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Genetic Information and Individual Rights

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Genetic research and exceptions to the protection of personal data

Roberto Cippitani

1. *Freedom of research and respect for fundamental rights*. Today one of the most powerful expressions of techno-science (i.e., scientific activities that affect the world through technology) is represented by genetic research.

Interest in genetics by scholars and the public has been growing ever since the manipulative power of techno-science has allowed it to not only gain greater meaning from genetic information[1] but also to be used for intervention in the structure of life through techniques such as cloning and genetic editing.[2]

On the other hand, the law governs individual rights and duties concerning genetic information depending on the typology of living beings and thus on the specific interests to be protected[3] (in respect to the EU and international legal instruments concerning animals and plants, see in this book V. Colcelli).

With reference to individuals, Article 1 of the Recommendation of the Committee of Ministers of the Council of Europe, No. R (97) 5 on the Protection of Medical Data (of 13 February 1997) considers genetic information to be "medical data", that is, "personal data concerning the health of an individual".

As medical data, genetic information is taken into consideration by the European Convention on Human Rights and Biomedicine (approved by the Council of Europe in 1997 in Oviedo), especially by its Chapter IV on the Human Genome and by additional protocols.[4] Within European Union law, the Charter of Fundamental Rights explicitly refers to genetic information in provisions Articles 3 and 21.

For individual nations, only some recently amended constitutions, such as those of

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1 An important milestone in the history of this sector is represented by the Human Genome Project, initiated by the US National Institutes of Health (NIH) along with a private undertaking, Celera Corporation, established and run by the biochemist Craig Venter.

2 See for example recent news (2 August 2017) concerning CRISPR, a technique that allows scientists to make changes to genomes in order to correct disease-causing mutations in human embryos. Ledford H., "CRISPR" fixes embryo error. Gene-editing experiment in human embryos pushes scientific and ethical boundaries, in Nature, 3 August 2017, Vol. 548, pp. 13–14.

3 See Janneke H. Gerards, General Issues Concerning Genetic Information, in GERANRDS J.H., HERINGA A.W., and JANSEEN H.L., Genetic Discrimination and Genetic Privacy in a Comparative Perspective, Itersentia, Oxford, 2005, 5 ff.

4 Several additional protocols refer to genetic information: Prohibition of Cloning Human Beings (1998); Human Rights and Biomedicine: Transplantation of Organs and Tissues of Human Origin (2001); Biomedical Research (2005); and Genetic Testing for Health Purposes (2008).

Switzerland (Article 24*nonies*) and Portugal (Article 26.3, para. 2), make specific reference to the protection of genetic data. More typically, legal issues concerning genetic information are regulated by legislation, such as in the legal systems of France and Austria,[5] and in other legislation.[6]

According to legal sources, there are at least two interests that are protected in the case of individuals' genetic information. First, genetic information is considered a particularly important component of personality, and therefore its use must respect the dignity[7] of individuals and in general their fundamental rights.[8] In particular, the protection of dignity is necessary to prevent or punish discrimination based on genetic characteristics (Article 11 of the Convention of Oviedo and Article 21 of the EU Charter).

Another interest taken into consideration does not concern the person but humankind: the intangibility of the human genome. The protection of the human genome is achieved from several perspectives: any alteration of human genetic patrimony (see Article 16-4 of the French Civil Code) in a transmissible manner (see the Universal Declaration on the Human Genome and Human Rights of UNESCO of 1997 and Article 13 of the Convention of Oviedo) is prohibited. This, in particular, concerns whether such modifications arise from scientific practices (see article 57 of the new Argentine Civil Code, which prohibits all scientific or therapeutic practices aiming at genetically modifying the human embryo).

Reproductive cloning of human beings is also considered unlawful under Article 3 of the EU Charter and Article 16-4 of the French Civil Code).

Furthermore, supranational sources consider it "important to exclude unequivocally from patentability processes for modifying the germ line genetic identity of human beings and processes for cloning human beings" (see recitals no. 40 Directive 98/44/EC of the European Parliament and of the Council of 6 July 1998 on the legal protection of biotechnological inventions; see also Article 6, para. 2.b of the directive).

Protection of genetic identity is considered as a safeguard of human identity.[9]

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5 In particular, French law regulates the use of genetic data through Chapter III of Title I of the Civil Code concerning "*De l'examen des caractéristiques génétiques d'une personne et de l'identification d'une personne par ses empreintes génétiques*" (examining the genetic characteristics of a person and his/her identification using genetic prints), which was introduced by laws concerning bioethics, the last one being Law 2011-267 of 14 March 2011. About the French *loi de bioétique*, see CIPPITANI R., "Principi e metodo nella revisione della normativa francese relativa alla bioetica", in Diritto di Famiglia e delle Persone, 2012, pp. 1836–1865; Id., "La nueva ley Francesa en tema de bioética en el contexto europeo", in Criminogenesis, 2011, pp. 199–214.

6 With respect to Swiss law, see the Federal Law on Human Genetic Testing, approved in 2004 and entered into force on 1 April 2007. In Germany in recent years a law concerning genetic diagnostics was approved (*Gendiagnostikgesetz* - GenDG) and entered into force on 1 February 2010. See DIURNI A., "Esperienze di regolamentazione della diagnostica genetica", in Danno e Resp., 2010, 7, 660.

7 FALCONE A., "La tutela del patrimonio genetico umano, fra Costituzione e diritti. Verso la Formazione di un Corpus Iuris sul genoma umano", Rubettino, Catanzaro, 2012, p. 17.

8 RUGGERI A., "Nuovi Diritti fondamentali e tecniche di positivizzazione", in Politica del Diritto, n. 2, 1993, p. 183.

9 ECJ, judgement of 18 October 2011, C-34/10, Oliver Brüstle/Greenpeace eV, ECLI:EU:C:2011:669, para. 33.

2. *Protection of genetic information through the discipline of privacy*. As mentioned in the previous paragraph, among the interests protected by law in respect to human genetic information, personality in its more intimate aspects is of primary importance.

Due to the ethical and legal issues concerning these kinds of interests, this chapter is focused on the legal aspects of scientific research carried out on genetic information. From that perspective, legal sources consider genetic information as "personal data" related to the health of a person. Therefore, the main legal instrument for the protection of fundamental rights associated with genetic data is represented by the discipline of privacy.

At the European level, early legal sources concerning the protection of personal data, such as the Strasbourg Convention no. 108 on the Protection of Individuals with regard to the Automatic Processing of Personal Data of 1981 of the Council of Europe (hereinafter referred to as "Convention no. 108"), as well as Directive 95/46/EC of the European Union of 24 October 1995, do not explicitly consider genetic information.

However, they include references to data that can also involve genetic data. Article 8, para. 1, of the directive, especially, takes into consideration "personal data revealing racial or ethnic origin, and (…) data concerning health". Such data are considered "sensitive" because they may reveal particularly intimate aspects of the life of a person. On these grounds, the processing of those data can be prohibited or subject to special control by authorities, in order to guarantee the reinforced protection provided by the directive (see also Article 6 of the Convention no. 108).

In any case, the qualification of genetic information as personal data has been confirmed in the literature[10] and by documents issued by supranational bodies.

The Explanatory Memorandum of Recommendation No. R (97) 5 of the Committee of Ministers of the Council of Europe on the protection of medical data states that "For the purposes of the recommendation, the drafters of the recommendation considered that most of the principles should apply to genetic data as well as to medical data" (para. 41). The appendix to that recommendation provides a definition of genetic data (among the medical ones) and affirms that the text "refers to 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".

Additionally, the Working Document on Genetic Data, adopted on 17 March 2004 by the Article 29 Data Protection Working Party,[11] states that genetic information must be considered as personal data (para. III, p. 5).

Today, the new Regulation (EU) 2016/679 of 27 April 2016 of the European Parliament and of the Council (General Data Protection Regulation), which will soon replace the directive, explicitly considers genetic information as "personal data" (Article 4, 1), defining the information as "data relating to the inherited or acquired genetic characteristics of a natural person which give unique information about the physiology or the health of that natural person" (Article 4, no. 13).

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[10] D'AMICO M., "Il trattamento pubblico dei dati sensibili: la disciplina italiana a confronto con il modello europeo", in Il diritto comunitario e degli scambi internazionali, Vol. n. 4, 2002, p. 817 ff.

[11] Available at http://ec.europa.eu/justice/policies/privacy/docs/wpdocs/2004/wp91_en.pdf.

Article 9, para. 2, of Regulation (EU) 2016/679 confirms the qualification of genetic data as "sensitive", establishing the prohibition of their processing if some conditions are not met.

As a consequence of the reference to the discipline of privacy, it is possible to apply to genetic information the rules concerning collection, processing, and storage of personal data, especially those that must be considered sensitive.

However, as explained in the subsequent paragraphs, the discipline of protection of personal data has deviated from the general discipline concerning privacy due to the fact that data are processed with scientific research purposes and, also, that research is carried out on genetic information.

