, the guiding principle must be the need to attain the objectives set by the patient when deciding to have the analyses performed.

- b) Privacy can be violated even when this guideline is followed. As we have stated, the accuracy of the information does not preclude that personal privacy or honour has been injured. This principle is particularly important in the case of predictive medicine, since the data obtained from genetic analyses are usually “accurate”, at least holding to the scientific knowledge existing at the time, and such accuracy would never warrant disclosure. Disclosure, as mentioned, is only warranted when the knowledge is needed to prevent risk to individual or collective health, and even in these cases, only when the content of the information and the individuals with a “need to know” are limited.
- c) In the measure that they refer to a person’s health and to that of his or her family, genetic data are part of what is considered to be sensitive information and, consequently, are subject to special protection³⁶ in civil and criminal regulations. “The essential core of privacy”³⁷ is the name given to data related to health, ideology, religious beliefs, racial

³⁶ In this regard, Directive 95/46/EC of the European Parliament and of the Council of 24 October 1995 on the protection of individuals with regard to the processing of personal data and on the free movement of such data includes special protective measures with respect to sensitive personal data.

³⁷ Anglo-Saxon doctrine uses this designation to refer to protection of the most sensitive aspects of privacy.

origins and sexual life.³⁸ Genetic information would be included among these particularly sensitive data, as it can provide information on the individual's health, racial or ethnic origin and sexual habits.

- d) Further protection of genetic information above and beyond that provided for sensitive personal data is still under debate.³⁹ The discussion focuses on whether or not a “genetic record” implies any truly new problems. Fears about “transparency”, a concept that could arise from full availability of genetic data would be much more justified if, as appears to be confirmed from a scientific perspective, this “genetic record” included psychological as well as biological data.⁴⁰

The performance of predictive analyses without consent is an intrusion in private life and, as mentioned earlier, could be considered coercion. The subsequent revelation or disclosure of data obtained from these analyses is an attack on privacy which, depending on the entity, could be considered a civil illegality or a crime consisting in the disclosure of secret information. The revelation of certain data affecting the essential core of the person, such as the suffering of certain diseases, could even be qualified as slander (whether civil or criminal in nature) and would affect the individual's personal honour.⁴¹

In the cases in which the data pertain to a minor or an incompetent person, the obligation to maintain confidentiality is not only present but is also accentuated. The law particularly protects any data affecting a minor or

³⁸ Art. 7 of the LORTAD, Organic Law 5/1992, of 19 October, on the automated processing of data of a personal nature (Official Spanish Gazette 147, of 21 June) considers the aforesaid data to be “specially protected”. Nevertheless, this LORTAD regulation was contradictory to the provisions of the Council of Europe Convention of 28 January 1981 for the protection of automatic data, ratified by the Instrument of 27 January 1984 (Official Spanish Gazette 274, of 15 November 1985), since Art. 6 establishes the general principle that these data are not computerisable unless they are subjected to a splitting process that prevents the data owner from being identified. As a result, Organic Law 15/1999 on the protection of personal data is intended to solve this problem by adapting the Spanish legislation to the European regulations.

³⁹ European Directive 95/46, which is mentioned in footnote #36, states that inasmuch as genome analyses are limited to the confirmation or exclusion of specific pathological conditions, current measures for the protection of medical data and secrecy would be sufficient.

⁴⁰ In European Directive 95/46, the sufficiency of current protection measures is based on the supposition that the “genetic record” does not include psychological data which, according to the directive, implies a greater risk of abuse than biological data.

⁴¹ A Spanish Supreme Court ruling has dictated that a journalist's disclosure that a particular person had AIDS was slander.

incompetent person, precisely because these needy individuals require greater protection. Legally, the protection of secrecy with respect to minors is evident in the removal of parents' and guardians' rights to open a minor's correspondence by the 1995 Criminal Code, in which protection of the secrecy of communications was not limited in any way.⁴² Likewise, Article 4.1. of Spanish Organic Law 1/1996 on the protection of minors also protects the secrecy of minors' correspondence and communications. Section 5 specifically states that parents are obliged to respect these rights and to require respect by third parties. Such protection of secrecy in the case of minors does not preclude the possibility that there may arise situations where secrecy is justifiably breached, provided that all applicable presuppositions are met. Specifically in relation to minors, this justification may derive from the parents and guardians' right to behavioural correction.⁴³

In relation to genetic data, this possibility does not preclude the parent's or guardian's responsibility to respect the privacy of the minor or incompetent person as much as possible. Thus, the parents' or guardians' knowledge of the information should be limited to that which will contribute to the "greater welfare of the minor or incompetent person". A basic assumption should be that the disclosure of a minor's genetic data is only warranted when the aim is to avoid risks to the minor's health,⁴⁴ or when nondisclosure of these data implies certain danger for third parties.

The reason why the disclosure and revelation of secrets is a more serious crime when the disclosed data pertain to a minor is based on the greater

⁴² O. Queralt Jiménez, J.J., DP. PE., 3rd ed., p. 197, understands although the Penal Code does not expressly mention the possibility that parents or guardians in exercising their right to raise their children can disclose their secrets, it is understood that when this is done, it must be done with full respect of the minor's personality and will be allowed with the sole purpose of training the minor. According to majority opinion, Lenckner in Schönke/Schröder, 25th ed. § 202/13 states that the secrecy of a person's mail is no longer protected when there is an expressly regulated right that allows the mail to be opened and, in relation to minors, §§ 1626 and 1631 BGB grant parents and guardians the right to supervise the mail of their children or wards.

⁴³ In regard to the disclosure of other types of data relating to training and education that affect the minor's privacy, Morales Prats in *Comentarios a la Parte Especial...*, p. 303, believes that, in the case of minors, the reason justifying compliance with an obligation or exercise of a right of Art. 20. 7° of the Spanish Penal Code applies in the case of legitimate exercise of rights in childrearing enjoyed by virtue of custody or guardianship of the minor.

⁴⁴ The disappearance of the specific rationale, along with the regulation set forth in Organic Law 1/1996, means that this is the only fitting interpretation for the spirit of these legal reforms, inasmuch as the purpose of these new regulations would be not achieved when the rationale is understood generally.

vulnerability of these individuals.⁴⁵ The potential harm is increased, since secrets about minors can affect their future life (i.e., free development of their personality). The implications are even more serious in the case of genetic data, the knowledge of which might modify their character and condition their decisions.

3.2. “Genetic privacy”

The specific nature of genetic information,⁴⁶ which has been designated “polymorphous”, has determined a precise aspect of privacy known as “genetic privacy”, establishing the basis for what is now called “ethics of genetic information”. The concept of “polymorphous” nature derives from the existence of three levels of information. The first level is known as “genetic identity” and comprises the genetic makeup of each individual person; the second level is the capacity to infer the genotypic expression of a person from his or her family, as a normally recessive hereditary condition. The third level is aimed at recognising a social sphere in human genetics. The third level is precisely where protective mechanisms designed to limit stigmatisation or discrimination mainly act, or should act. The enormous information potential (and consequently the potential to violate privacy) of genetic data should put the individual’s genetic code not only on the same as level as the individual’s clinical history⁴⁷ but afford it even greater protection than that provided for sensitive data affecting the “essential core” of privacy.⁴⁸

Greater protection is warranted because of the special nature of genetic information, in which infringement of the right to “genetic privacy” not only harms the right to privacy of the data owner but also that of third parties. Although the data owner can freely access other data affecting his or her privacy, the same cannot be said of genetic information.

⁴⁵ The majority opinion is expressed by Lozano Miralles, *Compendio de Derecho Penal. Parte Especial*. Volume II (Bajo Fernández, director) Madrid 1998, p. 222.

⁴⁶ Cf. Moreno, R.F., *Información genética (informgenics) e intimidad*, in *El derecho ante el proyecto genoma humano*, T. I, Fundación BBV-Universidad de Deusto, Madrid 1994, pp. 391-392, defines it as the “science of collecting, handling, sorting, storing and retrieving recorded genetic information”.

⁴⁷ Cf. Martínez Bulle Goyre, *Genética humana y derecho a la vida privada*, in *Genética humana y derecho a la intimidad*, Mexico 1995, pp. 34-35.

⁴⁸ See above 3.1. on the concept and protection of “sensitive data” and the “essential core”.

“Genetic privacy” not only requires special protection against the obtaining or dissemination of data without the owner’s permission. Limits must be established regarding the interference of legitimate public or private authorities, even when they are supposedly acting on behalf of the person or society, because only the individual has the right to decide what he or she considers beneficial for him or herself.

The situation is not, however, so simple. As stated earlier, an individual right cannot be absolutely recognised since conflicts between individual interests and the interests of society can and do arise. For this reason, a distinction is made between “strictly private genetic data” and “genetic data available for public access”.⁴⁹

3.3. Exceptions to the protection of privacy

An immediate consequence of the concept of “genetic data available to the public” is the acceptance that there are limits on the right to “genetic privacy” related to these data. Nevertheless, even in the extraordinary cases where certain data must be reported by legal imperative or for a justifiable reason, disclosure of this information must be limited by determining exactly what data is actually needed. The general rule is that these data are the exclusive property of their owner, although there can be exceptions grounded in public interest. However, public interest cannot be considered a general interest used to rationalise any interference in the privacy of individuals. In contrast, being an exception, the existence of an interest challenging the respect of privacy must be proven in each case. In order to prevail, it must consist in a certain danger to individual or collective health or to public safety.⁵⁰ The proportionality principle that should be used to weigh conflicting interests implies that a fundamental right (in this case, privacy) may not be harmed when there are less damaging ways to circumvent these risks. From a legal-penal perspective, these exceptions to protection of privacy are justifiable causes and specifically imply a situation of justifiable need.⁵¹

⁴⁹ Zimmerli, W., *Who has the right to know the genetic constitution of a particular person?*, in *Human Genetic Information: Science, Law and Ethics*, Chichester 1990, p. 93f.

⁵⁰ Cf. Casado González, *El conflicto entre bienes jurídicos en el campo de la genética clínica: exigencias de salud pública y salvaguardia de la dignidad humana*, *Rev. de Derecho y Genoma Humano*, no. 4, 1996, p. 31, in relation to the difficulty of reconciling the protection of individual rights with political interests relating to public health.

⁵¹ In relation to the state’s requirement of justifiable need, see Mir Puig, *Derecho Penal. Parte General*, 4th ed. Barcelona 1966, p. 453f., stressing the importance of using an “extremely strict version” of the proportionality principle.

Nevertheless, a rationale justifying interference in a person's privacy in certain cases does not mean that there are no limits to this interference. In this regard, Arts. 4ff. of the LOPDCP (Organic Law relating to the protection of data of a personal nature) serve as a guideline when they state that only the obtainment of appropriate, pertinent and non-excessive data within the legitimate scope and purpose for which they have been collected is admissible. These limitations on the type and amount of data that can be gathered in the public interest is particularly relevant in light of the enormous capacity of modern computer systems to interpret results. Computer power now allows scientists to sift through data on genetic differences and statistically associate them to health and disease.⁵² In order to construct the so-called "genetic citizen", it is necessary to have abundant data that can come from various tests (paternity cases, legal identification by DNA, prenatal analyses, medical examinations for employment contracts, analyses for obtaining loans or mortgages, analyses for obtaining insurance, etc.). The sources are numerous, but there are no accompanying mechanisms to prevent errors, ensuring the accuracy of the results. Predictive analyses function with statistical criteria and their accuracy depends largely on scientists' ability to cross a considerable amount of data from many individuals. This leads to the idea of an obligation to collaborate with respect to genetic data, since results with some degree of accuracy are only possible by looking at extended layers of the population. Nevertheless, extreme precaution is needed with this activity to prevent abuse by public officials or specific private sectors.

4. Positive and negative consequences of predictive medicine

4.1. Adverse consequences of predictive medicine

4.1.1. Biodiversity versus "perfection"

Along with the fears of psychological, job-related and financial problems arising from predictive analyses we mentioned earlier, one extremely important aspect related to these problems is the discrimination resulting from a disease, or rather from the probability of developing a disease.

The problem is accentuated when we do not have a positive response to a particular disease, in which case one solution would be elimination of the sick

⁵² The geometric increase in the danger of using computerised personal data has been considered by the legislation that protects these particular data and that provides special control measures, which in Spain pertain to the Data Protection Agency.

person. Though these fears may seem groundless, they are not so; as an example, for many psychological and neurological degenerative diseases, practically the only possible action is negative eugenics. In the case of predictive medicine in embryos (e.g., embryo screening) the problem is similar because, although many physiological factors can be controlled and analysed, we know nothing about the factors making up the personality. In this sense, ignorance is total. It is true that mankind has advanced physically and even culturally, but in the area of instincts, i.e., aspects beyond the cumulative cultural control, 21st century man is equal to prehistoric man.

It is difficult to predict the consequences of achieving a society of “perfect” individuals from a physical standpoint, but with all the negative aspects of character that we are already familiar with. This is irrespective of the concept of “perfection”, as this idea is subjective. The various models of perfection that have existed throughout history have been founded on diversity, namely on the contrast between what is perfect and what is imperfect. If diversity disappears, it will be difficult to conceive of perfection. We cannot overlook the importance of biodiversity (now accepted as an indisputable principle in the balance of natural systems) within the human race.

From the perspective of equality and in relation to the generalised application of gene therapies (positive and negative) in the developed world, predictive medicine could lead to an even greater imbalance than the huge distance now existing between developed and developing countries. Biotechnology will allow those with more financial resources to improve by combining biology, reproduction and genetics. Genetic improvements will be passed from one generation to the next, presenting an ethical dilemma in the following terms: only the well-to-do classes will have better offspring, further widening the enormous gap between rich and poor.⁵³

4.1.2. Genetic diseases, eugenics and discrimination

The rise in support for eugenics –even when it is positive, as in the case of “embryo screening”, with the idea of selection of species as an individual but common will of society– is contradictory to the will (also generalised) of the state, which promotes the prevention of discrimination on the basis of racial

⁵³ Lee F. Silver, *Vuelta al Edén*, Barcelona 1996.

differentiation, with the concept of racism including both cultural and biological racism. If genetic engineering can accentuate the inequalities between countries and citizens, it can also, and to a much greater degree, be a source of discrimination by differentiation –infringement of the principle of equality– for particularly horrible or repulsive reasons.⁵⁴ These reasons are considered horrible and repulsive when, in addition to the principle of equality, they harm human dignity. Discrimination derived from knowledge of genetic data becomes particularly important because the study of the human genome will allow us, in the not too distant future, to discover personality traits related to intellectual and manual aptitudes, temperament, etc.⁵⁵ As a result, the creation of “genetic records” would be detrimental to human dignity because of the social consequences of the “eugenic discrimination” that can result from complete genome analyses.⁵⁶

Non-discrimination is the object of many human rights declarations⁵⁷ and, specifically, of our Constitution (particularly, the Penal Code). The struggle against genocide and any other form of discrimination on the basis of race, creed, sex or disease can be viewed as a desire to defend differentiation and diversity. These regulations, which spare no punishment for these action, also imply a desire to prevent any conduct of this nature. They prohibit conduct which, if no action were taken, would be accepted as the response of a eugenic awareness which, in my opinion, is widespread in current society. Arts. 510 to 512 of the Penal Code prohibit discriminatory conduct and include activity of special interest in this regard set forth in Art. 511.1. and 3. This article punishes civil servants, particularly those responsible for a public service, who deny on the grounds of “disease or disability” any benefits to which an individual may be entitled. Among the crimes against workers’ rights, Art. 314 punishes discrimination in public or private employment because of “disease or disability”. In both cases, discrimination on the basis of disease would be

⁵⁴ Cf. Rodríguez Piñero, Fernández López, *Igualdad y discriminación*, Ed. Tecnos, Madrid 1986, p. 81.

