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The Legal and Ethical Framework of EU Rare Disease Policies

The Right to Health of Rare Disease Patients and the Legislative Framework for Rare Disease Registries in EU Policy Documents

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Contents

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Introduction

At the heart of the modern European Union is a commitment to the right to health of all citizens. It is an express goal for the policy and legislation of the Union. At the same time, the provision of healthcare for individual citizens remains primarily the concern of the individual Member States. For the vast majority of conditions, a local perspective is sufficient. In some matters of public health, a more international view is needed. Indeed, for the proper functioning of the supranational European single market, with its free-flow of people, the cross-border movement of individuals is not merely a matter of extending commercial and personal movement in theory, cross-border health provision is required as a part of enabling the free-movement of workers. Co-operation and joint planning for health is a necessary part of realising the economic goals of the European Union.

There are parts of medicine, however, that require international co-operation for very different reasons. Rare diseases appear sporadically within local populations, and in small numbers internationally. However, with global communications, and with the development of medicine internationally, there are enormous possibilities to gather together the small cohorts of those patients who suffer from, or who are at risk from, rare diseases so that they can become a viable group first for research purposes and then for the development of effective therapies, and for care. Rare disease registries are at the heart of the medical response to rare diseases

Rare disease registries are systematic collections of personal (socio-demographic) and clinical data, designed to improve knowledge about and understanding of rare diseases, and to translate that understanding into effective health policies (particularly social, medical, and research policies) to facilitate changes in the introduction of changes in the natural history of the disease from prevention to reduction in mortality. This points to a number of requirements for the activities of registries that present ethical and legal questions that must be addressed in the governance framework:

1. The epidemiological methods must be both scientifically and ethically sound;
2. Ethical robustness requires transparent and rigorous independent oversight of registries and research using information from registries;
3. Appropriate and robust personal data protection, including clarity in the ownership of data;
4. Protection of patient rights, including the right of access to health care, and the right against inappropriate genetic discrimination.

This list is not, however, already indicates some of the difficulties in developing the governance regime for rare disease registries. Two traditional safeguards for patients used in medical ethics, and that are to some extent expected without question, are ‘informed consent’ and ‘anonymisation’ (the deidentification of personal data). At the heart of the rare disease registry,

there is a need for on-going identification of individuals to ensure continuity and depth of information, and there is a need for wide inclusion of individuals within the registry. These competing needs from research and medical ethics cause a tension at the heart of the governance question. This is not simply a theoretical problem; individual citizens show a range of sensitivities towards the use of their personal data, and therefore the issue is a practical question for the development of the law and ethics in this area.

This said, within Europe, with our existing lines of cooperation and our aspirations for realising individual's health, we have the opportunity to lead the international community in developing the best environment for individuals with rare diseases. However, this requires a regulatory and political will to achieve this aspect of the right to health. These needs present challenges to the existing regulations to ensure that they accommodate the needs of individuals, without destroying other fundamental rights and freedoms. The starting point of such a consideration is to examine the existing law that is in place and how it impacts upon rare diseases. Thereafter, one must ask how the law can be made more effective to ensure the maximum opportunities that globalisation brings for the response to rare diseases.

At the heart of the rare disease problem is the development of sufficiently robust cohorts to understand and develop responses to the particular diseases. Therefore a first element in the development of this area is the creation of registries. These have to cover extensive populations to enable the identification of individuals. These, like biobanks, need to be registries that contain on-going, updated data about the individuals, so that the progress and expression of particular diseases can be tracked. Therefore, the need for the registries to contain and process identifiable data immediately suggests privacy dangers for the individuals whose data is contained on the registries. Registries, of different character and content, have already been created. Many of the registries have been set up in specific settings. These can relate to the particular medical information that is collected, but they can also relate to the regulatory environment in which they are set. For example, they differ because of following regional and national policies Patient organisations have played a crucial role in the past and remain important stakeholders in the future. However, EPIRARE also assesses to which extend European policies, such as the Patient Rights Directive or the Data Protection Directive, facilitate a common European information infrastructure for rare disease data.

Informed consent of patients and relatives is one possible regulatory pathway. Informed consent, however, is seen by many who work in the field of rare diseases as highly problematic, and having the potential to jeopardise the gathering of information about rare diseases. Alternative regulatory approaches are available, and used in different parts of Europe, exploring different, more solidarity and public interest based approaches to the gathering and processing of personal medical data. Exploring alternative bases for the registries and use of data for rare diseases, given the supremacy of informed consent as a safeguard in medical ethics and medical

law, are difficult issues to raise, even at an academic level. Since informed consent is so ingrained in democratic and ethical expectations, to depart from this regulatory platform would require extremely robust alternative safeguards for individuals, very strong and accepted philosophical arguments, and enormous political courage. However, it is now is an appropriate time to raise them.

The policy landscape will change in the next few years. The Data Protection Directive 95/46/EC is undergoing extensive review, as is the Clinical Trials Directive 2001/10/EC. The Patient Rights Directive 2011/24/EU These revisions give opportunities to explore very basic assumptions about the relationships between individuals in our society, and between autonomy and solidarity. The European Union, in its core Treaties, speaks of our common life together, and particularly about our solidarity. Rare diseases present a situation where our willingness to explore the meaning of solidarity is put to the test. It also tests our collective commitment to realising truly universal rights to healthcare.

In this, WP4's first paper for the EPIRARE project, we aim to explore the legal framework for rare disease registries in Europe, and to argue for a specific legal and ethical framework within which to support the development and operation of healthcare for those with rare diseases. Part 1 is an introductory evaluation of the range of law and ethics currently most relevant to the area. Part 2 considers how to develop a response to the existing law and ethics, and to evaluate the proposed Regulation on Data Protection in relation to the development and maintenance of rare disease registries.

Part 1. The Legal and Ethical Landscape

The rare diseases domain receives considerable support from the EU and its Member States. Unlike other areas of health care there seems to be a clearly defined and accepted need for European cooperation. Due to the number of different rare diseases and the scarce resources in many Member States, European cooperation has the potential to significantly improve the quality of care for rare disease patients. Still, European policies in rare diseases need to be embedded into the 'aquis' and the competence deficit of the EU in the health sector may potentially limit the number and impact of European actions in the field. The area of rare disease registries can serve as an exemplar that highlights the impact of other, primarily non-health policies on the quality of services. We do not aim to refer a lot to the "Health in all Policies" approach as it has a different connotation[1]. However, EU policies such as the orphan drug regulations or the data protection directives have a substantial impact on the policy framework in the field of rare diseases. In particular the Data Protection Directive 95/46/EC is often quoted as a bottleneck. Naturally such a Directive influences a field of healthcare in which patients, or rather their samples and data, travel to receive specialised diagnostics and care. We start our analysis with an overview of the Public Health significance of rare diseases and continue with the ethical and legal benchmarks. Finally we explore the current regulatory framework and set the scene for future actions that might be needed to facilitate better services for patients with rare diseases.

1.1 Rare Diseases – an important Public Health Issue in Europe

Within the policy documents of the European Union rare diseases are defined as diseases that do not affect more than 5 in 10.000 citizens[2]. With a population of 500 million citizens it is obvious that rare diseases are a major Public Health issue and require attention from both the Member States and the European Union. From a legal and ethical perspective we need to make an important differentiation: due to the fundamental rights of each citizen the sheer numbers should not disguise that each and every individual has a right to the highest attainable level of health. However, numbers do matter both in ethics and law when it comes to allocation decisions that determine which resources are raised for societal welfare. We will address this important ethical and legal aspect in more detail.

1.2 Biomedical Ethics and Rare Diseases

Biomedical Ethics has been centred on universal principles that developed during the 20th century. The debate in ethics has been flanked by important

[1] Puska P, Health in all Policies, Eur J of Public Health, 2007, 17 (4) : p 328
[2] Other definitions apply around the world, e.g. see Griggs R, Batshaw M et al, Clinical research for rare diseases, Molecular Genetics and Metabolism, 2009, 96 (1): pp. 20-26

legal decisions such as the Schloendorff case and by the historical review of the Nazi war crimes. The highly influential four principles approach of Biomedical Ethics as developed by Beauchamp and Childress are taken as a ethical-normative starting point / framework and benchmark to elucidate the specific importance of Biomedical Ethics for the rare diseases field.

Firstly, the principle Respect for Autonomy means that people should be able to exercise autonomy that entails that their "opinions and choices" and goals must be considered, respected and acknowledged "unless they are clearly detrimental to others"[3]. In other words, Respect for Autonomy refers to the acknowledgement of the will and value of every individual[4]. This also includes individuals "with diminished autonomy" which should be "entitled to protection" even though an autonomous individual is defined as being "capable of deliberation about personal goals and of acting under the direction of such deliberation" or in other words, "being capable of self-determination"[5]. In the case of rare diseases the notion of autonomy is slightly different as it rather implies the access to medical services and knowledge.

Secondly, the principle of beneficence relates to the need "to do good to every single person" and to the obligation "to act for the benefit of others"[6]. Hence, it adds to the first principle as it aims to strive for the well being of the individual. In other words, beneficence entails the obligation to "maximize possible benefits and minimize possible harms" of the individual[7]. Furthermore, the principle Beneficence is associated with three obligations. Firstly, it "ought to prevent evil or harm", it "ought to remove evil or harm" and lastly, it "ought to do or promote good"[8]. In the public health context the principle of beneficence implies that the benefits for the individual in particular concerning his/her health in implementing a policy or public health activity should or must outweigh the harm, cost and risks compared to the alternative policy options including no policy implementation.

Thirdly, the principle of non-maleficence calls all actors to avoid any harm for every single person and some of its essential aspects / meanings have been an integral part of the ethics of medical professions through the "primum non nocere" concept. Although the principles are partly overlapping, Beauchamp and Childress argue that "obligations of non-maleficence are more stringent than obligations of beneficence, and, in some cases non-maleficence

[3] The National Commission for the Protection of Human Subjects of Biomedical and Behavioral Research (US Congress) (1979). The Belmont Report; Ethical Principles and Guidelines for the protection of human subjects of research. http://ohsr.od.nih.gov/guidelines/belmont.html

[4] Schröder-Bäck P, *Principles for public health ethics – a transcultural approach*. Eubios Journal of Asian and International Bioethics, 2007, 4 (17): pp.104-108

[5] Supra idem Fn iii

[6] Beauchamp, T.L. & Childress, J.F., *Principles of biomedical ethics (6th ed.).* New York: Oxford University Press, 2009

[7] Supra idem Fn iii

[8] Supra idem Fn iii

overrides beneficence"[9]. The principle of non-maleficence is difficult to apply in situations which are characterised through the lack of medical / scientific certainty. Society strives for an acceptable risk/benefit ratio that protects patients with rare diseases from an undue exposure to risks. However, following the principle of autonomy rare diseases patients may also seek alternative treatments which are not (yet) the medical standard or may seek to be treated under off-label use conditions. Some jurisdictions, like the German courts have developed special frameworks for such treatments ("Heilversuch").

