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How can the Data Governance Act and Data Altruism Organizations facilitate innovation in the rare diseases sector?

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During the preparation of this master's thesis, the author utilized ChatGPT for the following purposes:

1. Synonyms: The ChatGPT tool was used to find synonyms to avoid repeating certain words in the text.

2. Reformulation: The ChatGPT tool was used to help rephrase certain sentences. I always used my own ideas but asked ChatGPT to formulate them properly in some cases. However, I did not use his suggestions as they were. I just based myself on them and adapted them.

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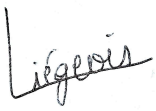
By signing this declaration, I affirm that the content of this master's thesis reflects my original work, augmented by the responsible use of AI.



Sarah Liégeois, 22/07/2024

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List of Acronyms

CEDS	Common European Data Space
DAO	Data Altruism Organization
DGA	Data Governance Act
EHDS	European Health Data Space
EU	European Union
GDPR	General Data Protection Regulation
R&D	Research and Development

Chapter 1: Introduction

1.1 Context

"Patients with rare diseases generally face a poor health status because of the disease itself but also because their health care pathway to accessing appropriate diagnosis and treatment for their condition can be lengthy and complicated." (Ali & Tubeuf, 2019a, p. 2)

In Europe, a rare disease is defined as one that affects less than one person in 2,000 (Rodwell & Aymé, 2015). In 2023, rare diseases affected 3 to 8% of the world population (Gaik & Münster, 2023). The number of rare disease patients was therefore estimated at 446 million worldwide and 30 million in Europe (Gaik & Münster, 2023). For Belgium more precisely, this number is estimated at 700,000 (*Institut des maladies rares*, n.d.). Most of the time, rare diseases have serious consequences on patients' lives but the cause of many of them remains unexplained (Wakap et al., 2019).

People affected suffer severe physical, emotional and family handicaps, and their vital prognosis is very often limited but, at present, none of these diseases has a cure (Lannoy, Vincent, Lohest & Hermans, 2019). Treatment options are very limited, mostly consisting of standard medication to reduce symptoms and provide relief to patients (Lannoy et al., 2019). Moreover, these drugs, known as "Orphan drugs", are very expensive and are only available to less than 5% of patients (Lannoy et al., 2019). The lack of curative treatments and the high price of Orphan drugs are justified by pharmaceutical companies by the rarity of rare diseases and the consequent lack of scientific knowledge and medical data (Ali & Tubeuf, 2019a). There are very few people affected, making it also difficult for pharmaceutical companies to find patients for clinical trials. (Ali & Tubeuf, 2019a). In addition, diagnosing these diseases is equally complex and time-consuming. It takes a very long time to get a precise diagnosis due to diversity and heterogeneity of rare diseases (Lannoy et al., 2019).

Research and innovation in this sector are therefore vital, in order to develop rapid diagnostic methods and more effective treatments for patients affected by a rare disease. But despite this great need of innovation, this remains complicated because of the large number of rare diseases – estimated at 7,000 –, their rarity and their complexity (Ali & Tubeuf, 2019b).

One way of boosting innovation in the rare diseases sector would be to exploit better the potential of health data (Thorogood, 2020). Data are increasingly omnipresent in our society and affect every aspect of our lives because they are at the heart of the digital transformation of our economy. According to the European Commission (n.d.-a), the volume of data available in the European Union is set to increase by 530% between 2018 and 2025. Moreover, data can have an enormous potential as data-driven innovation can bring considerable benefits to the European Union's economy and its citizens (Data Governance Act, 2022). Health data in particular can be the source of incredible benefits, if properly managed. They can be used and reused in ways that speed up research work and thus can help provide better and more effective healthcare (European Health Data Space, 2022). According to European Commission estimates, the reuse of health data is worth between €25 and €30 billion a year and could reach €50 billion over the next decade (European Health Data Space, 2022).

However, this incredible potential of data remains under-exploited, mainly due to regulatory fragmentation between Member States of European Union (EU), preventing optimal use of data (European Health Data Space, 2022). In 2020, the EU decided to take action and drew up a European Strategy for Data, with the aim of exploiting this potential better. In summary, the aim of this strategy is to create a single market for data to enable their flow between EU countries and their sectors, while ensuring data protection in accordance with the General Data Protection Regulation of 2016 (Penedo, 2024). The development of this single market will lead to the creation of various data spaces – the Common European Data Spaces (CEDSs) – in strategic sectors, the most important of which, according to the European Commission, is healthcare (Kari, Schurig & Gersch, 2023). That is why, in May 2022, the European Commission released a legislative proposal for the establishment of the very first CEDS in the healthcare sector: the European Health Data Space (EHDS) (Kari et al., 2023). The aim of the EHDS is to optimize the use of health data within the EU, by establishing an infrastructure and a framework to develop a European digital health industry (Hussein, Scherdel, Nicolet & Martin-Sanchez, 2023). To enable the creation of the different CEDSs and manage the flow of data, the European Commission has proposed a horizontal framework composed of two new legislative instruments: the Data Governance Act and the Data Act. In this research, we will focus on the former which aims to establish a legal framework to ensure the proper use and sharing of data (Data Governance Act, 2022). In the Data Governance Act (DGA), we will be particularly interested in the concepts of Data Altruism and Data Altruism Organizations. Data Altruism can be briefly defined as the willingness of a person, legal or

natural, to share their data in order to serve the general interest (Lalova-Spinks, Meszaros & Huys, 2023). This altruistic purpose may be to support research into healthcare, and particularly into rare diseases, for example. This sharing of personal data for general interest is managed by companies known as Data Altruism Organizations.

1.2 Objective and research question

A link can therefore be made between the lack of innovation in the rare diseases sector and the introduction of the DGA. In short, the aim of this regulation is to create a governance framework for data exchanges within the EU (Data Governance Act, 2022). In our context, it could therefore facilitate the exchange of health data on patients suffering from a rare disease and thus encourage and facilitate innovation in this sector. This shows us that our research topic is interesting, topical and relevant. The DGA defines a number of new concepts and rules for data governance but, for the purposes of our research, we will confine ourselves to the concept of Data Altruism, which may be of interest in our quest to understand how to boost innovation in rare diseases. The question that arises in this research is to what extent the DGA established by the EU can help boost innovation in this sector.

To the best of my knowledge, no research has yet explored the potential benefits that the DGA – and particularly the Data Altruism mechanism – could bring to the field of rare diseases. This report aims to fill this gap by providing a preliminary theoretical analysis of the benefits and challenges of the implementation of the DGA in this sector. This work will serve as a theoretical foundation for future empirical studies, aimed at verifying and deepening the hypotheses and proposals put forward here in the conclusion. In this sense, this research is an essential first step in understanding the potential impact of the DGA on innovation in the treatment of rare diseases.

This work aims to enrich academic research, but also to raise awareness of the lack of innovation in the field of rare diseases and the profound distress felt by patients suffering from one of these diseases and their families. It calls for concrete actions to improve research and investment in medical innovation to develop more efficient healthcare.

The ultimate objective of this master's thesis is to answer the following research question: “How can the Data Governance Act and Data Altruism Organizations facilitate innovation in the rare diseases sector?”.

1.3 Methodology, results and structure

As mentioned above, to date, no research has been carried out in this specific area, partly due to the recent development and implementation of the DGA. This is why the nature of this research is primarily exploratory and the method of gathering information will be based on a systematic review of the existing literature. Our research will therefore be more theoretical and be based on a qualitative study of the potential benefits and challenges/risks of implementing the DGA, with a particular focus on innovation in the context of rare diseases.

To answer our research question, we will use the analysis of Chapters 4 and 5, each including respectively the potential benefits and challenges/risks of the DGA in the rare diseases sector. Comparing the two chapters will therefore enable us to answer our research question. The approach chosen for our research is deductive. We have to deduce for ourselves some of the benefits and challenges of the DGA in order to answer our research question given that there is currently little or no explicit literature linking the DGA to rare diseases. Our deductive reasoning will be based on a series of publications demonstrating the benefits of health data sharing to support research and innovation in the pharmaceutical sector. On this basis, we will deduce the benefits of the DGA for rare diseases, highlighting that the aim of the DGA is to increase confidence in, and therefore encourage, data sharing.

Following this methodology, our analysis will allow us to conclude that the implementation of the DGA and the introduction of the Data Altruism mechanism can offer significant opportunities for innovation in rare diseases, thanks to the benefits of health data sharing that it allows and regulates, including the facilitation of clinical trials, collaboration and the reduction of redundancies and costs. However, certain challenges and risks associated with the DGA could hinder and complicate the health data sharing process and require action from different stakeholders, including the lack of clarity regarding consent, the potential lack of trust in data sharing and the risk of fragmentation.

This master's thesis is divided into six chapters, the first of which is the introduction. The second chapter will introduce all the central concepts of our research: innovation, rare diseases, the European Strategy for Data, the Data Governance Act, Data Altruism and Data Altruism Organizations. This chapter will also include some key definitions about data and an explanation of their enormous potential. Next, Chapter 3 will detail our research methodology to answer our question, before moving on to our analysis which will be contained in Chapters 4 and 5. Chapter 4 will aim to define the potential benefits of the DGA for rare diseases, while Chapter 5 will identify the challenges and risks associated with its implementation. Finally, Chapter 6 will conclude this work by answering the research question, and identifying some managerial implications, limitations and avenues for future work.

Chapter 2: General concepts and definitions

Before answering our research question, it is important to understand all the concepts that will be used in this work. This chapter will therefore define all the terms central to our research and the methodology used to gather this information.

2.1 Methodology

The methodology used to write this theoretical section was essentially based on the study of numerous peer-reviewed scientific articles, which were collected using various search engines, including Discovery, Google Scholar, ProQuest, Dial, …

An essential part of this literature review is also based on several legislative documents and communications from the EU, all available on the EUR-Lex website[1]. The main ones used in this thesis are: Regulation (EU) 2022/868[2], COM/2020/66[3], Regulation (EU) 2016/679[4] and

[1] https://eur-lex.europa.eu/homepage.html?locale=fr
[2] Regulation (EU) 2022/868 of the European Parliament and of the Council of 30 May 2022 on European data governance and amending Regulation (EU) 2018/1724 (Data Governance Act).
[3] Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions: A European strategy for data.
[4] Regulation (EU) 2016/679 of the European Parliament and of the Council of 27 April 2016 on the protection of natural persons with regard to the processing of personal data and on the free movement of such data, and repealing Directive 95/46/EC (General Data Protection Regulation).

COM/2022/196[5]. For the sake of simplicity, they will be referred to respectively as follows in the remainder of the thesis: Data Governance Act, European Strategy for Data, General Data Protection Regulation and European Health Data Space.

Finally, an asset in writing this master's thesis was the reading of Benoît Gailly's book: "Navigating Innovation: How to Identify, Prioritize and Capture Opportunities for Strategic Success", published in 2018 and read during the academic year 2022-2023. This book explores various aspects of innovation and offers practical advice on how to navigate the innovation process successfully.

The following theoretical part will be divided into five main parts. First, we will define what constitutes the basis of our work: innovation. Next, Section 2.3 will define the central concept of this research: rare diseases. In this section, we will also look at the main challenges of rare diseases, i.e., the difficulties of innovation in this sector and thus the difficulties in finding the most effective diagnostic methods and treatments for patients. Section 2.4 presents important definitions concerning data, as well as an explanation of their potential in order to better understand the context and the reason of the research. The following part will provide an overview of the European Strategy for Data, which is at the origin of the Data Governance Act. Finally, the last part of this review will explain this regulation and more precisely the concepts of Data Altruism and Data Altruism Organizations.

2.2 Innovation

2.2.1 Definition

The term "innovation" has many different definitions because it is rather complicated to define. Baregheh, Rowley & Sambrook (2009) carried out a study aimed at analyzing different definitions of innovation in order to propose a new version bringing together the various collected definitions. They propose the following definition: "Innovation is the multi-stage process whereby organizations transform ideas into new/improved products, services or processes, in order to advance, compete and differentiate themselves successfully in their marketplace" (Baregheg et al., 2009, p. 1334). This definition is consistent with that of Gailly,

[5] Communication from the Commission to the European Parliament and the Council. A European Health Data Space: harnessing the power of health data for people, patients and innovation.

who states that “innovation can be defined as the combination of newness and change, or as a change toward something new” (Gailly, 2018, p. 14). As part of our research into rare diseases, innovation is essential to develop diagnostic methods and treatments for these diseases, for which there are as yet no curative solutions. However, innovation in this sector is particularly complex due to a number of factors, which will be explained in Section 2.3.2.

