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Privacy and Data Protection Issues in The Era Of Precision Medicine And Personalized Healthcare

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ABSTRACT

The present research paper delves into the urgent issues pertaining to privacy and data protection within the context of precision medicine and personalised healthcare. The utilisation of sensitive personal health data has become indispensable in the delivery of customised medical treatments and the enhancement of patient outcomes due to the swift progressions in medical technology. Nevertheless, the emergence of this issue has presented considerable ethical, legal, and regulatory dilemmas pertaining to the confidentiality and safeguarding of individuals' health data. This study investigates the potential hazards associated with data breaches, unauthorised access, and the improper utilisation of health data, while also considering the impact on patient autonomy, trust, and confidentiality. The analysis also explores the current legal frameworks and data protection mechanisms in various jurisdictions, emphasising the deficiencies and the necessity for strong privacy safeguards. The primary objective of this study is to make a valuable contribution to the ongoing discourse surrounding the delicate equilibrium between healthcare innovation and safeguarding individuals' privacy rights. This will be achieved through the identification of significant privacy concerns and the proposal of potential remedies.

Keywords: *Privacy, Data Protection, Precision Medicine, Personalized Healthcare, Medical Data*

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1. INTRODUCTION

"*Implanted and internet-connected wearable and implantable devices are collecting more personal data as targeted therapies and personalised treatments progress. This creates serious privacy and data security concerns that must be addressed by law*." [1]

Researchers, ethical review boards, and national authorities are waiting for guidance on how to ethically balance privacy and scientific research, such as biomedical research, in a way that is GDPR-compliant. To get there, you must first understand privacy and the particular problems of future personalised medical advances, which are expected to use huge amounts of personal data. This chapter will teach you about genetic research, ethics, and the GDPR, as well as how to reconcile personalised therapy and data privacy. [2]

One of the biggest genetics advances in recent years is focusing on genotypes and environmental variables that affect illness risk, treatment efficacy, and prevention. This emphasis has dominated genetics in recent years. This is "personalised" or "precision" healthcare. Alternately, personalised healthcare. Risk profiling can help achieve this. Medical treatment and prevention must be tailored to each patient. This change contradicts the General Data Protection Regulation's stated goal of strengthening privacy safeguards for people. Individualised medicine raises the question of how much of the private domain will stay private. [3]

Academic and private researchers are creating genetic data at unprecedented rates. Medical and therapeutic uses drive this research. Research infrastructures include vast prospective biobanks, omics-databases, and medical and personal data. They want to "revolutionise" clinical and medical applications by personalising patient care. New disease-causing or disease-associated genes are found often now that genomics and next-generation sequencing are mature enough for investigation. Genetic and lifestyle-related risk information is important to present and prospective patients. The vision being created and implemented in clinics may allow us to switch from trial and error to evidence-based therapy. [4] Remember that "personalised" doesn't mean customised drugs. Instead, it divides patients into high- and low-sensitivity to pharmaceutical toxicity or responders before therapy. General practitioners' future medical consultations could benefit from pre-emptive genome screening, which is always available as part of a genetic examination, and medical record systems that can handle

large amounts of patient-specific genotypic information. Genome screening is always a possibility in preventative medicine. [5]

Genetic testing was previously exclusively offered through medical genetic services for rare hereditary disorders with a high risk of transmission. Dementia, heart disease, diabetes, and cancer are caused by numerous genes, each of which contributes a little proportion of inherent vulnerability, as well as the environment or epigenetics. [6] Some aspects of a person's environment, such as diet, physical activity, and whether they take potentially harmful medications, can be changed, but others, like air and water pollution and psychological or social stress, cannot. Genetic propensity may lead to risk factor avoidance. Even with normal therapy, such information may be hard to understand. The diagram below shows the present healthcare system problem. [7]

