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Public and patient involvement in decision-making about health data sharing, access, use, and reuse for rare diseases care and research

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Table of Contents

LIST OF TABLES

ABREVIATIONS

AI - Artificial Intelligence
CHUSJ - Centro Hospitalar Universitário de São João
CPRD - Clinical Practice Research Datalink
EPIUnit - Epidemiology Unit
EURORDIS - Economic Co-operation and Development
ESS - European Social Survey
FCT - Fundação para a Ciência e a Tecnologia
FMUP - Faculdade de Medicina da Universidade do Porto
GWAS - Genome Wide Association Studies
HIQA - Health Information and Quality Authority
IBM - International Business Machines Corporation
ISCTE-IUL - Centre for Research and Studies in Sociology, University Institute of Lisbon
ITR - Laboratório para a Investigação Integrativa e Translacional em Saúde Populacional
KPMG - Klynveld Peat Marwick Goerdeler
NIH - National Institute of Health
NY - New York
OECD - Economic Co-operation and Development
RCCHD - Reference Centre for Congenital Heart Diseases
RCIMD - Reference Centre for Inherited Metabolic Diseases
SMG - Scientific Management Committee
SHIP - Scottish Health Informatics Programme
SPMS - Serviços Partilhados do Ministério da Saúde
SPSS - Statistical Package for the Social Sciences
UK- United Kingdom
USA - United States of America

RESUMO

A utilização, em larga escala, de conjuntos de dados relativos à saúde das pessoas potencia desenvolvimentos promissores que podem melhorar a previsão do risco de doença, a fiabilidade e exatidão de diagnósticos e cuidados de saúde, e a descoberta de novas terapias para muitas doenças sem tratamento, como acontece na maioria das doenças raras. Atualmente, só 5% de todas as doenças raras conhecidas têm tratamento e a taxa de mortalidade infantil nalgumas dessas doenças atinge os 20%. É, por isso, fundamental promover investigação sobre doenças raras com recurso a dados intensivos e centrada nas necessidades das pessoas. No entanto, a acumulação de relatos sobre violações e má gestão dos dados tem gerado preocupações entre o público quanto à perda de privacidade, vigilância indesejada e discriminação. Estas preocupações podem fragilizar a confiabilidade das instituições de processamento de dados e limitar a predisposição das pessoas para partilhar os seus dados para efeitos de cuidados de saúde ou de investigação.

Agências políticas internacionais recomendam a participação dos cidadãos na governança de dados de saúde, de modo a assegurar enquadramentos alinhados com as expectativas dos indivíduos e com os valores sociais em geral. Argumenta-se, ainda, que o envolvimento de cidadãos leigos na governança de dados de saúde contribui para usos seguros, aceitáveis e benéficos dos dados ao promover o diálogo entre investigadores e públicos, ao reforçar a responsabilização e prestação de contas entre os supervisores de dados na cadeia de governança, e ao potenciar a confiabilidade na investigação, elementos centrais para melhorar a confiança pública nas iniciativas científicas. Porém, é ainda escassa a evidência empírica sobre como os titulares dos dados percecionam as oportunidades de envolvimento na governança dos seus dados e se a confiança pública na ciência e demais instituições influencia as suas visões. Esta dissertação pretende preencher esta lacuna e melhorar o conhecimento sobre as visões de pacientes com doenças raras e seus cuidadores informais quanto ao envolvimento nos processos de decisão acerca de diversos aspetos da governança de dados de saúde a nível individual, nomeadamente a partilha, acesso, utilização e reutilização de dados de saúde. Pretende-se, mais especificamente, avaliar como a confiança na ciência e nas instituições de investigação afetam a posição dos participantes.