Finally, the principle of justice implies that everybody is treated in a non-discriminatory way and that there is a fair and just distribution of benefits, costs, risks and resources. Consequently, nobody should be discriminated, stigmatized or excluded from access to diagnosis and care; in the field of rare diseases patients every patient needs to be given the opportunity to achieve the best possible provision of services based on the available resources and the need of other law subjects to use the same resources. Beauchamp and Childress also consider the influential justice approach of Norman Daniels in their work as one way to give further normative insights what the principle of justice would demand from a just health care system.[10] Following Daniels' "Just Health Care" approach[11] we have moral obligations to keep people in the "normal opportunity range" so that they have chances to live full human lives. The "normal opportunity range" of human beings is co- determined by health that is defined by Daniels as species typical functioning. Accordingly, just societies have to provide a health care system in which everyone has access to decent health care to keep or regain health. Further to questions of access to (often limited) resources and services of health care, justice also implies procedural justice, which refers to fair and transparent decision-making processes. Such a fair and transparent policy making process is of utmost importance in the rare diseases sector to make sure that each patient is given the opportunity to express his / her needs.

However the principle of justice is of particular importance as it builds a bridge from biomedical to public health ethics. Also Daniels recognised over the years that health is not only determined by questions of access to health care but further social determinants contribute to the normal opportunity range. Thus, he further elaborated his approach and his revision – "Just Health: meeting health needs fairly"[12] – includes the wider Public Health perspective. In this work Daniels demands that also social determinants of health are to be distributed in such a way that fair chances for health can be enjoyed by all members of a society.

1.3 Public Health Ethics and Rare Diseases

[9] Supra idem Fn vi
[10] Supra idem Fn vi
[11] Daniels N, Just Health Care, CUP, 1985
[12] Daniels, N. Just Health: Meeting Health Needs Fairly, CUP, 2007.

Next to the central position of "justice" for public health – that was also stressed by other authors of the field [13]– Public Health is often said to be a consequentialist and – other than bioethics – not clinically focused but population focused endeavour[14]. It not only looks at the just distribution of health but also wants to maximize health in a given population – an endeavour that can be held in check by justice considerations.[15] Thus, compared to the "biomedical ethics perspective" (from Beauchamp and Childress and also focused on by the early work of Norman Daniels) that looked at maximizing the benefit of the individual patient (beneficence), the public health ethics perspective captures the normative idea to maximize the benefit of the population. As beneficence is traditionally a principle of individual ethics for the patient-doctor-encounter, it is worth to mention this different normative social ethics perspective and to make it explicit. Next to this "health maximization" public health often considers aspects of efficiency. The public perspective on health encompasses the obligation to use scarce resources efficiently not to waste scarce goods. In short, extending the more clinical bioethics view to a public and population-based perspective, normative considerations of "health maximization" and "efficiency" broaden the normative spectrum of health. In case of conflicting norms – e.g. if respect for autonomy and health maximisation come in conflict when it is considered if immunization should be made mandatory[16] – a balance of this norms has to be found.

To take the consequentialist and social perspective of public health to the context of rare diseases, this might elucidate further ethical argumentation for giving priority to taking rare diseases in view of public health perspectives. it should be stressed that, while for other diseases, the biomedical approach and the public health approach are complementary ways to fulfill the human and patients' rights, in the case of rare diseases, the public health approach is a prerequisite to ensure a quality biomedical approach. In the latter case indeed, the individual physician knowledge is largely inadequate (as is correspondingly the current scientific knowledge) and any present and future approach to quality care (e.g. reference or expert networks) necessarily must take into account individual data collection and sharing. Taken the "burden of disease" of all combined rare diseases shows clearly from the "health maximisation" perspective that rare diseases have to be tackled to maximize population health. The justice perspective that is also very relevant in public health stresses that everyone shall get that kind of health care and live under relevant further determinants of health that allow one to be as healthy as possible. Here is a certain kind of normative overlap / concordance between the "health maximization" and "justice" perspective.

[13] Powers M, Faden R, Social Justice: The Moral Foundations of Public Health and Health Policy, OUP, 2006
[14] Kass NE, Public Health Ethics: From Foundations and Frameworks to Justice and Global Public Health, J of Law, Medicine and Ethics, 2004. 32 (2): 232 -242
[15] Schröder-Bäck, supra idem Fn iv
[16] Schröder-Bäck P, Brand H et al., Ethical Evaluation of Compulsory Measles Immunisation, Cent Eur J of Public Health, 2009, 17 (4): pp 183-186

One could argue now, though, that the public health perspective – looking at populations – considers also efficiency aspects. And once the population of people with rare diseases is stratified down to certain diseases and risk profiles, it becomes clear that improving the health of this population is relatively resource-intensive – and the ethical dilemma is that these are resources that could arguably have been used more efficiently to improve population health elsewhere (e.g. fighting common chronic diseases). Yet, the normative ethical framework of public health takes rare diseases in view and prioritises them for several reasons. Not only that – as one could argue from a consequentialist perspective – the omission of adequately caring for people with rare diseases might leave to overall negative effects because it undermines the feeling and practice of solidarity within a society which might result in more fear or social tensions that ultimately are bad for health and well-being in the population. To argue against prioritising rare diseases – given the high burden of disease that can be reduced – one can "only" lead resource and financial reasons. To invest in curing or preventing rare diseases leads, no doubt, to opportunity costs that reduce health and utility elsewhere. However, the gained utilities and life-years in the sector of rare diseases deliver a significant contribution to health maximisation. And not to do so would lead to significant ethical issues that would be unacceptable from a moral perspective – as the public health ethics principles of justice and respect for the individual's dignity remind us with their normative weight that is prima facie the same as the weight of health maximisation. From this perspective, saving effects shall rather be achieved where it is morally less controversial or even obligatory – e.g. by demanding more self-responsibility.

1.4 The Solidarity Principle and the Right to Health for Rare Diseases Patients

In its 2008 Communication on Rare Diseases[17] the European Union has stressed the importance of the solidarity principle. Referring to the Council Conclusions on Common values and principles in European Union Health Systems[18] the EU highlighted the need for an equitable and solidary access to high quality services in the field of rare diseases. These initiatives are part of a series of policy documents which underline the importance of rare diseases in EU policies: EU Regulations on Orphan Drugs (1999)[19], on Paediatric Use of Medicines (2006)[20], on Advanced Therapies (2007)[21] and

[17] Communication from the Commission to the European Parliament, the Council, The European Economic and Social Committee and the Committee of the Regions on Rare Diseases: Europe's challenges, COM(2008) 679 final, 11.11.2008

[18] Council Conclusions on Common values and principles in European Union Health Systems, OJ 2006/C 146/01.

[19] Regulation (EC) No 141/2000 of the European Parliament and of the Council of 16 December 1999

[20] Regulation (EC) No 1901 and 1902/2006 of the European Parliament and of the Council of 12 December 2006 on medicinal products for paediatric use

[21] Regulation (EC) No 1394/2007 of the European Parliament and of the Council of 13 November 2007 on advanced therapy medicinal products and amending Directive 2001/83/EC and Regulation (EC) No 726/2004

more recently the Commission Communication (2008) and the Council Recommendation on Rare Diseases (2009). The EU Directive on Cross Border Health Care (2011)[22] is another cornerstone as it includes rare diseases as a public health priority in its Article 13. This article may not directly award legally enforceable rights to patients with rare diseases but it paves the way for a common European service landscape in the rare diseases domain. Due to the low prevalence of rare diseases, patients have a need for special services which cannot be provided in all 27 Member States and in particular not in many of the other WHO Europe countries. Insofar, the solidarity principle, as expressed in the EU policy documents, requires a transformation to render it useful.

From a fundamental rights perspective the solidarity principle needs to be associated with the right to health of patients with rare diseases. In the preamble of the 1946 WHO Constitution[23] the right to health is explicitly reaffirmed: "The enjoyment of the highest attainable standard of health is one of the fundamental rights of every human being without distinction of race, political belief, economic or social condition." This right to health has been seen as a programmatic, political goal but it also implies duties for the WHO Member States; in the preamble the WHO underlines: "Governments have a responsibility for the health of their peoples which can be fulfilled only by the provision of adequate health and social measures." In the rare diseases domain this creation of the right to health implies that the special needs of rare diseases patients need to be taken into consideration when countries organise their health systems and provide health services.

In 1948 the United Nations adopted the Universal Declaration of Human Rights[24], which is not binding but has influenced the recognition of fundamental rights globally. In its Art 22 the Declaration states: "Everyone, as a member of society, has the right to social security and is entitled to realization, through national effort and international co-operation and in accordance with the organization and resources of each State, of the economic, social and cultural rights indispensable for his dignity and the free development of his personality." The UN links human dignity to the provision of basic social rights, which include the right to healthcare as one subset of social rights. The right to health of rare diseases patients is bridging the principle of human dignity and the solidarity principle. Further documents such as the 1966 International Covenant on Economic, Social and Cultural Rights (ICESCR)[25] and the 1966 International Covenant on Civil and Political

[22] Directive of the European Parliament and the Council 2011/24/EU on the application of patients' rights in cross-border healthcare of March 9th 2011

[23] The Constitution was adopted by the International Health Conference held in New York from 19 June to 22 July 1946, and signed on 22 July 1946 by the representatives of 61 states.

[24] Universal Declaration of Human Rights (UDHR) was adopted by the United Nations General Assembly (10 December 1948, Paris)

[25] The International Covenant on Economic, Social and Cultural Rights (ICESCR) was adopted by the United Nations General Assembly on December 16, 1966, and is in force since January 3, 1976

Rights (ICCPR)[26] have strengthened the right to health of patients with special needs.

Health in the holistic understanding of the WHO cannot be guaranteed by anyone and the rare diseases domain is a clear example for the virtual limits of the right to health. Thus, the 2000 General Comment No 14 of the UN Committee on Economic, Social and Cultural Rights (CESCR) rather referred to a fundamental right to the highest attainable standard of health: "Health is a fundamental human right indispensable for the exercise of other human rights. Every human being is entitled to the enjoyment of the highest attainable standard of health conducive to living a life in dignity."