⁵⁵ De Sola, C., *Privacidad y datos genéticos. Situaciones en conflicto* (I), *Rev. de Derecho y Genoma Humano*, no. 1, 1994, p. 179.

⁵⁶ Eser, A., *¿Genética, “Gen-Ética, Derecho Genético? Reflexiones político-jurídicas sobre la actuación en la herencia humana*, *Rev. Jurídica La Ley*, Vol. I, 1992, p. 1145, understands that this possibility of discrimination involves not ruling out the need for a specific regulation that safeguards against possible abuse.

⁵⁷ For example, see Art. 11 of the *Convention on Human Rights and Biomedicine*, ratified on 23 July 1999.

placed on the same level as discrimination on the basis of creed, religion, sex, race, etc. However, the application of these measures is practically impossible according to the interpretation suggested. In order for behaviour characterised by denial of service, employment, etc. to fall under this Article, a necessary condition is that it be done for discriminatory reasons and not simply because the individual in question is more or less apt for the job or service.

It is possible that discrimination derived from predictive medicine will be included under “traditional” racial discrimination, marginalising “unhealthy” persons as third-class citizens since they represent a social burden that the State is not willing to take on. It is starting to seem almost “normal” that surgical techniques are chosen and used (or not used at all) on the basis of an individual’s productive capacity, and that this activity is grounded on financial criteria that ignore the dignity of every human being regardless of race, health or age, which only a few years ago was considered to be paramount. This principle of human dignity is stated in all charters of fundamental rights but could become a mere declaration without content, because of the numerous State regulations that adhere exclusively to utilitarian criteria without attending to this right to dignity, which encompasses all other basic rights.

The apparently complete legal consensus on banning discriminatory conduct resulting from present or future disease is, in practice, not so complete. In reality, as mentioned earlier, this conduct is only understood to be prohibited when the reason that a service or job was denied is solely discriminatory and not because of the risks posed to the individual or to third parties by the service or job. The question would be: is the consideration of a person’s vulnerability to a certain disease discriminatory behaviour?⁵⁸ It is clear that new criteria must be introduced, permitting establishment of whether a decision (regardless of the alleged reason) is or is not based on individual or collective risk. This assessment must be performed from an objective perspective, determining whether a risk actually exists in the specific case. The idea of risk should include situations where individual health and/or the health of third parties is jeopardised, including in this concept a person’s aptitude to perform a specific job. If the assessment shows that these risks do not exist or that they are distant or uncertain, the refusal must be considered as discriminatory, without going against the principle of presumed innocence.

⁵⁸ Cf. Michaud, J., *Las nuevas leyes francesas sobre bioética*, RDGH, no. 3, 1995, p. 297f.

4.2. Positive results of predictive medicine

4.2.1. Health protection and predictive medicine

Much that has been described up to this point might be seen as an apocalyptic vision. This is because the intrinsic “goodness” of predictive medicine has not been called into question and, as mentioned earlier, will continue advancing. Predictive medicine (particularly genetic analyses) will undoubtedly lead to significant advances in medical research.⁵⁹ The positive, extremely beneficial nature of certain activities, such as “à la carte” treatment based on the individual’s genetic map, the possibility of treating a disease before the symptoms appear and the possibility of taking measures to prevent the development of the disease, is something that does not allow discussion from either an ethical standpoint or from utilitarian postures. Both economics and ethics agree on this point.

The advantages of predictive medicine, specifically genetic analyses versus traditional medicine, are based on the distinct nature of the information obtained in each case. Genetic information is understood to be “the set of messages contained in the molecular structures that carry hereditary information –nucleotide sequences of the DNA and RNA– comprising all the structures of the individual and its functioning”.⁶⁰ A genetic test is any technique used to determine the presence of inherited genetic alterations or genetic changes caused by personal genetic characteristics combined with the predisposition to develop certain diseases. An individual’s predisposition to a disease is not based on causality but rather statistics, since certain gene sequences are usually found in people who develop a specific disease, allowing this type of sequence to be used to identify risk groups.

We will group the various types of genetic analyses on the basis of this working objective, i.e., on the basis of the consequences of discrimination or attacks on the fundamental rights derived therefrom. Analyses of genetic data can be classified into three types of “genetic tests”: a) DNA analysis b) genetic monitoring, and c) genetic screening.

⁵⁹ Santiago Grisolia, chairman of the Scientific Co-ordinating Committee of UNESCO for the Human Genome Project, affirms that knowledge of the human genome will mean social revolution and will allow the practice of individualised, preventive medicine.

⁶⁰ Cf. Cadiet, L., Laberge, C.M., *La génétique humaine (De l’information à l’informatisation)*, Paris-Montreal 1992, p. 45.

Let's take a brief look at the most commonly used genetic tests:

- a) **DNA analysis:** This expert forensic test has already achieved a much higher level of reliability than the tests previously used for these purposes. The fact that DNA –*the genetic fingerprint*– reflects the unique character of each individual is proving to be of great service to justice, particularly in the context of criminal and family law. It permits individuals to be identified with a high degree of accuracy, much higher, in fact, than older methods such as fingerprints and blood tests. DNA analysis is being used in paternity suits and to determine who has committed a crime, since this test is accepted by positive law. Use of this kind of testing has recently been proposed for the identification of newborns, in order to prevent careless confusion or fraudulent exchanges of children. The positive aspects and advantages of this test versus previous methods cannot be questioned, as it is a highly valuable tool for justice and makes it possible to eliminate a number of legal errors.

The problems posed by this test arise from the abuse of DNA data banks; thus, this activity must be governed by stringent controls. There are numerous hazards. First abusive use within the legal context; that is, abusive use of these genetic data to “open a file” or indefinitely “label” individuals who have committed a crime, preventing or hindering social reintegration, an objective established for these individuals by Art. 25.2 of the Spanish Constitution for criminal law.⁶¹ Second, problems arise due to the fact that a DNA study on a individual can be used not only for identification but also to provide other types of genetic information that could be used illegitimately. In any case, the risk does not stem from the test itself but rather from misuse thereof. This is precisely the question leading to reticence about the aforesaid use of the test in identifying newborns, particularly because abuse of the data obtained would affect children who did not consent to these tests and who will also be “labelled” from birth.

The existence of DNA banks, as well as computer cross-referencing of data obtained for other purposes –hiring, loans, etc.– will allow

⁶¹ In France, a file has been created with the genetic fingerprints of sexual delinquents (National Automated File of Genetic Fingerprints) under the auspices of the Ministry of Internal Affairs, with the data kept for 40 years. In June 1998, a bill was passed in Spain whereby the government is urged to regulate the use of DNA analyses in the investigation of paternity cases and of certain crimes.

governments to control their citizens, such that we could end up being “transparent citizens”, with all the dangers this entails. Nevertheless, as we mentioned, this problem is avoidable through adequate control of the data banks and does not involve any risk of attacking fundamental rights.

- b) **Genetic monitoring:** These tests are intended to monitor damage that may have occurred in the chromosome (cytogenetic) structure or in the molecular structure of the DNA in the chromosomes (noncytogenetic). Through regular analyses, these tests make it possible identify the effects on cells of exposure to radiation or chemicals in the environment. The tests are designed to detect “biologically significant” genetic damage or mutations in the DNA induced by this kind of exposure. The possibility of learning more about the adverse effects on health of certain substances not only makes it possible to prevent further injury to individuals who have already experienced these alterations, but also to avoid injury to other individuals through the implementation of safety measures or bans on certain substances. Consequently, these tests have an essential importance in occupational risk prevention and generally, in the achievement by public authorities of a nonharmful environment for public health. These tests do not, however, identify the disease or risk caused by these substances in the individual, but only detect which components are harmful to health.

Such tests pose serious problems in the context of labour law since, since just as they promote prevention of occupational risk, they also allow the discrimination of workers whose genetic information make them more prone to a disease,⁶² without necessarily allowing the disease to be identified and treated.

- c) **Genetic screening:** Genetic screening is a one-time test, as opposed to genetic monitoring which is done regularly. This type of analysis looks at the genetic structure of a person, making it possible to detect the presence of genetic abnormalities. Based on the test results, the consequences derived from the genetic structure in relation to the probability of developing certain diseases can be assessed.

This test, although apparently enormously useful in financial terms as preliminary tests for hiring employees, granting loans or calculating

⁶² For more on the problems posed by genetic analyses in labour relations, see 6. below.

insurance premiums, raises important issues in terms of individual rights. The potential discrimination is substantially greater than the discrimination resulting from the other two types. The severity of the problem stems from the fact that there is no scientific certainty about apparently certain conclusions. For this reason, I have used the word “assess”. As I mentioned earlier, in most cases there is no solution for the problem revealed by the test.⁶³

Viewed as a whole, we can conclude that once it is sufficiently developed, predictive medicine will be enormously positive for individual and public health. Many of the problems we have mentioned are not intrinsic to predictive medicine but rather to the fact that it is still in an early stage. Predictive medicine –in the context of disease prevention and the use of gene therapy– makes it possible to prevent disease and implement faster, less invasive treatments than those now existing in conventional medicine. In relation to occupational risk, the area where predictive medicine is being most highly developed, the limitations of current medicine are being underscored precisely because its preventive potential comes into play only once the disease has developed. Effective occupational risk prevention requires tools that provide information on risks before the symptoms appear, establishing the necessary measures to avoid them.⁶⁴ Predictive medicine of a genetic nature allows truly effective prevention of employee health risks resulting from their profession and also risks to third parties resulting from a disease before the symptoms appear. Moreover, genetic predictive medicine enables workers more susceptible to developing certain professional diseases to be identified and is more reliable than conventional medicine in compulsory aptitude tests when hiring workers for jobs involving some risk of disease.⁶⁵

4.2.2. Predictive medicine and economics

As mentioned earlier, once genetic testing becomes sufficiently reliable and gene therapies are available after the tests are performed, genetic tests become beneficial from an economic standpoint, provided we understand economics

⁶³ See Section 6, which looks at the minimum requirements for the legitimacy of this type of genetic test.

⁶⁴ Cf. Goñi Sein, *Límites constitucionales a los tratamientos médicos obligatorios establecidos como medida de prevención de riesgos laborales*, *Rev. de Derecho Social*, no. 5, 1999, p. 50.

⁶⁵ Cf. Art. 196 of the General Social Security Act, that requires companies to perform a preliminary examination before hiring for job positions involving any risk of job-related disease.

in terms of the company or state, since individual economies (particularly the weakest) are severely affected by these tests. As will be seen in the next two sections, both in the context of certain employee relations which are identified by an actual material inequality that contrasts with the theoretical equality required when hiring employees in the private sector, as well as in the context of labour relations, one party is always financially benefited whereas the other party can be financially (and personally) harmed to a significant extent.

In terms of public economics, the benefits or damage to the State or its citizens will depend on the governmental model. In the case of governments with a liberal economy, genetic testing unquestionably favours private enterprise, whether it be banks, insurance companies or any firm with employees. The financial benefits enjoyed by the companies result not only from a lowering of costs but also from the possibility of avoiding future expenses, thereby allowing better planning. In this liberal economic model, the State assumes only a small portion of the social cost of health, unemployment and pensions. Therefore, the public economy will not experience a serious setback, at least not a financial one. However, politically the situation could indeed be disastrous because of the enormous social unbalance resulting from systematic use of genetic criteria for health.

In the case of welfare states, the government is obliged to care for public health and unemployment. This is the case in Spain, as stated in Art. 43 and 49 of the 1978 Constitution which recognises the right of all citizens to health protection. To be effective, this right requires that public officials adopt appropriate measures to meet this need. In response to the constitutional requirements, the Spanish General Healthcare Act of 25 April 1986 acknowledges that all citizens and foreign residents in Spain are entitled to healthcare from the health system. Nevertheless, the right to free healthcare is not implemented globally and immediately; rather it is implemented only gradually in order to carefully assess cost trends.

In my opinion, even with the current state of the art in genetics, predictive medicine may be essential to the aim of complying with the constitutional requirement of a universal public health system. From a regulatory standpoint, this statement is confirmed by Spain's General Healthcare Act where Art. 6.3 establishes that the government must ensure that the healthcare activity carried out addresses the prevention of disease, as well as the cure. In order to comply with these requirements, preventive medicine plays a key economic

role, as it reduces the cost of providing healthcare. Cost-cutting is possible through environmental improvements related to radiation and chemical exposure detected in genetic monitoring and through intervention before the disease develops, based on genetic screening. Costs can be substantially reduced as test reliability improves and as genetic techniques advance to become alternatives to conventional surgical and pharmacological medicine in the treatment of disease.

Genetic tests can also be enormously effective in terms of occupational risk prevention and other relevant aspects implied by the obligation to protect public health. They allow harmful agents to be detected and safety measures to be taken through genetic monitoring. Moreover, as genetic test results improve, it will be possible to reduce the incidence of job-related disease and the dangers resulting from imprudent actions by identifying the individuals susceptible to certain risks or with inadequate characteristics for the job they hold. The Occupational Risk Prevention Act 31/1995 of 8 November relates to the requirements set forth in Art. 40.2. of the Spanish Constitution, which involves the need to develop a policy of employer health protection through the prevention of risk derived from work. This law also transposes Council Directive 89/391/EEC (LCEur 1989, 854) on the introduction of measures to encourage improvements in the safety and health of workers at their place of employment, which specifies the general legal framework for the EU occupational risk prevention policy.⁶⁶

There will be little danger of discrimination due to the use of genetic testing if pertinent control measures are taken to prevent abuse by companies when hiring, reassigning or dismissing workers and if the social welfare systems for unemployment and pensions are maintained. Nevertheless, this optimistic vision is relative to the situation. Although the political and social system has not formally changed, trends in economic policy related to social costs are reversing and we are moving dangerously close to a system of liberal

⁶⁶ Employee health protection holds a prominent position in EU regulations, as is obvious from the modification of the treaty establishing the European Economic Community (LCEur 1986, 8) by the Single European Act (RCL 1987, 1562). Based on Article 118 A) thereof, the Member States have promoted improvements in the workplace since this legislation became effect, in order to achieve the aforesaid objective of harmonisation in the progress of employee health and safety conditions. This objective was strengthened by the Treaty on European Union (RCL 1994, 81 and 1659 and RCL 1997, 917) through the procedure contemplated therein for adoption through directives of the minimum regulations that must be progressively implemented.

capitalism that is increasingly similar to the one advocated in the 19th century. The response to this liberal trend can already be seen in some laws, particularly the Private Insurance Regulation. As we will see, even when it does not refer expressly to genetic testing, this law opens the door to legitimising these tests since its structure begins by assuming the fallacy that the parties involved in these contracts are on equal footing.

4.2.3. Predictive medicine and the health of offspring

Genetic engineering and assisted reproduction make it possible to eliminate genetic defects before or after conception and to select embryos with better genetic material. In this way, advances toward improvement of the human race could theoretically be achieved. One of the indisputable “benefits” of predictive medicine from a strictly scientific or medical standpoint is the ability to detect hereditary diseases and consequently, prevent them from being passed on. Through genetic analyses we can detect genes that transmit a hereditary disease before the symptoms develop and before the person has offspring (at least naturally). If, once a hereditary disease has been detected, assisted reproduction systems allowing embryo screening or gene therapy on embryos are used to avoid disease transmission, certain diseases of a strictly genetic nature can be eradicated.