3. *Scientific purposes*. According to Article 5, para. 1 of Regulation (EU) 2106/679, personal data must be collected lawfully (let. a) and only to achieve specific proposes, and must be processed in a way that is compatible with those purposes (so called "finality principle"). Not all purposes are acceptable.[12] Pursuant to Article 5 of the International Declaration of UNESCO on Human Genetic Data of 2003 (hereinafter referred to as "Declaration of UNESCO"), the scopes available for the use of genetic data are those concerning health and criminal investigations, that is to say, diagnosis and health care, including screening and predictive testing; and forensic medicine with regard to civil, criminal, and other legal proceedings.

Furthermore, any other purpose consistent with legal definitions and requirements is admissible if it does not violate fundamental rights (see for example Article 20 of the Recommendation CM/Rec(2015)5 of the Committee of Ministers to member states on the processing of personal data in the context of employment).

Among other purposes, research activities may represent a legitimate purpose to collect and process genetic information, as legal sources explicitly establish, at different levels. In particular, Article 5 (ii) of the Declaration of UNESCO considers "medical and scientific research", that is to say medical and other scientific research, including epidemiological research, especially population-based genetic studies, as well as anthropological or archaeological studies, to be legitimate.

Research concerning genetic information is also accepted by supranational legislation (Article 8, para. 3 of Directive 95/46/EC) and by national laws (see Article 16-10 and 16-11 of the French Civil Code; see also the Italian "*Garante per la protezione dei dati personali*", General Authorisation No. 8/2012 of 15 December 2016, para. 3).

Generally speaking, the acceptability of scientific purposes arises from the relevance assumed by science for society and legal systems. Today, national and supranational constitutions, as well as international legal agreements, consider academic activity, and especially research, as a fundamental freedom (see mainly Article 13 of the EU Charter).[13]

[12] The processing of genetic information for purposes not legally recognised may be punished by criminal law, as is the case in France for those requesting genetic testing on themselves or others, outside the cases authorized by law (see Article 226-28-1 penal code).

[13] For commentary on this disposition, see MOLINA DEL POZO F. and ARCHONTAKI C., "Libertad de artes y de Investigación Científica, Libertad de Cátedra", In ALVAREZ LEDESMA M.

This freedom is considered necessary for the benefit of humankind. As stated by Article 2 of the Universal Declaration on the Human Genome and Human Rights of UNESCO of 1997, the "benefits from advances in biology, genetics and medicine, concerning the human genome, shall be made available to all" (Article 2.a) and "Freedom of research, which is necessary for the progress of knowledge, is part of freedom of thought. The applications of research, including applications in biology, genetics and medicine, concerning the human genome, shall seek to offer relief from suffering and improve the health of individuals and humankind as a whole." (see letter b). According to Article 14 of that declaration, states have the obligation to grant the exercise of such freedom.

Sub-constitutional legislation also underlines the importance of research. This is particularly clear in the field of personal data and especially in the EU's General Data Protection Regulation. As in other EU directives during the last thirty years, research is considered as the fulcrum of European integration. This is explained by the institutional documents of the Lisbon strategy of 2000 and today in "Europe 2020".[14]

In particular, the General Data Protection Regulation (see recital no. 159 mentioned above) underlines the importance of the circulation of information for the building of the European Research Area (hereinafter referred to as "ERA"), as provided for by Article 179, para. 1, TFU, "in which researchers, scientific knowledge and technology circulate freely".

ERA is not only a dimension of the internal market, but also the expression of a cultural pillar on which the European integration process should be built. As a matter of fact, the regulation itself affirms that "the legitimate expectations of society for an increase of knowledge should be taken into consideration" (recital no. 113) and also points out that "To meet the specificities of processing personal data for scientific research purposes, specific conditions should apply in particular as regards the publication or otherwise disclosure of personal data in the context of scientific research purposes" (recital 159 as above).

Due to the above-mentioned reasons, and especially on the bases of the particularities of research activities, the European discipline concerning protection of personal data provides some specific derogations or exceptions to data use in the case of processing of personal data for scientific purposes.

4. *Scientific purposes and exceptions to the rule of consent.* On the ground of the qualification as "personal data" (see Article 4, nn. 1 and 13, Regulation (EU) 2016/679), genetic information should be under the control of the "data subject", who is entitled to give her/his "informed, free, express, specific and documented consent of the person" (Convention of Oviedo, see in particular Article 14) for processing such data (see also Article 6, letter d, Declaration of UNESCO).[15]

I. and CIPPITANI R. (coord.), Diccionario analítico de Derechos humanos e integración jurídica, ISEG, Roma-Perugia-México, 2013, pp. 361–367.

[14] Communication, Europe 2020, A strategy for smart, sustainable and inclusive growth, COM(2010) 2020 final, 3 March 2010. On the legal issues of a knowledge-based society, see CIPPITANI R. (editor), El Derecho en la Sociedad del Conocimiento, ISEG, Roma-Perugia, 2012.

[15] About informed consent to use personal genetic information, see CIPPITANI R., "Consent to the

According to the definition provided for by Article 2(iii) of the Declaration of UNESCO, consent is the "specific, informed and express permission that a person freely gives for his genetic data to be collected, processed, used and preserved" (see also Article 2 (j) of the Directive 2001/20/EC on clinical trials).[16]

Due to the qualification of genetic information as "personal health data", the subject's consent should be not only clear (see Article 4, no. 11 of the General Data Protection Regulation), but also explicit.[17] This is because legal texts state that the form of expression of consent should depend on the importance of the interests to be protected.[18]

Explicit written consent is needed in the case of the individual's participation in biomedical scientific research (see Convention of Oviedo, Article 16, v), especially when research activities are related to genetic information (see the General authorisation no. 8/2012 para. 6; see also Article 16-10 of the *Code Civil*, and also in French Law Article L. 1131-1 of the *Code Santé Publique*, hereinafter "CSP").

In particular, consent is needed when the genetic data are "stored for diagnostic and health care purposes and for medical and other scientific research purposes, unless otherwise provided for by domestic law for compelling reasons and consistent with the international law of human rights" (see Article 22).

Furthermore, Article 8, para. 2 of the EU Charter states that "Everyone has the right of access to data which has been collected concerning him or her, and the right to have it rectified".

However, the individual's consent and the linked rights on personal data may be subject to several exceptions to safeguard other interests recognised by constitutional norms. Privacy should be coordinated with these other important freedoms or rights recognised by constitutional norms.

National or supranational legislation may impose limitations to some rights in order to protect personal data, for reasons such as national security; defence; public security; prevention, investigation, detection, and prosecution of criminal offences or breaches of ethics for regulated professions; important economic or financial interests; or the protection of data subjects or of the rights and freedoms of others.

Moreover, legal sources provide an important set of exceptions in case personal information is used in scientific activities.[19]

The necessity for exceptions to the right of consent arises from the features of research, the development of which depends on the availability of data. In fact, public policies

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Use of Genetic Information: Between Respect of Privacy and Protection of Other Fundamental Interests", in Diritto e Processo/Right and Remedies/Derecho y Proceso, 2014, pp. 493–532.

16 See SASSI A., "Derechos patrimonialmente neutros", in Mario ALVAREZ LEDESMA M. I. and CIPPITANI R. (edit by), Diccionario analítico de Derechos humanos e integración jurídica..., pp. 213–218.

17 See WP131 - Working Document on the processing of personal data relating to health in electronic health records (EHR).

18 See Article 29 Data Protection Working Party, *Opinion 15/2011 on the definition of consent*, Adopted on 13 July 2011, para. III.A.3; available at http://ec.europa.eu/justice/policies/privacy/docs/wpdocs/2011/wp187_en.pdf.

19 See Commission, *Open Innovation, Open Science, Open to the World - a vision for Europe*, Bruxelles, 2016.

limiting access to data[20] may adversely affect scientific research, especially in the case of genetics.[21] For these reasons, legislation on privacy provides some limits to the rights of data subjects.

Directive 95/46/EC established that member states can be "authorized, when justified by grounds of important public interest, to derogate from the prohibition on processing sensitive categories of data where important reasons of public interest so justify in areas such as public health and social protection (…) scientific research and government statistics" (recital no. 34 of the Preamble).

Such derogations from the general rules were possible in two cases: rights of the data subject when the information is not obtained directly by the data subject her/himself (Article 11, para. 1); and the right of access in order to know how the data are processed, as well as rights of rectification, erasure, or blocking (see Article 12, para. 1). In those cases, legislation of member states was allowed to provide derogations from the data subjects' rights when these data were used for scientific purposes. In the first case, exceptions were possible if "the provision of (…) information proves impossible or would involve a disproportionate effort or if recording or disclosure is expressly laid down by law". In the case of the rights of access, limitations of the data subjects' rights were authorised by the EU directive for processing solely for scientific research purposes (see Article 13, para. 3).

The new regulation concerning protection of personal data aims at establishing a more general framework of derogations from the rights of the data subject.

Regulation no. 2016/679 considers the same case of Article 11, para. 1, of the directive, establishing for research activities a derogation from the rights of the data subject if the data are collected from sources other than the latter (see Article 14, para. 5, Regulation 2016/679; see also recitals nn. 61 and 62). Furthermore, in a wider perspective than the directive, the regulation establishes that when "personal data are processed for scientific or historical research purposes or statistical purposes", European and national laws may provide derogations from the rights normally belonging to the data subjects such as the right of access (Article 15); right to rectification (Article 16); right to restriction of processing (Article 18), and the right to object (Article 21).