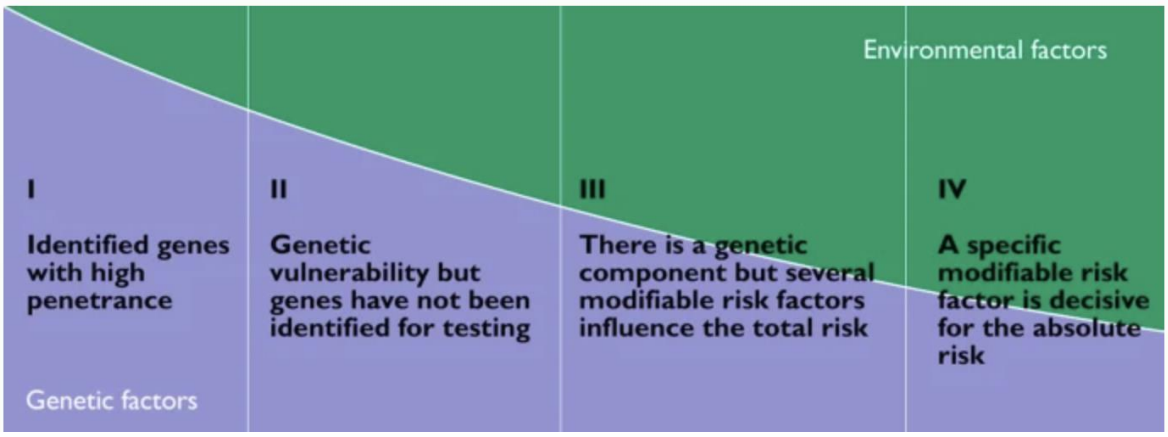

Figure 1: Whether a person's health risk may be reduced depends on hereditary versus environmental influences.

Important Definitions

Precision medicine

The term “precision medicine” refers to the practice of tailoring a patient's diagnosis and treatment to his or her specific set of symptoms, medical history, and genomic and other health data. Data science, data analytics, and data collection are indispensable to the practice of precision medicine. There are actual security, privacy, and integrity risks associated with the creation, storage, transmission, and analysis of precision medicine-required datasets. [8]

DNA Database

Due to its genomic foundation, precision medicine is highly reliant on genetic testing. There may or may not be laws protecting the privacy and security of this information, depending on who has access to these databases and why.

The HIPAA and state or local equivalents protect medical practitioners' diagnostic and treatment information. [9] 23andMe, Ancestry.com, and smartwatches that gather fitness data are exempt from privacy and data security rules. Genetic data collection, storage, and use are unregulated. Anyone with access to one of the many private DNA databases now publicly accessible and integrated with other genetic databases can identify DNA sample providers for ancestry and other purposes. [10]

Internet of Medical Things (IoMT)

Due to the requirement for large amounts of highly individualised data, precision medicine is appropriate for the “Internet of Medical Things” (IoMT). IoMT devices remotely monitor, diagnose, and treat patients. The Internet of Medical Things (IoMT) includes implantable cardio-defibrillators, pacemakers, and insulin pumps. Actuators, cloud storage, data analytics engines, wireless sensors, and transmitters are covered. [11]

Changes in Healthcare due to Onset of Personalised Medicine

Consider diagnostic procedures and treatments designed specifically for your illness and your body in order to obtain the most accurate picture. Everything in the modern medical system, including diagnostics, treatments, pharmaceuticals, and apparatus, is typically designed to “fit” the largest number of individuals conceivable. Included in this are treatment protocols, drug formulations, and device configurations. [12]

Receiving medical care in the modern era is more comparable to going clothing purchasing. If you give off-the-shelf solutions a chance, you can still use them as a jumping-off point for further investigation, even if they may not be a perfect match for your requirements. However, this is not necessarily always the case. Consider it akin to purchasing a custom-tailored suit that fits you perfectly in every way. Precision medicine refers to the practise of providing medical care that is

specifically tailored to the needs of a particular patient. [13]

With the use of precision medicine, both the quality of therapy and the results for each individual patient can be enhanced. Before a physician can determine the optimal treatment and pharmaceutical regimen for an individual patient, extensive trial and error is typically required. Utilising precision medicine is essential to resolving this issue. In precision medicine, a large quantity of diagnostic, historical, and patient data is analysed to determine the most effective treatment plan for each individual patient. No longer must you rely on trial and error or educated guesses to determine which candidates have the most pertinent experience and skills. [14]

This is of the uttermost importance in the treatment of rare diseases, cancer, and other chronic illnesses, where a "one-size-fits-all approach" would certainly impede the development of a potentially life-saving drug. Precision medicine seeks to illuminate the potential contributions of factors such as genetics, socioeconomic status, and other SDoH to disease diagnosis and treatment.