2.2.2 Innovation management

The literature highlights the benefits of data sharing for innovation and demonstrates that, in addition to advancing innovation, data sharing can also benefit society as a whole (Barczak, Hopp, Kaminski, Piller & Pruschak, 2021). But before demonstrating this and applying it to the field of rare diseases, it is important to note a few fundamentals for a proper understanding and management of innovation. For this, we draw mainly on Benoît Gailly's book (2018) and the “key insights” he identifies as the pillars of innovation management, which will be useful to support the reflections made in the analysis in Chapters 4 and 5. We will explain his recommendations here by setting out a theoretical framework for innovation. Gailly (2018) identified dozens of recommendations, but, for reasons of relevance, we have highlighted only the key insights of interest to our research.

a) Key insight I: “Innovation means much more than invention. Managing innovation means managing both newness and change, and the latter often matters the most.” (Gailly, 2018, p. 14)

To understand this key insight, we need to go back to Joseph Schumpeter's definition of innovation, a famous 19th century economist. He defined innovation as "new combinations of existing resources", and insisted on the distinction between novelty and innovation (Croitoru, 2017). Schumpeter also distinguished between two types of innovation according to their intensity: incremental innovations, i.e., modifications or improvements; and radical innovations, i.e., major changes (Croitoru, 2017). Several authors have based their understanding of innovation on Schumpeter's work, emphasizing that innovation is different from invention (Croitoru, 2017). Innovation corresponds to a result, but also to an activity/process to achieve that result (Nooteboom & Stam, 2008). It is therefore much more than the generation of new ideas; it is also the implementation of these ideas (Nooteboom & Stam, 2008). Gailly (2018) recommends that managers should not forget the change part of the

innovation concept, which he considers to be the most important one. Innovators do not just need to have a good idea; they also need to be able to convince people to implement it (Gailly, 2018).

b) Key insight II: “Most people resist change. As a consequence, the main job of an innovator will be to drive adoption, by convincing people to disrupt their routines.” (Gailly, 2018, p. 20)

This key insight is based in particular on the theory of conformity, according to which people tend to resist change and prefer stability (Berkun, 2010). This is known as “resistance to change”, and more specifically “resistance to innovation” (Claudy, Garcia & O’Driscoll, 2014). This resistance to innovation is a classic behavior in humans who perceive innovation as a disruption of a beneficial status quo (Claudy et al., 2014). They may see it as a source of difficulty and risk, which dissuades them from adopting it (Claudy et al., 2014). On the other hand, according to Berkun's theory (2010), the success of an innovation lies in the innovator's ability to convince people to adopt it. Gailly (2018) therefore makes the following recommendation for innovation managers. Firstly, people need to be convinced that the innovation is worthwhile and can bring significant benefits (Gailly, 2018). Secondly, managers need to convince them that the innovation will not be difficult or risky to adopt (Gailly, 2018).

c) Key insight III: “Innovation is about both making new things and making similar things in new ways.” (Gailly, 2018, p. 27)

Joseph Schumpeter distinguished five types of innovation: new products, new production methods, new sources of supply, exploitation of new markets and new ways to organize business (Fagerberg, 2004). Today, managers focus mainly on distinguishing between the first two types, which can be called "product innovations" and "process innovations" (Fagerberg, 2004). Product innovations are characterized by the launch of new or improved products, while process innovations are innovations in the way a company conducts its operations (Fagerberg, 2004). Gailly (2018) therefore recommends that innovation managers ensure that they allocate equal time and resources between "doing new things" and "doing the same things in new ways".

d) Key insight IV: “Partnerships offer opportunities to capture unique competitive advantages, by gaining scale and speed – pooling resources – and by developing unique assets – acquiring new capabilities.” (Gailly, 2018, p. 82)

The positive correlation between collaboration and innovation is not new. For years, the literature has emphasized the importance for an organization to look beyond its organizational boundaries and consider building networks of partners in order to leverage resources and advance innovation (Powell, Koput & Smith-Doerr, 1996). Theory suggests that this allows partners to save time and money while minimizing risk, which can be particularly important for very expensive and uncertain projects (Schilling, 2004). De Man & Duysters (2005) explain that collaboration can have two effects on innovation: either it encourages the emergence of an innovation, or it considerably accelerates its realization. In 2006, Henry Chesbrough formalized this and introduced the theory of “open innovation”, arguing that companies should exploit ideas from within as well as from outside the company (Kastelle & Steen, 2011). Gailly (2018) therefore recommends that organizations look beyond their own walls and consider collaborations to accelerate innovation by enabling partners to pool their resources to achieve synergies and economies of scale by sharing knowledge, ideas and data.

e) Key insight V: “Untamed free markets often fail to support sustainable innovation. Targeted and effective public interventions are also needed for strong innovation ecosystems to emerge and strive.” (Gailly, 2018, p. 93)

This key insight can be linked to the thinking of Douglass North, a famous American economist and one of the fathers of “institutionalist theory” at the beginning of the 20th century (Edquist, 2013). This theory asserts that institutions shape the performance of economy, distinguishing between formal institutions (laws, regulations, etc.) and informal institutions (conventions, behavioral norms, etc.) (Edquist, 2013). Several authors use this theory to define the crucial role of institutions and infrastructures for “innovation systems”, which they define as networks of public and private institutions that act together to develop innovation (Stam, 2015). These innovation systems need laws, regulations, standards and public interventions, which are necessary to create a robust innovation ecosystem (Stam, 2015). Gailly (2018) therefore recommends that effective institutions and policies are necessary because innovators need regulations. According to him, "strong innovation ecosystems require strong governance"

(Gailly, 2018, p. 97). Innovation also needs to be supported by public authorities, whether through loans or subsidies, or by providing the necessary infrastructures (Gailly, 2018).

2.3 Rare diseases

2.3.1 Definition

At present, there is no universal definition of a rare disease. There are a lot of different definitions, which are adopted depending on the country or institution. Richter et al (2015) carried out a study to obtain a summary of all the definitions of rare diseases used in the world and they found that in 2015 there were no fewer than 296 definitions from 1,109 different organizations. The reason why there is no single definition is that there is as yet no universal threshold above which a disease is considered rare (*Orphanet*, 2012). However, in Europe, the accepted threshold is one person in 2,000 affected (*Orphanet*, 2012). Therefore, in this research, we will use a definition that uses this threshold: "Rare diseases are those with a particularly low prevalence; in Europe, diseases are considered to be rare when they affect not more than 5 in 10,000 persons in the European Union" (Rodwell & Aymé, 2015, p. 1). Thus, rare diseases are diseases that affect a very small number of people and therefore pose problems linked to this rarity. Fifty percent develop in childhood, but anyone can be affected by these diseases, at any age (Fisher, 2011).

Most of the time, these diseases have serious consequences on patients' lives: life-threatening prognosis, severe handicaps, a negative impact on family life, and economic and societal issues (Fisher, 2011). Appendix 1, from Rare Diseases International[6] (2023a), provides a good illustration of the journey and difficulties faced by people affected by a rare disease and their families. Rare diseases are currently incurable and treatment options are very limited, mainly consisting of standard medication only to alleviate symptoms (Lannoy et al., 2019). These medicines, which reduce symptoms, are called "Orphan drugs" and are currently very expensive (Fellows & Hollis, 2013).

In terms of numbers, in 2023, rare diseases affected 3 to 8% of the world population (Gaik & Münster, 2023). Thus, that year, the number of people suffering from a rare disease

[6] Rare Diseases International is the global alliance of people affected by a rare disease worldwide (*Rare Diseases International*, 2023b).

was estimated at 30 million in Europe and 446 million worldwide (Gaik & Münster, 2023). According to Haendel et al (2019), about 7,000 rare diseases had been identified by 2019 and new diseases are regularly added and described in the medical literature. Moreover, some rare diseases can have several variants, so their diversity is immense (Haendel et al., 2019). The majority of these diseases have a genetic, infectious or autoimmune origin, but in most cases their cause remains unexplained (Wakap et al., 2019). Orphanet, an open-access internet portal, attempts to list and compile an inventory of rare diseases and their associated genes, with the aim of providing as much information as possible, and thus ultimately supporting research (Maiella, Rath, Angın, Mousson & Kremp, 2013). The most well-known rare diseases include cystic fibrosis[7], spinal muscular atrophy[8], Huntington's disease[9], Duchenne muscular dystrophy[10] and Marfan's syndrome[11] *(Orphanet*, n.d.). To illustrate the lack of understanding of these illnesses, none of these five rare diseases has a curative treatment (*Sciensano*, n.d.).

2.3.2 What are the challenges of rare diseases?

"Sixty-five percent of rare diseases are serious, chronic and incapacitating, and may require extensive and prolonged specialist care" (Lannoy et al., 2019, p. 96). This is why innovations are paramount to address the challenges posed by rare diseases, ensuring timely and accurate diagnosis and developing effective treatments. But despite this need for innovation, this remains very complicated because, for the moment, medical and scientific knowledge of rare diseases is still limited. The main reasons for this lack of knowledge are the large number of rare diseases in existence, their complexity, their great diversity and the small number of people affected (Ali & Tubeuf, 2019b). The challenges of rare diseases begin with the determination and setting of the rarity threshold, as mentioned earlier, and continue with access to scientific, medical and pharmaceutical innovations (Ali & Tubeuf, 2019b).

Concerning the diagnosis, the time needed to reach a precise diagnosis is much longer for rare diseases than for others. This is due to their rarity and their heterogeneity because, with

[7] Mucoviscidose in French, it causes respiratory and digestive problems (*Sciensano*, n.d.).
[8] Neuromuscular disease affecting part of the nervous system that controls voluntary muscle movement (*Sciensano*, n.d.).
[9] Neurodegenerative disease of the central nervous system characterized by involuntary movements, behavioral problems, psychiatric disorders and dementia (*Sciensano*, n.d.).
[10] Hereditary disease characterized by progressive muscle degeneration, generally affecting boys (*Sciensano*, n.d.).
[11] Hereditary disease affecting connective tissue (*Sciensano*, n.d.).

some 7,000 rare diseases, there is no standard approach to diagnose them (Lannoy et al., 2019). According to Centogene[12] (2019), it takes no less than 7 years to diagnose a rare disease on a patient. Scientists refer to this as "diagnostic odyssey" which is the inability to make a precise diagnosis (Lannoy et al., 2019).

As a reminder, rare diseases currently have no curative treatments, but only drugs to reduce symptoms, known as Orphan drugs. Currently, less than 5% of patients receive Orphan drugs which allows them to live more or less a normal life and these drugs are relatively expensive (Lannoy et al., 2019). Pharmaceutical companies justify these high prices and the lack of curative treatments by the high cost of developing them (Fellows & Hollis, 2013). They perceive costs as excessively high and risky, given the small number of people involved and the companies' difficulty in finding patients willing to take part in clinical trials (Ali & Tubeuf, 2019a). Researchers agree that clinical trials in rare diseases are complicated by the fact that they have to be carried out on small populations (Crow et al., 2018; Lannoy et al., 2019). Indeed, the low prevalence of rare diseases in the world population means that the recruitment process therefore takes significantly longer than for patients affected by other types of diseases (Lannoy et al., 2019). In addition, the geographical dispersion of patients means that several sites have to be set up in different countries, which further complicates logistics and increases costs. (Crow et al., 2018; Lannoy et al., 2019). This explains why the few Orphan drugs that are available are extremely expensive: as the prevalence of a disease decreases, the cost of the associated drugs increases (Fellows & Hollis, 2013).

Innovation in the field of rare diseases remains therefore complicated. One way of encouraging innovation and reducing costs in order to develop diagnostic methods and treatments can be health data sharing between hospitals, pharmaceutical companies and other institutions (Thorogood, 2020). This shows us the interest and relevance of our research question, which is, as a reminder, as follows: "How can the Data Governance Act and Data Altruism Organizations facilitate innovation in the rare diseases sector?".

[12] Centogene is an international company based in Germany specializing in the field of clinical genomics. It works to improve the understanding, diagnosis and treatment of rare diseases (Centogene, 2024).

2.4 Data: Some definitions and their potential

Before taking a closer look at the Data Governance Act (DGA), a few definitions are important to keep in mind for the rest of our research.

2.4.1 Data

First, it is important to define what data really are. According to the DGA of 30 May 2022, “data means any digital representation of acts, facts or information and any compilation of such acts, facts or information, including in the form of sound, visual or audiovisual recording” (Data Governance Act, 2022, p. 19).

2.4.2 Personal and non-personal data

A distinction made by the DGA and which is important for the rest of our research is the difference between personal data and non-personal data. The General Data Protection Regulation defines personal data as any information relating to an identified or identifiable natural person (a “data subject”, defined below), i.e., a person who can be identified by a name, an identification number or location data for example (General Data Protection Regulation, 2016). Non-personal data is defined as all data other than personal data (General Data Protection Regulation, 2016).