In health systems, resources are limited and need to be allocated in accordance with the needs of patients. The Solidarity Principle implies that patients with rare diseases receive a bigger share of the available resources. However, it is also a matter of solidarity that the rights and legally protected interests of individuals and society are balanced. Ideally, society will aim for a concordance[27] of the different rights and interests which is guided by (bio)medical and Public Health Ethics. Consequently, the right to the highest attainable standard of health is the same for every rare disease patient in Europe as it is a fundamental, and therefore universal, right. Nevertheless, the level of services and practical support differs from country to country as the translation of the right into actions depends on the available resources, competitive societal goals and cultural values. Solidarity, equity and justice have manifold dimensions in the case of rare diseases. Despite the policy framework processes are needed which ensure the practical translation of the right to the highest attainable standard of health into legally protected and enforceable rights of rare diseases patients. Solidarity forms the basis of this enterprise.

1.5 The Right to Health and the Solidarity Principle in EU Law

The Charter of Fundamental Rights[28] introduced a limited right to health that is not even as far reaching as the right to the highest attainable standard of health. In Art 35 the EU proposes a conditional right that is not harmonised within the EU. In principle EU citizens have the same right to access high quality health care but only under the conditions of the country they are legally residing in. Such a conditional right seems to contradict the underlying concept of European fundamental rights as it perpetuates a gap between the different Member States. This is clearly a side effect that is equally recognisable in Art 168 of the EU Treaty. The conditional right to access to

[26] The International Covenant on Civil and Political Rights (ICCPR) was adopted by the United Nations General Assembly on December 16, 1966, and is in force since March 23, 1976

[27] Hesse K, *Grundzüge des Verfassungsrechts der Bundesrepublik Deutschland*, 20. Aufl., Heidelberg 1999, Rn. 72

[28] Kokott J, Sobotta C, The Charter of Fundamental Rights of the European Union after Lisbon, 2010, EUI AEL; 2010/06; Distinguished Lectures of the Academy, online accessible at http://cadmus.eui.eu/handle/1814/15208 (last accessed Oct 17th 2011)

healthcare as it is defined in EU law also contradicts the solidarity principle in EU law. Solidarity in a very practical sense is limited to emergency situations and labour accidents. The new Patient Rights Directive seems to challenge these limits but has not yet been tested in real life. There are also important aspects that seem to support the current construction of the right to access to healthcare of the EU. Health is a necessary condition for the exercise of all other fundamental rights and this is in particular obvious in the case of severe rare disorders.

However, unlike human dignity the right to health has not been seen as an absolute right so far. Consequently, law and Public Health Ethics underline that the right to health needs to be balanced with other fundamental rights and legally protected interests. As we have discussed already society needs to aim for a concordance of rights and interests that are equitable balanced. With the current legal framework of Art 168 EU Treaty the EU does not have any right to govern into the national health care systems. Thus, EU law is very limited in its ability to structure and govern the concordance process that is evidently needed to translate the right to health into practice. From the perspective of rare diseases patients who cannot be bound to the limitations of their national health care systems, this is a clear deficit in the construction of the Charter of Fundamental Rights. Nevertheless, the limited and highly conditional fundamental right to access to healthcare is a logical consequence of the weak competence of the EU in the health sector.

1.6 Legal and Ethical Benchmarks for the Right to Health of Rare Diseases Patients (and their Families)

The Right to Health of patients with rare diseases is shaped by the core normative domains we have discussed in the paper. Biomedical Ethics stresses the respect for autonomy, human dignity and the beneficence of services for patients. Public Health Ethics adds a different perspective and highlights the equitable access to services which implies a fair distribution of chances and opportunities. The contribution of law is a schizophrenic one: the Right to Health has been merely evolved as a political “slogan” but with the increasing importance of the right to self-determination and the accessibility of more personalised services for rare diseases patients, the Right to Health is transforming into a real fundamental right which can be exercised by rare disease patients. European rare disease registries are needed to serve as an infrastructure for better services and research in the field of rare diseases. To some extend, such an infrastructure is a necessary condition for the transposition of the right to health into reality. Without the actual transposition of the fundamental right the ethical and legal aims of the right would remain unfulfilled and the right to the highest attainable standard of health would be pure lip service.

1.7 EU Policy Documents and Regulations relevant to the Field of Rare Diseases Registries

Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions on Rare Diseases COM (2008) 679

Rare diseases have been acknowledged by the European Commission as an important Public Health issue and the Commission has been supportive of rare disease registries from an early stage onwards. In its Communication to the other political actors from Nov 11th 2008 the Commission confirmed its long term commitment[29]. A Communication has no direct legal impact but it shapes the policy development and shall therefore be included in the list of important policy documents. In particular in the sections 4.3 and 4.4 the Commission highlights the urgent need for better, and more coherent, epidemiological data on rare diseases. Consequently, the document further elaborates on registries in the section 5.11 and calls the creation of better rare diseases registries as an important action point for European Public Health. The Commission also expresses certain conditions which need to be met to ensure the long term success of rare disease registries: "Collaborative efforts to establish data collection and maintain them will be considered, provided that these resources are open and accessible. A key issue will also be to ensure the long-term sustainability of such systems, rather than having them funded on the basis of inherently precarious project funding". These conditions, if agreed upon by the stakeholders, need to be woven into the legal framework for disease registries.

Council Recommendation on an action in the field of rare diseases (2009/ C 151/02)

As a follow up of the Communication from the Commission to the Committees and the Council, the Council agreed on recommendations for action in the rare disease field on June 9th 2009[30]. The Recommendation addresses the need for rare disease registries in Section II.5: "Consider supporting at all appropriate levels, including the Community level, on the one hand, specific disease information networks and, on the other hand, for epidemiological purposes, registries and databases, whilst being aware of an independent governance." The recommendation does not have a legally binding function but it underlines the commitment of the Member States and the Commission to support a regulatory framework which is supportive of rare disease registries.

Regulation on Orphan Drugs (141/2000) and Regulation on Criteria for Orphan Drug Designation (847/2000)

[29] Online accessible at http://ec.europa.eu/health/ph_threats/non_com/docs/rare_com_en.pdf (last accessed on Oct 17th 2011)

[30] Online accessible at http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:C:2009:151:0007:0010:EN:PDF (last accessed on oct 17th 2011)

On December 16th 1999 the European Parliament and the Council finalised Regulation 141/2000 on orphan drugs which has substantially helped to give incentives for research in the field of rare diseases[31]. The orphan drug regulation gave the field of rare diseases more visibility at the EMA (at that time EMEA) and has triggered the development of other policy documents. Beyond the specific regulation on orphan drugs, the common legal framework in the field of pharmacovigilance may also facilitate the coherent collection of data on rare diseases. Manufacturers are obliged to follow up on pharmaceutical products which are marketed and sold within the EU. Such data could be a valuable source of information for rare disease registries.

Data Protection Directive 95/46/EC

Whereas this legislation will be discussed in detail in the second part of this report, some observations can usefully be made at this point. Data protection is often highlighted as a major bottleneck for the interoperability of health information systems in Europe. The field of rare diseases is therefore not a special case but may need more support than other domains of the health sector to guarantee, in the end, an equitable access to high quality services. Other registries in the Public Health sphere might be able to operate with non-identifiable data that do not fall, or in a limited extend, under the scope of the Directive. However, rare diseases are special and registries may need to be prepared to comply with the Directive.

The Data Protection Directive is a constituent part of the Commission's public health policy, and it represents the core regulatory instrument serving both the protection of privacy and the access to information / data sharing[32]. The task is therefore twofold, as both dimensions need to be explored. The potential tension between privacy and the public good raises the question how the balancing process ought to be structured and whether costs deriving from data protection should be taken into account, too. It should not be forgotten that the Data Protection Directive was drafted and proposed by DG Market as a harmonising directive that aims to facilitate the exchange of data within the internal market. With the reallocation of responsibility and the shift from DG Market to DG Justice, Liberty and Security (now DG Justice) the implementation of the Directive changed its focus and a human rights approach was adopted.

The Data Protection Directive follows certain principles and differs between different degrees of data. Often, data entered into rare disease registries will qualify as sensitive data under Art. 8 of the Directive. The processing of such data is prohibited unless it is explicitly allowed. A justification can follow from the informed consent of the data subject, the law or, to a limited extend, from a preponderant interest of another data subject. Most registries under

[31] Online accessible at http://ec.europa.eu/health/files/eudralex/vol-1/reg_2000_141/reg_2000_141_en.pdf (last accessed on Oct 17th 2011)
[32] Zika E, Schulte in den Bäumen T et al, Sample, data use and protection in biobanking in Europe: legal issues, Pharmacogenomics, 2008, 9 (6): pp 773 – 781

national law are set up or confirmed by law to allow a maximum coverage of cases without any bias that would naturally follow from the application of the informed consent principle.

As indicated before, under the Data Protection Directive the level of protection depends on the contents of the data and the purposes the data are collected for. In Art .8 para. 1 the directive incorporates an enumerative list of sensitive data categories. The use of this categorisation has been questioned, and in particular in the field of genomics lawyers feel difficulties to differentiate between health data that are health data per se and "ordinary" data which turn into health data if certain diseases are analysed.

With regard to rare diseases we face the same dilemma that has been highlighted often in the fields of biobanking and genomic research. The samples of patients are not sensitive per se but the data contained are describing the physiological state of the donor and may qualify as sensitive data. The data that identify the sample are not sensitive as they do not contain health information. Any secondary data are either sensitive or not, depending on the purpose they are processed for. Thus the application of Art 8 of the Data Protection Directive depends on the epidemiological model/purpose of the registry and the work actually carried out with the samples and data. From a governance perspective it should be analysed whether the legal reasoning and the societal understanding are congruent or not. This refers to the views on genetic exceptionalism, bioethical reasoning and Public Health ethics. The given concept of the Data Protection Directive is highly questionable as it does not allow a case sensitive individual decision.

