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Omics in Society

Social, Legal and Ethical aspects of Human Genomics

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"The genomics revolution, proteomics, metabolomics, all of these 'omics' that sound so terrific on grants and on business plans. What we're doing is we are seizing control of our evolutionary future. I mean we're essentially using technology to just jam evolution into fast-forward" (Stock, 2003)

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List of abbreviations

ACMG	American College of Medical Genetics and Genomics
AI	Artificial Intelligence
APW	Associated Press in Washington
ARRIGE	Association for Responsible Research and Innovation in Genome Editing
ASHG	American Society of Human Genetics
ATCG	Adenine, Thymine, Cytosine and Guanine
BBC	British Broadcasting Corporation
BMJV	Bundesministerium der Justiz und für Verbraucherschutz
BVL	Bundesamt für Verbraucherschutz und Lebensmittelsicherheit
Cas9	CRISPR associated protein 9
CCMG	Canadian College of Medical Geneticists
DG	Directorate-General
CRISPR	Clustered Regularly Interspaced Short Palindromic Repeats
DIYbio	Do-It-Yourself Biology
DTC-GT	Direct-to-Consumer Genetic Testing
DNA	Deoxyribonucleic acid
EASAC	European Academies Science Advisory Council
EC	European Commission
ESHG	European Society of Human Genetics
ESHRE	European Society of Human Reproduction and Embryology
EU	European Union
GMO	Genetically Modified Organisms
HFEA	UK Human Fertilisation and Embryology Authority
ISSCR	International Society for Stem Cell Research
JRC	Joint Research Centre
NASEM	National Academy of Sciences Engineering Medicine
NIH	USA National Institutes of Health
OECD	Organisation for Economic Co-operation and Development
RCIGM	Rady Children's Institute for Genomic Medicine
UK	United Kingdom
UNCED	United Nations Conference on Environment and Development
UNESCO	United Nations Educational, Scientific and Cultural Organization
USA	United States of America
WHA	World Health Assembly
WHO	World Health Organization

Summary

This report was produced by the Foresight, Behavioural Insights & Design for Policy Unit of Joint Research Centre (JRC) under the initiative towards enhanced research in the area of "Omics in Society with a focus on Genomics". The work presented tries to address the main research areas in line with current European Commission (EC) policy priorities for foresight and citizen engagement planning. Through an extensive review of literature, corporate and media discourses, as well as do-it-yourself bio movement's Internet sites, several thematic narratives have been identified. These narratives come from different actors telling about on-going promises, interests, expectations and concerns across the human genomics field.

The present report maps also key players working in the human genomics field around the world, identifying the most expressive or emblematic companies. We observe co-existing narratives across the analysed companies' discourses in particular in relation to the claims and promises associated to the technological advances in human genomics. Attractive narratives are offered to citizens, appealing to personal *needs* and *interests* (e.g. ancestry, genetic make-up, or genetic risks), and often overlooking ethical considerations. The literature and other sources covered in the report suggest that the human genomics field appears to be strongly consolidated in North America, with a high number of institutes and companies operating in the field. The human genomics field also appears strongly connected to big data, artificial intelligence (A.I.) and blockchain technologies debates. This has particularly gained momentum due to the involvement of tech giants such as Google, Amazon and Microsoft.

We have identified relevant EU legislation and institutions in the genomics field and evidence suggests that the European Union lacks a coordinated and uniform regulation in this domain, in particular in relation to human genomics. Combined with an inability to accompany rapid advances of scientific fields, this sets a scenery of grey areas in the legislation that can be potentially exploited by practically anyone - be it companies, academia, or individuals. In fact, as we were finalising this report, Chinese scientist He Jiankui claimed he had produced the first babies with an edited genome.

The analysis of major ethical concerns on the human genomics field urges the need of inclusion of non-scientific groups into the ethical debate, as well as the need to address the complete non-compliance of any international guidelines by Direct-to-Consumer Genetic Testing (DTC-GT) companies, the fuzziness surrounding DTC-GT business model, and concerns surrounding confidentiality security in the age of *-omics* with possible social repercussions. Similarly, the lack of consistent ethical guidance on DIYbio needs attention.

A boost in the presence of genomics related topics in the news media is also evident, reflecting to a certain extent an upsurge of reports of *optimistic portrayal* that can lead to misleading and misinformed enthusiasm. The importance and influence of new media and social media is a major concern that needs to be addressed in the evaluation of information's quality and impact in the public.

Finally, this report does not present an exhaustive evaluation of citizen engagement on the social or ethical impacts potentially arising from the developments in the human genomics field, but it identifies common problematics transversal to the studies included in our analysis. We can say that not many citizen engagement activities about human genomics were found, suggesting that there is a need for the creation of dialogue spaces about this technology and its potential applications. However, it offers an updated mapping of DIYbio communities and activities, illustrating the growth of this type of grassroots engagement

activities. The report informs the next steps of implementation of citizen engagement activities in the human genomic field.

1. Introduction

1.1 Purpose of this report

The present report was produced by the Foresight, Behavioural Insights & Design for Policy Unit of Joint Research Centre (JRC) under the initiative towards enhanced research effort in the area of "*Omics in Society with a focus on Genomics*". The JRC initiative proposes to address four main research areas which are in line with current European Commission (EC) policy priorities:

Omics: A neologism for the constellation of an organism's "-omic" information, which includes the genome itself (genomic), transcription products (transcriptomic), protein products (proteomic) and metabolic products (metabolomic).

Genomics: Interdisciplinary field of biology focused on the study of the structure, function and expression of all the genes in an organism. Genomics aims to understand the organism's complete set of DNA (i.e. the Genome), including the mapping/sequencing of genes.

- The impact of the already wide-scale human genome sequencing and how it will influence health and sociological concepts and thinking.
- Better understanding of the environmental factors that affect the genetic predisposition and how it will ultimately determine the health status and well-being of humans, but also the physiology and safety of the food we consume (studied in-depth in the "*epigenomics*" project).
- Development of a program on biomarkers and biomonitoring to better estimate the exposure of the human body to external factors and thus to assess the associated risk factors.
- Assessment of the ethics of genomics technology and its applications in order to assure that the innovative outcomes of new technologies are fully respecting the individual's integrity.

The work presented in this report is based on an extensive literature review and starts with a mapping of actors, with special focus on companies operating in the human genomics field (**section 2**). *What are the promises, claims, and assumptions in their narratives by these companies in order to attract customers?* A better understanding of these narratives allows future citizen engagement initiatives to capture and extract the matter of concern of citizens in relation to businesses operating in the area of genomics.

Sections 3 and **4** of this report address, respectively, the legal and ethical discourses that are currently taking place in our society. The inherent complexity and different types of uncertainty in human genomics demonstrates the relevance of opening the debate to all actors of concern, including citizens. *What are the expectations and imaginaries from citizens in relation to the applications and future impact of human genomics? Do they think they will have access to these technologies? How do they envision their life in a world where human genomics is open and freely used?*

Next, **section 5** is dedicated to the media. *What were the main highlights about genomics in news media? What is the impact of social media on shaping our views and opinions on topics related to gene editing or CRISPR? Do movies and TV series also have a role in informing societies about gene editing technologies?*

Based on the aforementioned sections, a selection of possible future narratives in genomics were extracted (**section 6**). New narratives in human genomics were also explored to account with possible scientific and

technological advances in the genomics field. The report ends with a review of citizen engagement initiatives in this topic (**section 7**).

1.2 Setting the scene

The World Health Organization (WHO) defines genomics as the study of genes, their functions and related techniques, in order to understand the interrelations and combined influence of genes on the growth and development of the organism (WHO, 2002, 2004). As a result of the research that has been done during the last decades we are, nowadays, not only able to identify and understand how genes behave but also, able to manipulate genes in order to attempt to reverse or stop certain diseases. The latest achievements were largely facilitated by the use of sequencing technologies that provided access to the full genetic information of an organism within 20 hours and by the development of the CRISPR/Cas9 technology which provides a faster, cheaper, more accurate and efficient gene editing methodology than any previous technique (Cox, Platt, & Zhang, 2015; Krigel, 2018; Marchione, 2017; Qasim et al., 2017; Rady Children's Hospital, 2018).

Gene editing: Group of technologies that give scientists the ability to change specific DNA sequences by allowing genetic material to be added, removed, or altered in the genome (Merriam-Webster, 2018a).

CRISPR/Cas9 stands for '*Clustered Regularly Interspaced Short Palindromic Repeats*' and it is part of the defence mechanism found in the immune system of bacteria against viruses. After its discovery it was used as part of the CRISP-Cas9 genome editing technology which allows scientists to edit, cut or replace DNA at precise locations. This technology can be applied to permanently modify genes in living cells and organisms with the aim to correct mutations and treat disease.

The aforementioned technological achievements have also made human genomics affordable and accessible to the public which means that not only the advancement of science and biomedical innovation has been prompted, but it also leads to:

- Pharmaceutical companies offering gene therapy services using gene editing techniques.
- Direct-to-Consumer genetic testing services.
- Businesses working in the interface of genomics and other technologies such as, artificial intelligence, cloud computing and blockchain.
- Social movements such as the Do-It-Yourself Biology (DIYbio).

Together with these newly emerged actors, a number of legal, ethical and social implications also surfaced. In terms of regulatory discourses, there is a global uncertainty about the legislation that will regulate gene edited organisms, DTC-GT or DIYbio activities. Regarding ethical concerns, our findings point out to the most pressing concerns being related to *(1)* genetic modification of human embryos; *(2)* privacy and ownership of genetic data; *(3)* genetic counselling performed by professionals; and *(4)* animal ethics in case of usage during DIYbio experiments. Although several ethical committees produced statements or reports about these topics, little has been incorporated into the creation of policies or used in engagement with citizens. To this, we can also add the recent research papers demonstrating that CRISPR/CAS9 technique is not as efficient as it was initially thought and some collateral damage can be made by using it, such as promoting risk to develop cancer or deleting unwanted regions of DNA (Haapaniemi, Botla, Persson, Schmierer, & Taipale, 2018; Ledford, 2018; Richardson *et al.*, 2018).

1.3 The case of DIYbio

The Do-It-Yourself biology (DIYbio) movement has been gaining a great deal of attention from academia, policy makers and the media in the last years (Seyfried, Pei, & Schmidt, 2014). The interest and relevance within the human genomics debate is primarily due to the regulatory and ethical discourses (see sections 3.3 and 4.3). Despite the public narrative around DIYbio potential of democratisation, education, and community building for biotechnology, the movement faces a widespread concern from policy makers, journalists and the overall public regarding its safety procedures and security monitoring (Nascimento, Guimarães Pereira, Ghezzi, Guimaraes Pereira, & Ghezzi, 2014). Our particular interest in the DIYbio movement in the context of this report goes further that the intrinsic ethical and regulatory discourses. It falls also towards the fact that DIYbio has become a deep form of engagement with genomics for civil society.

The term DIYbio is usually used to refer to the experimental manipulation of genetic material for any number of purposes in non-traditional academic and industrial settings influenced by open source principles. Hence, DIYbio is mostly administered without an official and certified supervision and is coordinated by supporters and followers of the movement.

The birth of the DIYbio movement can be traced back to 2005, when Rob Carlson first mounted a bio laboratory on his own garage (Carlson, 2005; Ledford, 2010). Three year later, in 2008, Jason Bobe and Mackenzie Cowell founded one of the largest online DIYbio communities, DIYbio.org, with the mission of establishing a vibrant, productive and safe community of DIY biologists. The community rapidly expanded, and nowadays counts with 106 groups listed in their website (DIYbio, n.d.-b). Because of the openness to the public, the DIYbio movement gathers a plethora of enthusiasts from professionals with different backgrounds and varying levels of expertise to people with only an interest in biology. Whilst the majority of the DIYbio groups and practitioners are not conducting world changing experiments and pushing the boundaries of molecular biology, there are cases of sophisticated experiments in these nonconventional settings, many involving DNA profiling and genetic engineering, creating fluorescent bacteria for artistic purposes (Jorgensen, 2016). Others groups are more interested in creating standard instruments, such as PCR machines cheaper and easier to use outside the confines of a laboratory, ultimately promising to make DIYbio more accessible (Seyfried *et al.*, 2014). DIYbio has also merged with citizen science programs. Examples of collaborations can be seen in the DNA Barcoding Project in Hong Kong which intends to sample, identify and monitor the local biodiversity with the help of the community (Chan, 2016).

The DIYbio movement is intensely connected to the open-science[1] movement in the sense that it encourages an open exchange of data and equipment. It also brings together various fields from science, technology and innovation, to entrepreneurship, activism, education, and arts. Furthermore, nowadays, the DIYbio movement is as well a driving force within the open source movement. This was made possible mainly due to the following factors:

[1] Open Science: transparent and accessible knowledge that is shared and developed through collaborative networks.

- The continuous growth of the Maker Movement[2] (see Rosa *et al.*, 2017).
- A higher access to information and technological protocols which were provided mainly by platforms such as DIYbio.org and DIYbiosphere[3].
- The accessibility and affordability of technological equipment and biological reagents.

[2] Maker movement: Grassroots movement with a strong DIY approach mostly applied to emerging personal fabrication technologies such as 3D printing and laser cutting. It promotes distributed access to information across individuals of the same community and across different communities
[3] https://diybio.org/; http://sphere.diybio.org/

2. Human Genomics around the World: Who is who?

2.1 Companies

According to the report *Global Genomics Market* by Research and Markets (2017), the genomic business is an ever expanding market, expected to reach USD 23.88 Billion by 2022 from USD 14.71 Billion in 2017. The main companies in the sector primarily target the academia and research institutions sector but gradually are expanding their market by including in their scope of clients also small businesses, star-ups and the general public. Nowadays, we have business offering not only equipment and reagents for research but also providing gene therapy services, genetic testing for individuals and kits for DIYbio. Human genomics has also awakened the interest of companies working with blockchain, cloud computing, data mining and artificial intelligence. Various companies are also investigating into solutions to big human genomics challenges, such as the storage, analysis and management of small and large-scale genomic data or securing genomic data trading and its ownership.

As the genomics business grows and matures, it becomes necessary to understand who the key players are, where they are located, how the products/services are presented to the public, and what the social or ethical implications arising from these emerging businesses are. In order to answer to these questions, we first undertook the exercise of mapping different companies and research institutions working on genomics around the world, based on the information available from three sources: the *Genetic Testing Registry* website[4], the *Biopharmguy* website[5], and the *International Society of Genetic Genealogy* wiki[6]. After a detailed analysis of the companies referenced in these sources, a final selection of 653 organisations was documented, and visually represented in a world map based on their geographical location (Figure 1). Additional information of the geographical panorama of genomics companies is provided in Figure 2 and Figure 3, where it is illustrated the number of genomic companies and research institutes by geographical area, and by country, respectively. It must be pointed out that the data were retrieved from the mentioned sources during July-August 2018 and, consequently, it must be seen as a snapshot for that period of time. Moreover, it is not possible to guarantee the accuracy of every piece of information as the data come from self–reported online sources. As a second step, newly emerging companies and businesses are described based on the type of service offered and the discourses used to attract new customers (sections 2.1.1 to 2.1.8). The companies highlighted provide a picture of the variety and diversity of services available.

[4] https://www.ncbi.nlm.nih.gov/gtr/
[5] https://biopharmguy.com/
[6] https://isogg.org/wiki/List_of_DNA_testing_companies

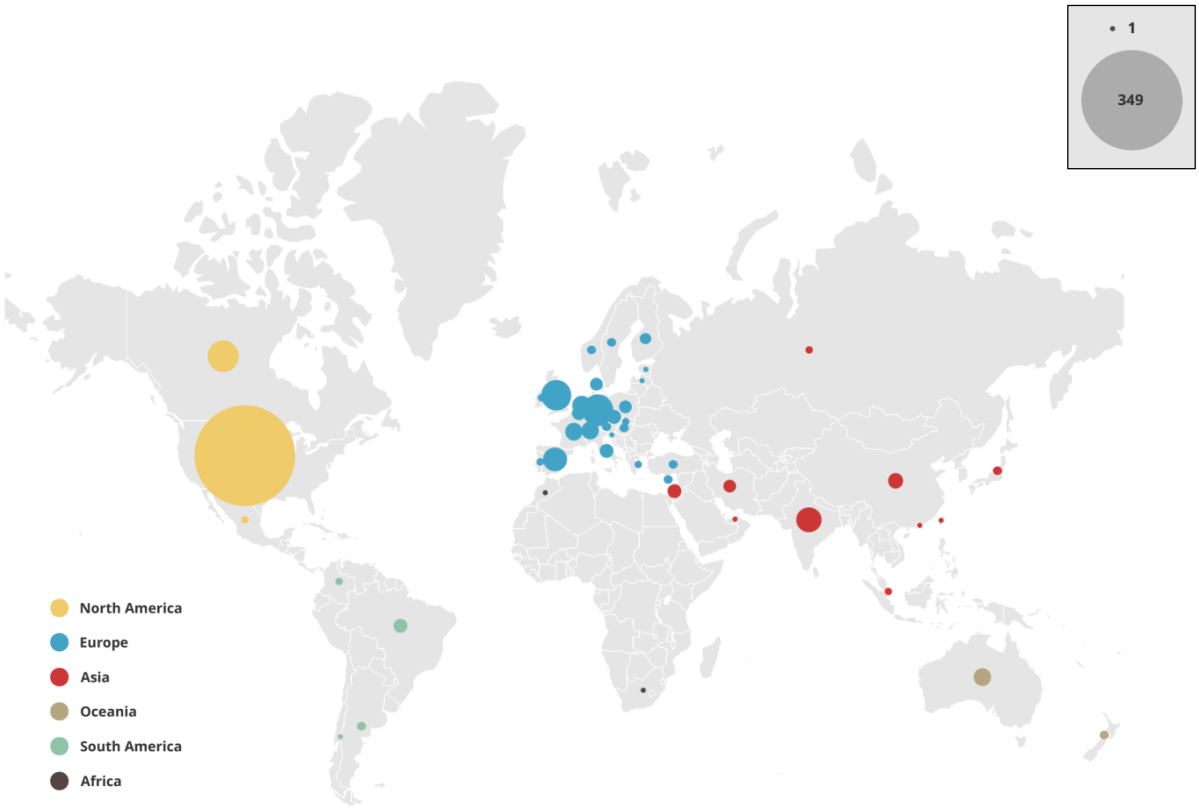

Figure 1: World map of genomic companies and research institutes (year 2018).

