Protection of Genetic Data in Medical Genetics: A Legal Analysis in the European Context*

Sergio Romeo-Malanda, LL.B., Ph.D.
Research Fellow. Centre for Law and Genetics. Law Faculty, University of Tasmania. Hobart, Australia.

Dianne Nicol, B.Sc., M.Sc., *LL.B., LL.M., Ph.D.*
Deputy Director. Centre for Law and Genetics. Associate Professor. Law Faculty, University of Tasmania. Hobart, Australia.

Resumen / Abstract:
Este artículo se basa en tres ideas, a saber: 1. Todas las cuestiones en el ámbito de la "intimidad" y de la "confidencialidad" que surgen a partir de los análisis genéticos únicamente deben ser tomadas en consideración cuando nos encontramos ante "datos personales". 2. Cuando se trata de datos genéticos personales, hay dos aspectos que hay que garantizar especialmente: a) la libertad y autonomía del individuo; y b) el deber de secreto en aras a la protección de la intimidad de la persona. 3. Entre estos dos aspectos, puede surgir algún tipo de conflicto, que habrá que resolver. El autor analiza la legislación europea de carácter supranacional relativa a esta cuestión, de acuerdo con la cual, debe protegerse la intimidad genética. También indica cómo la medicina genómica puede dar lugar a una variedad de conflictos e intereses, y señala como resuelven este dilema los diferentes textos legales objeto del estudio.

* We would like to thank Prof. Margaret Otlowski for her insightful comments on earlier drafts of this article. Responsibility for ignoring her best advice, and making mistakes anyhow is, of course, ours alone.

This article is based in three ideas, namely: 1. All the questions in the field of "privacy" and "confidentiality" derived from genetic tests only must be taken into account if we deal with "personal data". 2. When we are dealing with personal genetic data, two aspects must be especially guaranteed: a) the freedom and autonomy of the individual; and b) the duty of secrecy in order to protect the privacy of the person. 3. Some conflicts can appear between these two aspects and we have to deal with them. The author analyses the supranational European legislation referring to this topic, according to with, genetic privacy must be guarantee. He also notes how Genetic medicine can give rise to a variety of conflicts of interests, and points out how the different legal texts object of study deal with this issue.

Palabras clave / Keywords:

Datos genéticos / Derecho a la protección de datos / Derecho a saber y a no saber / Deber de confidencialidad / Descubrimientos inexperados / Conflictos de intereses / Legislación europea

Genetic data / Right to data protection / Right to know and not to know / Duty of confidentiality / Unexpected findings / Conflicts of interests / European legislation / Public trust

1. Introduction

One of the short- and medium term goals of human genome research is to gain knowledge of the characteristics of human DNA and its components, particularly genes. There is particular interest in learning how genes function and what their role is in the transmission of biological heredity. Progress in the field of biomedical science has made it possible to obtain greater knowledge of the human genome and the nature of genetic disorders.

Thanks to these advances in genetics, doctors now have the tools to understand how certain illnesses, or increased risks for developing certain illnesses, pass from generation to generation. According to some health experts, the definition of an inherited or genetic illness should be expanded beyond the classic inherited disorders (like hemophilia and sickle cell anaemia) to include many types of cancer, Alzheimer's disease, and other illnesses. They look toward a future where genetic test results are an important part of every healthy person's medical file. Currently, genetic testing has developed enough so that doctors can pinpoint missing or defective genes. However, in practically all cases, treatments for those diseases are still far off.

The term "genetic testing" refers to those analyses which serve[1]: a) to diagnose and classify a genetic disease; b) to identify unaffected carriers of a defective gene in order to counsel them about the risk of having affected children; c) to detect a serious genetic disease before the clinical onset of symptoms in order to improve the quality of life by using secondary preventive measures and/or avoid giving birth to affected offspring; d) to identify persons at risk of contracting a disease where both a defective gene and a certain lifestyle are important as causes of the disease.

Despite the paucity of available treatments for genetic conditions at the current time, genetic testing still has an important social value in informing people about genetic risk factors for them and their offspring (both present and future). Genetic testing of individuals can bring to light important personal and family information, such as biological information on a person's current and future health, including mental health, even though this may be limited to giving advance warning of a propensity or predisposition to certain disorders, or information on reproductive capacity and the future health of offspring. Knowledge of such matters gives individuals the capacity to plan for the future and to avoid lifestyle choices that may exacerbate the risk of adverse healthcare outcomes. And, of course, those who receive a negative test result will have their concerns about developing a genetic condition in the future or passing on genetic risk factors to their offspring alleviated.

But genetic testing could have significant detrimental social impacts as well. The information which can or could be obtained from genetic testing raises problems associated with the information itself, access thereto, and the uses of such data, given that the interests of the individual to whom these data refer (data subject) may be in conflict with those of other individuals (including their biological family), with collective research, health and safety interests, and even with interests of an entirely different kind (e. g. economic). Access to such information provides knowledge of highly important aspects of the tested individual and directly affects his or her innermost sphere[2]. Unless appropriate

[1] Recommendation No. R (92) 3 on genetic testing and screening for health care purposes, of 10 February 1992.

[2] Nevertheless, as Margaret Otlowski, "Protecting Genetic Privacy in the Research Context: Where to From Here?", *Macquarie Law Journal*, 2002, p. 91 notes, «the availability of human genetic information is not of itself new (as noted earlier, some genetic information has long been available through family history of genetic disease) – what has changed is the *means* by which genetic information is available and also the *extent* of information which can now potentially be obtained as a result of the advancements in relation to genetic testing».

procedures are put in place to protect this information, there is a risk that individuals may simply choose to refusing testing, even though it might otherwise provide significant benefits to them and their families. The role of the law in this area should be to provide an appropriate balance between the need to protect the interests of individuals who undergo testing and the need to take into account other legitimate interests.

The aim of this article is to point out the main questions and dilemmas that have arisen in the field of the processing of genetic data in medical genetics[3] and to examine the solutions that have been offered in the European framework. The rationale for examining these issues in the European context is that the European framework for personal data protection was established much earlier than in other jurisdictions and has undergone much greater scrutiny at the policy level. As such, the European approach provides one possible model that might be adopted much more broadly, especially if we take into consideration that the key legislative development in the field of genetic data is the recognition of the human right to privacy. In this respect, as we will see later in this article, of special interest are the works carried out since the early eighties by the Council of Europe.

The Council of Europe is an international organization of 47 member states in the European region, among whose purposes is the protection of human rights[4]. This organization has produced several documents in the field of right to privacy and data protection. From this same perspective, Recital 10 of Directive 95/46/EC on the Protection of Individuals with Regard to the Processing of Personal Data and on the Free Movement of Such Data, reminds us that 'the object of the national laws on the processing of personal data is to protect fundamental rights and freedoms, notably the right to privacy, which is recognized both in Article 8 of the European Convention for the Protection of Human Rights and Fundamental Freedoms and in the general principles of Community law; ... for that reason, the approximation of those laws must not result in any lessening of the protection they afford but must, on the contrary, seek to ensure a high level of protection in the Community'.

[3] That means that the possibility of misusing genetic information for non-medical purposes (i.e. insurance, employment, commercial transactions), or even its use with other social and legally admissible aims (i.e. DNA fingerprint in forensic or criminal justice, determination in parentage, etc.) is beyond the scope of this article.

[4] The main success of the Council of Europe has been the European Convention on Human Rights in 1950, which serves as the basis for the European Court of Human Rights. The Council of Europe is not to be mistaken with the Council of the European Union or the European Council, as it is a separate organization and not part of the European Union.

2. Public Views on Genetic Testing in Europe

The extensive debate at the policy level in Europe about protection of genetic data has not been matched by a detailed analysis of public concerns and needs with regard to the protection of their data. In broad terms, it would appear that public trust in genetic testing is far higher than for other aspects of biotechnology, as illustrated by the 2005 Eurobarometer survey on biotechnology. Eurobarometer surveys have been conducted six times in the European Union since 1991. The aim of the surveys is to seek opinions from a representative sample of 25,000 people across the European Union on such issues as stem cell research, GM crops, nanotechnology, pharmacogenomics and genetic testing.

The report on the 2005 survey makes the following statement about the portrait of Europeans that it presents, showing them to be: "increasingly optimistic about biotechnology, more informed and more trusting of the biotechnology system. The European public is not risk-averse about technological innovations that are seen to promise tangible benefits."[5] This finding followed on from a sharp decline in public confidence in biotechnology in 1999.

The 2005 survey includes specific questions relating to genetic testing and use of genetic information. In response to the question: "would you be willing to take a genetic test to detect any serious disease that you might get", 32% of participants said that they definitely would and 32% said they probably would, whereas only 16% said they definitely would not and 15% said they probably would not (leaving 5% undecided).[6] Similar levels of support were given in relation to donation of genetic information to a national data bank for research into the origins of disease. These and other data cause the authors of the study to conclude that while the European public is not overwhelmingly supportive of the use of genetic data for such matters as personal genetic diagnosis or for gene banks, nevertheless there is majority support for such uses. Conversely, use by government agencies and for commercial insurance does not receive broad support.[7] The authors of the report also note that while there has been a general increase in level of trust in biotechnology

[5] George GASKELL *ET AL*, *Europeans and Biotechnology in 2005: Patterns and Trends. Final Report on Eurobarometer 64.3* (European Commission's Directorate-General for Research; July 2006), p. 3.

[6] Ibid., p. 51.

[7] Ibid., p. 54.

actors across Europe since the 1999 survey, the European Union tends to be more trusted than respondents' national governments in the regulation of biotechnology.[8] This factor must be borne in mind in considering the adequacy of the regulatory regime for genetic testing in Europe.

There is a decided lack of data on the reasons why members of the public might be less than willing to undergo geentic testing. Privacy is generally thought to be of paramount concern by legislators, policy makes and academic commentators alike. It would be useful to have more empirical data on public perceptions to validate these assumptions (or not, as the case may be).

A Report from the UK Academy of Medical Sciences on the specific issue of use of helath information in medical research makes the following comment on the consequences of the lack of empirical evidence of public attitudes relating to this specific issue[9]: "The absence of such knowledge, and the lack of public debate forces regulatory and advisory bodies to make assumptions about what the public might find acceptable. Development of good practice should be informed, as far as possible, by empirical evidence on public and patients' awareness and attitudes".

