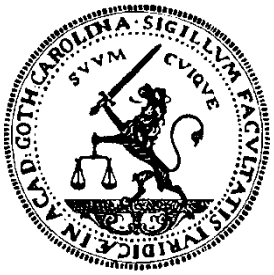


FACULTY OF LAW

Lund University

Seyed Adib Hosseini

The Impact of the GDPR on the Genomic Research

Striking a Balance Between Personal Data Protection and Genomic Research Interests

JAEM03 Master Thesis

European Business Law

30 higher education credits

Supervisor: Ana Nordberg

Term: Spring 2022

Table of content

Summary

The GDPR is a progressive regulation that preserves the right to data privacy and provides a high level of personal data protection. A wide range of data falls under the scope of the GDPR, and genetic data is not an exception. There is a great demand for various activities that involve processing genetic data, and the GDPR introduces specific conditions for processing them. Meanwhile, Member States have the power to maintain or introduce more requirements and limitations to ensure a high level of data protection. In this regard, genomic sequencing has several usages for health-related services in the public and private sectors. Among these, genomic research is highly important due to its impact on health care. It is essential that researchers can conduct research and ensure that their projects comply with the GDPR. Since public health is a shared competence, there are situations where the extra restrictions introduced by the Member states can be problematic and challenge genomic research projects.

This thesis explains the necessity of having a data privacy regime from a legal and ethical perspective. It looks into how the GDPR affects genomic research and whether further national measures can restrict the processing of genetic data. It also examines how different national rules can impede the free flow of personal data within the European Union. The analysis shows the interplay between Article 9(2)(j) and Article 9(4) GDPR concerning the right to introduce further measures by the Member States and the right to conduct research in light of the GDPR, respectively. Moreover, it discusses different solutions to mitigate the problem. This thesis concludes that the GDPR provides a sufficient legal basis for personal data protection and conducting genomic research in accordance with the current legal framework. While it is not possible to limit the Member State's discretion regarding public health, the Commission mitigates the situation through soft law mechanisms, such as promoting and developing policies that can accelerate collaboration and harmonization.

Keywords: Privacy, Data Protection, GDPR, Healthcare, Genomic Research, Precision Medicine, Personalized Medicine, Artificial Intelligence, Soft Law, Harmonization.

Preface

My Master's program has come to an end, and I am grateful that I had the opportunity to study European Business Law at Lund University. It has been an honor to spend two years with many excellent teachers, lawyers, and students from all over the world.

I would like to thank my supervisor Ana Nordberg for her great support and inspiration during my studies at the Faculty of Law. I dedicate this thesis to the memory of my grandmother, Amina, who was unable to see my graduation. I also dedicate this dissertation to my mother, Sara, for being there for me throughout my life. Finally, I dedicate my work to Iranian women for their constant human rights and equality efforts.

List of Abbreviations

AI	Artificial Intelligence
CFR	Charter of Fundamental Rights of the European Union
DTC	Direct-to-consumer
ECHR	European Convention on Human Rights
EHU	European Health Union
EU	European Union
GDPR	General Data Protection Regulation
TEU	Treaty on European Union
TFEU	Treaty on the Functioning of the European Union
PMI	Personalized Medicine Initiative
WP29	Article 29 Data Protection Working Party

"To preserve the values that we cherish, in science as in medicine, we must be careful never to become, consciously or unwittingly, the instrument of forces that would subordinate those values for selfish or misguided goals of their own. Ethics of knowledge and ethics of innocence must never become dissociated, lest either one without the other becomes a vehicle for ignorance or a tool of oppression".

Salvadore E. Luria
The Nobel Prize in Physiology or Medicine, 1969

1. Introduction

1.1. Background

Nowadays, various public and private healthcare entities employ precision medicine to provide personalized treatments based on patients' genomic data. The development of genomic sequencing offers an excellent opportunity for health-related services ranging from medical diagnosis to direct-to-consumer genetic tests (DTC GT). Meanwhile, biomedical research plays a critical role in genomic research developments through using different methods, such as cloud computing in large-scale genomic data processing and collaborative data sharing. Employing genomic data for treatment, research, or commercial purposes poses new ethical and regulatory challenges regarding genetic test consent and data protection.

These developments have many benefits, but they also bring responsibilities. It is necessary to introduce mechanisms that preserve the patients' rights, especially regarding data protection and safety. For many years, the legal concerns regarding human genetics have been complex, and new achievements in the last 50 years have increased this complexity more than before.[1] Scientific discoveries should not end up discriminating or endangering humans.[2] Research should have ethical intentions without harmful ideas.[3]

Regarding data protection, physicians are responsible for keeping their patients' information secret, and by any breach of this rule, they find themselves before the competent authorities.[4] With this in mind, there are situations where physicians are required to disclose the information, which makes the principle of confidentiality vague, and sometimes it gets difficult to know where to draw the line. In extreme examples,

[1] James R Sorenson, 'From Social Movement To Clinical medicine' in Aubrey Milunsky and George J Annas (2nd eds), *Genetics and the Law* (Plenum Press (Springer) 1976) 467
[2] Salvadore E. Luria, 'Biological Roots of Ethical Principles' in Aubrey Milunsky and George J Annas (2nd eds), *Genetics and the Law* (Plenum Press (Springer) 1976) 408
[3] Art 6-8 World Medical Association Declaration of Helsinki: ethical principles for medical research involving human subjects (Adopted by the 18th WMA General Assembly, Helsinki, Finland, June 1964)
[4] Jean V McHale, *Medical Confidentiality and Legal Privilege* (1st edn, Routledge 1993) 3

such as a crisis or in specific diseases like AIDS, where other people's lives are in danger, it is difficult to decide whether to preserve patients' information or public interests.[5]

When it comes to precision medicine and genomic testing, physicians face unique challenges that are more complicated. There are no generally accepted practices or standards in precision medicine because sometimes, even though genetic knowledge is available, it cannot be fully understood, or there are no treatments for some diseases.[6] It is challenging for an individual to fully understand the range and limits of a genomic test result since they can contain unclear clinical significance.[7] Some researches discuss that it is imperative to ensure that even data subjects should not self-identify themselves because any unexpected discovery of diseases could be stressful for the person.[8]

There is a similar situation in precision medicine and pharmaceutical companies. Although they are innovative health care models, they raise serious privacy concerns. One of the critical challenges is balancing the need to protect the privacy of data subjects and the need to give access to these data to the researchers. Personal data in the European Union has been recognized and protected by the EU Charter of Fundamental Rights,[9] the Treaty on the Functioning of the European Union,[10] and the EU general data protection regulation.[11] Several Articles of the GDPR explain why genomic data and relevant information could be considered 'Personal Data' and fall under the scope of the GDPR.

Article 4(1) defines personal data as any information relevant to an identifiable natural person who can be identified, whether directly or indirectly. Since genetic data

[5] Jean V McHale (no 4) 3
[6] Scott P Mcgrath and others, 'Legal Challenges in Precision Medicine: What Duties Arising From Genetic and Genomic Testing Does a Physician Owe to Patients?' [2021] 8(1) Frontiers in Medicine<https://www.frontiersin.org/articles/10.3389/fmed.2021.663014/full> 1-2, accessed 27 August 2022
[7] Leila Jamal, 'An Ethical Framework for Genetic Counseling in the Genomic Era' in Glenn Cohen and others (eds), *Consumer Genetic Technologies* (Cambridge University Press 2021) 239
[8] University of Oxford, 'The GDPR and genomic data - the impact of the GDPR and DPA 2018 on genomic healthcare and research'(2020)< https://www.phgfoundation.org/media/123/download/gdpr-and-genomic-data-report.pdf?v=1&inline=1> accessed 29 August 2022.
[9] Charter of Fundamental Rights of the European Union (adopted 2 October 2000, entered into force 7 December 2000) OJ C 326/291 (EU Charter) article 8.
[10] Ibid, article 16(1)
[11] Ibid, art 4(1)

is considered an identifier, Article 9 introduces controversial conditions regarding the process of personal data. It includes explicit consent of the data subject, the processing purpose, and the Member States' authority to maintain or introduce other conditions or limitations on processing genetic and health-related data.

Generally, processing anonymous data which is unrelated to an identifiable natural person for statistical or research purposes falls outside the scope of the GDPR.[12] However, personal data can be processed for scientific research by considering appropriate conditions and safeguards at Union or Member State level.[13] When it comes to clinical trials, the situation is slightly different. While EU Clinical Trials Regulation ensures patient safety purposes, including reports, archives, and inspections, it does not cover scientific research purposes. As a result, the data controllers must take different legal bases into account according to the nature of the clinical trial under Article 9 of the GDPR.[14] Unlike the United States, GDPR is not limited to a specific sector and applies to a wide range of activities as long as they are not the member states' sole responsibility, e.g., national security.[15] However, this differentiation can create challenges regarding genomic healthcare and research.[16]

[12] Regulation (EU) 2016/679 of the European Parliament and of the Council of 27 April 2016 on the protection of natural persons with regard to the processing of personal data and on the free movement of such data, and repealing Directive 95/46/EC (General Data Protection Regulation) (GDPR) [2016] OJ L119/1 Recital 26
[13] Ibid Recital 157
[14] Kristof Van Quathem and Dan Cooper, 'European Commission Issues Updated Q&A on Interplay between the GDPR and the Clinical Trials Regulation' (*Inside Privacy*, 4 July 2022) <https://www.insideprivacy.com/international/european-union/european-commission-issues-updated-qa-on-interplay-between-the-gdpr-and-the-clinical-trials-regulation> accessed 20 August 2022
[15] Art 4 TEU
[16] University of Oxford (n 6) 19

1.2. Purpose and Research Questions

The primary purpose of this thesis is to analyze two critical rights involved in precision medicine; protecting data subjects' privacy and giving access to the data for research purposes. The aim is to understand the conflict between protecting patients' data in light of the privacy rules and providing a sufficient basis to conduct research in genomic science. It is essential to clarify to what extent scientists are allowed to research genomic data under the scope of the GDPR exemption.[17] The main research questions shall be described as:

(i) Does Art 9(2)(j) GDPR meet genomic research needs

(ii) Does Art 9(4) GDPR restrict cross-border genomic research within the EU

(iii) Is it necessary to reform the GDPR

1.3. Delimitations

There are four delimitations that must be clear. Firstly, it must be noted that the main research interest of the thesis is data protection and privacy in the European Union concerning the GDPR and its application in genomic research. Secondly, providing information about artificial intelligence, direct-to-consumer tests, consent, and anonymous data was necessary for a better understanding of the subject; however, the main focus is on the application of Article 9 GDPR and its impact on genomic research, cross-border processing, and data sharing. Thirdly, the information about national provisions was for comparative perspective, but the main scope of the subject is the European Union. Fourthly, this thesis aims to analyze genomic research in light of Article 9 GDPR; therefore, healthcare-related matters like medical and treatment aspects or liability matters like the processor's responsibility fall outside this thesis's scope.

[17] Kärt Pormeister, Genetic data and the research exemption: is the GDPR going too far? Int Data Priv Law [2017] 7(2) International Data Privacy Law <https://academic-oup-com.ludwig.lub.lu.se/idpl/article-abstract/7/2/137/3798545> accessed 27 August 2022

1.4. Method

This thesis mainly benefits the legal dogmatic method. In legal science, "Dogma" is described as people's attitudes facing the law.[18] On the one hand, this method represents the systemic nature of law and identifies the applicable law to the subject. On the other hand, it focuses on its effectiveness through interpreting established legal sources.[19] The legal methodology has a functional characteristic with an open system that allows development in different conditions to describe, classify, and generalize the definitions of various legal facts based on the system of law.[20] Although the legal doctrine is not mainly an explanatory discipline, explaining the reason and rationale behind legal concepts, rules, and principles is necessary to provide a reliable interpretation.[21] This is in accordance with the non-doctrinaire approach of this thesis. Since the arguments and discussions have a legal and social context, this thesis also emphasizes challenges and solutions rather than only a state-centric approach.[22]

Genomic research requires legal, ethical, and empirical perspectives. It is impossible to provide a comprehensive analysis without considering the legal, ethical, and scientific aspects. This thesis includes the legal aspects of genomic research in light of the existing legal system at both EU and national levels, accompanied by ethical aspects, moral boundaries, and scientific projects. The ethical perspective offers a better understanding of why genomic research must be ethical and respect moral principles, followed by a few examples of how destructive unethical research could be. The primary objective of this thesis is to analyze the subject from a legal perspective; the definitions and explanations of the scientific concepts and terms are provided for the reader's understanding.

[18] AlexanderV Petrov and AlexeyV Zyryanov, 'Formal-Dogmatic Approach in Legal Science in Present Conditions' [2018] Journal of Siberian Federal University 969
[19] Aleksander Peczenik, 'Juridikens Allmänna Läror' [2005] SvJT 249 < https://svjt.se/svjt/2005/249> accessed 27 August 2022
[20] AlexanderV Petrov and AlexeyV Zyryanov, (n 16) 970
[21] Van Hoecke, 'Methodologies of legal research: what kind of method for what kind of discipline?' (Hart Oxford, 2011) 8.
[22] Irina Domurath, 'The Politics of Interdisciplinarity in Law' in Marija Bartl and Jessica C Lawrence (eds), *The Politics of European Legal Research* (Edward Elgar Publishing 2022) 143

Legal research is not an empirical social science; it should set out the norms that apply in a particular legal system and share approaches with an interpretative social science perspective.[23] It is important to integrate recent social and legal issues into the existing legal system through interpretation.[24] With a normative view, the research should provide answers and solutions for the problems according to the rules and regulations.[25] Therefore, the focus here is not only on laws and regulations but also on functioning these rules in practice, analyzing the problem, and providing possible solutions according to the existing legal framework.