What implications does this have for other forms of medical facilities, such as hospitals? Reduce your expenses. If new diagnostic tests and analytics are utilised at an earlier stage, then the frequent back-and-forth communication and multiple office visits required to identify causes and develop solutions can be avoided. If the outcomes improve, there will be fewer patient visits, fewer complications, and fewer journeys to the emergency room. [15]

Precision Medicine driven by Data

Trastuzumab (Herceptin) is a highly effective treatment for patients whose cancerous tumours overexpress the HER-2 gene. In contrast, cetuximab (Erbitux) and panitumumab (Vectibix) for colon cancer are less effective in patients with an altered KRAS gene. A computerised analysis of genomic data can assist in the identification of genetic overlap between individuals with the same prognosis or treatment response. This can be an effective weapon in the fight against disease. Even though there may be tens of thousands of significant genomic measures (such as hundreds of thousands of RNA transcripts or millions of single point mutations), only a few thousand of these genomic events are typically sampled. Due to the vast number of variables relative to the number of samples [16], it is challenging to conduct statistics in this context. Data sharing is one of the most effective methods for increasing the total number of cohorts and maximising the use of each sample. As a result, the transmission of data is an essential aspect of the development of this discipline. Due to these restrictions, the currently available techniques for analysing genomic data are severely constrained. New statistical models and machine learning strategies must be developed to aid computational biologists, statisticians, and machine learners in analysing these datasets for insights into these challenges. As an illustration of the increased privacy dangers associated with the sharing of genetic information, we will conduct a GWAS, a specific type of genomic research. They are one of the most common methods used to identify genetic abnormalities associated with an observable trait, such as the presence of a disease or the response to treatment.

These investigations collect the variations exhibited by large populations containing hundreds of thousands to millions of SNPs. SNPs are genomic locations where single nucleotide mutations may originate. These variations in the genetic code are referred to as single nucleotide polymorphisms (SNPs). In addition, we maintain a unique identifier for each individual (this identifier may be discrete, such as their current state of health, or continuous, such as their age at the onset of symptoms). Then, statistical analyses are conducted to determine whether or not there is a correlation between SNPs and product quality. [17]

Because they do not require the control of potentially confounding factors such as age, gender, or environmental influences, the 2 test and the Cochran-Armitage test are extensively used when the trait is binary. This is due to the fact that they are frequently used when the attribute is binary. In addition, it is believed that individuals derive from a genetically stable population and that differences exist independently of one another, unaffected by other differences. Researchers in the disciplines of statistics and machine learning are constantly searching for novel methods to reduce the amount of rigour required by these assumptions in application settings. [18]

Patient Privacy

Even though there may be significant scientific and societal benefits to exchanging genetic data, extra caution is essential owing to the delicate nature of the material. In actuality, an examination of a person's genome sequence can provide information about their history, medicine metabolism, and susceptibility to sickness. All of these are aspects of one's life that the great majority of individuals choose to keep private. [19] Furthermore, genetic databases contain sensitive health information such as a person's current medical issues, medical history, and environmental factors (which may include substance abuse and trauma). This information is only accessible to authorised workers. In an era where data is widely referred to as "the new oil," safeguarding individual privacy is becoming increasingly important. This must be done without impeding any scientific, technical, or social improvements that may result from the analysis of large data sets.[20]
Because of the unique features of genetic information, it is vital to keep this in mind. Because our understanding of genetics

is always expanding, predicting what information may be retrieved from an individual's DNA ten years from now is challenging. In addition to revealing a person's personal history, genetic information may also reveal a person's linguistic, cultural, and ethnic origins. Because genetic information encompasses a person's whole family tree. DNA, unlike credit card numbers, cannot be altered after it has been extracted from a person's body. For more than three decades, people have been concerned about genetic discrimination. [21]

This refers to circumstances in which a person is treated differently because of their genetic make-up. In 1993, the Bilbao Declaration1 denounced genetic information for its potential discriminatory nature. Since then, a host of legislation have been enacted across the world to enforce these restrictions. Article 21 of the "EU Charter of Fundamental Rights" makes discrimination based on genetic characteristics prohibited. Employers and health insurers in the United States are prohibited from discriminating against individuals based on genetic information under the "Genetic Information Non-Discrimination Act" (GINA). GINA was first introduced in the banking industry, but it has now been expanded to include a broader range of enterprises in a number of jurisdictions, including California. GINA was initially only applicable to the banking industry. However, in Canada, genetic testing results were not deemed reliable until January 2018. [22]