It is also evident from a human perspective that parents cannot be denied the right to procure the best for their children and this includes the right to prevent them from developing diseases, when science makes this possible. We could even claim that it would be ethically unacceptable for people to transmit a disease to their children, if it could be avoided.

Based on this definition, it appears to be difficult to point out any risk that could derive from the generalisation of these techniques. Nevertheless, in my opinion we should mention several risks briefly discussed earlier in relation to predictive medicine.⁶⁷ From the perspective of biodiversity and its consequences for the survival of the human race and the health of future generations, I feel that science is not in a position to guarantee that these techniques are innocuous at long-term. From ethical positions, we must look at the consequences that could arise from the implementation of prenatal gene therapies in developed countries, particularly, in certain strata of the

⁶⁷ See Point 4.1.

population. Financial, physical and even psychological inequality combined with cultural inequality resulting therefrom could create insurmountable barriers for the less fortunate. Both today and at mid-term, it is an idealisation to believe that these techniques could actually be “globalised”. Consequently, we must question whether it is legitimate to foster such a situation. Moreover, I feel that we are unable to predict what the social and political consequences of the resulting inequality would be.

5. Private law and predictive medicine

5.1. Private law, equality of parties and legitimacy of genetic testing

A central principle of private law consists in the equality and free will of the parties signing an agreement. Based on this principle, there should be few limitations on the freedom of the parties to define the clauses governing the agreement. If the freedom to establish clauses is based on the equality of the contractual parties and on the fact that neither party is compelled to sign the agreement, the principle of contractual freedom must be revised in the cases where these premises fail for any reason.

Insurance, particularly life, health and pension insurance, deserves particular attention. When the State does not guarantee compliance with the constitutional requirement of universal healthcare and pension, the public is obliged to obtain private insurance. At this moment, one of the premises –voluntary nature of contracts– begins to fail. The same occurs in the case of insurance required by law. An example is the case of car insurance where the obligation of a person to obtain insurance is in conflict with the insurer’s freedom to refuse to provide an insurance policy,⁶⁸ to set disproportionate premiums⁶⁹ or to unilaterally cancel the agreement if one of the payments for a period exceeds the limit of this period. When the legal or social obligation behind many insurance policies is combined with the effective inequality between parties also existing in these cases, the principle of freedom between parties should enter into discussion.⁷⁰ Limits should be posed on this

⁶⁸ A case in point is the current situation of young drivers.

⁶⁹ In the case of drivers, whether young or not so young.

⁷⁰ The Austrian Law 510/1994 on genetic technology provides coverage to anyone requesting insurance, in order to prevent the conclusion of a contract from depending on the results of a genetic analysis.

contractual freedom because of the existing inequality. This would mean applying the principle of equality in its material sense, namely treat equals as equals and unequals as unequals. This principle of material equality in relation to predictive medicine should lead to a ban on the limitation or refusal of basic services based on a person's predisposition to a disease.

5.2. Genetic analyses for insurance contracts. Insured's obligation to declare

The implementation of predictive medicine can have an extremely negative impact when it is related to the lack of equality and freewill of many insurance policies, since insurers may refuse to underwrite policies for individuals with adverse genetic health, implying that the individuals most needing coverage will be unprotected.

The qualitative differences in genetic analyses (inasmuch as they allow future diseases to be predicted) versus traditional analyses causes severe conflict with respect to the insured's obligation to make an accurate declaration on his or her health condition.⁷¹ According to current regulations in Spain, the insurer can provide direct information on the specific risk it intends to assume only with extreme difficulty and within a limited scope, with this depending on the information provided by the insured.⁷² As a result, the insurance contract is considered an agreement of utmost good faith (*uberrimae bona fidei*). In response to the investigative limitations of the insurer, the law imposes the duty to declare all known circumstances by the insured that could affect the assessment of risk.⁷³ Nevertheless, the insured party, or the policyholder, is exempt from this obligation when there are circumstances that could influence the assessment of risk that are not considered in the insurer's questionnaire.⁷⁴ Due to this limitation, the obligation to declare is said to have become an obligation to respond,⁷⁵ which

⁷¹ Sánchez Calero, *Efectos de la declaración exacta del asegurado*, in *Riesgo y Seguro*, no. 11, 1965, p. 54

⁷² Sánchez Calero, *Instituciones de Derecho Mercantil*, vol. II, 23rd ed., ed. McGraw-Hill, Madrid 2000, p. 384.

⁷³ See Arts. 10 and 89 Law 50/1980, of 8 October, on insurance contracts, in effect after Law 30/1995, of 8 November, on the regulation and supervision of private insurance.

⁷⁴ This key limitation on the obligation to declare risk circumstances was enacted by Law 21/1990, of 19 December, in order to adapt Spanish law to Directive 88/357/EEC.

⁷⁵ Spanish Supreme Court ruling (civil court) of 2 December 1997.

would imply that any omission in declaring a circumstance that would modify risk has no relevance. However, this is not true because along with this limit to the policyholder's obligation to declare, the same legislation allows the insurer to cancel the contract when it learns of omissions or inaccuracies in the declaration by simply notifying the policyholder within one month.⁷⁶ In addition, premiums for the period in progress must be paid by the insured when there is fraud or extreme culpability regarding omissions or inaccuracies at the time the declaration is made. If the catastrophe occurs within one month prior to the notification and fraud or extreme culpability also exist, the insurer is released from its obligation to pay the corresponding indemnity.⁷⁷ If there is no fraud or extreme culpability, the indemnity may be proportionally reduced to the difference between the agreed premium and the premium that would have been applied if the true risk had been known.⁷⁸

The situation becomes even more serious for individuals with "adverse genetic health" who, as part of their obligation to declare, must declare the risk of future disease.⁷⁹ In Spain there are no regulations on the content of the questionnaires given by insurance companies to the insured, nor is there any legislation about whether questions on genetic tests can be included.⁸⁰ In terms of the legitimacy of genetic analyses, we only have Art. 12 of the Convention on Human Rights and Biomedicine, ratified by Spain, whereby predictive genetic testing can only be performed for medical or research purposes. A literal interpretation of this legislation would lead to a banning of

⁷⁶ Art. 10 of Law 50/1980 on insurance contracts. In any case, this legislation relaxes the above regulation, laid down in Art. 381 of the Commercial Code, whereby an inaccurate declaration, although when made in good faith, allowed the possibility to cancel the insurance.

⁷⁷ In both cases, the jurisprudence understands that fraud exists by the mere fact that the subject, although he or she knew the circumstance, omitted it, without requiring any specific act performed with the intention of deceiving the other party. Cf. Spanish Supreme Court rulings (civil court) of 12 July 1993 and 31 December 1998.

⁷⁸ Cf. Spanish Supreme Court rulings (civil court) of 16 July 1990 and 22 December 1992.

⁷⁹ The possibility is not remote. In Great Britain, on 13 October 2000 the Genetics and Insurance Committee –a government institution created to regulate this area– approved insurers' use of tests related to Huntington's disease. Few people question that this will pave the way for insurance companies to request their clients' genetic data.

⁸⁰ Cf. Fernández Domínguez, *Pruebas genéticas en Derecho del Trabajo*, ed. Civitas, Madrid, Spain, 1999, p. 103, on the hiring of a genetically defective worker that increases the insurer's risks and "presents in all its harshness the obligation to declare risk imposed on the potential policyholder, which must also extend to the insured in cases where the contract is obtained through others".

predictive genetic testing for insurance or labour relations. Nevertheless, since the declaration is quite generic, I do not believe this interpretation is sustained in practice. This is especially true with the understanding that preliminary or periodic predictive analyses for an insurance or employee contract are to have immediate medical purposes, even when these immediate purposes are other than medical.

Given the current regulations on insurance contracts and in the light of the legal vacuum I have briefly outlined, when the practice of genetic analysis is generalised, the insured would be obliged to declare data on genetic makeup even when this information is not asked for on the company questionnaire. Awareness of a genetic disease indisputably leads to a variation in risk and consequently, the insurer can cancel the agreement and, if it is applicable and fraud or extreme culpability exist, require the insured to pay the premiums and then not provide the corresponding services. In any case, since the obligation to declare any change in risk continues throughout the agreement, if the insurer becomes aware of these genetic data at any time after the agreement is signed, it is equally entitled to cancel the agreement, require that the policyholder pay the premiums and not pay the service or pay only an amount proportional to the difference between the declared and the actual risk.

5.3. Predictive medicine and insurance fraud

The lack of legal regulations related to the legitimacy or illegitimacy of genetic testing prior to obtaining an insurance policy or during the contract term may imply not only that the weaker party in the contractual relationship is unprotected but also that he or she can be accused of fraud. If the policyholder does not report a known genetic alteration, even if it is not expressly indicated on the insurance company's protocols or questionnaires, the doubt arises as to whether this will have civil consequences, such as those mentioned in the previous section or even be considered fraud. In Spain I understand that this legal obligation does not exist and, consequently, the policyholder is only under the obligation to answer the stipulated questions, particularly because genetic information does not need to be declared at present.

In the cases where a person, based on examinations that are still not universally recognised and are not under established protocols, becomes aware that he or she will develop a disease relatively soon, although the symptoms

have not as yet manifested, one cannot consider that the deceit involved is great enough to constitute fraud. Fraud requires that there be considerable deceit leading to error and that this error determines a distribution of assets in detriment to the party holding such assets or to a third party. In the proposed case, I feel that considerable deceit cannot be said to exist. The omission of information may only be considered deceit when the subject gives biased information that leads to error rather simply omitting information, and not when the subject omits stating adverse circumstances, except when there is a specific duty to declare them.

The issue as to the ethics and/or legitimacy of obliging a person to provide information clearly in his or her detriment is an entirely different question. Based on Art. 24.2 of the Spanish Constitution, the criminal process establishes a guarantee derived from the principle of defence whereby “no one is obliged to declare against him or herself”. This principle should be analysed to determine if it could be extrapolated to other areas where a declaration may be detrimental to the person, as occurs in the proposed case.

6. Predictive medicine in labour relations

6.1. Genetic analyses in labour law

6.1.1. Genetic analyses in comparative labour law

The biggest problems related to genetic testing are unquestionably in the area of labour relations, with regard to both the supposed advantages to the company and employee and to the potential discrimination posed to the employee. In some countries, specific regulation limiting compulsory genetic analyses already exists. The European Social Charter of 18 October 1961 recognises several rights, among them those laid down in Art. 1.2. b), “the right of all to equitable working conditions, with prohibition of worker discrimination on the basis of their *genetic heredity*”. In an attempt to address the problems resulting from genetic analyses, the Council of Europe’s decision of 29 June 1990 indicates that the right to a genetic identity is part of a person’s right to integrity and dignity. It also admits that there is a risk that genetic analysis will be performed for control purposes and warns about the need for clear legislation in this respect. A number of legislative bills have been proposed by various European states in this regard, ranging from the Danish Ministry of Labour’s total ban on genetic testing in relation to employment and insurance

in 1992⁸¹ to the opinions of the German Society of Occupational Medicine⁸² which openly criticise any restriction on genetic tests as a preventive measure for occupational health.⁸³ In Italy the Penal Code theoretically prohibits genetic analyses intended to exclude workers prone to certain pathologies in any way related to the type of work involved.⁸⁴ In contrast, testing to determine the presence of a genetic disease for employment applications and to select the best candidates is allowed for reasons related to individual health and occupational risks and not financial purposes (the reason behind the criminal ban). As a result, this kind of analysis is used with increasing frequency.

6.1.2. Genetic analyses in Spanish labour law

In Spain genetic analyses are neither prohibited nor expressly permitted and, consequently, specific measures or procedures for the performance of these analyses are not regulated. Although there is no express declaration of their legitimacy, it is understood (although limited by certain suppositions) through Art. 25.2. of the Spanish Occupational Risk Prevention Act (“Ley de Prevención de Riesgos Laborables”, or LPRL): “Likewise, in the assessments the employer must take into account any risk factors that could affect the procreation function of employees and workers, particularly in relation to exposure to physical, chemical and/or biological agents that could have mutagenic effects and/or toxic effects for procreation, both in the aspects related to fertility as well as those related to the development of the offspring, in order to adopt the necessary preventive measures”.

Although genetic monitoring is needed to determine whether chemicals or radiation have “mutagenic effects”, no specific regulations exist. As a result, we must resort to the general laws containing principles that are also compulsory

⁸¹ Cf. *Ethics and Mapping of the Human Genome*, Danish Council of Ethics 1992 Report; the Norwegian regulation in the law on the medical use of biotechnology is analogous, being extremely restrictive of work-related genetic testing and allowing it only with the employee’s prior consent, the French regulation laid down in Laws 94/563 and 94/654 of 29 July, as Austrian Law 510/1994.

⁸² Ammon, U., *Pruebas genéticas en el mundo laboral. Contribución al debate sobre la necesidad de regulación en la República Federal Alemana*, in *El Derecho ante el Proyecto de Genoma Humano*, Vol. IV, Fundación BBV – Universidad de Deusto, 1994, p. 322.

⁸³ Likewise, UK legislation in this area leads to the conclusion that public interest prevails over the right to privacy.

⁸⁴ Cf. Catalano, G., *Análisis genético de los trabajadores italianos: un enfoque jurídico*, in *El derecho ante el proyecto de genoma humano*, Vol. IV, Fundación BBV – Universidad de Deusto, 1994, p. 329f.

in these cases, irrespective of the appropriateness of explicit legislation, given the unique characteristics involved in genetic tests. General criteria for medical exams in at work are included in Art. 22 of the Occupational Risk Prevention Act.⁸⁵ The general principle is that health supervision is allowed voluntarily and requires the worker's informed consent. Nevertheless, this general principle is distorted in practice by the voluntary exception of "...cases in which the performance of examinations is essential to assess the effects of working conditions on workers' health or to check whether or not the worker's health condition could pose a danger to him or her, to other workers or to other individuals related to the company or when so established in a legal regulation relating to the protection from specific risks and particularly hazardous activities". In these cases the legislation establishes that the examination will be handled after the workers' representatives are informed, but does not specify the effects of an adverse report, i.e., the connection of the employer with the report is not regulated. Moreover, the fact that this report comes from the worker's representatives does not mean that the decision-making capacity of the person entitled to the right to freedom has been delegated to his or her representatives.

In relation to compulsory examinations, the situation becomes more complicated due to the guarantor status assigned by law to the employer.⁸⁶ In

⁸⁵ Article 22. - Safeguarding of health

1. The employer shall guarantee regular monitoring of the health status of the workers employed, based on the inherent risks of the job.

Such monitoring may only be performed with the employee's consent. Exceptions to voluntary consent (with prior notice from the employees' representatives) shall consist in cases in which the performance of examinations is essential to assess the effects of working conditions on workers' health or to check whether or not the worker's health condition could pose a danger to him or her, to other workers or to other individuals related to the company or when so established in a legal regulation relating to the protection from specific risks and particularly hazardous activities.

In any case, the examinations or tests should cause minimal inconvenience to the worker and should be proportional to the risk.

3. The results of the health monitoring mentioned in the previous section shall be reported to the workers concerned.

4. Data related to the health monitoring of workers may not be used for discriminatory purposes or to the detriment of the worker.

Access to medical information of a personal nature shall be limited to the medical personnel and the health authorities handling the health monitoring of the workers, and the information may not be provided to the employer or to other individuals without the worker's express consent.