The main source of this thesis is EU law, as it is the primary source of legislation accompanied by relevant national rules of member states. At the EU level, the sources that have been used are the General Data Protection Regulation, the Treaties (TEU and TFEU), and the EU Charter of Fundamental Rights. At the national level, the sources are mainly used for comparative analysis in light of the EU law. The primary focus remains on the GDPR, especially Article 9. Other sources like guidelines, opinions from WP29, or information from the Commission's website have a complementary function, included for more clarification as secondary sources.

The comparative law approach in this thesis provides a better understanding of the issues from different legal system perspectives. Comparative law research is a more general form of legal research that helps to find the similarities and differences of several legal systems to compare the same concept from different legal perspectives.[26] For the purpose of this thesis, the data protection rules concerning genetic data in Portugal, Sweden, and Ireland have been discussed in detail regarding the application of the GDPR in those legal systems.

[23] John Bell, 'Legal Research and the Distinctiveness of Comparative Law' in Mark Van Hoecke (ed), *Methodologies of Legal Research: What Kind of Method for What Kind of Discipline?* (Hart Publishing 2011) 175

[24] Jan Bm Vranken, 'Methodology of Legal Doctrinal Research: A Comment on Westerman' in Mark Van Hoecke (ed), *Methodologies of Legal Research: What Kind of Method for What Kind of Discipline?* (Hart Publishing 2011) 112

[25] Jaap Hage, 'The Method of a Truly Normative Legal Science' in Mark Van Hoecke (ed), *Methodologies of Legal Research: What Kind of Method for What Kind of Discipline?* (Hart Publishing 2011) 27

[26] John Bell (n 21)175

Furthermore, many authors, researchers, and scholars from different disciplines provided informative comments, opinions, and analyses through documents, books, articles, and reports. These additional sources were beneficial in elaborating on the subject from different points of view.

1.5. Outline

This thesis is divided into six chapters. The first chapter includes information about the topic and the subject's background, followed by the purpose of the topic, the research questions, delimitations, and the method. The second chapter introduces precision medicine and AI, their role and application in healthcare, relevant public and private research projects at national and international levels, and direct-to-consumer tests. The third chapter is about the legal and ethical perspectives, including privacy and data protection rights, the objectives of the GDPR, and its scope. Chapter four describes the genetic data that is categorized as special data. Then it explains Article 9 GDPR, followed by privacy challenges, consent, and genomic research at the EU and national level. Chapter five is the central chapter of the thesis; it explains the difference between genomic medicine, precision medicine, and personalized medicine. Then it describes anonymous data, evaluates protecting research or personal privacy, as well as the efficiency and sufficiency of the GDPR. Finally, chapter six summarizes the discussion from previous chapters and ends with the conclusion.

2. The role of Precision Medicine & AI in Healthcare

2.1. AI and Healthcare

In the European Union, there is no legally binding definition for artificial intelligence.[27] However, The Commission stated that the definition of AI "will need to be sufficiently flexible to accommodate technical progress while being precise enough to provide the necessary legal certainty."[28] Maybe an acceptable definition would be "the systems that display intelligent behavior by analyzing their environment and taking actions – with some degree of autonomy – to achieve specific goals."[29] The meaning of 'intelligent' has been described as "the ability to acquire and apply knowledge and skills and to manipulate one's environment."[30]

Nowadays, using AI agents in providing medical care has increased diagnostic capabilities; the medical AI agent is an entity that can process data from patients, self-report the features, and communicate the finding and provide medical feedback to the patients.[31] Although AI agents are not likely to replace doctors and nurses completely, they transform the healthcare sector and improve outcomes.[32]

The digitization of health-related data is accelerating the development of AI applications within healthcare.[33] Some researchers have introduced three principles to successfully adopting AI in healthcare: data and security, analytics and insights, and

[27] Jenny Gesley, 'Legal and Ethical Framework for AI in Europe: Summary of Remarks' [2020] 114 Proceedings of the ASIL Annual Meeting 240 < https://www.cambridge.org/core/journals/proceedings-of-the-asil-annual-meeting/article/abs/legal-and-ethical-framework-for-ai-in-europe-summary-of-remarks/9FFEF2BE5DE62BFA5A974A7E565473B3> accessed 27 August 2022

[28] European Commission, 'White Paper on Artificial Intelligence - A European Approach to Excellence and Trust' (2020)16 <https://ec.europa.eu/info/publications/white-paper-artificial-intelligence-european-approach-excellence-and-trust_en>

[29] Eleanor Bird and others, 'The ethics of artificial intelligence: Issues and initiatives' ((EPRS), European Parliamentary Research Service 2020)

[30] Ibid

[31] Rao Kotesh and others, 'A Medical AI Agent as a Tool for Neuropsychiatric Diagnoses'[2021] 23rd International Symposium on Measurement and Control in Robotics (ISMCR) < https://ieeexplore.ieee.org/document/9263713> accessed 27 August 2022

[32] Arjun Panesar, *Machine Learning and AI for Healthcare* (2nd edn, Apress 2021) 10

[33] Kevin B. Johnson and others, 'Precision Medicine, AI, and the Future of Personalized Health Care' [2021] 14(1) Clinical and Translational Science 86, 87 accessed 27 < https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7877825> accessed August 2022

shared expertise.[34] To explain the first principle, data and security result in transparency and trust, meaning that AI and humans work together, so it is vital to trust the final result. Data security is essential because a large amount of information about people's health is linked to their lifestyle and environment. About 60% of the health data is related to behavioral, socio-economical, physiological, and psychological information, 30% for the gens, and the last 10 % for actual medical records.[35]

AI and big data are becoming more widely used in healthcare involving five main parties; payer, provider, policy maker/government, patients, and product manufacturers.[36] A secure system can prevent fraud, waste, and abuse in payer programs.[37] AI is also used to detect risks and predict patients who are at risk for readmission.[38] On a larger scale, governments and healthcare organizations hire AI to control or predict infections and outbreaks,[39] or more extensively in global pandemic situations like COVID-19, where AI helped clinicians target people at risk and provide more information for their treatment.[40]

AI technologies are used in various medical forms, but the primary focus of this thesis is diagnostic systems.[41] AI allows recognizing complex patterns and structures in diagnostic techniques in healthcare to perform at the same level or sometimes better than clinicians.[42] As a result, AI has several capabilities, such as reducing diagnostic errors, augmenting intelligence to support decision-making, and helping clinicians with administrative tasks.[43] Although AI has many applications in different sectors, precision

[34] Ibid 86
[35] Ibid
[36] Ibid
[37] Hossein Joudaki and others, 'Improving fraud and abuse detection in general physician claims: a data mining study' [2016] 5(3) International Journal of Health Policy and Management (IJHPM) 165 < https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4770922> accessed 27 August 2022
[38] Alvin Rajkomar and others, 'Scalable and accurate deep learning with electronic health records' [2018] 1(18) NPJ Digital Medicine 1 < https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6550175/> accessed 27 August 2022
[39] Adam Sadilek and others, 'Machine-learned epidemiology: real-time detection of foodborne illness at scale' [2018] 1(36) NPJ Digital Medicine <https://www.nature.com/articles/s41746-018-0045-1#citeas> accessed 27 August 2022
[40] Raju Vaishya and others, 'Artificial intelligence (AI) applications for COVID-19 pandemic' [2020] 14(4) Diabetes & Metabolic Syndrome: Clinical Research & Reviews < https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7195043> accessed 27 August 2022
[41] Eric Topol, '*Deep Medicine: How Artificial Intelligence Can Make Healthcare Human Again*' (Basic Books, 2019)
[42] Ibid
[43] Thomas H. Davenport and others, 'Using AI to improve electronic health records' [2018] Harvard Business Review< https://hbr.org/2018/12/using-ai-to-improve-electronic-health-records> accessed August 2022

medicine has a stronger influence on the healthcare direction[44] because it needs access to massive amounts of data and AI to personalize care for every individual and facilitate the prospective cases for healthcare providers in the future respectively.[45]

2.2. Application of Precision Medicine in Healthcare

One of the best definitions for Precision medicine would be "a new taxonomy of human disease based on molecular biology."[46] In other words, precision medicine is a new healthcare method based on human genome sequencing knowledge[47] which also takes other factors such as medical records, lifestyle, and the environment of patients into account.[48] Healthcare providers use precision medicine to gain understanding by linking genetic information to health and disease outcomes.[49] Through this mechanism, the medical decision connects the average patient's evidence to others based on their characteristics.[50]

Precision medicine functions in both predictive and preventive way by stopping diseases before it even starts, and it considers individual variations and tailors the treatment for each patient.[51] As a result, clinicians can provide personalized care to individuals and reveal possible diseases that usually remain hidden.[52] Precision medicine has a wide range of applications ranging from early detection of disease[53] to

[44] Samuel J Aronson and Heidi L Rehm, 'Building the foundation for genomics in precision medicine' [2015] Nature < https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5669797/> accessed 27 August 2022
[45] Kevin B. Johnson and others (n 31) 86
[46] National Research Council, '*Toward Precision Medicine: Building a Knowledge Network for Biomedical Research and a New Taxonomy of Disease*' (The National Academies Press, 2011)
[47] Ibid
[48] Roy C. Ziegelstein 'Personomics and precision medicine' [2017] 128(1) Transactions of the American Clinical and Climatological Association 160 < https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5525386/> accessed 25 August 2022, also see Annex 1
[49] University of Oxford (n 6) 9
[50] Kevin B. Johnson and others (n 31) 89
[51] Yudong Cai and Tao Huang, 'Accelerating precision medicine through genetic and genomic big data analysis' [2018] 1864(6), Biochimica et Biophysica Acta (BBA) - Molecular Basis of Disease< https://pubmed.ncbi.nlm.nih.gov/29548968/> accessed 25 August 2022
[52] Kevin B. Johnson and others (n 31) 89
[53] Marc van der Schee and others, 'Breath biopsy for early detection and precision medicine in cancer' [2018] 12(1) Ecancermedicalscience <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6070367> accessed 25 August 2022

personalized treatments[54] and genotype-guided treatment.[55] By way of explanation, precision medicine increases healthcare efficiency through precise diagnoses, predicting health risks before any symptoms, and providing customized treatment for each patient.[56] Different terms are used interchangeably regarding genetic data, such as genomic medicine, precision medicine, and personalized medicine. For the remainder of this paper, we will use the term precision medicine for the use of genomic variant data in treating patients, the term genomic research to reflect the projects that use genetic data for research, and the term genomic medicine for using patients' genotypic information in their clinical care.

2.3. Research Projects on Precision Medicine at Global Level

Several countries like the UK, USA, and France launched their national genomic medicine projects[57] in 2012, 2014, and 2015 respectively. The objective of these projects and data repositories, such as PMI in the United States[58], UK Biobank[59], BioBank Japan[60], and the Australian Genomics Health Alliance[61], is to establish precision medicine in the medical sector and develop a national framework.[62] However, human genome projects have been originally a global initiative. The primary steps were

[54] Ryan J Hartmaier and others, 'High-Throughput Genomic Profiling of Adult Solid Tumors Reveals Novel Insights into Cancer Pathogenesis' [2017] 77(9) Cancer Res. 2464< https://pubmed.ncbi.nlm.nih.gov/28235761> accessed 25 August 2022

[55] Andrea L. Jorgensen and others, 'Implementation of genotype-guided dosing of warfarin with point-of-care genetic testing in three UK clinics: a matched cohort study' [2019] 17(76) BMC Medicine. < https://bmcmedicine.biomedcentral.com/articles/10.1186/s12916-019-1308-7> accessed 27 August 2022

[56] Kevin B. Johnson and others (n 31) 88

[57] Catherine Lejeune and others, 'The Economic, Medical and Psychosocial Consequences of Whole Genome Sequencing for the Genetic Diagnosis of Patients With Intellectual Disability: The DEFIDIAG Study Protocol' [2022] Frontiers in genetics< https://europepmc.org/article/med/35444683> accessed 28 August 2022

[58] Pamela L Sankar and Lisa S Parker 'The Precision Medicine Initiative's All of Us Research Program: an agenda for research on its ethical, legal, and social issues' [2017] 19(7) Genetics in medicine: official journal of the American College of Medical Genetics, 19(7) <https://www.gimjournal.org/article/S1098-3600(21)02228-0/fulltext> accessed 28 August 2022

[59] Clare Bycroft and others, 'The UK Biobank resource with deep phenotyping and genomic data' [2018] Nature 203 < https://www.nature.com/articles/s41586-018-0579-z> accessed 28 August 2022

[60] Akiko Nagaiand and others, 'Overview of the BioBank Japan Project: study design and profile' [2017] Journal of Epidemiology 27(3S)< https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5350590/> accessed 29 August 2022

[61] Zornitza Stark and others, 'Australian genomics: a federated model for integrating genomics into healthcare' [2019] 105(1) American Journal of Human Genetics<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6612707/> accessed 29 August 2022

[62] Plan France Médecine Génomique 2025 (Aviesan, 2025; PFMG 2025)< https://pfmg2025.aviesan.fr/en/> accessed 29 August 2022

based on scientist-to-scientist interactions at an international level rather than a political action due to the lengthy process and lack of efficiency.[63] Twenty centers in six countries started cooperating on human genome sequencing: China, France, Germany, Great Britain, Japan, and the United States. Through this project, every center had the opportunity to visit other's genome centers and share their technical and experimental advances.[64] Another project in 2011 was called "the ORIENTplus;" even though non-EU member states were also participating, the project was funded by the European Commission and ran for 42 months to connect research and education communities in Europe and China.[65] The ORIENTplus had several outcomes, such as rapid advances in the medical sciences through decoding the human genome and improving real-time knowledge transfer and e-learning tools.[66] Many countries had started generating national genomic data projects in the EU by 2018. Since it is necessary to have systematic access and integration of research and healthcare data on a larger scale,[67] the Commission created a Europe-wide project called "1+ million genome groups by 2022".[68] Twenty-four countries have signed the declaration (22 EU member states, the UK, and Norway).[69] Although these collaborations and research cooperations are valuable, they also include data sharing and security challenges which will be discussed in the following chapters.