Realizou-se um estudo observacional transversal em dois centros de referência de doenças raras no Centro Hospitalar de S. João, localizado no Porto, Portugal. Os dados foram recolhidos através de um questionário estruturado autoadministrado, entre junho de 2019 e março de 2020. Um total de 651 participantes (162 pacientes com doenças raras e 489 cuidadores informais) responderam ao questionário (proporção de resposta de 89,4%). A importância atribuída ao envolvimento no processo de decisão acerca da partilha, acesso, utilização e reutilização de dados de saúde foi medida utilizando uma escala de Likert de cinco pontos variando de "1" - nada importante a "5" - muito importante. Testou-se a associação com características sociodemográficas, a confiança interpessoal, a confiança em instituições nacionais e internacionais, e a importância atribuída à confiança nas equipas de investigação e nas instituições de acolhimento. A análise baseou-se em 159 pacientes com doenças raras e 478 cuidadores informais (n=637), os quais tinham informação disponível para todos os outcomes supramencionados.

A maioria dos participantes considerou importante ou muito importante o seu envolvimento nas decisões sobre partilha (85,1%), acesso (87,1%), utilização (85%) e reutilização (79,9%) de dados de saúde. Esta tendência verificou-se tanto nos pacientes como nos cuidadores. Atribuir importância ao envolvimento nos processos de decisão sobre partilha e acesso de dados foi mais frequente nos cuidadores e nos participantes mais velhos. Os mais escolarizados tenderam a atribuir maior importância à participação nas decisões sobre a utilização dos seus dados do que os participantes menos escolarizados. Os participantes com menor confiança nas instituições nacionais e internacionais revelaram-se menos propensos a valorizar o envolvimento nas decisões respeitantes à utilização dos seus dados. Quem atribuiu muita importância à confiança nas instituições de investigação valorizou, mais frequentemente, o envolvimento em decisões sobre a partilha, acesso, utilização e reutilização de dados de saúde. Uma posição semelhante foi reportada, principalmente, pelos participantes que valorizaram a confiança nas equipas de investigação para a partilha, acesso e utilização de dados.

A elevada importância que os participantes atribuíram ao envolvimento na governança de dados a nível individual realça a necessidade de repensar as oportunidades de participação pública nos processos de tomada de decisão sobre

dados de saúde. Merece particular atenção o alargamento das opções de consentimento proporcionadas a comunidades de doenças raras, incluindo mecanismos dinâmicos de consentimento que favoreçam uma maior proximidade com os investigadores. A confiança na ciência e noutras instituições influencia as visões dos participantes, apelando a uma maior transparência na investigação e na governança. Esta poderá ser alcançada através da provisão de informação transparente sobre as implicações da partilha de dados, do auxílio na obtenção dos recursos necessários para tomar decisões informadas, e do desenvolvimento de instrumentos de compensação e redução de danos.

ABSTRACT

The use of large-scale health data sets carries potential to deliver beneficial developments that can be harnessed to predict disease risk, enhance accurate diagnosis and health service delivery, and to discover new therapies for many untreatable diseases, as is the case of most rare diseases. Currently, only 5% of all known rare diseases have treatment and some have a mortality rate during childhood as high as 20%. Fostering data-intensive and needs-driven research on rare diseases is therefore of critical importance. However, mounting reports of data breaches and mismanagement have generated concern for privacy loss, undisclosed surveillance, and discrimination among the public. These concerns can undermine the trustworthiness of data processing institutions and reduce people's willingness to share their data for care and research purposes.

International policy agencies recommend public involvement in health data governance to ensure that data frameworks are consistent with individuals' expectations and with society values more generally. Involving lay members of the public in the governance of health data is also argued to help promote safe, acceptable, and beneficial uses of data by fostering dialogue between researchers and publics, enhancing accountability among data stewards on the governance chain and increasing research trustworthiness, which are essential to improve public trust in research initiatives. However, there is still limited empirical evidence on how data holders perceive opportunities for getting involved in the governance of their data and if public trust in science and other institutions influence their views. This thesis aims to fill this gap and improve knowledge on the views of rare disease patients and their informal carers about getting involved in decision-making in a range of individual-level aspects of health data governance, including health data sharing, access, use and reuse. More specifically, it aims to assess how trust in science and research institutions influences participants' positions.