Laws may also establish a derogation from the right to erasure (the right to be forgotten), established by Article 17, para. 1, of Regulation (EU) 2016/679: "The data subject shall have the right to obtain from the controller the erasure of personal data concerning him or her without undue delay and the controller shall have the obligation to erase personal data without undue delay".

Derogations from the rights usually recognised to data subjects are also provided for by documents of the Council of Europe's bodies. For instance, Article 8, para. 2.d, of Rec-

[20] LOWRANCE W. and COLLINS F. S., "Identifiability in Genomic Research", in Science, 3 August 2007, vol. 317, pp. 600–602.

[21] See the conclusions of GYMREK M., McGUIRE A., GOLAN D., HAPERIN E. and ERLICH Y., "Identifying Personal Genomes by Surname Inference", in Science, 18 Jan 2013, vol. 339, Issue 6117, pp. 321–324; and also the editorial in Nature concerning research on science titled "Genetic Privacy". ("The ability to identify an individual from their anonymous genome sequence, using a clever algorithm and data from public databases, threatens the principle of subject confidentiality.") Nature, 24 January 2013, vol. 493, p. 451.

ommendation R(97) states that access to medical data (including genetic data) and the right of rectification may be refused when "the data are used for statistical or for scientific research purposes where there is clearly no risk of an infringement of the privacy of the data subject, notably the possibility of using the data collected in support of decisions or measures regarding any particular individual".

Therefore, according to EU law, once genetic information is processed within scientific activities, the data subject loses her/his power over the information, as provided for on the contrary in other cases of processing of personal data.

Such limitations are justified from both subjective and objective points of view: staff dealing with genetic information must be professionally qualified (see for example para. 14 of the Declaration of Helsinki, Article 3, para., lett. a, Directive 2001/20/EC)[22] and must respect "relevant professional obligations and standards" (see Article 14 of the Convention of Oviedo); the activities carried out must be qualified as "research".

According to the latter condition, due to the favourable legal and political context, "research" and "research purposes" should be considered in a broad manner, in accordance with EU law, therefore "including for example technological development and demonstration, fundamental research, applied research and privately funded research" (recital no. 159, Regulation 2016/679).

In order to avoid any doubt, research activities must be formalised in a project (see para. 4, Authorisation no. 8 of the Italian *Garante*) that has to be drawn up in accordance with the standards of the relevant disciplinary field, in order to provide evidence that the processing of data and the use of biological samples are carried out for suitable and effective scientific purposes.

5. *Further uses*. Normally, legal sources provide a "specific consent", meaning that the data subject is entitled to give her/his authorisation for any specific use of personal data, in order to achieve a more complete safeguard of the autonomy of persons. In addition to the aforesaid dispositions of the Convention of Oviedo and of the Declaration of UNESCO, the specificity of consent is provided within EU legislation, such as by Article 8, para. 2 of the EU Charter, which states that "[personal] 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." Article 4, no. 11 of Regulation (EU) 2016/679 provides likewise.

Therefore, the discipline of protecting personal data is based on the rules of "granularity",[23] that is to say the necessity that the consent should be given for limited aims and for specific situations.[24] When the purposes of processing or the situation of the data subject change, the person should be requested to express a new consent.

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22 Freedom of research is different from freedom of expression, because it is recognised only to qualified persons acting within academic institutions or undertakings, who have the necessary skills and instruments. See CIPPITANI R., "Academic Freedom as a Fundamental Right". In: 1st International Conference on Higher Education Advances, HEAd'15, Universitat Politècnica de València, Valencia, 24–26 June 2015, Universitat Politècnica de València, pp. 552–558.

23 See para. III.A.1 of the Advice 15/2011 *on the definition of consent*, ref.

24 *Ibidem*.

This is what emerges, for example, from the Recommendation of the Committee of Ministers of the Council of Europe Rec (2006) 4 of 15 March 2006, concerning research on biological material of human origin. Article 12, paragraph 1 required that biological material collected for purposes other than scientific research (i.e., for therapeutic purposes) could not be used without consent or authorisation. Thus, when the subsequent activity is "substantially different" as regards the authorised individual,[25] new consent should be given.

Consent should not be given without time limits. EU documents set forth that those responsible for the processing of personal data shall re-ask the person to confirm her/his consent[26] if the situation of the data subject has changed (e.g., because a child becomes a teenager).[27]

The granularity rule may constitute an obstacle for research activities. As a matter of fact, the collection of data is normally realised in the frame of other activities, such as for diagnostic analysis, and then processed for scientific purposes. Those purposes are not so specifically clear at the moment of data collection, and they can change over time. Furthermore, the same base of data may be useful for many types of research, even in different fields of research (genetic data can be processed in the medical, biological, anthropological, and sociological fields, for example). Therefore, it could be difficult to acquire a consent concerning specific programmes of research, and it can be problematic and expensive to require consent for each specific scientific activity.

This is especially true for bio-banks activities, that is to say large collections of biological samples (in particular of human origin) and associated data, such as genetic information.[28] Bio-banks are established for various reasons, such as criminal investigation, therapeutic treatments, and research activities. Public and private interests (e.g., those of pharmacological industries) need to maintain genetic information in bio-banks for many years. Many kinds of research activities with stored information could be carried out in the future, but they are not all known or at least foreseeable when data and biological material are collected. This makes it particularly difficult to require consent for a specific purpose and over the entire time that research could be undertaken with the samples and associated data.

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25 Council of Europe, *Explanatory report to the convention on human rights and biomedicine*, 1997, para. 214.

26 See also Article 29 Working Party, *Opinion 2/2010 on online behavioural advertising*, adopted on 22 June 2010, available at http://ec.europa.eu/justice/data-protection/article-29/documentation/opinion-recommendation/files/2010/wp171_en.pdf.

27 Article 29 Working Party, *Working document 1/2008 on the protection of children's personal data,* adopted on 18 February 2008. Available at http://194.242.234.211/documents/10160/10704/1531889.

28 For an overview of European, international, and national legislation relating to bio-banks, see, among others, TESÓN I. V., "Bioresearch, Biobanks and Informed Consent from Vulnerable Donors in Spanish Law", in Europa e Diritto private, 2013, p. 1069 ff.; SCAFFARDI L., "Legal Protection and Ethical Management of Genetic Databases: Challenges of the European Process of Harmonization", in European Legal Integration: The New Italian Scholarship, Jean Monnet Working Paper 19/08, New York University School of Law, New York, 2008; GODARD B., SCHMIDTKE J., CASSIMAN J.J. and AYMÉ S., "Data storage and DNA banking for biomedical research: informed consent, confidentiality, quality issues, ownership, return of benefits. A professional perspective", in European Journal of Human Genetics, 2003, 11, Suppl 2, S88–S122.

For those reasons, studies in the literature and praxis suggest more flexible approaches. Furthermore, from an institutional point of view, in recent years we can observe a tendency to mitigate the principle of granularity.

It is possible to find solutions that refer to enlarged or broad consent (for a range of broadly defined uses); to presumed consent (where people who do not want to be involved have to opt out voluntarily); and, in some cases, also "blanket consent", that is to say consent to whatever future use has been outlined. According to the latter, which seems the furthest removed from specific consent, the World Health Organisation, in a document of 1998, admits that "[a] blanket informed consent that would allow use of sample for genetic research in general, including future as yet unspecified projects appears to be the most efficient and economical approach, avoiding costly re-contact before each new research project".[29] It would seem that this approach should be put in place to grant protection of personal data[30]; the more widely used approach, however, is broad consent.

Therefore, the Recommendation of 2016 of the Committee of Ministers of the Council of Europe has replaced the obligation to give information concerning each research activity (as established by Article 10, para. 2 of the Recommendation of 2012) with the duty to inform the data subject about a more general "nature of any envisaged research use" (Article 10, para. 1, Recommendation of 2016).

Also, the Draft Explanatory Memorandum to the Draft Recommendation on Research on Biological Materials of Human Origin, of the Committee on Bioethics (DH-BIO) of the Council of Europe of 2015, specifies that when human biological materials or associated personal data are collected, it is good practice to obtain the consent to their use for future research, even in cases where the specific research is not known. If future research cannot be identified, the consent should not be unconditional (i.e., a blanket consent) but should be as specific as possible, given the knowledge at the time consent is obtained.[31]

At the national level, for example, the UK Ethics and Governance Framework provides explicitly that "[b]ecause it will be impossible to anticipate all future research uses, consent will be sought for research in general that is consistent with UK Biobank's stated purpose (rather than for specific research)". A "[f]urther consent will be sought for any proposed activities that do not fall within the existing consent".

Other examples of the implementation of broad consent can be found in the German

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29 World Health Organisation, Proposed international guidelines on ethical issues in medical genetics and genetic services, 1998, p. 13, available at http://www.who.int/genomics/publications/en/ethicalguidelines1998.pdf .

30 *Ibidem*.