2.4. DTC genetic testing

Genetic testing is not always for research or treatment. Direct-to-consumer tests are genetic tests that are for commercial purposes. Through these tests, consumers order tests and send their biological samples to the company. These companies are located in

[63] Francis S Collins and others, 'The Human Genome Project: Lessons from Large-Scale Biology' [2003] Science < https://pubmed.ncbi.nlm.nih.gov/12690187/> accessed 29 August 2022
[64] Ibid
[65] European Commission, 'Linking European and Chinese Research Infrastructures through the ORIENTplus project' [2015] <https://digital-strategy.ec.europa.eu/en/news/linking-european-and-chinese-research-infrastructures-through-orientplus-project> accessed 29 August 2022
[66] Ibid
[67] Gary Saunders, 'Leveraging European infrastructures to access 1 million human genomes by 2022' [2019] Nature reviews. Genetics < https://www-nature-com.ludwig.lub.lu.se/articles/s41576-019-0178-3> accessed 29 August 2022
[68] See Annex 2
[69] European Commission, 'The Roadmap of the 1+Million Genomes Initiative is now clearly illustrated in a new brochure'(European Commission, 28 September 2020) < https://digital-strategy.ec.europa.eu/en/library/roadmap-1million-genomes-initiative-now-clearly-illustrated-new-brochure>

different countries but provide services across the world. As a result, consumers' genetic data and other personal information are often processed, stored, and shared in other countries.[70]

Generally, there are nine main types of DTC GT introduced: ancestry tests, relatedness tests, nutrigenetic tests, athletic ability and talent tests, prenatal tests, diagnostic tests, personalized medicine, and Carrier Testing.[71] Each type of test has different ethical and public policy issues that should be considered in different researches.[72]

3. Legal and Ethical Perspectives

3.1. Introduction

Human rights have a biological dimension to protect each person's life and dignity regarding their living body. The question here is to what extent biological data should be protected under the scope of human rights and personal data. In this regard, we should not forget the impact of society and culture on defining the level of data protection. Although the right to protect one's biological data as a part of their identity is essential, this protection does not entitle the person to own or have an exact knowledge of their genetic data. In other words, human rights genomic data is a part of one's identity and should be protected under the CFR and the ECHR; however, this protection is only limited to the knowledge of their existent life.[73] In recent years, genetics and genomics have become more diverse, especially for commercial purposes. The complexity of new genetic discoveries brought up numerous ethical concerns.[74]

The principle of "Ethics of Knowledge" was introduced by Jacques Monod in his book "Chance and Necessity." This principle indicates the commitment to the scientific

[70] Alexander Nill and Gene Laczniak, 'Direct-to-Consumer Genetic Testing and Its Marketing: Emergent Ethical and Public Policy Implications' [2020] 175(4) Journal of Business Ethics 674
[71] Ibid 671-673
[72] For more information read: Paula Boddington, *Ethical challenges in genomics research a guide to understanding ethics in context* (Springer, 2012)
[73] Jill Marshall, *Human Rights Law and Personal Identity* (1st edn, Routledge 2014) 141
[74] Leila Jamal (n 5) 233

exploration of natural phenomena. The formulation introduces an intellectual framework that denies the existence of absolute or ultimate values; in other words, values are chosen rather than given, whether consciously or unconsciously. This way, ethics of knowledge promotes the idea of having an intelligent approach toward values. This principle caused various misunderstandings. Some interpreted it as an absolute set of values. Others claimed that it is about prioritizing scientific knowledge over other types of human activities that also serve as sources of value.

The idea of an ethics of knowledge has been criticized as a manifestation of scientific elitism. More dangerously, the ethics of knowledge is often embraced by some who interpret it as the right to pursue the quest for knowledge whenever one wishes, irrespective of consequences.[75] Salvadore E. Luria explains how difficult it is to balance the ethics of knowledge (the right to know) and the ethics of innocence (anything that harms other people). On the one hand, it is necessary to increase knowledge in different areas; on the other hand, research might harm other human beings, and the ethics of innocence requires us to refrain from that research.[76]

Although research and experiments benefit society, they should respect ethics and human rights. There were several numbers of research that were not ethical and still provided significant results and extensive knowledge. For example, there were a series of human experiments in Lund, Sweden, known as the Vipeholm experiments, where patients of a hospital who were intellectually disabled were subjected to unethical research to determine whether carbohydrates affected the formation of cavities. Even though the result of this experiment was significantly helpful, it violated medical ethics and patients' rights.[77] In another case, a thoracic surgeon and regenerative medicine researcher at Sweden's Karolinska Institutet (KI) was guilty of misconduct.[78] He performed unethically experimental surgeries on patients, which led to their death.

[75] Salvadore E. Luria (n 2) 407

[76] The ethics of knowledge and ethics of innocence are two complementary aspects of morality. The development of the human brain wants us to know more and transit knowledge. The power to identify ourselves and our feelings and sufferings ask us to be innocent. We cannot legitimately follow either one and ignore the other.

[77] Elin Bommenel, 'Sockerförsöket : kariesexperimenten 1943-1960 på Vipeholms sjukhus för sinnesslöa' (Arkiv förlag, 2020) < https://portal.research.lu.se/sv/publications/the-suger-experiments> accessed 28 August 2022

[78] 'The final verdict on Paolo Macchiarini: guilty of misconduct' [2018] Lancet <https://pubmed.ncbi.nlm.nih.gov/30047386> accessed 28 August 2022

The ethical aspect of research is vital, and "To preserve the values that we cherish, in science as in medicine, we must be careful never to become, consciously or unwittingly, the instrument of forces that would subordinate those values for selfish or misguided goals of their own. Ethics of knowledge and ethics of innocence must never become dissociated, lest either one without the other becomes a vehicle for ignorance or a tool of oppression."[79] Therefore, it is imperative to consider ethics before conducting any research or experiments, regardless of how beneficial or significant the results might be. Ensuring human life and fundamental rights should be considered before any research or experimentation that might negatively affect patients' life and privacy.

3.2. Privacy and Data Protection Rights

Nowadays, protecting the data and keeping individuals' information is vital in any sector. It has been claimed that up to 10% of worldwide health care expenditure is caused by fraud and abuse, and AI-based tools can worsen the situation.[80] Any negligence might turn into ruining people's lives, whether it turns into revealing patients' identities or, in severe cases, hacking patient databases to blackmail them.[81] Privacy protection is not a new concept and has been a fundamental right for many years.[82] Although New technologies and digital life provide great opportunities, they bring new demands and responsibilities in their train. In this regard, the EU is one of the pioneers in recognizing personal data protection as a fundamental right.

The EU Charter is a binding source of primary law that provides citizens and residents with a wide range of fundamental rights. Article 7 gives every individual the right to respect their private and family life, home, and communications. Then, Article 8 provides every individual with the right to protect personal data, followed by indicating conditions such as data must be processed fairly, for specified purposes, and

[79] Salvadore E. Luria (n 2) 407
[80] Hossein Joudaki and others (n 35) 165
[81] Vastaamo data breach in Finland, [2020] https://en.wikipedia.org/wiki/Vastaamo_data_breach accessed 28 August 2022
[82] Article 12 Universal Declaration of Human Rights, proclaimed by the United Nations General Assembly in Paris on 10 December 1948 (General Assembly resolution 217 A)

based on the individual's consent or by law. Similar to Article 8 CFR, Article 16(1) TFEU also gives everyone the right to protect personal data concerning them. However, the basis for legislating a data protection regulation is within the meaning of the second paragraph of Article 16 TFEU.

According to Article 16(2) TFEU, the European Parliament and the Council are competent bodies to lay down the rules governing the protection of individuals concerning the process of personal data. Processing the personal data by Union institutions, bodies, offices, and agencies, as well as by the Member States, when their actions fall under the scope of EU law, and the rules relating to the free movement of those data are subject to this rule. The Data Protection Directive[83] was the first act to regulate EU personal data protection in 1995. It was repealed by Regulation (EU) 2016/679, known as the GDPR, which was adopted in 2016 but became enforceable in 2018.

3.3. The GDPR

The GDPR might not be the first legislation that regulates personal data protection; however, it is the first thorough reform of the EU personal data protection regime since Directive 95/46/EC was adopted. The importance of the GDPR is not limited to superseding the Directive because it is a progressive document. The GDPR, as a primary source of law, provides privacy protection not only for processing personal data but also for the free movement of data within the EU.

3.3.1. The Objectives and Scope of the GDPR

Two main objectives can be understood from Article 1 GDPR:

- Protecting fundamental rights and freedoms of natural persons and, in particular, their right to the protection of personal data.

[83] Parliament and Council Directive 95/46/EC of 24 October 1995 on the protection of individuals with regard to the processing of personal data and on the free movement of such data [1995] OJ L 281/31

- The free movement of personal data within the Union shall be neither restricted nor prohibited for reasons connected with the protection of natural persons with regard to the processing of personal data.

To understand the scope of GDPR, it is necessary to read Article 1 GDPR in conjunction with Article 4(1) GDPR and recital 14. Personal data does not cover information about legal persons; only information about natural persons is subjected to this concept regardless of their nationality or place of residence. A unified data protection standard is established alongside the free movement of personal data within the Union.[84] According to Article 2(1) GDPR, if personal data is being processed wholly or partly by automated means or which is part of a filing system or is intended to form part of a filing system, then it goes under the scope of the GDPR. It is essential to know what activities are considered 'processing' in order to conclude whether GDPR applies. Article 4(2) GDPR defines processing as any operation performed on personal data, whether or not by automated means. Since precision medicine involves various factors ranging from genetic data to location, or personal interests, it falls under the definition of 'profiling.' Article 4(4) GDPR defines profiling as "any form of automated processing of personal data consisting of the use of personal data to evaluate certain personal aspects relating to a natural person, in particular, to analyze or predict aspects concerning …, health, personal preferences, interests,…, location or movements."

In accordance with the general requirements, Article 5(1) GDPR stipulates the qualities for processing personal data. Article 5(1)(a) GDPR requires the processing to be lawful, fair, and transparent. To elaborate on this part, processing personal data should be in accordance with the law, respect the legitimate interests of the data subjects, and be transparent to them.[85] Article 5(1) GDPR also limits the personal data to be collected and processed in a way that is enough for the initial purpose and does not go further than necessary. Article 5(2) GDPR puts the responsibility on the controller to ensure the lawfulness of data processing and, in particular, its compliance

[84] Mariusz Krzysztofek, *GDPR: Personal Data Protection in the European Union* (Kluwer Law International 2021) 14
[85] Ibid 59

with the principles of data protection and data quality.[86] It could be understood from Article 5(1) GDPR that it has a primary connection to the ethical concerns as well as the consent of the data subjects that will be discussed later.

Moreover, it is necessary to mention that the GDPR has categorized genetic data under the special categories of personal data[87], which is different from personal data introduced in Article 4(1) GDPR. In other words, genetic data require more protection due to the unique characteristics of processing since there is a higher risk to the patient's privacy rights.[88] While the legal basis for processing ordinary data is Article 6 GDPR, Article 9 GDPR is the basis for special data categories (including genetic data). The processing of personal data in Article 6 GDPR is permissive, meaning that personal data can be processed as long as it meets at least one of the conditions in Article 6(1) GDPR. By contrast, processing special personal data under Article 9 GDPR is restrictive unless it meets one of the conditions in Article 9(2)[89] that we will discuss more deeply in the next chapter.

4. GENOMIC Medicine and the GDPR

4.1. GDPR AND SPECIAL CATEGORIES OF PERSONAL DATA

As a general rule for special data, Article 9(1) GDPR stipulates:

> '[…] Processing of *genetic data*, biometric data for the purpose of uniquely identifying a natural person, *data concerning health* or data concerning a natural person's sex life or sexual orientation shall be prohibited.'