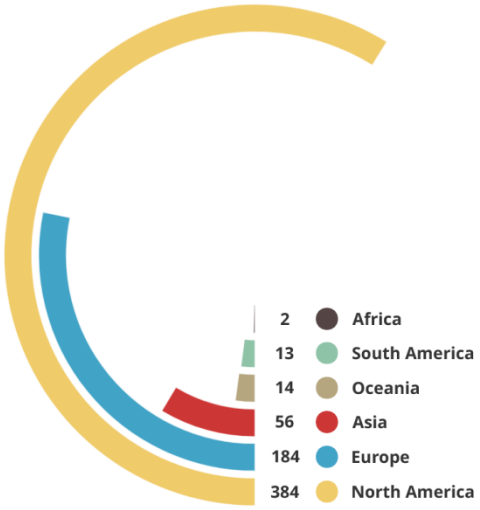

Figure 2: Number of genomic companies and research institutes by geographical area (year 2018).

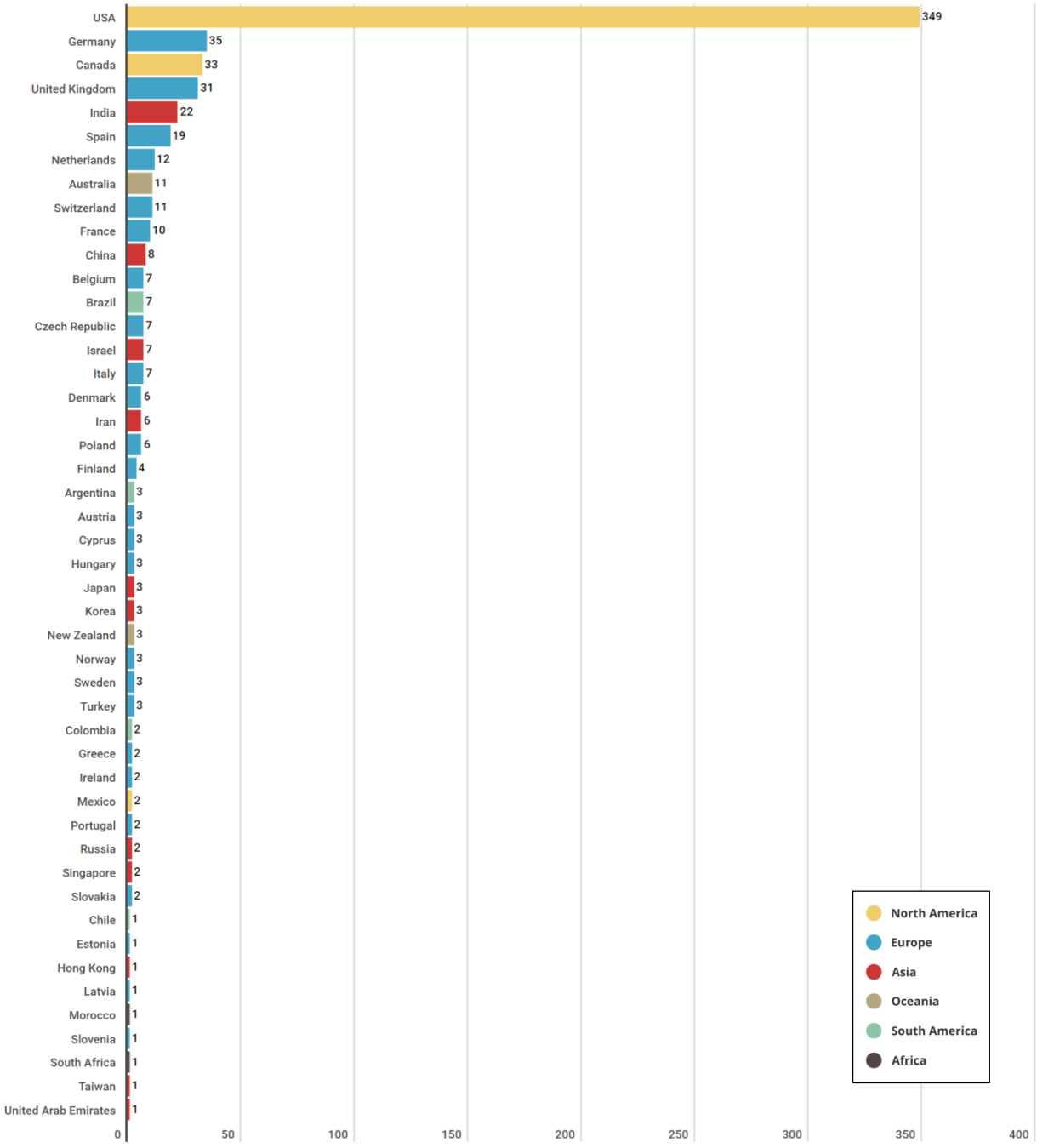

Figure 3: Number of genomic companies and research institutes by country (year 2018).

Table 1: Data quality box for Figure 1, Figure 2 and Figure 3.

Data Source	– *Genetic Testing Registry* website. – *Biopharmguy* website. – *International Society of Genetic Genealogy* wiki.
Data Year	Data retrieved from original source in July 2018.
Data Processing	– Data cleaned for duplicate entries. – Data grouped into geographic areas.
Data Type	Numeric (integer).
Data Items	Data from 653 unique spaces.

2.1.1 Companies that sell material for academic research

In terms of sequencing technologies, *Illumina*[7] (USA) is one of the leading global companies in selling life science tools and integrated systems for large-scale analysis of genetic variation and function, providing cheaper DNA sequencing analysis for detection of mutations in different genes in a wide range of areas from cancer research to agriculture. Their narrative includes for example, from producing agricultural products that were not possible to be cultivated before, extending the live spam of patients in order to make personalised healthcare a reality. *Illumina* uses the slogan "(We) *improve human health by unlocking the power of the genome*" (Illumina, 2018) which begs the question "*does the customer wonder how genomics can transform their own lives and control their own future?*".

Sequencing technologies: since the discovery of the double helix structure of DNA in 1953 by Watson and Crick it was learnt that DNA was formed by four molecular bases (A-T-C-G) and this was the code that contained all the information about our genome and the genome of any specie. Since this publication many technologies have aimed to read the genome of humans and other species in a faster, cheaper and more efficient way with the aim to understand and decrypt the code to all biological life. Sequencing technologies have been used to characterize pathogens, identify mutations that cause disease, guide the diagnosis or/and treatment of cancer, monitor pathogen outbreaks and more recently for recreational purposes such as heritage assessing.

Similarly, *Nanopore Technologies*[8] (UK, USA) is also focused on sequencing equipment and analysis of any type of biological sample. *GeneScript*[9] (USA, China, and Japan) offers multiple kits to assemble your own Cas9 constructs, as well as purified Cas9 enzymes for *in vitro* and *in vivo* use. *Integrated DNA technologies*[10] (USA) offer new variations of CRISPR to outperform current technologies and to open up CRISPR editing to additional areas in genomes. *Cellectics*[11] (France, USA) design next generation immunotherapies based on gene-edited CAR T-cells.

[7] https://www.illumina.com/
[8] https://nanoporetech.com/
[9] https://www.genscript.com/crispr-products.html
[10] https://eu.idtdna.com/pages/products/crispr-genome-editing
[11] http://www.cellectis.com/

2.1.2 Companies that offer gene therapy services

Among the companies offering CRISPR technologies we have *Intellia Therapeutics*[12] (USA) which although their products are currently at the pre-clinical stage, they motivate investors by stating that conventional medicines only treat the symptoms but not the genetic cause, therefore, *Intellia Therapeutics* is "*revolutionizing medicine by harnessing the power of genome editing*" (Intellia Therapeutics, 2018).

> **Gene therapy**: Experimental technique that allows the replacement of a mutated gene (that causes disease) with a healthy copy of the gene. It also allows to inactivate or "knocking out" a mutated gene that is functioning improperly by introducing a new gene into the body to help fight a disease (NIH, 2018a).

Another company, *Editas Medicine*[13] (USA), mentions that finally now "*a new technology known as CRISPR (Clustered Regularly Interspaced Short Palindromic Repeats) has the potential to achieve accurate, directed changes in DNA and fulfil the promise that started with the sequencing of the human genome – the potential to treat diseases at their source, at the DNA level*" (Editas Medicine, 2018). Their current pipeline is at the very beginning (discovery phase).

CRISPR Therapeutics[14] (Switzerland, USA, and UK) sells itself as a company that will develop new and transformative gene-based medicines that will cure diseases at the molecular level. The company underlines that they follow the recommendations of the *International Society for Stem Cell Research* (ISSCR, 2015) and thus their focus is only on the treatment of somatic cells and not in germ cells, which are inheritable. *CRISPR Therapeutics* also states that they work closely with patients and families, healthcare professionals, regulatory agencies and other groups dedicated to improving healthcare. Together with *Intellia Therapeutics, CRISPR Therapeutics* has released a joint statement on their position regarding human germline gene editing. In this statement, they "[refrain] *from directly modifying germline cells, including sperm, egg or embryonic tissue, or developing any clinical applications of germline gene editing*" (Intellia & CRISPR Therapeutics, 2015, p. 2).

2.1.3 Companies that offer direct-to-consumer genetic testing

The direct-to-consumer genetic testing (DTC-GT) has become a blooming business in the last years. It provides people access to their genetic information without necessarily involving a healthcare provider or health insurance company in the process. The most popular tests use genetic variations to make predictions about health, provide information about common traits, and offer clues about a person's ancestry (NIH, 2018b, 2018a). Companies such as *Consyl*[15] (USA) offer genetic screening to identify couples at risk to pass down inherited conditions (around 175) to their children. In addition, they also offer prenatal and cancer genetic screening. Their main target audiences are women under the idea that they are the responsible ones of their families' fate by accessing "*early awareness about genetic conditions, so they can live informed and prepare for the future*" (Counsyl, 2017).

Futura Genetics[16] (Canada with offices in Middle East and Europe) offers individuals the service to screen one's DNA against 28 common conditions such as, Alzheimer, melanoma, migraine obesity, and prostate cancer. A final report sent to the patient specifies a number of behaviours that should be avoided in order

[12] https://www.intelliatx.com/overview/
[13] http://www.editasmedicine.com/
[14] http://crisprtx.com/
[15] https://www.counsyl.com/
[16] https://www.futuragenetics.com/en/

to live a healthier life. *Veritas Genetics*[17] (USA, China) offers similar information. They state that they help individuals making lifestyle adjustments in order to potentially reduce genetic risks. Their service includes the joint creation of a plan with the individual's doctor, to keep him/her informed of his/her genetically-influenced response to certain medications. The company makes the link between the genetic data they offer to the clients and their medical practitioner, who together with the client's data will, in principle, make "smarter" choices about the client's health.

Global Gene Corp[18] (Singapore, India, and UK) was created to "[democratise] *genomic studies to positively disrupt health outcomes for everyone*"(Global Gene Corp, 2018). According to this company, diverse populations hold different mutations however most genomic data and research are based on samples originating from Western countries. Hence, their research is focused on extracting genetic data from other populations around the world, starting on individuals from India and South Asia.

Another growing market in the realm of direct-to-consumer genetic testing is the business of offering personalised nutrition, fitness and beauty creams tailored to your DNA. *Pathway Genomics*[19] (USA) is one of the leading companies in this field. Their argument goes towards uncovering the hidden information coded in the DNA in order to take control of your personal journey to a different lifestyle: "*Knowledge is power and nothing knows you better than your DNA*" (Pathway Genomics, 2018).

MyDNA[20] (Australia) offers personalised counselling on which type of medication, diet, sport and exercises one should do depending on the DNA analyses. In this case, the company provides a report on the seven genes analysed per category (diet and sports). *MyDNA* also offers corporate wellness packages where through the genetic insights of employees, companies can empower them on a truly personalised level (beyond team building) by motivating them to have a healthier lifestyle and by increasing engagement "*by showing you care about their well-being*" (myDNA, 2018).

DNAFit[21] (UK) is another company operating in the area of fitness tailored to your DNA. The company recently started to include professional football players (namely former England national team footballer Rio Ferdinand) as part of their online campaign to compare the client's DNA to the professional football player and see how their endurance, production of muscle fibres, levels of inflammation after a hard training sessions (among others) are suitable to play football or another sport, and how the client can improve their nutrition in order to achieve the maximum efficiency from workouts.

The *Superhero*[22] genetic test from *ORIG3N* proposes their customers to unravel their "*unique superhero traits*", such as above-average intelligence, speed and strength (Orig3n, 2018). The *Behavior*[23] test, also from *ORIG3N*, offers customers the possibility to understand the role their DNA may play in their personality (e.g. tolerance to stress and pain, impulsive behaviour and feelings of empathy and euphoria). *LifeDNA*[24] goes a step further in a different field, and provide cosmetics that are supposedly tailored to your DNA results.

[17] https://www.veritasgenetics.com/
[18] https://globalgenecorp.com/
[19] https://www.pathway.com/
[20] https://www.mydna.life/
[21] https://www.dnafit.com/
[22] https://shop.orig3n.com/products/superhero
[23] https://shop.orig3n.com/products/behavior
[24] https://www.lifedna.com/

AfricanAncestry[25] (USA) provides genetic screening services aimed at determining individuals' African ancestry, namely their country and ethnic group of origin. The company attracts customers by claiming to hold the largest database of African lineages and ethnic groups. In the same topic, the *Genographic Project*[26]*,* an initiative from *National Geographic,* also offer ancestry assessments based on one's DNA. The *Genographic Project* is advertised as part of a wider scientific study to better understand how our ancestor migrated from Africa to populate the world, and how each one of us can be involved on this study as a citizen scientist.

23andMe[27] (USA) is probably the most widely known company in the area of genetic screening for unravelling ancestry. The company provides their costumers the possibility to know their ancestry, broken-down by region and by how much of a Neanderthal inherited. It also offers the possibility to find relatives by connecting the client's data with other costumers that share DNA with them. This last product is mostly sold as recreational due to its easy to read reports, where they can tell how much percentage of DNA the client shares with near relatives (e.g. father, cousin, sister, etc.). Since women can only trace their maternal haplogroup (mitochondrial DNA) and not their paternal (due to lack of Y chromosome), it is suggested for females customers to encourage their brothers to also take the test so that it can be inferred the paternal ancestors from the results. Worth noticing that initially *23andMe* also offered testing of 254 health related conditions, including heart disease, breast cancer and diabetes, however the U.S. Food and Drug Administration (FDA) suspended these tests on legal basis (MacKenzie, 2013). Since then, *23andMe* has been allowed to prove genetic reports for only 10 health conditions (Kolata, 2017).

2.1.4 Companies that sell kits for DIYbio

Amino Lab[28] (Canada) is a DIYbio start-up company with the aim of making genetic engineering accessible to everyone. The company argues that they make science "*fun, accessible and affordable*" by teaching their customers, primarily kids and teenagers, how to perform biotechnological experiments such as genetic engineering, as easy as "*learning maths, programming or even cooking*" (Amino Labs, n.d.). Their products include kits for engineering and growing bacteria that fluoresce in the dark or produce pigments, or as *ink* to do art on a *petri* dish. *Amino Lab* complements their kits with pay online courses, where they teach costumers how to use the kits proficiently.

DIYbio (Do-It-Yourself Biology): Citizen science-driven movement that aims to find innovative solutions by studying life sciences related topics in non-traditional academic and industrial settings, namely in self-made laboratories (e.g. assembled in kitchens and garages) that are not ruled by the policies of a research or academic institution.