Regardless of the foregoing, the employer and the individuals or bodies responsible for prevention shall be informed of the conclusions resulting from the examinations in relation to the worker's aptitude to perform his or her job or in relation to the need to implement or improve protective and preventive measures, in order to carry out their responsibilities with respect to prevention.

⁸⁶ See Goñi Sein, *Rev. de Derecho Social*, no. 5, 1999, pp. 49ff.

Art. 47.2 of the Occupational Risk Prevention Act the non-performance of compulsory medical examinations and regular monitoring tests designed to monitor workers' health conditions set forth in the occupational risk prevention regulations is considered a serious administrative offence. In addition, as stated in Art. 197.2. of the General Social Security Act the employer is considered responsible for all healthcare services derived from job-related disease when the company has not complied with its obligation to perform preliminary or periodic examinations.⁸⁷

Compulsory examinations include regular check-ups designed to monitor health as well as examinations performed prior to hiring the employee and aimed at assessing the candidate's psychological capacity to perform the job.⁸⁸ The aim in these cases is to verify that the potential employee has the necessary aptitude to perform the job. The major limitation on freedom implied in these compulsory examinations means they should be regulated by law, with the range depending on the degree of coercion authorised for the employer.⁸⁹ In no case should the limitation of rights be performed through instruments such as collective bargaining agreements. Art. 43.2 of the Spanish Constitution establishes a legal reserve to determine healthcare responsibilities, including compulsory employee examinations.⁹⁰

⁸⁷ Cf. Goñi Sein, *Rev. de Derecho Social*, no. 5, 1999, p. 50, which states that the employer is liable for damage to the worker's health that would have been avoided if these compulsory examinations had been performed.

⁸⁸ Article 25.- Protection of workers particularly susceptible to certain risks

1. The employer shall specifically guarantee the protection of workers who, because of their inherent personal characteristics or known biological condition, including those with an acknowledged physical, psychological or sensorial disability, are particularly susceptible to the risks derived from the job. Hence, these aspects must be taken into account in the risk assessments and, based on the assessments, the necessary preventive and protective measures must be implemented by the employer.

Workers shall not be employed in jobs where –because of their duly recognised personal characteristics, biological status or physical, psychological or sensorial handicap– they could place themselves, other workers or other persons related to the company situations in jeopardy, or in general, be placed in manifestly temporary conditions or situations that do not meet the psychological and physical requirements associated with the respective jobs⁷.

Likewise, Art. 196 of the General Social Security Act requires that companies perform a preliminary examination prior to hiring an employee for a job involving any risk of work-related diseases.

⁸⁹ The Spanish Constitutional Court ruling 140/86 advocates the need for an organic law in the case of acts that deprive a person of freedom, as laid down in Art. 81.1. of the Spanish Constitution.

⁹⁰ Cf. Goñi Sein, *Rev. de Derecho Social*, no. 5, 1999, p. 66f.

6.1.3. Requirements of pre-hiring and regular compulsory examinations

As can be seen, in practice our regulations allow a company to carry out any type of genetic test it deems desirable. Moreover, systematic interpretation of these regulations indicates that in many cases the employer is obliged to perform compulsory examinations of the workers before hiring, as well as subsequent periodic check-ups while the individual is employed and a final examination upon completion of his or her term of employment.⁹¹ For this reason, it is important to analyse the limits to these compulsory examinations until specific regulations addressing the issue are formulated. The limit should be set on the basis of another requirement laid down in this legislation: “that they be essential”. This idea of “essential” must be related to the reason that makes them essential, which can only consist in certain risk to the health of the individual or of third parties (employees and non-employees). This rephrases the question to the concept of justification due to need, in which the proportionality principle between the violation of freedom resulting from examination without consent and the predictable risks to health must be strictly adhered to.⁹² Moreover, these examinations must be made in the least harmful way for the worker, “cause the least inconvenience and discomfort”, and include only the tests strictly needed for the objectives pursued.

The idea of “functionalising” the examinations in view of the physical and psychological risks and requirements of the specific job follows these same lines. This measure is laid down in Art. 14.1. of the Occupational Risk Prevention Act which requires that the measure be taken “on the basis of the risks inherent to the job”.⁹³ Consequently, the objectives of the analyses must be limited to those strictly required as dictated by the nature of the job.

Equally important is the confidentiality needed for examinations without consent, with such confidentiality being at least similar to that required in voluntary examinations. Art. 22 of the same law establishes respect for privacy

⁹¹ Art. 22. 5. of the Occupational Risk Prevention Act “In the event that the nature of the risks inherent to the job require it, the workers’ right to regular monitoring of their health condition shall extend beyond termination of the labour relation under the terms set forth by the regulations.”

⁹² Cf. Mir Puig, *Derecho Penal...*, p. 453f., on the importance of the proportionality principle in the condition of justifiable need.

⁹³ Cf. Goñi Sein, *Rev. de Derecho Social*, no. 5, 1999, p. 54.

and dignity as a general rule, but contemplates an exception that, as occurred with regard to consent, is likely to become the rule in practice: “Regardless of the foregoing, the employer and the individuals or bodies responsible for prevention shall be informed of the conclusions resulting from the examinations in relation to the worker’s aptitude to perform his or her job or in relation to the need to implement or improve protective and preventive measures, in order to properly carry out their responsibilities with respect to prevention”. The arguments put forth for consent also stand in terms of protecting privacy, understanding that only aspects that are actually relevant to achieving the purpose of prevention and that warrant this interference may be reported.⁹⁴

Although it may seem obvious, I mention that examinations must be performed in such a way that they minimise any harm to the worker’s privacy and dignity, and naturally, to their health.. Moreover, the results must be reliable, with the tests carried out properly using appropriate techniques and trained personnel.⁹⁵ Therefore, specific protocols must be developed and used on the basis of the risk to which the worker is exposed.⁹⁶

6.2. Employer arguments in favour of genetic analyses

The direct and/or indirect financial benefits potentially enjoyed by the company because of the general use of genetic testing during the hiring process and throughout the employee’s working life are unquestionable, particularly as genetic tests become less expensive and easier to use with standardisation and improvements in the techniques. When implementing the use of genetic analyses the company first assesses their cost and reliability. Secondly, the

⁹⁴ Generally speaking, collective bargaining agreements usually establish that the employee must agree to any medical examination prescribed by the medical staff. These agreements do not usually refer to the need to justify the obligatory nature of a specific examination in each case. For clauses contained in various collective bargaining agreements, see Goñi Sein, *Rev. de Derecho Social*, no. 5, 1999, p. 61, n. 19.

⁹⁵ Cf. Art. 22.6. of the Occupational Risk Prevention Act: “Measures undertaken to monitor and control employee health shall be carried out by healthcare staff with accredited technical competence, training and skills”.

⁹⁶ Art. 10. a) of the Occupational Risk Prevention Act entrusts government bodies with establishing “...appropriate means for assessing and controlling healthcare activities carried out in the prevention departments acting within the companies. To do so, they must draw up guidelines and protocols for their activities, heeding the opinion of scientific societies and submitting the aforesaid services to their scrutiny”.

various legal difficulties established (whether greater or smaller) are considered. Depending on the reliability and affordability of the tests, multiple benefits can be obtained since genetic analysis allow exclusion of individuals without “genetic health” rather than those already suffering from a disease. This yields a number of benefits, which can be strictly financial, or ethical and social as well.

6.2.1. Strictly financial employer arguments in favour of genetic analyses

Companies that use genetic analyses and exclude employees at risk for genetic disease obtain high job performance, lower absenteeism, decreased staff turnover and therefore, lower costs for professional retraining, ensuring adaptation to the job and a return on investments in training. This also avoids increases in welfare payments, particularly in systems where the employer’s share is based on the number of job-related accidents and diseases. Savings are obtained by avoiding the lawsuits and indemnities for accidents by and between employees resulting from negligence in choosing and in supervising workers.

Genetic analyses taken to the extreme will provide power to control and manage the employee above the current limits, since the worker would be a person whose qualities are “crystal clear”.⁹⁷ Genetic analysis of a worker’s phenotype allows the employer to take advantage of the potential and real ability (whether favourable or unfavourable) to assign the most appropriate tasks, the most appropriate location, and the most appropriate pay for his or her productivity and, if applicable, to dismiss the employee when allowed for objective-genetic reasons. In combination, this means that the employer will be able to maintain or increase productivity at a low cost.

6.2.2. Social and ethical employer arguments in favour of genetic analyses

The improvements in productivity brought about by predictive medicine are beneficial to economic and social progress, as well as to full employment, as a guiding principle of economic and social policy laid down in Art. 40.1. of the Spanish Constitution. A competitive advantage in the domestic and international markets is attained by improving product quality and cutting production costs, hence predictive medicine could be tremendously useful.

⁹⁷ Cf. Fernández Domínguez, *Pruebas genéticas en Derecho del Trabajo ...*, pp. 95ff.

Genetic analyses, as we saw from our list of types of genetic testing, allow the employer to adopt safety measures that eliminate risk to worker health resulting from exposure to radiation or chemicals. An awareness of the worker's genetic makeup provides a more precise idea of probable injury and is helpful for seeking ways to prevent it.

6.3. Genetic analyses and workers' rights

Employees are also benefited by an awareness of their genetic makeup, as it allows them to steer their professional career in keeping with their physical and psychological aptitudes and prevents them from running risks that could compromise their health. This awareness also allows them to highlight favourable aspects for attaining a specific job or better compensation. Nevertheless, the risks are also evident. We have already shown the risks of test results that identify an individual with "poor genetic health", where in fact the difficulty of finding employment or performing certain jobs could become insurmountable. "Legal" discrimination of workers with poor "genetic health" will mean that these individuals will be hard-pressed to find a job offering them a minimal degree of satisfaction. The employee's psychological burden resulting from an awareness of genetic testing results cannot be overlooked, particularly when there are no means to minimise or prevent development of the disease.

6.4. Genetic analyses and public interest

Arguments for genetic analyses are all favourable when we look at them from the standpoint of public interest:

- a) Firstly, the State is enormously aided by predictive medicine in meeting the general needs regarding health prevention required by Art. 43 of the Spanish Constitution.
- b) The interest of Social Security in predictive medicine as a way to reduce the financial burdens associated with healthcare for job-related disease and for genetic disease is also indisputable.
- c) For the employee's colleagues, predictive medicine can decrease or prevent the risk involved in working with a person who is genetically incompetent or dangerous because of a predisposition to develop certain diseases or behaviour patterns.

- d) It is also useful for the employees' representatives, who are assigned the responsibility for "monitoring and controlling safety and hygiene conditions in the workplace"⁹⁸ In this case, in addition to safeguarding this collective interest, the employees' representatives must assume the responsibility for preventing the company from using genetic tests exclusively to cut costs and for preventing discrimination against the worker when the risk is not real but merely a predisposition.
- e) For the benefit of the customers, clients and users of the company or the company's products, it is extremely important to prevent individuals with certain susceptibilities from holding jobs requiring a high degree of reliability (e.g., pilots, drivers, etc.).
- f) Lastly, generalised usage of genetic analyses will evidently benefit researchers and society in general, by allowing them to use the results obtained from this research.

Conclusion

Several conclusions can be drawn, although some are merely provisional. The provisional nature of the conclusions is inevitable, since it is generally agreed that our knowledge of genetics in medicine is only at an early stage. This situation will change as scientific knowledge advances. Thus, we must first make a distinction between the problems resulting from a lack of knowledge and the problems which will not be solved with scientific advances but will worsen unless control measures are introduced (and I feel my opinion is not overly pessimistic).

Regardless of whether we believe genetic medicine will make any qualitative (or only quantitative) differences with respect to the current situation, this is still medicine and as such, it must comply with all the legal, ethical and moral requirements currently in effect. The question rests on whether it is necessary to emphasise this protection, particularly in relation to consent and confidentiality. In my opinion, even if we accept that these differences are quantitative, they are so great that they prompt the need to improve and adapt current systems protecting an individual's autonomy and privacy in relation to predictive medicine.

⁹⁸ Art. 64.9. b) of the Workers' Statute and Art. 34.2. of the Occupational Risk Prevention Act.

Predictive medicine also raises new issues related to collective interests that often run counter to individual interests. The nature of predictive medicine (where research requires data on large population sets to further knowledge) has led to the idea that some personal data is of collective interest, a fact that contravenes our understanding of personal data as pertaining solely to the individual. Similar conflict arises between the interests of the law in creating genetic data banks and the rights of a criminal to social reintegration and human dignity.

Moreover, there is a utilitarian perspective that places collective and individual economic criteria in opposing positions. For the economy of the State or public bodies (social security and national healthcare programmes) or semi-public institutions (insurers, banks, etc.), there are almost exclusively benefits, whereas for the private economy, particularly that of genetically unhealthy individuals, it may be extremely harmful.

Based on the current state of predictive medicine, I feel that a guiding criterion for authorising certain analyses (along with the criteria of essential nature, confidentiality and functionality that must be present at all times) is the reliability of the results and particularly the existence of treatment when the results are positive. Any test that simply reveals a person's susceptibility to a disease but leaves significant doubt about the probability of developing the disease and about the measures that must be taken to decrease or eliminate this risk, should be used only with comprehensive informed consent.

The problems that can be aggravated by advances in predictive medicine are discrimination and inequality. Discrimination may involve personal, social, occupational or financial aspects, or all of them at once on many occasions. Inequality can lead to insurmountable increases in current gaps between the various social levels in developed countries and particularly between these countries and those in the third world. As a result, these aspects must be taken into consideration when controlling or limiting the use not only of genetic analyses (which particularly affect discrimination) but also gene therapies, which can determine insurmountable inequalities between genetically healthy and genetically weak individuals.

CONTRIBUTIONS TO THE DEBATE

Josep Egozcue

Professor of Cellular Biology, Universitat Autònoma de Barcelona.

I would now like to give the floor to the experts participating in this debate. The participants will make their contributions in groups, according to their area of expertise. However, I would first like to comment briefly on the excessive use that is made of the term “eugenics”.

According to Galton (1883), eugenics is social intervention in the inherited characteristics of the population. First of all, eugenics is obviously impossible because the elimination of affected individuals does not decrease the frequency of the gene in the population. Moreover, forced in-breeding would be necessary to increase the frequency of genes considered to be “favourable”, a tactic that would foster the appearance of undesirable recessive genes and is unacceptable in a democratic state. Secondly, because genetics always tends toward the mean, the genotypes considered to “best” at any specific time cannot be permanently established in organisms that reproduce sexually.

The selection of healthy embryos or the detection of foetuses affected by a pathology in order to prevent the birth of diseased children is not eugenics, since it is performed at an individual level. In any case, it could be described as fostering health.

Moreover, the barbaric practices of sterilising physically or mentally handicapped persons frequently performed in the early 20th century, or the criminal policies of eliminating ethnic groups (Jews, gypsies) or those with certain sexual orientations (homosexuals) practised by some political regimes should in no case be considered eugenic practices.

Galton, F. (1883). *Inquiries into Human Faculty and its Development*. McMillan, London.

Opinion of the Scientists

Ángel Carracedo

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I would first like to express my appreciation to the Foundation for organising this seminar and for inviting me, as well as to the speakers for the excellent work they have done.

As to Xavier Estivill's talk, I agree with each and every point he made. I would simply like to highlight the problem posed by the fact that medical genetics does not exist as a speciality and that there is a lack of specific courses on medical genetics in many medical schools within Spain. In a field such as medicine which experiences enormous changes on an ongoing basis, this situation is causing a handicap that must be dealt with urgently. The genetic information being generated will lead to a number of practical problems that will require adequate answers from both the medical and healthcare standpoint, as well as the educational and social point of view.