2.4.3 Data subject, data holder, data user and data sharing

Other terms will also be important in the context of our research. First of all, the data subject is a natural person to whom personal data relates (General Data Protection Regulation, 2016). Secondly, a data holder is a legal or natural person who is not a data subject, who can authorize access to and sharing of personal and non-personal data (Data Governance Act, 2022). Then, a data user is a natural or legal person who has legal access to data, whether personal or not, and who is authorized to use it for commercial or non-commercial purposes (Data Governance Act, 2022). Finally, data sharing refers to the act whereby a data subject or a data holder provides data to a data user, with a view to joint or individual use of this data (Data Governance Act, 2022).

2.4.4 Primary use and secondary use of data

Another important distinction for our research is that between primary and secondary use of data. The primary use of data is considered to be the use of data to provide a “basic” service (European Health Data Space, 2022). The secondary use is defined as the exploitation of data for other secondary purposes, such as research and innovation (European Health Data Space, 2022). Later in this report, we will give more specific examples in the case of health data to illustrate this distinction.

2.4.5 The potential of data

As today's environment becomes increasingly digital, the volume of data is increasing (Shan, Obwegeser, Teracino & Wade, 2020). We can see this in Appendix 2, giving some projected figures from the European Commission (n.d.-a). According to the European Commission (n.d.-a), the volume of data available in the EU is indeed set to increase by 530% between 2018 and 2025. Moreover, the economic and social potential of data is becoming immense, as they can be used for a wide range of applications in many different fields. Data can be used to create new products and services based on innovative technologies, to increase production efficiency or to provide solutions to societal challenges for example (European Commission, n.d.-b). Data are therefore a powerful catalyst for innovation.

More precisely, in the field of health, data can have incredible benefits, when used ethically and legally. Health data can be a key factor in facilitating and accelerating the research for new drugs and medical treatments for the patients who need them most, those suffering from rare diseases in our context (European Health Data Space, 2022). Health data can be used in primary and secondary ways, two concepts defined above in Section 2.4.4 and illustrated in Figure 1. Firstly, obviously, medical data enables primary use, i.e., the provision of healthcare to people who need it (Saelaert, Mathieu, Van Hoof & Devleesschauwer, 2023). But at a later stage, these data can also be used for secondary purposes, such as scientific and medical research, and can provide considerable benefits such as improved methods of diagnosis and treatment and more efficient and higher quality care (Saelaert et al., 2023). The secondary use of health data can also reduce costs and therefore potentially increase the scope for medical innovation (Baines et al., 2024). According to European Commission estimates, the reuse of

health data is worth between €25 and €30 billion a year, and could reach €50 billion over the next decade (European Health Data Space, 2022).

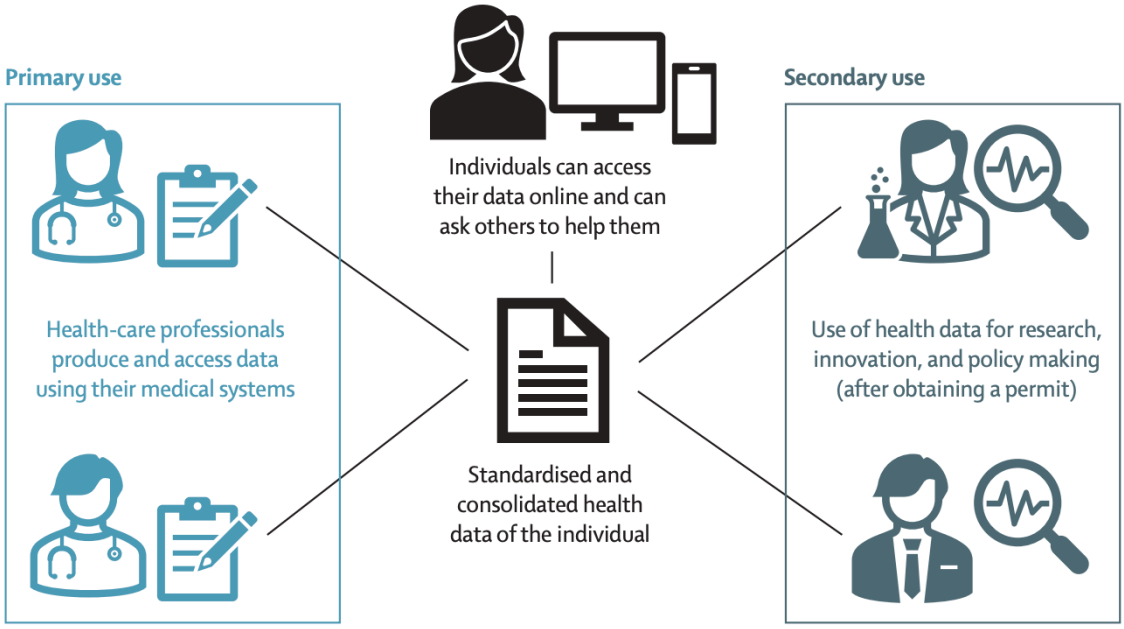

Figure 1: Primary and secondary uses of health data (Raab et al., 2023)

However, the potential of health data is under-exploited (European Health Data Space, 2022). The main obstacle to the under-exploitation of health data is the difficulty for citizens to access and control their data (European Health Data Space, 2022). Although some countries are attempting to develop digital infrastructures to facilitate this access and control, a uniform regulatory and legislative framework across the EU is sorely needed. Several Member States do not yet have the resources to digitize data and enable it to be exchanged, with the result that patients' medical information is still often recorded on paper, making it untraceable and scattered across various medical locations (European Health Data Space, 2022). This also complicates access to health data for healthcare professionals. On a larger scale, this lack of access to data and of regulatory fragmentation between Member States limit research and innovation based on health data (European Health Data Space, 2022). The majority of EU Member States do not have specific legislation regarding the reuse of electronic health data for research, policy or regulatory purposes (European Health Data Space, 2022). This restricts the reuse of health data and hinders innovation. Figure 2 summarizes all these problems that prevent health data from being exploited optimally. EU action is therefore needed to put in place a regulatory framework allowing the free circulation of data in general, and health data in

particular, in order to develop a digital single market. This is why, in 2020, the European Commission has developed a strategy aimed at exploiting this potential to the full and facilitating innovation (European Strategy for Data, 2020).

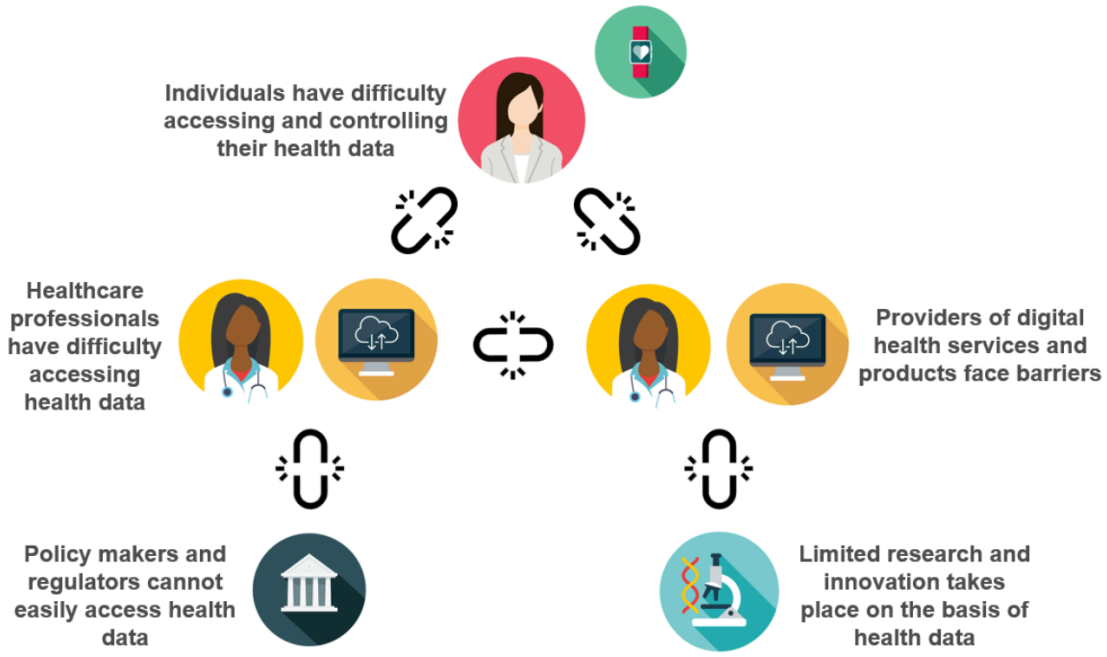

Figure 2: Problems in controlling, using and sharing health data (European Health Data Space, 2022)

2.5 The European Strategy for Data

2.5.1 Definition, goals and actions of the strategy

To exploit the incredible potential of data, the European Commission is defining a strategy called the "European Strategy for Data". This strategy was published in a communication – the COM/2020/66 – on 19 February 2020 to put in place a data management plan for the next five years. The Commission's vision originates from European values and fundamental rights, emphasizing the principle that citizens are central in all European strategies (European Strategy for Data, 2020). In this way, the overall focus is to defend and promote European values and rights in the digital world (European Commission, n.d.-a). The actions implemented in the context of the strategy are based on four pillars, briefly described in Appendix 3.

In practical terms, the European strategy aims to make more data available and to improve the way in which this data is managed and used (European Strategy for Data, 2020). The EU wants to balance the flow of data while guaranteeing high standards of protection in terms of confidentiality, security and ethics for the citizens who generate data (European Strategy for Data, 2020). The European Strategy for Data is creating a "single market for data" that will guarantee Europe's competitiveness and data sovereignty (Curry, Scerri & Tuikka, 2022). The objective is to create a single European data space, which will be made up of several data spaces for certain strategic areas such as health, agriculture and mobility, for example (Curry et al., 2022). This will be explained in more depth in Section 2.5.2. The creation of this single market will enable the unrestricted movement of data, both personal and non-personal, within the EU and across its different sectors (Penedo, 2024). The goal is ultimately to foster innovation driven by data and thus generate societal and economic value for the whole EU and its citizens (Sarretta & Minghini, 2021).

2.5.2 Common European Data Spaces

The European Strategy for Data (2020) mentions that the establishment of the single data market will lead to the creation of various Common European Data Spaces (CEDSs) in order to ensure that more data will be available for the economy and the society and to keep the control of the companies and individuals responsible for generating the data. These data spaces are created in strategic sectors and domains of public interest, i.e., sectors and domains in which the potential of data can have a systemic impact on the entire ecosystem of the EU (Curry et al., 2022). The Commission has decided to support the development of nine CEDSs in 2020, and to add five more in 2024. The complete list of these CEDSs can be found in Appendix 4 but, in the context of our research on rare diseases, we will obviously be interested in the CEDS of the health sector, explained below.

The main role of the CEDSs is ultimately to enable the development of innovations based on data, by making it available and enabling it to be exchanged reliably and securely within the EU (Curry et al., 2022). They "should make data findable, accessible, interoperable and re-usable (the 'FAIR data principles'), while ensuring a high level of cybersecurity" (Data Governance Act, 2022, p. 2). Thus, CEDSs can be seen as vast reserves of data in different sectors and domains, coupled with the essential technical resources required for data utilization and exchange, and suitable governance mechanisms (Margoni, Ducuing & Schirru, 2023).

The European Commission has decided to create different data spaces for different sectors, given that the progress of problems and discussions on data availability varies between the different fields (European Strategy for Data, 2020). Another key factor driving this division is the degree of public interest and involvement in the different sectors (European Strategy for Data, 2020). In fact, this degree may be higher in areas such as health and energy, and lower in sectors such as industry and public administrations (European Strategy for Data, 2020). All CEDSs will be governed by a horizontal framework – composed of the Data Governance Act and the Data Act –, which will be supplemented by sectoral legislations for each sector, given the difference in maturity between them (European Strategy for Data, 2020).

Obviously, in the context of our research on innovation in the rare diseases sector, we will be focusing on the data space in the health sector. It was the very first CEDS to be proposed and established by the European Commission, which considered it to be the most important of the nine originally envisaged (Kari et al., 2023). As explained earlier, health data can have an incredible potential for innovation in the EU, if they are used and reused correctly and legally. This is why the European Commission has decided to set up a CEDS in this sector: the "European Health Data Space". According to the European Strategy for Data (2020), a European Health Data Space (EHDS) is crucial to enable progress in the prevention, diagnosis and treatment of diseases and it contributes to the competitiveness of the EU in the health sector. This CEDS makes it possible to exploit the potential of health data for citizens, patients and innovation within the EU (European Health Data Space, 2022). In addition to the DGA and the Data Act to regulate all CEDSs, the European Commission has decided to establish a sectoral legislative measure for the EHDS in May 2022 (Horgan et al., 2022). The ultimate aim of this proposal is to establish standards for better use of health data in the coming years, in order to take full advantage of their potential (Horgan et al., 2022).