The ODIN[29] (USA) is another example of a DIYbio company with the aim of making genetic engineering accessible to everyone. However, in this case, *The ODIN* market and target audience is mostly adults. Josiah Zayner, CEO of *The ODIN,* is extensively known in the DIYbio arena for his YouTube channel and videos on genetic engineering and biohacking (which is also the main source of advertisement for the company). Zayner videos promote the idea that any person can "*genetically engineer a human on their own garages*" or that you can "*create CRISPR DNA to target your genes*" (Zayner, n.d.). *The ODIN* sells kits that, for example, allow customers to genetically engineer yeast to fluoresce, or advocate experiments using frogs.

[25] http://www.africanancestry.com/home/
[26] https://genographic.nationalgeographic.com/
[27] https://www.23andme.com/en-int/dna-ancestry/
[28] https://amino.bio/
[29] http://www.the-odin.com/

Another example is provided by *Biorealize*[30] (USA), a company that sells an automatic bio lab, called *Microbial Design Studio*, capable to design, culture, and test genetically modified organisms. The mini bio lab allows users to genetically engineer bacteria at home for approximately USD 5000. Akin, *Bento Lab*[31] (UK) is a DNA analysis laboratory that combines the essential tools for molecular biology. It allows users to take a biological sample, extract the DNA, and conduct a simple genetic analysis.

2.1.5 Companies that buy/sell genetic data

In the last years, the way genetic testing companies were handling and selling personal genetic data come under the public scope, opening a new debate on the significance of personal data. This issue became a pressing matter after becoming publicly known that DTC-GT companies were entering into million dollar deals with pharmaceutical companies. This is the case, for example, of the company *23andMe* (USA) that facilitated access to data worth 60 million USD of Parkinson's patients to *Genentech* (Herper, 2015). In relation to this issue, it must also be pointed out, that the terms and conditions of DTC-GT companies, at that time, were found to provide ambiguous information, and when consenting to their forms, consumers could be allowing companies to share their *anonymised* data to third parties (Adam & Friedman, 2016; Niemiec & Howard, 2016).

The interest (and market) for personal genetic data continues to grow and companies such as *Pfizer*[32] (USA, around the world) and *Lundbeck*[33] (Denmark) have already shown interest, for example, on the genetic data coming from *23andMe*, as specified by the findings of genetic links to depression by both companies (Lundbeck, 2017; Pfizer, 2016). The interest in genetic data by big companies has also encouraged the appearance of several start-up companies that aim to make the consumer the owner of its own genetic and operate as intermediaries in a new market for companies that are willing to buy this data.

2.1.6 Companies that monetise genetic data using blockchain and cryptocurrencies technologies

With the advent of Big Data and genetic information availability, several were the methods intended to keep genetic information safeguarded while keeping it available to be shared. The use of blockchain technology for trading genetic data is described in this section. The narratives and business models used by these emerging companies raises the question: *is giving the total control to users over their own genomic data the ethical and legal solution for empowering individuals*? Indeed, companies operating in this type of market often claim that their sole aim is to ensure transparency, equal access to market and overall privacy (DNA protection from misuse and genetic discrimination). If this is truly the case, needs to be ascertained.

> **Blockchain**: Open, distributed ledger that can record transactions between two parties efficiently in a verifiable and permanent way. Transaction records are embedded in digital code that is stored in transparently shared databases managed by cluster of computers and thus not owned by any single entity. Individuals, organisations, machines, and algorithms would freely transact and interact with one another with little friction (Iansiti & Lakhani, 2017).

[30] http://www.biorealize.com/
[31] https://www.bento.bio/
[32] https://www.pfizer.com/
[33] http://www.lundbeck.com/

EncrypGen[34] (USA) aims to address the issues of privacy and ownership on genomics by allowing individuals to directly sell their genomic data to researchers and pharmaceutical companies while keeping ownership. In return, they earn DNA tokens, a new cryptocurrency created by the company. Likewise, *Zenome*[35] (start-up in Russia), provides a service where genetic data is exchanged by tokens. In this case, the company ZNA tokens are used to stimulate individuals to upload their genomic data to the company database. *Zenome* also claims that using their services will help to establish equal conditions for drug development among companies and promote the de-monopolisation of DTC-GT companies profiting from selling their users genomic data.

Another example is *Nebula Genomics*[36] (USA), a company founded by famous geneticist George Church (Professor at Harvard University and Massachusetts Institute of Technology (MIT)). The novelty of *Nebula Genomics*, in addition to ownership of the data, the company also offers whole genome sequencing services, thus eliminating in this way companies acting as intermediary between data owners and data buyers. In the future, customers will be able to buy personal genome sequencing at *Nebula Genomics* and pay with Nebula tokens whereas data buyers will use the same tokens to purchase access to genomic data.

Luna DNA[37] (USA) was created in 2017 as a community-owned genomic and medical research database that empowers people. In its conception, *Luna DNA* was established as a Public Benefit where individuals can share their genomic and health information "*for the greater good of the community*" (LunaDNA, 2017). It is based on the concept that all members are viewed as equitable partners, reason why they are awarded shares of ownership (Luna coins) in exchange for their DNA data and other medical record contributions. Dividends for the community are generated when companies and researchers pay to get access to the genetic data stored in the platform. *Luna DNA* already counts with millionaire investments from companies such as *Illumina Ventures* and *Arch Ventures* (LunaDNA, 2018).

2.1.7 Companies that store genetic data

The business of storing data is currently an ongoing competition among the big technological companies, namely Microsoft, Google, IBM, and Amazon among others (

Table 2). Google Genomics[38], a product from Google Cloud, offers customers cloud computing services to store, process, explore, and share large complex genomic datasets. Their slogan promotes the idea that by using Google technology you can power up your science and organise the world's genomic information. Likewise, Amazon Web Services[39] allows customers to simplify and securely scale genomic analysis. Microsoft Azure Genomics[40] also offers the performance and scalability of a world-class supercomputing centre.

> **Cloud computing**: On-demand delivery of computing power, database storage, applications, and other IT resources through an online services platform with according to a usage-based pricing model (Zhang, Cheng, & Boutaba, 2010).

Start-ups such as *DNAnexus*[41] are already using the computer power of these companies to accelerate their genomics services by combining it with their extra features for genomic data handling, analysis and

[34] https://www.encrypgen.com/
[35] https://zenome.io/
[36] https://www.nebulagenomics.io/
[37] https://www.lunadna.com/index.html
[38] https://cloud.google.com/genomics/
[39] https://aws.amazon.com/health/genomics/
[40] https://azure.microsoft.com/en-us/services/genomics/

manipulation. *Seven Bridges*[42], which is a data company specialising in software and data analytics to drive healthcare research, is also using both Amazon Web Services and Google cloud to run their business.

Table 2: Main cloud service providers for genetic data. Source: (Langmead & Nellore, 2018).

Platform	Website	Notes
AWS	https://aws.amazon.com/	IaaS
Google Cloud Platform	https://cloud.google.com/	IaaS
Microsoft Azure	https://azure.microsoft.com/	IaaS
IBM Cloud	https://www.ibm.com/cloud/	IaaS
Alibaba Cloud	https://www.alibabacloud.com/	IaaS
DNA nexus	https://www.dnanexus.com/	SaaS
Illumina BaseSpace Sequence Hub	https://basespace.illumina.com/	SaaS
Seven Bridges	https://www.sevenbridges.com/platform/	SaaS
Globus Genomics	http://globusgenomics.org/	SaaS

IaaS – Interface as a Service.
SaaS – Software as a Service.

2.1.8 Companies that provide deep analysis through Artificial Intelligence of genetic data

The continuously growing field of genomics is currently generating data at an exponential rate (Cook *et al.,* 2016). Subsequently, managing and processing these data is becoming a major hurdle for scientists. In the quest for making sense of all the new knowledge, the scientific community has turned to Artificial Intelligence for support (Kersting & Meyer, 2018). The ever-evolving and ever-improving machine learning technologies are expected to have a major role in the automation and efficient analysis of the enormous amount of data already available.

Yin *et al*. (2018) published a paper explaining how AI can bring the biological and medical science research to a new level. The authors argue that through AI we can improve the efficacy and precision of CRISPR technology, especially into better understanding multifactorial disorders and designing therapeutic targets by combining machine learning and pattern recognition capabilities. The idea of combining genomics and AI is significantly being taken by technological companies. For example *Deep genomics*[43] (Canada) claims that the future of medicine will rely on artificial intelligence because "*biology is too complex for humans to understand*" (Deep Genomics, 2018). In one of their flagship projects, *Deep genomics* is using AI to perform in silico studies to predict genetic drugs efficiency. With a similar marketing strategy, *Atomwise*[44] (USA) uses AI-deep learning approaches to extrapolate information from a large database of compounds to determine which subset has higher specificity for synthesis and testing. Through this type of data analysis a time consuming and strenuous process can be simplified into a faster and more efficient drug design, finding data patterns which a human being would never be able to see.

Within the realm of drug research and development, AI is also being used as a tool for the analysis of genomic data. For example, *IBM Watson*[45] (USA, global) uses an AI-based approach to unravel connections among drugs, genes, diseases and life sciences knowledge to provide personalised medicine with evidence-

[41] https://www.dnanexus.com/
[42] https://www.sevenbridges.com/
[43] https://www.deepgenomics.com/
[44] https://www.atomwise.com/
[45] https://www.ibm.com/products/watson-drug-discovery

based predictions. The open-source *DeepVariant*[46] (a deep learning neural network technology) was created by the *Google Brain* team[47] (USA, global), in collaboration with *Verily Life Sciences*[48], and allows to identifying genetic variants (SNPs) by using a software that translates genomic information into image-like data. According to *Google*, *DeepVariant* is currently outperforming state-of-the-art methods, winning in 2016 the *PrecisionFDA Truth Challenge*[49] award. Since then, the *Google Brain* team has reduced the percentage of errors by 50% (DePristo & Poplin, 2017).

2.2 DIYbio spaces and groups

The rapid expansion of the DIYbio movement is noticeable in the propagation of DIY community laboratories in recent years, eithers as standalone facilities or associated to existing makerspaces[50]. Data published by the Brookings Institution (Kolodziejczyk, 2017) demonstrates that, for example, in Europe in the last 5 years (2013 to 2017) there was a growth of 139%, from 23 to 55 DIYbio spaces. Similarly, in North America, for the same period the growth was of 77%, from 35 to 62 spaces; and in Asia of 214%, from 7 to 22 spaces (Figure 4).

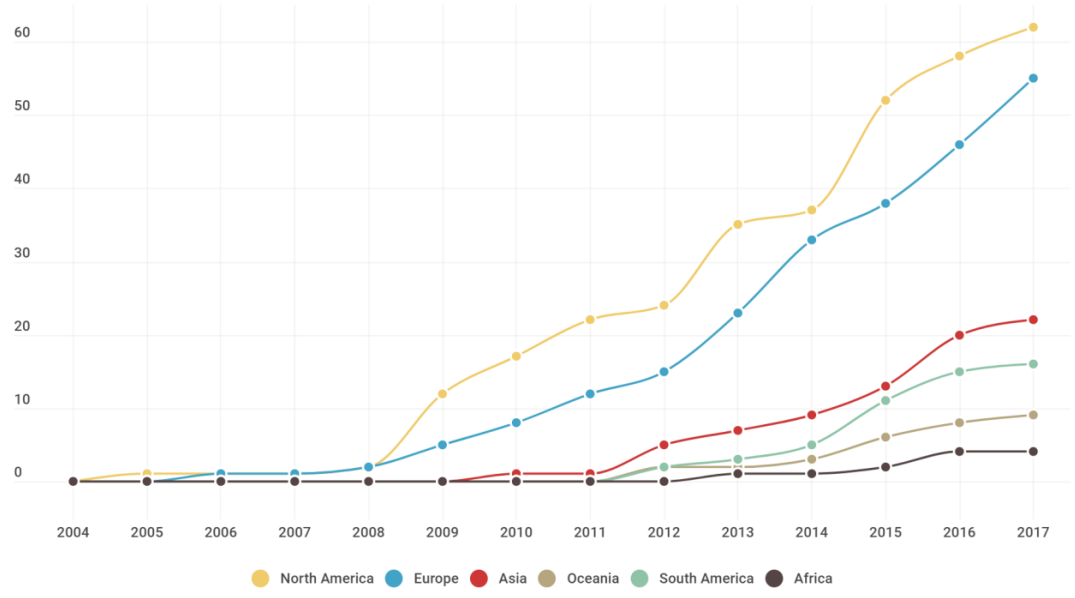

Figure 4: Evolution of the number of active DIYbio groups by geographical area. Source: Adapted from (Kolodziejczyk, 2017).

[46] https://github.com/google/deepvariant
[47] https://ai.google/research/teams/brain
[48] https://verily.com/
[49] https://precision.fda.gov/challenges/truth
[50] For the purposes of this report, the term makerspace is inclusive of FabLabs and Hackerspaces, representing collaborative community physical spaces.

Table 3: Data quality box for Figure 4.

Data Source	Brookings Institution (Kolodziejczyk, 2017).
Data Year	2004 to 2017.
Data Processing	None. Raw data not available.
Data Type	Numeric (integer).
Data Items	735 data points. 168 unique spaces for the year of 2017.

Data available from the *DIYbio.org* website[51] provides slightly different numbers, but sustains the overall dimension of the DIYbio movement in the world. In Figure 5, it is possible to observe the geographical spread of the DIYbio movement, with a higher significance, as also illustrated in Figure 4, in North America and Europe.

Regarding the data relative to the DIYbio movement, it must also be pointed out that it is not possible to guarantee the accuracy of the information since the data come from self–reported online sources. Moreover, as the movement is undergoing a rapid growth, it is very likely that the data retrieved misses out new spaces that are not yet registered.

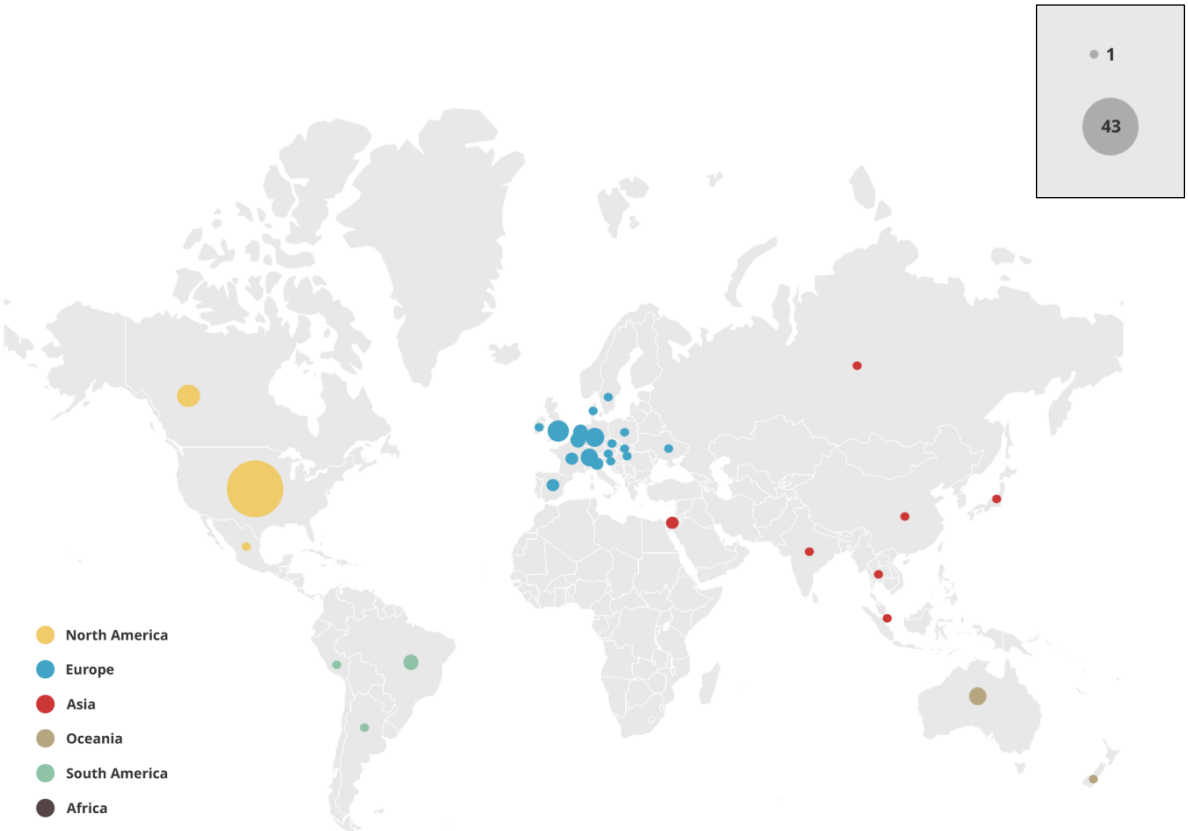

Figure 5: World map of DIYbio communities (year 2018).

[51] https://diybio.org/local/

Table 4: Data quality box for Figure 5.