31 See Article 12, para. 48 of the Draft Explanatory Memorandum to the Draft Recommendation on Research on Biological Materials of Human Origin of Steering Committee on Bioethics: "When biological materials of human origin and personal data are collected it is best practice to ask the sources for their consent to future use, even in cases where the specifics of the future research projects are unknown. If future research use of biological materials of human origin and personal data cannot be specifically anticipated, the consent should not be framed too broadly in order to prevent unconditional, "blanket" consent. The request for consent should be as explicit as possible in regard to the future research uses of the biological material of human origin and personal data".

Nationaler Ethikrat of 2004[32] as well as in the Code of Practice of the UK Human Tissue Authority of 2006 and in Swedish,[33] Icelandic, and Estonian laws that allow a broad description of the purposes of research. The Spanish law on biomedical research[34] provides the possibility to give consent for specific research projects even if they are carried out by other subjects.[35]

Additionally. the EU regulation concerning privacy considers the hypothesis that it is not possible to fully identify the purpose of personal data processing for scientific research at the time of data collection. In that case, data subjects should be allowed to give their consent within certain areas of scientific research, if recognised ethical standards for scientific research will be observed (recital no. 33, Regulation (EU) no. 2016/679).

On the other hand, Regulation no. 2106/679 and other European sources extend the effectiveness of consent. If the principle of purpose limitation prescribes that "the processing of personal data for purposes other than those for which the personal data were initially collected should be allowed only where the processing is compatible with the purposes for which the personal data were initially collected" (recital no. 50), nevertheless "further processing for archiving purposes in the public interest, scientific or historical research purposes or statistical purposes shall, in accordance with Article 89(1), not be considered to be incompatible with the initial purposes" (Article 5, para. 1, let. b) Regulation 2016/679).[36] For these types of purposes, to be a sort of presumed consent is given.

The same approach is chosen by the Council of Europe in the draft of the Recommendation on the Protection of Health-Related Data, which will replace the above-mentioned recommendation of 1997 (see Article 4.1.b); this also seems to consider it difficult to provide detailed information to the data subject about the use of health-related data at the time of collection (see Article 11.2).

In application of the model of presumed consent, the Italian Authority of Privacy, in its General Authorisation No. 8/2014 for the Processing of Genetic Data, allows processing research for scientific purposes "directly linked" to the original one. Otherwise, processing is authorised only if samples are anonymised or in the case of a new consent, but in the absence of the latter consent can be authorised by the relevant ethics committee

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32 Nationaler Ethikrat, Biobanken für die Forschung. Stellunghame, 2004, Berlin, available at www.ethikrat.org/_english/publications/Opinion_Biobanks-for-research.pdf.

33 The Recommendation R(2006)4 of the Council of Europe was inspired by the UK Human Tissue Act of 2004 and by the linked code of practice issued by the Human Tissue Authority of January 2006. In particular, point 106 of the Code of Practice Consent provides that "consent can be general, i.e. if someone consents to the use of tissue for research, it need not be limited to a particular project". See also para. 90 stating that "consent should be generic where appropriate".

34 Ley no. 14/2007, de *Investigación biomédica*, of 3 July 2007.

35 See Article 60, para. 1: "*El consentimiento sobre la utilización de la muestra biológica se otorgará, bien en el acto de obtención de la muestra, bien con posterioridad, de forma específica para una investigación concreta. 2. El consentimiento específico podrá prever el empleo de la muestra para otras líneas de investigación relacionadas con la inicialmente propuesta, incluidas las realizadas por terceros. Si no fuera este el caso, se solicitará al sujeto fuente que otorgue, si lo estima procedente, un nuevo consentimiento*".

36 By EU and national laws. An example of such national provisions is the Austrian Data Protection Act (*Datenschutzgesetz*), Federal Law Gazette No. 165/1999, para. 46, available in English at www.dsk.gv.at/DocView.axd?CobId=41936.

and authority. However, it may not be simple to identify either the meaning of the "link" or who has control over the compliance.

Another solution regarding research can be found in the Recommendation of 2016 of the Committee of Ministers of the Council of Europe in case of collection of biological material. In that hypothesis, the material "should only be used in a research project if the latter is within the scope of the consentor authorisation given by the person concerned" (Article 21, para. 1). However, if the proposed use will not be within the scope of prior consent or authorisation, if any, given by the person concerned, reasonable efforts should be made to contact the person concerned (para. 2.a), and the process must be subject to an independent evaluation (para. 2.b).

6. *Storage of genetic data and data retention.* Storage and retention of personal data are regulated in a special manner when they are put in place within research activities. Although the discipline of privacy does not establish a fixed term for storing data, it provides for rules that are incompatible with long-time storage, such as the above-mentioned right to be forgotten and the right to withdraw, as well as the principle of "storage limitation", according to which data must "[be] kept in a form which permits identification of data subjects for no longer than is necessary for the purposes for which the personal data are processed" (Article 5, para. 1.e, Regulation (EU) 2016/679). Also in this case, scientific purposes allow exceptions from rules concerning data processing in general.

As explained above, it is very important for current scientific activities to have access to data and materials included in long-term collections. Regulation no. 2016/679 takes into consideration the need of science to collect information and to store it (also for historical research purposes, see recital no. 160). The reason to improve archiving is explained by recital no. 157 of the new regulation: "By coupling information from registries, researchers can obtain new knowledge of great value with regard to widespread medical conditions such as cardiovascular disease, cancer and depression. On the basis of registries, research results can be enhanced, as they draw on a larger population. Within social science, research on the basis of registries enables researchers to obtain essential knowledge about the long-term correlation of a number of social conditions, such as unemployment and education with other life conditions. Research results obtained through registries provide solid, high-quality knowledge which can provide the basis for the formulation and implementation of knowledge-based policy, improve the quality of life for a number of people and improve the efficiency of social services".[37]

Public interest and the use for research are considered legitimate grounds for storing health-linked personal data, including genetic data, for a longer period (see Article 4.1.f draft Recommendation on the Protection of Health-Related Data). In respect to the rule that personal data cannot be stored longer than it is necessary, national law should lay

[37] In the case of historical research purposes, the value of the archiving is underlined by recital no. 158, where it states that "Member States should also be authorised to provide for the further processing of personal data for archiving purposes, for example with a view to providing specific information related to the political behaviour under former totalitarian state regimes, genocide, crimes against humanity, in particular the Holocaust, or war crimes".

down "more detailed provisions, including the necessary safeguards, to reconcile the interest in scientific research with the right to data protection",[38] and "Keeping data for future scientific, historical or statistical use is explicitly exempt from the principle of limited data retention."[39]

With respect to the right to erase, as mentioned above, the regulation provides a specific exception in consideration of research purposes. As a matter of fact, Article 17, para. 1.b provides the right to erase (the "right to be forgotten") "where there is no other legal ground for the processing". As stated above, scientific purposes are considered the ground for not applying the rights provided under Article 17. According to the right to withdraw, Article 7, para. 3 establishes that "The data subject shall have the right to withdraw his or her consent at any time". However, the same provision states that "The withdrawal of consent shall not affect the lawfulness of processing based on consent before its withdrawal". Therefore, it seems that ongoing research activities at least should not be affected by the withdrawal of consent.

When biological material is collected in addition to data, Recommendation CM/Rec(2016)6 of the Committee of Ministers on research on biological materials of human origin provides for the right to withdraw "in the manner foreseen by law" (see in particular Article 13), but it also states, in regard to informing the person prior to removing the material, that "This information should also include any possible limitation on withdrawal of the consent or authorisation" (Article 10, para. 2).

It should be emphasised that the guarantee of withdrawal of consent, due to the considerable size achieved by bio-banks and the continuous exchange of materials and data among researchers, is weak and difficult to concretise, especially as far as the information dimension is concerned. In this regard, Spanish law no. 14/2007 concerning biomedical research states that, in case of withdrawal, biological samples will be destroyed. However, the data obtained in the preceding phases can be maintained.[40]

7. *Specificity of genetic information.* As research activities are carried out using genetic information, it is necessary to consider some further specific issues. Although genetic information is protected by legislation concerning personal data, the Declaration of UNESCO as well as other documents (see paragraph 2 of the Working Document on Genetic Data of 2004) recognise it as having a "particular status". Some scholars do not agree with the presumed particularity of genetic information, increasing the resistance of public opinion in respect to genetic technologies.[41]

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[38] European Union Agency for Fundamental Rights, Council of Europe, *Handbook on European data protection law*, Luxembourg: Publications Office of the European Union, 2014, p. 31.

[39] European Union Agency for Fundamental Rights, Council of Europe, *Handbook on European data protection law*, … p. 73.

[40] Article 60.3: "*El consentimiento podrá ser revocado […] en cualquier momento. Cuando la revocación se refiera a cualquier uso de la muestra, se procederá a su inmediata destrucción, sin perjuicio de la conservación de los datos resultantes de las investigaciones que se hubiesen realizado con carácter previo*".

[41] RICHARDS M. P.M., "How distinctive is genetic information?", in Studies in the History and Philosophy of Biological and Biomedical Sciences, 2001, 32, pp. 663–687.

Indeed, so-called "genetic exceptionalism" has been criticised due to the exaggerated view of the significance of genetic information in people's lives, based on an unacceptable genetic determinism and genetic reductionism.[42] Nevertheless, the special status of genetic data may be observed in relation to several cases.