One recent study provides useful insight about public views on the confidential use of identifiable information by the British National Cancer Registry.[10] The authors of the study surveyed 2955 respondents and based on their findings they conclude that: "most of the British public considers confidential use of personal, identifiable patient information by the National Cancer Registry for the purposes of public health research and surveillance not to be an invasion of privacy"[11]. Similar findings have also been reported in Australia.[12]

In sum, what data there are on public trust in relation to genetic testing and use of genetic information suggest that individuals do have some confidence that their genetic data will be used in ways that respect

[8] Ibid.

[9] Academy of Medical Sciences, *Personal Data for Public Good: Using Health Information in Medical Research (January 2006).*

[10] Geraldine Barrett/Jackie Acassell/Janet L Peacock/Michael P Coleman, "National Survey of British Public's Views on Use of Identifiable Medical Data by the National Cancer Registry", *British Medical Journal,* 332 (2006), p. 1068.

[11] Ibid., p. 1068.

[12] David C Whiteman/Cathy Clutton/David Hill, "Australian Public's Views on Privacy and Health Research", *British Medical Journal,* 332 (2006), p. 1274

their privacy and confidentiality, and are willing to allow their genetic information to be used on this basis. In moving forward, it is important that this level of trust is not compromised. Much more evidence needs to be collected on public attitudes more generally. In the interim, probably the safest option in considering the appropriate mechanisms for regulating genetic testing and use of genetic information is probably to rely on the perceptions of experts in the field as to the issues that are most likely to concern members of the public. On this basis, adequate protection of privacy and recognition of self determination are likely to be pivotal issues. This article discusses these and related matters within the context of the European legislative and regulatory framework.

3. The European Legislative and Regulatory Framework

There are several legal and non-legal documents dealing with the protection of genetic data that must be taken into account at the European level. Firstly, documents of international organizations like UNESCO[13] have a repercussion world-wide and should be respected both by the European Union (EU) and by the States. In this group, the International Declaration on Human Genetic Data (IDHGD), of 16 October 2003, is particularly important.

Secondly, there are a number of important documents of the Council of Europe, namely, the European Convention on Human Rights, of 4 November 1950; the Convention for the Protection of Individuals with regard to Automatic Processing of Personal Data, of 28 January 1981[14]; Recommendation No. R (92) 3 on Genetic Testing and Screening for Health Care Purposes, of 10 February 1992; the Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine: Convention on Human Rights and Biomedicine (CHRB), of 4 April 1997[15]; and Recommendation No. R (97) 5 on the Protection of Medical Data, of 13 February 1997.

[13] UNESCO (United Nations Educational, Scientific and Cultural Organization) is a specialized agency of the United Nations established in 1945. Its stated purpose is to contribute to peace and security by promoting international collaboration through education, science, and culture in order to further universal respect for justice, the rule of law, and the human rights and fundamental freedoms proclaimed in the UN Charter.

[14] This Convention was the first international legally binding text on data confidentiality.

[15] Unlike the European Convention on Human Rights which applies to all EU Member States, the Convention on Human Rights and Biomedicine has not been signed or ratified by many States, including most of the larger States. In spite of it not applying directly to many EU States, it is nevertheless significant in that it has been drawn upon by the

Thirdly, the legislation of the European Union is also relevant, particularly the Charter of Fundamental Rights of the European Union, of 7 December 2000, and Directive 95/46/EC of the European Parliament and of the Council, on the Protection of Individuals with Regard to the Processing of Personal Data and on the Free Movement of Such Data, of 24 October 1995. The latter is especially relevant because it generates in all the EU Member States an effective obligation to act according to the terms stipulated in it[16].

Finally, we cannot overlook the legislation of every European country[17]. At regulatory level the situation across Europe appears to be uneven. Indeed, while some States either have explicitly listed genetic data as sensitive data in their Data Protection laws, thereby incorporating all the safeguards and restrictions associated with this categorization (Italy[18], Poland[19]), or have passed specific legislation on protection of genetic data (Switzerland[20], France[21], Spain[22]), in most States the issue of

European Court of Human Rights in making judgments involving States who are not parties to this Convention. See in this respect, *Glass v. The United Kingdom,* 9 March 2004 (paragraph 58); *Evans v. The United Kingdom*, 7 March 2006 (paragraph 40).

[16] Directives are a legislative act of the European Union which requires member states to achieve a particular result without dictating the means of achieving that result. It can be distinguished from European Union regulations which are self-executing and do not require any implementing measures. Directives normally leave member states with a certain amount of leeway as to the exact rules to be adopted. Directives can be adopted by means of a variety of legislative procedures depending on the subject matter of the directive.

Of course, this legal text is only applicable to EU member States (currently 27: Austria, Belgium, Bulgaria, Cyprus, Czech Republic, Denmark, Estonia, Finland, France, Germany, Greece, Hungary, Ireland, Italy, Latvia, Lithuania, Luxembourg, Malta, The Netherlands, Poland, Portugal, Romania, Slovakia, Slovenia, Spain, Sweden, United Kingdom).

[17] At this level, we will have to take into account that the European Court of Justice has made clear that nothing prevents the Member States from extending the scope of the national legislation implementing the provisions of the Directive to areas not included within the scope thereof, provided that no other provision of community law precludes it. It may therefore very well happen that certain situations not involving processing of personal data as defined in the Directive 95/46/EC are nevertheless subject to protective measures under national law. See. DATA PROTECTION WORKING PARTY, *Opinion 4/2007 on the concept of personal data*, p. 24.

[18] Personal Data Protection Code (*Codice in materia di protezione dei dati personali*). Legislative Decree No. 196, of 30 June 2003. Section 90 deals with the "processing of genetic data and bone marrow donors".

[19] Act on the Protection of Personal Data, of 29 August 1997, Article 27.

[20] Human Genetic Testing Act (*Loi fédérale sur l'analyse génétique humaine*), of 8 October 2004.

[21] Public Health Code (*Code de la Sante Publique*), amended by Law No. 2004-800 on bioethics, of 6 August 2004, Article L1131.

[22] Law 14/2007, of 3 July, on Biomedical Research.

the processing of genetic data is not as such regulated by specific legislation. However, as national authorities become increasingly aware of the risks associated with the processing of genetic data, a general trend towards new initiatives at the national regulatory level is anticipated.

Appart from these legislative sources, several guidelines and reports have been elaborated both at international and national level. It is beyond the scope of this work to discuss all of these instruments. However, special attention will be paid to the "Working Document on Genetic Data", adopted on 17 March 2004 by the *Data Protection Working Party*[23] and the report entitled "Ethical, Legal and Social Aspects of Genetic Testing: Research, Development and Clinical Applications", elaborated by the *Expert Group on Genetic Data* at the request of the European Commission, also in 2004.

4. Personal Genetic Data and Protection of Privacy. The Meaning of "Personal Data" in the European Context

As noted above, the most significative legal text related to the protection of personal data in the European Framework is the Directive 95/46/EC. Its object is, as stated in Article 1.1, to "protect the fundamental rights and freedoms of natural persons, and in particular their right to privacy with respect to the processing of personal data".

This Directive states that *personal data* "shall mean any information relating to an identified or identifiable natural person ('data subject')", (Article 2 a)). In the same vein, Recommendation R (97) 5 states that "the expression 'medical data' covers any information relating to an identified or identifiable individual" (Principle 1). This definition of personal data

[23] The Working Party has been established by Article 29 of Directive 95/46/EC. It is the independent EU Advisory Body on Data Protection and Privacy. Its tasks are laid down in Article 30 of Directive 95/46/EC and in Article 14 of Directive 97/66/EC, of 15 December 1997, concerning the processing of personal data and the protection of privacy in the telecommunications sector.

The Working Party was set up to achieve several primary objectives: a) To provide expert opinion from member state level to the Commission on questions of data protection; b) To promote the uniform application of the general principles of the Directives in all Member States through co-operation between data protection supervisory authorities; c) To advise the Commission on any Community measures affecting the rights and freedoms of natural persons large, and in particular to Community institutions on matters relating to the protection of persons with regard to the processing of personal data and privacy in the European Community.

incorporates any information regarding a person, whatever his or her nature or origin, being intimate or not, even if it affects several people at the same time or a family group (an aspect that can be of great importance in relation to data concerning health and genetics)[24].

In this respect it is important to make a brief reference to a recent document adopted by the Data Protection Working Party, Opinion 4/2007 on the concept of personal data. This advisory body notes that the above-mentioned definition reflects the intention of European lawmakers for a wide notion of personal data[25]. It points out specifically that, from the point of view of the nature of the information, this definition "covers 'objective' information, such as the presence of a certain substance in one's blood"[26]. In addition, from the point of view of the content of the information, the concept of personal data includes data providing any sort of information[27].

According to this definition, data can be classified into one of three categories, depending on the greater or smaller possibility of identification of the person from whom the data are obtained[28]: a) data relating to an identified person; b) data relating to an identifiable person; and c) anonymous data. However, the nomenclature may vary from one text to another and from one author to another, so that it is more important to pay attention to the concept rather than to the wording.

Data relating to an identified person ("identified data") are data that appear clearly and directly linked with the person from whom they were obtained (data subject). The IDHGD refers to such data as *data linked to an identifiable person*, being "data that contain information, such as name, birth date and address, by which the person from whom the data were derived can be identified" (Article 2.ix).

[24] Carlos M. ROMEO-CASABONA, "Anonymization and Pseudonymization: The Legal Framework at a European Level", in D. Beyleveld/D. Townend/S. Rouillé-Mirza/J. Wright (Eds.), *The Data Protection Directive and Medical Research Across Europe*, Ashgate, England, 2004, pp. 36 f.

[25] DATA PROTECTION WORKING PARTY, *Opinion 4/2007 on the concept of personal data*, pp. 4 and 6.

[26] Ibid., p. 6.

[27] The DATA PROTECTION WORKING PARTY, *Working Document on Genetic Data*, p. 5, notes that «there is no doubt that genetic information content is covered by this definition».

[28] See DATA PROTECTION WORKING PARTY, *Opinion 4/2007 on the concept of personal data*, pp. 12 ff.

The question of whether the individual to whom the information relates is identified or not depends on the circumstances of the case, bearing in mind that a person may be identified directly or indirectly. In terms of "direct" identification, the name of the person is indeed the most common identifier, and, in practice, the notion of "identified person" implies most often a reference to the person's name[29]. With regard to "indirect" identification, this category typically relates to the phenomenon of "unique combinations". Some characteristics are so unique as to render an individual readily identifiable ("King of Spain"), but a combination of details from various categories (age category, regional origin, etc) may also be fairly conclusive in some circumstances, particularly if one has access to additional information of some sort.