Another problem is the need to organise genomic medicine and to implement quality assurance systems in the laboratories. In view of the predictive component of this new medicine and its importance, the patient must have the right to know that the laboratory offers full quality assurance for genetic analysis. This right is not yet guaranteed, however. Nevertheless, the first step consists in solving the issue of creating a medical speciality and proper training in this field.

As to Mirentxu Corcoy's talk, I concur with most of her opinions, although at times the legal and genetic language do not coincide completely. In this regard, I would like to clarify that the DNA analysis used for forensic studies does not provide any information that is useful for predictive medicine and cannot be utilised to deduce any characteristic of a person except his or her identity. Nevertheless, the information is not completely neutral since details regarding family relationship can be deduced in some cases, thereby affecting privacy.

One of the main problems is the genetic files created for individuals by means of DNA polymorphism analysis. Although these files can be positive in terms of more efficient pursuit in crimes, particularly sexual and repeated

crimes, at the very least they affect fundamental individual rights, such as the right to privacy (in the meaning used earlier) and freedom. For this reason, the issue must be addressed urgently by legislation with an Organic Law. Spain is one of the few countries in Europe (along with Portugal and Greece) where there is no specific legislation on the subject, a fact that contrasts with the excellent quality of the forensic genetics laboratories.

I would also like to clarify that, as was stated in the speech, probability is not synonymous with uncertainty, and this causes considerable confusion in the thought on this matter. Probability is merely a measure of the uncertainty of an event (or an analysis, for example), and the safest estimates in existence are the ones governed by probability, in which uncertainty can be estimated accurately and is very low or practically null.

Miguel Ángel Peinado

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New knowledge on the molecular basis of a disease and the availability of methodologies capable of decoding the genome are bringing encouraging expectations for improvement in quality of life and health. Nevertheless these advances are not always accompanied by the possibility of implementing them on behalf of patients and therefore, they present ethical problems. It appears to be absolutely clear that the use of genetic information must be restricted only to cases in which there is an obvious benefit for the persons affected or for possible carriers. In other words, in diseases where the genetic diagnosis will not affect treatment strategies or survival rates, the technique should be limited to well-circumscribed research lines under the strict supervision of specialists.

In any case, the results of genetic studies should always be reported by professionals specialising in genetic counselling, who can discuss the implications and restrictions of these studies with the individuals involved. This information must be provided before the analyses are performed. Another important issue is personalised psychological counselling, which should be provided before and after genetic analysis by psychologists with an understanding of genetics.

Therefore, the public health system must utilise the benefits provided by the advances in genetics, but must also offer the structures needed for this

information to be used in a framework aimed at improving life expectancy and quality of life. The access to genetic information must be avoided if it is not approved by a team of professionals that offer individuals not only quality information but also medical and psychological support.

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Genetic testing allows the study of an individual's genotypical characteristics, which can be valuable for predicting and diagnosing disease. Hence, genetic analyses have unquestionable clinical value and allow precise advice to be given on the health aspects of those individuals in whom they are applied. Ethical dilemmas are raised by the potential use of such information by third parties (employers and insurance companies, other political or military institutions) for purposes that are not directly related to these ends.

It is not simple to establish the difference between a genetic disease and other types of disease. Genes are the determining factors for some pathologies. In many others, there is an interaction between genetic predisposition and environmental factors. Likewise some tests are clearly considered genetic (e.g., mutation or chromosome studies) whereas many other analyses used for the diagnosis of genetic diseases are useful for other purposes. These include the identification of biochemical or hormonal alterations (e.g., phenylalanine or TSH determinations from filter paper). This demonstrates that there is no clearly defined boundary between genetic diseases and other types of diseases, nor between genetic testing and non-genetic testing.

In principle, genetic testing should be considered in the same way as any other diagnostic medical test with or without additional prognostic value. The ethics governing indication or contraindication must be similar to those established in medical practice and should be based on the principle that they will be performed with regard to benefits and autonomy and without malice. In addition, they must be subject to the same confidentiality criteria required of the physician-patient relationship. It is true that there is general tacit agreement that genetic testing is looked at with special care because of the social fear concerning potential improper usage. Nevertheless, within a few years genetic testing will probably be regarded in exactly the same way as other medical tests, making these special considerations only provisional.

These principles of benefits, absence of malice, autonomy and confidentiality are applicable to all tests (genetic and non-genetic) that are used in population studies and social activities such as occupational health and insurance contracts. In keeping with these principles, it may be not much different to indicate the use of a microchip to analyse DNA for determining an individual's risk of developing cardiovascular disease instead of or along with taking their blood pressure or analysing serum levels of lipids. Ethical considerations must be evaluated on an individual basis for each type of study to determine if they are adequate.

The most important consideration to be addressed is that in most cases genetic tests should be prescribed, performed and validated by specialists trained in medical genetics because of their complexity. Adequate information must be provided before the analysis is indicated, informed consent must be obtained from the individual being tested and a system must be in place to report and act according to the results. Adequate infrastructure and qualified staff are needed to ensure that all medical procedures are performed in a rigorous and ethical manner. Unfortunately there is a serious problem in our setting posed by the lack of formal specialised training programmes and the lack of any recognition of the medical speciality of genetics in Spain. High priority should be assigned to solving this problem, in order to ensure that genetic testing is performed in the most suitable manner for individual healthcare, health planning, occupational health and regulation of insurance company requirements.

Opinion of the Legal Experts

Agustín Jorge Barreiro

Professor of Criminal Law, Universidad Autónoma de Madrid.

First of all, I would like to thank the Grífols Foundation for the invitation they extended, thereby allowing me to be here today among renowned experts to discuss the key issue of “Predictive Medicine and Discrimination”. I would like to express special appreciation to Dr. Mirentxu Corcoy for her kind invitation.

1. This morning we have had the opportunity to listen to the interesting speech presented by Dr. Xavier Estivill “On the road toward predictive medicine?”. The thoughts of a physician and geneticist have been extremely clarifying for me as a legal expert.

1.1. As a layman, I would like to highlight several comments made by Dr. Estivill:

a) Research into the human genome has made it possible to identify the genes of the main hereditary diseases, and in upcoming years information may be obtained on aspects related to an individual’s genetic susceptibility to develop diseases such as asthma, cancer, diabetes, hypertension, schizophrenia, etc.

b) These advances in genetics will obviously have positive aspects, as they will increase our understanding and make it possible to “diagnose, prevent, treat and even cure” disease. Nevertheless, information on the risk of developing certain diseases and knowledge of aspects related to a citizen’s “character and personality” entail dangers or potential drawbacks with respect to safeguarding personal privacy. In other words, the advances resulting from human genome research will bring new expectations to humanity concerning the prevention, treatment or cure of diseases. However, they may also be a source of danger to fundamental individual rights such as those concerning freedom –right to self-determination (consent)– and privacy. In addition, information on the risks of developing certain diseases affects both the individuals directly involved and their family or certain groups of individuals related to that person.

As Dr. Estivill has stated, the law, or rather legislation, must lay the legal foundation needed to handle this fast-approaching scenario (less than ten years away) with the foreseeable advances in the study of the human genome

and with the contributions –benefits and risks– of predictive medicine. Legislation will be needed to avoid “genetic discrimination”, guarantee the “privacy” of genetic information and respect the individual’s “right to self-determination”.

c) Lastly, Dr. Estivill’s points about the difficulties of establishing the reliability of the “predictive capacity” of genetic tests depending on the type of disease and genetic test, should be highlighted. The development of diseases depends not only on genetic factors, but also cultural and environmental factors. As mentioned by Dr. Estivill, we must admit that our knowledge about the “causes of disease” is still extremely limited. Valuable information on individual genetic defects can be obtained at present. However, as Prof. Jens Reich, a molecular biology expert at the Max Delbrück Centre in Berlin pointed out recently, some time must elapse before it will be possible to cure or prevent these defects and therefore, many more diagnoses will be made without any potential therapy in sight.

The progressive and impressive advance of biomedical investigation is undeniable, as evidenced by the presentation of the map of the human genome on June 26 2000, the so-called Book of Life, in which 97% of the human genome is deciphered. However, attention must be drawn to the paucity of knowledge regarding the results that may be produced from the application of data from the human genome. In any case, predictive medicine –with all its advantages and disadvantages– that allows some prediction of what diseases a person is susceptible to develop, is destined to become the medicine of the 21st century, as foreseen in 1993 by Jean Dausset, the French doctor who won the 1980 Nobel Prize in Medicine. In this new historic context, the field of law –in co-ordination with the scientific and international community– will be required to offer adequate solutions to ensure freedom in scientific research and prevent genetic information from becoming a discriminatory tool, preserving individual human dignity, privacy and the right to self-determination at all times.

2. As to the brilliant speech given by my colleague, Dr. Mirentxu Corcoy, I would like to underscore several issues, adding an occasional personal comment in line with the debate.

I agree with the speaker about the need to listen to the scientific community’s opinion on making decisions related to this new, complex field of

predictive medicine. Nevertheless, our understanding of the adverse consequences of research in this field is still very limited. We should take a closer look at this worrisome trend of negating the adverse effects of genetic research techniques which, in any case, would be justified by the inherent value of scientific research. In this final aspect, the European Parliament's Resolution on "the mandate of the Group of Advisors from the European Commission entrusted with analysing the ethical aspects of biotechnology" of 13 June 1997 (Official Journal of the European Communities, 30 June 1997) has stated that "until now the interests of research have received much more attention than the possible consequences in society".

2.1. With regard to limitations on genetic research and the necessary original dependence of legal experts with respect to scientific advances, the first major filter consists in the basic ethical-social considerations of modern society, among them the inevitable respect for human dignity laid down in Art. 10.1 of the 1978 Spanish Constitution, and in the need for the scientific community specialising in this field to draft a "Code of Ethics" for self-monitoring of its research activities (see below European Parliament Resolution of 20 Sep 1996). I would like to draw your attention to one of the points of discussion that came up in the debate: the lack of linguistic co-ordination in the communication between physicians and legal experts. Use of a "common language" has been proposed to facilitate the communication and collaboration needed between genetics and legal scholars. In my opinion, this suggestion should be qualified and reviewed in the following terms: on the one hand, it must be remembered that scientific language has been coined by the medical and legal sciences and this most certainly means that we cannot renounce it on behalf of a "common language". And, although we accept this, we must acknowledge the need for reciprocal interchange and familiarisation of terminology between genetics and legal specialists so as to allow adequate collaboration. Moreover, this should be done without relinquishing the scientific rigor of our own language or neglecting the distinct nature of these disciplines (experimental in the case of genetics and evaluative in the case of law).

2.2. Limits must be set on biotechnology applications, but this control is only effective if there is international agreement, as Dr. Corcoy mentions. In contrast with certain positions expressed in this discussion that are contrary to international declarations of generic bans, this approach is correct in my opinion for two reasons: first, because these declarations allow us to create

international opinion and consensus on basic issues related to human beings and the future of humanity; second, because the signing, ratification and implementation of international conventions on the issue will condition the internal legislation of the countries making this type of commitment.

2.3. From a legal standpoint, the solutions provided by predictive medicine should be based on respect for the individual's fundamental rights. The perspectives of the constitutions and international law, and particularly the principle of individual autonomy or the right to self-determination, as well as the right to (genetic) privacy, must be taken into account at this point. I would simply like to underscore the most relevant aspects and add a brief point related to the issues raised by Dr. Corcoy's speech.

2.3.1. The starting point and basis for any legal assessment of human genetics must be the constitution (in our case the 1978 Spanish Constitution), as mentioned by Albin Eser (ADPCP 1985, p. 350). Art. 20.1.b of the Spanish Constitution is particularly relevant, as it acknowledges and protects "the right to scientific production and creation" as a fundamental right. The legitimacy of scientific research (including research into human genetics) has limits and cannot be allowed when it infringes other fundamental rights, such as those related to life and health –physical and moral integrity– (Art. 15 of the Spanish Constitution) and personal privacy (Art. 18 of the Spanish Constitution) (*Cf.* Art. 20.4 of the Spanish Constitution). Moreover, the supraindividual outlook of scientific research, particularly in the use of genetic techniques, also has limits in the general interest of society (Art. 44.2 of the Spanish Constitution). Lastly, within this constitutional perspective we must underscore the importance and the key role of "human dignity", which is basic to political order and social peace (Art. 10.1 of the Spanish Constitution).

2.3.2. International law includes the resolutions passed by the European Parliament as well as conventions signed and ratified by various countries.

Here we can mention certain resolutions of the European Parliament and the important 1997 Convention of Oviedo:

a) The European Parliament resolution of 16 March 1989 "on the ethical and legal problems of genetic engineering" (Official Journal of the European Communities of 17 April 1989) acknowledges the risks potentially derived from the analysis of the genome, such as using genetic testing as a tool for social control, and emphasises the relevance of "the principle of freedom of

science and research” and the restraints imposed by the rights of third parties and of society, without neglecting the key role of human dignity. The analysis of the human genome must be exclusively for the well-being of the person concerned and be based on his or her voluntary agreement, with the priority being the principle of self-determination. The possibility of weeding out employees according to genetic criteria is rejected and, in an attempt to prevent discrimination, insurance companies are denied the right to require genetic analyses when providing insurance contracts.”

b) The European Parliament’s resolution of 20 September 1996 on “the protection of human rights and dignity of the human being with regard to the application of biology and medicine” (Official Journal of the European Communities of 28 October 1996) considers it essential to establish ethical standards for biology, biotechnology and medicine based on respect for human dignity. The ban on “any transmission of the results of genetic testing to other individuals or institutions –such as insurance companies or firms– except in the event of legal requirement” is described as fundamental. In addition, it states that “the performance of tests aimed at predicting the appearance of genetic diseases or that indicate a predisposition to a certain disease or a disability may only be authorised in severe cases when an effective treatment can be taken...”.

c) The Convention on “the rights of humans and of biomedicine” (Oviedo, 4 Apr 1997, ratified by Spain on 1 Sep 1997 and effective 1 Jan 2000) includes several significant general and specific points about the topic we are discussing.

On the one hand, it states generally that the “interests and welfare of the human being shall prevail over the sole interest of society or science” (Art. 2); “intervention in the health field may only be carried out after the person concerned has given free and informed consent to it” (Art. 5.1); and that “everyone has the right to respect for private life in relation to information about his or her health” (Art. 10.1).

In addition, Art. 12 refers expressly to “tests which are predictive of genetic diseases” and states that “tests which are predictive of genetic diseases or which serve either to identify the subject as a carrier of a gene responsible for a disease or to detect a genetic predisposition or susceptibility to a disease may be performed only for health purposes or for scientific research linked to health purposes, and are subject to appropriate genetic counselling”.

2.3.3. As part of the fundamental legal problems that may arise from the performance of predictive tests –apart from those already mentioned in reference to discrimination in the labour world and in obtaining insurance– I would like to comment on certain aspects related to the basic issues of the right to freedom of self-determination (principle of autonomy) and the right to personal privacy, which could be directly and seriously affected by the practice of predictive genetic testing.

2.3.3.1 Any medical intervention in the context of health, and any intervention related with predictive tests, must have the approval or consent of the person concerned, when he or she is competent. This means that a preliminary, essential requirement for performing predictive tests is the freely expressed, informed consent of the individual concerned, provided he or she is capable of giving it. In the event that the individual concerned is a minor or is incompetent, and does not have sufficient capacity to understand and natural discernment to decide on the medical intervention to be performed, his or her legal representative(s) or close family members must give their consent, acting in the benefit or interest of the person they represent. The consent may be revoked freely at any time prior to the performance of the medical procedure.