Legislation that protects people' personal information and laws that ban discrimination based on a person's genetic composition are two options for preserving genomic privacy. "The Health Information Technology for Economic and Clinical Health" (HITECH) Act mandates data custodians in the United States to offer acceptable solutions in the areas of physical security, administrative management, and technical security in order to safeguard biological data. The newly formed General Data Protection Regulation (GDPR) of the European Union (EU) classifies genetic and biometric information as sensitive. This act aims to prevent discrimination based on, among other things, a person's genetic composition or present condition of health. This law is supplemented by the Network and Information Security Directive. This directive identifies health databases as a critical information technology system that requires additional security protections to prevent cyberattacks. Even though these limits have been implemented and there is no evidence that genetic discrimination is widespread, many people throughout the world are concerned about the privacy of genetic information.[23]

The law, however, does not protect persons from all types of genetic discrimination, particularly in personal relationships. This is true whether or not there is a law. Furthermore, just because all forms of bias are prohibited does not mean that they are never practised. As a result, ethical and regulatory safeguards must be included into the implementation of technological solutions. We will examine the most recent techniques, as well as mathematical and computational tools, that enable researchers to share genetic data without jeopardising patient privacy. Let us begin with the things that our anonymity will not protect us against. Then, employing a random number generator, we provide two mathematical models for preserving personal information, one that eliminates information and the other that generates noise. The last section of this lesson will go over several cryptographic hardware alternatives as well as fundamental data encryption procedures. [24]

AI and Big Health Data

Machine learning algorithms must have access to a large quantity of data in order to be successful. The more data artificial intelligence systems can analyse and analyse, the more accurate their projections and recommendations will be. 31 In the next paragraphs, we will give data demonstrating that adding additional information increases complexity. Due to time and financial constraints, physicians could only afford to collect the bare minimum of data in the past. The last three years have been extremely important for artificial intelligence because of the exponential increase in the amount of data generated and processed globally, as well as the significant decrease in the cost and time required to analyse this data. These variables have conspired to make recent years particularly significant.[25] The modern era's information gathering has advanced in a number of significant ways, the most significant of which are as follows: (i) there is now a significantly larger quantity of data available; and (ii) the breadth of information contained within this data has expanded significantly. The phrase "big data" is commonly used as an operational notion when seeking to define this movement. The use of big data has resulted in significant changes in the healthcare industry. [26]

Previously, the phrase "health data" solely referred to information gathered by medical specialists. However, in today's world, "health data" refers to any piece of information that can be extrapolated to reveal something about a person's health (including, but not limited to, data about physical, environmental, or biological aspects; data about social, economic, or individual status; data about lifestyle and commercial preferences; and so-called "big health data"). Furthermore, data can be collected from non-medical sources such as social networks, which collect information about people's preferences, interests, and contacts; smartphones, which can track people's movements, activities, and social interactions; "mHealth apps," which are intended to store health-related data and keep track of users' health and physical conditions; and Internet of Things (IoT) devices, which receive external input via sensors. Each of these gadgets is useful for gathering information. This means that algorithms trained on massive amounts of health data may be able to do more than just confirm hypotheses and aid in the diagnosis process; they may also be able to offer innovative hypotheses and provide prognoses before symptoms occur. [27]

Instead of causality, which has traditionally served as the ground for scientific inquiry, correlation acts as the foundation for these predictions. Developing a scientific frame of view used to entail first discovering the causal relationships between causes and effects. When dealing with large volumes of data, academics often employ the correlational technique. This type of output, which is based on statistical analysis, may and should be created by computers. In order to generate reliable predictions about the occurrence of health-related events, the data are linked in accordance with the patterns established in the training set. [28] This does not prove that the two are connected in any way that could be deemed causal. Because it has been difficult to establish a causal relationship between genetic variations and disease, genomics researchers have had to rely on correlation rather than causation. Correlation offers the benefits of speed, mechanisation, and low cost over other techniques to detecting causality that require a long time and a lot of labour. [29] Correlations between variables and variations are more likely to occur because, while correlations are frequently understood intuitively, providing an analytical explanation for them can be difficult. However, there are a few disadvantages to consider. The most difficult hurdle will be demonstrating that a specific alteration in a person's genetic composition enhances that person's likelihood of developing a certain disease. Furthermore, because there is no theory that can explain the prediction, the given result can only be used in the context that was presented. In conclusion, however this is not the end of the subject, when there is no stable foundation, the potential of errors and blunders increases. [30]