Data relating to an identifiable person (as known as *dissociated data*) are data that seem not to be directly attributable to a certain person, since he or she does not appear to be identified or there is no link between the data and the person. However, the linking of such data to the person is possible by diverse procedures, which can normally be easily carried out[30].

Directive 95/46/EC gives a definition of identifiable data in the following terms: "An identifiable person is one who can be identified, directly or indirectly, in particular by reference to an identification number or to one or more factors specific to his or her physical, physiological, mental, economic, cultural or social identity" (Article 2 a)). According to this, the Directive considers a person identifiable in connection with his or her data, if the data are identifiable by mean of an identification number (coded data)[31]. In such cases it is possible to use the code to re-identify the person to whom the data relate, so that the process of de-identification is reversible.

[29] In order to ascertain this identity, the name of the person sometimes has to be combined with other pieces of information (date of birth, names of the parents, address or a photograph of the face) to prevent confusion between that person and possible namesakes.

[30] In these cases, as ROMEO-CASABONA, "Anonymization and Pseudonymization", p. 38 points out, "the connection of the data with the person to whom it belongs can also be obtained by other indirect procedures, such as, for example when the data reveals certain personal or social characteristics that only one person or a very reduced group of people possesses, and those characteristics could be known by others".

[31] Article 2.x IDHGD names *data unlinked to an identifiable person* those data that are not linked to an identifiable person, through the replacement of, or separation from, all identifying information about that person by use of a code.

Finally, *anonymous data* can be considered as data where the identity of the data subject is not known, and identification is not possible because the data were collected as such, or because although collected with identification, they have later been anonymized[32]. To be classified as anonymous, the data must have been subjected to a process of dissociation from the data that refers to that person, in such a way that it no longer allows the person's identification. Consequently, it is necessary that such a dissociation process should be irreversible, that is to say, that the data cannot return to the form taken previously[33].

The IDHGD speaks of *data irretrievably unlinked to an identifiable person*, being data that cannot be linked to an identifiable person, through destruction of the link to any identifying information about the person who provided the sample (Article 2.xi). However, Directive 95/46/EC does not mention this category of data explicitly. It only sets down that "an identifiable person is one who can be identified, directly or indirectly", without mentioning the level of difficulty of the identification. But Recital 26 specifies the reach of Article 2 a) since it points out that "to determine whether a person is identifiable, account should be taken of all the means likely *reasonably* to be used either by controller or by any other person to identify the said person"[34] (emphasis added).

Thus, Recital 26 has restrictive effects on what should be understood as "identifiable people's data", because it adds that "account should be taken of all the means likely reasonably to be used" to identify a person. When those means are not reasonable, the person will no longer be considered legally identifiable and the data will move into the category of anonymous data. What "reasonable" means is not easy to say. Recommendation R (97) 5 is a little more concrete in this respect, as it

[32] Carlos M. ROMEO-CASABONA, "Anonymization and Pseudonymization: The Legal Framework at a European Level", in D. Beyleveld/D. Townend/S. Rouillé-Mirza/J. Wright (Eds.), *The Data Protection Directive and Medical Research Across Europe*, Ashgate, England, 2004, p. 38.

[33] The DATA PROTECTION WORKING PARTY, *Opinion 4/2007 on the concept of personal data*, p. 21, also notes that "the assessment of whether the data allow identification of an individual, and whether the information can be considered as anonymous or not depends on the circumstances, and a case-by-case analysis should be carried out with particular reference to the extent that the means are likely reasonably to be used for identification as described in Recital 26".

[34] It should be note that the recitals in the Directives do not have a statutory value, but they are very useful to interpret the rules contained in the legal text.

states that "an individual shall not be regarded as 'identifiable' if identification requires an unreasonable amount of time and manpower" (principle 1)[35].

The principles of data protection do not apply to data rendered anonymous in such a way that the data subject is no longer identifiable. This statement implies that protection will not be given to *any* personal data that have been subjected to an anonymization process[36]. However, if these anonymized data were processed and it became possible to identify the data subject again, they would regain the status of personal data and the principles of data protection would be applicable to them again[37].

5. Medical Data and Genetic Data: Genetic Exceptionalism?

Within the category of personal data, there are some data that have special protection: the so called *sensitive data*[38]. Sensitive data have been defined as "data in connection with which the data subject is more vulnerable when the data is known or used by a third party because of its potential for causing discrimination and other misuse, especially when accessed, used or illicitly disclosed"[39]. As a consequence, these data are regarded as needing more intensive protection. What are usually regarded as sensitive data are data that reveal racial or ethnic origin, political opinions and religious or philosophical convictions of the data subject, as well as concerning health and sexuality. Within this category of sensitive data, data concerning health, and genetic data, require closer scrutiny.

The expression *medical data* refers to all personal data concerning the health of an individual. It refers also to data which have a clear and close link with health as well as to genetic data.[40] But genetic data are subject to a specific definition. G*enetic data* are "all data, of whatever type,

[35] In the same sense, the German Federal Data Protection Act, of 20 December 1990, gives a definition of "despersonalization" as follows: "modification of personal data so that the information concerning personal or material circumstances can no longer or only with a disproportionate amount of time, expense and labour be attributed to an identified or identifiable individual" (Section 3.6). On relevant factor will also be the purpose pursued by the data controller in the data processing. See DATA PROTECTION WORKING PARTY, *Opinion 4/2007 on the concept of personal data*, pp. 15 ff.

[36] ROMEO-CASABONA, "Anonymization and Pseudonymization", p. 34.

[37] Ibid., p. 42.

[38] Cf. Directive 95/46/EC, Recitals 34 an 70.

[39] ROMEO-CASABONA, "Anonymization and Pseudonymization", p. 37.

[40] Recommendation R (97) 5, Principle 1.

concerning the hereditary characteristics of an individual or concerning the pattern of inheritance of such characteristics within a related group of individuals. The definition also includes all data on the carrying of any genetic information (genes) in an individual or genetic line relating to any aspect of health or disease, whether present as identifiable characteristics or not. The genetic line is the line constituted by genetic similarities resulting from procreation and shared by two or more individuals".[41]

Thus, the term "human genetic data" (or "human genetic information") is used to describe information about an individual's genetic make-up. However, there is ongoing debate about the real reach of the term "genetic information"[42]. In the light of the major biotechnological developments in the field of genetics, there is a tendency to assume that acquiring human genetic information necessarily entails genetic testing. Nevertheless the term is broad enough to also cover genetic information available through other means (for instance, family history)[43]. In this sense, Article 2.i) IDHGD describes human genetic data as "Information about heritable characteristics of individuals obtained by analysis of nucleic acids *or by other scientific analysis*" (emphasis added).

In a nutshell, medical data are data concerning health, and information about a person's genetic make-up obviously come within the meaning of "health" or "medical" information[44]. As a consequence,

[41] Ibid.

[42] See Pilar NICOLÁS JIMÉNEZ, *La protección jurídica de los datos genéticos de carácter personal*, Cátedra Interuniversitaria de Derecho y Genoma Humano-Comares, Bilbao-Granada, 2006, pp. 66 ff.; Janneke H. GERARDS/Heleen L. JANSEN, "Regulation of Genetic and Other Health Information in a Comparative Perspective", *European Journal of Health Law*, No 13 (2006), pp. 347 ff.

[43] OTLOWSKI, "Protecting Genetic Privacy in the Research Context", p. 89. The NATIONAL CONSULTATIVE ETHICS COMMITTEE FOR HEALTH AND LIFE SCIENCES (France), *Opinion n° 76 regarding the obligation to disclose genetic information of concern to the family in the event of medical necessity* (2003), p. 2 f., also points out that a test is not the only way of diagnosing a genetic disease, putting as examples the haemoglobin electrophoresis (for sickle cell disease and thalassemia), renal ultrasound (polycystic kidney disease), coloscopy (polyposis of the colon), or cholesterol assays, and affirms that "all of these diagnostic criteria would well generate the same legal consequences as a genetic test". See also, GERARDS/JANSEN, "Regulation of Genetic and Other Health Information in a Comparative Perspective", p. 349 («Genetic information may be derived from family medical history, from testing (tests either directed immediately to genetic information or tests that are directed to other health information but that may also yield genetic information, such as blood tests or urine tests) but also from information derived from observance of an individual's behaviour»).

[44] DATA PROTECTION WORKING PARTY, *Working Document on Genetic Data*, p. 5; OTLOWSKI, "Protecting Genetic Privacy in the Research Context", p. 92.

genetic data are considered to be part of this category[45]. The concern that genetic data may be particularly prone to misuse has fuelled perceptions that genetic information is fundamentally different from other forms of medical data, and has led to calls for policies to treat such information differently from all others medical information ("genetic exceptionalism")[46].

The Report "Ethical, Legal and Social Aspects of Genetic Testing: Research, Development and Clinical Applications", affirms that "genetic information is not, as such, different from any other personal medical data, and should therefore be treated in the same way"[47]. However, the potential information obtained from genetic tests conducted on a person is different from any other information: its source is indestructible since it is present in almost all the cells of the body while alive and usually even when dead; it is permanent and unalterable, save for spontaneous genetic mutations or ones triggered through genetic engineering or as the result of other external agents (e. g. radioactive ones). In any case, such mutations will always be partial and limited. Lastly, genetic tests give information not only about the subject, but also about his or her biological family.

Likewise, according to the IDHGD (Article 4.a)), human genetic data have a *special status* because:

[45] It has been discussed if information provided by non-coded DNA (for instance, in order to obtain the DNA fingerprint with forensic or paternity purposes) can also be considered as health data. As scientists still do not have a great knowledge of the purpose of this non-coding DNA, the *Agencia Española de Protección de Datos* ("Spanish Data Protection Authority") has set out that this information must be processed as health data. Cf. AGENCIA ESPAÑOLA DE PROTECCIÓN DE DATOS, *Tratamiento de datos genéticos para la localización de personas desaparecidas o en investigación criminal*, 2000. See also, NICOLÁS JIMÉNEZ, *La protección jurídica de los datos genéticos de carácter personal*, pp. 85 ff.