Data Source	DIYbio.org website.
Data Year	Data retrieved from original source in August 2018.
Data Processing	Data re-grouped into new geographical areas.
Data Type	Numeric (integer).
Data Items	Data from 106 unique spaces.

With the aim of further exploring the DIYbio landscape in Europe, the data available from the *DIYbio.org* website was merged with other existing data sources on the subject, namely with the data available in the *DIYbiosphere* website[52] and the authors own research on the maker movement (Rosa et al., 2017). Table 5 provides the results obtained, and it too gives an initial glimpse of the dimension of the DIYbio movement in Europe. It lists by country, *(1)* DIYbio Labs; *(2)* Citizen Science labs with focus on synthetic biology; *(3)* DIYbio associations; *(4)* DIYbio local communities; and *(5)* start-ups that had their origins or are related to the DIYbio movement. An analysis of which spaces and/or groups are offering genetic engineering[53] or genetic testing activities was done by the authors.

[52] http://sphere.diybio.org/

[53] Genetic engineering: Group of applied techniques of genetics and biotechnology used to cut up and join together genetic material and especially DNA from one or more species of organism and to introduce the result into an organism in order to change one or more of its characteristics (Merriam-Webster, 2018b)

Table 5: DIYbio labs, communities and start-ups in Europe. Source: Adapted from (DIYbiosphere, n.d.; Rosa *et al.*, 2017).

Name	Website	Country	Type	Genomics Activities
Kilobaser	http://www.kilobaser.com/	Austria	Start-up/Equipment	Yes
Open bioLab Graz Austria	https://realraum.at/wiki/doku.php?id=olga:olga	Austria	DIY Bio Lab	No
DIYbio Belgium	http://www.diybio.be/	Belgium	Association	Yes
Open BioLab	https://www.erasmushogeschool.be/nl/labs/open-biolab	Belgium	DIY Bio Lab	Unclear
ReaGent	https://reagentlab.org/	Belgium	DIY Bio Lab	No
Brmlab	http://brmlab.cz/index.html	Czech Republic	Citizen Science Lab	Unclear
BiologiGaragen	http://biologigaragen.org/	Denmark	Bio Lab	No
Biome Hack Lab	https://lebiome.github.io/	France	Community	Unclear
Eligo Bioscience	http://eligo.bio/	France	Start-up	Yes
La Paillase	http://lapaillasse.org/	France	Citizen Science Lab	No
Biogarage	http://biogarage.de/	Germany	DIY Bio Lab	No
BIOLAB Eberswalde	https://plattform-n.org/project/biolab-eberswalde-offenes-forschungsla/	Germany	DIY Bio Lab	Unclear
Biotinkering Berlin	https://www.biotinkering-berlin.de/	Germany	Community	Unclear
BioTop Community Lab	http://www.biotop-heidelberg.de/	Germany	Community	No
Dezentrale	https://dezentrale-dortmund.de/	Germany	DIY Bio Lab	No
FAU FabLab	https://fablab.fau.de/diybio/	Germany	DIY Bio Lab	No
MikroBiomik	https://mikrobiomik.org/en	Germany	Association	No
Forma Biolabs	https://twitter.com/formabiolabs	Ireland	DIY Bio Lab	Unclear
OWL - Open Wet Lab	http://www.openwetlab.it/	Italy	Association	Unclear
Bio Art Laboratores	http://bioartlab.com/	Netherlands	DIY Bio Lab	No
DIYbio Groningen	https://www.facebook.com/DIYBioGroningen/	Netherlands	DIY Bio Lab	No
Open Wetlab	https://waag.org/en/labs/open-wetlab	Netherlands	DIY Bio Lab	Yes

Name	Website	Country	Type	Genomics Activities
Biohacking Poland	https://www.facebook.com/groups/BiohackingPoland/	Poland	Community	No
DIYbio Bratislava	https://www.facebook.com/groups/diybiobratislava/about/	Slovakia	Community	No
BioTehna	http://kersnikova.org/biotehna/	Slovenia	DIY Bio Lab	Unclear
Symbiolab	https://www.irnas.eu/projects/	Slovenia	Start-up	No
DIY Bio Barcelona	http://www.diybcn.org/	Spain	DIY Bio Lab	Unclear
PechBlenda Lab	https://pechblenda.hotglue.me/	Spain	Citizen Science Lab	Unclear
BioNyfiken	https://www.facebook.com/BioNyfiken/	Sweden	DIY Bio Lab	Yes
Bioscope	http://bioscope.ch/	Switzerland	Citizen Science Lab	Yes
GaudiLabs	http://www.gaudi.ch/GaudiLabs/	Switzerland	DIY Bio Lab	Unclear
Hackteria	https://www.hackteria.org/	Switzerland	Community	No
Hackuarium	http://www.hackuarium.ch/en/	Switzerland	DIY Bio Lab	No
L'Eprouvette	http://www.eprouvette.ch	Switzerland	Citizen Science Lab	Yes
Bento Lab	https://www.bento.bio/	UK	Start-up/Equipment	Yes
BioMakeSpace	https://biomake.space/home	UK	DIY Bio Lab	Yes
London Biohackspace	https://biohackspace.org/	UK	DIY Bio Lab	No
MadLab	https://madlab.org.uk/	UK	Citizen Science Lab	Yes
OpenGenx	https://opengenx.wordpress.com/	UK	DIY Bio Lab	Yes
DIYBio Kiev	https://groups.google.com/forum/#!forum/diybio-kiev	Ukraine	Community	Unclear

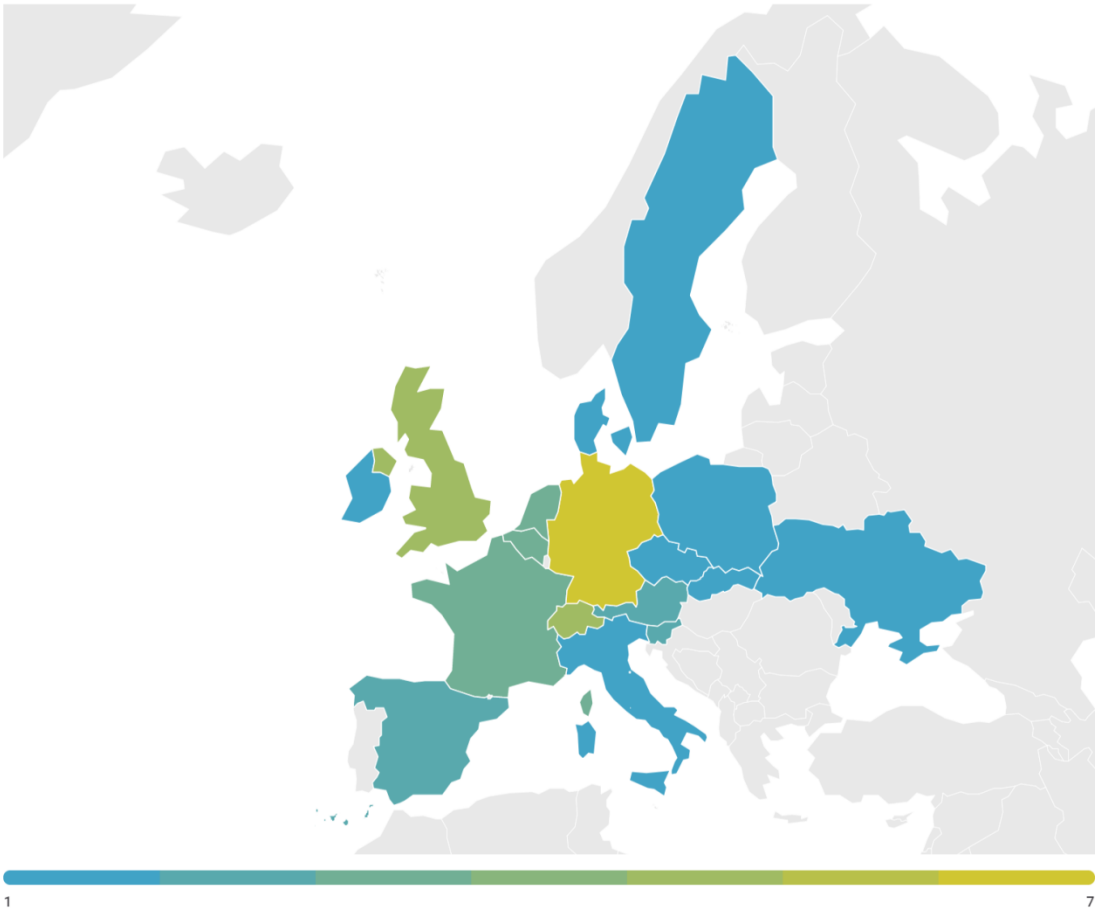

Figure 6: Map of the total number DIYbio spaces and groups in Europe, by country (year 2018).

Table 6: Data quality box for Figure 6.

Data Source	– DIYbio.org website. – DIYbiosphere website. – Database from the JRC Technical Report "Overview of the Maker Movement in the European Union" (Rosa *et al.*, 2017)
Data Year	Data retrieved from original sources in August 2018.
Data Processing	– Data cleaned for duplicate entries. – Data checked for non-relevant entries. – Data grouped into types of space/group.
Data Type	Numeric (integer).
Data Items	Data from 40 unique spaces/groups.

2.3 Chapter summary

The main findings of chapter 2 are:

- The Genomics market is blooming and it is expected to produce revenues of around 20 billion USD by 2022.
- The large majority of genomic businesses are based in North America.
- Human genomics has awakened the interest of companies working with blockchain, cloud computing, data mining and artificial intelligence.
- Nowadays genomics businesses clients are not only academia and pharmaceutical companies but also citizens.
- Direct-to-consumer genetic testing companies have developed different narratives and strategies to attract a wide variety of consumers targeting their needs. This ranges from targeting the ones interested in knowing their genetic inheritance and ancestry, to the ones focused on getting personalised exercise, cosmetics and diet programs tailored to their own genome.
- Through the purchase of online DIY Bio kits for genetic engineering, citizens can perform these experiments in their houses and garages.
- Monetising genetic data has turn into a profitable business performed by direct-to-consumer genetic testing companies and even by individuals through blockchain technology.
- The DIYbio community also presented a gradual presence, possibly indicating that the movement is becoming anchored and steadier across Europe.

3. Regulatory discourse

3.1 Governance

The genomics debate has always been controversial and multifaceted, covering a wide scope of subjects ranging from genetically modified organisms (GMO) to human cloning (Callaway, 2018; Green, 2014). Indeed, the advances in biomedicine witnessed in the early nineties, caused concern to the Council of Europe which undertook the task to outline a set of common general standards for the protection and preservation of human dignity across the field of biomedicine - for an historical perspective see, for instance, Sacchi & Holmes (2016). In April 1997, the *Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine*, also known as the *Oviedo Convention* (Council of Europe, 1997), was opened for signature to parties, entering officially into force on December 1999. The *Oviedo Convention* can be seen as a unique effort to implement the foundations for an international bioethics standardisation approach and a minimum commitment throughout the countries that signed it. Since its establishment, the *Oviedo Convention* has been complemented with four additional protocols targeting: (1) the prohibition of human cloning (Council of Europe, 1998), (2) the right to transplantation (Council of Europe, 2002), (3) biomedical research (Council of Europe, 2005), and (4) genetic testing for health purposes (Council of Europe, 2008).

For the purposes of this report, we highlight the following chapter of the *Oviedo Convention*:

Chapter IV – Human Genome

Article 11 – Non-discrimination

> Any form of **discrimination** against a person **on grounds of** his or her **genetic heritage** is prohibited.

Article 12 – Predictive genetic tests

> Tests which are **predictive of genetic diseases** or which serve either to **identify** the subject as a carrier of a **gene responsible for a disease** or to detect a genetic predisposition or susceptibility to a disease may be **performed only for health purposes or for scientific research linked to health purposes**, and subject to appropriate genetic counselling.

Article 13 – Interventions on the human genome

> An intervention seeking to **modify the human genome** may only be undertaken for **preventive**, **diagnostic or therapeutic purposes** and only if its aim is **not to introduce any modification in the genome of any descendants**.

Article 14 – Non-selection of sex

> The use of techniques of medically assisted procreation shall not be allowed for the purpose of choosing a future child's sex, except where serious hereditary sex-related disease is to be avoided.

Although the establishment of the *Oviedo Convention* was promoted by states such as Japan, Australia, USA, Mexico, Canada and the Holy See, it has only been signed and ratified by 29 European parties. From these signatories, six countries - Denmark, Turkey, Moldova, Croatia, Norway and Switzerland - have added their own *reservations and declarations* to the *Oviedo Convention* in order to be implemented in their countries. Noticeable absences include the European Union as an institution, Germany, Ireland, Italy, The Netherlands, Poland, Sweden, United Kingdom, Israel, and the Russian federation. It is worth mentioning that the UK and Germany did not sign the *Oviedo Convention*, each for its own specific reasons. The UK considered the *Oviedo Convention* to be too restrictive, whereas Germany assumed it to be too permissive, especially in cases of embryo research and non-therapeutic interventions on persons unable to give their consent. The reasons behind Germany's decision not to sign the *Oviedo Convention* were also rooted on reactions generated by a strong public opposition and responsive legislation to human experimentation (in part related to historical events). Consequently, the German Embryo Protection Act states that both artificial alterations and use of human germ cells are prohibited (BJV, 1990). On the other hand, the UK implemented the Human Fertilisation and Embryology Act (UK Department of Health, 2008). Among several other modifications to the *Oviedo Convention*, the amendment bans gender selection for non-medical reasons, allows keeping human embryos *in vitro* for research purposes, and licenses the creation, keeping and use of human admixed embryos[54].

3.2 Legislation regulating gene editing in humans in the EU

The absence of comprehensive legislation regulating gene editing, common and applicable to all European countries has already raised a series of issues. As a direct consequence, nowadays, there is still uncertainty about which type of legislation should be applied to the practices of gene editing in humans. In 2015, the novel technique of gene-editing CRIPR/Cas9 appeared and allowed for a much cheaper and easier way to do precise modifications on the genome, setting it apart from the genetic engineering techniques existing so far (Reis, Hornblower, Robb, & Tzertzinis, 2014). One of the most pressing concerns in Europe is whether this new technique falls under the scope of the GMO legislation or not (Abbott, 2018), a controversial topic especially when the current legislation has been claimed to present double standards and missing opportunities for business within the EU (Tagliabue, 2017).

A report produced by the Nuffield Council on Bioethics (Nuffield Council on Bioethics, 2018) widely reviewed current genomics related legislation in Europe, pointing out that "*there is no prohibition in European community or international law that would make heritable genome editing interventions unlawful*" (Nuffield Council on Bioethics, 2018, p. 100). However, these prohibitions take place at the country-level, where, on one side there are countries that prohibit the use of human embryos on research (e.g. Italy) or that prohibit heritable genetic modification of human embryos (e.g. Sweden) and, on the other side, there are countries that permit the creation of embryos for research (e.g. UK). Even though the focus of the Nuffield report is on germline genetic modifications[55], it also pointed out that no regulation exists for controlling genetic modification of adult humans (somatic cells modifications[56]): "*there is no international treaty of general application that directly regulates the human genome or the possibilities for its modification*" (Nuffield Council on Bioethics, 2018, para. 114). This legislation twilight takes place in a

[54] Admixed Embryos: types of embryo that contain both human and animal DNA
[55] Germline Genetic Modification: genetic modification of eggs and sperms that are inheritable to the next generation.
[56] Somatic cells: Any cell of a living organism other than the reproductive cells (eggs and sperms). Genetic modifications in these cells are not inheritable.

world where patients are undergoing gene therapy trials at pharmaceutical companies and citizens are injecting themselves with CRISPR/Cas9 kits (Mullin, 2018).

In terms of human genomics, CRISPR/Cas9 technique was widely debated among the public when Chinese scientists used it to gene edit unviable human embryos (Liang *et al.*, 2015). These news fuelled once again the discussion around the genomic law and the ethical and social implications that such techniques could have in the future (Cyranoski & Reardon, 2015). Following the publication of two articles from Chinese scientists using human embryos gene-edited by means of CRISPR, the UK Human Fertilisation and Embryology Authority (HFEA) gave green light to an experiment using human embryos to be gene-edited in what would be the beginning of an arms race for genetic editing technology (Callaway, 2016; Mukherjee, 2018). The recent episode of the Chinese biophysics researcher He Jiankui rekindled the discussion[57]. Jiankui claims he helped to create the first babies born with edited genomes - twin baby girls were produced with genetically modified embryos in order to made them resistant to HIV and thus remove the risk of inheriting HIV from one of the parents. The case raised much attention and debate from media and academia, exemplifying the ethical and legal ramifications of human gene editing. Though the experiment was seen by most as unethical, it shows how gene-editing practices are not addressed properly by current international legislation or the lack of an international framework governing this type of work.