As a matter of fact, genetic information is different from other types of information, due to the fact that it identifies a specific individual in a permanent way ("immutability") and it is predictive of predisposition to diseases ("predictability"). Furthermore, genetic information belongs not only to the concerned person but also to people sharing the same genetic patrimony ("familiarity").[43] For example, in regard to the use of genetic data in criminal investigations, it has been argued that "DNA samples or profiles are intrinsically 'more private' objects or their collection involves greater infringement of bodily integrity than, for example, fingerprints or photographs." [44]

DNA is akin to a "future diary" of persons (it includes information about our present and future medical conditions), and the right of protection from unwanted "readership" must be imperative in order to maintain autonomous control of personal and sensitive information.[45]

The above-mentioned features of genetic information should lead to a specific regulation, also taking into account the great risks of misuse and/or re-use for various purposes and the risks of discrimination and stigmatization that may affect the individual. Moreover, some authors underline that the discipline of privacy can cover only some aspects of the protection of genetic information and related rights.[46] At least some issues may arise from the use of genetic information especially in the context of research activities: information to be provided to the data subject, relativity of anonymisation, and the rights of other subjects.

A) Information to be provided to the data subject.

Despite the limitations of the rights of data subjects in the context of scientific activity, the fact that research is carried out on genetic information may lead to solving other problems. The special informative content of genetic data has important consequences on the right of the data subject to know or not to know the implications of such data for future health.-

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42 MURRAY T.H., "Genetic Exceptionalism and Future Diaries: Is genetic Information Different from Other Medical Information", in ROTHSTEIN M.A., Genetic Secrets: Protecting Privacy and Confidentiality in the Genetic Era, Yale University Press, New Haven, 1997, pp. 60–73, in particular p. 71.

43 On the co-shared nature of genetic information, see TAYLOR M.J., "Data Protection, Shared (Genetic) Data and Genetic Discrimination", in Medical Law International, 8, 1, 2006, p. 51.

44 WILLIAMS R., JOHNSON P. and MARTIN P., "Genetic information and crime investigation: social, ethical and public policy aspects of the establishment, expansion and police use of the National DNA Database. Project Report", Durham University, School of Applied Social Sciences, Durham, 2004, para. 6.2.2, p. 78

45 ANNAS G.J., "Genetic Privacy", in LAZER D., DNA and the Criminal Justice System: The Technology of Justice, Cambridge, MA: MIT Press, 2004.

46 In particular, see TAYLOR M., "Genetic Data and the Law: A Critical Perspective on Privacy Protection", Cambridge, Cambridge University Press, 2012, *passim*.

A first problem arises if the development of a new technique may give more information in comparison with the past. With reference to biomedical research, such a hypothesis seems to be covered by Article 24 of the Additional Protocol to the Convention of Oviedo on Biomedical Research, which provides for the re-examination of a research project in the "light of scientific developments or events arising in the course of the research", when "research participants, or if applicable their representatives, need to be informed of the developments or events" (para. 2.ii). When information does not refer to the health of persons, it does not seem mandatory to inform the data subject.

Another problematic aspect is represented by so-called "unexpected findings", that is, information that was not expected to be found during research or diagnostic practices, such as information on ongoing diseases or predispositions to diseases, or information concerning biological parenthood, and so on.

For example, the general authorisation no. 8/2012 of the Italian *Garante* mandates that the individual, before any genetic testing, must also be informed on the possible results of such testing, especially "with regard to unexpected findings" (para. 5.b). This caution should not be necessary in the processing of other kinds of sensitive data (as with political opinions). It is not clear what happens if information on a health situation or other information (for example concerning filiation or paternity) arises from research activities. Probably, in these cases, the data subject has to be requested to give her or his authorisation to be informed, including about any unexpected findings. However, if such authorisation was not requested, or could not be acquired (on the ground of some above-mentioned rules), the problem remains whether researchers have an obligation to inform the concerned persons. No obligation in this regard seems to be provided for by legislation, even if the importance of health would suggest the prudence of informing the affected individuals at least on the existence of findings concerning diseases, and in particular about treatment options.

B) The problem of anonymisation.

Recital no. 26 of the General Data Protection Regulation clearly states that "This Regulation does not therefore concern the processing of such anonymous information",[47] including for statistical or research purposes. Thus, if data might not be associated with a specific person, it is outside the protection of the legislation and it can be processed without the consent of the data subject.

Personal data may be collected in a non-anonymous way and be anonymised subsequently. Data "are anonymised if all identifying elements have been eliminated from a set of personal data. No element may be left in the information which could, by exercising reasonable effort, serve to re-identify the person(s) concerned".[48]

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[47] Data are considered anonymous taking into account "means reasonably likely to be used, such as singling out, either by the controller or by another person to identify the natural person directly or indirectly. To ascertain whether means are reasonably likely to be used to identify the natural person, account should be taken of all objective factors, such as the costs of and the amount of time required for identification, taking into consideration the available technology at the time of the processing and technological developments" (see 26th recital of Regulation (EU) 2016/679).

[48] European Union Agency for Fundamental Rights, *Handbook on European data protection law*, Luxembourg, 2014, p. 44.

In addition, Recommendation CM/Rec(2016)6 of the Committee of Ministers on research on biological materials of human origin supports the use of anonymisation. In fact, the recommendation states that "non-identifiable biological materials" (see the definition under Article 3, according to which they are "those biological materials which, alone or in combination with data, do not allow, with reasonable efforts, the identification of the persons from whom the materials have been removed") "may be used in a research project provided that such use does not violate any restrictions defined by the person concerned before the materials have been rendered non-identifiable and subject to authorisation provided for by law" (Article 21, para. 4) and "Biological materials previously removed for another purpose and already non-identifiable may be stored for future research subject to authorisation provided for by law" (Article 11, para. 3).

In respect to the specific case of genetic information, the Declaration of UNESCO states that genetic data when "collected for the purposes of scientific research should not normally be linked to an identifiable person. Even when such data or biological samples are unlinked to an identifiable person, the necessary precautions should be taken to ensure the security of the data or biological samples" (Article 14c).

Otherwise, European law considers the alternative technique of pseudonymisation (see Article 4, no. 5 Regulation (EU) no. 2106/679). This occurs when the identifiers are replaced by pseudonyms, and the data cannot be identifiable without possession of a decryption key.[49]

Tissue and Cells Directive no. 2004/23/EC obligates member states to take all necessary measures to ensure that all data, including genetic data, have been rendered anonymous so that neither donors nor recipients remain identifiable (see Article 14, para. 1).

According to the General Data Protection Regulation, anonymisation and pseudonymisation are considered ordinary measures to protect personal information in research activities (see Article 89).

Additionally, the Appendix to Recommendation R(97) of the Committee of Ministers considers that "Whenever possible, medical data used for scientific research purposes should be anonymous" and that "Professional and scientific organisations as well as public authorities should promote the development of techniques and procedures securing anonymity" (see para. 12.1).

However, the option of anonymisation, as an alternative to consent, may encounter some problems in the case of genetic information. First, anonymisation is never the better option from a scientific viewpoint. As shown by legal sources (see for example the Declaration of UNESCO on genetic data), the link to an identifiable person may be acceptable "if necessary to carry out the research and provided that the privacy of the individual and the confidentiality of the data or biological samples concerned are protected in accordance with domestic law" (Article 14d) and for a period that does not exceed the time needed for achieving the purposes for which they were collected or subsequently processed (Article 14.e).

Complete anonymisation implies some serious consequences: both data subject and

[49] See Council of Europe, Explanatory Report to the Convention for the Protection of Individuals with regard to Automatic Processing of Personal Data, Article 42.

researcher will lose important information and will not be able to yield follow-up results,[50] often fundamental to optimal performance of a research project.

Second, anonymity is always relative because of technical reasons. The anonymisation processes are likely reversible, and in principle any anonymised genetic data can be linked to a person. *A fortiori*, the situation might also occur in the case of pseudonymisation.[51]

As underlined within the scientific community, "No responsible scientist can guarantee absolute privacy" and "Privacy and confidentiality are important principles. But being identifiable has some benefits, and being anonymous has some costs; science will be better off when it acknowledges this reality."[52]

According to some authorities, the risk of re-identification posed by genetic data would be considered low. As stated by Article 29 of the Working Party, treating the matter of pseudonymisation, "In that case, although data protection rules apply, the risks at stake for the individuals with regard to the processing of such indirectly identifiable information will most often be low, so that the application of these rules will justifiably be more flexible than if information on directly identifiable individuals were processed."[53]

However, this interpretation refers to the current state of the technique and does not take into consideration that it is possible to establish an association between the genetic information and other pieces of information, in a way leading to identification of a person. As demonstrated by a study published in *Science*,[54] it is possible, through the sequencing of genetic data without identifiers, to recover surnames of the data subjects by profiling short tandem repeats on the Y chromosome and querying genetic genealogy databases (as for example www.ysearch.org and www.smgf.org). Then, a specific person can be targeted by combining the surname with other types of metadata, such as age and state, easily and freely available in Internet resources.