An observational cross-sectional study was carried out at two reference centres for rare diseases located at University Hospital Centre S. João (UHCSJ), in Porto, Portugal. Data was collected using a self-administered structured questionnaire, between June 2019 to March 2020. A total of 651 participants (162 rare disease

patients and 489 informal carers) responded to the hospital-based survey (response rate of 89.4%). Participants rated the importance of involvement in decision-making concerning health data sharing, access, use and reuse in a 5-point Likert scale ranging from “1” - not important to “5” - very important. Its association with sociodemographic characteristics, interpersonal trust, trust in national and international institutions, and the importance attributed to the trust in research teams and host institutions was tested. This thesis uses data from 159 patients and 478 carers (n=637), with available data on all the above-mentioned outcomes.

Most participants considered it important or very important to be involved in decisions concerned with health data sharing (85.1%), access (87.1%), use (85%) and reuse (79.9%). This trend was observed among both patients and carers. Carers and older participants stated more frequently the importance of being involved in decision-making regarding data sharing and data access, while more educated participants revealed a statistically significant tendency to attribute more importance to participation in decisions about data use. Participants with the lowest levels of trust in national and international institutions were less likely to value involvement in decisions about data use. Those who considered trust in research host institutions as very important rated higher the importance of being involved in decisions about data sharing, data access, data use, and data reuse. A similar position was primarily expressed by participants who valued trust in research teams for data sharing, data access, and data use.

The high value attributed by participants to involvement in individual-level data governance stresses the need to rethink opportunities for public participation in health data decision-making. Extending consent options for rare disease communities to include dynamic consent mechanisms which allows more direct engagement with researchers deserves thorough consideration. The influence of trust in science and other institutions in our participants’ views also calls for more transparency in research and governance. This can be achieved through provision of transparent information about the implications of data sharing, assistance with obtaining the resources needed to make informed choices, and the development of harm mitigation tools and redress.

1 INTRODUCTION

According to Forbes, about 175 trillion gigabytes of new data will be created globally by 2025 (Gil Press, 2020). Health data is expected to follow this trend, both in growth and value. Following the technological development of health apps and wearable devices, there has been a huge rise in the amount of health data being digitally collected and processed. The uses of health data have thus grown beyond the management of patients' immediate health conditions to seeking to enable the anticipation and prevention of disease (Benson, 2016). Moreover, the data used in public health research has diversified to include not only clinical data, but also biological, social, and environmental information related to the health and well-being of both single individuals and large cohorts (Knoppers & Thorogood, 2017). The use of large-scale health data sets holds the potential to deliver beneficial developments that can be harnessed to predict disease risk, enhance accurate diagnosis and health service delivery, and to discover new therapies for many untreatable diseases, as is the case of most rare diseases (Saxena, 2019; Benson, 2016; Flores et al., 2013).

60 million people are estimated to be living with rare diseases in Europe and North America (Shah et al., 2019). Yet, out of 7000 known rare diseases worldwide, only 5% have treatment (Brasil et al., 2019) and some have a mortality rate that reaches 20% in the early years (Goreta et al., 2012). Fostering data-intensive and needs-driven research on rare diseases is of critical importance. However, limited access to quality health care, delayed and/or misdiagnosis, low patient numbers and geographical dispersal make it challenging to gather sufficient health data on rare diseases (Saxena, 2019; Boulanger et al., 2020; De Freitas et al., 2017; Decherchi et al., 2021). Pooling data from rare disease registries and sharing it within and between countries can help to address these challenges (Thorogood, 2020; Decherchi et al., 2021). This practice has been recently endorsed by the European Commission which is sponsoring the creation of European Research Networks that aim to gather cross-national data into interoperable registries (European Commission, 2019)

Growing national and transnational data flows raise new challenges for all parties involved. For people affected by rare diseases, these may entail making decisions about data reuse when the purposes and implications of research are difficult to grasp