Finding the Balance: Personalised Medicine Data and Patients' Privacy

The European Union's Bill of Rights places a high value on the right to personal privacy. Furthermore, Articles 34 and 35 of the Charter provide the right to preventative health care as well as the right to medical treatment within the boundaries of national laws and traditions. These liberties are governed by country laws and norms. [31] The right to health care and social aid in times of illness, as well as the right to privacy, are recognised as essential individual rights in the founding text of the European Union. This is the case, notwithstanding the potential that social and public health interests are also at stake in this argument. In general, we do not place a high value on a privilege that is not accompanied by a comparable obligation. To actualize health rights, relevant duties must be accepted, resources must be made available, and the right itself must be monitored to ensure that it is upheld. Each member state will be responsible, within the framework of the European Union, for ensuring that its residents have appropriate access to medical care and other types of social support. Because their jurisdiction does not extend to this territory, the European Union and the European Commission have no control over it. They, on the other hand, have the intellect and power to choose the criteria that will be utilised to strike a balance between opposing interests. This is the purpose of the General Data Protection Regulation. [32]

Participation in data processing is priceless. Privacy must be proportionally balanced against other fundamental rights and the public good. Following the discussion of privacy, this guiding notion elegantly conveys the need to find an ethical middle ground between competing objectives, such as doing research and using genetic data for current and future patients. This concept weighs the immediate medical benefits of using genetic information against the potential long-term benefits for patients. This article addresses some of the GDPR's particular restrictions and what they may entail for genetic and other personal data-based scientific research. The proportionality principle underlines the need of considering both privacy concerns and the use of personal data for vital purposes such as research. Privacy safeguards and legitimate uses of personal data, such as scientific research, should be evaluated in a balanced manner.

To get insight into how scientific research and individual privacy should be balanced, review the case law of the European Court of Human Rights and the Court of Justice of the European Union. While these conditions exist, governments, authorities, ethical review bodies, and researchers must follow the core ethical values outlined and shown in GDPR. It is critical to remember that the great majority of researchers are law-abiding people who would never wilfully breach the law, such as by suing a government entity over its management of personal information or its informed consent methods. In the framework of academic inquiry, the goal of this study is to provide a basis that might serve as the cornerstone for GDPR nationwide implementation.

REFERENCES

[1] Karoliina Snell, "*Health as the Moral Principle of Post-Genomic Society: Data-Driven Arguments against Privacy and Autonomy*", 28 CAMBRIDGE Q. HEALTHCARE ETHICS 201 (2019).

[2] HANSSON, M.G. "*Striking a Balance Between Personalised Genetics and Privacy Protection from the Perspective of GDPR*". in: "SLOKENBERGA, S., TZORTZATOU, O., REICHEL, J. (EDS) "GDPR AND BIOBANKING. LAW, GOVERNANCE AND TECHNOLOGY SERIES", vol 43. (Springer, Cham, 2021). https://doi.org/10.1007/978-3-030-49388-2_3.

[3] Hansson MG, Dillner J, Bartram CR, Carlsson J, Helgesson G, "*Should donors be allowed to give broad consent to future biobank research?*" Lancet Oncol 7:266–269 (2006).

[4] T Graeme Mary and Dixon-Woods, "The Social Licence for Research: Why Care.Data Ran into Trouble".

[5] Kanu Song, "*Warning Third Parties of Genetic Risks in the Era of Personalized Medicine*", 49 U.C.D. L. REV. 1987 (2016).