Therefore, it is possible, at least, to provide practical suggestions, such as those included in para. 4.2 of Authorisation no. 8 of the Italian Privacy Authority, which states that where the genetic information arises from biological samples and the "temporary" identification of the subject is necessary, specific measures should be adopted to keep identification data separated from biological samples and genetic information at the time of collection, unless this is impossible due to the particular characteristics of the treatment or to the necessity to use manifestly disproportionate means.

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[50] MACILOTTI M., IZZO U., PASCUZZI G. and BARBARESCHI M., "La disciplina giuridica delle biobanche (The Legal Aspect of Biobanks)", in Pathologica, 2008, v. 100, pp. 86–108, particularly p. 87.

[51] Article 29, Data Protection Working Party, Opinion 4/2007 on the concept of personal data, adopted on 20th June 2007, p. 18, stating that "Retraceably pseudonymised data may be considered as information on individuals which are indirectly identifiable. Indeed, using a pseudonym means that it is possible to backtrack to the individual, so that the individual's identity can be discovered, but then only under predefined circumstances."

[52] ANGRIST M., "Genetic privacy needs a more nuanced approach", in Nature, 7 February 2013, vol. 494, p. 7.

[53] Article 29, Data Protection Working Party, Opinion 4/2007 on the concept of personal data, ref.

[54] See GYMREK M. et al., "Identifying Personal Genomes by Surname Inference", in Science, ref.

C) Rights of other subjects.

As mentioned above, genetic information belongs not only to a specific person, but it is shared among persons of the same genetic group. According to Article 14 of Regulation (EU) 2016/679, the data subject also has the right to receive information from the controller (or his/her representative) when the data have not been obtained from the aforesaid data subject. In consequence, a physician or other health professional, who found a risk of a genetic disease while examining the biological material of a person, might face the following dilemma: on the one hand he/she could be bound by the obligation of secrecy, as well as the right to not inform the individual. On the other hand, he/she could be obliged under Article 11 to provide information to the data subjects, who include relatives sharing the same genetic line.

There is not a clear answer to that question, neither within the discipline concerning privacy nor in the supranational and international legal sources. According to Article 18 of the Additional Protocol to the Convention of Oviedo on genetic testing, "Where the results of a genetic test undertaken on a person can be relevant to the health of other family members, the person tested shall be informed."

However, the consequences and conditions arising from that information are not clear. According to the above-mentioned working document on privacy, at least two scenarios may be imagined: "One is that other family members could also be considered as 'data subjects' with all the rights that follow from this. Another option is that other family members would have a right of information of a different character, based on the fact that their personal interests may be directly affected."

At the national level, legislation is focused on the protection of the personal data subject's privacy, requiring his or her consent to disclose the information to relatives.[55] Within Europe, an interesting solution is provided by French law, even if it does not directly refer to scientific activities. Before the last version of the law concerning bioethics (Law 814-2011), the legislation previously in force already established a procedure for communicating the results of genetic testing to family members (s. Article L. 1131-1, para. 5 CSP), without providing any consequence in case the person had not informed her/his relatives.[56]

Such an exclusion of liability appeared in conflict with constitutional principles.[57] Thus, the *Conseil d'Etat* in its document on the review of law concerning bioethics proposed to make explicit the responsibility to inform family members about genetic abnormalities, while respecting medical confidentiality.[58] Therefore, Article 1 of the new law

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55 GODARD B., HURLIMANN T., LETERNDRE M. and ÉGALITÉ, "INHERIT BRCAs, Guidelines for disclosing genetic information to family members: From development to use", in Familial Cancer, 2006, 5, pp. 103–116.

56 See BINET J.R., "Le nouveau droit de la bioéthique: Commentaire et analyse de la loi n° 2004-800 du 6 août 2004 relative à la bioéthique", LexisNexis, Paris, 2005, p. 30 ss.

57 See the judgement of the *Conseil constitutionnel* n. 82-144 DC of 22 October 1982, in www.conseil-constitutionnel.fr. As affirmed by the Constitutional Council, "*le droit français ne comporte, en aucune matière, de régime soustrayant à toute réparation les dommages résultant de fautes civiles imputables à des personnes physiques ou morales de droit privé, quelle que soit la gravité de ces fautes.*"

58 Conseil d'État, *La révision des lois de bioéthique*, Paris, 2009, Cap. IV "*Examen des caractéristiques génétiques: respecter la volonté des personnes et renforcer leur information.*" According to the *Conseil*,

adds Article L.1131-1-1 to the *Code de la santé publique*, which states a specific duty of the physician to inform the person of the risks for family members in cases of diagnosis of a serious disease, if they were not properly informed (para. 1).

The disposition also states the duty of the person concerned to prevent the consequences of genetic abnormalities for her/his relatives, when measures of prevention can be adopted (para. 3). The person may also decide not to be informed about the results of the diagnosis. In this case, if the persons concerned do not feel able to make the communication, the physician is requested to inform the relatives (para. 4). However, the doctor will not reveal either the name of the patient or the genetic abnormality, or the risk associated with it. Basically, the physician has to invite family members to take a genetic test, if he/she envisages the existence of a potential risk.

8. *Ethical principles and freedom of research on genetic information.* According to the legal sources quoted within the previous sub-paragraphs, processing of personal data, in particular genetic information, for scientific purposes implies an exception to the discipline of protection of personal data. This situation is due to the characteristics of the scientific activities and depends on the position of science within the legal systems.

However, as a fundamental right, freedom of research also cannot be considered as absolute, and therefore it must be subject to legislative limitations,[59] in order to protect other fundamental rights. Such limitations are provided for by national constitutions (normally those most recent or recently amended, such as Article 118b of the Swiss Constitution; Article 29 of the Constitution of Bulgaria; Article 18 of the Constitution of Slovenia; and Article 23 of the Constitution of Croatia), and by supranational fundamental legal texts.

The Declaration of UNESCO of 1997 affirms the "responsibility" of researchers and their obligation to comply with principles of primary importance (such as meticulousness, caution, intellectual honesty, and integrity in carrying out their research as well as in the presentation and utilization of their findings; see Article 13), taking into consideration particular attention to research on the human genome. On the other hand, it affirms that states "should take appropriate steps to provide the framework for the free exercise of Research on the human genome with due regard for the principles set out in this Declaration, in order to safeguard respect for human rights, fundamental freedoms and human dignity and to protect public health. They should seek to ensure that research results are not used for non-peaceful purposes" (Article 15).

At the continental level, the need to face the potential collision between research freedom and other fundamental rights can be found within the preambles of the EU Charter of Fundamental Rights as well as the Convention of Oviedo. Furthermore, Article 26 of the Convention of Oviedo permits restrictions to the right to consent in biomedical research, if such restrictions are provided for by law and if they constitute necessary meas-

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the Swiss approach—allowing the physician to be authorised by the public authorities to contact the relatives if the patient refuses to inform them—might affect the trust relationship between the professional and the patient.

[59] See the Italian *Corte costituzionale*, judgment 4 June 1958, n. 36.

ures, in a democratic society, for public safety, prevention of criminal offenses, protection of public health, or of the rights and freedoms of others.[60] Such limits to freedom of research have to be applicable also in case of research on genetic information.

According to Article 89 of Regulation 2016/679, both EU and national laws shall provide "safeguards" in order to implement exceptions due to "public interest, scientific or historical research purposes or statistical purposes" (see also recital no. 34 of Directive 96/46/EC). The apparently wide derogations from privacy law, justified by scientific purposes, have to be implemented on the basis of "ethics principles", which are those rules aimed at making freedom of research consistent with the protection of other fundamental interests of the society, such as the principles of necessity, proportionality, and precaution.

In application of the principle of necessity, derogation from the law concerning data privacy is acceptable only when such rights likely render impossible or seriously impair the achievement of the objectives of the processing (see Article 14, para. 5.b; 13, para. 3.d; 89, para. 2, Regulation (EU) 2016/679). More in general, processing of genetic data is allowed only when their protection is guaranteed (see recital no. 52 of Regulation no. 2016/679) and where it respects "the essence of the right to data protection and provide for suitable and specific measures to safeguard the fundamental rights and the interests of the data subject" (see Article 9, para. 2, let. j).

Article 9 of the regulation implicitly quotes Article 52s, para. 1 of the EU Charter, which, indeed, states that limitations to the exercise of the rights and freedoms recognised by the charter must be provided for by law and must be necessary. In respect to the protection of personal data, this principle is affirmed in the case law of the Court of Justice, such as in the judgement Tele2 Sverige AB (see in particular para. 100).[61]

With reference to biomedical research, necessity entails that there is no alternative to involving persons (especially vulnerable ones) in research activities (see Article 16 of the Convention of Oviedo, points iv and v). Also, the principles of necessity imply that the actual benefits have to be evident, taking into account that "the very nature of biomedical research means that it is uncertain whether an individual will benefit from research participation and any benefit to the person is not the main purpose of research."[62]

(per evitare ripetizione con "in the case" qllq riga sotto) In any case, the conditions of absence of alternatives and evidence of benefits should be applicable only in the case of

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60 ANDORNO R., "The right not to know: an autonomy based approach", in Journal of Medical Ethics, 2004,30, pp. 435–440, especially p. 437. In regard to the conditions and limitations of human rights, see also ÁLVEREZ LEDESMA M. I., "La libertad de expresión en el sistema electoral mexicano desde una perspectiva jurídica", in MONTIEL G. L. and TAMÉS MUNOZ E. (edit by), Libertad de expresión en el proceso electoral 2012, México, PNDU/ONU, 2013.