(e.g. to improve machine learning that can introduce new biases in care delivery), choosing whether to be sent back research incidental findings that can compromise future choices (e.g. offspring's g predisposition for late onset genetic diseases), and having to deal with the consequences of data breaches that can cause them to loose and even face discrimination in workplaces or by health insurance companies (e.g. disclosure of personal, social or health characteristics considered undesirable by employers and insurance companies such as a diagnosis of chronic disease) (Aitken et al., 2016; Kleiderman et al., 2014; Powles & Hodson, 2017; Sterckx et al., 2016). Failing to provide data governance frameworks that are guided by the provision of transparent information to, the protection of, and accountability towards data holders (Thorogood, 2020) may result in the loss of public trust in data processing organisations and limit people's willingness to share data for research and care purposes (Aitken et al., 2016; Oderkirk & Ronchi, 2018; Trinidad et al., 2010). There is, therefore, an impending need for better governance to ensure safe and beneficial uses of health data.

International policy organisations assert that involving lay members of the public in health data governance can improve data stewardship and ensure that data frameworks are more consistent with individuals' expectations and with society values more generally (OECD, 2015, 2016). However, there is still limited empirical evidence on how data holders perceive opportunities for getting involved in the governance of their data and if trust in science and other institutions influence their views. In this thesis, I seek to contribute to reduce this gap by inquiring rare diseases patients, and their informal carers, about their views regarding involvement on decisions concerned with four aspects of individual-level data governance: whether they want to share their data (data sharing), with whom they want to share their data with (data access), for which purposes (data use) and whether their data can be used for purposes other than those for which it was initially collected (data reuse). In this chapter, I delve into public involvement in health data governance, explaining the rationales that underline it and current opportunities for participation. Subsequently, I explore the existing literature on public views about lay people's involvement in decision-making about health data, which is a central aim of this thesis.

1.1 Public involvement in health data governance

1.1.1 Rationales for public involvement in health data governance

With the growing risk of data breaches and cyber-attacks comes the need for new technologies and governance measures that can help to secure data while at the same time promote informed data sharing and allow easy data access, use, and reuse by those who legitimately seek to mobilise it for the greater good of patients and the public, including researchers, care providers, and policymakers. International organisations such as the Economic Co-operation and Development (OECD) have devoted considerable attention to the issue of health data governance and produced guidelines for strengthening data protection among member states. Among other measures, the OECD recommends public involvement in data governance schemes. This is to ensure that the processing of data under the governance framework serves the public's interest and is commensurate with societal values and the reasonable expectations of individuals for both the protection and the benefits that may derive from the uses of their data (OECD, 2016). Similarly, the European Organisation for Rare Diseases (EURORDIS) has stated key principles of building and maintaining rare disease patients' registries. EURORDIS also recommends the involvement of rare disease patients' representatives in the development, management, and maintenance of these registries to ensure that their needs are acknowledged, to enhance the quality of information in the registries and to increase stakeholders' awareness about and participation in registry activities (EURORDIS, 2012).

At individual country level, involving the public in health data governance has been recognized and adopted in health data governance policies, for example, in Canada. In the Pan-Canadian Health Data Strategy report, putting people in the centre of health data governance is recognized as one of the principles that can foster data interoperability and collaboration between government, stakeholders, and the public. (Public Health Data Agency, 2021). And the Portuguese Ministry of Health in their report on the data strategy for the next generation has stated that the design of a plan for secondary data use in population health and personalised medicine must be driven by the public's needs. This is to ensure that investments in secondary use of health data, advanced analytics, and Artificial Intelligence (AI), support a digital health system

that captures the complete range of the public's needs for data-driven decisions (SPMS, 2019).

Public involvement in health data governance is also substantiated by ethical arguments that are centred around the fair distribution of the benefits arising from data use (Kaye et al., 2018), as well as on the need to promote accountability in data management (Kariotis et al., 2020; Young et al., 2018). Under bioethics debates, public and patient involvement is shown to contribute to alleviate the feeling of being at disadvantage among patient groups and to empower patients to take greater control over decisions regarding their own health. Additionally, public involvement aligns with the principles of democracy and equity by promoting fairness and justice within the scope of data management and use (Conklin et al., 2010).