Article 9(2) GDPR provides several exceptions to the rule, but some of them are more related to our subject. According to Article 9(2)(a), it is possible to process the

[86] Ibid 60
[87] Recital 35 and Article 9(1) GDPR
[88] Recital 51 GDPR
[89] Mariusz Krzysztofek (n 80) 77

data if data subjects give explicit consent for one or more specified purposes as long as the EU or Member State law does not allow such an exception. Article 9(2)(g) makes data processing possible if there is substantial public interest; Still, it should be proportionate and provide suitable and specific measures to safeguard the data subject's fundamental rights and interests. Article 9(2)(h) allows the processing of the data if it is necessary for preventive or occupational medicine, medical diagnosis, the provision of health or social care or treatment, or the management of health. Article 9(2)(i) allows processing based on the necessity of public interest in public health, such as protecting against serious cross-border threats to health or ensuring high standards of quality and safety of health care and medicinal products or medical devices based on Union or Member State law which provides for suitable and specific measures to safeguard the rights and freedoms of the data subject, in particular, professional secrecy. Last but not least, Article 9(2)(j) mentions that data processing is permitted where it is necessary for archiving purposes in the public interest, scientific or historical research purposes. However, it should be proportionate and provide suitable and specific measures to safeguard the data subject's fundamental rights and interests.

4.2. Privacy Challenges in GENOMIC RESEARCH

Unlike the US, which has a sectoral approach for different subjects, some researchers say that the GDPR has a broad policy, which applies as a general law to a wide range of processing operations within public and private organizations.[90] There are several bases for processing personal data under Article 9(2), but it is still not without challenges. While Article 9(2)(a) allows data processing after the data subject has given explicit consent, Article 9(2)(j) GDPR indicates that personal data, including sensitive data, could be processed for scientific research under the conditions in Article

[90] Christopher F Mondschein and Cosimo Monda, 'The EU's General Data Protection Regulation (GDPR) in a Research Context' in Kubben and others (eds), *Fundamentals of Clinical Data Science* (Springer, 2018) 57

89(1). The details of how these rules can affect research will be discussed in the subsections.

4.2.1. The Scope of Consent

'Consent' is any freely given, specific, informed, and unambiguous indication of the data subject's wishes through a statement or a clear affirmative action that signifies agreement to the processing of personal data relating to them.[91] It is essential to know that informed consent is not a right per se, but it is required for the lawfulness of processing and as one of the adequate safeguards under Article 89(1).[92] The data subject's consent is required by Article 6(1)(a) GDPR, and Article 7 GDPR introduces the conditions for consent. The second condition in Article 7 GDPR is that personal data concerning a unique person who is distinguishable from others can also apply to genetic data since the GDPR does not govern biological or familial genetic information unless it can be related to one specific individual from a group.[93] The data subjects keep the right to withdraw their consent at any time; however, the data processing performed before the withdrawal remains lawful.[94] Finally, it is essential to know that if the controllers decide to go with consent as a legal basis for any part of the processing, they cannot switch from consent to another ground mentioned in Article 6 GDPR. In other words, the controller must decide the legal basis in advance, and they cannot change the legal basis to justify the processing once they face problems with the validity of the consent.[95]

The GDPR indeed recognizes the possibility for broad consent to cover unspecified research projects in the future.[96] Still, the broad definition of data processing does not mean that consent to processing gives another controller access to the data or

[91] Article 4(11) GDPR
[92] Santa Slokenberga and others, *GDPR and Biobanking* (1st edn, Springer International 2021) 23
[93] University of Oxford (n 6) 60
[94] Article 7(3) of the GDPR
[95] Article 29 Data Protection Working Part (WP29), *Guidelines on consent under Regulation 2016/679*, [2017] (WP259 rev.01) Page 23
[96] Recital 33 GDPR

permission to send data to third countries.[97] If consent is the only legal basis for processing different and unrelated purposes, it should not be put together in one clause. In other words, the data subject must be able to distinguish the necessary purposes from those that merely serve the controller's interest. Therefore, different aims of data processing demand separate consents.[98]

From the data subjects' perspective, it is vital to ensure that first, patients understand the difference between the clinical and research use of data, and then they have suitable options for refusing secondary use of their data.[99] Moreover, when special categories of data are processed, they require explicit consent, and applying the flexible approach of Recital 33 faces a strict interpretation that requires higher scrutiny.[100]

One of the significant challenges regarding consent is the concept of 'Imbalance of power.' According to Recital 43, consent cannot be considered a valid legal basis for processing personal data in specific cases if there is a clear imbalance between the data subject and the controller. In other words, the controller can benefit from the consent as a legal basis for processing personal data only if there is a balance between the controller and the data subject. Any imbalance between the controller and the data subject undermines the validity of consent; the validity of consent has cumulative criteria under Article 4 (11), Article 7, and recitals 32, 33, 42, and 43 of GDPR.[101] Without a shred of doubt, achieving these requirements is complex in health research projects[102], mainly when the controller is a public authority, and it is unlikely that consent was freely given.[103] Still, public authorities are not entirely excluded under the legal framework of the GDPR, and under certain circumstances, it is possible to use consent.[104] The main concern about the imbalance is more directed to the consent to be

[97] Mariusz Krzysztofek (n 80) 78
[98] Ibid 79
[99] Emilia Niemiec, 'Consenting Patients to Genome Sequencing' in Aad Tibben and Barbara B biesecker (eds), *Clinical Genome Sequencing* (ScienceDirect 2019) 51
[100] Santa Slokenberga and others (n 88) 78
[101] Mary Kirwan, 'What GDPR and the Health Research Regulations (HRRs) mean for Ireland: "explicit consent" a legal analysis' [2020] Springer < https://link.springer.com/article/10.1007/s11845-020-02331-2> accessed 18 August 2022
[102] Ibid 518
[103] Recital 43 GDPR
[104] Article 29 Data Protection Working Part (WP29), *Guidelines on consent under Regulation 2016/679*, [2017] (WP259 rev.01) Page 6

'freely given' rather than 'who' is the controller or the data subject because. In other words, an imbalance of power can also occur in the employment context or any other relationship than can affect the data subject's freely given consent.[105]

The difference between consent and explicit consent is not very clear, and there is no definition for 'Explicit consent' provided by the GDPR. The term explicit is related to how the GDPR consent is expressed by the data subject and raises the consent standard where there is a severe data protection risk.[106] In this regard, there might be confusion about whether explicit consent is also necessary for conducting research. This might come from the difference between consent as a research ethics principle and consent as a lawful basis in data protection law.[107] To answer this question correctly, it should be divided into three parts:

First, it is important to know the aim of processing the special data. If the special data is to be processed for a specified purpose, explicit consent is necessary, and Article 9(2)(a) is applicable. If the data is to be processed for the public interest, scientific or historical research purposes, then Article 9(2)(j) GDPR is applicable but in accordance with the appropriate safeguards under Article 89(1) GDPR.

Second, it should be examined whether there are any Union or Member State laws provide that the prohibition referred to in paragraph 1 may not be lifted by the data subject.[108] Even though Article 9(2)(a) allows processing the special data via explicit consent, the last part ceases the exception if any EU or national rules restrict the data subject from giving consent.

Third, in light of Article 9(4) GDPR, the Member States can maintain or introduce other conditions, including limitations, concerning the processing of genetic data, biometric data, or data concerning health. Although explicit consent is a lawful basis

[105] See also Article 88 GDPR, where the need for protection of the specific interests of employees is emphasized, and a possibility for derogations in Member State law is created, see also Recital 155 GDPR
[106] Mary Kirwan (n 97) 516
[107] Edward S Dove and Jiahong Chen, 'Should consent for data processing be privileged in health research? A comparative legal analysis' [2020] 10(2) International Data Privacy Law 118 < https://academic.oup.com/idpl/article/10/2/117/5757981> accessed 18 August 2022
[108] Article 9(2)(a) GDPR

for processing under Article 9 GDPR and is not a mandatory requirement,[109] it must be considered if processing the data is subjected to additional national conditions or limitations. Therefore, before processing the data, it is necessary to check whether the Member State has introduced any further requirements.[110]

Another division of consent is informed consent and broad Consent. Although informed consent and broad consent are introduced as opposites, they might be neither opposites nor points on a continuum or spectrum. They solely reference different matters within consent. Informed consent concerns the quality of the consent, whereas broad consent concerns the subject matter of the consent. [111]

To summarize, researchers can benefit from the exemption if the public interest in carrying out the research significantly outweighs the public interest in requiring explicit consent.[112] Some researchers think that under these requirements, many health research projects could be considered disproportionate, which can negatively affect health research, research competitiveness, and patient access to new medical discoveries.[113] However, this also varies from one Member State to another, which will be briefly discussed in the next chapter.

4.2.2. GENOMIC Research UNDER THE EU LAW

Article 13 CFR and Article 35 CFR ensure the freedom of scientific research and a high level of human health protection, respectively; still, GDPR established a research regime, which to some degree can be seen as research harmonization through the back door.[114] Although the GDPR did not define scientific research, Article 29 Working Party has indicated that 'the notion may not be stretched beyond its common meaning and understands that "scientific research" in this context means a research project set

[109] Mary Kirwan (n 97) 516
[110] Article 9(4) GDPR
[111] Carla Barbosa and Andreia da Costa Andrade, 'Biobanks and GDPR: A Look at the Portuguese Panorama' in Santa Slokenberga and others (ed), *GDPR and Biobanking* (Springer 2021) 359
[112] Santa Slokenberga and others (n 88) 406
[113] Edward S Dove and Jiahong Chen (n 103)
[114] Ibid 14

up in accordance with relevant sector-related methodological and ethical standards, in conformity with good practice.'[115]

Some researchers claim that Article 9(4) GDPR gives enough space to the Member States to restrict the processing of genetic data, which has the potential to impact the research path significantly.[116] Even though European countries were initially entitled to regulate biomedical research, nowadays, such activities have legal and ethical aspects involving data protection and participant privacy rights. These rights are classified as human rights under Article 8 CFR and Article 16 TFEU; as a result, the EU has legislative competence.[117]

4.2.3. NATIONAL DATA PROTECTION RULES AND GENOMIC RESEARCH

From a comparative perspective, EU Member States have taken different approaches. In Portugal, various legislations regulate the establishment and functioning of biobanks which imposes specific protection duties on scientific research activities.[118] The informed consent should be in writing and not an oral statement. Also, it is necessary to obtain two consents: the first consent is in order to get the biological sample, and the second one is to include that sample in the biobank.[119] An interesting fact in Portugal's regulation is that if someone gives consent that their biological sample is incorporated into a biobank, not only they still have can exercise their privacy rights but also if they die, this right is transferred to their relatives and family members.[120]

Even though the GDPR clearly stated that it does not apply to the personal data of deceased persons,[121] Recital 27 entitles the Member States to introduce rules regarding processing the personal data of deceased persons. Additionally, there are biobanks in

[115] Article 29 Data Protection Working Part (WP29), *Guidelines on consent under Regulation 2016/679*, [2017] (WP259 rev.01) 27–28
[116] Christopher F Mondschein and Cosimo Monda (n 86) 59
[117] Santa Slokenberga and others (n 88) 13
[118] Carla Barbosa and Andreia da Costa Andrade, (n 107) 345
[119] Ibid 348
[120] Ibid 349
[121] Recital 27, 158, and 160 GDPR

private and public institutions for research purposes, and the legislation prohibits the latter from having identified samples.[122] Portugal has been favoring public biobanks, and it is expected to continue supporting public biobanks and increase them, not in terms of number but their size.[123]

In Sweden, the legislator has a minimalistic approach concerning the research exception in Article 89 GDPR with limited general exceptions to the data protection rules.[124] The implementation of GDPR in Sweden has a general exception from the data protection rules concerning the right to access official documents, which also covers researchers.[125] Some researchers find Sweden's regulatory regime 'permissible but complex' due to data protection and secrecy rules. The reason was explained as, on the one hand, the rules are permissive by giving researchers broad access to registries but, on the other hand, a bit ambiguous and complex.[126] In light of the transparency tradition and the principle of public access, which are a part of the national constitutional identity of Sweden, researchers have broad access to publicly-held health data held by Swedish authorities.[127] According to Recital 154 GDPR, Sweden allows personal data in official documents to be disclosed to 'reconcile public access to official documents with the right to the protection of personal data.' As a result, the GDPR and the Swedish Data Protection Act will not apply in a way that conflicts with the Freedom of the Press Act or the Freedom of Expression Act.[128] Still, personal data remain secret if there is a risk that after the document has been released, it will be processed in conflict with the GDPR, the Data Protection Act, or the Ethical Review Act.[129]

In Sweden, Etikprövningsmyndigheten has the authority to examine applications for ethics review if the research involves processing special categories of personal data or human biological material.[130] In other words, obtaining ethical approval before any

[122] Ibid 360
[123] Ibid 361
[124] Magnus Stenbeck and others, Swedish Law on Personal Data in Biobank Research: Permissible But Complex. in Santa Slokenberga and others (ed), *GDPR and Biobanking* (Springer 2021) 379
[125] Ibid
[126] Ibid 394
[127] Ibid 383
[128] Chapter 1, 7 § Data Protection Act 2018
[129] Chapter 21, 7 § Public Access to Information and Secrecy Act
[130] Magnus Stenbeck and others (n 120) 386

research that involves human biological samples taken from a living person or linked to that person is necessary.[131] As a general rule, Sweden's legal ground for research is public interest rather than consent, which strongly focuses on public interest as the default legal basis for processing personal data in research. However, there are also exceptions to the rule. There were situations where research by pharmaceutical companies did not fall under this general rule, so in those cases, the processing of personal data may be allowed by consent or under Article 6(1)(f).[132] Moreover, even if the legal basis for processing personal data is based on public interest, consent might still be required to grant ethical approval of a research project. Last but not least, there is a mandatory consent requirement for collecting and preserving samples in biobanks and their general availability for medical treatment and research purposes.[133]