61 In that judgment the court points out that "terrorism may depend to a great extent on the use of modern investigation techniques, such an objective of general interest, however fundamental it may be, cannot in itself justify that national legislation providing for the general and indiscriminate retention of all traffic and location data should be considered to be necessary for the purposes of that fight (see, by analogy, in relation to Directive 2006/24, the Digital Rights judgment, paragraph 51)."

62 Council of Europe, Steering Committee on Bioethics, Guide for Research Ethics Committee Members, January 2012.

medical interventions,[63] due to the particular position of vulnerability of the patient. In other cases, those conditions are not essential, or their respect should be ascertained with less rigor. Furthermore, Article 52, para. 1, EU Charter allows limitation of fundamental rights, such as privacy, subject to the respect of proportionality,[64] which is another primary principle of the EU legal system.[65] This is true, in particular, when personal life must be protected, including personal data.[66]

In respect to the considered matter, the principle of proportionality imposes a minimisation of the quantity of gathered and processed data (see Article 89, para. 1, Regulation 2016/679).[67] Such data must be relevant and limited to what is necessary in relation to the purposes allowed by law (see Article 5, para. 1, c). This principle also constitutes a limitation on the length of data storage (see Article 5, para. 1, let. e).

Furthermore, an evaluation in respect to proportionality and legitimacy is necessary, taking into account the principle of precaution,[68] i.e., risks for the protection of fundamental rights and freedoms of individuals and notably whether or not the intended purpose could be achieved in a less intrusive way.

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63 Biomedical research is defined by the Additional Protocol to the Convention of Oviedo of 2005 as "research activities in the health field involving interventions on human beings", and also as research concerning genetic information. Use of genetic information for research activities not linked to medical interventions should be prohibited by the discipline of the Convention of Oviedo and its additional protocols (see Article 2, para. 2.b, Additional Protocol concerning Genetic Testing for Health Purposes). However, we can observe the tendency to mitigate the link between data and medical intervention, considering health data "all personal data concerning the physical or mental health of an individual, including the provision of healthcare services, which reveals information about this person's health" (Article 3 of the draft Recommendation on the Protection of Health-Related Data).

64 See ECJ, jud. 15 February 2016, J. N./ Staatssecretaris voor Veiligheid en Justitie, C-601/15 PPU, EU:C:2016:84, paragraph 50. In general, on principle of proportionality ECJ, judg. 9 November 2010, C-92/09 y C-93/09, *Volker und Markus Schecke y Eifert*, ECLI:EU:C:2010:662.

65 The principle of proportionality is also used by the case law of the European Court of Human Rights. See, in particular, ECHR, judg. *Gillow vs. UK*, 24 November 1986, series A nº 109, para. 55, y the ECJ, judg. 20 May 2003, Österreichischer Rundfunk and Others, C-465/00, C-138/01 and C-139/01, EU:C:2003:294, para. 83)

66 See ECJ, judg. 16 December 2008, Satakunnan Markkinapörssi and Satamedia, C-73/07, EU:C:2008:727, para. 56; Id., judg. 9 November 2010, Volker und Markus Schecke and Eifert, C-92/09 and C-93/09, EU:C:2010:662, para. 77; Id., judg. 8 April 2014, Digital Rights Ireland Ltd, joined Cases C-293/12 and C-594/1, ECLI:EU:C:2014:238, para. 52; Id., judg. 6 October 2015, Schrems, C-362/14, EU:C:2015:650, para. 92.

67 According to Authorisation no. 8 of the Italian Authority for Privacy (par. 4.1), the collection of genetic data for carrying out genetic testing and screening is limited to personal and family information which is strictly necessary for the performance of the analysis.

68 According to the principle of precaution in science and technology, see for example ANDORNO R., "The Precautionary Principle: A New Legal Standard for a Technological Age", in Journal of International Biotechnology Law, Vol 1, I, 2004, pp. 11–19; COLCELLI V., "Precautionary Principle Liability in the Food Industry: the search of a general regime in vertical and horizontal Liability", in Rainer Arnold and Valentina Colcelli, (eds), Europeanization through private law instruments, Regensburg, Universitätsverlag, 2016, pp. 249 ff.

In the specific field of research, the application of the principle of precaution implies a risk assessment and a comparison with direct or indirect benefits: As a matter of fact: "Although the anticipated overall benefits of the research project must clearly be higher than the potential risks, the research may not be considered justified if there is a particularly high risk of serious harm."[69] Research activities must also observe other principles such as "distributive justice". As stated, "In biomedical research involving human beings, this implies that the distribution of risk and burden on the one hand and benefit on the other be fair—a principle known as distributive justice". Such a principle should be applied for example to the research participants, who should be those who actually may benefit from experimentation.[70]

9. *Consent and the balance between freedom of research and rights concerning genetic information.* As mentioned above, research activity is considered as a lawful reason to deviate from privacy concerns, especially in respect to consent. Such deviations have to be implemented in compliance with ethics principles that work in order to put in equilibrium freedom of research and other interests protected by the legal system.

However, to achieve a balance between interests, the idea that privacy is an absolute value should be subjected to revision. Legislation about privacy derives from a "proprietary"[71] logic concerning the whole human body and its parts, including genetic data. As a matter of fact, propriety is at the base of the meaning of "privacy" itself, since the origin of the notion can be found in the famous work of Samuel Warren and Louis Brandeis, "The Right to Privacy", published in the *Harvard Law Review* in 1890. In that paper, the notion of privacy was drawn up within the proprietary paradigm, even if from a "spiritual" and not a "physical" viewpoint.

According to European law, we can observe the tendency to overprotect privacy in comparison with other interests. The Court of Justice may be considered as the guardian of this tendency. For example, in regard to the "right to be forgotten", in a leading case regarding Google Spain,[72] the Court of Luxemburg held that the fundamental rights recognised by Articles 7 and 8 (i.e., protection of personal data) of the EU Charter "override, as a rule, not only the economic interest of the operator of the search engine but also the interest of the general public" (para. 97 of the judgement). According to the Court of Justice, the public interest should be "preponderant" in order to overtake individual rights arising from the protection of personal data. In contrast, legislation seems to be less demanding when a deviation from the right to erase requires the existence of "public interest" (see Regulation (EU) 2016/679, recital no. 65).

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69 Guide for Research Ethics Committee Members, issued by the Steering Committee of the Council of Europe on January 2012.

70 Guide for Research Ethics Committee Members, ref.

71 See for example DE WITTE J. and TEN HAVE H., "Ownership of genetic material and information", in Social Science & Medicine, 45(1), August 1997, pp. 51–60. See also CIPPITANI R., "Property paradigm" and protection of rights concerning genetic information, in Diritto e processo/Derecho y Proceso/Right and Remedies, 2016, pp. 261–288.

72 ECJ, judg. of 13 May 2014, Google Spain SL Google Inc., C-131/12, ECLI:EU:C:2014:317.

This approach is justified by the attempt to protect persons from the great risks arising from the massive use of techno-science, and in particular of the ITC or biomedical technologies. Nevertheless, as stated "In the European Convention on Bio-medicine as well as in the Universal Declaration on Human Genome, the approach to protecting data confidentiality would appear to be based on an individualistic concept" (Working Party, Working Document on Genetic Data, p. 8). Indeed, it was also stated that "If we protect privacy effectively, we will not reduce ethics to autonomy, and autonomy to data ownership. Reducing ethics to ownership comes at a high price: ethics that care only about ownership and consented transfers are, by exclusion, indifferent to distributional justice and optimizing social outcomes."[73]

Privacy should be coordinated with other important freedoms or rights recognised by constitutional norms, such as freedom of research (see, for example, the above-mentioned Article 13 of the Charter of the Fundamental Right of the European Union).[74] The Court of Justice itself, in the above-mentioned judgement regarding Google Spain, seems to consider scientific purposes adequate *per se* to deviate from the rights of the data subject (see paras. 72 and 92 of the judgement). The features of genetic data, and the specificity of science from a legal viewpoint, have as a consequence that research on genetic information cannot be reduced to a question of privacy. In particular, as stated above, legal techniques to provide free and informed consent or anonymisation do not always represent solutions to problems arising from the processing and storage of genetic data.

The relevance of scientific activities for society—especially, but not exclusively, for therapeutic reasons—should lead to a different approach. It would be advisable, also in respect to the balance of different interests in so complex a field, to put in place various strategies and new instruments.[75]

On the one hand is the idea that consent serves only as an instrument to prevent external invasions, without taking into consideration the reasons or the interests at the base of such an intervention. As a matter of fact, "The core of both 'privacy' and 'property' involves the same abstract right: the right to exclude unwanted interference by third parties. The only real difference between the two concepts is the kind of relationship that is protected from interference: 'property' principally protects market relationships while 'privacy' protects more spiritual ones".[76]

Consent may be conceived as a set of legal instruments for participating in activities which may concern not only the interests of the "data subject", but also those of third parties and of the community. It should not be considered an instantaneous act, but rather a continuous process, useful for establishing a trusted link among data subject,

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73 TAYLOR P., "When consent gets in the way", in Nature, 6 November 2008, vol. 456, pp. 32–33.