3.3 DIYbio legislation

EU legislation presently in force in relation to genomics is primarily directed to regulate research institutes, official organisations and companies. Hence, genomics research in academia is to a certain extent controlled and regulated; however, this is not the case for DIYbio. When existing legislation was first designed, policy makers did not foresee the raise of the DIYbio movement and neither that genetic engineering experiments could be done at an unconventional space such as a garage. Due to the lack of specific legislation tailored to DIYbio activities, EU governments are normally restricted to the application of general laws when aiming to govern these actions. For example, governments can use GMO legislation for supervising gene edited organisms created at DIYbio labs, more specifically, the Directive 2001/18/EC of the European Parliament & of the Council of 12th March 2001, which states that GMOs cannot be released into the environment (European Parliament, 2001). Recently, in July 2018, the European Court of Justice also considered that genetic editing organisms fall under the scope of the same regulations that rule over GMOs (Callaway, 2018). The only EU government that has taken any specific action towards the regulation of DIYbio activities was Germany (Brown, 2017). In January 2017, the German government emitted a statement (BVL, 2017) reminding DIYbio suppliers and consumers about *Article #1 of the Genetic Engineering Act* (BJV, 1993), which states that genetic engineering activities can only be performed by "*officially monitored laboratories under the supervision of a knowledgeable project manager*" and misconduct can be penalise with a fine of EUR 50000 and imprisonment.

An interesting case illustrating the lack of specific legislation involving DIY activities (in this case, in the USA) involves the company *The ODIN*. *The ODIN* initially started selling affordable genetic engineering kits to the public, including one particular kit that allowed individuals to produced mead (type of alcoholic beverage) using genetically modify fluorescent yeast (Brown, 2016). The US Food and Drug Administration (FDA) promptly intervened and banned the company from selling unsafe products targeted at human consumption. *The ODIN* response to the FDA action was to change the product description on their

[57] See for example https://www.nature.com/articles/d41586-018-07573-w

website, removing any mention to being directed to human consumption. The final result was that the FDA did not have any more legal grounds to intervene in the regulation of these kits. Currently, *The ODIN* continues to sells similar kits, now also under the flag of *educational DIYbio kits*, thanks to the ambiguity surrounding the regulation of DIY biology (Olmstead, 2017).

3.4 Chapter summary

The key findings of chapter 3 are:

- There are several laws and organisms designed to target omics-related issues, yet a lack of a central and unanimous regulation is found across the member-states of the European Union.
- The developing of national legislation to handle such emerging technologies also depends of the socio-demographic and historical character of the involved nations, which ends up influencing the design of these laws.
- Gene edition of human embryos is regulated differently in Europe - there is no unified legislation.
- Somatic gene edition of adult humans is not regulated by any specific legislation.
- The lack of legislation consensus and inability to keep pace of scientific progress sets up a landscape unable to regulate emerging technologies.
- There is a general lack of regulation for the DIYbio community, creating a legislative grey area that can be (and is) exploited by some of its members.

4. Ethical discourses

4.1 Ethics committees

Ethical concerns surrounding human genetic modifications have been for a long time under the public and political scope, driven primarily by academia, science councils, associations and societies. In this section we examine the most relevant documents (published in English) related to ethical recommendations on human genomics by different world organisations (listed in Table 7). The reports and statements produced by each of these organisations converge in one decisive point: ***the need of regulations for human gene editing***. Equally important, they also target the need to create a specific committee formed by a diverse variety of professionals that can evaluate emerging technologies in a multi-approachable and multi-disciplinary way, where the ethical and social implications are also considered.

It is also important to notice how the various organisations throughout the years have realised that it is important to incorporate the views and recommendations coming from non-scientists groups. This change can be particularly observed in the reports produced in 2018 such as the one from the Nuffield Council on Bioethics. Similar concerns are also visible on reports from newly formed organizations such as ARRIGE, where not only scientists have been consulted but also patient organisations, representatives of the economic and communication sector, disability rights advocates, and the *general* public.

Regarding the ethics of embryonic gene editing, basic research is generally supported whereas clinical applications are considered too risky and unethical to explore further. It is believed that basic research can allow the development and discovery of enhanced techniques and therapies that in the future may be used on clinical applications. The report by UNESCO (UNESCO & IBC, 2015) states that further advances should be halted until a more clear understanding of the ethical implication are understood. Even though the prohibition of gene editing of human embryos for reproduction is agreed, it is also specified that gene editing of embryos "*should not be advisable*". In cases trying to avoid inherited genetic disease, where other techniques are not possible, it leaves space for the possibility to perform gene editing in embryos.

In this section of the report, we have also included ethical statements related to DTC-GT, which can be visualised in Table 7 in shading. For an in-depth description of the analysis of ethical statements on DTC-GT, please refer to Rafiq *et al.* (2015) and Laestadius *et al.* (2017). Overall, the main concerns raised by these reports are the lack of public understanding of the implications on consumer data privacy, the right to ownership, what companies do with the data and the validity of anonymity of data.

Table 7: Ethic committees involved in human genomics.

Organisation	Region	Report/Statement Title	Year	People involved in the report	A	B	C	D
Nuffield Council on Bioethics	UK	Genome editing and human reproduction: social and ethical issues (Nuffield Council on Bioethics, 2018).	July 2018	Experts in reproductive genetics , genomics, bioethics, reproductive and disability rights advocates and public online questionnaire	X			X
Association for Responsible Research and Innovation in Genome Editing (ARRIGE)[58]	France	CRISPR–Cas9: A European position on genome editing (Hirsch, Lévy, & Chneiweiss, 2017).	March 2018	Patient organizations, representatives of Ethics committees, of the economic sector, and communication sector		X		
The European Society of Human Genetics (ESHG) and The European Society of Human Reproduction and Embryology (ESHRE)	Europe	Human germline gene editing: Recommendations of ESHG and ESHRE (de Wert et al., 2018).	January 2018	Diverse universities in Europe in the topics of health, ethics, medicine, gene technology, clinical genetics, reproductive medicine, stem cell, fertility, molecular biology and gynaecology.	X			X
European Academies Science Advisory Council (EASAC)	Europe	Genome editing: scientific opportunities, public interests and policy options in the European Union (EASAC, 2017).	March 2017	Physician, geneticist, microbiologists, ethicists, molecular biologist, chemist, evolutionary biologist.	X			X
National Academy of Sciences Engineering Medicine (NASEM)[59]	USA	Human Genome Editing: Science, Ethics, and Governance (NASEM, 2017).	2017	Physician, geneticist, policy advisor, biologists, lawyer, political scientist, molecular biologist	X			X
Bioethics and Law Observatory (University of Barcelona)	Spain	Document on bioethics and gene editing in humans (Santaló & Casado, 2016).	December 2016	Experts in Bioethics, philosophy, reproduction, medicine and genetics.	X			X
American College of Medical Genetics and Genomics (ACMG)	USA	Direct-to-consumer genetic testing: a revised position statement of the American College of Medical Genetics and Genomics (ACMG Board of Directors, 2016).	February 2016	Medical geneticists and other healthcare providers	X		n/a	n/a
The European Group on Ethics in Science and New Technologies (EGE)	Europe (EU)	Statement on Gene Editing (EGE, 2016).	2016	Ethicists, geneticist, lawyers, biologist, philosophers and political scientists		X		X
The European Group on Ethics in Science and New Technologies (EGE)	Europe (EU)	Opinion No 29: The ethical implications of new health technologies and citizen participation (EGE, 2015).	2015					
UNESCO International Bioethics	International	Report of the International Bioethics	October 2015	Geneticists, bioethicist, medical doctor, lawyer,	X			

[58] https://arrige.org/
[59] https://www.nap.edu/

Committee (IBC)		Committee on Updating Its Reflection on the Human Genome and Human Rights (UNESCO & IBC, 2015).		philosopher, psychologist, social scientist, pharmacologist	X		n/a	n/a
Society for Developmental Biology	USA	Position Statement from the Society for Developmental Biology on Genomic Editing in Human Embryo (Society for Developmental Biology, 2015).	April 2015	Biologists	X		X	
International Society for Stem Cell Research (ISSCR)	USA	The ISSCR statement on human germline genome modification (ISSCR, 2015).	March 2015	Experts in stem cell research		X		X
Canadian College of Medical Geneticists (CCMG)	Canada	CCMG Statement on Direct-to-Consumer Genetic Testing (Nelson *et al*., 2011).	January 2011	Geneticists	X		n/a	n/a
European Society of Human Genetics (ESHG)	Europe	Statement of the ESHG on direct-to-consumer genetic testing for health-related purposes (European Society of Human Genetics, 2010).	December 2010	Not specified	X		n/a	n/a
The European Group on Ethics in Science and New Technologies (EGE)	Europe (EU)	Opinion No 25: Ethics of synthetic biology (EGE, 2009).	2009					
American Society of Human Genetics (ASHG)	USA	ASHG Statement on Direct-to-Consumer Genetic Testing in the United States (Hudson, Javitt, Burke, Byers, & Committee, 2007).	September 2007	Molecular Biologist, geneticists, lawyer, expert in Bioethics and Humanities, medical doctor.	X		n/a	n/a
The European Group on Ethics in Science and New Technologies (EGE)	Europe (EU)	Opinion No 18: Ethical aspects of genetic testing in the workplace (EGE, 2003).	July 2003	Ethicists, geneticist, lawyers, biologist, philosophers and political scientists	X		n/a	n/a

A: Advise the need of regulation and gives recommendations.
B: Advise the need of regulation without giving specific recommendations.
C: Complete ban on embryonic gene editing (Basic Research, Pre-clinical and Clinical applications).
D: Selective ban on embryonic gene editing.

4.2 Ethical issues related to genomics businesses

Businesses providing DTC-GT and gene therapy services are currently facing strong critics from news media and ethical committees regarding multiple issues. *Who owns the genetic data? How is the anonymity of genomic data collected handled? Is the genetic data being sold to third parties? Is the quality of the genetic results and counselling appropriate? Who is handling the patients and user's DNA data and delivering the genetic results? Are they experts in the field? Who is auditing these businesses?* In the following subsections, we describe in detail these critical issues.

4.2.1 Data ownership

In 2017, Phillips (2017) analysed the contracts (terms and conditions) of several DTC-GT companies and found several concerning issues. In particular, the study unveiled:

1) The presence of clauses that allow unilateral variation of contracts;
2) Exclusion clauses for liabilities when personal injuries are caused by the company's negligence;
3) Clauses indicating that their services are not provided for medical purposes;
4) Clauses purporting to bind the consumer to resolve any disputes in another jurisdiction.

In addition, often these contracts give the ownership of the genetic data to the company instead of the customer, unless expressly stated otherwise. For example, Adam & Friedman (2016) in an article published in the journal *Genetics in Medicine*, addresses the concrete situation around the deal between *23andMe* and *Genentech,* where DNA samples collected by *23andMe* from more than 800000 people, including about 12000 with Parkinson disease were sold to *Genentech*.

In sum, as another author states, "*DTC-GT companies do not consistently meet international transparency guidelines related to confidentiality, privacy, and secondary use of data*" (Laestadius et al., 2017, p. 1).

4.2.2 Data anonymity

Genetic data privacy and anonymity are nowadays serious pressing issues, in particular, with the increasing number of genetic tests being done and the subsequent increase of sales of genetic data throughout the word. Even though DTC-GT companies anonymise the DNA data collected, frequently there is still an internal code that links the genetic information to the individual. Moreover, it is as well possible to uncover the identities of customers or patients by cross-referencing data from recreational genetic genealogy databases with information freely available on the internet such as surnames, age and state (Gymrek, McGuire, Golan, Halperin, & Erlich, 2013). Genomic data is the ultimate source of identity for humans, and this was visibly demonstrated when a serial killer from the seventies was caught in April 2018, thanks to the genetic data of a relative stored by the DTC-GT company *GEDmatch* (Arango, Goldman, & Fuller, 2018).

Although the use of genetic data to caught criminals can be seen as beneficial for society, it should not halt us from raising questions about the doors that are being opened when anonymity is not an issue. If we recall the case of Henrietta Lack (Callaway, 2013), the original donor of the HeLa cancer cell line which

nowadays is widely used worldwide for scientific research, no consent form was signed (or for that matter even existed), resulting in her full identity being published in two separate scientific studies. Needless to say that genomics companies and pharmaceutical companies promptly offer *Terms of Agreement* that are mandatory to be signed by costumers, yet we continue to wonder ***how many of the individuals sharing their DNA data really understand the implications and risks of the procedure?***

Another pressing concern is the possibility of having genetic databases hacked. Indeed, this was partly what happened in October 2017, when 92 million people had their accounts data leaked from the genealogy company *MyHeritage* (Rahhal, 2018). Even though *per se* no genetic data was stolen, usernames and encrypted passwords were revealed, demonstrating possible fragilities and threats in terms of cybersecurity associated to the storage of genetic data. The trade of genetic data is today an extremely lucrative business, and as such the monetisation of hacked data has become an attractive practice. The thread is even more alarming as it is possible to discover one's identity via the possibility of cross-referencing "pseudo-anonymised" genetic data. If such information is made public, ***it can have a considerable impact in people's employability, insurability and daily life interactions with friends and family***.

4.2.3 Business model

To date, it is still not entirely clear what the business models of certain genomics companies are, neither the main source of revenues, in particular of DTC-GT companies - *does it derive from selling products to consumers or from selling genetic data to pharmaceutical companies?* According to a white paper published by *Nebula Genomics* (Grishin *et al.*, 2018), genomic companies mainly rely on businesses agreements done with big pharmaceutical companies (Figure 7).

Figure 7: Business model for genomics companies such as 23andMe, Helix, and Ancestry. Source: (Grishin et al., 2018)

As stated by *Nebula Genomics*, "*people pay to sequence or genotype their genomes and receive analysis results. Personal genomics companies keep the genomic data and sell it to pharma and biotech companies that use the data for research and development*" (Grishin et al., 2018).

4.2.4 Genetic counselling

According to Schaper & Schicktanz (2018), most of companies in the genomics field tend to imply medical legitimacy by using suggestive publicity material. Moreover, they state that by using DTC-GT services, consumers are empowering themselves and taking their health's future on their own. However, the lack of professional genetic counselling is one of the major ethical standpoints that needs to be addressed, in particular, when a consumer can obtain different results when taking genetic tests from different companies (Ramsey, 2018). A recent paper by Schleit *et al*. (2018) also illustrated this particular issue by evidencing a case study where an impactful difference between two genetic reports was verified. On one side, the results received from the company *23andMe* tested negative for breast cancer markers, whereas on the other side, the results obtained from *Adult Genetic Medicine Clinic* tested positive for the same DNA sample. The disparity was attributed to the difference of methods and algorithms used by the different companies but it could also be due to an inappropriate data analysis. It is evident that qualified and trained personal is needed for performing analysis and for delivering results to consumers.

4.3 DIYbio code of ethics

The non-profit organisation *DIYbio.org* (founded by Mac Cowell and Jason Bobe) was the first to target one of the major concerns regarding the DIYbio movement: the lack of regulation. In May 2011, a number of individuals and delegates from five countries (Denmark, England, France, Germany, and Ireland) of the *DIYbio.org* European community gathered at a conference organised by the *DIYbio.org* organisation, in London, UK, with the goal of generating an aspirational code of ethics for the emerging do-it-yourself biology movement (DIYbio, n.d.-a). Following this example, in July 2011, a similar but fundamentally different code of ethics was drafted in a second congress in North America. Although both codes of ethics (see Table 8) are important milestones in establishing some guidelines for the DIYbio moment, they are not legally binding.

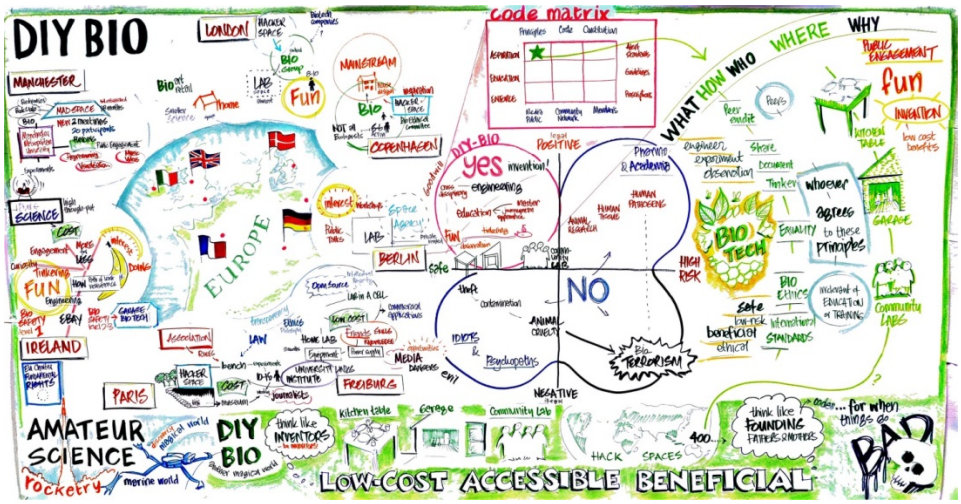

Figure 8: Sketch of the European DIYbio code of ethics. Source: (DIYbio, n.d.-a).