Furthermore, it has been revealed that public involvement is considered one out of a set of conditions that are essential to ensure the trustworthiness of research, which, in turn, is key to foster public trust in science (Aitken et al., 2016). The other two conditions entail the provision of transparent information about research initiatives to participants and accountability among data stewards on the governance chain, including the presence of mechanism to mitigate the impacts of any arising harms (Aitken et al., 2016; Buyx et al., 2017; Kaye et al., 2018; Young et al., 2018).

The role of trust in the public's acceptability and support for research cannot be overemphasised. Some studies have explored the nature of the relationship between the publics' trust and research describing it as 'conditional'. In other words, a "simple trust/distrust binary relationship" of the public with science does not appear to exist (Aitken et al., 2016; Resnik, 2011). Instead, public support for science seems to co-exist together with scepticism and ambiguity towards science (Aitken et al., 2016; Cunningham-Burley, 2006). This may be attributed to the changes in public interest in and scrutiny of science as new uses of data emerge. Also, the group referred to as the "public" is not static and can change with research themes. The impact of research participation therefore will not necessarily be the same for every group, as some groups can get a better hold of the resources needed for involvement (De Freitas et al., 2017; MacFarlane et al., 2021; Understanding Patient Data, 2022). Accordingly, public trust is dependent on how much researchers and research institutions can be

trusted with data management and governance (i.e. on research trustworthiness), as well as on the degree to which the expectations and needs of the research participants are imbued into data frameworks, namely through participatory initiatives (Aitken et al., 2016; Kerasidou, 2016). When these two align in a research project, trust is more likely to be achieved.

A study involving the scientific management committee (SMG) of the Scottish Health Informatics Programme (SHIP) that inquired about the role of public involvement in enhancing trust in health research elicited different views: while some committee members indicated that providing more information about research and its benefits to the participants would qualify as a way of involving them and, consequently, would earn trust; other members argued that involving the public would entail the accommodation of specific approaches to identify and address the concerns of the public and that, in turn, would help to improve research trustworthiness and earn their trust (Aitken et al., 2016). This implies that not only are there many interpretations of what public involvement means but also of how it relates to public trust in science.

Following the OECD recommendations and the view of the SHIP's SMG members, the aim of public involvement, more than just to provide information about the benefit of research, needs to be targeted at understanding lay people's interests, values and concerns, and to possibly adapt them into data governance frameworks to meet public expectations and deserve the trust of different publics. Some large-scale health data projects have failed to accomplish their goals because they could not achieve public trust and acceptability despite promising benefits to health care and the public. Examples are the *care.data* and the *Clinical Practice Research Datalink* (CPRD) projects in the UK. Studies of the public's opinion suggest that these projects failed due to concerns about informed consent, trust in privacy and data security, lack of communication on how data linkage would work, and the undisclosed involvement of commercial and private companies. These concerns have also been identified through focus group studies as influencing factors for achieving public trust in health care and research (Aitken et al., 2016; Van Staa t al., 2016; Williams & Fahy, 2019). Public involvement could therefore assist in increasing research trustworthiness by seeking and addressing these issues to a reasonable length while also being transparent with communication of research procedures and findings.

1.1.2 Opportunities for public involvement in health data governance

Aitken et al. (2019) assert that "the public should not be categorised as a problem to be overcome but a key part of the solution to establish socially beneficial data-intensive health research for all". In line with this motto, many opportunities for public involvement in data governance arose in the last decade (Darquy et al., 2015; Riordan et al., 2015; Nisha Shah et al., 2019). Public involvement can entail: 1) consultation to obtain information from the public regarding their values, interests, and expectations from research; 2) raising awareness by sharing information about research process and governance mechanisms with the public in a clear and transparent manner; 3) empowering the public by improving its access to resources, building its capacity, and increasing its control over shared data (Aitken et al., 2019) and 4) a combination of all the above.