74 See MOLINA DEL POZO F. and ARCHONTAKI C., "Libertad de artes y de Investigación Científica", Libertad de Cátedra, … ref.

75 See VILLANI L., "Biobanche e test rivelatori di informazioni genetiche: spunti di riflessione per un nuovo consenso informato", in Responsabilità civile, 2010, 2, pp. 140 ff.

76 See ACKERMAN B., "Liberating Abstraction", in University of Chicago Law Review, vol. 59, 1992, pp. 317–348, in particular p. 347.

researcher, and the institutions.[77] The consent could also include the decision to voluntarily share information as a common good.[78]

As stated above, scientific activities need a model of consent which is different from the specific one, in particular a broad-based approach. However, it would be consistent with a unilateral and asymmetric logic of consent. A bilateral approach should be elaborated, according to which, for example, consentors are constantly informed on the follow-up of the research, so that they may participate in other research and be invited to events or to participate in associations. On the other hand, the centrality of consent should not be carried to extremes. In order to achieve a balance among the different types of interests, including those related to the data subject, consent may not be considered either as a sufficient or a necessary condition. In many cases, not all personal data have the same value or importance for the individual.

The following aspects should also be stressed: the procedural aspect of the consent, the quantity and quality of information to be provided, the time to make the decision, and the kinds of decisions to be taken should be adequate to the situations.[79]

For example, the International Bioethics Committee of UNESCO, in its document Human Genetic Data: Preliminary Study by the IBC on its Collection, Processing, Storage and Use of 15 May 2002, affirms that "Many tests which reveal genetic information will not have a great deal of significance for the person tested (…). Other tests, however, will have major implications, both for the individual and for relatives. The principle stated above sets out the consent requirements. For practical reasons, it would be unrealistic and unnecessary to require that there be specific consent to the genetic component in any test unless the consequences of this are sufficiently serious enough to justify this" (para. 59 p. 15).[80] Consent, considered alone, could be not sufficient.

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77 AZZINI S., "Biobanche, consenso e fonti del diritto: un caso di eccezionale disordine?", 2010, available at http://www.biodiritto.eu/sito/images/stories/azziniforum2010papersito.pdf.

78 See the document Ethical, legal and social aspects of genetic testing: research, development and clinical applications, ref., p. 41 ff., especially p. 42.

79 See BUNNIK E.M., CECILE A., JANSSENS J.W. and SCHERMER M. H.N., "Informed Consent in Direct-to-Consumer Personal Genome Testing: The Outline of a Model Between Specific and Generic Consent", in Bioethics, 2012, pp. 1–9. The paper, in respect to personal genome testing, uses a "combined tiered-layered-staged model for informed consent", which may be more suitable. This combination "is tiered to provide consumers with options, so as to enable them to choose what types of information on what (categories of) diseases they wish to receive, and especially to opt out of receiving information they do not wish to receive. Layering of information will help limit the otherwise overwhelming quantity of information offered to all consumers in the first layer of the consent process, while it also strives for an 'individual consumer-based' consent, as it offers additional information for those who need that information in order to consent. Finally, a staged set-up of the pre-test information provision process can serve educational purposes and improve the quality of consent. Moreover, subsequent renewal of consent will be required as new test outcomes become available as a result of ongoing genomics research. A combined tiered-layered-staged model for informed consent in PGT would allow for relevant information provision that is both sufficiently complete and sufficiently understandable."

80 As affirmed by the UK Human Genetics Commission, "the difficulties involved in tracing and securing re-consent for different forms of medical research may make obtaining fresh consent imprac-

When issues arising from research reach a general dimension, it would be advisable to define consent of the members of a group and techniques to involve communities and to establish a sort of "collective consent" instead of the individual one. This is the case for the establishment of the program called "deCODE Genetics", approved by the Icelandic state, to gather the genetic profile of all Icelandic citizens.[81]

Furthermore, consent is not sufficient because of the vulnerability of the individual in respect to professionals and/or institutions carrying out research or other activities concerning personal data, or because it is necessary to access the research activities in order to make them consistent with the ethical principles and with legal rules. In those cases, consent in itself is not sufficient to ensure proper protection of individual interests[82] and therefore needs further tools to integrate its effectiveness.

Another important instrument to face the ethical problems concerning the use of genetic information is the control carried out by ethics committees or other third parties. For example, documents dealing with genetic screening for the recruitment of employees recommend requiring the prior assent of the appropriate labour organisation and a specific ad hoc authorisation by an independent committee. Indeed, the person may be compelled to consent to the screening in order to be recruited by the employer.[83]

According to some legal authorities in the field of health, such as the discipline of clinical trials, the expression of consent has to be subject to independent bodies' control, through ethical committees, agencies, or other bodies that allow the evaluation of the activity (see Article 6, para 3, Directive 2001/20/EC). The role of the ethics committee is affirmed by many documents of the Council of Europe in critical situations, such as when health data cannot be anonymised for technical reasons (see para. 12.2, Appendix to Recommendation (97), mentioned above), which normally is the case for genetic data; or if it is not possible, with a reasonable effort, to contact the person who has not given her/his consent to carry out research activities concerning biological material (see Article 21.2, Recommendation (CM/Rec(2016)6). In those cases, the scientific purposes together with an external and independent evaluation carried out by an ethics committee allows for the research institution to overcome the lack of consent.

In this respect, we can also see the draft Recommendation on the Protection of Health-Related Data, which establishes that "The conditions in which health-related data are processed for scientific research must be assessed, where necessary, by the body or

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tical and would seriously limit the usefulness of large-scale population databases" (Human Genetics Commission Inside Information, May 2002).

81 See ÁRNASON V. and ÁRNASON G., "Informed Democratic Consent? The Case of the Icelandic Database", in Trames, 2004, vol. 8/12.

82 OTLOWSKI M., "Developing an appropriate Consent Model for Biobanks: In Defence of 'Broad' Consent", in KAYE J. and STRANGER M. (eds), Principles and Practice in Biobank Governance, Surrey, Ashgate Publishing Limited, 2009, Chapter 5, pp 79–92.

83 See the European Group on Ethics in Science and New Technology in its Opinion no. 18 concerning "Ethical Aspects of Genetic Testing in the Workplace" of 2003, para. 2; see also the document "Ethical, legal and social aspects of genetic testing: research, development and clinical applications" of 2004, elaborated for the General Directorate of Research Commission by a group of independent experts.

bodies designated by domestic law" (Article 16.6).[84] For example, Italian law provides for situations when consent is not necessary if research activity is established explicitly by law or when the processing is foreseen in a biomedical research programme approved on the ground of Article 12-bis Legislative Decree no. 502 of 30 December 1992 and referred to the Authority of Privacy (see Article 110, para. 1, Legislative Decree no. 196/2006).

Another possible solution to achieve a balance of interests involved could be drafting a code of conduct (see Article 40 Regulation (EU) no 2016/679),[85] as highlighted by the General Data Regulation, according to which it is necessary to "calibrate the obligations of controllers and processors, taking into account the risk likely to result from the processing for the rights and freedoms of natural persons" (see Recital no. 98).

EU discipline encourages the adoption of other instruments arising from private autonomy, although it is subject to the control of authorities, such as standard contractual clauses between controllers and processors and between processors, technical standards, and mechanisms for certification (see Recital no. 167).

Other instruments for ensuring accountability and the quality of the institutions and professionals dealing with genetic information need to be refined and developed.[86]

More generally, it is necessary that the consent process be part of a governance framework of "trust, responsibility and accountability", in which the involvement of institutional review boards would be essential.[87]

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[84] With respect to the position of independent authorities, in the judgement Tele2 Sverige AB et oth. of 21 December 2016 (in Joined Cases C-203/15 and C-698/15), the exceptions due to the justification to fight crime are admissible only where they will be reviewed by an independent administrative authority (see para. 120 and 125; see, by analogy, Directive 2006/24, the Digital Rights judgement, paragraph 62; see also, by analogy, Article 8 of the ECHR, ECtHR, 12 January 2016, Szabó and Vissy v. Hungary, CE:ECHR:2016:0112JUD003713814, §§ 77 and 80).

[85] See, for example, United Kingdom Information Commissioner's Office (2012), *Anonymisation:managing data protection risk. Code of practice*, available at www.ico.org.uk/ for_organisations/data_protection/topic_guides/anonymisation.

[86] Article 5 of the Additional Protocol to the Oviedo Convention concerning Genetic Testing for Health, adopted in Strasbourg on 27 November 2008, already stipulates that states must ensure that "a) genetic tests meet generally accepted criteria of scientific validity and clinical validity; b) a quality assurance programme is implemented in each laboratory and that laboratories are subject to regular monitoring; c) persons providing genetic services have appropriate qualifications to enable them to perform their role in accordance with professional obligations and standards."

[87] CAULFIELD T., UPSHUR R.E.G. and DAAR A., "DNA databanks and consent: A suggested policy option involving an authorization model", in BMC Medical Ethics, 2003, 4:1.