Art. 10.5 of Spain's General Healthcare Act 14/1986 (LGS) of 25 April requires "prior written consent by the user for the performance of any surgery, except in the following cases: a) when failure to operate represents a public health risk; b) when the patient is incapable of making decisions, in which case the right pertains to the patient's relatives or close family members; and c) when the urgency of the condition does not allow delay because of the possibility of irreversible lesions or a life-threatening situation."

The doctrine has pointed out the shortcomings of this provision of the LGS. On the one hand, it underscores the disadvantages of requiring prior written consent for any intervention, as strict compliance with this requirement (with all the consequences thereof) would make it a mere bureaucratic formality, as if the idea were to mechanically complete a form (see in this regard, Cobreros Mendazona). The most recent health regulations state with a more effective line that "consent must be given in writing in the cases of surgeries, invasive diagnostic procedures and, in general, when procedures are performed involving obvious, predictable risks and disadvantages that could have repercussions on the patient's health (Art. 6.2 of Law 21/2000, of 29 December, of the Catalan Parliament relating to "the rights to information concerning the

patient's health and autonomy, and the clinical documentation" (Official Spanish Gazette of 2 Feb 2001). The performance of predictive tests involving a diagnosis shall require prior consent of the individual concerned in writing. Nevertheless, there has been criticism (by Bajo Fernández and Díez Ripollés, among others) that the General Healthcare Act does not discuss "the right to oppose medical treatment" with enough clarity (*cf.* Arts. 10.9 and 11.4 of the General Healthcare Act), nor is there any reference to the conditions needed for the consent to be valid, such as a lack of malice, deceit, error and coercion on the part of the patient, or to the conditions under which the legal representatives or close family members may give consent.

The validity of the consent of the person subject to any intervention in the context of healthcare requires that the person concerned be previously informed (expressly required by Art. 6.1 of Catalan Law 21/2000, of 29 December), i.e., informed consent is required. This refers to the patient's right to be aware of the medical treatment to which he or she consents, hence the physician's duty to provide the information. Informed consent is required to perform predictive tests and it is to be granted by the person concerned, or his or her legal representatives or close family in the case of minors or incompetent individuals.

In terms of the conditions involved in obtaining informed consent, the following must be considered:

1) In principle, the physician responsible for the patient must ensure the patient's right to receive information (see Art. 10.7 of the General Healthcare Act; this gives the physician assigned to the user the role of "liaison" with the healthcare team. However, the healthcare professionals attending the patient or applying a specific technique or procedure must also assume the responsibility for providing information (see also, Art. 2.3 of Catalan Law 21/2000, of 29 December).

2) As to the form of informed consent, Art. 10.5 of the LGS requires that the information be provided "in understandable language, that is complete and continuous, verbal and written" (*cf.* Art. 2.2 of Catalan Law 21/2000, of 29 December). Basic formal informed consent is preferably verbal. Only in certain cases of risk pertinent to the patient's life or health –surgeries such as a coronary bypass or examinations such as catheterisation involving some risk– is written consent considered essential (see Art. 6.2 of Catalan Law 21/2000, of

29 December). What will happen with the necessary form for free, informed consent when performing predictive tests? Based on the special nature of predictive analyses, their enormous relevance for the individual concerned and for third parties, and the dubious effectiveness of the diagnostic procedure and the therapeutic approach to be taken, we must develop careful processes for information and consent and make use of the written word. Undoubtedly, the requisite of written informed consent is unchallengeable if the predictive tests involve the collection of tissues from the individual concerned, as this would imply a surgical procedure.

There should be specific documents on informed consent for predictive tests (*cf.* proposal on informed consent by the Consejo Interterritorial, 6 Nov 1995: 1.5.5°), which include a series of sections such as the following: who provides the information and who receives it; the nature, risks and consequences of predictive tests; the possibilities of effective treatment of the disease being prevented or cured and, therefore, the indication for performing the predictive analysis; protective measures and assurance of confidentiality of genetic data; the possibility of revocation or relinquishment and the ability to exercise the right not to know...

3) Limits and exceptions to the obligation to inform. Medical information must discuss “the process, including the diagnosis, prognosis and treatment alternatives” (see Art. 10.5 LGS). The problem lies in setting the limits and defining any possible exceptions with respect to the physician’s duty to inform. Certain guidelines or basic criteria can be mentioned in terms of limits. One is the need to inform of typical or predictable risks associated with the medical intervention to be performed, with this duty not including atypical risks. Another is the principle of “therapeutic privilege” in English or “assistance” in German, with this being extremely relevant for information on the diagnosis and limiting the information when it is contraindicated for therapeutic reasons. Finally, the precision and extent of the information must be inversely proportional to the urgency of the medical intervention; according to this rule information on predictive tests must be detailed and comprehensive. The following exceptions to the duty to inform must be taken into account: emergency cases where the patient cannot be informed and his or her life or health is endangered; when the patient is incompetent and his or her legal representatives or close family members cannot be informed; when the patient waives the right to receive information, highlighting the patient’s “right not to know” in predictive tests

based on the refusal of the person concerned to learn about his or her own genetic data and the physician's obligation to respect this decision.

Possible conflicts between the patient's right to self-determination and the rights to life or health of third parties should be resolved, particularly in view of the person's need where the principle of weighing assets, the criterion of proportionality, and constitutional considerations play important roles, with the latter including the impassable limit on human dignity that –according to Art. 10.1 of the Spanish Constitution– is the basis of our political order and social peace.

A medical procedure (e.g., predictive test) that is carried out against the will or without the valid consent of the person concerned would be an attack against personal freedom and could be qualified –according to a major area of Spanish penal doctrine– as a crime of coercion (Art. 172 of the Penal Code, which states that “Whoever, without being legitimately authorised to do so, prevents another by violence from doing what the law does not prohibit, or compels him or her to do what he or she does not want to do, whether just or unjust...”). Nevertheless, it will be very difficult –if not impossible– to include certain arbitrary medical therapies in Art. 172 of the Penal Code, e.g., medical procedures carried out with non-effective consent from the patient because of a lack of sufficient information. Hence, express regulation in the Penal Code of a new criminal offence is perhaps warranted to handle these cases of arbitrary curative medical treatments as crimes against freedom along the lines of the Penal Codes of Austria (paragraph 110) and Portugal (Art. 156).

2.3.3.2. The performance of predictive tests may involve a risk for the fundamental right to personal privacy recognised in Art. 18.1 of the Spanish Constitution. Respect for this right will be one of the limits to the exercise of the right to freedom enjoyed by scientific research (*see* Art. 18.4 of the Spanish Constitution).

Personal data relating to health, such as data from predictive tests, pertain to the “particularly sensitive essential core of privacy” and require special protection (*see* Arts. 4ff. of the Organic Law 15/1999, of 13 December relating to the protection of data of a personal nature).

The performance of predictive tests can lead to an intolerable, illegitimate interference in the personal privacy of the person concerned, when there is disclosure of his or her genetic data without consent. The principle of

autonomy of will is fundamental in this regard, and the express consent of the person concerned must be given in order to obtain access to genetic data pertaining only to him or her, unless set forth by law (see Arts. 6.1, 7.3 and 7.6, and 11.2f of the Organic Law 15/1999 of 13 December for general aspects on data processing related to health). In any case, it is also necessary to guarantee the secrecy –hence, the physician’s duty to maintain confidentiality– of the results of genetic tests. However, no specific regulation (law) on genetic databases yet exists (Dr. Carracedo has already made this valid point). A mere reference point or collective guideline could be the provisions of regulations on the protection of automated data of a personal natural, with the protective measures enhanced in the case of custody of genetic information (see Romeo Casabona, “Aspectos específicos de la información en relación con los análisis genéticos y con las enfermedades transmisibles”, in *AAVV Información y Documentación Clínica*, Vol. I, Consejo General de Poder Judicial and Ministry of Health and Consumer Affairs, Madrid 1997, p. 355 on the situation in Spanish hospitals in recent years with respect to the protection of personal data, and condemning their serious shortcomings in light of the reflections made by the 1995 and 1996 Reports issued by the Data Protection Agency, see Lucas Murillo de la Cueva, P. “El tratamiento jurídico de los documentos y registros sanitarios informatizados y no informatizados”, in *AAVV Información y Documentación Clínica*, vol. II, 1997, pp. 591 and 592).

Personal privacy and therefore “genetic privacy” is unique in that it affects both the person concerned and third parties –family members– and in that it has a public facet –data permitting public access. As a result, it can be legally protected by the aforesaid Organic Law 15/1999 of 13 December, as well as by civil law (Organic Law 1/1982 of 5 May on the civil protection of the right to honour, personal and family privacy and self-image) and by penal law (infringement of the ban on disclosing and exposing secrets: Arts. 197ff., particularly Arts. 197.2 and 197.5, Art. 199 on professional secrecy, Art. 198 –if the public authority or official, outside of cases allowed by law and availing him or herself of his or her office, were to incur in any conduct set forth in Art. 197–, as well as Art. 417 –the public authority or official were to reveal secrets or information to which they are privy by virtue of their profession or position and which must not be disclosed– from the Penal Code. These phrases highlight the fact that physicians who practice medicine at public healthcare centres are considered public civil servants for the purposes of the Penal Code –Art. 24.2 of the Penal Code–).

3. In conclusion, predictive medicine broadens the horizon of medical research in the field of biomedicine. This has advantages, such as the prevention of disease and the possibility of curing genetic disease, as well as disadvantages, such as serious risks for the patient's fundamental rights of self-determination and privacy, and potential discriminatory situations. At present predictive tests function within certain margins of probability when issuing a diagnosis and scientific knowledge is very limited regarding effective medical therapies to prevent or cure the diseases that can be detected. As in everything related to the advances of modern biotechnology, here we must follow the standards of a reasonable, gradual legislative policy along the lines laid out by Albin Eser (*Anuario de Derecho Penal y Ciencias Penales* 1985, pp. 363 and 364). In other words, we must use the moral self-monitoring of the scientific community's code of ethics and move toward the establishment of governmental guarantees for procedures (e.g., legislation relating to genetic data banks, specific regulations on informed consent for predictive tests) in Spanish hospital praxis, eventually introducing –if necessary on the basis of current legislation– civil regulations or penal bans. Since the Spanish regulations are obsolete in light of the new challenges of predictive medicine and biotechnology in general, the Spanish Committee for Ethics in Scientific Research and Technology should take steps to promote the adoption of appropriate measures which, while ensuring that scientific research exercises its right to freedom, will also ensure the patient's fundamental rights to self-determination and personal privacy by preventing discrimination on the basis of his or her genetic heredity.

José María Goerlich

Professor of Labour Law, Universidad de Valencia.

From my perspective as a labour fraud expert, the problems posed by predictive medicine are by no means new. Rather they are a manifestation of a classic problem: namely, potential discrimination of workers by their employers. The extraordinary possibilities available in the future as scientific knowledge progresses will increase the possibility that this risk and the causes that explain it will be fulfilled. Not only will personal or social circumstances or the exercise of public rights or freedoms be taken into consideration as occurs now, but abundant information obtained from genome analysis on future pathologies, predispositions or tendencies will also become available. It is no less certain, however, that since the greatest possibilities of discrimination

are only a question of degree, it is precisely in the area of anti-discriminatory protection where the solutions and remedies will be found.

Anti-discriminatory protection in labour relations has progressed dramatically in the last twenty years, mainly due to the jurisprudence of the Constitutional Court. The jurisprudence is reasonably mature in relation to discrimination during employment but is more limited in terms of discrimination occurring during the hiring process. In both areas (particularly the latter), an essential guarantee of the principle of non-discrimination undoubtedly affecting the issue at hand consists in limiting the possibility that the employer can gain access to sensitive data and information on the employee. This involves preserving the employee's privacy and particularly, protecting him or her from discrimination by preventing others from learning about facets of his or her personality that could lead to discrimination. Consequently, since the employer has been prevented until now from asking about or investigating in depth the employee's opinions on unions, ideologies or religions at the time of hiring, we should start thinking that this type of restriction also affects genetic information and related investigations.

There are significant differences between the two, however. In fact, there are legal provisions that allow the employer to require the employee to undergo medical examinations, but no regulation that expressly allows the employer to make enquiries about the employee's ideology. The legislation relating to the prevention of occupational risks (Art. 22 of the Occupational Risk Prevention Act) outlines possible cases where the employee must undergo such testing to be hired, and also afterwards, during the employment relationship. Although this is logical since medical monitoring is an essential part of any preventive policy, it paves the way for genetic information on the employee to reach the company, with the latter using it in ways outside of this purpose, ways that may be illegal. This might be to discriminate against the worker on the basis of data unrelated to the job or to assess the fact that the worker could develop a particular condition in the future, either alone or partially due to the work environment, making the employee unsuitable for the job.

This risk is well-recognised and has been mentioned in the speeches. Nevertheless, it must be prevented. In my opinion, we should emphasise the following ideas which, once developed in regulations and/or interpretations, could avert this situation:

1. First of all, medical examinations are essential to any preventive policy, as only adequate medical monitoring makes it possible to identify existing risks and evaluate the effectiveness of measures against them. However, the tests are clearly much more ambitious and their priorities are legally established (Art. 15 of the Occupational Risk Prevention Act). The principles of preventive action do not allow elimination of the risk by removal of the worker posing the risk when he or she comes into contact with the potentially adverse situation. Our first and main goal should be to prevent this at the root. On this basis, we could discuss the possibility of declaring an employee unsuitable because of a potential incompatibility between his or her genetic makeup and the work environment, but only when the possibilities of making the environment innocuous have not been exhausted.

2. Secondly, we must be extremely careful about the possibility of obtaining health information in general and genetic information in particular about the employee. Art. 22 of the Occupational Risk Prevention Act does allow medical examinations to be required. But this must be interpreted fairly. Generally speaking, the employee voluntarily submits to medical monitoring, and any exceptions must be interpreted strictly and restrictively, as occurs when hazards exist for the health of the employee (adequately informed) that he or she has, at least up to a certain point. Moreover, consultation with the employees' labour representatives is needed for compulsory examinations, and these representatives should provide the employee with information.

Moving to another point, health monitoring is instrumental for one purpose. Art. 22.1 of the Occupational Risk Prevention Act subordinates it to the function of assessing work-related risks. General health investigations are not allowed, and only examinations performed for this purpose may be undertaken. Based on this idea, the aforesaid precept lays down a principle of minimum understanding of the employee's freedom, establishing the proportionality of acknowledgement of risks existing in the workplace.

Lastly, the resulting information is not available for free use. Art. 22.4 of this act is extremely important, as it prevents the information from being used for discriminatory purposes and only allows it to be disclosed to the worker and the public authorities. Neither the employer nor the workers' representatives have access to the information unless the worker consents, and only the final information concerning whether the worker is apt or not or concerning the need to improve preventive measures will be obtained, in order for them to comply with their responsibilities.

3. In short, the professionals who devise the preventive measures and handle the medical monitoring play an essential role. I am referring to the company's medical services which are entrusted by law with these responsibilities (Art. 22.6 of the Occupational Risk Prevention Act) and therefore, are in charge of defining the type of examinations the workers must undergo and of assessing the information obtained.

In one of the speeches these professionals were not fairly viewed and were compared with staff serving private interests (e.g., that of insurance companies), and were excluded from the possibility of prescribing genetic analyses. This is probably necessary. But I feel we should be fair to the company medical staff. In my opinion, in view of the protection against the reprisal they usually receive (see Art. 30.4 of the Occupational Risk Prevention Act), they are not subordinate to private interests but can act impartially in the interest of health within the company.