These opportunities for involvement can be put into practice through various methods including deliberative polls, forums, focus groups, public meetings, or citizen juries (Aitken et al., 2019). They can also participate via electronic platforms such as PatientsLikeMe, which is a community of patients with chronic health conditions that aims at promoting interaction between members. They share their de-identified data within the community, to vendors, and partners. It is one of the major platforms for crowdsourced research (Kariotis et al., 2020). Other examples include Open Humans and Night Scout Data Commons that allow members to import and aggregate different types of personal data including wearable devices and genomic data. In this platform, governance occurs at multiple levels with individuals having control over their shared data at the basic level and at the functional level, the larger community invites members, reviews prospective new projects and votes for board members. The platform employs an extended consent procedure at the basic governance level such that when members share their data, they also give permission for diverse re-use (Aitken et al., 2016; Beier et al., 2019; Buyx et al., 2017; Jo & Nabatchi, 2020; Kariotis et al., 2020; Murtagh et al., 2018).

At the individual level, lay members of the public can participate by making decisions about different aspects of data governance, which include data sharing, access, use

and reuse. These individual-level decisions are typically enacted through different types of informed consent procedures (Beier et al., 2019). Broad consent offers data subjects limited opportunities for decision-making beyond the initial decision of sharing data. Specific consent enables data subjects to play a more active role in deciding who uses their data and for what purposes. And finally, dynamic consent enables the highest degree of involvement in decision-making by allowing data subjects to define and modify consent preferences over time, including decisions about the possible reuse of their data (Budin-Ljøsne et al., 2017; Kaye et al., 2015). The latter implies the creation of interactive platforms that enable data subjects to be notified of requests to use their data and to proceed with making a decision regarding consent over the duration of a specific project and beyond, when data is used for other purposes (Kaye et al., 2015).

1.2 Public views about involvement in health data governance

There has been growing support for public participation in health data governance, and studies have focused on whether patients, and other members of the public, value involvement in individual-level decisions about health data sharing, access, use and reuse. However, with a few relevant exceptions (HIQA, 2021), most studies tend to address the four key dimensions of individual-level data governance independently and only a few explored how trust in research initiatives influences the value attributed by different publics to involvement in health data decision-making (HIQA, 2021). In what follows, I present the key findings of these studies.

A national survey and focus group study done in Ireland sought the general publics' perspectives on sharing and access of personal health information for direct patients' care and health planning, as well as their opinion about secondary data use for digital health records and research. It found that people generally have a positive experience with the national health services and support the use of personal health information for direct care. They also support secondary data use for digital health records and research that seeks to improve quality of care if identifiable information is removed. Despite the enormous trust in health professionals held by participants, they were also concerned about data security and expressed their wish to have access to the data being shared and to be informed about the ways that their data may be used beyond

direct care (HIQA, 2021). Another study explored UK residents' and patients' (i.e. mental and physical health clinic users) opinion regarding secondary use of their data for clinical and research purposes without explicit consent. The study found that participants are more willing to share identifiable data without explicit consent for clinical care locally, and within the UK, however, they require explicit consent and de-identification of their data when it is used for research. They also showed more willingness to share physical health data than mental health data and this was found to be associated with people who have had mental ill-health experience (Jones et al., 2021).

Looking further into public perceptions in Australia about the sharing of health records held by government with private companies for research and drug development, Mayer, and colleagues' (2021) study reveals that most people are willing to share if their data would not be used for profit. However, they expressed concern about the ability of the Australian government to influence how private companies would use their data. When asked about their most preferred consent approach, the participants chose the opt-in as opposed to opt-out consent option. The former allows them to choose whether or not to share data, while the latter only notifies participants after sharing their data and gives them an option to withdraw in case they are not willing to continue with the research (Braunack-Mayer et al., 2021). A focus group study on data reuse probed participants of Genome Wide Association Studies (GWAS) who were contacted to provide a re-consent for allowing their data to be submitted to the US National Institute of Health (NIH) data bank, to understand their motivation for allowing reuse of their health data and their views about being contacted for a re-consent prior to submitting their data to NIH. The most stated reason for allowing reuse of data is to contribute to general patients' care, and to add to scientific knowledge. They highly rated the importance of being recontacted for consent before reuse and stated that it would have been inappropriate for their data to be submitted to the data bank without their consent (Ludman et al., 2010).