[46] According to Thomas H. MURRAY, "Genetic Exceptionalism and 'Future Diary': Is Genetic Information Different from Other Medical Information?", in Mark A. Rothstein (Ed.), *Genetic Secrets: Protecting privacy and Confidentiality in the Genetic Era*, Yale University Press, New Haven, 1997, pp. 60 f., genetic exceptionalism means «the claim that genetic information is sufficiently different from other kinds of health-related information that is deserves special protection or other exceptional measures».

See also, Koichi SETOYAMA, "Privacy of Genetic Information", *Osaka University Law Review*, No 52 (2005), pp. 94 ff.; GERARDS/JANSEN, "Regulation of Genetic and Other Health Information in a Comparative Perspective", pp. 341 ff.

[47] EXPERT GROUP ON GENETIC DATA, *Ethical, legal and social aspects of genetic testing: research, development and clinical applications*, p. 42. The Group believes, therefore, "that genetic exceptionalism is both scientifically unjustified and not helpful in addressing ethical and societal issues" (at p. 43). In the same sense, MURRAY, "Genetic Exceptionalism and 'Future Diary'", pp. 64 ff.

a) They can be predictive of genetic predispositions concerning individuals;

b) They may have a significant impact on the family, including offspring, extending over generations, and in some instances on the whole group to which the person concerned belongs;

c) They may contain information the significance of which is not necessarily known at the time of the collection of the biological samples;

d) They may have cultural significance for persons or groups.

Certainly, the aforementioned features may, in themselves, not be unique to genetic information, but may also be relevant to certain types of non-genetic health information. Nevertheless, genetic information can be considered different from other health information, as all factors mentioned above appear in combination[48].

Thus, genetic data, although medical data (and for that reason, part of medical information), seem to have specific dimensions which are not necessarily common to all medical information[49]. Therefore, it has been argued that genetic information should be treated differently and given special legal protection[50]. However, it cannot be flatly rejected that similar protection should also be given to other kinds of predictive health information[51].

6. Genetic testing and genetic counselling. The duty to provide "non-directive" counselling

It is important to distinguish genetic counselling from clinical genetics services and genetic testing or screening[52]. The former is a

[48] Cf. GERARDS/JANSEN, "Regulation of Genetic and Other Health Information in a Comparative Perspective", p. 352, where they affirm that «genetic information in exceptional in that it shows a unique combination of features».

[49] See DATA PROTECTION WORKING PARTY, *Working Document on Genetic Data*, pp. 4 f.; Guilherme FREIRE FALCÃO DE OLIVEIRA, "Juridical implications of genome knowledge (Part II)", *Law and the Human Genome Review*, No 7 (1997), p. 88; Loane SKENE, "Patients' Rights or Family Responsibilities? Two Approaches to genetic Testing", *Medical Law Review*, No 6 (1998), pp. 5 ff.; OTLOWSKI, "Protecting Genetic Privacy in the Research Context", p. 92 f.;

[50] DATA PROTECTION WORKING PARTY, *Working Document on Genetic Data*, p. 4; George ANNAS, "Genetic Privacy: There Ought to Be a Law," *Texas Review of Law and Politics*, No 4 (1999), pp. 9 ff. According to FREIRE FALCÃO DE OLIVEIRA, "Juridical implications of genome knowledge", p. 88, «in view of these special characteristics, tighter confidentiality must be guaranteed».

[51] Cf. GERARDS/JANSEN, "Regulation of Genetic and Other Health Information in a Comparative Perspective", pp. 351 f.

[52] *Vid.* Barbara Bowles BIESECKER/Theresa M. MARTEAU, "The future of genetic counselling: an international perspective", Nature Genetics, No 22 (1999), p. 133.

communication and, in some cases, a psychotherapeutic process, while the latter are diagnostic or prognostic services. Current clinical genetics services and accompanying genetic counselling commonly involve the diagnosis (and prediction) of what are for the most part rare and untreatable conditions in fetuses, children and adults. Genetic diagnosis has traditionally been based on physical examination or family history but increasingly relies on molecular testing.

According to Article 12 CHRB, "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 subject to appropriate genetic counselling*" (emphasis added). This is not new. Recommendation R (92) 3 also states that "any genetic testing and screening procedure should be accompanied by appropriate counselling, both before and after the procedure" (Principle 3).

Genetic counselling can be defined as "a procedure to explain the possible implications of the findings of genetic testing or screening, its advantages and risks and where applicable to assist the individual in the long-term handling of the consequences; It takes place before and after genetic testing and screening" (Article 2.xiv IDHGD).

The development of new diagnostic methods has resulted in major advances in our ability to detect microscopic and submicroscopic chromosome abnormalities as well as single gene disorders. This often allows to provide a person with accurate information regarding the aetiology, prognosis, the risk of recurrence and the options available to deal with such finding. Genetic counselling is important because this information should be communicated in simple language, with care and sensitivity, so that the person or the family can make decisions that are fully informed.

Thus, the main goal of genetic counselling is to help individuals or families understand or cope with genetic disease, not to decrease the incidence of genetic disease[53]. In addition, the counsellor should adopt a non-directive approach. In this respect, Principle 3 of the Recommendation R (92) 3 states that "such counselling must be non-

[53] EXPERT GROUP ON GENETIC DATA, *Ethical, legal and social aspects of genetic testing: research, development and clinical applications*, p. 78.

directive"[54]. The information to be given should include the pertinent medical facts, the results of tests, as well as the consequences and choices. It should explain the purpose and the nature of the tests and point out possible risks. It must be adapted to the circumstances in which individuals and families receive genetic information[55].

7. Right to privacy and right to data protection

Privacy refers to the general interest in control of one's private sphere, broadly conceived. The concept of "privacy" is huge and complex and has many meanings. Allen identifies four dimensions of privacy[56]: a) informational privacy; b) physical privacy; c) decisional privacy; and d) proprietary privacy. In summary, *informational privacy* concerns access to personal information; *physical privacy* concerns access to persons and personal spaces; *decisional privacy* concerns governmental and other third party interferences with personal choices; and *proprietary privacy* relates to the appropriation and ownership of interests in human personality.

This work focuses on "informational privacy" interests, that is, the interest a person has in controlling access to and use of their personal information[57]. This is the aspect of privacy which is most commonly referred to in the discussions about genetic privacy. As we will mention later, this sphere of the right to privacy has turned towards a specific right, the right to data protection.

[54] See also IDHGD, Article 11.

[55] The Expert Group on Genetic Data, *Ethical, legal and social aspects of genetic testing: research, development and clinical applications*, p. 79, notes that "[t]he most difficult task for the counsellor is to communicate the precise meaning of genetic risk ... Different methods – verbal, numerical and graphical – have been developed to communicate risk in an understandable way to the person being counselled ... [T]he provision of simple, printed information that can be consulted by the individual after leaving the counselling session has been shown to be essential. Moreover, the existence of genetic support groups for the particular disease or problem, to which the person can be referred will, in many cases, provide information complementary to that given during the counselling session and can provide further support in the understanding of, or the coping with, a genetic problem". See also, Carlos M. Romeo Casabona, "Legal Aspects of Genetic Counselling", *Law and Human Genome Review*, No 1 (1994), pp. 164 ff.

[56] Anita L. Allen, "Genetic Privacy: Emerging Concepts and Values", in Mark A. Rothstein (Ed.), *Genetic Secrets: Protecting privacy and Confidentiality in the Genetic Era*, Yale University Press, New Haven, 1997, pp. 41 ff. This sorting is also accepted by Otlowski, "Protecting Genetic Privacy in the Research Context", p. 88; Setoyama, "Privacy of Genetic Information", pp. 81 ff.

[57] Mark A. Rothstein, "Genetic Secret: A Policy Framework", in Mark A. Rothstein (Ed.), *Genetic Secrets: Protecting privacy and Confidentiality in the Genetic Era*, Yale University Press, New Haven, 1997, p. 453; Otlowski, "Protecting Genetic Privacy in the Research Context", p. 88.

The right to privacy is a well established right in the European tradition. From a European perspective, we should begin by quoting Article 8 of the European Convention on Human Rights (which has its precedent in Article 12 of the Universal Declaration of Human Rights, of 10 December 1948)[58]: "1. Everyone has the right to respect for his private and family life, his home and his correspondence. 2. There shall be no interference by a public authority with the exercise of this right except such as is in accordance with the law and is necessary in a democratic society in the interests of national security, public safety or the economic well-being of the country, for the prevention of disorder or crime, for the protection of health or morals, or for the protection of the rights and freedoms of others".

Here we have all the essentials concerning the right to privacy: on the one hand, society must guarantee respect for privacy as essential to the individual's development. On the other hand, many limitations are listed in the Convention itself, so that privacy is not an absolute but a relative right. In particular, the protection of health is mentioned.

The right to privacy in relation to health has been established in Article 10 CHRB, in the following terms: "Everyone has the right to respect for private life in relation to information about his or her health". In similar terms, Recommendation R (97) 5 states that "The respect of rights and fundamental freedoms, and in particular of the right to privacy, shall be guaranteed during the collection and processing of medical data" (Principle 3.1).

More recently, the right to data protection, considered as a right linked but different to the right privacy has been recognized at the

[58] Article 12 of the Universal Declaration of Human Rights states that "No one shall be subjected to arbitrary interference with his privacy, family, home or correspondence, nor to attacks upon his honour and reputation. Everyone has the right to the protection of the law against such interference or attacks".

In addition, other international documents on human rights also deal with this issue. For instance, International Covenant on Civil and Political Rights, of 16 December 1966, according to which, "1. No one shall be subjected to arbitrary or unlawful interference with his privacy, family, home or correspondence, nor to unlawful attacks on his honour and reputation. 2. Everyone has the right to the protection of the law against such interference or attacks" (Article 17). And more recently, the UNESCO's Universal Declaration on Bioethics and Human Rights, of 19 October 2005, states in its Article 9 that "The privacy of the persons concerned and the confidentiality of their personal information should be respected. To the greatest extent possible, such information should not be used or disclosed for purposes other than those for which it was collected or consented to, consistent with international law, in particular international human rights law".

European level. While the Convention for the Protection of Individuals with regard to Automatic Processing of Personal Data, refers to the "right to privacy, with regard to automatic processing of personal data relating to him", the Charter of Fundamental Rights of the European Union recognizes the "protection of personal data" as a right in itself (Article 8)[59]. And Directive 95/46/EC develops this right extensively.