In this context, the existing restrictions may be related to their potential lack of training, in both scientific and ethical terms, on the use of genetic testing. The solution does not lie, in my opinion, in excluding them from their practice but in including these disciplines in their training, so they are aware of the ethical and scientific issues involved (reliability, etc.). We must not forget that, although extremely worrisome from the perspective of worker discrimination, they may be immensely useful in the area of preventive action.

Carlos María Romeo Casabona

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In my opinion, the innovation of predictive medicine as a subject of interest for law lies in the information it provides. This information has several characteristics that differentiate it from other kinds of information: it refers to a field of considerable importance for humans, namely, health and other biological characteristics (e.g., ethnic features); it is personal and individual, but also related to the family (biological family); it is not generated by the individual and does not depend on his or her will; it is permanent in the sense that it cannot, in principle, be changed; and, since it is predictive, it also concerns the future in terms of events that will occur unless the person interferes (e.g., to avoid or prevent the manifestation of a disease) or that will probably occur if human behaviour plays any role (e.g., with lifestyle, diet, etc.).

Consequently, law must intervene to establish certain guarantees in relation to information in which the individuals are extraordinarily vulnerable. At this time, this is the aspect related to the advance of genetic knowledge that constitutes the greatest, most urgent challenge for law.

First of all (although this does not appear to be innovative for law and is only up to a certain point), free, informed consent of the person involved must be guaranteed for any genetic test, except when urgent circumstances weighing on behalf of the individual preclude the possibility of first obtaining consent. Consequently, the prohibition of compulsory imposition (much less coercive) in benefit of any public or private interest must be established. Exceptions to the obligation of samples being taken for obtaining non-coded DNA profiles in a criminal procedure are allowed, in accordance with the provisions of law.

The information provided to the person concerned before he or she consents to undergo any genetic testing must be complete, in particular on the nature of the analyses and the results (whether expected or unexpected) that may be obtained. Likewise, respect for the individual's refusal to receive information about his or her own genetic data (right "not to know" or not to be informed) must be guaranteed, although also with certain limits.

In my opinion, genetic analyses should be allowed when other diagnostic health tests are imposed by law, to the extent that they are more reliable for the legally established objective and do not provide any other kind of information, particularly predictive information. The law may establish exceptions, but only if there are proven interests of a general nature, e.g., reasons relevant to public health or safety –or of clearly prevailing third parties.

In any case, the secrecy of genetic test results must be guaranteed. In addition, restrictions on access to these results must be established, even regarding the person's family members, notwithstanding the solutions provided by the law in the event of any conflict of interest, even those that could be qualified as a clash of responsibilities. Genetic information must be protected by using similar guidelines and measures (although enhanced) to the ones laid down for data of a personal nature (data quality: pertinence, accuracy, purpose, cancellation, right of access and correction by the party concerned, etc.; physical and logical security, splitting of data to allow partial, selective access, etc.). Moreover, this should be done regardless of whether the data have been obtained to fulfil the law (if required) or with the consent of the

interested party. The responsibility to maintain secrecy must also extend to anyone whose job or duties imply access to this information.

In my opinion, the resulting information should also be used only for the originally intended purpose or purposes. For this reason, more restrictive specific measures must be provided for the transfer of genetic data to other files and for the cross-border flow of these data, as current legislation on the automated processing of data of a personal nature do not generally provide sufficient protection.

The results of genetic tests performed for health reasons must not lead to discriminatory practices or interference in private life. In addition, access to appropriate healthcare or social services should not be conditioned and reproductive restrictions should not be implemented (e.g., abortion, sterilisation) except for the conditions established by law for access to assisted reproduction techniques.

Juan Antonio Segarra

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The brilliant speeches made by Professor Xavier Estivill and Professor Mirentxu Corcoy in the meeting held raised a number of issues for those who have heard or are familiar with them.

Due to the short time available, I would like to make only a brief comment on one of the many aspects raised by Dr. Corcoy in her study.

Recent scientific advances in the area of what we have called “predictive medicine” are so vast that they entail obvious risks for citizens as a whole, as well as countless advantages.

These risks will not halt scientific progress which, in principle, should have no other limit than that of the ethical postulates widely recognised as absolutely essential by the scientific community. This does not necessarily mean that the legislator, the courts and all legal players in general must not be particularly sensitive about the implementation of these advances to ensure that indiscriminate use thereof cannot injure the fundamental rights of individuals.

The fact that these advances could be useful for all mankind would imply that they should be used in the same manner for everyone. Although international

texts relating to human rights do exist, there are considerable differences in the interpretation and application of these within each country. For this reason each territorial entity must find a way to assure essential safeguards against the improper use of the scientific knowledge that “predictive medicine” offers us until a paradigm of legislative harmonisation in this field is reached.

In Spain, the Constitution acknowledges a set of fundamental rights and freedoms which are founded on human dignity, including the right to life and health. Many conclusions can be drawn as a result. However, we agree with Dr. Corcoy’s opinion that the sacredness of the right to freedom, or better yet the person’s own dignity, requires that what the speaker called “analysis” or any other predictive practice be preceded by informed consent as broad, detailed and personalised as possible for the target individual. This is already required in the diagnostic and/or treatment context within conventional medicine. In fact, some laws state that failure to obtain informed consent is an offence against freedom. This should be stressed even more, if possible, in the case of predictive techniques, in which their early stage of development means that a diagnosis is frequently obtained, but unfortunately, there is no satisfactory treatment in sight. Thus, consent given by adults must be accompanied by the utmost guarantee concerning the information, and this includes the information that many of the findings may lack an adequate treatment or that there may be no treatment at all at the present.

Since diagnosis and/or treatment in this field can affect the scope of the individual’s self-determination, in the case of minors and/or incompetent persons there should not be an automatic mechanism for granting consent by the parents or guardians of these individuals.

Along these lines and given the inherent risks of the field, we do not feel that it warrants the same application as that set forth in Art. 10 of the Spanish General Healthcare Act for conventional medicine. To the extent that the minor is naturally capable, he or she should be heard. Moreover, additional guarantees by some official body, authorisation from the Minors’ Office, disability courts, etc. or even Ethics Committees in hospitals or other healthcare centres should be provided in order to restrain the use of these practices to strictly necessary situations where they are intended to improve the minor’s health, thereby avoiding abuse by parents aimed at identifying what we could call a “transparent child”.

The essential core of human dignity must encompass the possibility that the person refuse part or all of the information of his or her genetic map, without this necessarily representing any hindrance to access to certain legal relationships, even governed by private law, because of the discrimination that this might imply.

In short, this is how discrimination can be prevented. In our opinion, the possibility to hold a job in the public or private sector, obtain a mortgage or sign an insurance policy –to mention only a few of the examples cited by the speaker– cannot be indiscriminately linked to compulsory genetic screening.

Since a conclusion of this nature may seem debatable, particularly in relationships governed by private law, we feel that only a constitutional reading of these relationships, and how they are affected by fundamental rights to dignity, freedom and even health, will lead to this thesis. In effect, and only mentioning a few examples, it is unacceptable that in a social, democratic state, a health insurance policy can only be obtained after undergoing all existing predictive practices in fulfilment of requirements laid down by the insurer. It would be different if a possibility for the individual to obtain basic healthcare from a public, compulsory (not necessarily free) system already existed. In this case, the requirement would not be a problem, as the individual would still be freely able to exercise his or her right to self-determination.

Prehiring medical examinations also pose a specific problem, as there are wide discrepancies among the legislations enacted in the various countries.

Although the employer is, to some extent, the guarantor of the employee's health, he or she cannot require that the potential employee undergo a battery of "predictive medicine" tests and link the final results thereof to hiring. In this field (as in so many others where the fundamental rights of the parties must be weighed), the practice of medical examination must be restricted to tests related to specific, immediate risks connected with the job. Any limit on hiring because of a mere diagnosis that the individual will predictably develop a disease not directly related to the tasks of the job is legally reproachable.

Lastly, in terms of the essential content of the right to privacy, the guarantees on the confidentiality of data obtained from the tests must be enhanced, even if consent was given by the person concerned. As a result, we feel that disclosure of these data must be extremely well-protected. The regulation in the 1995 Penal Code for the disclosure of health data without consent appears plausible.

Although consent may be granted for one or more tests, this does not mean the data can be used (even anonymously) for any other ends, including scientific purposes, unless express consent is given in this regard.

Ana Victoria Sánchez Urrutia

Professor of Constitutional Law, Universitat de Barcelona.

In my opinion, our concern that the development of predictive medicine and genetic diagnosis techniques could lead to discrimination on the basis of genetic characteristics is warranted. In fact, discrimination simply means *treating a person differently without justifiable cause*. The ignorance and malice of individuals with decision-making power can result in unfair, arbitrary, discriminatory treatment.

Genetic analyses offer numerous advantages, as they allow personalised medical therapies. Once the genetic mechanisms behind a disease have been discovered, research can be performed to design drugs that work directly on the pathology without adverse effects for the patient. Nevertheless, poor use of genetic information could lead to the labelling of individuals and cause discrimination, understood to be unjustified different treatment of an individual. A report issued by the Labor Department of the U.S. federal government mentions, for example, discrimination against employees of African origin who are carriers of the genetic defect for sickle-cell anaemia. In this case, the American report states that there may have been hidden racial discrimination.

For this reason, I concur with the cautionary and preventive measures mentioned by Dr. Corcoy in her speech. Genetic information on a person can affect his or her personal and family privacy, since the results of an individual's genetic diagnosis is also pertinent to his or her biological family. In terms of medical data, particularly on the genome and other information that can be generally derived from the DNA, the right to privacy concerns the protection of family privacy in most cases.

Genetic privacy normally concerns confidentiality, secrecy or respect for the privacy of information. Nevertheless, as the circumstances change and genetic testing possibilities develop, genetic privacy is cited in terms of an individual's right to control his or her personal data, i.e., once they are generated, the right to determine what genetic data on a person can be kept, and what data he or she wants known and the right of third parties affected by

an individual genetic test to know the results. The legitimacy of public officials or individuals to store genetic information is also questioned.

Genetic information can be stored in one of two ways: as the results of a genetic test, in which computer analysis of the sample is stored, or as potential genetic samples; that is, tissue samples which could be used for genetic testing.

There is no specific regulation on the use of genetic data and therefore, the protection of medical information must be enforced until special regulation on this type of information exists. In the final analysis, genetic information is actually medical information, irrespective on whether it is related to healthcare or personal identification. Certain additional precautions can be taken with genetic data as indicated by international jurisprudence, treaties and recommendations:

The Universal Declaration on the Human Genome and on Human Rights states that any “research, treatment or diagnosis affecting an individual’s genome shall be undertaken only after rigorous prior assessment of the potential risks and benefits pertaining thereto”. According to the declaration, “In all cases, the prior, free and informed consent of the person concerned shall be obtained.”

The Council of Europe recommends that any genetic or screening test procedure must be accompanied by appropriate consulting, both before and after the procedures. The counselling must be of a nondirective nature: the information must include pertinent medical facts, the test results and the consequences and options. The object and nature of the tests must be explained, and any risks must be mentioned. The information must be adapted to the circumstances under which the individuals and families receive the genetic information. When necessary, every effort must be made to provide continued support to the individuals who have been tested.

Health legislation lays down the obligation to perform epidemiological control of certain diseases, designated as requiring compulsory reporting. In this context, healthcare personnel are required to report the diagnosis of these diseases to the health officials having jurisdiction. The reporting may require numbers or names, or urgent completion. In the latter two cases, the diagnosis must be accompanied by the particulars of the patient. The health authorities of the state or autonomous community handling personal data obtained from the diagnosis of these diseases requiring compulsory reporting can transfer these data for epidemiological studies, statistical processing and scientific research. Both the government and the ones receiving the data must protect

them with the extreme security measures set forth in the regulation on the safety of personal data. The processing of genetic data is justified in this context. Nevertheless, despite the fact that epidemiological studies exist on genetic diseases, normally these diseases are not expressly named in the decrees of the autonomous communities that list diseases requiring compulsory reporting. Moreover, it is surprising that only one file on genetic disease studies is recorded in the Data Protection Agency. Hence, in most cases, the creation of data banks on genetic diseases and the storage of tissues for subsequent study take place outside the law and therefore, lack any legal guarantees.

Opinion of the insurers

Màrius Berenguer

General Director of CaiFor (Insurance Group of “la Caixa”).

Advances in genetic research –the possibility to predict with more or less accuracy the probability that a person will develop a particular disease, and society’s use of this information– opens many doors; most are positive (possible identification, prevention and cure of hereditary diseases), but some are negative, including potential discrimination resulting from their use.

In the case of insurance, particularly life insurance, a clear understanding of the technical principles and bases is needed in order to clear up certain concepts and foster a richer, more objective discussion on the actual and potential effects of this information.

1. Introduction: Risk and insurance activity

The insurance business is based on combining risk situations as a whole, in order to allow compensation or mutual protection, in which many individuals pay relatively small amounts so that a few (those affected by an adverse event) receive significant amounts.

Thanks to a combined treatment of homogenous risks explained mathematically and represented by statistical laws, the insurance industry can provide previsionary mechanisms that offer solutions in terms of indemnity or lump sum payments in the case of an adverse event.

In practice, this means that no problem is solved and no situation is resolved by an individual simply knowing that his or her probability of dying next year is 0.6%, i.e., that six of every one thousand individuals of that age will die. An insurer who has insured one thousand individuals in the same age bracket does find this useful, because it knows that there will be about six deaths. On the basis of these data, it can define the respective premium for the amount of the policy; for example, a capital of 10,000,000 pesetas would have a premium of 60,000 pesetas per year.

This compensation or financing system by sharing the risk is the foundational principal of insurance and is supported by the mathematical and statistical theories we mentioned earlier on a general basis: the larger the

population set exposed to risk, the greater the probability that the projected population outcome will be met.

Aggregate treatment is what distinguishes insurance from a simple game of chance. Although both share a stochastic behaviour of the variables, the aggregate treatment of a set of risks is the rationale behind the insurance business. This approach is used to set insurance premiums, with the premium encompassing the statistical experience plus a safety margin and representing the expectation that the statistics applied to the population will be fulfilled. The determining factors for the safety margin parameters are, among others, the total number of policyholders and the reliability of the contract process.

The influence of the number of policyholders is obvious; the actual behaviour of the insured variable approaches the initial projections as the number of elements increases. In contrast, fewer policyholders means that chance is more likely to skew the expected results and that the actual situation will vary considerably. The same thing occurs during the contract process, which is intended to measure the insurability of the risk and then to set the price of the risk being insured, based on available information.

Obviously, the term “risk” refers to the possibility that something might occur, not to something that has already occurred or will undoubtedly occur. In the latter case, the event cannot be insured, or if it were, fraud or malice would exist on the part of the person or entity attempting to pass off a past event as a simple risk.

Transfer of risks; the extent of the variables

In other types of financial or business transactions, the proportion between goods or services provided and the payment is around five or ten per cent, in the form of the interest rate or profit margin.

In insurance, the difference between the price and the benefit is enormously disproportionate in comparison with other businesses, since the premium is 60,000 pesetas in our example and the capital paid out is 10,000,000 pesetas.

Naturally this is the result of the low probability that the insured adverse event will occur, but this also means that the contract process and premium calculation must be rather meticulous since an error in a transaction would only be offset by hundreds of transactions.