The EHDS promotes the use and reuse of EU citizens' health data. Its precise objectives are listed in Appendix 5, but we can sum them up in two main objectives: optimizing the primary use of health data, and facilitating their secondary use. These two concepts were defined in Section 2.4.4 and are represented in Figure 1. In terms of primary use, the EHDS aims to exploit health data to provide faster, more continuous, effective and less costly healthcare for patients (Saelaert et al., 2023). This data space gives indeed EU citizens the right to control and access their medical data and pass it on to any medical staff, enabling better-quality diagnosis and more personalized treatment (European Health Data Space, 2022).

Secondly, the EHDS promotes the secondary use of health data for scientific and medical research purposes, in order to stimulate the development of new diagnostic and treatment methods (European Health Data Space, 2022). The European Commission would like to establish better health policies by making data more accessible to researchers, innovators and political decision-makers (Saelaert et al., 2023).

The aim of the EHDS is therefore to develop an environment in which electronic health data can be accessible and shared securely and in a way that protects the privacy of individuals in accordance with the GDPR of 2016 (Lalova-Spinks et al., 2023). In this way, the EHDS makes it possible to strengthen EU citizens' access to their health data and to reassure them about the ethical use that the healthcare system makes of it (Lalova-Spinks et al., 2023). This public confidence will in turn allow their data to be used to improve the quality of healthcare, enabling the EU to fully exploit the potential of secure and efficient data exchange and re-use (European Health Data Space, 2022).

2.5.3 What has already been done?

Before establishing its data strategy, the EU had already put in place a number of measures concerning the management and protection of data issued within the EU. Appendix[13] 6 shows a timeline of the most important pieces of legislation adopted in this context. The most important thing to mention for the rest of our research is the General Data Protection Regulation (GDPR). The purpose of the GDPR adopted in 2016 is to improve the protection of EU citizens' personal data through very strict regulations imposed on companies and institutions that have access to these data (Hu & Wei, 2020). This gives individuals control over the use of their personal data by requiring companies to obtain explicit consent from them before collecting and using their data (Hu & Wei, 2020).

Following the development of the European Strategy for Data, the European Parliament and Council accepted two new legislative proposals put forward by the European Commission: the Data Governance Act and the Data Act (European Strategy for Data, 2020).

[13] This is a non-exhaustive list. The dates chosen for this timeline are those used for the adoption of these laws.

2.6 The Data Governance Act

In order to create and regulate the different CEDSs, the European Commission has developed a horizontal framework, consisting of the Data Governance Act (DGA) and the Data Act (European Strategy for Data, 2020). Here we will focus on the DGA, which aims to create a harmonized legal framework for data exchanges within the EU (Data Governance Act, 2022). Indeed, good legislation for data governance is essential for the sharing of data and then for the use of this data in the interest of the population, and rare disease patients in our context. A little schema from European Commission (2024), see Appendix 7, shows this well.

2.6.1 Goals and actions

The DGA, adopted on 30 May 2022 and applicable from 24 September 2023, establishes a structure aimed at enhance confidence in voluntary data exchange for secondary use, to the advantage of business and citizens, and thus the whole EU (European Commission, n.d.-b). The main objective of the DGA is to enable the implementation of the CEDSs in several strategic areas and ultimately to promote the evolution of the borderless digital marketplace and to encourage innovation, by setting a wide range of basic requirements for data governance within the EU (European Commission, n.d.-c). The act should help to complete the basic regulatory framework in order to remedy certain problems that prevent the sharing and re-use of data from being optimized (Shabani, 2021).

The regulation defines conditions for the sharing and the utilization of all types of data, whether personal or not, but it is primarily the growing demand for the sharing of health data that has prompted the European Commission to propose a strategy and this legislation (Shabani, 2021). The objectives of the DGA can be summarized as follows (Von Ditfurth & Lienemann, 2022):

i. Optimize the reuse of protected data held by the public sector, while ensuring their confidentiality (Chapter II, Art. 3-9);
ii. Define and regulate Data Intermediation Services that facilitate the exchange of data between legal persons (Chapter III, Art. 10-15);
iii. Enable natural and legal persons to share their data voluntarily for altruistic purposes in the public interest (Chapter IV, Art. 16-25);

iv. Create the European Data Innovation Board, a new body responsible for overseeing the activities of Data Intermediation Services and Data Altruism Organizations (Chapter VI, Art. 29-30).

In the following of the thesis, we will concentrate on point (iii). The entire structure of the DGA with the different chapters and articles is available in Appendix 8, and for further explanations on points (i), (ii) and (iv), see Appendix 9.

2.6.2 Data Altruism

The DGA refers to and promotes the concept of "Data Altruism", a concept that has emerged over the last few decades. The DGA defines Data Altruism as the voluntary sharing of personal and non-personal data for altruistic purposes (Data Governance Act, 2022). More specifically, Data Altruism refers to a situation where a natural or a legal person decides to share their data, without seeking a reward and with the aim of serving general interest objectives such as combating climate change, improving healthcare or mobility for example (Lalova-Spinks et al., 2023). In its articles, the DGA identifies several actors for the Data Altruism mechanism: the data subject, the data holder, the data user, the Data Altruism Organizations and the competent authority for registration (Paseri, 2024). The first three actors were defined in Section 2.4.3, while the next two will be defined immediately afterwards.

An important aspect here is consent, on which the sharing of data is based. The GDPR defines consent as follows:

> Consent of the data subject means any freely given, specific, informed and unambiguous indication of the data subject's wishes by which he or she, by a statement or by a clear affirmative action, signifies agreement to the processing of personal data relating to him or her (General Data Protection Regulation, 2016, p. 34).

As part of the DGA and Data Altruism, a new concept linked to consent has been developed: the "European data altruism consent form" (Piachaud-Moustakis, 2022). This form is intended for people who wish to share their data altruistically, so as to ensure total transparency regarding the use of their data, in accordance with the rules laid down by the GDPR (Data Governance Act, 2022). The organizations responsible for gathering and

overseeing consents and permissions for sharing data are called Data Altruism Organizations and are explained just below (Lalova-Spinks et al., 2023).

2.6.3 Data Altruism Organizations

The DGA establishes the conditions required for persons choosing to share their data voluntarily to be managed by organizations called Data Altruism Organizations (DAOs) (Data Governance Act, 2022). DAOs will therefore be responsible for collecting data that data subjects and data holders wish to share altruistically (Lalova-Spinks et al., 2023). After collecting shared data, they must be able to make them available for use by data users, who can be natural or legal persons, for objectives of general interest (Lalova-Spinks et al., 2023).

For an entity to be recognized as a DAO, it must undergo a registration process imposed by the European Commission in order to obtain the status of “Data Altruism Organization recognized in the Union” (Paseri, 2024). During this process, it must prove that it meets the general conditions for registration, defined in Article 18 of the DGA. These conditions are as follows. These entities must be non-profit-making and must guarantee the protection of the rights and interests of those who share their data voluntarily (Paseri, 2024). Moreover, organizations must be a legal person in order to carry out Data Altruism activities (Data Governance Act, 2022). They must also carry out these activities via an entity that is distinct from its other operations and be completely independent of any other entity that carries out activities for profit (Data Governance Act, 2022). To manage this registration process, one or more competent authorities will be designated by each EU Member State and will be responsible for maintaining a national register of DAOs in their country (Data Governance Act, 2022). If the entity submitting the application for registration as a DAO meets the above requirements, it will be entered in this register and will be able to use the label "Data Altruism Organization recognized in the Union" (Data Governance Act, 2022).

Natural persons wishing to share altruistically their data must express their consent in compliance with two conditions: the absence of recompense and the public interest purpose (Paseri, 2024). For legal entities, if the data is personal, they must obtain the explicit consent of the individuals concerned – data subjects – before sharing these data with a DAO (Paseri, 2024). Individuals must be informed as to how their data will be used, why it will be shared and which entities will have access to this information (Paseri, 2024). Appendices 10 and 11

show the data sharing infrastructure before and after the introduction of the DAOs concept in the case of clinical trials. These figures show that citizens can share their data with a DAO anywhere in the ecosystem, as long as they give their consent.

Chapter 3: Methodology

Having defined all the concepts essential to our research, it is now time to start looking for the answers to our research question, which, as a reminder, is: "How can the Data Governance Act and Data Altruism Organizations facilitate innovation in the rare diseases sector?". Before we can answer this question, we need to define the methodology used.

To the best of my knowledge, there is as yet no research into the specific benefits that the DGA could offer to the field of rare diseases. This report seeks to fill this gap by providing an initial theoretical analysis of the benefits and challenges of the DGA in this sector. This preliminary approach is essential to assess the real impact of the DGA on rare diseases innovation. Consequently, our approach is primarily exploratory with a method of gathering information based mainly on research and documentary study. Thus, our research will be more theoretical and qualitative, highlighting the potential benefits as well as the challenges and risks of implementing the DGA, with a focus on innovation in the context of rare diseases.

To answer our research question, we will use the analysis of Chapters 4 and 5. Firstly, Chapter 4 aims to link the two most important concepts in our research – rare diseases and the Data Governance Act – and will present the potential benefits that the implementation of the DGA could bring to address the lack of innovation in this sector. Chapter 5 will present an analytical comparison between this theory and practical reality, based on studies of articles that have investigated challenges and risks of the implementation of the DGA. Comparing the two chapters will therefore enable us to answer our research question and draw up a summary of the potential benefits and challenges/risks associated with the DGA in the context of rare diseases. Both chapters will also be linked to the theoretical framework on innovation developed in Section 2.2 in order to support the discussion.

The approach chosen for our research is deductive. We have to deduce for ourselves some of the benefits and challenges of the DGA in order to answer our research question, given the recent implementation of the DGA.

Chapter 4: Potential benefits of the Data Governance Act for rare diseases

Our reasoning for defining the benefits of the DGA for rare diseases will be as follows. There is currently no literature linking rare diseases and the DGA, but there are many publications on the benefits of health data sharing. We will base our reasoning on these. Given that the DGA promotes data sharing, we will build on this foundation to infer the potential benefits of this Act. In Section 4.1, we begin by defining the benefits of health data sharing for rare diseases, in order to deduce the potential benefits of the DGA for this sector. Then, in Section 4.2, we will explain how the DGA promotes and can increase health data sharing, either directly through the EHDS or via the Data Altruism mechanism and DAOs, in order to support the benefits deducted above.

4.1 Benefits of data sharing for rare diseases

4.1.1 Facilitation and acceleration of clinical trials

As we saw earlier in the theoretical section, one of the challenges to innovation in rare diseases is the development of clinical trials to test new medical devices, new diagnostic methods or treatments (Lannoy et al., 2019). The authors all agree that clinical trials are complicated to set up in this sector due to the lack of affected patients and their geographical dispersion, making the logistics of clinical trials complicated (Crow et al., 2018; Kempf, Goldsmith & Temple, 2017; Lannoy et al., 2019; Pariser & Gahl, 2014). Another problem put forward by researchers complicating clinical trials is the clinical variability of rare diseases and the lack of data concerning it (Lannoy et al., 2019; Kempf et al., 2017; Pariser & Gahl, 2014). Indeed, among patients suffering from the same disease there can be immense variability in terms of symptoms and disease progression (Lannoy et al., 2019; Kempf et al., 2017; Pariser & Gahl, 2014).

One solution to facilitate clinical trials would be data sharing within the EU. Data exchange could help to reduce the time and cost involved in recruiting participants for clinical

trials, making it more efficient. Some authors have confirmed this argument, arguing that the accessibility of data would allow more innovative design and execution of clinical trials in the field of rare diseases, thanks to a huge European database accessible to researchers (Courbier, Dimond & Bros-Facer, 2019; Denton et al., 2022; Thorogood, 2020). Courbier et al (2019) also emphasize the importance of this exchange of health data, allowing a broader understanding of the clinical variability of rare diseases and therefore an improvement in clinical trials and results and ultimately an improvement in the diagnostic process.

Putting this in parallel with the DGA, which defines the regulation for data sharing within the EU, we can therefore deduce that the DGA would stimulate the exchange of health data, which in turn would facilitate clinical trials for rare diseases. Health data could be shared between clinical trial sites, facilitating logistics and speeding up research. Data Altruism, which enables patients suffering from rare diseases to share altruistically their health data, could therefore also facilitate and accelerate clinical trials in this sector.

4.1.2 Facilitation of collaboration

To explain the lack of innovation in the rare diseases sector, researchers also point to the lack of collaboration as one of the reasons that slows down the research and development of new diagnostic methods and treatments (Amorim et al., 2022; Denton et al., 2022; Kempf et al., 2017; Ranganath & Sireau, 2023; Thorogood, 2020). Denton et al (2022) advocate the need of a "culture of collaboration" for rare diseases. Authors argue that innovation in this field requires total international collaboration, particularly for genomic research[14], which is essential for diagnosing these diseases (Amorim et al., 2022; Thorogood, 2020). Kempf et al (2017) showed that this cooperation is essential in this sector, as it facilitates and speeds up clinical trials.