Table 8: Side-by-side comparison of the European [60] and North America[61] DIYbio codes of ethics.

DIYbio code of ethics values	European Congress	North American Congress
Transparency	Emphasises transparency and the sharing of ideas, knowledge, data and results.	Emphasises transparency, the sharing of ideas, knowledge and data.
Safety	Adopt safe practices.	
Open access	Promote citizen science and decentralised access to biotechnology.	
Education	Help educate the public about biotechnology, its benefits and implications.	Engage the public about biology, biotechnology and their possibilities.
Modesty	Know you don't know everything.	
Community	Carefully listen to any concerns and questions and respond honestly.	
Peaceful Purposes	Biotechnology must only be used for peaceful purposes.	Biotechnology should only be used for peaceful purposes.
Respect / Environment	Respect humans and all living systems.	Respect the environment.
Responsibility	Recognise the complexity and dynamics of living systems and our responsibility towards them.	
Accountability	Remain accountable for your actions and for upholding this code.	
Tinkering		Tinkering with biology leads to insight; insight leads to innovation.

As observed in Table 8, the main differences between both codes of ethics rely on the permissiveness mainly evidenced by the use of the words *must* and *should*. These differences and the cultural origins of both codes of ethics have widely been analysed by Keulartz & van den Belt (2016) in the article "*DIY-Bio – economic, epistemological and ethical implications and ambivalences*". According to the authors there is a clear tension between Europe and the USA. Where the model of DIYbio for USA is oriented to market driven entrepreneurship and personal enhancement, the EU model is focused on open access, social empowerment and community

[60] https://diybio.org/codes/draft-diybio-code-of-ethics-from-european-congress/
[61] https://diybio.org/codes/code-of-ethics-north-america-congress-2011/

4.4 Chapter summary

The main findings of chapter 4 are:

- The reports and statements produced by ethical committees call for a need of regulations for gene editing in human embryos, , direct-to consumer genetic testing and also for the creation of a committee that can evaluate emerging technologies in a multi-approachable and multi-disciplinary way, where the ethical and social implications are also evaluated.
- Since 2018 ethical committees are incorporating the voices of non-scientific groups in their statements, reports and within the organizations themselves.
- DTC-GT companies do not always meet international transparency guidelines related to confidentiality, privacy, and secondary use of data.
- Even though DNA sample from clients is anonymised, it is still possible to discover one's identity via cross-referencing and hacking. If this happens, the consequences on people's employability, insurability, privacy and daily life interactions with friends and family can become compromised.
- DTC-GT business models are not entirely clear; it is unknown if their main revenues come from selling services and products to consumers or from selling genetic data to third parties (or both).
- Different DTC-GT companies can provide different results for the same client, the disparity is attributed to the difference of methods and algorithms but it could also be due to an inappropriate data analysis making it more evident the need for trained personal for performing analysis and for delivering results to consumers.
- The DIY Bio movement does not have any type of binding legal regulation to control it, possessing only a simple and self-regulated code of ethics.

5. Media discourse

The media can be considered the principal source of information (or misinformation) of scientific and technological advances of the public. In a society striving to increase science standards and scientific literacy, the means by which information gets to the public can have a major impact on citizen's opinion and the future reception of policies in regards to science, technology and innovation. The use of accessible, often non-technical, language by the media allows to explain and to open the debate about diverse techno-scientific issues to the public. These raise several questions:

- *What happens when certain scientific information about genomics never make the highlights of news media?*
- *How would the public opinion be shaped if only certain types of scientific breakthroughs in human genomics are communicated?*
- *And, in the lack of strong news media efforts to inform citizens, what is the impact of social media on shaping our views and opinions on topics related to gene editing or CRISPR?*
- *Do movies and TV series also have a role in informing societies about gene editing technologies?*

Posing these and other related questions, but also thinking about possible answers and solutions, is paramount, especially now with the rise of fake news and the postulation of the post-truth society[62].

5.1 Online news media

In the last four years, the topic of genomics has been progressively more frequent in the news media as it can be observed in Figure 9. The graph was created based on the results obtained by querying the *European Media Monitor* system. In this particular case, the query was done to analyse traditional news media addressing the subject of genomics in humans with special interest on gene editing and genetic modification technologies.

[62] The post-fact / post-truth society refers to our current modern culture where the gap between truth and lies has shortened to the point where lying became commonplace and tolerated. Facts are presented inaccurately and deception becomes an acceptable modern way of life with little or no consequences for those who lie. See for instance: "The Post-Truth Era: Dishonesty and Deception in Contemporary Life" (Keyes, 2004)

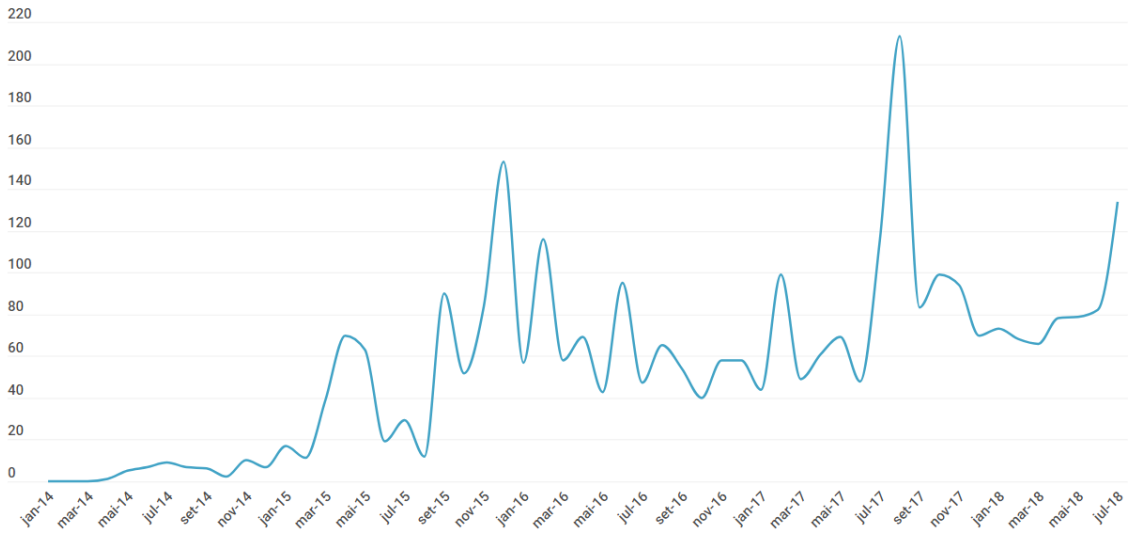

Figure 9: Number of articles published in media from January 2014 to July 2018[63].

By analysing the graph in more detail, it is possible to identify roughly eight peaks that correspond to relevant moments in genomics research, namely:

- April 2015: Chinese scientists edit human embryos for the first time (Cyranoski & Reardon, 2015).
- September 2015: (1) British scientists seek permission to gene edit embryos for research (Briggs, 2015); and (2) the *Hinxton Group* (international group of scientists and policy experts), deemed gene edited research as "*essential*" (Dunham, 2015).
- December 2015: Aftermath of the International Summit on Human Gene Editing convened in Washington D.C., USA, where it was concluded that it would be "irresponsible to proceed with any attempt to create a pregnancy or a baby from human eggs, sperm or embryos that have been altered, because of safety and ethical concerns." (Associated Press in Washington, 2015).
- February 2016: Britain gives scientists approval to conduct gene-editing experiments on human embryos (Callaway, 2016).
- June 2016: First human CRISPR therapy reviewed and given go-ahead in the USA (Le Page, 2016).
- February 2017: Publication of a report by the *National Academies of Sciences* and the *National Academy of Medicine* stating that scientific advances make gene editing in human reproductive cells a "*realistic possibility that deserves serious consideration*", giving a green light to gene editing therapies in germ cells to correct disease or disability (Steenhuysen, 2017).
- July/August 2017: First gene-editing experiment successfully carried out in the USA editing the DNA of human embryos to erase a heritable heart condition (Cha, 2017).
- July 2018: *Nuffield Council on Bioethics* publishes a report stating that there is nothing morally wrong with choosing the characteristics of your child (see for instance Matthews-King, 2018)

[63] Data retrieved on 13th August 2018 using the European Media Monitor (http://emm.newsbrief.eu/overview.html).

The increasing interest of the news media on genomics offers the possibility for the public to be informed and follow recent scientific and technological advances. The accurate transmission of scientific knowledge and information is thus critical but so it is the understanding of possible social and ethical implications. Even though the potential social impact of genomics should also be part of the common interest, unfortunately, that type of discussion is frequently absent or not enough critical. In addition, there have been cases where journalists end up misinforming the public by omitting relevant facts that can lead to misconceptions about the applicability of genetics research, by delivering stories that lack quality or do not have enough references to provide the full picture of the topic in discussion (Geller, Bernhardt, & Holtzman, 2002; Robinson, Coutinho, Bryden, & McKee, 2013). This can be due to a lack of understanding of the scientific evidence or by failing to present the information in an accessible and understandable manner. In other cases, scientific information does not seem to be relevant enough to be taken by news media. This was the case, for example, observed in Japan where a study demonstrated a "*significant gap between newspaper coverage and scientific articles in scientific journals*" (Sugawara, Narimatsu, & Fukao, 2012, p. 1).

The positive bias of journalists towards the benefits of novel genomics technologies and treatments when describing the potential outcomes of genetic research has also been noticed. Ostergren, Dingel, McCormick, & Koenig (2015) criticise this positive bias, stating that the "*unwarranted optimism can be misleading*", adverting that the media should strive for an equilibrium between tempered enthusiasm and caution through improved communication with scientists. Akin, an article by Marcon *et al.* (2018) showed how the term "*personalised/precise medicine*" was associated to optimistic stories and to a positive healthcare trend in the news media (verified in 82% of the news articles analysed). Only 6% of the news articles analysed in this study raised critics about the lack of data surrounding the terminology. Marcon *et al.* research intended to demonstrate how the news coverage of scientific issues can have implications on the perception of consumers, public expectations, and ultimately on policies. In addition, it can also serve to illustrate how easy the promotion of DTC-GT products can be which are usually marketed as the next step towards personalised medicine.

There are also good examples of the news media articles and documentaries resonating with scientific evidence on the topics of human gene-editing. This is for example, the case of the video documentary "*Futureproof: Gene editing and the rise of the home DNA test*" by Reuters News (Reuters, 2017), or the BBC News radio episode on gene editing in embryos and the ethical considerations of creating "designer babies" on the radio show "*Everyday Ethics*" (BBC, 2018), broadcasted after the publication of the *Nuffield* report on genome editing and human reproduction.

5.2 Social media

The importance and relevance of social media platforms such as Twitter and Facebook as an instrument for delivering and spreading information to certain publics is well-known and acknowledged today. Indeed, social media gave "voice" to millions of people by enabling them to spread the information they feel is relevant. However, it also allowed for companies to easily target their consumers with their products.

Using the Altmetric[64] tool, we looked at the number of times the word "*genomics*" has been used in different social media platforms, with special focus on Twitter, Facebook, and Google+. In Figure 10, it is possible to observe, for the Twitter platform, how the contextualised use of the term "*genomics*" has been gradually growing over the years. This growth is not so keen in the other social media platforms (Figure 11 and Figure 12). The overall number of posts shared in Facebook and Google+ addressing the topic of genomics is also smaller.

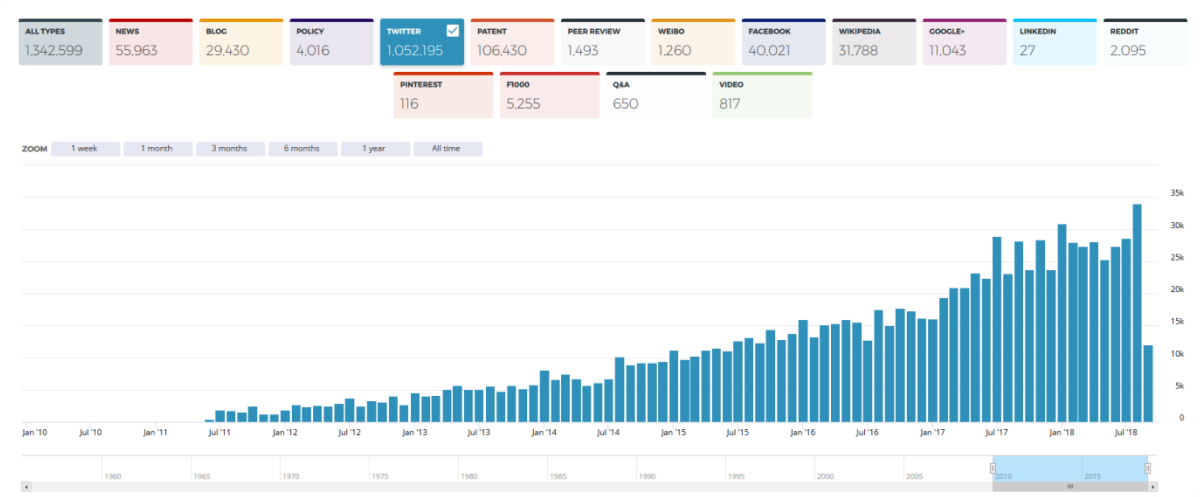

Figure 10: Use of the term *Genomics* in Twitter (2010-2018).

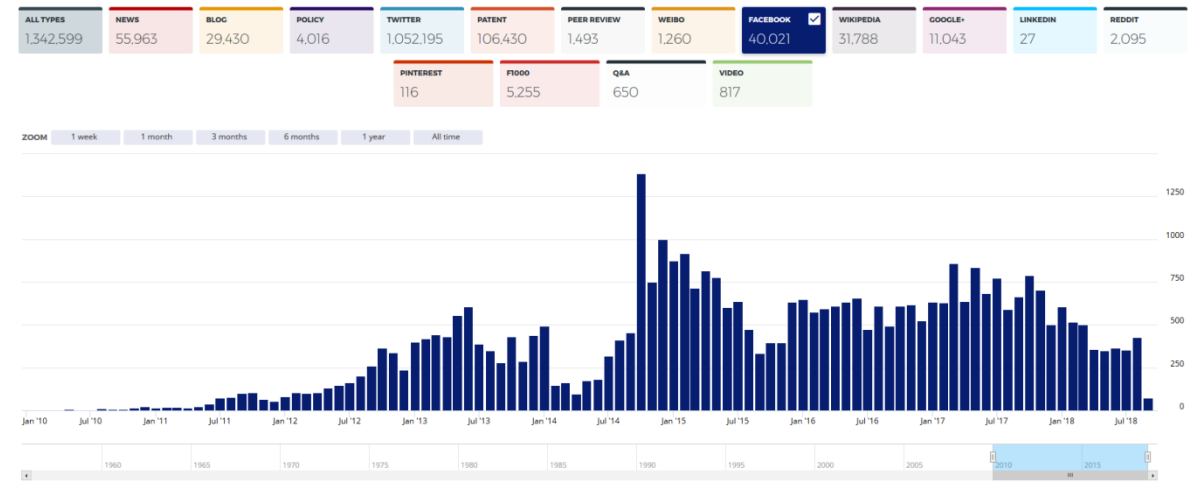

Figure 11: Use of the term *Genomics* in Facebook (2010-2018).

[64] https://www.altmetric.com/

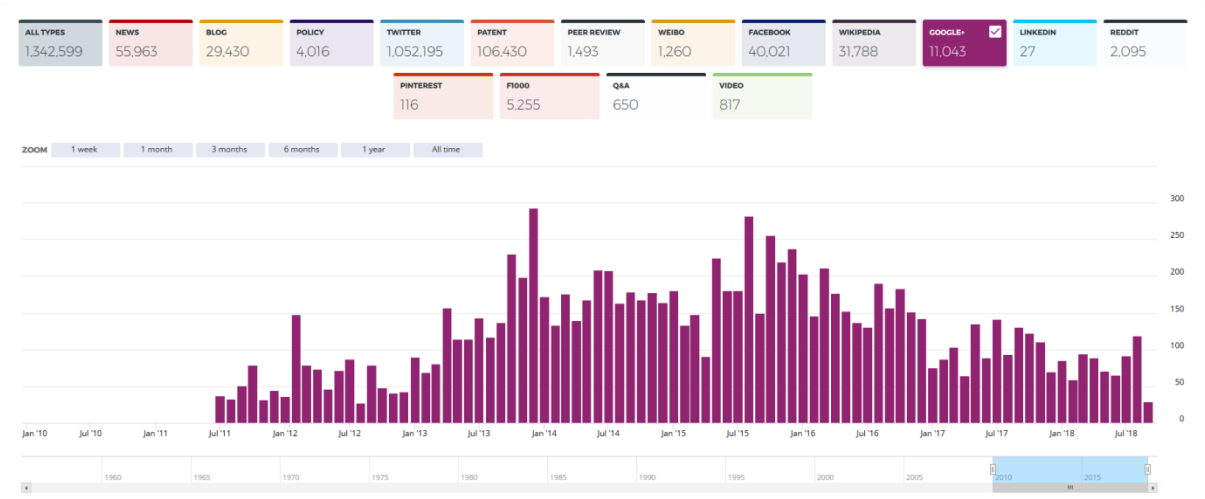

Figure 12: Use of the term *Genomics* in Google+ (2010-2018).