[6] Leroy Hood and Mauricio Flores, "*A Personal View on Systems Medicine and the Emergence of Proactive P4 Medicine: Predictive, Preventive, Personalized and Participatory*"
[7] Horgan, Denis, Mario Romao, Servaas A. Morré, and Dipak Kalra. "*Artificial Intelligence: Power for Civilisation – and for Better Healthcare*." PUBLIC HEALTH GENOMICS 22, no. 5/6 (2019): 145–61. https://www.jstor.org/stable/27012984.
[8] Mark D. Rasch, "*Privacy and Security: The World Precision Medicine*" GPSOLO MAGAZINE (August 2021), https://www.americanbar.org/groups/gpsolo/publications/gp_solo/2021/july-august/privacy-and-security-the-world-precision-medicine/.
[9] Mark D. Rasch, "*Privacy and Security: The World Precision Medicine*" GPSOLO MAGAZINE (August 2021), https://www.americanbar.org/groups/gpsolo/publications/gp_solo/2021/july-august/privacy-and-security-the-world-precision-medicine/.
[10] Chiara Rauccio, "*Artificial Intelligence and Genomics: The Data Protection Implications in the Use of AI for Genomic Diagonstics*", 2021 EUR. J. PRIVACY L. & TECH. 115 (2021).
[11] Jacob S. Sherkow & Jorge L. Contreras, *Intellectual Property, Surrogate Licensing, and Precision Medicine*, 7 IP THEORY 1 (2017).
[12] Sabatello, Maya, and Paul S. Appelbaum. "*The Precision Medicine Nation*." THE HASTINGS CENTER REPORT 47, no. 4 (2017): 19–29. http://www.jstor.org/stable/26625159.
[13] Mählmann, Laura, Matthias Reumann, Nikolaos Evangelatos, and Angela Brand. "*Big Data for Public Health Policy-Making: Policy Empowerment*." PUBLIC HEALTH GENOMICS 20, no. 6 (2017): 312–20. https://www.jstor.org/stable/26778352.
[14] Philippe A, Georges D (eds) "*A history of private life*", vol I–V. The Belknap Press of Harvard University Press, Cambridge (1989).
[15] O'Donnel PH, Ratain MJ Germline pharmacogenomics in oncology: decoding the patient for targeting therapy. Mol Oncol 6(2):251–259 (2012).
[16] "Title II of the Genetic Information Non-discrimination Act", 2008 (GINA); 2008.
[17] Genetic Non-Discrimination Act—Parliament of Canada; 2017. s-201.
[18] Donoho DL, "*High-dimensional data analysis: the curses and blessings of dimensionality*" In "AMS CONF. ON MATH CHALLENGES OF THE 21ST CENTURY", Los Angeles, CA, (7–12 August 2000) https://statweb.stanford.edu/∼donoho/Lectures/AMS2000/AMS2000.html.
[19] Dankar FK, El Emam K. 2012 "*The application of differential privacy to health data*" In Proc. 2012 Joint EDBT/ICDT Workshops (EDBT-ICDT '12), Berlin, Germany, 26–30 March 2012, pp. 158–166. New York, NY: ACM.
[20] Ball MP et al. 2012 "A public resource facilitating clinical use of genomes" Proc. Natl Acad. Sci. USA 109, 11 920–11 927. (doi:10.1073/pnas.1201904109)
[21] Hansson MG, Dillner J, Bartram CR, Carlsson J, Helgesson G, "*Should donors be allowed to give broad consent to future biobank research?*" Lancet Oncol 7:266–269 (2006).
[22] Niemiec E, Howard HC. 2016 "Ethical issues in consumer genome sequencing: use of consumers" samples and data. Appl. Transl. Genomics 8, 23–30. (doi:10.1016/j.atg.2016. 01.005)
[23] Regulation (EU) 2016/679 of the European Parliament and of the Council of 27 April 2016; 2016.
[24] NIH. "Policy for sharing of data obtained in NIH supported or conducted Genome-Wide Association Studies" (GWAS); 2007.
[25] WN Price, IG Cohen, "Privacy in the Age of Medical Big Data" (2019) 25 NATURE MEDICINE.
[26] S Hoffman, "*Big Data's New Discrimination Threats Amending the Americans with Disabilities Act to Cover Discrimination Based on Data-Driven Predictions of Future Disease*" in G Cohen, A Hoffman and W Sage (eds.), 'BIG DATA, HEALTH LAW, AND BIOETHICS' (Cambridge University Press, 2017).
[27] TZ Zarsky, "*Incompatible: The GDPR in the Age of Big Data*" (2017) 47 SETON HALL LAW REVIEW.
[28] A Romei, S Ruggieri, "*A Multidisciplinary Survey on Discrimination Analysis*" (2014) 29 The Knowledge Engineering Review, 582
[29] D Schönberger, "*Artificial intelligence in healthcare: a critical analysis of the legal and ethical implications*" (2019) 27 INTERNATIONAL JOURNAL OF LAW AND INFORMATION TECHNOLOGY 171–203.
[30] WN Price II, "Black-Box Medicine" (2014) 28 Harvard Journal of Law & Technology, 419.
[31] B Mittelstadt, "*From Individual to Group Privacy in Biomedical Big*" in G Cohen, A Hoffman and W Sage (eds.), Big Data, Health Law, and Bioethics (Cambridge University Press, 2017).
[32] O'Donnel PH, Ratain MJ, "Germline pharmacogenomics in oncology: decoding the patient for targeting therapy" MOL ONCOL 6(2):251–259 (2012).