With this in mind, one might think that data exchange could encourage collaboration between various stakeholders in the field of rare diseases. The exchange of health data could enable researchers to have access to the same data and thus work together by coordinating their efforts, in order to boost innovation. Some authors have confirmed this idea by explaining that

[14] Genomic data are considered to be the most important for R&D in rare diseases (Rhem, 2022). Genome sequencing is the process of determining the exact order of the nucleotide bases (A, T, C and D) in an individual's DNA (Costain, Cohn, Scherer & Marshall 2021). As each organism has a different genome, sequencing can help scientific research into rare diseases (Costain et al., 2021).

one of the many benefits of data sharing is cooperation (Amorim et al., 2022; Courbier et al., 2019; Darquy et al., 2025 Lacaze et al., 2017; Thorogood, 2020; Van Panhuis et al., 2014). Data sharing allows researchers to access the same databases and enables them to avoid duplication and maximize the impact of their research (Amorim et al., 2022; Courbier et al., 2019; Darquy et al., 2025 Lacaze et al., 2017; Thorogood, 2020; Van Panhuis et al., 2014). Courbier et al (2019) cite striking examples proving that data sharing and the international collaboration it enables can lead to major scientific and medical advances. In particular, they mention the UK 100,000 Genomes Project, launched in 2012, which aimed to sequence 100,000 patient genomes and provide this database to researchers to encourage collaboration. The project resulted in a major breakthrough for medicine (Courbier et al., 2019).

We can therefore deduce that the DGA, by providing a legal framework to encourage and improve data sharing within the EU, could facilitate collaboration between various stakeholders, thereby accelerating innovation in the field of rare diseases. More specifically, the altruistic sharing of health data via DAOs could enable international collaboration between researchers and scientists in the search for treatments for these diseases. A link can be made here with key insight IV, defined in Section 2.2.2. This key insight, identified by Gailly (2018), recognizes the importance of collaborations and partnerships in driving innovation forward. Collaborations offer significant benefits through the pooling of partners' resources and the sharing of ideas and data, allowing for time and cost savings while minimizing risk. This is particularly important for costly and uncertain research projects in the context of rare diseases.

4.1.3 Redundancy reduction

Data sharing and the collaboration it fosters could also reduce the redundancy of studies and clinical trials trying to find new medical devices for patients suffering from rare diseases. For example, we could imagine a case where, without the possibility of sharing health data, two researchers undertake the same study, unaware that it has already been carried out and that it has failed. If data sharing is authorized and properly regulated, this situation could be avoided. This argument has been confirmed by authors, who point the finger at the waste of resources during research projects and clinical trials (Courbier et al., 2019; Thorogood, 2020). Courbier et al (2019) explain that international data sharing can reduce the risk of duplication of clinical studies and therefore avoid wasting resources, whether financial, human or material. Thorogood (2020) discusses the concept of "clinical trial transparency", which means making available all

information relating to clinical trials, i.e., in particular the dissemination of results and the data on which these results are based. It is this transparency in the data that makes it possible to reduce redundant testing (Thorogood, 2020). Some authors are critical on this transparency of individual patient data, which raises concerns about the protection of privacy, but Thorogood (2020) plays down these fears by saying that all that is needed is good data governance.

This is where we can draw a parallel with the DGA, which sets out rules for data governance in the EU. We can therefore deduce that, thanks to the sharing of data enabled by the DGA, it is helping to reduce the redundancy of clinical trials and the waste of resources during research projects seeking to develop new medical devices for rare diseases.

4.1.4 Costs reduction

Earlier in the theoretical section, we mentioned the high cost of Orphan drugs and healthcare in general for rare disease patients (Lannoy et al., 2019). Another benefit of health data sharing for innovation in rare diseases may be the reduction in these costs. By sharing data and coordinating their efforts, researchers will be able to reduce the costs of research projects, which are so costly in the field of rare diseases. This argument is confirmed by some authors who claim that there is evidence that health data exchange can reduce technical costs (Courbier et al., 2019; Lacaze et al., 2017; Van Panhuis et al., 2014). Courbier et al. (2019) explain that the world has an increasing capacity to produce and store health data, the sharing of which is essential for innovation in rare diseases. This sharing is a way of speeding up the diagnostic process and therefore reducing costs for affected patients (Courbier et al., 2019).

We can therefore deduce that the DGA, by enabling health data to be shared, could reduce the costs of research projects and clinical trials for companies, and therefore reduce costs for patients suffering from rare diseases. DAOs in particular, by collecting the health data needed for research and making them available to data users, reduce data collection costs for companies and research institutions. A link can be established here with key insight IV, defined in Section 2.2.2, which recognizes that collaborations enable economies of scale. Here, for example, it is collaboration between data subjects, DAOs, hospitals and data users that can reduce research costs, and therefore the cost of developing treatments.

4.2 Enhancing health data sharing through the Data Governance Act

For many years, the literature has highlighted the lack of medical data as one of the main obstacles to innovation in the rare diseases sector and authors and researchers all agree that setting up effective data sharing mechanisms is crucial to boosting innovation (Amorim et al., 2022; Brownstein et al., 2015; Courbier et al., 2019; Darquy et al., 2015; Denton et al., 2022; Lacaze et al., 2017; Rehm, 2022; Thorogood, 2020). A few years ago, researchers have already been calling for databases and regulations to be put in place in the field of data governance. In particular, some authors have called for the establishment of "registries"[15] and databases to collect clinical data that can be used to stimulate innovation in order to develop diagnosis methods and treatments for rare diseases (Pariser & Gahl, 2014; Lacaze et al. 2017; Darquy et al., 2015; Brownstein et al., 2015). Several years later, the authors still agree that access to and sharing of large genomic datasets are essential to fill the medical data gap faced by researchers and pharmaceutical companies (Amorim et al., 2022; Thorogood, 2020).

According to Shabani (2023), the DGA is a "EU's move towards facilitating data sharing". By implementing a legal framework, the DGA is seeking to increase confidence in data sharing and thus optimize the use and reuse of EU citizens' data, in particular health data (Shabani, 2021). Hospital databases are a crucial resource for medical research and, if shared across the EU, could generate considerable benefits for healthcare in general including the facilitation of the diagnosis, the development of new medicines and treatments, and the personalization of healthcare (Shabani, 2021). A large number of authors and researcher also seem to agree with this analysis (Carovano & Finck, 2023; Lalova-Spinks et al., 2023; Lalova-Spinks et al., 2024; Paseri, 2024; Richter, 2023; Schneider, 2021, Thorogood, 2020). Links can be made with key insights I and III defined in Section 2.2.2. With regard to key insight I, the DGA illustrates that innovation means much more than invention, and that it involves managing both novelty and change, by introducing new mechanisms for sharing data within the EU. The DGA can be seen as an incremental innovation, improving what already existed thanks to the GDPR: data sharing. In this context, the DGA is not reinventing the concept of data sharing, but improving it by introducing new and innovative mechanisms aimed at increasing confidence and security in data sharing. This illustrates how similar things (sharing data) can

[15] Registries are systems of data collection (Lacaze et al., 2017).

be done in new ways (new governance and sharing mechanisms) as explained in key insight III.

Putting these elements in parallel in the context of rare diseases, we can deduce that the implementation of the DGA could facilitate the exchange of health data and give researchers and pharmaceutical companies access to comprehensive and diversified health data which are crucial to the study of rare diseases. This would make it possible to fill the data gap which is holding back research and the development of diagnostic methods and treatments for these diseases. The European Commission itself confirms this argument, saying that the DGA, by enabling and regulating the health data sharing, will make it possible to provide better quality healthcare and help diagnose and cure rare or chronic diseases (European Commission, n.d.-d).

The DGA enables health data to be shared across EU through several mechanisms, but two of these are important in the context of our research question: the establishment of the EHDS and the introduction of the Data Altruism mechanism. Let us imagine a patient suffering from a rare disease who wants to share his medical data to advance research. He can do this in two ways: directly through the EHDS via hospitals, researchers or other institutions, or altruistically via a DAO. Figure 3[16] clearly illustrates the two cases, which are described in detail in Sections 4.2.1 and 4.2.2 below.

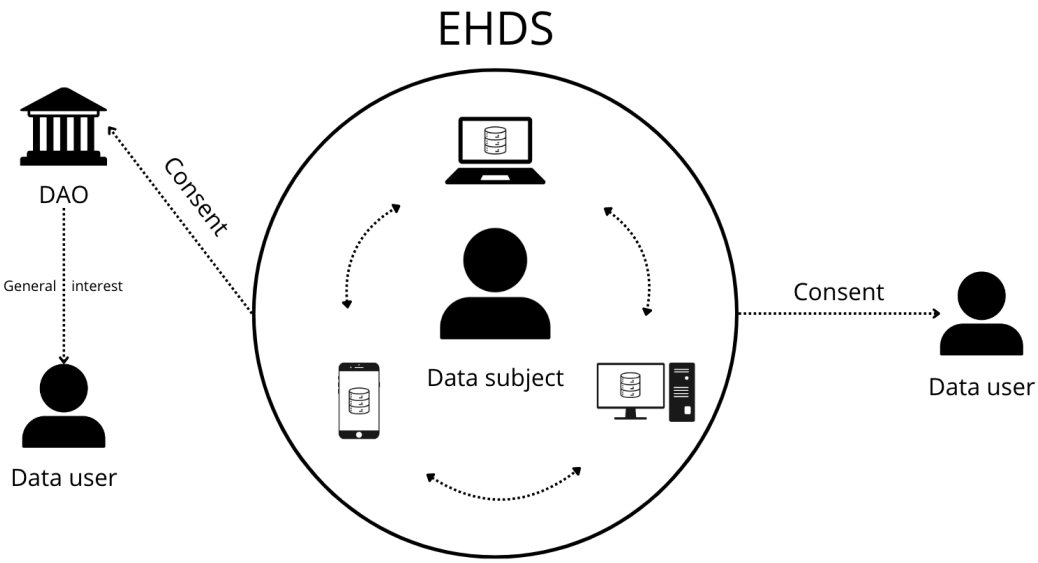

Figure 3: Health data sharing in European Union

[16] Here we have chosen to illustrate the case of a natural person, but the reasoning is the same for a legal entity with different consent requirements, as explained in Section 2.6.3.

4.2.1 Increased health data sharing through the European Health Data Space

As explained in the theoretical section, the European Commission is establishing the EHDS, the aim of which is to optimize the primary use of health data and facilitate its secondary use (Marelli et al., 2023). In our research on innovation in rare diseases, it is this secondary use of health data that interests us most. To fully understand how this system of data exchange through the EHDS works, it should be emphasized that health data is managed and stored in a decentralized manner by hospitals and other health entities in each Member State, and not in a centralized infrastructure (European Health Data Space, 2022). It should also be remembered that in order to use and process someone's health data, the data subject must give his or her consent (Paseri, 2024). In simple terms, the process of sharing data directly through the EHDS works as follows. Let us imagine a patient suffering from a rare disease and having an electronic medical record in a hospital that contains all his health data. If he or his hospital wishes to share these data with other hospitals, researchers or pharmaceutical companies in order to use them for scientific research purposes, the hospital must ask the patient for consent. If consent is given, the data can be shared in compliance with the requirements of both the GDPR and now the DGA. By creating a harmonized legal framework for the operation of the EHDS, the DGA should increase data sharing within the EU.

4.2.2 Increased health data sharing through Data Altruism

Another way of sharing data implemented by the DGA is via the Data Altruism system and therefore via the DAOs. As Figure 3 illustrates, this data sharing mechanism requires the involvement of three players: the data subject, the DAO and the data user. Let us imagine a patient with a rare disease who has an electronic medical record in a hospital containing all his health data. These data are under his control and are accessible thanks to the implementation of the EHDS. If the patient wants to share his data altruistically in order to support research and innovation in rare diseases, he must complete the European data altruism consent form in order to give him consent. The DAO must obtain this consent before collecting the health data. The DAO then sends the data to the data user, who will use it as agreed to serve the altruistic cause chosen, in this case research into rare diseases.

Chapter 5: Challenges of the Data Governance Act for rare diseases

Even if most of our arguments are confirmed by the authors, some do not see the implementation of the DGA so easily. Some point to certain challenges and uncertainties surrounding the DGA. This chapter will present a comparison between theory of Chapter 4 and practical reality, based on studies of articles that have investigated challenges and uncertainties of the implementation of the DGA.