Within the core medical setting Art 8 par 3 of the Directive is applicable which is an important exemption to the prohibition of the processing of sensitive data. Art 8 par 3 is part of the harmonised field covered by the Directive. Contrasting to this, Art 8 par 4 covers the field of Public Health, and due to Recital 34, also research carried out for Public Health purposes: *"Subject to the provision of suitable safeguards, Member States may, for reasons of substantial public interest, lay down exemptions in addition to those laid down in paragraph 2 either by national law or by decision of the supervisory authority".*

As the word "may" in 8 para. 4 indicates, this paragraph is not fully harmonising as Member States are free to decide whether and to what extend they use the exemption. Even if they use the exemption Member States still have a substantial discretion to set up standards for the "appropriate safeguards" mentioned by the Directive[33]. The further assessment of the legal framework, and in particular the analysis of the re-cast of the Data Protection Directive, will have to show whether Art 8 par 4 has the potential to serve as a common legal base for European rare disease

[33] Schulte in den Bäumen T, Paci D, Ibarreta D, Data Protection and Sample Management in Biobanking – a legal dichotomy, Genomics, Society and Policy, 2010, 6(1), pp 33-46

registries. In a worst-–case scenario all 27 Member States may have to negotiate and agree to a specific contractual framework which addresses the special needs of the rare disease community. We call this worst case scenario as it would imply that all 27 Member States and possibly also the EFTA/EEA countries would have to agree upon a common data protection standard. If further collaboration with partners from overseas is intended legal experts would have to draft a global gold standard which fulfils all the data protection regulations. However, Art 8 par 4 also enables the rare disease community to work on a best practice that can be translated into a normative framework in the field of data protection. Such a best practice would need to respect the core data protection.

Patient Rights Directive 2011/24/EU

On March 9th 2011 the Directive on Patient Rights in Cross-border Healthcare has been finalised. The Directive is now in the transposition phase. which ends in October 2013. The Directive aims to transform the jurisdiction of the European Court of Justice into a normative framework[34]. Since the mid-1990's the European Court of Justice had to decide several cases which demonstrated the tensions between the Four Freedoms of the European Union and the competence of the Member States in the field of health. While the Court used principles of competition and consumer protection law to extend the impact of the market freedoms on health, the Treaties after Maastricht confirmed the very limited role of the EU in health policies. In principle the Directive only affects a small margin of the healthcare expenditures in the 27 Member States; it might be of particular importance in the field of rare diseases. This has been acknowledged by both the EU and the Member States and led to the inclusion of a special article on rare diseases in Art 13 of the Directive.

Patients with rare diseases may often require health services from other Member States. The Directive fosters the establishment of networks of reference, it clarifies the reimbursement rules and it provides patients with better access to information regarding health services in other Member States. The rare diseases domain is relatively well organised and should be able to use the new opportunities. Consequently, Art 13 stresses the access to information and the provision / reimbursement of services which are not provided by the country of residence. In addition, Art 15 strengthens European cooperation in the field of health technology assessment that can be of further added value for the rare disease community. However, the impact on rare disease registries needs further elaboration and research. In principle the Patient Rights Directive implies the creation of a common European health data infrastructure but this will not be achieved within the time of the transposition of the Directive. It does not follow from the Directive under which condition registries will have access to data generated within

[34] Sauter W, Harmonisation in Healthcare: The EU Patient Rights Directive, TILEC Discussion Paper Series, 2010, available at SSRN: http://ssrn.com/abstract=1859251 (last accessed online on Oct 17th 2011)

this system. Registries for rare diseases that already use or aim to use data sets from other data sources may not fall under the scope of the Directive at all.

Miscellaneous

Additional policy documents issued by the European Union may serve as benchmarks for the development of best practices in the field of rare disease registries. Such documents shape the work of Commission agencies, in particular the European Centre for Disease Prevention and Control (ECDC) in Stockholm or the Eurostat regulations that ensure a high quality of statistical data for policy making[35].

Despite these technical regulations the overarching policy frame of the European Union such as the Charter of Fundamental Rights plays a vital role for the policy making process in the field of rare diseases. Furthermore the jurisdiction of the European Court of Justice, the European Court of Human Rights and the jurisdiction of the national courts need to be observed to get a full picture of the policy situation. Beyond the positive law the courts have developed a substantial body of decisions that strengthen the self-determination autonomy of law subjects[36]. The field of medical law, and in particular the area of patient rights, has been promoted by courts around the world since the beginning of the 20th century. These different legal traditions in the various Member States need to be included into a policy analysis before a common European approach for rare disease registries is finalised.

[35] An overview is provided by Eurostat at http://epp.eurostat.ec.europa.eu/cache/ITY_OFFPUB/KS-31-09-254/EN/KS-31-09-254-EN.PDF (last accessed on Oct 17th 2011)

[36] In particular the European Court of Justice is also stressing the role of patients as consumers in a "health market", see Sindbjerg Martinsen D, Vrangbeak K, The Europeanization of Health Care Governance: Implementing the Market Imperatives of Europe, Public Administration, 2008, 86 (1): 169–184.

Part 2: Changing the Law to Respond to Rare Disease Registers

2.1 Understanding harm and the fear of harm.

Rare diseases can affect many individuals. Whilst they could be individuals who suffer from the disease, they could equally be carriers of genes that relate to the disease, or they could be relatives of those who have have the diseases or genetics relating to the disease. They represent a range of individuals and cannot be assumed to be of one type. There is not a large amount of public opinion survey data specifically about individuals with rare diseases, who are in the penumbra of individuals who are touched by rare diseases, or in relation to the public opinions about dealing with rare diseases generally and the implications of participating in health information registries to assist medical research.

When one considers the surveys that are available about genetic privacy or about the development of biotechnology, then one can draw analogies from the data. The EC FP6-funded research PRIVILEGED project undertook a literature survey about public attitudes towards genetic privacy and biobanking.[37] What became clear from that survey was first that there was a great need for specific research in the area to understand how individuals perceive their privacy in relation to this type of information. Second, it was equally clear that in relation to questions about the role of science in society, about trust in different researchers and professionals, in the development of commercial research and the questions of benefit sharing, and in relation to the right to know (and not to know) about conditions, and the duty to tell family members (or the right not to tell family members), individual participants in the research gave answers that displayed a range of sensitivities (attitudes) towards their privacy.[38]

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[37] http://www.privilegedproject.eu/ (last visited 20th May, 2012).

[38] European Commission (2005) *Social Values, Science and Technology.* Special Eurobarometer 225/Wave 63.1. http://ec.europa.eu/public_opinion/archives/ebs/ebs_225_report_en.pdf (last visited 20th May, 2012). Relevant questions: Q15.a.3, Q17; Gaskell, G., Stares. S., Allansdottir, A., Allum, N., Corchero, C., Fischler, C., Hampel, J., Jackson, J., Kronberger, N., Mejlgaard, N., Revuelta, G., Schreiner, C., Torgersen, H. and Wagner, W. (2006) *Europeans and Biotechnology in 2005: Patterns and Trends.* Final report on Eurobarometer 64.3 http://ec.europa.eu/public_opinion/archives/ebs/ebs_244b_en.pdf (last visited 20th May, 2012). Relevant question: Q5.3; Gaskell, G., Stares. S., Allansdottir, A., Allum, N., Castro, P., Esmer, Y., Fischler, C., Jackson, J., Kronberger, N., Hampel, J., Mejlgaard, N., Quintanilha, A., Rammer, A., Revuelta, G., Stoneman, P., Torgersen, H. and Wagner, W. (2010) *Europeans and Biotechnology in 2010: Winds of Change?* Brussels: European Commission DG Research http://ec.europa.eu/public_opinion/archives/ebs/ebs_341_winds_en.pdf (last visited 20th May, 2012). See particularly chapter 5. See also Gaskell, G., Allansdottir, A., Allum, N., Castro, P., Esmer, Y., Fischler, C., Jackson, J., Kronberger, N., Hampel, J., Mejlgaard, N., Quintanilha, A., Rammer, A., Revuelta, G., Stares. S., Torgersen, H. and Wagner, W. (2011) "The 2010 Eurobarometer on the Life Sciences." *Nature Biotechnology* 29(2): 113–114 http://www.nature.com/nbt/journal/v29/n2/full/nbt.1771.html (last visited 20th May, 2012).

From this analysis, one can see that one cannot assume that individuals will take one position, because they have or are touched by a rare disease. It is safer to assume that individuals will take a range of opinions about how their data can and should be used. For example, in relation to whether or not informed consent is necessary for the processing of their personal data. If 58% of individuals indicate that they would not require specific informed consent for each research processing of their data contained on a rare disease register, or even to place their data on such a register, 42% hold an opposite view. And both views, in our society, are held to be legitimate if arguable views. One can see, for example in Scandinavian countries, that there is a greater acceptance of the operation of public registers for epidemiological studies, and that this is seen within individuals' concept of their 'privacy', but even there the range of views is still apparent.[39]

Why is it important, when considering the governance of rare disease registries, to think about the range of expressed opinions? In creating rare disease registries we want to include as many people as possible in a project that is on-going and for a good end. However, coercing individuals into participation against their will seems to be neither ethical, within the general construction of our common values as indicated in the human rights legislation and our European Union foundation treaties. Further, it could be massively counterproductive in terms of inclusion. When individuals feel threatened, they are unlikely to participate freely and early in projects. When this is about going to seek medical help for rare diseases, the treatment of personal information should not become something that deters individuals from seeking help, no matter how logical inclusion on a registry seems to be to those who wish to undertake research and find cures. The governance structure needs to be more sympathetic, and the job of creating robust, reliable and trustworthy registries needs to be undertaken over time. Trustworthy implies a practical proof that the scientific community creating registries is worthy of trust; we cannot assume that we are worthy, we must show that we are by being trustworthy. Unfortunately, in many jurisdictions, that record is not well established and the public may be right to be wary initially.[40]

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[39] For further discussions about this area, see Høyer, K. (2003) "'Science is really needed – that's all I know': Informed consent and the non-verbal practices of collecting blood for genetic research in northern Sweden." *New Genetics and Society* 22(3): 229–244; Høyer, K., Olofsson, B-O., Mjörndal, T. and Lynöe, N. (2004) "Informed Consent and Biobanks: a population-based study of attitudes towards tissue donation for genetic research." *Scandinavian Journal of Public Health* 32: 224–229; Høyer, K., Olofsson, B-O., Mjörndal, T. and Lynöe, N. (2005) "The Ethics of Research Using Biobanks. Reasons to Question the Importance Attributed to Informed Consent." *Archives of Internal Medicine* 165: 97–100; Høyer, K. and Lynöe, N. (2006) "Motivating Donors to Genetic Research? Anthropological reasons to rethink the role of informed consent." *Medicine, Health Care and Philosophy* 9: 13–23. For a general discussion, see Townend, D., Taylor, M. J., Wright, J., and Wickins-Drazilova, D. (2009) "Privacy Interests in Biobanking: A Preliminary View on a European Perspective." In: Kaye, J. and Stranger, M. *Principles and Practice in Biobanking Governance*. Farnham: Ashgate Publishing Ltd, pp. 137–159.