Several principles of data protection come from this right[60]. Among them, in relation to genetic data, the following will be considered: a) the principle of self-determination, that is, freedom and autonomy of the individual, which also implies initial freedom of choice and consent before undergoing genetic testing and even before providing biological samples for the test[61]; b) the right of access to information.

7.1. Right to Self-determination and Consent

Each person is entitled to decide to whom, when, and to what extent personal information relating to him or her can be processed. Thus, the processing on the information obtained by means of genetic testing should be prohibited unless consent is given by the data subject.

According to Article 8.2.a) of Directive 95/46/EC, the data subject must give his or her explicit consent to the processing of personal data concerning health[62]. Considering the extremely singular characteristics of genetic data and their link to information that may reveal the health condition or the ethnic origin of the subject, they can be considered as falling within the scope of Article 8 Directive 95/46/EC[63].

[59] Charter of Fundamental Rights of the European Union, Article 8: "1. Everyone has the right to the protection of personal data concerning him or her. 2. Such data must be processed fairly for specified purposes and on the basis of the consent of the person concerned or some other legitimate basis laid down by law. Everyone has the right of access to data which has been collected concerning him or her, and the right to have it rectified. 3. Compliance with these rules shall be subject to control by an independent authority". The more general "right to respect for his or her private and family life, home and communications" is recognized in Article 7. See DATA PROTECTION WORKING PARTY, *Opinion 4/2007 on the concept of personal data*, p. 7.

[60] See NICOLÁS JIMÉNEZ, *La protección jurídica de los datos genéticos de carácter personal*, pp. 176 ff.

[61] See Recommendation No. R (92) 3, Principle 8; IDHGD, Article 8; Recommendation Rec (2006) 4 on research on biological materials of human origin, of 15 March 2006.

[62] However, domestic legislation of the Member States can foresee some exceptions to this principle. For instance, when the processing is necessary to protect the vital interests of the data subject or of another person where the data subject is physically or legally incapable of giving his consent. See Directive 95/46/EC, Article 8.2 and 3.

[63] See DATA PROTECTION WORKING PARTY, *Working Document on Genetic Data*, pp. 5 and 7.

Equally, Principle 5 of Recommendation R (92) 3 states that "the provision of genetic services should be based on respect for the principle of self-determination of the persons concerned. For this reason, any genetic testing, even when offered systematically, should be subject to their express, free and informed consent". And Article 8 IDHGD states that "prior, free, informed and express consent, without inducement by financial or other personal gain, should be obtained for the collection of human genetic data, human proteomic data or biological samples, whether through invasive or non-invasive procedures, and for their subsequent processing, use and storage, whether carried out by public or private institutions"[64].

When a person is incapable of giving informed consent, authorization should be obtained from the legal representative[65]. The legal representative should have regard to the best interest of the person concerned[66]. In any case, an adult not able to consent should as far as possible take part in the authorization procedure[67].

In regard to Recommendation R (92) 3, its Principle 5 states that the testing of minors, persons suffering from mental disorders and adults placed under limited guardianship should be subject to special safeguards. In particular, testing of these persons for diagnostic purposes should be permitted only when this is necessary for their own health[68] or if the information is imperatively needed to diagnose the existence of a genetic disease in family members[69]. That means

[64] See also Recommendation (97) 5, Principle 6.1.

[65] "Legal representative" refers to a person provided for by law to represent the interests of, and/or take decisions on behalf of, a person who does not have the capacity to consent. That is the case of a parent o guardian, for instance.

[66] See IDHGD, Article 8.b); and Recommendation (97) 5, Principle 5.5; Charter of Fundamental Rights of the European Union, Article 8.2.

[67] Cf. *Explanatory Report* on CHRB, paragraphs 45 and 46 ("the participation of adults not able to consent in decisions must not be totally ruled out. This idea is reflected in the obligation to involve the adult in the authorisation procedure whenever possible. Thus, it will be necessary to explain to them the significance and circumstances of the intervention and then obtain their opinion").

[68] One good example of this is the genetic testing of newborn babies (neonatal screening) for treatable diseases such as phenylketonuria and hypothyroidism (available in all UE countries). As we said above, the testing of children must be strictly limited to those cases in which a diagnosis is important for disease management or therapy. See EXPERT GROUP ON GENETIC DATA, *Ethical, legal and social aspects of genetic testing: research, development and clinical applications*, p. 75 ("susceptibility screening is not justified as the benefit is remote and often uncertain. Carrier screening can also wait ultil the child can make his or her own decision").

[69] See also, IDHGD, Article 8. c) and d).

that genetic testing on newborns and children should be confined to treatable disorders, for which early treatment has a substantial positive impact on the health status and where delay would reduce benefits[70].

When human genetic data are collected for medical purposes, consent may be withdrawn by the person concerned unless such data are irretrievably unlinked to an identifiable person (Article 9. a) IDHGD). When a person withdraws consent, the person's genetic data should no longer be used (Article 9. b) IDHGD).

There is some debate as to whether the anonymization process of itself requires the affected person's prior consent or whether it is necessary at least to inform him or her of this process. According to Directive 95/46/EC, processing of personal data "shall mean *any operation or set of operations which is performed upon personal data*, whether or not by automatic means, such as collection, recording, organization, storage, adaptation or alteration, retrieval, consultation, use, disclosure by transmission, dissemination or otherwise making available, alignment or combination, blocking, erasure or destruction" (Article 2. b)) (emphasis added).

There is no doubt that this paragraph includes the process of anonymization, since personal data is subjected to an alteration or mutilation in order to avoid the identification of the person from whom this data originates. The process of anonymization itself is therefore still an act of data processing[71]. This means that, until anonymization is carried out in fact, the data will still be considered as personal data so the principles of data protection will be applicable.

[70] H.D.C. Roscam Abbing, "Genetic Information and third party interests. How to find the right balance?", *Law and the Human Genome Review*, No 2 (1995), p. 37.

[71] Romeo-Casabona, "Anonymization and Pseudonymization", p. 43.

See also the document entitled *European Standards on Confidentiality and Privacy in Healthcare* (2006), whose paragraph 3.3.5 sets out that "the Data Protection Directive requires data subjects to be informed of the purposes of all processing of personal data and rendering data anonymous is itself a process performed on personal data". These European Standards on Confidentiality and Privacy in Healthcare were developed through the work of the EuroSOCAP Project (funded by European Commission). Document available in: www.eurosocap.org.

7.2. Right of access to information

Articles 10 and 11 of Directive 95/46/EC state that the data subject has a right to receive information from the controller[72] (or his representative), both when the data is collected directly from said data subject and when the data has not been obtained from said data subject.

This right is exercised through the so-called *right of access*[73], that is, the right to obtain from the controller without constraint at reasonable intervals and without excessive delay or expense: a) confirmation as to whether or not data relating to him are being processed; and b) communication in an intelligible form of the data undergoing processing and of any available information as to their source[74]. Given the sensitivity of genetic data, the right to information is particularly relevant in the context of processing of such data[75].

8. Right to know and right not to know

Article 10.2 CHRB, provides that "everyone is entitled to know any information collected about his or her health ...". However, the last paragraph of Article 10 sets out that in exceptional cases, domestic law may place restrictions on the right to know or not to know in the interests of the patient's health[76]. There might be a situation where the harm to the data subject which is expected to be caused by the information is such that it clearly justifies withholding the information or part of it (for example a prognosis of death which might, in certain cases if immediately passed on to the patient, seriously worsen his or her condition). This is the so-called "therapeutic exception"[77].

In respect of the right to know, special mention should be made of *unexpected findings*. It is not uncommon in medicine, during operations

[72] According to Article 2 (d) of Directive 95/46/EC, "controller" shall mean the natural or legal person, public authority, agency or any other body which alone or jointly with others determines the purposes and means of the processing of personal data.

[73] Carlos de Sola, "Privacy and Genetic Data. Cases of Conflict (I)", *Law and the Human Genome Review*, No 1 (1994), p. 177.

[74] Cf. Directive 95/46/EC, Article 12. See also Recommendation R (97) 5, Principle 8.1; IDHGD, Article 13; Charter of Fundamental Rights of the European Union, Article 8.2.

[75] Data Protection Working Party, *Working Document on Genetic Data*, p. 6.

[76] Article 10.3 CHRB: "In exceptional cases, restrictions may be placed by law on the exercise of the rights contained in paragraph 2 in the interests of the patient".

[77] Roscam Abbing, "Genetic Information and third party interests", p. 39. In general, such a therapeutic exception is only justifiable in very exceptional circumstances.

or tests, for personal data of varying importance to be discovered in addition to the information actively sought. For example, a person may be tested for one disease and found to be suffering from another; or a genetic test carried out for medical purposes may reveal that the genetic relationship is not the same as the legal one.

In conformity with the right to know, subjects should be informed of "unexpected findings" when they are of medical relevance[78]. However, except in some clear situations, and always that the patient has not been informed of this eventuality, the physician will face important doubts in taking a decision and whether to inform the patient about any unexpected findings. In this decision not only objective factors must be taken into account, such as the possibility of therapy, but also subjective ones, such as the personality of the patient, the consequences of receiving this information, and other familiar circumstances.

Recommendation R (92) 3 points out that "In conformity with national legislation, unexpected findings may be communicated to the person tested only if they are of direct clinical importance to the person or the family. Communication of unexpected findings to family members of the person tested should only be authorised by national law if the person tested refuses expressly to inform them even though their lives are in danger" (Principle 11).

This position has been developed by Principle 8.4 of Recommendation R (97) 5. According to this Principle, the person subjected to genetic analysis must be informed of unexpected findings if the domestic law does not prohibit the giving of such information; or the person himself has asked for this information; or the information is not likely to cause serious harm to his/her health; or to his/her consanguine or uterine kin, to a member of his/her social family, or to a person who has a direct link with his/her genetic line, unless domestic law provides other appropriate safeguards. The person should also be informed if this information is of direct importance to him/her for treatment or prevention.

Thus, Recommendation R (97) 5 restricts the circumstances in which unexpected findings should not be communicated. In conclusion, the doubtful cases that remain at the moment of deciding on whether to report or not of an unexpected finding are very limited. The cases are

[78] Cf. DE SOLA, "Privacy and Genetic Data", p. 178.

those in which this information does not have health repercussions because the mutation found does not have a great significance or there is not a therapy or prevention known, or it is not going to be transmitted to the offspring. In these cases, it is not necessary to inform. That means that this duty to inform would not extend to data which were not directly medical, such as the discovery of the lack of a genetic link between father and son. But it must not be forgotten that these data can also be medically relevant and may be of considerable importance to the subject when taking important decisions of other kinds (for instance, when a donor for a transplant is required)[79].