5.1 Unclear concepts surrounding the consent

Some authors have their doubts and believe that the DGA is likely to complicate matters even further (Lalova-Spinks et al., 2023; Paseri, 2024; Shabani, 2021). One of the reasons for their view is the lack of clarity regarding the interaction of the DGA with the GDPR Lalova-Spinks et al., 2023; Paseri, 2024; Shabani, 2021). Lalova-Spinks et al (2023) have carried out an empirical and legal study of the application of DGA and particularly Data Altruism in clinical research, which provides a solid basis for our analysis. They have identified several aspects of the DGA that lack clarity, particularly in relation to the concept of consent. Some of their analyses were confirmed by Paseri (2024), as we will see.

5.1.1 Lack of guidelines for the use of a “broad consent”

Firstly, Lalova-Spinks et al. (2023) noted a lack of clarity in the DGA regarding “broad consent” for the processing of data for general interest purposes. We will explain this briefly. Broad consent is distinct from consent as defined by the GDPR, particularly in Articles 9 and 13. Article 9 of the GDPR sets out the rules for processing specific categories of personal data, such as health data (General Data Protection Regulation, 2016). A crucial point in this article is that explicit consent is required from the data subject before its data can be used (General Data Protection Regulation, 2016). Article 13 goes further by specifying the conditions required to obtain this consent, requiring that the data subject be informed in detail about all aspects of the processing of his or her data (General Data Protection Regulation, 2016). Where consent is given, it is very specific and limited to particular studies (General Data Protection Regulation, 2016). Any other processing of their data for other studies will require new consent from data subjects. However, in some cases, it is difficult to fully define the purpose of data processing

at the time of collection (General Data Protection Regulation, 2016). This is why, in recital[17] 33 of its regulation, the GDPR refers to a broad consent for data processing, defined as a consent where data subjects authorize the use of their data for general areas of research rather than for specific studies (General Data Protection Regulation, 2016). However, this is not repeated in the articles of the GDPR, which constitutes a certain ambiguity and lack of clarity that had already been identified when it came into force by the European Data Protection Board[18] (Lalova-Spinks et al., 2023). According to Lalova-Spinks et al. (2023), the DGA should have included in its articles additional guidelines necessary for the use of this broad consent for data altruism but it is not the case.

If we follow the reasoning of Lalova-Spinks et al. (2023), in our context of research into rare diseases, it would have been beneficial if the use of broad consent had been authorized and regulated by the DGA. It is important to remember that medical research in this field is very hard and time-consuming. If the DGA had included additional provisions to the GDPR regarding broad consent, this would have saved valuable time. Indeed, with these provisions, it would not have been necessary to ask data subjects for explicit consent several times, as they would have been able to give permission for their data to be used altruistically in various medical researches, thus maybe speeding up research projects for rare diseases. Our argument is validated by Thorogood (2020) who argues that broad consent is essential in the rare diseases sector. He explains that without this broad consent, there is a risk of losing the data if each use requires a new consent from the data subjects (Thorogood, 2020). This argument is also supported by key insight II, defined in Section 2.2.2. According to Gailly (2018), for an innovation to be accepted, innovators must prove that it is worthwhile and not difficult or risky to adopt. Here, the lack of guidelines for broad consent complicates the process of collecting and using health data, which could discourage patients from sharing their data.

5.1.2 Ambiguity between “purpose” and “processing operation”

Another aspect related to consent that lacks clarity in the DGA according to Lalova-Spinks et al. (2023) is the ambiguity between the notions "purpose" and "processing operation". According to Article 9(2)(a) of the GDPR, in order to authorize the processing of his or her

[17] European regulations are divided into two parts: recitals and articles. The recitals are a preamble that explains the background and general principles of the regulation, while the articles set out the specific rules and legally binding obligations (European Union, 2015).
[18] It is a board that ensures consistency in the application of the GDPR (Lalova-Spinks et al., 2023).

health data, the data subject must give his or her consent for one or more specific “purposes”, as explained above (General Data Protection Regulation, 2016). On the other hand, for Data Altruism, Article 25(3) of the DGA states that data subjects must give their consent for a specific data “processing operation”, and not for a specific purpose (Data Governance Act, 2022). Lalova-Spinks et al. (2023) note that these two concepts are different. The purpose can be defined as the objective for which the health data is collected, for example developing a cure for a rare disease; while the processing can be defined as the manipulations of the data to achieve that purpose, for example collecting, analyzing and sharing the data (Lalova-Spinks et al., 2023). In fact, several processing operations can be carried out to achieve a single purpose, and a single process can be used to achieve several purposes (Lalova-Spinks et al., 2023).

Lalova-Spinks et al (2023) raise the question of whether the legislator wanted to regulate consent to Data Altruism in this way or whether there is a lack of clarity. Either way, it complicates medical research into rare diseases. As we saw above, one of the challenges of innovation in rare diseases is the lack of affected patients and therefore the lack of data for research (Douglas et al., 2022). In the context of rare diseases, the DGA should therefore have based altruistic data consent on the purpose rather than the specific operation, as this would allow more efficient use of the data. By focusing on the general purpose, researchers would have been able to carry out various necessary data processing operations without having to obtain new consent for each separate operation, thus facilitating and speeding up innovation. This argument is supported by key insight II, defined in Section 2.2.2. If consent has to be sought for each processing operation, this will complicate the process, which may discourage people from sharing their health data. It therefore represents a missed opportunity for innovation.

5.2 Potential lack of trust in Data Altruism

Some researchers have identified people's trust as a key driver in their willingness to share their data (Courbier et al., 2019; Darquy et al., 2015; Lalova-Spinks et al., 2024). Establishing and maintaining this trust can encourage patients to share their data in a sustainable way and thus increase the chances of success of research projects (Courbier et al., 2019). In the context of our research into rare diseases, the trust of EU citizens in the data sharing infrastructures established by the DGA could be an essential key to encouraging them to share their health data altruistically to boost innovation in this sector and thus meet unmet medical

needs. In fact, the European Commission has explained that setting up the DGA would be a way of increasing confidence in data sharing (Data Governance Act, 2020). However, some authors think that some missing aspects of the DGA could compromise data subjects' confidence in data sharing. According to Lalova-Spinks et al. (2023), the current DGA regulation does not present enough convincing arguments to inspire confidence, especially for the following reasons.

5.2.1 Lack of control over data users

One of the missing aspects of the DGA recognized by several authors is the lack of control by DAOs over the data users who have access to the data in order to use it altruistically (Lalova-Spinks et al., 2023; Paseri, 2024). Article 21(2) of the DGA simply establishes the requirement that a DAO may not use the data to which it has access for purposes other than those for which the data subject has authorized the processing, but makes no mention of any checks on data users to ensure that they are processing data lawfully (Data Governance Act, 2022). Paseri (2024) confirms this idea, but goes further by saying that, even if this control were put in place, it would not be easily achievable in practice. In her opinion, the DGA does not define precisely enough what is meant by "general interest objectives" (Paseri, 2024). The DGA does not provide a precise list of what falls into this category, which would make the control of data users subjective and complicated (Paseri, 2024).

This lack of control could result in a lack of trust from people and it could slow down some people in their desire to share their health data altruistically. In the context of our research, this could represent missed opportunities to collect health data for research into rare diseases. This is supported by key insight II, defined in Section 2.2.2. According to Gailly (2018), one of the conditions for getting people to adopt an innovation is to convince them that it is not risky. However, here, this lack of control can make patients think that it is risky to share their health data, and therefore this can slow down the effective implementation of the innovation.

5.2.2 Lack of patient involvement

Another weakness of the DGA and the Data Altruism system identified by Lalova-Spinks et al. (2023) is the lack of compulsory presence of citizen representatives within the DAOs. This lack can translate into a lack of trust among EU citizens, discouraging them from

sharing their data altruistically. This weakness of the system is even more important for clinical research as patient representatives are essential to ensure the smooth sharing of sensitive health data, such as genomic data in our context for innovation in rare diseases (Lalova-Spinks et al., 2023).

Several authors have stressed the importance of patient involvement in scientific research and even more so in research into rare diseases (Amorim et al., 2022; Lalova-Spinks et al., 2024; Thorgood, 2020). Lalova-Spinks et al. (2024) carried out a study showing that patients are more likely to share their health data if they are involved in the data sharing process in an altruistic manner. This lack of patient representatives in DAOs could well prevent rare disease patients from sharing their health data, which are essential for the research and development of treatments. Lalova-Spinks et al. (2023) argue that the addition of a patient advisory committee would also be welcome within DAOs. This argument is supported by key insight II, defined in Section 2.2.2, because the lack of patient involvement in the Data Altruism mechanism may discourage patients from sharing their health data if they consider it risky.

5.2.3 Concerns about security and privacy

Paseri (2024) raises also concerns about the security of DAOs. These DAOs have to collect, store and transfer a large amount of data, which makes them central to the data management ecosystem (Paseri, 2024). This centralization of data in DAOs makes them both powerful and highly vulnerable, as they are more susceptible to cyber-attacks and therefore to breaches of security and privacy (Paseri, 2024). Thorogood (2020) confirms this idea, pointing out that the more data are shared with various stakeholders, the higher the risk of a breach.

These concerns about data security may worry EU citizens and make them wary of the solidity of the infrastructures that the DGA is putting in place. In our context, rare disease patients may be skeptical about their possibility of sharing their health data altruistically with DAOs. This argument is supported by key insight II, defined in Section 2.2.2. If patients perceive that sharing their data is risky, they will not adopt innovation and will not share their health data, which will represent a missed opportunity to advance research and innovation in rare diseases.

5.3 Risk of fragmentation

Another risk of implementing the DGA is the risk of fragmentation, identified by Paseri (2024). Although the European Commission has confirmed that the DGA will reduce fragmentation between Member States in terms of data management, it is the Member States that will have to implement the concept of Data Altruism and create DAOs, and some will encounter more difficulties than others (Paseri, 2024). This argument is confirmed by Lalova-Spinks et al. (2023) who explain that the DGA may lead to an uneven sharing of Data Altruism in the EU as Member States may have different sensitivities regarding the donation of personal data and trust in government authorities.

This risk of fragmentation may not only slow down the uniform implementation of the DGA, but also compromise its overall objective: stimulating innovation. This is particularly true in the context of research into rare diseases, where data sharing is essential for the discovery of new treatments. The inequality in the practice of Data Altruism between EU countries can lead to a missed opportunity to foster innovation in this sector. A link can be made with key insight V defined in Section 2.2.2, according to which innovation requires adequate policies, institutions and infrastructures. If some Member States do not have these elements or have them insufficiently, innovation will be made more difficult.

Chapter 6: Conclusion

As a reminder, the aim of this master's thesis was to answer the following research question: "How can the Data Governance Act and Data Altruism Organizations facilitate innovation in the rare diseases sector?".

Following our analysis, we can state that the implementation of the DGA and the introduction of the Data Altruism mechanism offer significant opportunities for innovation in the field of rare diseases. By defining a regulation for data sharing within the EU, the DGA aims to increase health data sharing directly via the EHDS or via the Data Altruism mechanism and DAOs. This increase in health data sharing can then benefit the rare diseases sector, thanks to the benefits of data sharing that we have identified. It can therefore be summarized and deduced that the implementation of the Data Governance Act has the potential to a) Facilitate and accelerate clinical trials; b) Foster international research collaboration; c) Reduce

redundancies in scientific studies; and d) Reduce research costs. Ultimately, the DGA could help to boost innovation in the rare diseases sector, in order to develop new diagnostic methods and treatments.

However, we have identified some remaining challenges to the implementation of the DGA in the EU. Firstly, unclear concepts regarding patient consent can hinder and further complicate the implementation of health data sharing. The lack of clear guidelines for the use of a broad consent, as well as the ambiguity between the terms "purpose" and "processing operation" in the consent requirements for Data Altruism, can be major obstacles to innovation in the field of rare diseases due to the complexity involved. Secondly, the potential lack of trust of citizens in the Data Altruism mechanism represents a significant barrier to effective adoption of DGA, while trust is one of the key drivers for people to share their data and while the primary objective of DGA is to build trust in data sharing. The main reasons for this potential lack of trust are the lack of control over data users, insufficient patient involvement and concerns about security. Finally, although the European Commission argues that the DGA will reduce regulatory fragmentation between EU Member States regarding data management, we have identified that the DGA could actually lead to fragmentation, due to the different sensitivities of Member States regarding data sharing.

In summary, the DGA has enormous potential to encourage and improve innovation in rare diseases thanks to the increased data sharing it offers, but major challenges remain and could slow down and complicate data sharing even further.

6.1 Managerial implications

Following this conclusion to our research question, certain managerial implications for different stakeholders can be identified.