Shah and colleagues (2018) focused on research participants' preferences regarding the use of data beyond the purpose for which it was originally collected and found that most participants were supportive of secondary data use if they were involved in decision making about data sharing, namely by retaining control to withdraw their data

at any time (Shah et al., 2018). In a follow-up study about secondary data use, the authors also found that about half of the research participants valued the opportunity for involvement in decisions about who has access to their data (Shah et al., 2019). These findings portray the growing support and acceptability of the public for the advancement of health research beyond traditional patients' care. However, it also shows the inadequacy of the existing governance structures in ensuring data privacy and safety and maintaining public trust while sharing data for enhancement of health care and research.

In the field of rare disease, there is a limited but growing number of studies about the views of rare diseases patients and their informal carers about involvement in decisions concerned with individual-level health data governance. Darquy and colleagues (2016) studied the views and motivation of rare disease patients about sharing, access, and storage of their data for a research project focused on their specific disease (i.e. leukodystrophies), as well as their perception of the role of ethical committees in such research projects. Participants expressed their motivation to share data to: getting more knowledge of the disease; having access to the clinical trials; contributing to discovery of new therapies; and to belong to a community. However, concerns about data security and confidentiality remain a major reservation despite the respondents' support for data sharing. Nevertheless, participants stated that access to researchers outside the scope of the project can be permitted if their research is conducted for public good. Pharmaceutical companies can gain access to that data if anonymity, confidentiality, and security are maintained and if data ownership is not transferred to the pharmaceutical companies. And health professionals would also be able to access the data if they are specialists in the area of Leukodystrophies. Furthermore, most participants support data storage even after the death of a patient to enable research continuity and help those who are still facing the disease as a way of keeping a legacy of good cause for the deceased. Participants' expectations of an ethical committee are that it will ensure data safety and privacy and promote trustworthiness in the research project through transparency and avoidance of financial drift, as well as to listen to the problems and expectations of the patients and their families (Darquy et al., 2015). Another study focusing on data sharing by rare disease patients and their family members found that most participants would like to keep control over their shared data and that about half would not delegate to an ethics

committee the decision about whom their data will be shared with (Courbier e al., 2019). Finally, a study about data sharing found that most rare diseases patients would like to be re-contacted to decide on their data reuse and that not being given the opportunity to re-consent would be perceived as a threat to individual and group autonomy. It also found that participants' willingness to be involved in decision-making was motivated by concerns with data security and misuse (McCormack et al., 2016).

So far, it is evident in these studies that members of the public including rare disease communities are willing to support data usage and sharing that aims at harnessing the benefits of health data for research and advancement of health care. However, they also share common concerns about privacy and data security risks at various aspects of governance, and as such, they value consent mechanisms that allow more control of data and its accessibility. The vast majority of studies, as mentioned above, are based on independent analysis of multiple aspects of data governance and have not assessed public views about involvement in decisions concerning data sharing, access, use and reuse simultaneously. Moreover, there is still a scarce literature assessing the role played by trust in science and other institutions on the views of rare diseases patients and their carers about getting involved in individual-level health data governance This thesis aims to fill this gap and contribute to improve knowledge on a topic that appears to be of interest and concern to this community.

2 OBJECTIVES

This main aim of this dissertation is to improve the evidence base on public involvement in individual-level health data governance by inquiring people affected by rare diseases. Its specific objectives are:

1. To assess the views of rare diseases patients and their informal carers about involvement in decision-making regarding health data sharing, access, use and reuse.

2. To examine the influence of trust in science and other institutions on participants' views about involvement in decision-making regarding health data sharing, access, use and reuse, as well as their association with sociodemographic characteristics.

3 METHODS

This study entails a hospital-based survey that took place at two rare disease Reference Centres, namely the Reference Centre for Congenital Heart Diseases (RCCHD) and the Reference Centre for Inherited Metabolic Diseases (RCIMD), located at the University Hospital Centre S. João (UHCSJ), in Portugal.

3.1 Study design

This is an observational cross-sectional study that comprises a survey with rare diseases patients and their informal carers. The study draws on data collected in the scope of the larger project “DATAGov - Public and patient involvement in health data governance: a people-centred approach to data protection and sharing in genetic diseases” whose protocol is described elsewhere (De Freitas et al., 2021).