In Ireland, the Health Research Regulations (HRRs) introduced additional regulatory requirements in accordance with Article 9 (4) of GDPR for health research regarding governance, processes, and procedures.[134] Among these requirements, one is that 'identified or identifiable personal data cannot be included in health research' unless: (i) GDPR "explicit consent" exists or (b) a consent declaration has been granted.[135] As it is evident, informed consent is central to the fundamental rights of the data subject and the core of health research. Even though the process is straightforward, it still has complexity in practice. While explicit consent is another lawful basis for processing under Article 9, it is not a mandatory requirement. However, the HRRs requires mandatory GDPR explicit consent as a safeguard to the processing of personal data in health research.[136] In other words, researchers must obey Article 6 and 9 GDPR requirements as well as the GDPR explicit consent safeguard and informed consent as a lawful basis which can be seen as an unnecessary burden.[137] With this in mind, various European bodies and institutions have stated that GDPR consent could not always be

[131] 4 § p. 3 and 6 § Ethical Review Act (2003: 460 [Lag (2003:460) om etikprövning av. forskning som avser människor Act].
[132] Magnus Stenbeck and others (n 120) 386
[133] Ibid
[134] Health Research Consent Declaration Committee < https://hrcdc.ie/about-us/>
[135] Mary Kirwan (n 97) 516
[136] Ibid
[137] Ibid 517

appropriate for health research.[138] Several problems have been mentioned for the HRRs approach, such as achievability problems regarding the criteria for valid consent, technical and bureaucratic burden, and blanket application of GDPR explicit consent.[139]

Despite the fact that the GDPR provides the legal basis for scientific research, each Member State has tailored its approach according to its standards in light of the GDPR. Although the general frame is similar, details are different in regard to what legal basis is acceptable, the scope and conditions for consent, and the administrative process.[140]

5. THE STATUS OF GENOMIC RESEARCH UNDER THE GDPR

5.1. Are Genomic Medicine, Precision Medicine, and Personalized Medicine the same?

Before discussing the subject, it is vital to provide a comprehensive definition of genomic medicine, personalized medicine, and precision medicine. Although many assume that these are different terms for the same concept, they are not. One of the standard definitions for genomic medicine is "using an individual patient's genotypic information in their clinical care."[141] In other words, genomic medicine is the application of diagnostic or therapeutic tools for personalizing the components of patient care.[142] Regarding precision medicine, some researchers tend to use precision medicine and personalized medicine interchangeably and define them as "individual characteristics related to genotype and environmental factors that are decisive for

[138] Ibid
[139] Ibid 518
[140] For more comparative details read: Olga Tzortzatou and others, Biobanking Across Europe Post-GDPR: A Deliberately Fragmented Landscape in Santa Slokenberga and others (ed), *GDPR and Biobanking* (Springer 2021) 405-408
[141] Teri A Manolio and others, 'Implementing genomic medicine in the clinic: the future is here' [2013] Genetics in Medicine (GIM) < https://pubmed.ncbi.nlm.nih.gov/23306799> accessed 20 August 2022
[142] Leigh Ann Simmons and others, 'Personalized medicine is more than genomic medicine: confusion over terminology impedes progress towards personalized healthcare' [2012] 9(1) Personalized Medicine <https://pubmed.ncbi.nlm.nih.gov/29783292/> accessed 20 August 2022

diagnosis, treatment, and prevention of disease."[143] Others define precision medicine as "the use of genomic variant data in caring for patients" and consider precision medicine and personalized medicine as two separate terms.[144] Since some officials and public authorities have defined these terms differently, it is unsurprising that someone might misunderstand them.[145] The concept is sometimes also misunderstood with personalized health care, which is generally an approach to patient care that involves systems biology and personalized predictive, preventive, and participatory care.[146] A definition of personalized medicine could be "tailoring medical treatment to a specific subset of patients who are usually identified by genetic markers."[147]

Nowadays, precision medicine is more than solely genomic data of the patients. It also includes medical records, lifestyle, and environmental factors.[148] In other words, personal data originating from health and medical records, as well as research and clinical, are considered alongside biological samples. Personal data may include genetic and genomic data, other epistemological biomedical information, and environmental, lifestyle, or social data.[149]

[143] Mats G. Hansson, 'Striking a Balance Between Personalised Genetics and Privacy Protection from the Perspective of GDPR' in Santa Slokenberga and others (ed), *GDPR and Biobanking* (Springer 2021) 31
[144] Roden, D. M and Tyndale, R. F, 'Genomic medicine, precision medicine, personalized medicine: what's in a name?' [2013] 94(2) Clinical pharmacology and therapeutics 169–172 < https://ascpt-onlinelibrary-wiley-com.ludwig.lub.lu.se/doi/full/10.1038/clpt.2013.101> accessed 20 August 2022
[145] For more details, read: Leigh Ann Simmons and others (n 138)
[146] Leroy E Hood and others, 'Systems biology and new technologies enable predictive and preventative medicine' [2004] Science 306(5696) < https://pubmed.ncbi.nlm.nih.gov/15499008> accessed 20 August 2022. Also, Andrea D Weston and others, 'Systems biology, proteomics, and the future of health care: toward predictive, preventative, and personalized medicine' [2004] 3(2) Journal of Proteome Research 179-196 < https://pubmed.ncbi.nlm.nih.gov/15113093> accessed 20 August 2022
[147] Thomas J Newman and Jeffrey J Freitag, 'Personalized Medicine Development' [2011] 20(7) Applied Clinical Trials 30 < https://appliedclinicaltrialsonline.com/view/personalized-medicine-development> accessed 20 August 2022
[148] Roy C. Ziegelstein (n 46) 160-168
[149] Ana Nordberg, 'Biobank and Biomedical Research: Responsibilities of Controllers and Processors Under the EU General Data Protection Regulation' in Santa Slokenberga and others (ed), *GDPR and Biobanking* (Springer 2021) 72

5.2. Is Anonymous Data Achievable?

Anonymity is an essential condition of biomedical research, making it impossible to identify the person behind the data. Data subjects donate their DNA samples for three main reasons.[150] The first reason is to support personalized medicine studies. The second reason is to get knowledge about their genetic predispositions to diseases and their genetic compatibilities with potential partners. Third, to identify their distant patrilineal relatives and the potential surnames of their biological fathers. Most respondents have positive attitudes towards genomics research and donating their DNA samples;[151] preserving their data is very important.

The GDPR does not contain a definition of what constitutes anonymous data, and as a result, it is not easy to know where to draw the line and whether the GDPR applies.[152] From a legal perspective, the GDPR has a broad interpretation of the scope of personal data, and it is unlikely that utilizing technological advancements like homomorphic encryption (FHE)[153] or secure multi-party computing (SMC)[154] will be exempt from applying the GDPR. Data Privacy and Bioinformatic specialists claim that current technical solutions are insufficient to preserve data subjects' long-term privacy.

In conclusion, it is necessary to collaborate on technical solutions, policies, and legislation to ensure the privacy of the data subjects sufficiently; otherwise, anonymity in its absolute meaning would be almost impossible, and it is only a matter of time before every individual can be identified in a so-called anonymous set.[155] A revision to the privacy rules may be necessary for a higher level of protection under Art 9 GDPR;

[150] Mohammed Alser, 'Can you Really Anonymize the Donors of Genomic Data in Today's Digital World?' in Joaquin Garcia-alfaro and others (ed), *Data Privacy Management, and Security Assurance* (Springer 2015) 237
[151] Ibid
[152] Recital 43 GDPR
[153] For more details read: Avradip Mandal and others, 'Comprehensive and Improved Secure Biometric System Using Homomorphic Encryption' in Joaquin Garcia-alfaro and others (ed), *Data Privacy Management, and Security Assurance* (Springer 2015) 183
[154] For more details read: Sabrina De capitani di vimercati and others, 'Data Protection in Cloud Scenarios' in Joaquin Garcia-alfaro and others (ed), *Data Privacy Management, and Security Assurance* (2015) 6
[155] Paula Jansen, 'Research Data Stewardship for Healthcare Professionals' in Pieter Kubben (ed), *Fundamentals of Clinical Data Science* (Springer 2019) 52

however, any further requirement can complicate cross-border processing and data sharing.[156]

5.3. What Matters Most, Research or Personal Privacy?

Medical service is becoming more precise and personalized, especially in the precision medicine sector. Meanwhile, privacy rules preserve personal information, affecting processing data at both clinical and research levels. Still, the question is to what extent privacy rules should apply in order to protect personal privacy but give enough space to the researchers. In the past, genetic testing was more focused on relatively rare and high penetrance inherited diseases. However, in some disorders such as dementia, heart disease, diabetes, and cancer, genetics has a minor influence, acting together with environmental or epigenetic factors.[157] As a result, genetic, medical records, lifestyle, and environmental data are critical for developing precision medicine due to the scarcity of research participants, samples, data, resources, and researchers.[158] Since genomic research involves various data ranging from patients' genetics to their environment and lifestyle, it is essential to consider privacy rules because noncompliance can lead to failing ethical or privacy reviews or even obtaining funding for research, particularly from European Union grants.[159]

On the one hand, privacy is a value that gives everyone the right to determine who can access their private information. The CFR has recognized this value as the right to respect an individual's private life, protect personal data, and respect physical and mental integrity in medicine and biology fields.[160] On the other hand, there is the right to social security benefits and social services to protect individuals against illness,[161]

[156] University of Oxford (n 6) 61
[157] Mats G. Hansson (n 139) 32
[158] Deborah Mascalzoni and others, 'International Charter of principles for sharing bio-specimens and data' [2014] 23(6) European journal of human genetics 721< https://www.nature.com/articles/ejhg2014197> accessed 20 August 2022
[159]Christopher F Mondschein and Cosimo Monda (n 86) 56
[160] Article 3(2), 7, and 8 of The Charter of Fundamental Rights of The European Union (2000/C 364/01)
[161] Ibid Article 34

the right to access preventive health care, and the right to benefit from medical treatment, as well as a high level of human health protection.[162]

In this regard, one might argue that these rights are for individuals, not researchers, and it does not necessarily imply the necessity of research projects. The answer is that it would be meaningless if there were no corresponding duties towards a right; the right to health and medical treatment requires further research to fulfill them in practice.[163] In addition, the right to conduct research is also protected under the CFR to ensure the freedom of academic research, which shows the importance of research development alongside other fundamental rights.[164]

It is not easy to determine which right overrides the other. However, it is possible to strike a balance between the right to conduct research and personal privacy rights in accordance with the principle of proportionality. While genomic research has contributed so much to health and society, it should respect individuals' privacy and ethics. As was mentioned in the ethical section, science can bring various benefits to the patients and the healthcare community; however, we should not ignore the fact that any compromises on fundamental human rights can lead to disaster.

Generally, public health is a shared competence between the EU and the Member States. In other words, EU Member States have the primary responsibility in the health care system, but the EU has competence in public health and policymaking.[165] The European Commission, the Member States, and the European Parliament must overcome barriers, avoid duplication, and support the best projects for patients and citizens.[166] Therefore, the EU is entitled to lay down principles that can strike a balance

[162] Ibid Article 35
[163] Mats G. Hansson (n 139) 35
[164] Article 13 of The Charter of Fundamental Rights of The European Union (2000/C 364/01)
[165] Treaty on the Functioning of the European Union [2007] OJ 1 115/122
[166] 'A European health union - increasing EU competence in health - Coping with Covid-19 and looking to the future' (Progressive Alliance of Socialists and Democrats, November 2020) <socialistsanddemocrats.eu/sites/default/files/2020-11/european_health_union_sd_position_en_30512.pdf> accessed 25 October 2021

between different rights; here, the GDPR has the role of protecting privacy and making a balance.[167]

There are derogations introduced by the GDPR. For example, Article 89(2) GDPR gives more space to scientific research by derogating from data subject rights; however, this derogation does not include the duties found in the rights to information, the obligations to keep records, or the general responsibilities that attach to processors.[168] Similarly, Article 85(1) GDPR facilitates processing for academic purposes. Last but not least, in Article 11 GDPR, even though the data remains identifiable and not anonymized, it is allowed to derogate from data subject rights where identification of the data subject is not required for processing.[169]

According to the GDPR, the right to protect personal data is not absolute and must be considered as per its function.[170] The right to personal data protection should be balanced against other fundamental rights under the principle of proportionality.[171] From a doctrinal legal perspective, it is not possible to know where to draw the balance line between privacy protection and scientific research; maybe the Court of Justice of the European Union can clarify the balance in future cases.[172]

In conclusion, the situation varies from case to case, depending on the subject's importance and the project's possible outcomes. There is no one-size-fits-all solution because not every project has the same conditions or privacy risk; what matters most is to make a balance and reach the point that ensures personal data in a proportionate way that gives enough space for research activities according to their circumstances.