In our example, 167 policies are need to bring in the full 10,000,000 pesetas being insured and paid out for a single event. If the probability is fulfilled and six people out of every 1,000 die, the balance is maintained. However, if there is fraud and a person who is not simply exposed to the risk but is a carrier of the adverse event joins the insured population, two deaths will result. The final cost will be 20,000,000 pesetas and the premium that should be collected from the policyholders will be twice as large to offset the fraud.

It is precisely the insurance contract process that must prevent this type of alteration, since the situation is not managed by simply adjusting the premium by 10% but is an upset to the entire balance of the operation. For the insurer, the process must guarantee that the known risk characteristics mean that the individual can be insured; and the insured must have the guarantee that, in the measure necessary, the insurance company is solvent and will be able to pay the policy amount if there is an adverse event.

Information and good faith in the insurance contract process

Due to the legal characteristics of the agreement and based on the good faith of all contractual parties, the insurer bases its acceptance of the risk on the information provided by the candidate for insurance. Only in the case of extremely high policy amounts or deviations from the standard risk parameters (e.g., health condition, profession, athletic activities) may the insurer ask for additional information.

In other words, if a portfolio comprises 1,000 policies with a mean policy amount of 2,000,000 pesetas and 10 policies with a policy amount of 80,000,000 pesetas each, mathematical compensation of the risk is not possible to the same degree: the 1,000 policies of small capital represent a sufficient number of risk units to be well-adjusted to the six deaths forecast by the statistics. The remaining ten policies could be more seriously influenced by chance, with one, three or no deaths. Here the insurer must manage carefully to prevent collapse resulting from exposure to chance, because the limited size of the portfolio does not permit stable predictions.

Changes in the insuring process

The balance that should result from the good faith on which the agreement is based can be upset when the policyholder already knows that an event has occurred or will occur, and conceals this fact to gain coverage that would normally be accepted only at a higher premium or perhaps even denied.

The insurer can only handle these situations by analysing or checking the information provided when the policy is being taken out, such that if fraud or bad intentions are proven and information was withheld, the agreement becomes null and void. For this reason, the Insurance Contract Act requires that the insurer must be informed by the policyholder and the insured of any circumstances that could affect the assessment of risk and which, if known by the insurer, could mean that the insurance policy would not be granted or would be granted at another price.

When the law establishes this criterion, it does not protect the insurer, as might appear, but protects the solvency of the insurance institution by preventing fraudulent acts that could injure the other policyholders. The same goal is sought when the law establishes the principles of equity and sufficiency in defining the premium amount so that each person pays the risk that he or she represents.

2. Life insurance policy rates

The greater the likelihood that the risk will materialise and the more serious the consequences, the higher will be the premium. When assessing the risk in order to set the premium amount, the company uses two kinds of information:

1. Statistical information, which gives an idea of average risk, and
2. Specific information concerning individual risks.

In the case of life insurance, the statistical information is presented in mortality or survival tables used to calculate the probability of death and the probability of survival.

Along with the great majority of individuals who have a life expectancy that fits the tables, there are also some with enhanced risk (sick individuals, or individuals with a hazardous occupation, to cite two obvious examples). These individuals have a lower life expectancy and special interest in obtaining life insurance is foreseeable.

The insurer cannot systematically reject higher risks because it would be depriving insurance benefits to the individuals who most need it. Moreover, the company can assume the risk without any problem, provided the rate is appropriate to the increased risk.

Exceptional risks must be specially analysed, assigned rates and offset by staff with training in actuarial and medical life insurance techniques using specific information.

This information consists in medical screening which is simply the compilation of medical information aimed at determining the insured's current health condition and the factors that could determine his or her future health. The information is compiled on a "health status declaration", a questionnaire included in the insurance application that must be filled out and signed by the insured. It includes questions on the applicant's height and weight (body mass), personal history (e.g., prior accidents or diseases), whether he or she smokes, etc.

In relation to certain policy amounts or when there are affirmative answers to some of the questions on the simplified medical questionnaire, the company performs a more extensive questionnaire which contains questions on family medical history. This is important because the answers indicate the probability of developing a hereditary disease, as does the genetic information now under debate.

The insurance applicant may even be required to undergo a medical examination, which will be meticulous and can include a traditional medical examination (physical examination), urinalysis, EKG, etc.

Acceptance or refusal and "additional premiums"

After the health condition declarations are filled out and/or the medical examinations are completed, the results are analysed by specialised medical staff employed by the insurance companies. These staff members accept or reject the application.

When the insurance contract is signed, the premium is finally set by comparing the information on the mortality or survival tables, the medical data and any other information, the type of insurance requested, the policy amount and the term of the insurance policy.

3. Genetic tests and life insurance

Let's look at how genetic information can affect the process of selection and assignment of rates. For some diseases, genetic tests can show that there is an above-average probability of developing this disease in the future. For others, the result of the tests is less certain. Nevertheless, every indication is that in the future genetic tests will provide more information on a larger number of diseases.

To some extent, the factors currently examined by the insurance company (family medical history or certain medical tests) are indirect indications of

genetic characteristics; that is, such information forms a part of the medical data. Use of this information when setting rates for life insurance is beneficial for the insured population as a whole.

For example, if companies could not ask life insurance applicants any questions about any diseases such as cancer or AIDS, all the individuals affected by these diseases would be able to obtain insurance without additional premiums and the overall cost of these deaths would have to be assumed by the premiums of all the policyholders.

In line with the above, a potential concurrence of policyholders with genetic problems would result in a technical imbalance in the insurers' system unless these institutions were aware of the higher risk and could apply the respective additional premium. This situation would mean that the premium payments would be insufficient to pay the benefits due for actual deaths. If this circumstance continued, the premiums would quickly rise. If the increase were not too high, new insurance policies would not be affected too much but if the premium were to increase significantly, the increase could hinder the inclusion of "healthy policyholders", jeopardising the insurance institution by compromising its future viability and thus, its social function.

4. Legislation on insurance and genetics in Europe and Canada

Some European countries (Austria, Belgium, Denmark or Norway) have specific legislation that prevents insurers from gaining access to genetic information. In France and the Netherlands, there is no formal legislation; however, a voluntary moratorium does exist. In other countries such as Spain, Finland or Iceland to mention a few, there is not sufficient specific legislation. Nevertheless, the companies have not included any kind of question relative to genetic information in their questionnaires.

The ban on their use is generally a result of the willingness of legislators and politicians' to impose a period of waiting and reflection until genetic science is more advanced, and not of their desire to prohibit it categorically.

In the United Kingdom, the government has drawn up a code of conduct to which all institutions can adhere voluntarily. This code of conduct is based mainly on two principles:

1. The person who requests a life insurance policy should always have the option to undergo a genetic test or not. **No insurer may oblige an applicant for an insurance policy to undergo a genetic test as a condition to be insured.**

2. Nevertheless, when the insurance applicant has undergone a genetic test, the test results should be provided to the insurer unless the latter indicates that the information is not required.

This obligation is logical, since an individual who has already undergone genetic testing may be more likely to want to insure a specific risk from that time on. Therefore, this measure is intended to avoid an adverse selection.

This principle is consistent with the current operation of insurance companies. Once all the information needed to assess the risk has been compiled, the premium is set and the policy is issued. After this point, the companies are not allowed to request any additional information from the insured individual.

The UK case is certainly an example of the maturity and tradition of the insurance industry in that country. British society understands that, as analysis offers increasingly better, more reliable results, genetic information becomes to some extent an additional component that improves the assignment of rates to risk and prevents adverse selection and the resulting general increases in premiums.

The Canadian system is also particularly interesting. Canada has a single independent clearinghouse for all genetic tests that is used by any institution that needs information for its regular activities. In response to the requests for information, the centre does not provide a copy of genetic test results, as this would not provide any protection against potential misuse; for example, in the case of the insurers the answer is limited to an indication of increased risk and contains no further information.

5. Conclusions

Genetic information can be beneficial to the insurance business and its social function for the following reasons:

- It makes it possible to obtain predictive information on diseases that could allow the life expectancy to be extended through a more accurate estimation of expected death or survival rates.
- Discrimination is treating “equals” as different. However, treating different individuals as “equals” is also discriminatory; it is presently well-understood that a person should pay higher or lower premiums on the basis of age or the diseases they have. Why should genetic information be any different? We should not confuse the right to information needed for

doing business in the insurance industry with potential misuse of this information. Misuse must be avoided at all costs, but without reaching the extreme of withholding relevant information.

- This prevents restraints on the use of genetic information from leading to fraud or adverse selection that would jeopardise the balance and solvency of the insurance industry.

In short, genetic information helps to improve assignment of rates, as it raises and lowers the cost of insurance, thereby benefiting the entire insured population and aiding the solvency and continuity of insurance firms.

Its use requires:

- Implementation of mechanisms and processes needed to ensure that the interpretation, utilisation and confidentiality of genetic information is adequate.
- Increased development for proper use of genetic information.
- Flexible regulation that can be systematically adapted.

In addition, we could mention the following points:

- Genetic information is simply additional medical information, which allows a person's life expectancy to be assessed more accurately.
- Insurers already assess the family medical history when screening for persons at risk.
- Life insurance policies are often long-term contracts and genetic information can help to improve rate-assigning on behalf of the policyholders.
- The use and correct interpretation of genetic tests will be possible in the long run.
- Legislation relating to genetic information must be flexible and must adapt to the advances in science and medicine.

Companies use medical and scientific advances as they become popular and available. The objective is to improve risk assessment to the benefit of the entire insured population.

All fields, including the insurance industry, must handle these medical advances in a **responsible, prudent** manner in order to maximise their impact on the improvement of mankind and on the well-being of society in general.

Opinion of the Philosophers

María Casado

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The ethical, social and legal implications of predictive genetic medicine and of the information obtained through genetic diagnosis are evident, and each new step is naturally accompanied by bioethical reflection. The genetic information available today can be used in a wide variety of fields and for a wide variety of purposes.

First of all, advances in the study of genetic pathologies enable us to understand their molecular causes and provide new possibilities for diagnosis. This will have repercussions on the prevention of these diseases and on their potential treatment. Hence, an awareness of the individual's genetic makeup will enable us to design new personalised therapies that are appropriate for each case, thereby decreasing the harmful effects and enhancing the efficacy of new treatments. The possibilities of these treatments will gradually increase as new functions of the genes and their relationship to diseases are identified. These developments have irrefutable advantages for public health but can also entail disadvantages for individuals and be a source of new forms of discrimination.

Nevertheless, the capacity to accurately identify individuals is useful for the State and can also be useful for people. This is the case of identification in civil and criminal procedures (e.g., paternity cases or the analysis of samples found at the site of a crime). This is reassuring for the population, but it can also generate fear of a world in which we are converted into completely "transparent" individuals with no privacy whatsoever. The public is just as concerned about the insurance companies' use of these data when they take out policies, or when they are applying for mortgages.

These advantages and disadvantages lead to the perception that biotechnologies are a two-edged sword and cause the public to have many fears and hopes concerning their potential. This ambivalence can be seen in the bioethics debate, and is particularly obvious in the media and public opinion. As a result, much emphasis is placed on the need to draw up regulations that prevent and curb potential abuse. Regulations already exist at the national and

international level; however, the mere existence of rules is not enough. Regulatory measures from the top are insufficient; rigorous information and prior debate on the implications of biotechnology are needed in order to establish agreements, even if they are partial, provisional and revisable.

The reference framework for decision-making is not comprised of personal beliefs (which are worthy of respect but not universal). Rather it consists in a respect for human rights and the promotion of such rights, which are the ethical and legal basis of our coexistence. The new possibilities of infringement of rights already recognised in key international protective documents and the need to protect new rights have led to the adoption of the Universal Declaration on the Human Genome and on Human Rights under the auspices of the UNESCO on the 50th anniversary of the Universal Declaration of Human Rights (1948) and the Council of Europe Convention on Human Rights and Biomedicine, which was published in the Official Spanish Gazette of 20 March 1999 and is currently in effect in Spain. The aim of these agreements is to protect human dignity and human rights against the new potential of biomedicine by establishing the voluntary nature of genetic testing, the confidentiality of the results, the ban on discrimination for genetic reasons and on the modification of non-pathological genetic heredity, the equitable access to the benefits obtained from biotechnology, the solidarity and respect for freedom in research, as well as the obligation of countries to encourage interdisciplinary dialogue and the creation of ethics committees that foster public debate and information.

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As has been mentioned throughout our discussion, the results of predictive medicine can have positive and negative aspects. In terms of positive implications, we have highlighted the prevention and cure of disease, but we have also mentioned that, although we can measure the probability of developing certain diseases we still do not have suitable therapies to cure them. It is not surprising, therefore, that we find it necessary to point out the threats of genetic testing for both individuals and society; anxiety and frustration for the individual and possible attempts to justify discriminatory social practices, particularly in labour relations and in life and health insurance contracts. Since the tests are still expensive, their general use is not of interest to companies as

yet. However, the costs are bound to drop with time and an in-depth social debate will be needed to prevent the misuse of genetic information.

From my perspective, genetic diagnoses and the information they provide offer more advantages than disadvantages within the context of reproduction. Thus, these techniques can be widely used by the entire population. A social and democratic state under law must guarantee access to preconception, preimplant and prenatal diagnoses, particularly for individuals with more limited capacity to use their resources for health. In my opinion, it is essential to prevent the use thereof from being influenced by the ideological choices of the physician.

Information on the results must be clear, confirmed, complete and impartial. In any case, the decisions made by individuals on the basis of the information provided by the tests should be respected.

My rationale for using these tests is not based on any eugenic idea. I do not feel there is any moral obligation to perfect humanity through the selection of certain biological characteristics which could be considered more desirable than others at any particular point in time. I also do not feel it is fair to prevent the births of human beings with serious impediments on the basis that, although this does not consist in a prejudice to them, it is an injustice to introduce a specific ill in the world without cause, as Feinberg affirms.

Conceiving a child has never been considered illicit simply because it could have a serious disease. It is hard to allow, therefore, that it would be worse to live with these impediments than not to have lived. Many countries do not allow eugenic abortion, and the legislation allowing it does not make its practice *compulsory*, making it safe to assume that continuing with the pregnancy is *allowed*. This means that we give more value to life than to preventing someone from being born with serious anomalies. We cannot say, therefore, that bringing children with problems into the world is an *injustice* if the alternative is something worse: not having given them life. Of course, this has nothing to do with cases where the anomalies are caused by improper behaviour on the part of someone who has harmed an initially problem-free pregnancy.

The reason behind the advisability of genetic diagnoses is that parents will satisfactorily attend to the physical and psychological welfare of a child with severe or not-so-severe disabilities once born, only when the birth is the result

of an informed, voluntary decision. I feel that the State should provide all the resources needed for its citizens to take responsible, autonomous decisions in the area of reproduction. Naturally this should not preclude that significant economic assistance from the State be granted for handicapped children with special needs once they are born.

The greatest danger behind predictive medicine in general and prenatal and postnatal diagnoses in particular is that of biological reductionism. Eradication requires a well-developed cultural critique that exposes the relationship between disease/disability and the socio-political environment. In most cases, diseases are the result of an interaction among the different genes as well as between the genes and the environment. Focusing only on the genetic aspects of diseases has a negative side, as seen in the eugenics movement of the early 20th century, in that disease is considered basically the individual's problem, social integration of individuals with disabilities is prevented, and no effort is made to investigate into or transform the specific socio-political structures that favour and condition diseases.

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19. **Anna Veiga**, Head of the Biology Section, Reproductive Medicine Department, Institut Universitari Dexeus de Barcelona

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