For the legislator, an essential managerial implication is to clarify certain aspects of the DGA that we have identified as being ambiguous, in particular certain concepts relating to the consent that patients must give in order to share their data altruistically. In addition, the legislator should ensure that the DGA directives are harmonized in the Member States, in order to minimize the risk of fragmentation that we have identified and thus optimize the sharing of data that the DGA allows. This recommendation is supported by key insight V, which

emphasizes that for innovation to flourish, it is essential to have the right policies, institutions and infrastructures to support it.

For patient organizations, a managerial implication is to educate and inform people living with rare diseases about the possibility of sharing their health data in an altruistic way, via the Data Altruism mechanism and DAOs. The aim is to make patients aware of this opportunity and encourage them to share their data to promote scientific and medical research, thereby facilitating the development of new methods of diagnosis and treatment. This initiative can also be supported by the public authorities through national awareness campaigns aimed at strengthening confidence in the Data Altruism mechanism. This recommendation is supported by key insights II and V, which state that people need to be convinced before adopting an innovation, and that an innovation ecosystem needs support mechanisms.

Another implication for public authorities is to provide financial support to optimize the management of health data. This could take the form of subsidies to help hospitals, research institutions and pharmaceutical companies to modernize their digital infrastructures, which are necessary for the secure sharing of health data. National authorities could also allocate funds for companies wishing to become DAOs for example. This recommendation is supported by key insight V.

For research institutions, a major implication would be to maximize the opportunities for international collaboration offered by the DGA, which enable them to work in partnership not only with other national institutions but also with those of other Member States. This recommendation is supported by key insight IV, which recognizes the importance of collaboration for innovation.

6.2 Limitations

Although the work provides new and interesting insights, it is important to mention certain limitations of this report.

A first important limitation is that our work focuses on only one part of the DGA, that of Data Altruism and DAOs. However, the DGA also encompasses other significant initiatives. For example, it includes provisions facilitating the exchange of data between the public and

private sectors, as well as requirements for data intermediation services. By neglecting these aspects, our study does not take into account the whole data sharing ecosystem within the EU.

In the same perspective, although we have mentioned the sectoral legislative regulation related to the establishment of the EHDS, we have not studied it in depth. This omission limits our analysis, as this legislation introduces new bodies and infrastructures that work in concert with the DAOs.

A last limitation of this research is the newness of the DGA initiative. Since it only came into force in September 2023, existing literature on this subject is still limited. This lack of resources makes it difficult for the moment to assess the potential concrete impact of the DGA on innovation in the rare diseases sector. This work is therefore a preliminary study for assessing the real impact of the DGA for rare diseases, and is largely based on deduction and supposition.

6.3 Future research

Given the limitations, some recommendations for future research on the impact of DGA in rare diseases can be considered.

Firstly, given the theoretical nature of this research, a more empirical study of the impact of the implementation of the DGA could be considered. Such a study could be conducted over the long term, given the recent adoption of this initiative. It would consist of analyzing the real impact of the DGA on innovation in rare diseases by collecting data. This approach would make it possible to test the hypotheses formulated about the potential benefits in this theoretical research. An in-depth investigation of the effectiveness of DAOs could be undertaken to assess their specific contribution to facilitating data sharing and innovation in the rare diseases sector. In the same perspective, a cost-benefit analysis of the implementation of DAOs could be relevant to assess the benefits compared with the costs of implementing and maintaining these structures.

Secondly, in this work, we have focused exclusively on the Data Altruism section of the DGA. For future research on innovation in rare diseases, it would be interesting to examine another aspect of the DGA or to consider all aspects of the DGA simultaneously. In addition, it

would also be interesting to look at the sectoral legislation for establishing the EHDS in greater depth.

Finally, an empirical analysis of the willingness of rare disease patients to share their health data with DAOs could be of interest. This could help to understand the motivations and concerns of patients regarding the sharing of their health data, and thus help to develop strategies to strengthen patient confidence and thus improve participation in the Data Altruism mechanism.

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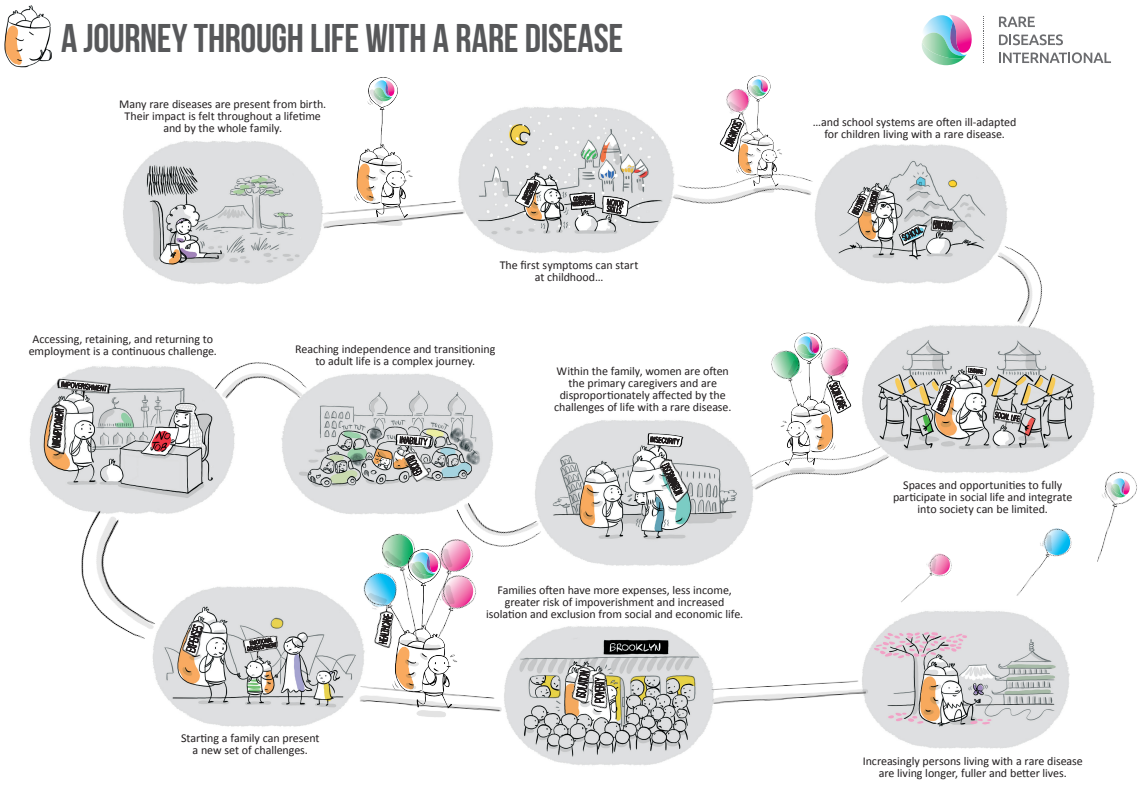
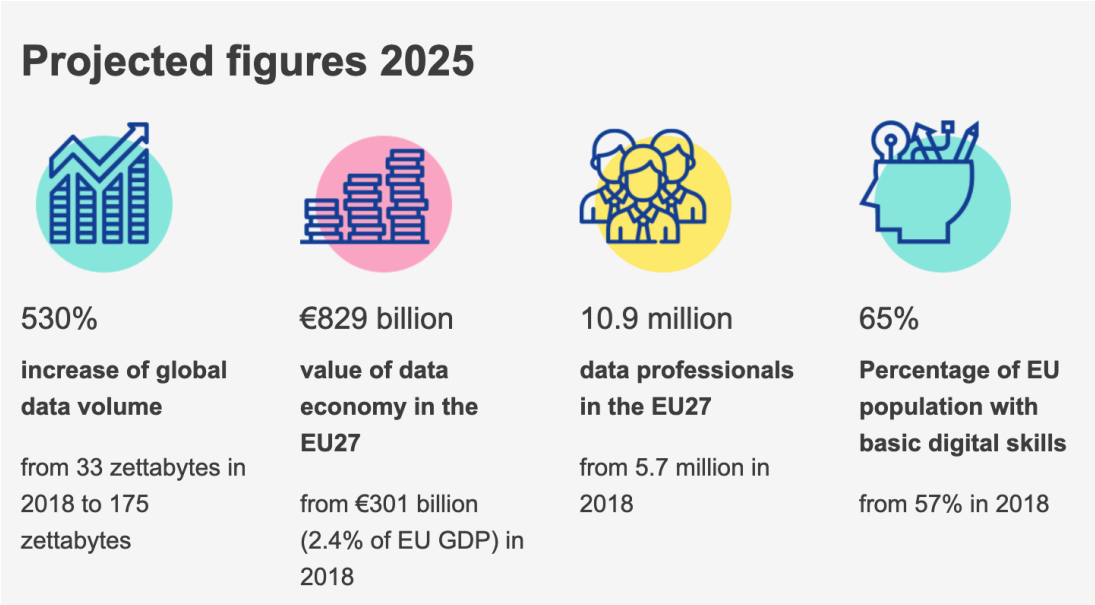
Appendices

Appendix 1: A journey through life with a rare disease (*Rare Diseases International*, 2023a)



Appendix 2: Projected figures 2025 (European Commission, n.d.-a)



Appendix 3: Pillars and actions of the European Strategy for Data (European Strategy for Data, 2020)

The actions implemented in the context of the strategy are based on four pillars, briefly described (European Strategy for Data, 2020):

a) A cross-sectoral governance framework for data access and use

The strategy aims to put in place cross-sectoral (or horizontal) measures relating to access to and use of data in order to create a data-agile economy. To make this vision concrete, the EU will be putting in place a legislative framework for the governance of all the Common European Data Spaces. This governance will make it possible to establish rules to define which data can be used in which situations, and to facilitate the exploitation of data across borders. This strategy must be done in accordance with the principles aimed at making data Findable, Accessible, Interoperable and Reusable (FAIR).

The aim is also to help make decisions about how data will be used and by whom for scientific research and to help people to permit the exploitation of their data for public interest ("Data Altruism"), while complying with the GDPR.

b) Enablers: Investments in data and strengthening Europe's capabilities and infrastructures for hosting, processing and using data, interoperability

To develop an environment conducive to data-driven innovation, EU plans to invest money. The Commission will be using EU funding programs to consolidate Europe's technological competitiveness and will be investing in a High Impact Project on European data spaces between 2021 and 2027. Funding for this project will enable the development of infrastructures and tools for data sharing, as well as governance mechanisms to regulate this sharing. The Commission is prepared to finance €2 billion out of a total of €4-6 billion. The remaining 2 to 4 billion euros will have to be invested by the Member States.

c) Competences: Empowering individuals, investing in skills and in SMEs

The actions put in place following the development of the strategy must make EU citizens more responsible. People must be authorized to access and control the data they generate, in accordance with the GDPR. Secondly, the strategy aims to build the capacity of SMEs by using data, which is an important asset in this context. Start-ups are playing a crucial role in the creation of new data-driven business models.

d) Common European Data Spaces in strategic sectors and domains of public interest

The horizontal framework described above in point a) must be supplemented by sectoral legislations to regulate access to and use of data in certain strategic sectors, as defined by the European Commission. These strategic areas will be the basis for creating different Common European Data Spaces, environments where data can be shared in complete security.

Appendix 4: List of Common European Data Spaces (Staff Working Document II, 2024)

In 2020, the European Commission has decided to implement the following nine CEDSs:

i. A Common European Industrial (manufacturing) Data Space;
ii. A Common European Green Deal Data Space;
iii. A Common European Mobility Data Space;
iv. A Common European Health Data Space;
v. A Common European Financial Data Space;
vi. A Common European Energy Data Space;
vii. A Common European Agriculture Data Space;
viii. Common European Data Spaces for Public Administration;
ix. A Common European Skills Data Space.