And of course, such discoveries should be passed down to the patient when he or she was aware of this possibility and wanted to receive such information, whatever the content was. That is why the information provided at the time of the consent should indicate the possibility of revealing unexpected findings and the right of the person concerned to decide if he or she wants to be informed about them or not. In this respect, Recommendation R (97) 5 also states that "before a genetic analysis is carried out, the data subject should be informed about the objectives and *the possibility of unexpected findings*" (Principle 5.4) (emphasis added).

In addition to the right to information, or right to know, which also covers individual genetic information, reference tends to also be made to the reverse, i.e., the right "not to know". According to Article 10 IDHGD, "the person concerned has the right to decide whether or not to be informed of the results". In the same sense, Article 10.2 CHRB states that "the wishes of individuals not to be so informed shall be observed". Patients may have their own reasons for not wishing to know about certain aspects of their health. A wish of this kind must be observed.

The right not to know includes the right not to undergo genetic testing, so that the subject can avoid knowing whether he or she is carrying a genetic condition or whether he or she might do so in the future. This right is important because not every hereditary disease can be treated or even prevented at present. In many cases, the only certain prediction is that diseases will develop and that nothing whatever can be done to prevent or delay them. If there is no medical technique to cure a

[79] See NICOLÁS JIMÉNEZ, *La protección jurídica de los datos genéticos de carácter personal*, pp. 289 f.

specific genetic disease, such as Huntington's disease, some people may not want to know their future fate and short life span[80].

For this reason, a genetic test should be performed or offered only where the expected benefits for the individual outweight the potential risks[81]. That is to say, some kind of action must be available (treatment, prevention, reproductive choices). In effect, to make a person conscious of a genetic disease can help him or her to take important measures in the field of reproduction. That is especially significant in some populations highly affected by any concrete illness. In effect, some populations are known to have a higher frequency of a gene that is known to be associated with a disease. Genetic testing of couples in such populations would permit them to take their carrier status into account before planning a family[82].

However, in some circumstances, the right to know or not to know can be restricted in the patient's own interest or else on the basis of Articles 10.3 ("In exceptional cases, restrictions may be placed by law on the exercise of the rights contained in paragraph 2 in the interests of the patient") and 26.1 CHRB[83].

[80] A study developed in the US concludes that suicide rate for Huntington's disease sufferers is much greater than the national average. See Jane S. PAULSEN/Karin FERNEYHOUGH HOTH/Carissa NEHL/Laura STIERMAN, "Critical Periods of Suicide Risk in Huntington's Disease", *American Journal of Psychiatry*, No 162 (2005), pp 725–731 (http://www.huntington-assoc.com/Critical%20ab05.pdf).

[81] EXPERT GROUP ON GENETIC DATA, *Ethical, legal and social aspects of genetic testing: research, development and clinical applications*, p. 75.

[82] One example of population-based testing is the screening for the carrier status of 'thalasemia' in certain Mediterranean communities, where genes for this disease appear to be more frequent. While this testing is not technically mandatory, individuals are not permitted to marry without a pre-marriage certificate. And a pre-marriage certificate is not issued without the requisite of genetic testing.

See Minas G. HADJIMINAS, "The cyprus experience-screening to combat a serious genetic disease", *Ethics and Human Genetics*, Council of Europe Press, Strasbourg, 1994, pp. 26-48; Ann CAVOUKIAN, "Confidentiality issues in genetics: the need for privacy and the right 'not to know'", *Law and the Human Genome Review*, No 2 (1995), pp. 62 ff.; Luis ZARRALUQUI SÁNCHEZ-EZNARRIAGA, "Genetic testing and matrimony", in *The Human Genome Project: Legal Aspects*, Vol. I, Fundación BBV, Bilbao, 1995, pp. 391 ff.; Barbara PRAINSACK, "The Rise of Genetic Couplehood? A Comparative View of Premarital Genetic Testing", *BioSocieties*, No 1 (2006), pp. 17 ff.

In this respect, according to Recommendation No. R (92) 3 on genetic testing and screening for health care purposes, "(...) marriage requirements (...) should not be made dependent on the undergoing of genetic tests or screening" (principle 6.a)).

[83] Article 26.1 CHRB: "No restrictions shall be placed on the exercise of the rights and protective provisions contained in this Convention other than such as are prescribed by law

The *Explanatory Report* CHRB points out that "it may be of vital importance for patients to know certain facts about their health, even though they have expressed the wish not to know them. For example, the knowledge that they have a predisposition to a disease might be the only way to enable them to take potentially effective (preventive) measures. In this case, a doctor's duty to provide care, as laid down in Article 4 [CHRB], might conflict with the patient's right not to know. It could also be appropriate to inform an individual that he or she has a particular condition when there is a risk not only to that person but also to others ... At the same time, certain facts concerning the health of a person who has expressed a wish not to be told about them may be of special interest to a third party[84] ... In such a case, the possibility for prevention of the risk to the third party might, on the basis of Article 26, warrant his or her right taking precedence over the patient's right to privacy, as laid down in paragraph 1, and as a result the right not to know, as laid down in

and are necessary in a democratic society in the interest of public safety, for the prevention of crime, for the protection of public health or for the protection of the rights and freedoms of others").

Therefore to be compatible with the CHRB, any interference with the right to privacy must meet certain conditions. It must be "in accordance with the law", which means that any interference must have some basis in national law, and the law must be precise enough so that people can reasonably understand its requirements and consequences. It must be "necessary in a democratic society", which means that the interference must also both correspond to a "pressing social need" and be "proportionate to the legitimate aim pursued". See further, Sergio ROMEO MALANDA, "Relación del presente Convenio con otras disposiciones (Capitulo IX)", en Carlos María Romeo Casabona (ed.), *El Convenio de Derechos Humanos y Biomedicina: su entrada en vigor en el ordenamiento jurídico español*, Cátedra de Derecho y Genoma Humano-Comares, Bilbao-Granada, 2002, pp. 387 ff.

According to the *Explanatory Report* on CHRB, "The reasons mentioned in Article 26.1 should not be regarded as justifying an absolute exception to the rights secured by the Convention. To be admissible, restrictions must be prescribed by law and be necessary in a democratic society for the protection of the collective interest in question or for the protection of individual interests, that is the rights and freedom of others. These conditions must be interpreted in the light of the criteria established with regard to the same concepts by the case-law of the European Court of Human Rights. In particular, the restrictions must meet the criteria of necessity, proportionality and subsidiarity, taking into account the social and cultural conditions proper to each State. The term "prescribed by law" should be interpreted in accordance with the meaning usually given to it by the European Court of Human Rights, that is a formal law is not required and each State may adopt the form of domestic law it considers most appropriate" (paragraph 159).

84 See also EXPERT GROUP ON GENETIC DATA, *Ethical, legal and social aspects of genetic testing: research, development and clinical applications*, p. 81. For example, if a deceased grandparent is known to have suffered from Huntington's chorea and his grandchild asks to be tested for carrier status (with a view to making decisions about his reproductive choices, for example), the information that he is a carrier will also be information about the parent who did not want to know.

paragraph 2. In any case, the right not to know of the person concerned may be opposed to the interest to be informed of another person ..."[85].

9. Right to privacy and (the corresponding) duty of confidentiality

9.1. Privacy and confidentiality

As a starting point in this section, it is important to distinguish the terms "privacy" and "confidentiality". Although they are connected in some sense, they are not synonyms at all[86]. Privacy is a much broader concept, involving the right to be free from intrusions, or simply to be left alone. It involves the right to control one's own personal information (this perspective is also known as "informational self-determination")[87]. Confidentiality, however, is only one means of protecting information, usually in the form of keeping that information protected from disclosure[88]. Despite these distinctions, confidentiality is still the standard safeguard to protect privacy and medical information[89].

Each person is entitled to decide to whom, when, and to what extent personal information relating to him or her can be disclosed. Thus, the passing on the information obtained by means of genetic testing should be prohibited unless consent is given by the data subject or legal representative in the case of minors or legally incapacitated persons[90].

[85] *Explanatory Report* on CHRB, paragraph 70. The EXPERT GROUP ON GENETIC DATA, *Ethical, legal and social aspects of genetic testing: research, development and clinical applications*, p. 82, also analyses the case of a couple that reject doctor's advise to andergo a genetic testig. In its opinion, "the couple's decision to remain ignorant would be fully reasonable and in accordance with the principle of autonomy".

[86] See, for instance, Ann CAVOUKIAN, "Confidentiality issues in genetics: the need for privacy and the right 'not to know'", *Law and the Human Genome Review*, No 2 (1995), p. 53.

[87] Felix THIELE, "Genetic tests in the insurance system: criteria for a moral evaluation", *Poiesis Prax* (2003), pp. 193 f. («The right to informational self-determination (...) should enable an individual to make decisions concerning personal data without being exposed to the coercion of a third party»). See also NICOLÁS JIMÉNEZ, *La protección jurídica de los datos genéticos de carácter personal*, pp. 163 ff.

[88] Margaret OTLOWSKI, "Protecting Genetic Privacy: An Overview", in *Regulating the New Frontiers: Legal issues in Biotechnology*, Centre for Law and Genetics, Hobart-Melbourne, 2001, pp. 66 and 72, defines confidentiality as "a specific obligation arising in certain relationships whereby the recipient of personal information about another is under an obligation not to use that information for any purpose other than that for which the information was given"

[89] José ELIZALDE, "Confidentiality, Privacy and Genetic Data", in *The Human Genome Project: Legal Aspects*, Vol. I, Fundación BBV, Bilbao, 1995, p. 307.

[90] Carlos M. ROMEO CASABONA, "Genetic Information: Collection, Access and Use", in Baltimore (Ed.), *Frontiers of Life, Vol. IV: The Living World*, Academic Press, San Diego, 2001, p. 781.

In some cases, it may be necessary to share some information from a patient with other professionals involved (directly or indirectly) in his or her care. In this respect, Recommendation R (97) 5 states that "medical data may only be communicated to a person who is subject to the rules of confidentiality incumbent upon a health-care professional, or to comparable rules of confidentiality ..." (Principle 7.2). It refers to the so-called "shared confidentiality"[91], which means that the processing of medical data may only be performed by health-care professionals[92], who are bound by the profession's rule of confidentiality[93].