[40] Indeed, in our modern society, we are encouraged to take a position of 'distrust' in society. It is arguably part of our social duty in free markets as individual consumers to be distrustful of institutions in order to call them to account and ensure that they follow their duties. See,

Individuals indicate that they have a range of concerns about their privacy in relation to this sort of registry. Specific research is needed in relation to rare disease registries, but the analogies available support this conclusion and would suggest that individuals will give a range of opinions that should be respected. The second question about harms is the type of harm people are concerned about. There seem to be two sorts of harm: direct harm and a consequential harm. Consequential harm is perhaps easiest to identify and address. Individuals are concerned about their data being disclosed to individuals who then use it to discriminate against them in an unacceptable manner. Many of the opinion surveys indicate that some individuals have concerns about, for example, insurance companies, their employers, or perhaps parts of the State, gaining access to their genetic (or by extension medical) data, and using it to discriminate against them. This is a fear of a consequential loss resulting from the breach of data security. This manifests itself to different levels. Some report no concerns about such sharing of data, whereas for others it is a major concern. There is also, of course, a debate about who should have access to different types of personal data.[41] The second type of concern is a more abstract sense of breach; one's right (to privacy) is violated by the breach, the disclosure of data to an unauthorised person, of itself, regardless of any consequential damage.

This more abstract sense of harm is more difficult to address. It can be understood, however, when one considers the analysis provided by Thomas Murray on this point. In the 2009 Nuffield Council of Bioethics Public Lecture, Dr Murray drew on feminist analysis of the abortion debate in the US, and identifies that disputes either relate to individuals' 'interests' or to their 'identity'.[42] He argues that where the dispute is about interests, there is the opportunity to negotiate and compromise; where the dispute is about 'identity', i.e. that the sense of injustice felt goes to the way that the individual constructs their personal sense of identity, then it is not a matter that can be resolved by negotiation and compromise. Whereas this does not offer solutions, it is highly instructive in understanding the differences in individuals' responses to breaches in their privacy. Some see the breach as a question of their interests, and can negotiate a settlement; others see the

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for example, Luhmann, N. (1988) 'Familiarity, Confidence, Trust: Problems and Alternatives,' in *Trust. Making and Breaking Cooperative Relations*, ed. D. Gambetta. Basil Blackwell, 94–107; Sztompka, P. (1999) *Trust. A Sociological Theory*. Cambridge University Press; Warren, M.E. (1999) *Democracy and Trust*. Cambridge University Press.

[41] See, for example, Ashcroft, R. (2007) "Should Genetic Information be Disclosed to Insurers? No." ('Head to Head' discussion with Holm, 2007 below). *British Medical Journal* 334: 1197; Holm, S. (2007) "Should Genetic Information be Disclosed to Insurers? Yes." ('Head to Head' discussion with Ashcroft, 2007 above). *British Medical Journal* 334: 1196; Van Hoyweghen, I. and Horstman, K. (2008) "European Practices of Genetic Information and Insurance: Lessons for the Genetic Information Nondiscrimination Act." *Journal of the American Medical Association* 300(3): 326–327.

[42] See, Nuffield Council on Bioethics, 2009 Public Lecture: http://www.nuffieldbioethics.org/video/new-genetic-recipes-are-we-cooking-trouble-synthetic-biology (last visited 20th May, 2012); Murray, T. H. (2011) "What Synthetic Genomes Mean for Our Future: Technology, Ethics and Law, Interests and Identities." *Valparaiso University Law Review* 45(4): 1–28; Murray, T. H. (2011) "Interests, Identities, and Synthetic Biology." *Hastings Centre Report* 41(4): 31–36.

matter as damaging their identity, and then the matter has much greater significance for the individual.

2.2 Making the ethical case for solidarity

Autonomy has a primary place in modern medical ethics. As has been shown above, the origins of ethical concerns stem from concerns about the instrumentalisation of individuals, often for claims about 'the greater good'. Thus, in the archetypal expression of biomedical ethics devised by Beauchamp and Childress, autonomy is first in a list of four elements: "autonomy", "non-malevolence", "beneficence", and "justice".[43] In relation to rare disease registries, privacy is often seen as the fundamental right that is in play, and at risk. The primacy of autonomy can have the effect of raising privacy to an absolute right. This, however, runs contrary to its construction in international, and European, law. Under the Universal Declaration of Human Rights, Article 12, "No one shall be subjected to arbitrary interference with his privacy, family, home or correspondence, nor to attacks upon his honour and reputation. Everyone has the right to the protection of the law against such interference or attacks".[44] Here the key word is "arbitrary". There are clearly, within this construction, times when overriding a right to privacy is appropriate. Likewise, in the European Convention on Human Rights, Article 8, whereas subsection 1 establishes that "Everyone has the right to respect for his private and family life, his home and his correspondence", subsection 2 provides that "There shall be no interference by a public authority with the exercise of this right except such as is in accordance with the law and is necessary in a democratic society in the interests of national security, public safety or the economic well-being of the country, for the prevention of disorder or crime, for the protection of health or morals, or for the protection of the rights and freedoms of others". Privacy is not an absolute right for the individual, it is a right that is held in balance with other human rights, and with the rights of others. The question becomes when the rights of others override the privacy (or other fundamental rights and freedoms of particular individuals).

2.2.1 The Utilitarian approach

Commonly, in the operation of 'public interest' appeals, one can see rather Utilitarian considerations being used. Courts are asked to see the public interest as a justification for overruling the rights of an individual, and there is very little structure to the remainder of the decision. Following the theories advanced by Bentham and Mill, a calculation designed to ensure that the decision accords the greatest happiness is made. Of itself, this is difficult to criticise. However, the exercise of this calculation can have devastating effects for individuals, because the calculation does not take effect of the multiplying effect of considering the benefit accrued to the sum of the

[43] Beauchamp, T. L. and Childress, J. F. (2008) *Principles of Biomedical Ethics* (6th Edition). Oxford: Oxford University Press.
[44] United Nations (1948) *Universal Declaration of Human Rights.* http://www.un.org/en/documents/udhr/ (last visited 20th May, 2012).

majority. A single individual cannot benefit from the multiplication of the number of individuals enjoying the benefit, except in the most abstract sense; the effect of aggregating small individual benefits for each member of the majority, however, can be disproportionate to the loss of amenity suffered by the individual whose rights are overridden.

Thus, in rare disease registries, a Utilitarian approach might be to suggest that the public benefit to be had by including individuals on the registry, regardless of whether they do or do not agree with their inclusion, overrides the harm and suffering endured by the individual who is included against his or her will. The underlying problem is that Utilitarianism, as a consequentialist approach, does not recognise 'rights' as overriding norms in and of themselves. The guiding factor in the ethical probity of an action is entirely the sum of utility produced by the action. In this way it is unsatisfactory in rights-based communities.

2.2.2 A rights-based approach to the public interest

Rights-based approaches often become grounded on the issue of competing rights. When two or more individuals assert rights in a particular situation, systems based upon the overriding supremacy of rights find the resolution of rights rather difficult. Townend has argued that it is possible to construct a rights-based approach to the public interest.[45] He uses a three-step approach. The first step is a refining of the Utilitarian approach taken above. Townend suggests that the problem in the Utilitarian approach, as characterised above, is that the individual's loss is calculated against the mass of individuals. By considering the individual (A) who stands to lose his or her rights in a balance with a single, foreseeable individual (B) who stands either to benefit from this overriding of A's interests (or to be harmed by not overriding A's interests) a greater fairness in the balance can be achieved. In that equation, the rights of individuals are considered and the multiplication effect is disengaged.

This, however, is arguably simply a refinement of the Utilitarian calculus, and therefore remains problematic to rights based theories. The second step is to consider how A makes a claim to his or her interests. Ethics and morality concern human decision making; all human action is open to moral (or ethical) scrutiny. This includes the act of making a rights claim. Townend argues that when one considers the equation of asking whether it is appropriate to override the interests of A when confronted with B, A must consider within the particular rights based argument, whether that claim is morally justified. Thus, if A is operating within Kant's categorical imperative, A must ask whether asserting his or her right when the evidence is that

[45] Townend, D. (2004) "Overriding Data Subjects' Rights in the Public Interest." In: Beyleveld, D., Townend, D., Rouillé-Mirza, S. and Wright, J. (eds.) *The Data Protection Directive and Medical Research Across Europe*. Farnham: Ashgate Publishing Ltd, pp. 89–101; see also Townend, D. (2012) *The Politeness of Data Protection: Exploring a Legal Instrument to Regulate Medical Research Using Genetic Information and Biobanking.* Maastricht University Press.

making such an assertion will cause a greater harm to B is universalisible or whether it uses B merely as an end to A's rights claiming rather than treating B as an end in his or her own right. Similarly, if A is following a Rawlsian model, A must consider if the choice to opt for maintaining the rights of an individual even when the result is to cause greater harm to another is an acceptable position to take within the veil of ignorance. This, Townend argues, grounds the consideration of the public interest firmly in rights-based approaches.

The third step in Townend's approach is to ask whether the law can require this calculation from an individual and, indeed, make the calculation on behalf of the individual. First, the creation of laws, based on the reasoning used in step two about the necessity of morality in rights based decision making requires that law's, where they have to be made, must be made morally and require moral outcomes. Second, if the society has to legislate, it is imperative that it requires an individual to act in the way that he or she would require him or herself to act if he or she considered the options. In this way, although it is controversial, an individual can be assumed to take the moral course, where a law is required in society. Arguably, in relation to the development of rare disease registries, it is necessary for the law to act. The question then becomes, is this an appropriate harm situation to presume to override the privacy rights of individuals in the public interest (i.e. in the rights of particular foreseeable individuals)?