[167] Mats G. Hansson (n 139) 36
[168] University of Oxford (n 6)
[169] Ibid
[170] Recital 4 GDPR
[171] Ibid
[172] Mats G. Hansson (n 139) 36

5.4. Is the GDPR Sufficient and Efficient?

The GDPR is sector-neutral legislation,[173] and as was mentioned previously, it has a broad approach.[174] Even though GDPR is a comprehensive tool that covers a wide range of subjects concerning personal data protection, sometimes this approach causes legal uncertainty and requires expertise to ensure compliance.[175] Similar to Directive 95/46/EC,[176] the objective of the GDPR is to provide an equivalent level of protection for natural persons and the free flow of personal data throughout the EU. The Union introduces measures to achieve this objective better at the EU level according to the principle of subsidiarity and proportionality.[177] Be that as it may, some researchers claim that it is not clear what level of harmonization the GDPR seeks since it has several opening clauses which make enough space for the Member States to make decisions at the national level, which may undermine this objective.[178]

According to Articles 85(1) and Article 89(2) GDPR, Member States are entitled to introduce specific derogation for academic and research purposes. At first glance, these derogations can facilitate research projects; however, they may lead to a fragmentation of the rules governing research in the long term because each Member State takes a different approach.[179] It is also possible that the GDPR does not intend to harmonize national provisions in detail and only aims to encourage the Member States to find harmonized solutions themselves. This approach can be seen in Article 29 Working Party[180] approach as well; although it promotes the Member States to search for a harmonized solution for age in Paragraph 131, it explicitly mentions the scope of the GDPR does not harmonize national provisions in paragraph 151. National governments are indeed competent to provide the resources needed to implement rights to health, medicine, and social services. However, it does not mean that the EU does not have the

[173] Santa Slokenberga and others, *GDPR and Biobanking* (1st edn, Springer International 2021) 14
[174]Christopher F Mondschein and Cosimo Monda (n 86) 57
[175] Ibid 58
[176] Recital 3 GDPR
[177] Recital 170 GDPR
[178] Christopher F Mondschein and Cosimo Monda (n 86) 58
[179] Ibid
[180] Article 29 Data Protection Working Part (WP29), *Guidelines on consent under Regulation 2016/679*, [2017] (WP259 rev.01)

power to introduce principles that guide the balancing of the different rights and interests.[181] In practice, the GDPR's approach is ambiguous, leaving the researchers uncertain, particularly with cross-border flows of personal data due to widely divergent national standards.

One of the critical elements of genomic research is the ability to share data, especially in rare diseases.[182] Recital 53 of GDPR stipulates that the Member States are allowed to maintain or introduce further conditions or limitations to ensure cross-border flows of genetic data. Still, it should not impede the free flow of personal data within the Union when it comes to cross-border processing of such data. Notwithstanding this option, the process of personal data should be lawful and fair, ensuring appropriate security and confidentiality of the personal data.[183] Therefore, the GDPR is trying to balance preserving data subjects' privacy and the possibility of cross-border data processing and sharing.

The GDPR ensures a sufficient level of personal data protection alongside further facilitating the free flow of personal data within the Union and the transfer to third countries and international organizations. Yet the question is whether introducing general principles at the Union level and authorizing the Member States to lay down national provisions is efficient. Article 9(2)(j) GDPR indicates that special categories of personal data, including genetic data, could be processed for scientific research under the conditions in Article 89(1). Meanwhile, Article 9(4) GDPR gives the Member States the power to maintain or introduce further requirements, including limitations, concerning the processing of genetic data, biometric data, or data concerning health. On the one hand, processing personal data under Article 9(2)(j) GDPR is permissive, meaning that genetic data can be processed as long as it meets the conditions in Article 89(1) GDPR. On the other hand, processing special personal data under Article 9(4) GDPR is restrictive because the Member States have the last word. As it was apparent from experience in different Member States, this can lead to uncertainty and imbalance;

[181] Mats G. Hansson (n 139) 36
[182] Ibid
[183] Recital 39 GDPR

as a result, the GDPR might be neither an efficient nor successful tool for balancing the protection of personal data and the right to conduct genomic research.

More tangible cases show that the answer to the question is not positive in practice either. In Portugal, "the scientific research carried out by the national research centers is in unequal circumstances vis-à-vis their peers."[184] In Sweden, "the GDPR has not been implemented in a clear and unequivocal manner, thus leaving researchers with an imprecise and ambiguous framework."[185] In Ireland, "the HRRs (Health Research Regulations) have heavily impacted on Ireland's capacity to conduct health research, including clinical trials (both interventional and noninterventional) and caused significant damage to Irish research."[186]

As a general rule, the EU does not take action unless it is more effective at the EU than at the national level.[187] Public health is a shared competence between the EU and its Member States. In other words, Member States have the primary responsibility in the health care system, but the EU has competence in public health and policymaking.[188] While the EU and the Member States were almost satisfied with their approach to healthcare policy, the outbreak of Covid-19 startled them with a huge impact that was not limited to public health but also affected the economy. The pandemic highlighted the fragility of current policy for collaborations between the Member States and EU institutions.[189] As a result, an initiative called 'the European Health Union (EHU)' started after Ursula von der Leyen, the President of the European Commission, invited the Member States to work together to detect, prepare and respond collectively and to build the foundations of a stronger European Health Union where.[190] Some researchers argue that without changing the Treaty, the Commission's proposal for making the EHU is unlikely to succeed, and the EU and the Member States should

[184] Carla Barbosa and Andreia da Costa Andrade, (n 107) 361
[185] Magnus Stenbeck and others (n 120) 394
[186] Mary Kirwan (n 97) 520
[187] Article 5 TEU
[188] Article 168(1) TFEU
[189] Vytenis Andriukaitis, 'A European Health Union' in Maria João Rodrigues (ed), *Our European Future Charting a Progressive Course in the World* (London Publishing Partnership 2021) 25
[190] Ursula von der Leyen, President of the European Commission, speaking at the World Health Summit (25 October 2020)< https://ec.europa.eu/commission/presscorner/detail/en/speech_20_1983> accessed 22 August 2022

make more ambitious decisions rather than slow developments.[191] Regarding integration, other researchers have controversial extreme liberal views which ask for a full negative integration supported by positive integration,[192] which might not be realistic under the current legal framework concerning public health.

6. Analysis and Discussion

Genetic data is being widely collected and processed by public and private healthcare entities for treatment, research, and commercial purposes. Collecting and processing genetic data brings responsibility in every respect and from an ethical aspect. On the one hand, physicians and researchers are spending so much time studying and researching to benefit humanity and science. On the other hand, patients and data subjects have fundamental rights regarding personal data protection, and, more importantly, their consent is needed as a general rule for any data processing.[193]

From an ethical perspective, the processing should also be ethical and respect fundamental rights and freedoms. Although there is a presumption that science and research are beneficial and not harmful, there have been cases that severely undermined fundamental rights and human dignity.[194] Therefore, it seems necessary that science and ethics go hand in hand to attain new scientific developments and preserve fundamental rights.

From a legal perspective, the EU General Data Protection Regulation (GDPR) preserves personal data privacy and establishes a research regime that aims for research harmonization.[195] The key provision for processing genetic data for scientific research is Article 9 GDPR. This provision allows the processing of genetic data, one of the special personal data categories, if it meets one of the required conditions.[196]

[191] Vytenis Andriukaitis (n 185) 26
[192] Desmond Dinan, The Historiography of European Integration. in Desmond Dinan (ed), Origins and Evolution of the European Union (Oxford University Press 2006) 319
[193] Section 1.1 and 3.1
[194] Section 3.1
[195] Section 4.2.2
[196] Section 1.1, 3.3.1, and 4.

Researchers and physicians can generally process genetic data under one of these Article 9(2) requirements. Among these conditions, two are more commonly chosen as a legal basis for genetic data processing. The first one is Article 9(2)(a), which requires explicit consent, and the other one is Article 9(2)(j), which applies to processing genetic data for scientific research purposes with two additional conditions. First, conducting such processing should be necessary, and second, enact Article 89(1) requirements.[197] Both Article 9(2)(a) and 9(2)(j) face challenges in practice. Under Article 9(4) GDPR, the Member States have the authority to keep or introduce more conditions and limitations on processing genetic data. The rationale behind Article 9(4) GDPR may be the Member States' competence and primary responsibility in health care. This can severely affect the situation for genomic research since the Member States might introduce more strict rules that can affect such research directly or indirectly in different stages.[198] These restrictions and limitations can occur before the research for ethical reviews, during the project for consent requirements, or after the research for cross-border flow of personal data.[199] Meanwhile, although there is a presumption that Recital 33 facilitates research projects, processing special categories of personal data like genetics require explicit consent, which causes more scrutiny.[200] As a result, researchers are trapped in a vicious circle of rules that look permissive but are restrictive.

Two hypothetical cases will be explained below to elaborate on the challenges of genomic research at national and cross-border:

- Scenario A is where a group of Swedish researchers intends to process genetic data in Sweden, which is beneficial from a research perspective. They receive ethical approval from Etikprövningsmyndigheten, and they choose Article 9(2)(j) as a legal basis for their project and ensure that the purpose of the project is proportionate and in accordance with Article 89(1). This approval is not an ethical permit or a safeguard for research conducted outside Sweden. As a result, these researchers can not rely on

[197] Section 4
[198] Section 4.2.3
[199] Ibid
[200] Section 4.2.1

the authorization issued by the Swedish Ethical Review Authority in the other EU Member States.[201]

- Scenario B is where a group of Swedish researchers intends to process genetic data in Sweden, which is beneficial from a research perspective. They receive ethical approval from Etikprövningsmyndigheten, and they choose Article 9(2)(a), explicit consent, as a legal basis for their project. In the middle of their project, they neede to process data in Ireland. Although their lawful basis for processing is the explicit consent of data subjects, they are not allowed to process data. The reason is a mandatory requirement by Irish rules, which requires processing special categories of data in Ireland only on the grounds of public interest. Therefore, they cannot proceed with their project in Ireland because, as the Article 29 Working Party Guidelines on consent requires, the controller cannot swap from consent to other lawful bases.

By reading the rule of different Member States in detail, there are more examples of how national provisions can, in practice, impede cross-border genomic research in the European Union. There are several suggestions for tackling this problem. Maybe the first solution that comes to mind is the revision of the GDPR or even reforming the Treaties.[202] There have not been any signs of decisions or intentions regarding a change to neither the Treaties nor the GDPR. The limitations of genomic research under the GDPR are indeed important but not deep enough to challenge the Treaties. The GDPR is also limited to the competence of the Union, and there is no legal basis for expanding the scope of EU Law where the subject, here Public Health, is shared competence. A reformation is not solely a legal decision but rather political.

[201] 5 §§ Ethical Review Act.
[202] Section 5.4

From a constitutional perspective, it is reasonable to say both the Treaties and the GDPR are sufficient.[203] It is not logical to attempt a reformation while there is enough potential for further refining a good law. In other words, the Treaties function as they should, and the GDPR protects the processing of personal data. Nonetheless, the second task of the GDPR is to ensure the free movement of such data. In regard to the cross-border flows of personal data, more collaboration and harmonization are needed, which affects research activities. This can be attained through different mechanisms that do not require a central change to the rules.

The ambiguous framework of the GDPR regarding the genetic data process has burdened researchers at the national and international levels.[204] The fact that under Article 9(4) GDPR, each Member State can introduce further conditions and limitations on processing genetic data has faded the potential of Article 9(2) GDPR in practice. The Commission's approach was to encourage the Member States for more collaboration and cooperation through non-binding acts. Notwithstanding, there have been promising initiatives by the Commission that can mitigate the situation for researchers.

One of these mechanisms is through new policies like the European '1+ Million Genomes' Initiative that support the European Union's genomic collaboration and research that benefits researchers, healthcare professionals, and all citizens.[205] This project is awe-inspiring since it is not limited to the EU Member States but includes the non-EU Member States that can contribute to the project. Through this remarkable plan, researchers will have secure access to genomics and clinical data across Europe.[206]

Another mechanism is through EU strategies based on the necessities within the Union, which can promote integration and harmonization through different methods.[207] The European Health Union is one of the Commission's strategies in the healthcare

[203] Ibid
[204] Section 4.2.3
[205] European Commission, 'European '1+ Million Genomes' Initiative' [2020] <https://digital-strategy.ec.europa.eu/en/policies/1-million-genomes> accessed 22 August 2022
[206] Section 2.3
[207] Section 5.4, also see Annex 3

sector.[208] The Proposal for a Regulation on the European Health Data Space was published in May 2022 that helps Natural persons exercise their rights over their electronic health data, including accessing and transmitting it nationally and cross-borders.[209] Patients can receive healthcare across the EU by controlling their health data in their home country or the other Member States. It also provides a consistent, secure, trustworthy, and efficient framework for using health data and, under certain conditions, gives access to researchers to large amounts of high-quality health data.

One possible solution may be through delegated acts mechanism by the Commission under Article 290 TFEU to supplement or amend Article 9 GDPR and balance the right to protect special categories of personal data and the right to conduct research and process genetic data. Two significant problems with this idea are that, first, it falls under the scope of Article TFEU, and it is shared competence. Second, delegated acts should have a general application. In other words, amending Article 9 GDPR only for processing genetic data and genomic research does not have the general application condition, and it is unlikely to receive the European Parliament or the Council's approval.