According to this, genetic data must also be protected from other persons even in case of a biological relative who seeks information concerning the possible presence in him or her of a pathological gene similar to that discovered in the data subject[94]. It is necessary to maintain confidentiality in these cases. Since the person disclosing the information will be the doctor, the answer is complicated by the very high value that our societies have placed on medical secrecy since time immemorial. Doctor-patient relations are based on trust, guaranteed by the doctor's duty not to disclose information of any kind about his or her patients. The latter would not confide in their doctor if they were afraid that he or she might then reveal their secrets[95]. So disclosure of confidential information not only breaches individual rights but undermines the very foundations of medical practice and therefore endangers a general interest[96].

In this respect, The European Court of Human Rights has held that "the protection of personal data, not least medical data, is of

[91] Ibid., p. 780.

[92] Directive 95/46/EC authorizes the processing of the data if it is required for the purposes of preventive medicine, medical diagnosis, the provision of care or treatment or the management of health-care services, and where those data are processed by a health professional (Article 8.3).

[93] See Santiago RIPOL CARULLA, "The protection of medical and genetic data in the Council of Europe's normative texts (Part I)'", *Law and the Human Genome Review*, No 5 (1996), p. 116.

[94] ROMEO CASABONA, "Genetic Information", p. 781; ROSCAM ABBING, "Genetic Information and third party interests", p. 40.

[95] DE SOLA, "Privacy and Genetic Data", p. 182; FREIRE FALCÃO DE OLIVEIRA, "Juridical implications of genome knowledge", p. 90; Ismini KRIARI-CATRANIS, "Genetic data and confidentiality, the Estonian experiment", *Revista de Derecho y Genoma Humano/Law and the Human Genome Review*, No 19 (2003), p. 148.

[96] In addition, Madison POWERS, "Justice and Genetics: Privacy Protection and the Moral Basis of Public Policy", in Mark A. Rothstein (Ed.), *Genetic Secrets: Protecting privacy and Confidentiality in the Genetic Era*, Yale University Press, New Haven, 1997, p. 358, notes that when genetic information obtained from one patient or subject is revealed to other

fundamental importance to a person's enjoyment of his or her right to respect for private and family life as guaranteed by Article 8 of the Convention (art. 8). Respecting the confidentiality of health data is a vital principle in the legal systems of all the Contracting Parties to the Convention. It is crucial not only to respect the sense of privacy of a patient but also to preserve his or her confidence in the medical profession and in the health services in general. Without such protection, those in need of medical assistance may be deterred from revealing such information of a personal and intimate nature as may be necessary in order to receive appropriate treatment and, even, from seeking such assistance, thereby endangering their own health and, in the case of transmissible diseases, that of the community"[97].

According to Article 14. b) IDHGD, human genetic data should not be disclosed or made accessible to third parties, in particular, employers, insurance companies, educational institutions and the family, except for an important public interest reason in cases restrictively provided for by domestic law or where the prior, free, informed and express consent of the person concerned has been obtained[98]. "Public interest" should be here understood in the sense of Article 26.1 CHRB, that is to say, for reasons of public safety, for the prevention of crime, for the protection of public health or for the protection of the rights and freedoms of others.

In practice, it is likely that the subject will consent to the disclosure of the information to members of the family or may even provide the information himself or herself[99]. However, it is important in the genetic counselling process to stress to the subject the importance for others to partially relieve the doctor of his or her professional duty of secrecy for the subject to give the information.

family members without his/her consent, «the ability of persons to shape their most intimate relationship is compromised, and the trust and patterns of usual communication within families can be compromised».

[97] *Z. v. Finland*, 25 February 1997 (paragraph 95). Cf. also, *M.S. v. Sweden*, 27 August 1997 (paragraph 41).

[98] See also Principle 9 of Recommendation R (92) 3: "Persons handling genetic information should be bound by professional rules of conduct and rules laid down by national legislation aimed at preventing the misuse of such information and, in particular, by the duty to observe strict confidentiality. Personal information obtained by genetic testing is protected on the same basis as other medical data by the rules of medical data protection".

[99] Cf. National Consultative Ethics Committee for Health and Life Sciences (France), *Opinion No 76 regarding the obligation to disclose genetic information of concern to the family in the event of medical necessity* (2003), p. 5; Moniz, "Privacy and intra-family communication of genetic information", p. 108.

9.2. Conflicts of Duties and Interests. Privacy and Intra-family Communication of Genetic Information

Genetic medicine can give rise to a variety of conflicts of interests. On the one hand, the data subject will have an interest in their protection. On the other hand, certain legal entities or individuals may show an interest of apparently equal legitimacy, although in conflict with the first, in gaining knowledge or making use of the very data which the subject would like to safeguard[100]. In the field of medical genetics, the most obvious situation of conflict is probably that between a subject and his or her blood relatives.

Genetic features are transmissible from one generation to another. If a person carries a gene responsible for a certain disease it is virtually certain that the gene will also be found in other members of that person's close family. From the medical standpoint, the patient is the entire family rather than just one single person[101]. Whenever a gene is discovered in a given individual, therefore, the question arises as to whether this information should be communicated to the individual's relatives for the purposes of diagnosis, prevention or therapy, etc. However, from a legal point of view, the data subject is only the individual person whose DNA is being analysed[102].

In the huge majority of cases, information can be passed on without much difficulty. As noted above, the subject himself or herself will agree to inform close family members. In some circumstances, however, problems will arise if the subject refuses to do so[103]. Although infrequent, such cases

[100] These conflicts may be classified in the following categories, according to the person or institution seeking access to the data: a) Biological relatives might unknowingly be healthy carriers of the same genetic anomaly as the subject and consequently have a direct interest in the information; b) Legal entities or individuals have entered or plan to enter into a contractual relationship with the subject, especially an employment, service-related or insurance contract; c) The use of genetic data may be required by society as a whole (collective interest), for example as a vital clue in identifying the perpetrator of a crime; d) The advancement of medical research may be dependent on the greatest possible knowledge of data relating to subjects belonging to families within certain hereditary diseases occur. See de Sola, "Privacy and Genetic Data", p. 175.

[101] de Sola, "Privacy and Genetic Data", pp. 179 f.; Freire Falcão de Oliveira, "Juridical implications of genome knowledge", p. 88; Moniz, "Privacy and intra-family communication of genetic information", p. 109.

[102] See Freire Falcão de Oliveira, "Juridical implications of genome knowledge", p. 89 («[genetic] information refers only to the subject and hence the right to privacy can be assured»).

do occasionally arise, sometimes because the subject does not get on with his or her family or, more often, because the subject would rather that his or her relatives did not find out that he or she is a carrier of the gene[104].

If an individual refuses to inform relatives of any relevant genetic data, we can wonder whether doctor has a duty to inform the interested parties. This is a frequent dilemma in medicine and there is no universal answer to it. In fact, there are two different approaches relating to the passing on of information to relatives[105]: the *legal model* and the *medical model*. The legal model is based on patients' right to privacy, based on the idea that each person has the right to control their own body and genetic information; while the medical model rejects the language of individual rights and instead stresses the need to treat patients who have risks of a genetic nature.

The so-called legal model has prevailed in Europe so far, based on the preservation of human rights, particularly the right to privacy, in defence of the principle of patient autonomy and self-determination. As mentioned above, in the countries that have adopted the "legal model", doctors may only disclose such information with the express consent of the patient, even if a family member is at risk[106]. This does not mean that doctors do not have the possibility of disclosing this information in very closely defined circumstances. In fact, they are allowed to. This is not because a duty to forewarn exists, but because there is a justification for the breach of confidentiality[107].

[103] Actually, given the nature of genetic information, it has been argued as to whether there is an obligation (legal duty imposed by civil law or even criminal law) to notify genetic information to members of the family. See in this respect, FREIRE FALCÃO DE OLIVEIRA, "Juridical implications of genome knowledge", pp. 89 f., according to whom, a «recognition of a legal duty to notify family members would (...) be a curtailment of the individual's right to privacy»; Harold EDGAR, "Is there a legal duty to disclose genetic characteristics to a future spouse?", in *The Human Genome Project: Legal Aspects*, Vol. I, Fundación BBV, Bilbao, 1995, pp. 360 ff.; NATIONAL CONSULTATIVE ETHICS COMMITTEE FOR HEALTH AND LIFE SCIENCES (France), *Opinion n° 76 regarding the obligation to disclose genetic information of concern to the family in the event of medical necessity* (2003).

[104] DE SOLA, "Privacy and Genetic Data", p. 180.

[105] SKENE, "Patients' Rights or Family Responsibilities", pp. 1 ff.

[106] On the contrary, when we refer to the medical model, the general principle is not respect for the right to privacy, but respect for each person's health. Consequently, genetic information must be shared between family members and under no circumstances can this be prevented by the patient. According to this model, a person does not have the right to control his o her own genetic information, and doctors have a duty to disclose it.

The CHRB offers no precise solution to this dilemma. It only provides the general guidance set out in Article 10.1 in conjunction with Article 26. The former lays down the right to privacy ("Everyone has the right to respect for private life in relation to information about his or her health"), whereas Article 26.1 states that there *may* be exceptions to this right when necessary to protect the rights of others.

The question of when these exceptions are justified and when they are not remains unanswered. Since there is no general answer, all that can be said is that the solution will depend on the circumstances surrounding each case[108]. The only possible criterion is an assessment of the respective consequences of each solution, bearing in mind in particular the seriousness of the disease in question[109'110].

Thus, "exceptional circumstances, whereby disclosure could prevent serious harm to the health of the relative, and provided there are no other less-intrusive alternatives with respect to the privacy of the patient,

[107] MONIZ, "Privacy and intra-family communication of genetic information", p. 111. Cf. *European Standards on Confidentiality and Privacy in Healthcare* (2006), paragraph 3.4.2.