2.2.3 Is the informed consent choice a free choice?

A variation of this discussion relates to a different aspect of this area. Let us assume that, on balance, a public interest approach is not seen as politically appropriate, and we remain with an informed consent model for the determination of whether an individual will participate in a rare disease registry. Is the choice that the individual has one that he or she can exercise considering only his or her own sensitivities and feelings. How far, when exercising an informed consent decision of whether or not to participate in a rare disease registry, is this a free choice, and how far is it one that must consider other people. Again, Townend has argued from the position taken above, and from analogies with the English law of Equity, that individuals have responsibilities to others when considering participation in medical research.[46] Steps one and two above are applicable here. An individual should consider his or her own interests against the interests of foreseeable others, and as a matter of morality, should consider the harms that others will suffer as a result of his or her decision. This does not necessarily require the individual to respond in a particular way. It does, however, require the individual to make the calculation and to take the decision-making seriously.

[46] Townend, D. (2012) *The Politeness of Data Protection: Exploring a Legal Instrument to Regulate Medical Research Using Genetic Information and Biobanking.* Maastricht University Press.

2.3 Data Protection and Rare Disease Registries: what sort of reform?

We argue very strongly that sui generis legislation for rare disease registries is not appropriate. They concern sensitive personal data, and this is already the concern of the Data Protection Directive in the EU, and we will argue here that this can be, and is being revised, to make a very effective piece of legislation for rare disease registries. More importantly, there are major practical concerns that mitigate against advocating sui generis legislation. First, the time that it will take to gain sufficient political momentum for new law at an international level will be very long. Legislation is needed now, not in five to ten years. Second, any legislation that is created will have to fit within the existing framework of rights; new law will have to dovetail with existing law, and even a sui generis piece of legislation will require amendments and accommodations by other law. If the Data Protection regime, and its the revisions suggested by the European Commission to that regime, were disastrous for rare disease registries, or gave no effective route through the Directive and its successors for the effective development and management of rare disease registries, then there would be an argument for seeking to overcome the practical problems of sui generis legislation. We will argue that this is not necessary within the proposed and current regimes.

We will consider the current proposals for revision of the Data Protection Directive (95/46/EC) first, because they indicate how the regulatory future could look, and because they indicate the direction that the Commission, following extensive consultation, wishes to take in the next years, and thereby could be amenable to in the interpretation of the current law before the new law is implemented.

2.4 The Proposals for European Data Protection Reform

The European Commission published its proposals for the reform of the Data Protection Directive 95/46/EC on 25th January 2012.[47] The reforms seek to build upon the current Directive, particularly in ensuring much greater harmonisation of approach to the fair and lawful processing of personal data across the EU, EEA, and when dealing with third countries. Since their publication in January 2012, the proposals have been commented upon by the Council, and are, as of July 2013, awaiting debate in the European Parliament. Since the January 2012 text is not formally amended yet, this report will comment first upon the Commission's text, and then briefly upon the subsequent developments. There will be a further amendment to this report following the voting in the European Parliament on its tabled amendments.

The first, and most striking, proposal is for the creation of a Data Protection Regulation in place of the Directive.[48] This would have direct effect in each

[47] http://ec.europa.eu/justice/data-protection/index_en.htm (last visited 20th May, 2012).
[48] http://ec.europa.eu/justice/data-protection/document/review2012/com_2012_11_en.pdf (last visited 20th May, 2012).

Member State,[49] reducing the opportunities for implementational discretion between the States.

At one level, much of the architecture of the proposed Regulation is very similar to the structures of the current Directive. The players are essentially the same: data subjects, data controllers, data processors remain in place, with the addition of "recipient" being "a natural or legal person, public authority, agency or any other body to which personal data are disclosed".[50] adding greater clarity in the scope of 'personal data' under the current law, "Genetic data", "biometric data", and "data concerning health" are all separately defined within PArt. 4.[51] This clearly indicates that rare disease registries concern data regulated by the Regulation. Again, the regime operates on the basis that personal data must be processed fairly and lawfully, that data controllers owe duties to data subjects, and data subjects have rights to protect their interests. However, the Regulation also recognises that there are situations where the public interest can and should prevail over data subjects' rights, and that in such situations alternative safeguards are required.

The proposed Regulation maintains the distinction between information provisions and lawful processing. Indeed, a separation is also maintained between gathering data directly from an individual and gaining the data indirectly, and then again between processing data for a first, identified and disclosed purpose and processing it for unforeseen, secondary purposes. Finally, processing data for health purposes and for research purposes are given separate attention under the proposed Regulation. The route that is taken for a particular category of processing is crucial. This has, arguably, under the Directive 95/46/EC produced different approaches between Member States, particularly as have been raised under the current Directive's Article 8(4). The implementation of a Regulation will go some way to reducing these differences; the requirement for the European Commission to develop 'best practice' for different types of processing under the Regulation will give a further opportunity for the development of a effective rare disease registries if there is a political will.

[49] Whereas Directives must be implemented in the Member State's domesitc law, Regulations are binding upon each Member State's legal regime in the form that they are passed by the European Union.

[50] Proposed Article (PArt.) 4(7).

[51] PArt. 4(10)"'genetic data' means all data, of whatever type, concerning the characteristics of an individual which are inherited or acquired during early prenatal development;"
4(11) "'biometric data' means any data relating to the physical, physiological or behavioural characteristics of an individual which allow their unique identification, such as facial images, or dactyloscopic data;"
4(12) "'data concerning health' means any information which relates to the physical or mental health of an individual, or to the provision of health services to the individual".

2.4.1 Gathering data directly from an individual

Much of the gathering of data for rare disease registries will be a secondary gathering. However, it is likely that as such registries become more prominent, the inclusion of patients will be foreseeable. Therefore, it is equally arguable that when individuals seek treatment for, what become identified as rare diseases, it is not inappropriate to indicate to them that their data could be included on a rare disease registry. This is not to prejudge the question of whether or not their informed consent is need for such a processing. However, when data are gathered directly from an individual, PArt. 14 indicates the information that must be given to a data subject.[52] Arguably, it is not difficult or inappropriate to supply the necessary information to an individual in this face-to-face data gathering situation. However, the question of informed consent will be dealt with below.

2.4.2 Gathering data indirectly about an individual

There will be situations where data is passed to a second person for processing. In the first instance, the majority of such transfers will be foreseeable in the original gathering of the data. For example, when an individual presents him or herself for treatment for a rare disease, the transfer of personal data about that individual to the rare disease registry will be foreseeable, and the necessary information about that transfer should be given to the individual at the initial gathering of the data. The data can similarly be given at any point in the treatment relationship.

It is possible that for some reason that transfer is unforeseen at the initial gathering of the data, or perhaps after the patient has completed treatment. For example, a new register could be developed, or a new test could identify the disease information in a new way. These are issues about secondary processing of data, and they will be dealt with below. There is an indirect gathering of data situation that is not related to a direct gathering of data from the immediate data subject. When data is gathered from a patient, it is highly likely that data will also be gathered at that point about members of the data subject's family. The current and proposed law is not clear on the 'group nature' of genetic or other medical data, and so one must rely on a simple interpretation of the general law. Personal data under PArt. 4(2) "means any information relating to a data subject", and a data subject under PArt. 4(1) is

> "an identified natural person or a natural person who can be identified, directly or indirectly, by means reasonably likely to be used by the controller or by any other natural or legal person, in particular by reference to an identification number, location data,

[52] PArt. 14 (1)(a) "the identity and contact details of the controller...; (b) the purposes of the processing...; (c) the period for which the personal data will be stored; (d) [rights concerning access, rectification, erasure, or to object to processing]; (e) to lodge a complaint to the supervisory authority...; (f) the recipients or categories of recipients of the personal data; (g) [details about intended third country transfers of the data]; (h) any further information necessary to guarantee fair processing...".

> online identifier or to one or more factors specific to the physical, physiological, genetic, mental, economic, cultural or social identity of that person".

It is strongly arguable that relatives of a patient are included in this definition of data subjects. At first sight, this would seem catastrophic to the creation of rare disease registers. However, such data subjects are only identified indirectly. This is then covered by PArt. 14(5), and particularly 14(5)(b). In such cases, giving information about processing of personal data about what we might call 'family data subjects' will only be necessary where it does not prove "impossible or would involve a disproportionate effort". Arguably, those family members who are possible to identify without disproportionate effort will be those who the registry will want to include anyway.

2.4.3 Informed consent.

The second part of the requirements concerns the conditions under which processing is lawful. This is found in Proposed Chapter II of the proposed Regulation. As in the current law, the general processing of personal data is treated separately from the processing of special personal data, and again, there is a ban on processing special personal data that is lifted in particular circumstances. As under the present law, the conditions for the lawful processing of personal data (as they appear in PArt. 6) are: 6(1)(a), where the data subject has given consent; (b) where consent can be inferred from a contract made by the data subject that requires the processing of the data; (c) that the data controller is under a legal obligation that requires the processing; (d) that the "processing is necessary in order to protect the vital interests of the data subject;" (e) that the "processing is necessary for the performance of a task carried out in the public interest or in the exercise of official authority vested in the controller;" (f) "processing is necessary for the purposes of the legitimate interests pursued by a controller, except where such interests are overridden by the interests or fundamental rights and freedoms of the data subject which require protection of personal data, in particular where the data subject is a child. This shall not apply to processing carried out by public authorities in the performance of their tasks." PArt. 6(3) indicates that the legal 6(1)(c) and (e) must be carried out in relation to EU law or Member State law, where "The law of the Member State must meet an objective of public interest or must be necessary to protect the rights and freedoms of others, respect the essence of the right to the protection of personal data and be proportionate to the legitimate aim pursued." Further, PArt. 6(2) indicates that "Processing of personal data which is necessary for the purposes of historical, statistical or scientific research shall be lawful subject to the conditions and safeguards referred to in Article 83."

We will address PArt. 83 below, but even if that route is not available, in terms of the general processing of personal data for the creation of rare disease registers, there is a route that is available through PArt. 6 that does not require informed consent. With Union Law or Member States legislation, processing of rare disease information could be made lawful by accepting

that it is in the public interest. This is a point where specific lobbying is needed to ensure a political decision to harmonise on this point is taken, where informed consent is not possible. However, there may be another route using secondary processing, again, this will be addressed below.

The construction of the provision for the protection of sensitive personal data is familiar to those who know the current law. PArt. 9(1) creates a moratorium on the processing of sensitive personal data.[53] The remainder of PArt. 9 indicates where 9(1) will not apply. Again consent is prominent in 9(2)(a) and (b), as is (c) processing "to protect the vital interests of the data subject or of another person where the data subject is physically or legally incapable of giving consent", (d) processing by certain associations and not-for-profit organisations for their purposes, (e) where the data are published by the data subject, (f) where the data are needed in the course of legal actions or (j) in relation to criminal convictions. Particularly pertinent to rare disease registries, beyond informed consent, are PArt. 9(2)(g) to (i). Again, the moratorium is lifted in the public interest where that is a matter of EU or Member State law, and again where sufficient safeguards protect the "data subject's legitimate interests".[54] The same arguments made above hold for this requirement. PArt. 9(2)(h) indicates that the prohibition is lifted when the processing is for health purposes under Article 81 or, under PArt. 9(2)(i), for research purposes under Article 83. These provide interesting routes for processing of sensitive personal data, in much clearer forms than under the current law.