The analysis of this thesis is a critical evaluation of the situation. However, it is unlikely to see a significant change in the GDPR or an amendment to the Treaties to increase EU competence in public health in the near future. The Commission wisely monitors the situation and tackles the problem through soft law in various ways ranging from President of the European Commission speeches[210] to initiatives like EHU or 1+ Million Genomes. Harmonization has different techniques and strategies; according to the current situation, the Commission's actions may be promising and help mitigate the problem. One realistic solution is inviting the Member States to collaborate more and encouraging them to facilitate genetic research can approximate national rules. Additionally, promoting and developing policies that can accelerate collaboration, such

[208] European Commission, 'European Health Union' <https://ec.europa.eu/info/strategy/priorities-2019-2024/promoting-our-european-way-life/european-health-union_en#latest> accessed 22 August 2022
[209] Proposal for a regulation - The European Health Data Space COM(2022) 197/2
[210] European Commission, 'State of the Union Address by President von der Leyen at the European Parliament Plenary' [2020] < https://ec.europa.eu/commission/presscorner/detail/en/SPEECH_20_1655>

as EHU, or 1+ Million Genomes, are desirable. Otherwise, any attempt to amend the GDPR or the Treaties not only is a lingering process but also causes political tensions in practice, which cannot be of any help to genomic research.

6.1. CONCLUSION

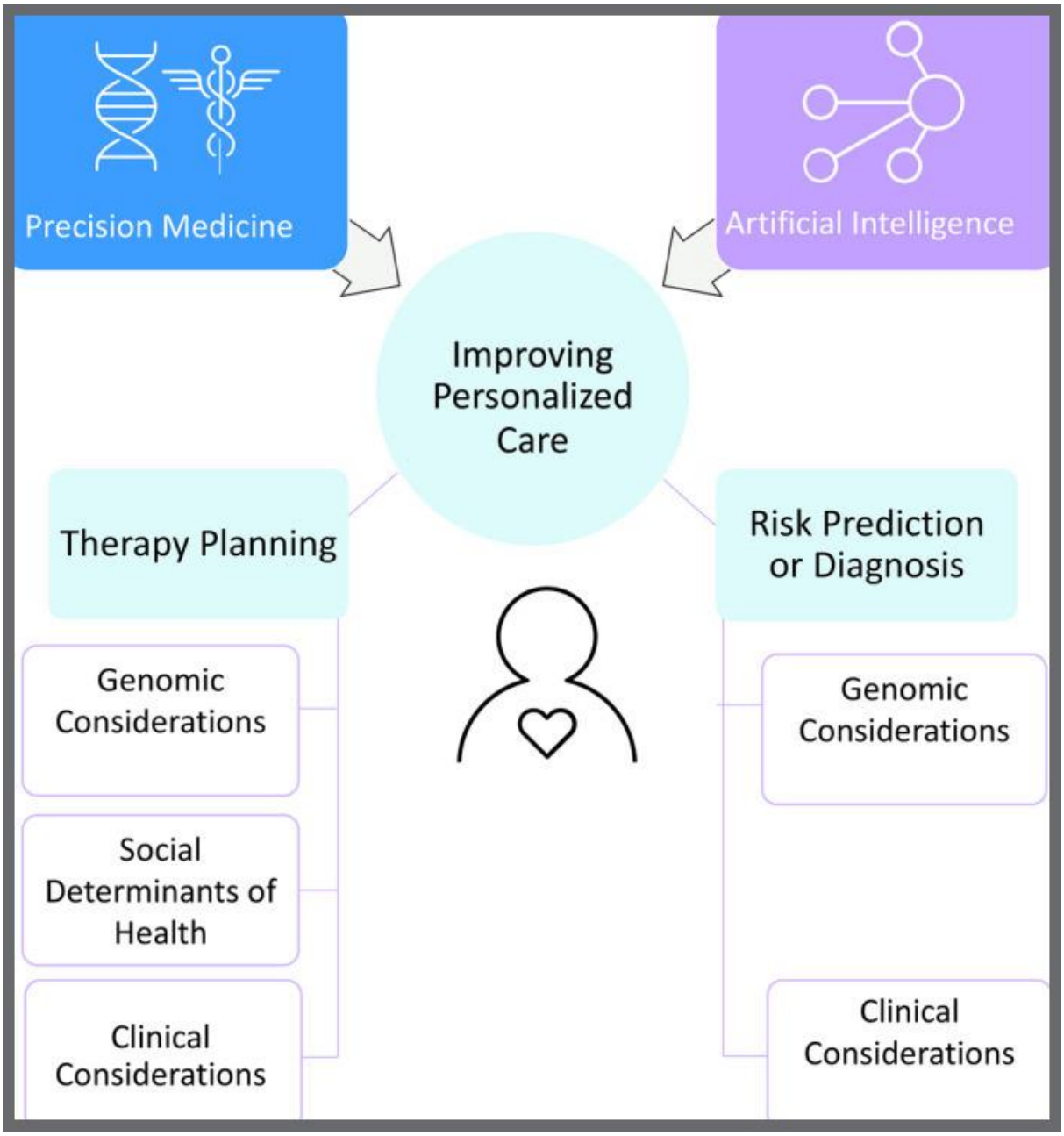
Striking a balance between the need to protect the privacy of data subjects and the need to give access to data for research purposes is indeed a challenge. The current situation has made researchers face barriers to proceeding with their projects. While Art 9(2)(j) GDPR meets genomic research needs and provides the basis to conduct genomic research and process genetic data, Art 9(4) GDPR gives enough space to the member states to restrict genomic research, which affects cross-border research.

Public health is a shared competence and also a sensitive political matter. Within the current legal framework and in theory, the GDPR allows balancing Art 9(2)(j) and Art 9(4) in a way that does not undermine national competence or deprive genomic research. In practice, researchers are trapped in a vicious circle of rules that look permissive but are restrictive. Although this mechanism is not the most efficient tool, and national requirements vary from one member state to another, amending the GDPR or the Treaties is not also an efficient solution.

A deep dive into the matter is needed to solve the problem. It is unlikely that the Member States will refrain from enjoying their competence regarding public health in the near future. Therefore, the complication is not related to the GDPR itself but to the fact that it cannot regulate what goes outside its scope. Both Art 9(2)(j) and Art 9(4) are functioning as they should; however, it is for the Member States to decide how much they are willing to cooperate at the EU level. Meanwhile, the Commission plays a critical role in harmonizing national rules by inviting the Member States to collaborate and encouraging them to facilitate genetic research.

To conclude, the priority is to preserve the privacy of data subjects. In light of the GDPR, it is essential to facilitate genomic research due to the numerous benefits it brings. In the current situation, reforming the GDPR or the Treaties is neither helpful nor likely to happen. The Commission’s approach should be trusted since it is more likely to mitigate the problem through soft law by inviting the Member States and encouraging them to collaborate.

ANNEX 1[211]



Note: This figure describes how precision medicine and artificial intelligence techniques impact the goal of personalized care.

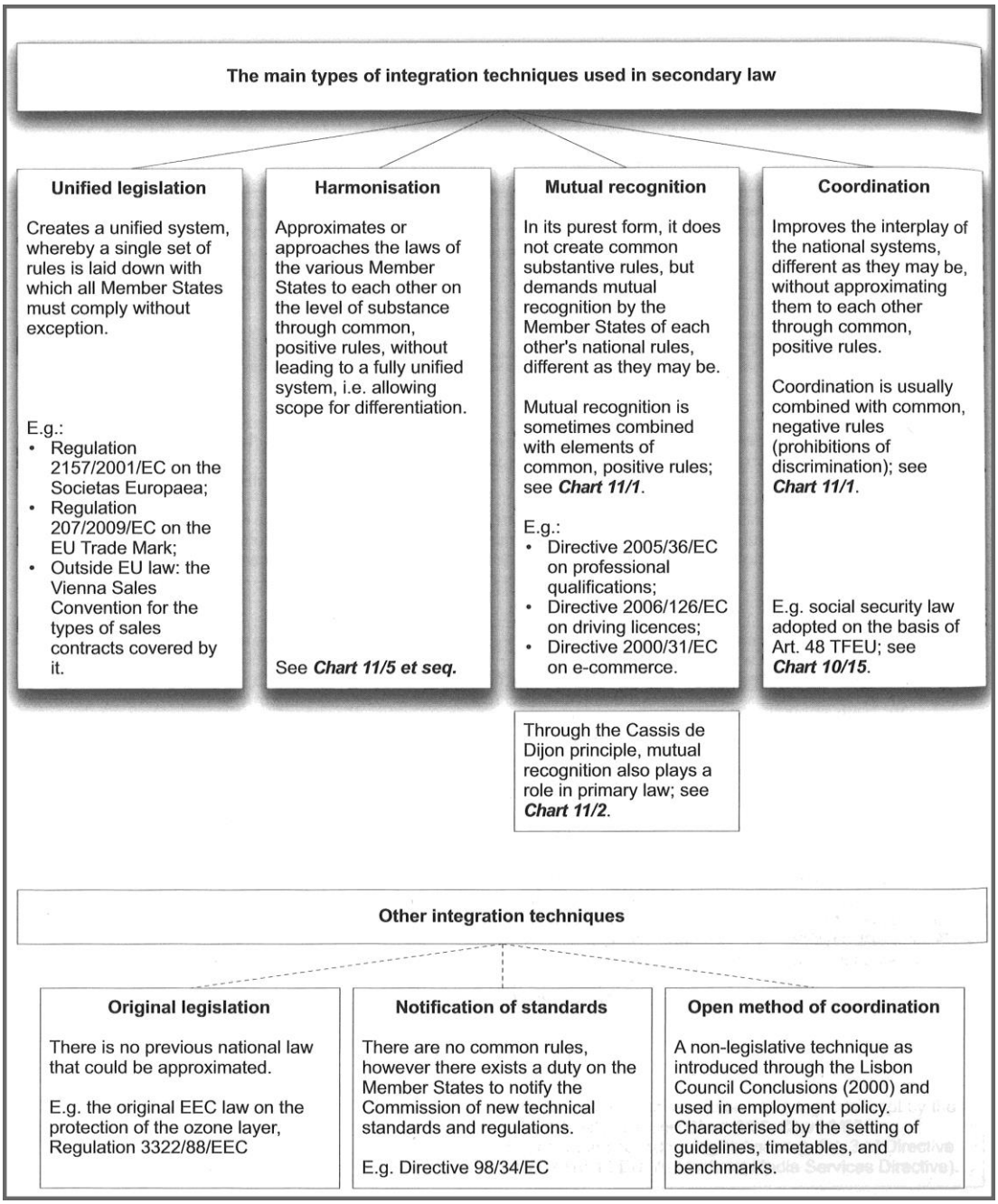
[211] Kevin B. Johnson and others (n 31)

ANNEX 2[212]

Note: This figure shows how many countries have signed the 1+MG Declaration since 2018

[212] European Commission (n 66)

ANNEX 3[213]



Note: This figure shows different types of integration, including the coordination method.

[213] Christa Tobler and Jacques Beglinger, Essential EU law in charts / Christa Tobler, Jacques Beglinger (2nd edn, HVG-ORAC 2013) 282

Bibliography

Legislations

Treaties

Charter of Fundamental Rights of the European Union (2000/c 364/01) (CFR), OJ C 2012/C 326/02

Consolidated version of the Treaty on the European Union (TEU), OJ C C115/13

Consolidated version of the Treaty on the Functioning of the European Union (TFEU), OJ C 326/01

EU Regulation

Regulation (EU) 2016/679 of the European Parliament and of the Council of 27 April 2016 on the protection of natural persons with regard to the processing of personal data and on the free movement of such data, and repealing Directive 95/46/EC (General Data Protection Regulation) OJ L 119/1

Parliament and Council Directive 95/46/EC of 24 October 1995 on the protection of individuals with regard to the processing of personal data and on the free movement of such data [1995] OJ L 281/31

Official Documents

Article 29 Data Protection Working Part (WP29), *Guidelines on consent under Regulation 2016/679*, [2017] (WP259 rev.01)

European Commission, 'Linking European and Chinese Research Infrastructures through the ORIENTplus project' [2015] <https://digital-strategy.ec.europa.eu/en/news/linking-european-and-chinese-research-infrastructures-through-orientplus-project> accessed 29 August 2022

European Commission, 'The Roadmap of the 1+Million Genomes Initiative is now clearly illustrated in a new brochure'(European Commission, 28 September 2020) <https://digital-strategy.ec.europa.eu/en/library/roadmap-1million-genomes-initiative-now-clearly-illustrated-new-brochure> accessed 29 August 2022

European Commission, 'White Paper on Artificial Intelligence - A European Approach to Excellence and Trust' (2020)16 <https://ec.europa.eu/info/publications/white-paper-artificial-intelligence-european-approach-excellence-and-trust_en> accessed 29 August 2022

Books

Alser M, ‘Can you Really Anonymize the Donors of Genomic Data in Today’s Digital World?’ in Joaquin Garcia-alfaro and others (ed), *Data Privacy Management, and Security Assurance* (Springer 2015)