The NATIONAL CONSULTATIVE ETHICS COMMITTEE FOR HEALTH AND LIFE SCIENCES (France), *Opinion n° 76 regarding the obligation to disclose genetic information of concern to the family in the event of medical necessity* (2003), maintains a much more strict position with regard to the duty of confidentiality. It says that "the doctor will never take the initiative of warning family members unless the proband [the data subject] request him to do so" (at p. 8), although he or she must do everything in his power to convince a patient that his or her family should be informed "but he must not step in personally and breach medical confidentiality" (at p. 6). Cf. also, Virgine COMMIN/Emmanuelle RIAL-SEBBAG, "UNESCO and Council of Europe's contribution to the question of the confidentiality of the genetic information of an individual towards his family, in the case of genetic tests", in Marcelo Palacios (Ed.), *IV World Conference on Bioethics: summary of Lectures and oral presentations*, Sociedad Internacional de Bioética, Gijón, 2005, p. 474.

108 DATA PROTECTION WORKING PARTY, *Working Document on Genetic Data*, p. 9 ("Given the complexity of the issues described above, the Working Party takes the view at this stage that consideration should be given to a case by case approach in deciding how to address possible conflicts between the interests of the data subjects and those of their biological family").

[109] DE SOLA, "Privacy and Genetic Data", p. 182.

[110] A case was addressed in this respect in Italy, in 1999, through a decision issued by the *Garante per la Protezione dei Dati Personali* (Personal Data Protection Authority), which granted a lady the possibility to access her father's genetic data although the latter had denied his consent. This request was granted by considering that the father's right to privacy was to be overridden by the lady's right to health – the latter meaning her "psychological and physical well-being". The lady had requested disclosure of the data to carry out a genetic test and subsequently take a fully informed reproductive decision – upon assessing the risk of transmitting a genetic disease that affected her father. See DATA PROTECTION WORKING PARTY, *Working Document on Genetic Data*, p. 9.

may justify a breach of confidentiality in the doctor-patient relationship, and disclosure of the information by the health professional against the wishes of a patient"[111].

In other words, before deciding whether or not to pass on the subject's personal information to the rest of the family against his or her wishes, doctors must not only weigh up the private interests at stake (the subject's right to privacy *versus* the family members' right to health) but must also take into consideration the loss of confidence in the medical profession that their action could cause.

Thus, having established that a person has rights over his or her own genetic information, and that said rights must prevail over the interests of others, it is nevertheless possible to consider the rights of others when their health or lives are seriously affected. This protection of essential rights of third parties can be achieved on grounds of the *conflict (or collision) of duties*[112] or, as the case may be, the *state of necessity*[113], institutions which exist in the majority of legislations[114].

In our opinion, the above-mentioned approach is the correct way to resolve this conflict between the protection of personal data, on the one hand, and the protection of health, on the other, when disclosure of

[111] EXPERT GROUP ON GENETIC DATA, *Ethical, legal and social aspects of genetic testing: research, development and clinical applications*, p. 80. See also, Recommendation No. R (92) 3, Principle 9 ("In the case of a severe genetic risk for other family members, consideration should be given, in accordance with national legislation and professional rules of conduct, to informing family members about matters relevant to their health or that of their future children"); Recommendation No. R (97) 5 on the Protection of Medical Data, principle 7.3.

However, the right to privacy will not be necessarily at stake because it is not always necessary to reveal the identity of the data subject when there are contacts with relatives. See in this respect, ROSCAM ABBING, "Genetic Information and third party interests", p. 39.

[112] In order to use the conflict of duties solution, it is necessary to first determine the obligation of information towards others on the part of the person who performed or proposed the tests (e.g. a doctor in relation to his or her patient). That is to say, if the doctor is simultaneously doctor to other family members of the patient in question, then he or she may have the duty to communicate the fact to them.

[113] When the clinician is doctor only to the patient having the test. In these cases, the doctor does not have to violate confidentiality, but when he or she does so, taken into account certain clearly defined circumstances, his or her conduct is not consider illicit.

[114] ROMEO CASABONA, "Genetic Information", p. 784. See also Herman NYS/Carlos M. ROMEO CASABONA/Christophe DESMET, "Legal aspects of prenatal testing for late-onset neurological diseases", in G. Evers-Kiebooms/M. W. Zoetewelj/P. S. Harper (Eds), *Prenatal testing for late-onset neurological diseases*, Bios, UK, 2002, p. 98; ROSCAM ABBING, "Genetic Information and third party interests", pp. 40 ff.

confidential information is required to achieve this aim. In fact, as we have noted, this solution has been widely supported at different levels in the European context.

However, the recently adopted *Opinion 4/2007 on the concept of personal data*, by the Data Protection Working Party, makes us put this assertion in question. In effect, in this Opinion the Working Party has provided guidance on the way in which the concept of personal data in Directive 95/46/EC and related community legislation should be understood and how it should be applied in different situations. The Working Party's analysis is based on the four main "building blocks" that can be distinguished in the definition of "personal data": i.e. "any information", "relating to", "an identified or identifiable", "natural person".

One of the key findings of the Working Party is that the second building block ("relating to") has been too often overlooked, and that it should play a crucial role in determining the substantive scope of the concept as a whole, especially in relation to objects and new technologies. The Opinion provides three alternative elements (namely, content, purpose or result) for determining whether information "relates to" an individual[115]. These three elements (content, purpose, result) must be considered as alternative conditions, and not as cumulative ones.

The Opinion 4/2007 provides the following guidance with regard to the third "result" element: "A third kind of 'relating' to specific persons arises when a "result" element is present. Despite the absence of a "content" or "purpose" element, data can be considered to "relate" to an individual because their use is likely to have an impact on a certain person's rights and interests, taking into account all the circumstances surrounding the precise case. It should be noted that it is not necessary that the potential result be a major impact. It is sufficient if the individual may be treated differently from other persons as a result of the processing of such data"[116].

On this basis, genetic data could be considered to be personal data, not only of the person source of the sample, but also of those members of his or her biological family. As was pointed out above, the IDHGD itself

[115] See DATA PROTECTION WORKING PARTY, *Opinion 4/2007 on the concept of personal data*, pp. 9 ff.

[116] DATA PROTECTION WORKING PARTY, *Opinion 4/2007 on the concept of personal data*, p. 11.

states that human genetic data "may have a significant impact on the family, including offspring, extending over generations"[117]. This also seems to be the understanding of the Working Party, because in another extract from this document, in relation to the data on dead persons, they say that "the information on dead individuals may also refer to living persons. For instance, the information that the dead Gaia suffered from haemophilia indicates that her son Titius also suffers from the same disease, as it is linked to a gene contained in the X-chromosome. Thus, where the information which is data on the dead can be considered to relate at the same time also to the living and be personal data subject to the Directive, the personal data of the deceased may indirectly enjoy the protection of data protection rules"[118].

We support the Working Party's approach in adopting a broad interpretation of the concept of "personal data", given that the ultimate purpose of the rules contained in the Directive 95/46/EC is to protect the fundamental rights and freedoms of natural persons, and in particular their right to privacy. Nevertheless this should not be done in such a way that surpasses the barrier of logic and common sense, making the concept impractical. Regarding genetic data, if we apply strictly the wide interpretation given by the Working Party, then we should admit that all the members of the biological family of the tested subject (parent, children, siblings, etc.)[119] must be considered as "data subjects", which means not only that they can have access to all data, but also that the must consent every act of processing. That is not acceptable, simply because it is not practicable. For this reason, it is argued that the "result" element in the interpretation of the element "related to", must be understood restrictively, and that access to genetic information of a third party should only be possible when it is in accordance with the above-mentioned rules of conflict resolution.

[117] IDHGD, Article 4.a).

[118] DATA PROTECTION WORKING PARTY, *Opinion 4/2007 on the concept of personal data*, p. 22. However, to be granted access to data of a deceased person it is not required to be considered the data subject. As a dead person is not a "natural person", information relating to dead individuals is in principle not to be considered as personal data. Even though the data of the deceased may still indirectly receive some protection, it does nor come from the right to data protection, so that it is possible that some national legislator may decide to grant access to those data in some circumstances without failing to fulfil the principles of data protection. So that happens in Spain, where Article 48.2 of Law 14/2007 on Biomedical Research allows a member of the biological family to have access to data of the dead person if they are of medical interest for him or her.

[119] They all are easily identifiable natural persons.

10. Conclusion

The European regulatory system is made up of a complex web of legislation and other regulations, which at first sight seem to lack consistency. However, this analysis has shown that there is in fact coherence in the broad themes associated with genetic testing and the use of genetic information across Europe. Some of the key principles are highlighted below.

Although genetic information fits within the definition of medical information, genetic data seem to have specific dimensions which are not necessarily common to all medical information. Therefore, it is necessary to treat genetic information differently and give it special legal protection. For example, tests should be performed only for health purposes or for scientific research linked to health purposes, and must be accompanied by genetic counselling to help individuals or families understand or cope with genetic disease. The term "genetic information" must be understood broadly, encompassing data also available through means other than a genetic testing, namely medical history or information derived from observance of an individual's behaviour.

Medical genetics also raises particular privacy concerns. The aspect of privacy which is most commonly referred to in the discussions about genetic privacy is the so-called "informational privacy". In recent times, especially in Europe, this sphere of the right to privacy has turned towards a specific right, the right to data protection.

The principles of data protection (such as the need of consent to the processing of personal genetic data and the right of access to personal genetic information) do not apply to data rendered anonymous in such a way that the data subject is no longer identifiable. This means that protection will not be given to personal data that have been subjected to an anonymization process. The process of anonymization itself is therefore still an act of data processing. This means that, until anonymization is carried out in fact, the data will still be considered as personal data so the principles of data protection will be applicable.

Every person has the right to decide whether or not to be informed of test results. Before a genetic analysis is carried out, the data subject should be informed about the objectives and the possibility of unexpected findings. If they occur, they should not be communicated if

they have no repercussion in the health because the mutation found does not have a great significance or there is no a therapy or prevention known, or it is not going to be transmitted to the offspring.

The right to privacy involves a corresponding duty of confidentiality. This means that genetic data must also be protected from other persons even in case of a biological relative who seeks information concerning the possible presence in him or her of a pathological gene similar to that discovered in the data subject. However, this does not imply that doctors do not have the possibility of disclosing this information in very closely defined circumstances.

Exceptional circumstances, whereby disclosure could prevent serious harm to the health of the relative, and provided there are no other less-intrusive alternatives with respect to the privacy of the patient, may justify a breach of confidentiality in the doctor-patient relationship, and disclosure of the information by the health professional against the wishes of a patient.

These clear principles should ease public concerns about the consequences of genetic testing, at least to some extent. Time will tell whether they engender a sufficient level of trust to encourage those individuals who would benefit from undergoing testing to take up the opportunity to do so when it is made available to them.