Therefore, leaving aside any other research or specifically health routes, it is possible to use the public interest as a mechanism for developing a route for processing by rare disease registries that does not require informed consent. The question of whether this is desirable seems, from the literature, to be contested. This is a matter that requires open discussion within EPIRARE.

2.4.4 Processing for unforeseen purposes.

One route to the processing of data in a rare disease registry could be through secondary processing. Many epidemiological studies look at large data sets, for example, cancer records, to see how a disease is progressing in the population. Under the current Directive such 'further processing' is ambiguous because of the drafting of the section. Directive 95/46/EC current Article 6 provides:

> "(1) Member States shall provide that personal data must be:
>
> [...] (b) collected for specified, explicit and legitimate purposes and not further processed in a way incompatible with those purposes. Further processing of data for historical, statistical or scientific

[53] "The processing of personal data, revealing race or ethnic origin, political opinions, religion or beliefs, trade-union membership, and the processing of genetic data or data concerning health or sex life or criminal convictions or related security measures shall be prohibited."
[54] PArt. 9(2)(g).

> purposes shall not be considered as incompatible provided that Member States provide appropriate safeguards".

This is awkward because it is unclear as to whether the 'further processing' relates to the original purpose - that the personal data are collected for stated purposes and then further processed, for example, analysed, coded, anonymised, stored, destroyed, for those stated purposes. If this was the case, why would processing for "historical, statistical or scientific purposes" require special consideration? The inclusion of these categories could be interpreted to mean that the further processing could be for other, unspecified but compatible purposes.

The proposed Regulation makes this much clearer. PArt. 6(4) indicates that:

> Where the purpose of further processing is not compatible with the one for which the personal data have been collected, the processing must have a legal basis at least in one of the grounds referred to in points (a) to (e) of paragraph 1. This shall in particular apply to any change of terms and general conditions of a contract.

As can be seen, this is much clearer, with a focus directly on the processing being for unforeseen purposes at the point of the original collection of the data. With this provision, it is arguable that the processing of data gathered for the diagnosis and treatment of patients with rare diseases (or, indeed, any disease) could be further processed, quite compatibly with the original stated purposes, for the creation of a rare disease registry. This would have been a great advance on the current law but for one thing. Whereas in the current law, Article 6(1)(b) applies to both personal and sensitive personal data, as that distinction is only drawn in Articles 7 and 8, in the proposed Regulation the clause dealing with the further processing of personal data is located in PArt. 6, which contains the provisions for the lawful processing of personal data and not sensitive personal data, which is separately dealt with in PArt. 9. It is very strongly arguable that this means that the further processing provision does not apply to sensitive personal data under the proposed Regulation. This is a point that needs to be clarified and lobbied upon in relation to the new Regulation. All that is required, if there is the political will, is for PArt. 6(4) to be repeated in PArt. 9.

2.4.5 Proposed Article 81.

Processing of Sensitive Personal Data for health purposes are particularly addressed in the proposed Regulation under PArt. 81. First, under PArt. 81(1), the processing of

> "personal data concerning health must be on the basis of Union law or Member State law which shall provide for suitable and specific measures to safeguard the data subject's legitimate interests, and be necessary for:

> (a) the purposes of preventive or occupational medicine, medical diagnosis, the provision of care or treatment or the management of health-care services, and where those data are processed by a health professional subject to the obligation of professional secrecy or another person also subject to an equivalent obligation of confidentiality under Member State law or rules established by national competent bodies; or
>
> (b) reasons of public interest in the area of public health, such as protecting against serious cross-border threats to health or ensuring high standards of quality and safety, inter alia for medicinal products or medical devices; or
>
> (c) other reasons of public interest in areas such as social protection, especially in order to ensure the quality and cost-effectiveness of the procedures used for settling claims for benefits and services in the health insurance system."

The coverage again does not make rare disease registries explicitly part of the health processing provision of the Regulation. This is disappointing, and should be brought to the attention of the legislators. It is arguable that rare disease registries could be included under the public interest discretion in parts (b) or (c), but it would be better to have this as a distinct inclusion.

There is a greater problem, however. PArt. 81(1) clearly poses the same problem as Article 8(4) under the current Directive, namely that this open textured discretion allows different Member States to take different approaches to the law in this area. PArt. 81(3) allows that:

> "The Commission shall be empowered to adopt delegated acts in accordance with Article 86 for the purpose of further specifying other reasons of public interest in the area of public health as referred to in point (b) of paragraph 1, as well as criteria and requirements for the safeguards for the processing of personal data for the purposes referred to in paragraph 1."

Arguably, this is a much better route for lobbying for rare disease registries, as it would ensure a harmonised approach to the issue.

2.4.6 Proposed Article 83.

PArt. 81(2) makes it clear that processing of personal health data for research purposes is regulated by PArt. 83. Arguably rare disease registries fall under this provision rather than PArt. 81. Under PArt. 83(1),

> Within the limits of this Regulation, personal data may be processed for historical, statistical or scientific research purposes only if:

> (a) these purposes cannot be otherwise fulfilled by processing data which does not permit or not any longer permit the identification of the data subject;
>
> (b) data enabling the attribution of information to an identified or identifiable data subject is kept separately from the other information as long as these purposes can be fulfilled in this manner.

This seems to be a very general permission for the processing of data for research purposes. Indeed, it connects to both PArt. 6 and 9 (the conditions for the lawful processing of personal and special personal data, respectively), and to PArt. 5(e) on the length of time for which data can be stored. It seems to clarify the position that is rather opaque in the current Directive, where the research provisions are scattered around the Directive. Thus, in the proposed Regulation, it seem that processing for research purposes is lawful in its own right, although the information provisions apply.[55] PArt. 17 establishes "the right to be forgotten and to erasure". However, PArt. 17(3)(b) and (c) make it clear that this shall not apply "to the extent that the retention of the personal data is necessary" for issues of public health (PArt. 81) or research (PArt. 83), respectively.

There is one remaining question about research. When is research not research? Arguably, when it is for a commercial purpose and not simply for its own sake. it is difficult to imagine, in the current economic climate, that research, particularly scientific research, is not connected to commercial outcomes. The same is true in medical science. The purpose of research is to make therapy products, or at least to feed into that chain of development. Proposed Recital 126 gives an indication that this reality is understood, although it could be more explicit:

> "Scientific research for the purposes of this Regulation should include fundamental research, applied research, and privately funded research and in addition should take into account the Union's objective under Article 179(1) of the Treaty on the Functioning of the European Union of achieving a European Research Area."

2.4.7 Impact Assessment, Prior Authorisation and Prior Consultation.

Arguably one of the most important developments in the proposed Regulation is about the management of risk. Under the current law, all data controllers should register their proposed processing and then comply with the law. Data protection authorities (Supervisory Authorities) are required, under Article 20 of the Directive, to identify "processing operations likely to

[55] See also the wording of Proposed Recital 88, "For the purposes of processing for historical, statistical and scientific research purposes, the legitimate expectations of society for an increase of knowledge should be taken into consideration."

present specific risks to the rights and freedoms of data subjects and shall check that these processing operations are examined prior to the start thereof". Supervisory Authorities are under-resourced, generally, and prior checking has not been vigourously pursued in many jurisdictions. The proposed Regulation attempts to address this reality. Data controllers are no longer required to register all processing. However, PArt. 33 requires them, where processing presents particular risks to the data subject, to make an 'impact assessment' of their proposed processing, and then comply with PArt. 34 requirements for "prior authorisation" and "prior consultation".

PArt. 33(2)(b) indicates that health data, and the sorts of data likely to be included in rare disease registries, present the sorts of risks that will require an "impact assessment". The details of the impact assessment are outlined generally in PArt. 33, and include a need to evaluate the views of data subjects or their representatives about the proposed processing. Data controllers (or processors) are required to consult the supervisory authority prior to processing, where the impact assessment, PArt.34(2)(a) "indicates that processing operations are by virtue of their nature, their scope or their purposes, likely to present a high degree of specific risks; or (b) the supervisory authority deems it necessary to carry out a prior consultation on processing operations that are likely to present specific risks to the rights and freedoms of data subjects by virtue of their nature, their scope and/or their purposes..." Under both PArt. 33 and 34, the Commission is empowered to create "standards", "standard forms" and "procedures" within which these requirements operate.

This new proposal could be seen as an onerous imposition. However, one of the key themes in the development of public trust and confidence is transparency and taking individual's concerns seriously. We suggest that the rare disease register community should seize these aspects of the new Regulation enthusiastically, ensuring that the impact assessment and prior authorisation and prior consultation are not just minimal compliance approaches, but are robust and respectful devices to ensure not only that data subjects rights are protected, but that they are seen to be protected.

2.4.8 Best Practice

Throughout the proposed Regulation, there are requirements or opportunities given to the Commission to create what are effectively 'best practice' documents and requirements for different aspects of the operation of the Regulation or for different sectors. In line with the comments made in relation to prior authorisation, consultation, and impact assessment, the rare disease registries community should offer rare disease registries as a case study for the development of such best practices. Using expert opinion and public opinion surveys, both general and directed to those with or affected by rare diseases, an evidence-based best practice could be established to coincide with the implementation of the new law.

2.4.9 Miscellaneous provisions

Multi-centre and Cross-border Registries. Under the proposed Regulation, the issue of multi-centre, particularly cross-border processing is addressed. The current law is criticised because where processing occurs between different jurisdictions, there can be a great deal of confusion about the managing the different data protection requirements. In line with the EC's desire to see greater harmonisation, PArt. 51(2) establishes

> Where the processing of personal data takes place in the context of the activities of an establishment of a controller or a processor in the Union, and the controller or processor is established in more than one Member State, the supervisory authority of the main establishment of the controller or processor shall be competent for the supervision of the processing activities of the controller or the processor in all Member States, without prejudice to the provisions of Chapter VII of this Regulation.