Andriukaitis V, ‘A European Health Union’ in Maria João Rodrigues (ed), *Our European Future Charting a Progressive Course in the World* (London Publishing Partnership 2021)

Barbosa C and Andrade A, Biobanks and GDPR: ‘A Look at the Portuguese Panorama’ in Santa Slokenberga and others (ed), *GDPR and Biobanking* (Springer 2021)

Bell J, ‘Legal Research and the Distinctiveness of Comparative Law’ in Mark Van Hoecke (ed), *Methodologies of Legal Research: What Kind of Method for What Kind of Discipline?* (Hart Publishing 2011)

Dinan D, The Historiography of European Integration. in Desmond Dinan (ed), Origins and Evolution of the European Union (Oxford University Press 2006)

Marshall J, *Human Rights Law and Personal Identity* (1st edn, Routledge 2014)

Niemiec Ed, Consenting Patients to Genome Sequencing. in Aad Tibben and Barbara B biesecker (eds), *Clinical Genome Sequencing* (ScienceDirect 2019)

Nordberg A, ‘Biobank and Biomedical Research: Responsibilities of Controllers and Processors Under the EU General Data Protection Regulation’ in Santa Slokenberga and others (ed), *GDPR and Biobanking* (Springer 2021)

Krzysztofek M, *GDPR: Personal Data Protection in the European Union* (Kluwer Law International 2021)

Jamal L, ‘An Ethical Framework for Genetic Counseling in the Genomic Era’ in Glenn Cohen and others (eds), *Consumer Genetic Tehnologies* (Cambridge University Press 2021)

Jansen P, ‘Research Data Stewardship for Healthcare Professionals’ in Pieter Kubben (ed), *Fundamentals of Clinical Data Science* (Springer 2019)

Vranken J, ‘Methodology of Legal Doctrinal Research: A Comment on Westerman’ in Mark Van Hoecke (ed), *Methodologies of Legal Research: What Kind of Method for What Kind of Discipline?* (Hart Publishing 2011)

Hage J, ‘The Method of a Truly Normative Legal Science’ in Mark Van Hoecke (ed), *Methodologies of Legal Research: What Kind of Method for What Kind of Discipline?* (Hart Publishing 2011)

Sorenson J, ‘From Social Movement To Clinical medicine’ in A Milunsky and G J Annas (eds), *Genetics and the Law* (Plenum Press (Springer) 1976)

Stenbeck M and others, Swedish Law on Personal Data in Biobank Research: Permissible But Complex. in Santa Slokenberga and others (ed), *GDPR and Biobanking* (Springer 2021)

Luria S, ‘Biological Roots of Ethical Principles’ in Aubrey Milunsky and George J Annas (eds), *Genetics and the Law* (2nd ed, (Plenum Press (Springer) 1976)

Domurath I, ‘The Politics of Interdisciplinarity in Law’ in Marija Bartl and Jessica C Lawrence (eds), *The Politics of European Legal Research* (Edward Elgar Publishing 2022)

McHale J, *Medical Confidentiality and Legal Privilege* (1st edn, Routledge 1993)

Mondschein C and Cosimo Monda, 'The EU's General Data Protection Regulation (GDPR) in a Research Context' in Kubben and others (eds), *Fundamentals of Clinical Data Science* (Springer, 2018)

National Research Council, '*Toward Precision Medicine: Building a Knowledge Network for Biomedical Research and a New Taxonomy of Disease*' (The National Academies Press, 2011)

Panesar A, *Machine Learning and AI for Healthcare* (2nd edn, Apress 2021)

Santa Slokenberga and others, *GDPR and Biobanking* (1st edn, Springer International 2021)

Topol E, '*Deep Medicine: How Artificial Intelligence Can Make Healthcare Human Again*' (Basic Books, 2019

Articles

Aronson S and Rehm H, 'Building the foundation for genomics in precision medicine' [2015] Nature < https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5669797/> accessed 27 August 2022

Bird E and others, 'The ethics of artificial intelligence: Issues and initiatives' ((EPRS), European Parliamentary Research Service 2020)

Bommenel E, 'Sockerförsöket : kariesexperimenten 1943-1960 på Vipeholms sjukhus för sinnesslöa' (Arkiv förlag, 2020) < https://portal.research.lu.se/sv/publications/the-suger-experiments> accessed 28 August 2022

Bycroft C and others, 'The UK Biobank resource with deep phenotyping and genomic data' [2018] Nature 203 < https://www.nature.com/articles/s41586-018-0579-z> accessed 28 August 2022

Cai Y and Huang T, 'Accelerating precision medicine through genetic and genomic big data analysis' [2018] 1864(6), Biochimica et Biophysica Acta (BBA) - Molecular Basis of Disease< https://pubmed.ncbi.nlm.nih.gov/29548968/> accessed 25 August 2022

Collins F and others, 'The Human Genome Project: Lessons from Large-Scale Biology' [2003] Science < https://pubmed.ncbi.nlm.nih.gov/12690187/> accessed 29 August 2022

Davenport T and others, 'Using AI to improve electronic health records' [2018] Harvard Business Review< https://hbr.org/2018/12/using-ai-to-improve-electronic-health-records> accessed August 2022

Dove E and Chen J, 'Should consent for data processing be privileged in health research? A comparative legal analysis' [2020] 10(2) International Data Privacy Law 118 < https://academic.oup.com/idpl/article/10/2/117/5757981> accessed 18 August 2022

Gesley J, 'Legal and Ethical Framework for AI in Europe: Summary of Remarks' [2020] 114 Proceedings of the ASIL Annual Meeting < https://www.cambridge.org/core/journals/proceedings-of-the-asil-annual-meeting/article/abs/legal-and-ethical-framework-for-ai-in-europe-summary-of-remarks/9FFEF2BE5DE62BFA5A974A7E565473B3> accessed 27 August 2022

Hartmaier R and others, ‘High-Throughput Genomic Profiling of Adult Solid Tumors Reveals Novel Insights into Cancer Pathogenesis’ [2017] 77(9) Cancer Res. 2464< https://pubmed.ncbi.nlm.nih.gov/28235761> accessed 25 August 2022

Hoecke V, ‘Methodologies of legal research: what kind of method for what kind of discipline?’ (Hart Oxford, 2011)

Hood L and others, ‘Systems biology and new technologies enable predictive and preventative medicine’ [2004] Science 306(5696) < https://pubmed.ncbi.nlm.nih.gov/15499008> accessed 20 August 2022.

Kotesh R and others, ‘A Medical AI Agent as a Tool for Neuropsychiatric Diagnoses’[2021] 23rd International Symposium on Measurement and Control in Robotics (ISMCR) < https://ieeexplore.ieee.org/document/9263713> accessed 27 August 2022

Johnson k and others, ‘Precision Medicine, AI, and the Future of Personalized Health Care’ [2021] 14(1) Clinical and Translational Science< https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7877825/> accessed 27 August 2022

Jorgensen A and others, ‘Implementation of genotype-guided dosing of warfarin with point-of-care genetic testing in three UK clinics: a matched cohort study’ [2019] 17(76) BMC Medicine < https://bmcmedicine.biomedcentral.com/articles/10.1186/s12916-019-1308-7> accessed 27 August 2022

Joudaki H and others, ‘Improving fraud and abuse detection in general physician claims: a data mining study’ [2016] 5(3) International Journal of Health Policy and Management (IJHPM) < https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4770922> accessed 27 August 2022

Kirwan M, 'What GDPR and the Health Research Regulations (HRRs) mean for Ireland: “explicit consent” a legal analysis' [2020] Springer < https://link.springer.com/article/10.1007/s11845-020-02331-2> accessed 18 August 2022

Lejeune C and others, ‘The Economic, Medical and Psychosocial Consequences of Whole Genome Sequencing for the Genetic Diagnosis of Patients With Intellectual Disability: The DEFIDIAG Study Protocol’ [2022] Frontiers in genetics< https://europepmc.org/article/med/35444683> accessed 28 August 2022

McGrath S and others, ‘Legal Challenges in Precision Medicine: What Duties Arising From Genetic and Genomic Testing Does a Physician Owe to Patients?’ [2021] 8(1) Frontiers in Medicine<https://www.frontiersin.org/articles/10.3389/fmed.2021.663014/full> 1-2, accessed 27 August 2022

Manolio T and others, 'Implementing genomic medicine in the clinic: the future is here' [2013] Genetics in Medicine (GIM) < https://pubmed.ncbi.nlm.nih.gov/23306799> accessed 20 August 2022

Mascalzoni D and others, ‘International Charter of principles for sharing bio-specimens and data’ [2014] 23(6) European journal of human genetics accessed 20 August 2022 https://www.nature.com/articles/ejhg2014197< https://www.nature.com/articles/ejhg2014197> accessed 20 August 2022

Nagaiand A and others, ‘Overview of the BioBank Japan Project: study design and profile’ [2017] Journal of Epidemiology 27(3S)< https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5350590/> accessed 29 August 2022

Nill A and Laczniak G, ‘Direct-to-Consumer Genetic Testing and Its Marketing: Emergent Ethical and Public Policy Implications’ [2020] 175(4) Journal of Business Ethics

Newman T and Freitag J, ‘Personalized Medicine Development’ [2011] 20(7) Applied Clinical Trials 30 < https://appliedclinicaltrialsonline.com/view/personalized-medicine-development> accessed 20 August 2022

Pormeister K, Genetic data and the research exemption: is the GDPR going too far? Int Data Priv Law [2017] 7(2) International Data Privacy Law <https://academic-oup-com.ludwig.lub.lu.se/idpl/article-abstract/7/2/137/3798545> accessed 27 August 2022

Quathem K and Cooper D, ‘European Commission Issues Updated Q&A on Interplay between the GDPR and the Clinical Trials Regulation’ (Inside Privacy, 4 July 2022) <https://www.insideprivacy.com/international/european-union/european-commission-issues-updated-qa-on-interplay-between-the-gdpr-and-the-clinical-trials-regulation> accessed 20 August 2022

Rajkomar A and others, ‘Scalable and accurate deep learning with electronic health records’ [2018] 1(18) NPJ Digital Medicine 1 < https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6550175/> accessed 27 August 2022

Sadilek A and others, ‘Machine-learned epidemiology: real-time detection of foodborne illness at scale’ [2018] 1(36) NPJ Digital Medicine (2018) <https://www.nature.com/articles/s41746-018-0045-1#citeas> accessed 27 August 2022

Saunders G, ‘Leveraging European infrastructures to access 1 million human genomes by 2022’ [2019] Nature reviews. Genetics < https://www-nature-com.ludwig.lub.lu.se/articles/s41576-019-0178-3> accessed 29 August 2022

University of Oxford, ‘The GDPR and genomic data - the impact of the GDPR and DPA 2018 on genomic healthcare and research’(2020)< https://www.phgfoundation.org/media/123/download/gdpr-and-genomic-data-report.pdf?v=1&inline=1> accessed 29 August 2022

Petrov A and Zyryanov A, ‘Formal-Dogmatic Approach in Legal Science in Present Conditions’ [2018] Journal of Siberian Federal University

Peczenik A, ‘Juridikens Allmänna Läror’ [2005] SvJT 249 < https://svjt.se/svjt/2005/249> accessed 27 August 2022

Plan France Médecine Génomique 2025 (Aviesan, 2025; PFMG 2025) <https://pfmg2025.aviesan.fr/en/> accessed 29 August 2022

Sankar P and Parker L ‘The Precision Medicine Initiative's All of Us Research Program: an agenda for research on its ethical, legal, and social issues’ [2017] 19(7) Genetics in medicine: official journal of the American College of Medical Genetics, 19(7) <https://www.gimjournal.org/article/S1098-3600(21)02228-0/fulltext> accessed 28 August 2022

Schee M and others, ‘Breath biopsy for early detection and precision medicine in cancer’ [2018] 12(1) Ecancermedicalscience <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6070367> accessed 25 August 2022

Simmons L and others, ‘Personalized medicine is more than genomic medicine: confusion over terminology impedes progress towards personalized healthcare’ [2012] 9(1) Personalized Medicine <https://pubmed.ncbi.nlm.nih.gov/29783292/> accessed 20 August 2022

Stark Z and others, ‘Australian genomics: a federated model for integrating genomics into healthcare’ [2019] 105(1) American Journal of Human Genetics<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6612707/> accessed 29 August 2022

'The final verdict on Paolo Macchiarini: guilty of misconduct' [2018] Lancet <https://pubmed.ncbi.nlm.nih.gov/30047386/> > accessed 28 August 2022

Vaishya V, and others, ‘Artificial intelligence (AI) applications for COVID-19 pandemic’ [2020] 14(4) Diabetes & Metabolic Syndrome: Clinical Research & Reviews < https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7195043> accessed 27 August 2022

Ziegelstein R, ‘Personomics and precision medicine’ [2017] 128(1) Transactions of the American Clinical and Climatological Association 160 < https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5525386/> accessed 25 August 2022

Vastaamo data breach in Finland, [2020] https://en.wikipedia.org/wiki/Vastaamo_data_breach accessed 28 August 2022

Weston A and others, ‘Systems biology, proteomics, and the future of health care: toward predictive, preventative, and personalized medicine’ [2004] 3(2) Journal of Proteome Research 179-196 < https://pubmed.ncbi.nlm.nih.gov/15113093> accessed 20 August 2022