In 2024, the European Commission has decided to add five more to this list:

i. Common European Cultural Heritage Data Space;
ii. Common European Language Data Space;

iii. Common European Media Data Space;

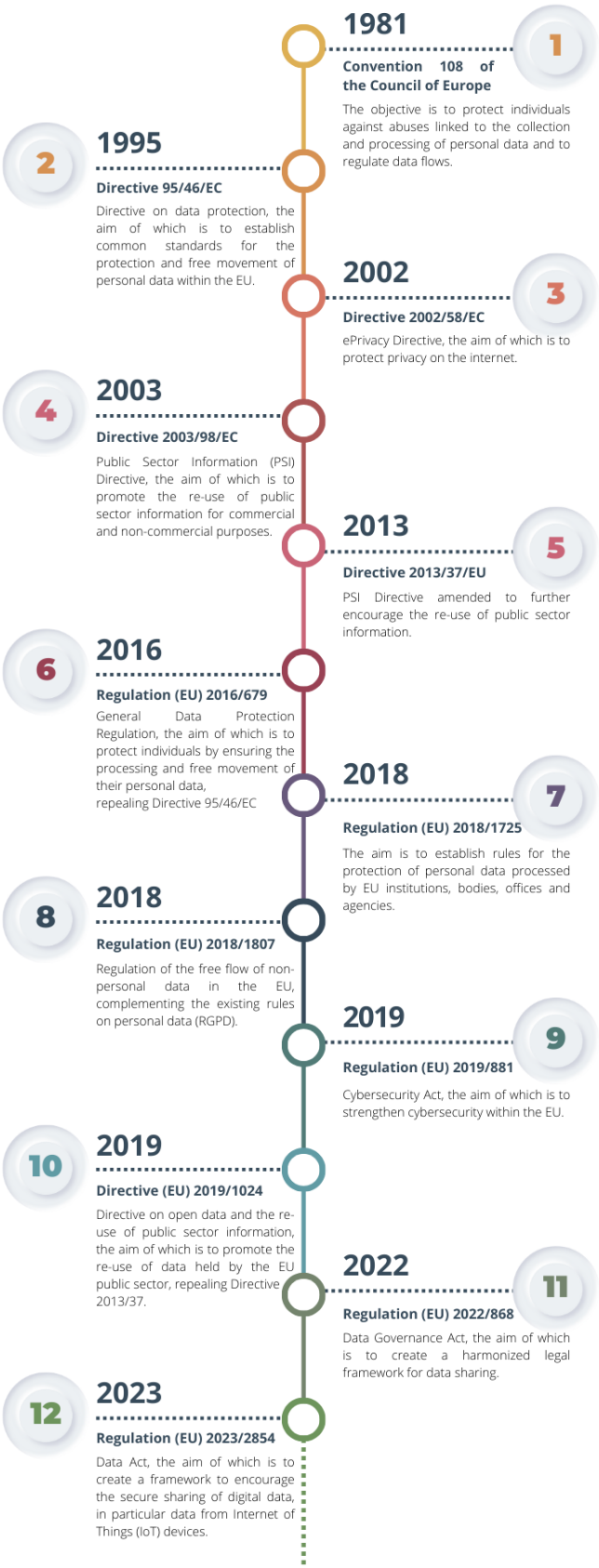
iv. Common European Tourism Data Space;

v. Common European Research and Innovation Data Space.

Appendix 5: Main objectives of the European Health Data Space (European Health Data Space, 2022)

Appendix 6: Timeline of important data management legislations

TIMELINE OF IMPORTANT DATA MANAGEMENT LEGISLATIONS



Appendix 7: Steps before data usage (Staff Working Document II, 2024)

Appendix 8: Content of the Data Governance Act (Data Governance Act, 2022)

CHAPTER I: ***General provisions***

Article 1: **Subject matter and scope**

Article 2: **Definitions**

CHAPTER I: ***Re-use of certain categories of protected data held by public sector bodies***

Article 3: **Categories of data**

Article 4: **Prohibition of exclusive agreements**

Article 5: **Conditions for re-use**

Article 6: **Fees**

Article 7: **Competent bodies**

Article 8: **Single information points**

Article 9: **Procedure for requests for re-use**

CHAPTER III: ***Requirements applicable to data intermediation services***

Article 10: **Data intermediation services**

Article 11: **Notification by data intermediation services providers**

Article 12: **Conditions for providing data intermediation services**

Article 13: **Competent authorities for data intermediation services**

Appendix 9: Brief explanations about point (i), (ii) and (iv) of Section 2.6.1

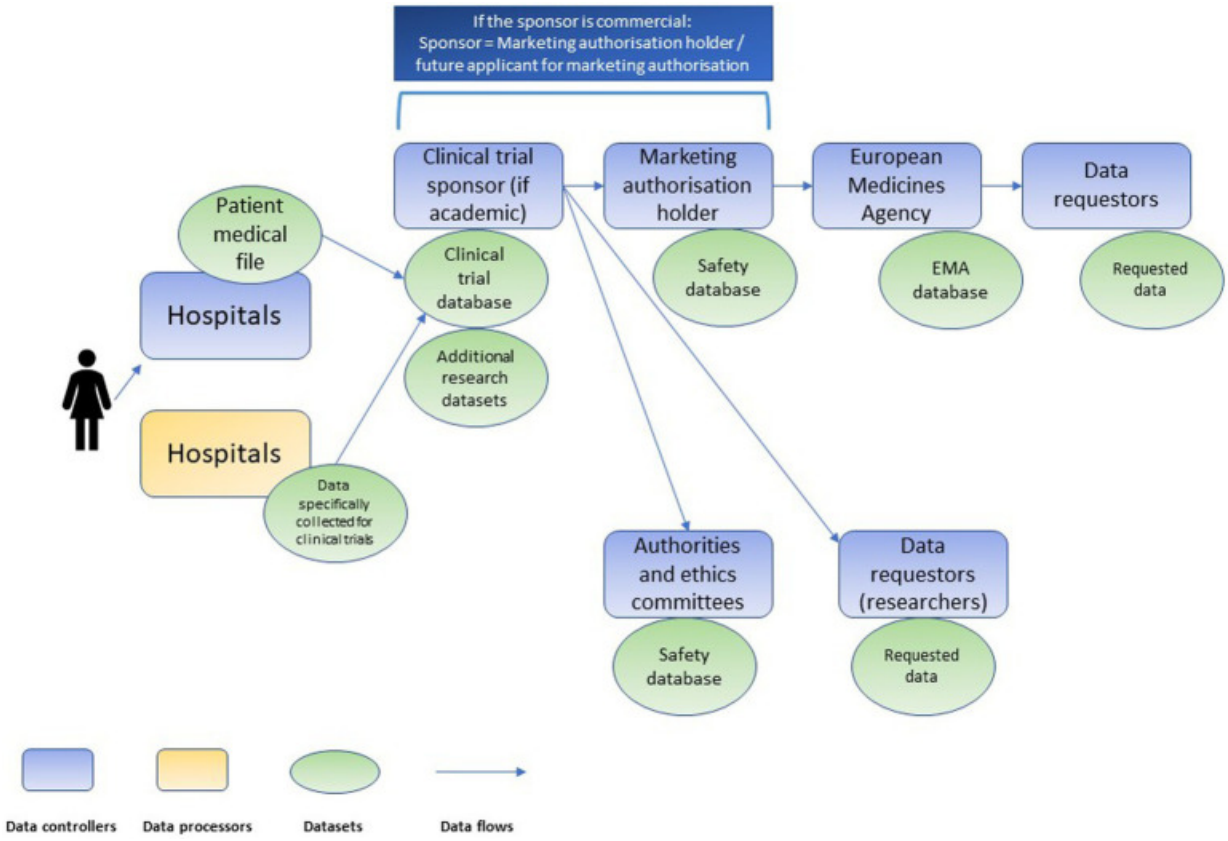
Concerning point (i), the DGA in fact provides for the establishment of a framework for the reutilization of certain categories of data protected by the public sector and which are subject to others' rights such as intellectual property and commercial confidentiality (Buttow & Weerts, 2023). It is because of the impact of these other rights that these data, which we call "sensitive data", need more precise rules governing their re-use (Buttow & Weerts, 2023).

Secondly, with regard to point (ii), the DGA creates a legal framework for "Data Intermediaries Services", i.e. services used to establish commercial relations between data holders and data users for the purpose of sharing data, through a Data Intermediary (DI) (Richter, 2023). DIs are defined as neutral intermediaries between data holders and data users, to facilitate the secure exchange of data between the two parties (Carovano & Finck, 2023).

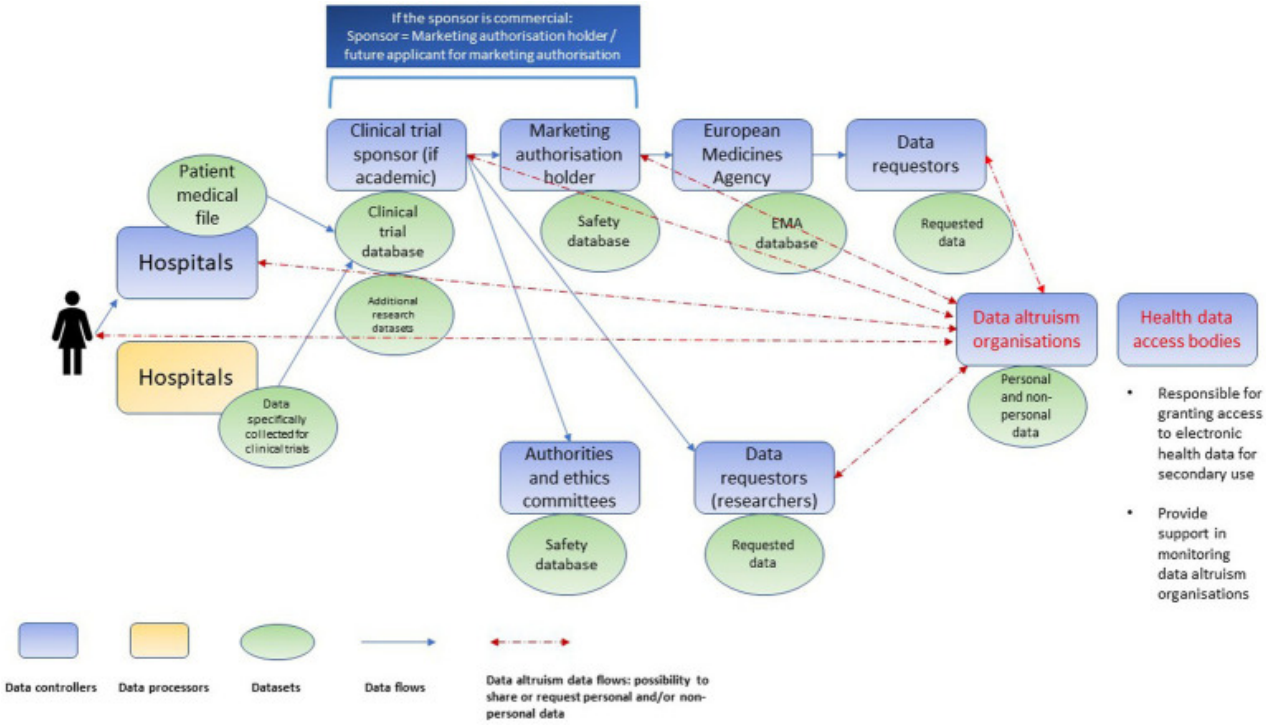
Finally, concerning the point (iv), the European Parliament and the Council have created the European Data Innovation Board (EDIB), a new consultative body (Data Governance Act,

2022). This body will in fact be an expert group, made up of representatives of the competent authorities for the registration of DAOs and for data intermediation services from all EU Member States (Kamocki, Linden, Puksas & Kelli, 2023). Article 30 of the DGA describes EDIB's various tasks in thirteen points, which can be summarized as follows for the context of our research. The main role of EDIB will be to advise and assist the European Commission in various tasks to improve data interoperability and implement best practice in data sharing (Kamocki et al., 2023). For example, the EDIB will help for the development of a practice for handling requests for the re-use of public sector data or the development of a practice for Data Altruism within the EU (Kamocki et al., 2023). The board will also be able to propose guidelines for CEDSs and thus assist the European Commission in commissioning the creation of these CEDSs (Data Governance Act, 2022).

Appendix 10: Chain of independent data controllers in clinical trials (Lalova-Spinks, Meszaros & Huys, 2023)



Appendix 11: Chain of independent data controllers in clinical trials after the introduction of DAOs and health data access bodies (non-exhaustive illustration) (Lalova-Spinks, Meszaros & Huys, 2023)



Abstract :

In Europe, a rare disease is defined as one that affects less than one person in 2,000. People affected suffer severe handicaps and their prognosis is very often life-threatening. However, to date, none of these diseases can be cured. Pharmaceutical companies and research institutes justify this lack of curative treatments by the rarity of rare diseases and the consequent lack of scientific knowledge. Innovation in rare diseases is therefore essential but remains complicated. One way of stimulating innovation in this sector would be to exploit the potential of health data, which can have enormous benefits for advancing scientific research. In 2023, a new legislative instrument from the European Commission came into force – the Data Governance Act – by putting in place a legal framework that aims to enable and facilitate the sharing of data, particularly health data. The aim of this master's thesis is to understand how the implementation of the Data Governance Act could facilitate innovation for rare diseases, by providing a preliminary theoretical analysis of the benefits and challenges of implementing this law in this sector. Our analysis will allow us to conclude that the Data Governance Act can offer significant opportunities for innovation in the field of rare diseases, thanks to the benefits of health data sharing that it regulates, including the facilitation of clinical trials, collaboration, reduction of redundancies and costs. However, certain challenges and risks of the law could hinder and complicate the health data sharing process, such as the lack of clarity regarding the notion of consent, the lack of confidence in data sharing and the risk of fragmentation.

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