Given the growing usage of social media platforms for spreading genomics related news and articles, it becomes also important to reflect how social media are influencing and shaping our views on the topic. Moreover, it becomes increasingly relevant to understand who is indeed influencing us and why. Hence, it is important to understand where the information comes from, and also to critically think about what is being presented.

5.3 Books, movies, and TV series

Nowadays, the majority of the population gets their first introduction to novel techno-scientific breakthroughs through television programs, popular series or cult films. Books that included genetic narratives appeared as soon as the 19th century in the literary world. These books provided entertainment to the masses but were also the initiators of the public debate regarding the strong political, ethical and social implications of genetics: a debate that initially started by posing the question of *if,* and quickly shifted towards a *when* scenario.

Frankenstein (1818) by Mary Shelley can be pointed as one of the early examples of science fiction novels portraying a scientist conducting unorthodox scientific experiments for artificially creating life. Other well-known books are *The Island of Dr Moreau* (1896) and *Jurassic Park* (1990), which depict a world where humans could manipulate nature to its own accord. Besides showing a dystopian outcome that resulted from manipulating genomes, it also raised ethical concerns as if it is ethical to revive extinct species for human entertainment purposes, and how much responsibility do humans have on the fate and treatment of laboratory animals. *A Brave New World* (1931) presents a world where society is structured based on your genetic manipulation, whereas the trilogy of the *Atlantis Gene* (2013) uses science-fiction together with genomics to explain alternative origins of human evolution, human enhancement and pandemics. Both books, even though published far from each other in terms of year, bring to the table the concept of eugenics and which type of genetic profiles should/could survive and/or dominate earth or even the galaxy. *Jar City* (2002) is a book that fits in the interface of science fiction and an open critic to an organisation dealing with genomics (Decode Genetics Inc.). More critical books such as *Home Deus*, a best seller from

2017, described and challenged the ethical and social impacts of human enhancement, artificial intelligence and genomics pushing the public to question and practice foresight.

Themes such as human cloning, mutation and hybridisation are also widely used as plot devices in science-fiction movies. For example, the movie *The Fly* (1996) depicts the life and physical transformation of a scientist after his genes are mixed with the genes of a fly. Similarly, the movie *Splice* (2009) shows what could happen after merging human DNA with animal DNA in order to create a new species, and the mayhem generated by its escape. Movies such as *Blade Runner* (1982) and *The Island* (2005) critically address the topic of cloning, portraying a scenario where the engineered human confronts his/her creator. *X-men* (2000-2018), both the comic book series and the movie franchise, portrays a world where a group of humans has reached the next step in evolution by having their genes evolved and now have superhuman powers that can also include altering their genetic information and taking the shape of any human. *Rampage* (2018) is a movie that sells based in the popular view of genetic experiments going awry and mutating species into raging creatures of enormous sizes creating havoc. Similarly to the book *A Brave New World*, the movie *Gattaca* (1997) portrays a society built around genetic improvements and a person's genetic make-up to determine their worth to society.

Series such as *Orphan black* (2013-2017) show the social conflicts that could arise when cloning humans. The *Black Mirror* (2011-ongoing) series is also well known for creating near-to science fiction episodes where technology is misused. For example, in the episode *USS Callister*, the dangers of having your saliva samples (DNA) misused for dreadful purposes, namely to create virtual clones of the consciousness of different individuals is addressed.

Scientific and technological discoveries can also find their way to the public in another format, namely through satire. Popular examples are late night-talk shows where politics and other relevant issues are discussed. For example, the American talk and news television show *Last Week Tonight with John Oliver* produced an episode around gene editing (LastWeekTonight, 2018) where highly controversial topics were presented in an accurate way with a quota of humour, making it more engaging to the public (by the end of August, the episode had been accessed 4.6 million times).

5.4 DIYbio in the news media

The DIYbio movement has been portrayed in the news media as a "*double-edged sword*": on the one hand, DIYbio "*will enable the democratisation of science*" being seen as a source of technological innovation. On the other hand, as a movement full of risks and few regulations that threatens global safety, to the point that some people believe it could be a place to create bioweapons or originate the next pandemic (Nai, 2018; Olmstead, 2017; Regalado, 2016). News titles such as "*How DIY gene editing could lead to a global catastrophe*" (Baumgaertner, 2018) or "*Amateur biohackers could build a biological weapon*" (Gray, 2016) are examples of the latest case. The response of the DIYbio community has continually been that their work is on "*science outreach and education* [rather] *than the scary-sounding research*" (Grushkin, 2018).

The growing interest of news media in DIYbio is also fuelled by the publicity acts of certain individuals operating in the limits of the community. For example, Josiah Zayner, CEO of *The ODIN* (see section 2.1.4), received a high extension of media attention due to the disputes his company had with the FDA surrounding the sales of genetic engineering kits targeted at products for human consumption (see section

3.3). The self-publication of videos in the YouTube platform where Zayner injected himself with CRISPR in order to genetically modify his muscles (Smalley, 2018) also contributed greatly to his media popularity.

Another example was Aaron Traywick, CEO of the start-up *Ascendance Biomedical*[65]. Traywick self-administered several times with experimental DIY gene therapies. The most know case was during a biohacking conference in the USA, where Traywick injected himself with an experimental herpes treatment developed by his company (Mullin, 2018). In October 2017, *Ascendance Biomedical* was also in the news when it live broadcast one of Traywick's associates, HIV-positive patient Tristan Roberts, self-injecting with an experimental and untested HIV therapy over Facebook's live-streaming service (Lussenhop, 2017). Although neither of the therapies worked, the media coverage was considerable. The death of Aaron Traywick in April 2018 (by accidental drown and not by something related to DIYbio) was intimately connected to the destiny of the DIY Bio as it is shown in the news article : "*What Does an Infamous Biohacker's Death Mean for the Future of DIY Science?*" (Brown, 2018). Again, the implications and possibilities of an unregulated movement stirred the media, leading to public debates regarding the safety of DIYbio initiatives.

On the other side, it is also possible to find positive depictions of the DIYbio community in the media. For example, in an article by *The Guardian*, it is illustrated how London's *biohackers* are conducting novel research and innovating in various fields, from the production of buildings out of fungi, the development of new vegan cosmetics, the creation of sustainable bioplastics for use in the fashion industry, and the conception of systems that extract nutrients and energy from wastewater (Ireland, 2018).

[65] https://ascendance.io/

5.5 Chapter summary

The key findings of this chapter are:

- An increased general interest from the media in the genomic field can be identified, in particular regarding gene editing experiments in human embryos and human genetic experiments.
- Journalism presenting an optimistic portrayal of certain topics such as personalised medicine, sometimes can lead to a misleading enthusiasm and to a subsequent misinformation.
- Scientific accuracy is still portrayed in several journal articles as a relevant subject to be explored and studied.
- The potential social impact of genomics should also be part of the common interest, unfortunately, that type of discussion is frequently absent or not enough critical in the news.
- The importance and influence of social media in citizens' views on human genomics is evident. Hence, it is becoming increasingly important to evaluate the quality of information shared on social media and its impact in the public.
- Books, movies and TV series can influence people perspectives and guide their interest into diverse scientific topics and make them think about possible implications of genetic advances in science and society.
- Scientific and technological discoveries can also find their way to the public through podcasts, documentaries and late night-talk shows where politics and other relevant issues are discussed.
- The DIYbio community has been portrayed in the media at two different extremes, as the actors that democratize science but also as a latent risk for society.

6. Narratives around human genomics

The work developed in section 2 allowed us to classify and better understand the promises, claims and assumptions created by each type of businesses. Based on the information collected, we compiled in Table 9 the main narratives used by the various types of genomics companies for acquiring the trust and approval of citizens. Likewise, we extracted the narratives put forward by online news, bestselling books, blockbuster movies, and mainstream TV Series (Table 10). In this analysis, it is also included narratives used to describe the DIYbio movement. The compilation of these narratives enables us to compare and see how these diverge and converge with the aim to use them to create improved citizen engagement initiatives. It is worth to keep in mind that the research work compiled and presented in both tables was done using sources written in English.

6.1 Narratives from companies

Table 9: Main narratives explored by companies when marketing their genomics related products/services.

Companies	Narratives
Companies selling genomic material for academic research	– Unlock the power of DNA. – Improve your health. – Personalised healthcare. – Extend life spam of patients. – Innovation for cancer drug development. – Analyse DNA samples anywhere, whoever you are. – Produce agricultural products do not exist.
Companies offering gene therapy services	– Harness the power of genome editing. – We revolutionise medicine. – We create gene-based medicine. – Cure diseases at the molecular level. – Treat the genetic cause and not only the symptoms. – We work together with a myriad of stakeholders. – We treat only somatic cells, not embryonic cells.
Companies that offer direct-to-consumer genetic testing	– No one knows you better than your genes, and knowledge is power. – Screen your DNA against common conditions (diseases) so you can live a healthier life by avoiding certain behaviours. – Reduce genetic risks and create a plan with your doctor where him/her knows your genetically-influenced response to certain medications. – Women are responsible for their families' fate by accessing early awareness about genetic conditions. – Democratise genomic studies to achieve health outcomes for everyone because diverse populations hold different mutations. – You can empower your employees on a truly personalised level by motivating them to have healthier lifestyle after analysing their genetic insights. – What is the role your DNA may play in your personality? Test your genes for tolerance, stress, impulsive behaviour, empathy and euphoria. – Compare your DNA to famous football players to see which type of nutrition and exercises are better to do considering your genetic profile. – Be part of a scientific study to better understand how our ancestor migrated from Africa to populate the world. – How much genetic data you share with your relatives, or with a Neanderthal? Find relatives (we connect you with other customers that share DNA with you).

	– We hold the largest database of African Lineages and ethnic groups. – We analyse your DNA so we can create cosmetics tailored to your DNA results.
Companies that buy/sell genetic data	– Using crowd-sourced data we are able to find genetic links to depression. – The crowd-sourced collection of genetic data has revolutionised our ability to detect subtle genetic effects on complex diseases. – Thanks to the recruitment of 25,000 participants 23andMe we have a flying start to huge depression/genetics-study. – Our collaboration with 23andMe has led to the discovery of 15 regions of the human genome that appear to be linked to major depressive disorder.
Companies that monetise genetic data using blockchain and cryptocurrencies technologies	– The Future of Genomic Data Encryption. – We are a decentralised market of genomic data and services. – Protect, share and sell your genomic data. – We will promote the de-monopolisation of DTC-GT companies profiting from selling their users genomic data. – We will help to establish equal conditions for drug development. – Eliminate companies acting as intermediary between data owners and data buyers. – We are established as a Public Benefit and are Community-owned. – The public can share their genomic and health information “for the greater good of the community”.
Companies that store genetic data	– Power up your science and organise the world’s genomic information and make it accessible and useful. – Simplify and securely scale genomic analysis. – Access the performance and scalability of a world-class supercomputing centre. – Ask bigger questions by efficiently processing petabytes of genomic data. – Efficiently and dynamically store and compute your data, collaborate with peers, and integrate your findings into clinical practice. – Innovate and collaborate with colleagues around the world. – Reduce time to insight and increase the pace of scientific discovery. – Accelerate scientific discovery with fast, secure, and collaborative genomic and healthcare data analytics.
Companies that provide deep analysis through Artificial Intelligence of genetic data	– Creating a new Universe of genetic medicines together with AI and automation. – The future of medicine will rely on artificial intelligence because Biology is too complex for humans to understand. – Machine learning improves hit rates for drug discovery by up to 10,000 times. – We are able to deliver accuracy comparable to wet lab experiments. – We offer high throughput screening (10 and 20 million compounds a day). – You can generate new hypotheses using our evidence-backed predictions which are generated by analysing multiple sets of life sciences knowledge. – We analyse the massive amount of knowledge produced in a more comprehensively and faster way than simple search tools. – By making machines intelligent we can improve people’s lives.
Companies that sell kits for DIYbio	– We make science fun, accessible and affordable. – It is as easy as learning maths, programming or even cooking. – You can genetically engineer a human on your own garages. – You can create CRISPR DNA to target your own genes. – We can find cures to diseases because we are not regulated as universities are. – The most innovative solutions will come from citizens doing biotechnological experiments in their own garages. – The future is going to be dominated by genetic engineering and consumer genetic design will be a big part of that.

6.2 Narratives from the media

Table 10: Main narratives explored by the media when addressing genomics related products, services and developments.

Companies	Narratives
Online news about genomics, gene editing, and direct to consumer genetic testing	– The safety of CRISPR-Cas9 gene editing is being debated but the caveats are attainable. – How knowing your genetic code could lengthen your life. – Personalised Medicine made available in a way that each person can have their own treatment according to their genetic profile. – Misdiagnoses: A hidden risk of genetic testing. – Knowing you genetic information may impact your lifestyle and psychological health. – Genetic makeup dictates your worth as an employee or mate. – Genetic disease risk screening is becoming a popular employee benefit. But the tests may not be all that beneficial for the general population. – Why did I risk my privacy with home DNA testing? – Gene editing opens world of 'man-gods'.
Books, Movies, and TV Series in relation to genomics	– Humans could manipulate nature to its own accord and entertainment unleashing a grim future. – Society can be better structured if based on genetic manipulation. – Genetic storage of citizens could allow the identification of murderers, rapists or serial killers. – Transhumanism can be achieved faster through genomics. – A society can be built around genetic improvements and a person's genetic makeup. – Who will survive and rule the world, humans or genetically modified/edited organisms? – Genetic experimentation in animals can lead to mutating species into creatures that are dangerous for humans. – Collection of DNA samples for genetic testing can lead to the creation of virtual clones of people's consciousness.
Online news about DIYbio	– As DIY gene editing gains popularity, "someone is going to get hurt". – Citizens start taking uncalculated, or misinformed, risks for personal entertainment or publicity stunts. – Become a strongly regulated or completely forbidden movement in the context of biological experiments. – Becomes a source of technological innovations cost-effective and context-appropriate. – Can become a bridge between scientific knowledge and the layman. – The public will be able to use these techniques to whatever uses they want - the free use of biology. – Can be taken advantage by bioterrorist threatening the public.

6.3 Exploring new narratives on human genomics

Based on the work presented in Table 9 and Table 10, we extracted the main recurring themes from the various contributions, deriving a subset of seven narratives (Table 11). In the context of this report, we present these seven narratives as discussion cases to be used in social research enquiries in order to explore the desirability and plausibility, as well as relevance, in relation to possible scientific and technological advances in the genomics field.

Table 11: Exploratory narratives produced based on the analysis of the narratives put forward by companies and media in relation to genomics.

Title	Narrative
Personalisation of medicine and drugs	Advances in the genomics field enable de design of medicines and drugs that are specifically tailored to your genetic code. Perfect diagnoses of diseases and optimisation of medication and treatment are a reality. Diseases and conditions that here previously considered fatal or untreatable are eradicated. As a result, the quality of life and expectancy of humans increases drastically.
Ready-to-use gene manipulation kits	Ready-to-use gene manipulation kits are sold in global supermarkets retail chains. Individuals gain widespread and unrestraint access to gene manipulation tools, enabling them to easily tinker with their own DNA from the comfort of their home.
Selection of genetic characteristics	The selection of physical, social and cognitive genetic traits on embryos becomes a reality. Parents are able to choose not only their new-born colour eyes and sex but also eliminate genetic diseases and reduce susceptibilities to alcohol and drugs.
Personal card of genetic data	Genetic data becomes part of every citizen identity card. Individuals have their genetic information readily available, which is used to gain admittance to certain social benefits. Genetic profiling is also used to concede access to certain education programs, jobs, insurance packages, and medicines.
Monetisation of genetic data	Genetic data is raised to the status of currency in society. Individuals are able to use their genetic information as currency to purchase goods and services from companies specialised in harvesting genetic information. The high profitability of such business inevitably leads to the creation of parallel markets that sell genetic data of individuals illegally obtained.
Merging of genomics and machine-learning	The analysis of large data sets of genetic data from the world population by advance machine-learning algorithms greatly accelerates the impacts of genetic research on society. The identification and interpretation of trends and variations in the world population genetic data by machine-learning changes the notion of what it means to be human: beyond genomic predictions, it is possible now to know the answer to the meaning of life.
Gene-tailored bio-terrorism weapons	The threat of bio-terrorism is high. The widespread availability of low-cost genetic engineering technologies and knowledge on the topic facilitates the development by anyone of tailored weapons based on genetic profiles of individuals or specific populations. It also becomes impossible to distinguish between naturally occurring epidemics and alleged tailored biological attacks.