Data Protection Officers. In cases of large companies (more than 250 employees) or where "the core activities of the controller or the processor consist of processing operations which, by virtue of their nature, their scope and/or their purposes, require regular and systematic monitoring of data subjects", the controller or processor is required to appoint a "Data Protection Officer".[56] That officer has duties to ensure that the controller and processors are aware of their regulatory duties, to develop the necessary records and monitoring of the regulatory observance, particularly the impact assessment and to responses to the supervisory authority.[57]

2.4.10 Developments in the Council and Parliament to date

At this time – July 2013 – there is no formal amendment of the text upon which to comment. The Council's response to the Commission's text showed a range of opinions on all the points discussed above. However, it is the Parliament's response that arguably causes more concern for rare disease registries and research. Over 3,000 amendments have been tabled. Whilst some, for example that of A Voss show awareness of the needs of epidemiological and other medical research, a large number of tabled amendments take a very strong privacy as individual autonomy line. Thus, there are a number of amendments that seek to change PArts 81 and 83 such that informed consent would be required in all but the most exceptional circumstances. Amendments are tabled that would remove the clarifying effect of PArt 6(4), and further amendments would remove the discretion proposed for the Commission in the development of 'best practice'. It is yet to

[56] PArt. 35.
[57] PArt. 37

be seen whether the revisions to the data protection regime will be achieved through a Regulation, or whether a revised Directive will be used.

Since these amendments are tabled for discussion and are currently under negotiation, further discussion of them in this report is not particularly useful. A further revision of this Report is, however, necessary once the changes are clearer, and the shape of the revision to European data protection is clear.

2.5 The Current Law: the Data Protection Directive 95/46/EC

Until a revised Law is passed and implemented, Directive 95/46/EC is in place (and was discussed in this report in part one). We argue that it should now be interpreted in line with the proposed Regulation. Those familiar with the current law will already have seen the similarities between the Directive and the proposed Regulation. We argue that the proposed Regulation indicates the policy thinking and direction that should inform the interpretation of the current law. First, this means that Member States should seek to harmonise their approaches to the current law. We argue that this is particularly important in areas of public health research and, in the area of rare disease registry regulation. It will be necessary to agree the position of rare disease registries within the Regulation - particularly how they will sit in relation to public health activity, whether they are seen as governed by PArt. 81 or 83, or whether they are matters that require special legislation under PArt. 6 and 9. As soon as this is resolved, this should prompt a harmonised approach to the current Directive 95/46/EC Article 8(4).

The shape of the current law is very similar to that described above. Personal data is very broadly defined, arguably already covering genetic data and other medical data included in rare disease registries. Data subjects can include both patients with rare diseases and carriers of conditions, and their relatives. Again, the operation of the Directive is separated through the fair and lawful processing of personal data into the conditions required to inform individual about the processing of their data (Articles 6, 10 and 11), and the conditions that make processing lawful (Articles 7 for personal data, and 8 for sensitive personal data). Data subjects have rights to access their data, and to demand rectification, or erasure, or to prevent the processing of their data, but these are generally limited and in the case where alternative safeguards are in place in national law these rights can be severely curtailed.

The information provisions, in relation to the information that must be given to the data subject from whom data are gathered directly and indirectly, are much the same as explained in relation to the proposed Regulation above. The difference in ‘further processing’ has also been explained. Using the interpretation of PArt. 6(4) in the proposed Regulation as an indication of how the Directive’s Article 6(1)(b) should be interpreted, it is arguably the case that the current law, where data are transferred from treatment to registry, is a compatible further processing of the data.

2.6 The Ownership of Data.

Data Protection law both in its current and proposed forms does not address the 'ownership' of data issue. This is, in large part, because the protection of personal data has its roots in privacy, and whilst there is the concept of 'proprietary privacy', personal data is much more firmly lodged in the literature of 'informational privacy' and 'decisional privacy'.[58] Ownership of medical data traditionally becomes an issue with its commercialisation, particularly in intellectual property. There are arguments that such information should be owned by the data subject, and that this has implications for the possible exploitation of such information.[59] Indeed, the use of personal data for commercial ends is a concern to some respondents in public opinion surveys. At this point, however, we argue that the ownership of medical data question is not a central issue.

Where it becomes an important issue is in the ownership of the data sets themselves. This is particularly the case in relation to what happens if data sets are privately owned and the company owning the information collapses economically. We would then argue that the 'nemo data quod non habet' rule should apply - that one cannot give what one has not got - and in particular, that it is imperative that the purchaser of the bankrupt stock (i.e. the data set) should be bound by the same duties to the data subjects that the original owner had. Indeed, there is a very strong argument that in such situations, new purchasers should be subject to authorisation from a public authority (e.g. the data protection Supervisory Authority) and if a suitable purchaser cannot be found that the data set should become public property. By extension, where a data set is sold, or where a company owning a registry is taken over, in those situations the suitability of the new owners should be tested by the Supervisory Authority prior to purchase.

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[58] See, for example, Allen, A. L. (1997) 'Genetic Privacy: Emerging Concepts and Values.' In: Rothstein, M. A. (ed.) *Genetic Secrets: Protecting Privacy and Confidentiality in the Genetic Era* Yale: Yale University Press, pp. 31–60; DeCew, J. (2008) "Privacy." *The Stanford Encyclopedia of Philosophy.* Edited: Zalta, E. N. http://plato.stanford.edu/archives/fall2008/entries/privacy/ (last visited 20th May, 2012).

[59] See, for example, Townend, D. (2003) "Who Owns Genetic Information?" In: Sandor, J. (ed.) *Society and Genetic Information: Codes and Laws in the Genetic Era*, Budapest: CPS–Central European University Press, pp. 125–144; Beyleveld, D. and Brownsword, R. (2001) *Human Dignity in Bioethics and Biolaw*. Oxford: Oxford University Press.

2.7 Conclusion

- The overwhelming sense that the WP4 team has about the current law is that, with specific tailoring to the needs of the rare disease community, the existing and proposed Data Protection law provides an excellent starting point for a governance regime that will give confidence to the public to participate not only in the registries but in healthcare generally.

- The rare disease registry community must be clear in the position that it wishes the law to take. For example, it must resolve the differences of opinion expressed about informed consent.

- Thereafter it must lobby to develop the political will to interpret the Directive and then the Regulation in a way that facilitates the valuable public health work of rare disease registries that are clearly in the public interest.

- This immediately requires clear, harmonised approaches to the current Article 8(4) to produce standard safeguards for the treatment of data subjects' data in rare disease registries such that Member States will have confidence to provide a specific lifting of the prohibition on the processing of special personal data for rare disease registries.

- A decision must be taken that clearly shows that medical research that could lead to commercial therapies remains 'scientific research' for the purpose of the Directive and the proposed Regulation.

- The placement of rare disease registries within PArt. 81, 83, or 6 and 9 must be clarified.

- Before a full lobbying approach can be devised, a number of further questions need to be answered in the second part of WP4's work towards a legal and ethical best practice model.

2.8 Initial Questions for the development of best practice.

1. How far can confidentiality be maintained when data is kept in an identifiable (or coded) state?
    a. What is the difference between confidentiality and anonymity?
    b. How far is 'anonymous' (deidentificatied) data possible in rare disease registries?
        i. Is an asumption of identifiability a better starting point for the design of safeguards for participants in rare disease registries?

2. How far is informed consent possible and desirable for the development of rare disease registries?
    a. Why should the collection of data for rare disease registries by compulsory?
    b. Why is informed consent such an important safeguard?
    c. What is the meaning of an informed consent? (Is it a licence, a contract, a permission - what status does and should the informed consent take in relation to rare disease registries?)
    d. Is informed consent an effective safeguard?
    e. Are there alternative safeguards that are as effective?
    f. What are the dangers of removing informed consent as a safeguard?
    g. Is only "informed, specific consent" applicable or is "informed, broad consent" appropriate?
    h. How should unforeseeable research proposals be addressed in consent?
    i. Who is in a position to give consent on behalf of vulnerable individuals?
    j. When giving consent, is it a free choice, or is an individual constrained by considerations for other people?
    k. Are dynamic consent models applicable?
    l. How far can personal data about people who have died be used when the consent they have given does not cover a new scenario?
    m. How far can the wishes of family members influence the questions about using personal data from dead people?

3. If informed consent is used, what information should be given?
    a. Whilst the general purpose of the registry is possible to describe, how much detail is necessary?
    b. Who determines the sufficiency of information that is given to individuals?
    c. What is the role of research ethics committees (and other authorities) in the monitoring of informed consent?

   d. How far is informed consent an effective licence to the registry, or does it have to operate to standards beyond the contractual obligations it has made to participants?

4. What data security measures must be in place?
   a. What is the purpose of data security, and what are the available options?
   b. How far can economic cost be an option in developing data security measures? What constitutes (and who determines) an acceptable level of risk?
   c. How can data security be successfully communicated to participants (without breaching data security)?

5. Who should have access to the data and for what purposes?
   a. What level of qualification and professional association is required for access to the registry?
   b. What level of vetting is necessary, and by whom?

6. What sort of sanctions should be used for breaches? (cf. what sort of sanctions should be available?)
   a. What sort of harms are breaches of rare disease registries comparable to in society?
   b. How far should sentencing guidelines or tariffs for fines reflect local considerations?
   c. How far should the views of individual participants be taken into account when determining the level of punishments for breaches?
   d. Is this a matter for professional bodies alone?

7. What sort of practical and ethical review and monitoring should be in place? Who should undertake this work?
   a. How far is this within the remit of currently available research or clinical ethics committees?
   b. Is an ‘in-house’ ethics committee acceptable?
   c. How far should the review be linked to other authorities, for example, the national data protection authorities?
   d. How far is it necessary to have international ethics committees with international standards for rare disease registries?

8. How should the development of registries relate to the development of new treatments and the availability of new therapies?
    a. Should registries operate within the normal economic expectations and practice of intellectual property and pharmaceutical development?
    b. Does participation in a registry imply a share in the economic operation of that registry and the work that is made possible through it?
    c. Who should own and develop rare disease registries?
        i. Are they necessarily public bodies, or could private bodies develop such registries?
        ii. Would such bodies pose particular ethical and legal problems?
        iii. What mecahnisms and expectations should be in place in the event of the economic failure of a rare disease registry?

9. What special measures should be taken to ensure that international data sharing is both technically and ethically possible?
    a. Is data sharing desirable?
    b. How far is data sharing possible between independent registries?
        i. is it necessary to create registries to an international data standard, or can there be individual variations between registries and still effective data sharing?