7. Citizen engagement activities

Collectively reflecting on and anticipating social impacts that might arise from current scientific and technological innovations is essential to the creation of trusting relationships among citizens and the institutions that govern them. As depicted in this report, genomics technologies present an increasing variety of applications and impact multiple technological and social fields. With this in mind, it is of interest to understand *which formats* of citizens engagement activities were carried out in this field, *how* they were implemented and how they influence *policy making* (if at all).

Within the European Commission, the "*Science with and for Society*" initiative (2014 -to date) proposes that all societal actors (researchers, citizens, policy makers, business, third sector organisations, etc.) work together during the whole research and innovation process. The Responsible Research and Innovation lemma that permeates the Horizon2020 research framework recognises as well dimensions of quality assurance of science, technology and innovation that are beyond the traditional framings of excellence (European Commission, n.d.). Even so, the topic of human genomics, in particular genetic testing and screening, genetic patents, genetic databanks, human enhancement and human-machine interactions is addressed solely in the Horizon2020 call entitled "*The Ethics of technologies with high socio-economic impact and Human Rights relevance*" (European Commission, 2017c). The aim of the call was to build concrete ways of "*better reconciling the needs of the research teams and the legitimate concerns of the citizens while stimulating innovation and contributing to the reduction of socio-economic inequalities including in health treatment and social status*" which ideally should result in "*the creation, with operational guidelines for research ethics committees as well as proposing a code of responsible conduct for researchers, taking into account the expectations of the different stakeholders*" (European Commission, 2017a, p. 37). The only project funded in this call was the "*Stakeholder-informed ethics for new technologies with high socio-economic and human rights impact project (SIENNA)*" which focuses on ethical and human rights challenges posed by human genomics, human enhancement and human-machine interaction technologies (European Commission, 2017b).

7.1 Why citizen engagement in human genomics is important

Literature review shows there are already calls from medical ethics academics to increase public participation[66] when debating research or policies in human genomics, highlighting its added value. Schicktanz, Schweda, & Wynne (2012) point out that "*the implementation and further development of inclusive and reasonable forms of public exchange constitute a central bioethical concern*" whilst Lemke *et al.* (2010) state that "[community engagement] *can enhance research quality, improve protection of participants, and address local public health needs*". Other reasons advanced by other scholars in the fields of health and ethics issues include:

- Increase legitimacy (Deverka et al., 2012; Schicktanz *et al.*, 2012);
- Ensure a responsive approach to public concerns (Terry *et al.*, 2012);
- Answering faster and comprehensively identify emergent concerns (Murphy, Garrett, Boyd, Dry, & Dohan, 2017);

[66] In this report we use the term citizen engagement and public participation interchangeably.

- Address this concerns in a responsive manner (Murphy *et al.*, 2017);
- Measure their impact (Deverka *et al.*, 2012);
- Inform future improvements (Deverka *et al.*, 2012).

In Table 12 we list the most prominent peer-reviewed papers in this topic. The statements presented make a clear call for more open citizen engagement initiatives when debating human genomics.

Table 12: Scientific articles debating citizen engagement in human genomics.

Title and source	Highlight
Rethink public engagement for gene editing (Burall, 2018).	**Lack of public engagement for altering policymakers and scientists decisions.** "*Over the past three years, thousands of articles have been published about editing genes and genomes. Apart from a public dialogue run by the Royal Society at the end of last year, there's been little attempt to engage the public on the implications of the technology in a way that could alter the decisions of scientists and policymakers*"
U.S. attitudes on human genome editing (Scheufele et al., 2017).	**Need for increasing the diversity of evaluators.** "*In sum, our findings show a broad mandate for public engagement, even across groups who otherwise differ in their evaluation of potential applications of human genome editing and in their assessment of the scientific community's ability to navigate emerging science independently of public input.*"
Engaging patients in health research: identifying research priorities through community town halls (Etchegary et al., 2017).	**Patients can help planning and aid priority-setting process of research.** "*Public research priorities and suggestions for improving the provision of healthcare provide valuable information to guide Support Units' planning and priority-setting processes. A range of research areas were raised as priorities for patients that are likely comparable to other healthcare systems. These create a number of health research questions that would be in line with public priorities. Findings also provide lessons learned for others and add to the evidence base on patient engagement methods.*"
Community engagement with genetics: public perceptions and expectations about genetics research (Etchegary, Green, Parfrey, Street, & Pullman, 2015).	**Bi-directional communication with the public enhances the research process.** "*If researchers can engage the public in regular, transparent dialogue, areas of public (and researcher) concern could be identified and discussed. Such two-way communication could help open the way for greater understanding of the research process and the design of efficient and effective genetic health services, informed by the public that will use them.*"

Akin, the following examples of citizen engagement initiatives in relation to human genomics (from around the world) provide some insights and recommendations. It is perceptive in present bibliography that the most well reported initiatives are projects targeting biobanks[67] as they actively engaged with the community in order to thrive.

[67] Biobanks: Biorepository of biological samples (usually human) for use in research which are gathered from donors.

Table 13: Citizen Engagement case example – "Let's talk about Genomics".

Initiative Name	**Round Table: "Let's talk about Genomics"**
Organisers	Department of Social Studies of Science. University of Vienna (Austria)
Date	N/A
Location	University of Vienna, Austria.
Scope	– Investigate hypotheses about participatory events by modifying some key elements and dimensions of classical citizen engagement designs. – Observe how social relations between scientists and citizens would develop, and how they affect citizens' evaluation of the ideal of scientific self-governance and how the scientists view on the idea of public participation is influenced. – Understand how the participants would discuss 'upstream issues', such as the "*values underlying specific techno scientific trajectories, and how they would imagine possibilities and limits of governing them*".
Methodology	– Adaptation of the Swiss foundation Science et Cité: Round Tables where the "*precise structure of the engagement design is not predefined, but may and should be developed by the participants in the ongoing process*". – Qualitative Interviews.
Participants	– 13 scientists and 14 citizens. – Heterogeneous group of citizens with a very even distribution concerning age and gender, however bias towards higher education levels (only one below A-level). – From the total of 14 citizens, four were between 18-30 years old, five 31-45, four 46-60 and one above sixty.
Outcomes	– "*Engagement with the scientists actually doing the research may be crucial for building sustainable relations between technoscience and publics. Proxy labs and public relation experts will not suffice, because they do not allow the public to test whether the ethos displayed on these 'facades' is actually 'incorporated' by the researchers themselves.*" – "*If science is to meaningfully contribute to the growing debates on innovation governance, it will have to adopt a more integrated vision of science's role in society. For the single scientist, this may imply more actively taking the hybrid role of the scientist/citizen concerned about the impacts of scientific knowledge beyond the narrow short-term risks of his or her own work.*"
Challenges	– Gender ratio challenges reflected on the hierarchical position (project heads were male while nearly all doctoral students were female). – Younger students were less likely to contribute (unless addressed) than senior counterparts.
Recommendations	– Do an open national wide call for attracting citizens. – Do an internal call for recruiting scientists; make sure to have a heterogeneous group from doctoral students to project leaders. – Make sure to have a heterogeneous selection of participants taking into account gender, education and age. – Use the scientists' research projects as an example to discuss the social and ethical aspects of genome research. – Let both the citizens and the scientists to equally engage in the process of public engagement. – Allow time for citizens and scientists to interact and to reflect on the discussions. – Make use of a round table where participants engage in discussing the social and ethical aspects of the research project of the scientists. Besides the round table make sure to also have small group discussions. – Number of sessions = six whole-day sessions over the period of eight months where the first three are dedicated to discussing the research project itself including a laboratory visit. – Let the participants collectively identified topics to be discussed in the remaining three meetings. Some examples are science and the media, ethical issues of genome research and regulatory issues. – Invite experts for each topic, for example a journalist, an ethicist, a policymaker. – At the seventh meeting give autonomy to the lay-participants to reflect on their experiences on a broader level. – Conduct qualitative interviews with all participants at the beginning and at the end of the citizen engagement initiative.
Source	(Felt & Fochler, 2008)

Table 14: Citizen Engagement case example – "Deliberative public forum on biobanking".

Initiative Name	**Deliberative public forum on biobanking**
Organisers	Office of Population Health Genomics, in collaboration with a team from the University of British Colombia and local experts (deliberative facilitation, law, bioethics, anthropology and political philosophy).
Date	2008
Location	Perth, Australia.
Scope	Gather a small group of citizens (15-20) to create a democratic and deliberative exercise.
Methodology	– 4-day forum over 2 weeks. – 36 page booklet with literature review delivered to the participants prior to the forum. – 13 interviews performed and transcripts done in the following 2 weeks of the forum.
Participants	15 citizens made recommendations (+1 researcher)
Outcomes	– Although limited diversity, the perspectives presented were different between same social-demographics, and the forum results were translated to policy recommendation (facilitated due to the recommendations elaborated being similar to the existing OECD guidelines). – Draft policy was elaborated and circulated among the participants, which was further used to create the guidelines for bio banking. – Inclusion of the participants' recommendations to the already followed OECD guidelines by direct or indirect reference.
Challenges	Lack of social-demographic diversity
Recommendations	Implement multiple forum session to increase the number of participant citizens (when cost are not prohibitive).
Source	(Molster et al., 2013)

Table 15: Citizen Engagement case example – "CARTaGENE".

Initiative Name	**CARTaGENE**
Organisers	Quebec CARTaGENE Project
Date	2001 and 2003
Location	Canada, Quebec (four regions).
Scope	– Report public concerns surrounding biobanking. – Report social-ethical implications and social perceptions of the Project.
Methodology	– Qualitative Phase: 23 focus groups of 7-8 – Quantitative Phase: interviews with 1800 respondents
Participants	Individuals aged 25-75; random sample stakeholders; 18 regions of the province
Outcomes	Reports of consultation: – Confidentiality concerns and ensuring of diversity of the population; – Transparency in the handling of data; – The right to feedback of the research results; – Right to withdraw from the project Commercialization.
Challenges	– Difficulty to access policy implications. – Religious, cultural and linguistic barriers.
Recommendations	– Targeted communications with appropriate information-communication ratio. – Considering community's values and personal interests.
Source	(Godard, Marshall, & Laberge, 2007)

Table 16: Citizen Engagement case example – "Deliberative public forum on biobanking".

Initiative Name	**Public Engagement in Genetic Variation and Haplotype Mapping Issues Project (PEGV)**
Organisers	University of Michigan School of Public Health in collaboration with Genetic Alliance (conceptualisation); Iona College (liberal arts college) and Village Team Project (members include New Rochelle's police, fire and school departments, members of various religious communities, drug rehab centres, and healthcare professionals); Community Advisory Committee members; two outside academics.
Date	2003 to 2004
Location	New Rochelle, New York, USA.
Scope	– Test a community engagement model; – Document concerns about genetic variation research – Make recommendations designed to address these concerns
Methodology	A six stage process: (1) identify possible partnerships in community-based organisations; (2) establish community oversight and advisory boards; (3) develop an initial list of genetics issues from focus groups; (4) carry dialogue sessions (2 h) from community-based selected issues of concern; (5) hold a 1-day conference; and (6) Disseminate results.
Participants	38 members of the community, with social, ethnical and age diversity divided in 4 focus-groups
Outcomes	– The objectives of the work were accomplished, and a 176 page report was produced and delivered to the NHGRI's HapMap project team which included a list of recommendations for further genetic research. – Dissemination of finding through conferences and Iona College's website.
Challenges	– Evaluation of participants' satisfaction. – Involve more community members less interested in the thematic. – Maintain momentum after initial enthusiasm in the community.
Recommendations	The PEGV Project provides a model to consider for citizen engagement.
Source	(Terry et al., 2012)

7.2 Main challenges

Our review of citizen engagement initiatives in the human genomics field made us realise *(i)* there are very few activities around the world; *(ii)* only occasionally different stakeholders are included; and *(iii)* most results are not marshalled into policy processes. Furthermore, the specific objectives of each initiative render follow-ups difficult to nearly impossible. As Lemke & Harris-Wai (2015) point out, "*detailed methods and outcomes of stakeholder engagement* [about human genomics] *for policy development are rarely published"* and "*little evidence regarding how to best incorporate stakeholder feedback into policy-making processes is currently available*". Terry *et al.* (2012) provide similar observations regarding the scarce reporting and engaging guidelines: *"many scholars call for increased community engagement and ongoing discussion to ensure that genetic research is responsive to the concerns of the public. However, only a few examples exist that demonstrate how to do so*" (Terry et al., 2012). The aftermath of citizen engagement initiatives is also addressed with the lack of effective measurement tools to evaluate their efficiency: "*Although a number of organisations have set out general principles, frameworks, and best practice for successful community engagements currently no standardised measures exist for evaluating CE* [citizen engagement] *goals and outcomes in biobank-based research"* (Lemke et al., 2010). Moreover, according to Bickerstaff, Lorenzoni, Jones, & Pidgeon (2010) *"society is not responding effectively to the outputs of*

dialogue or internalizing key messages [about human genomics]". Likewise, Hawkins & O'Doherty (2010) point out that "*these engagements are merely used to gain public support of a particular project, and that they fail to use the engagement results to represent public interests in the formation and governance of the projects*" (Hawkins & O'Doherty, 2010). These statements show that the need to improve citizen engagement initiatives so they are seen for what they truly are and contribute to the final goal to aid policymaking and scientific research.

Sperber *et al.* (2017), when performing a multi-analysis of six genomic medicine projects of the National Human Genome Research Institute (NHGRI) network, identified "*engaging patients*" as one of the common challenges. The authors proceed by point out three main strategies to overcome this issue:

- Use of mass media to educate potential users, by promoting news articles in TV, radio, local newspapers, emailing press releases that were forwarded to local and national media, and keeping a robust online presence;
- Involving patients in implementation activities, employing focus panels with patients to create a patient-oriented education tool, giving public speaking opportunities, providing patient engaging materials and strategies;
- Prepare patients to be active participants through tailored approaches, such as risk reports, template appeal letters and booklets

Another study by Avard *et al.* (2009) enumerates six essential issues central to public participation, obtained from comparing three case studies of public participation about public health and genomics:

- To define what is public participation (particularly important to define due to lack of conceptual uniformity at national and international levels);
- The goals of public participation;
- Which public needs to be involved;
- Timing of the involvement;
- Tools to promote public participation;
- Evaluation of public participation.

7.3 Citizen engagement translated into policy

It is paramount to, first, promote citizen engagement initiatives which should aim at providing recommendations to govern the human genomics field; secondly, that these recommendations are actively translated or addressed into policies. Few detectable events were identified on this subject as actively reported. We would like to highlight the outcomes of a study conducted by Lemke *et al.* (2010) that analysed six United States biobanks that have developed community engagement initiatives. Our focus will be only on two of the initiatives:

1. The Mayo Clinic biobank, which succeeded to write specific policy recommendations that have been implemented in the design and operation of the institution. These recommendations made the organisation modify and simplify the consent process; include an option for participants to

withdraw from the biobank; the creation of a community newsletter and website for keeping the participants informed; and promoted the creation of a Community Advisory Board.

2. The Northwestern University biobank initiative for citizen engagement has altered the set of communication material; also it has guided the paediatric expansion of the university and is continuously being a critical component of the programme evaluation and evolution.

Another noteworthy example is a deliberative public forum on biobanking (Molster et al., 2013), which produced a list of deliberated principles and important issues to be addressed as a list of recommendations. Using the OECD guidelines as a template, the recommendations from this forum were included by direct or indirect reference and presented to policy makers to create the Western Australia guidelines for the establishment, governance and use of human biobanks for research purposes. Since then, the document has been local, national and internationally recognized (for more information see Table 14).

Expanding our research work, our suggestions and recommendations for engaging citizens in a regular and transparent dialogue with regards to the topic of human genomics are along the following lines:

- As seen throughout this report, the field of genomics is of great complexity and uncertainty. Citizen engagement initiatives directed at this field, and in particular human genomics, should seek to incorporate the insights and expectations of wider societal collectives in relation to the governance of human genomics techniques, and current and envisaged applications. The dialogue should be opened to different actors, namely to citizens and civil society organisations.

- Citizen engagement initiatives around genomics should mobilise the imagination of those involved in order to understand the potential needs, values, fears and understandings of the publics. Both material participatory methods, such as those based on making and experimentation, as well as discursive participatory methods such as Citizens Juries, Delphi Surveys, Focus groups, 21st Century Town Meetings, Scenario Building Exercises, World Cafés, and Citizens Panels can be deployed as a way to co-create the necessary knowledge to approach the challenges posed by developments and applications of human genomics. Each approach needs to be tailored to local needs and audience.

- Finally, this report can be seen as a source of information regarding the current state-of-the-art in human genomics. The narratives presented in section 6.3 can be used in citizen engagement initiatives to stimulate extended conversations around the topic of human genomics and as source of inspiration about possible re-imaginations of the governance of human genomics.

7.4 Chapter summary

- Not many citizen engagement activities in human genomics were found, suggesting the need for the creation of dialogue spaces.
- There is also a lack of evaluation of the initiatives which could help understand what needs to be debated in this domain with citizens.
- It is not clear if activities identified have contributed to inform or alter scientific practices or policy processes.

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