3.2 Participants

Between June 2019 and March 2020, patients with rare diseases and their informal carers who had medical appointments at two Reference Centres for Rare Diseases at the University Hospital Centre S. João (UHCSJ) were recruited to participate in the study. Following a consultation, patients aged 12 years and above, who can read and write Portuguese, and have no cognitive impairment, together with their informal carers, were handed a study information leaflet by a health professional (Annex 1). Subsequently, they were invited to take part in the study by a researcher who clarified any arising doubts or questions. All eligible participants who decided to participate were accompanied to a private setting, where they read and signed the informed consent form. Underage participants gave verbal consent, and their legal representatives signed the written consent form. All participants were asked to fill in a self-administered questionnaire individually (Annex 2). From the 728 people invited, 77 refused to take part in the study due to unwillingness to participate (n=37), lack of time (n=34), lack of consent from the legal tutor (n=3), limited literacy (n=2) and emotional distress following diagnosis (n=1). In total, 651 including 489 carers and 162

patients agreed to participate (response rate: 89.4%). This thesis included 637 participants (159 patients and 478 carers), with available data on all the outcomes.

3.3 Data collection

The structured questionnaire was developed by the interdisciplinary research team of the DATAGov project to assess public and patient involvement in health data governance, based on a review of literature and existing instruments related to the research topic. The questionnaire was pretested by specialists with combined experience as professionals, informal carers, and researchers (from the social and health sciences) and subsequently piloted by a group of patients and carers. Among others, it included questions about involvement in decision-making concerned with health data sharing, access, use and reuse, as well as sociodemographic characteristics (e.g. sex, age, educational level, marital status, occupation, and perceived income adequacy). Data on participants' involvement with patient organisations and trust (interpersonal trust and institutional trust) was also collected. To assess the importance attributed to involvement in decisions about health data sharing, access, use and reuse, participants answered the following four questions: 1) how important is it that you decide whether your data is shared for research purposes (data sharing); 2) how important is it that you decide whom your data is shared with (data access); 3) how important is it that you decide for what purposes your data is used for (data use); and, 4) how important is it that you decide whether your data can be used for purposes other than those for which it was initially collected (data reuse). The level of importance was rated using a 5-point Likert scale, ranging from "very important" to "not important" (range 1–5). For the analysis, the variables were categorized into "important" (including participants who answered "important" and "very important") and "other" (including "not important", "slightly important" and "moderately important").

Occupations were classified according to the Portuguese Classification of Occupations 2011 (Instituto Nacional Estatística, 2011) and grouped into four categories: 1) upper-white-collar (executive civil servants, industrial directors and executives, professionals and scientists, middle management and technicians); 2)

lower-white-collar (administrative and related workers, service and sales workers); 3) blue-collar (farmers and skilled agricultural workers, fisheries workers, skilled workers, craftsmen and similar, machine operators and assembly workers, and unskilled workers); and 4) other (students, unemployed, domestic workers, participants on disability pension or on paid/unpaid leave, retired and informal carers or members of a foster family). Perceived income adequacy was measured through the question "When thinking of your household income, would you say that your household is able to make ends meet?". Participants could check one of the following answer categories: insufficient, caution with expenses, enough to make ends meet, and comfortable.
Interpersonal and institutional trust were measured through ten self-administered questions based on the European Social Survey (ESS) rated on a scale from 0 to 10. Three questions were used to measure interpersonal trust: "Generally speaking, would you say that most people can be trusted or that you can't be too careful in dealing with people?"; "Do you think that most people would try to take advantage of you if they got the chance, or would they try to be fair?"; and "Would you say that most of the time people try to be helpful or that they are mostly looking out for themselves?". As reported in another study (Zmerli & Newton, 2008), principal component analysis to these three questions produced a single component, explaining 70% of the variance. Institutional trust was measured by asking the participants their level of trust in national institutions such as a country's parliament, the legal system, the police, politicians, and political parties, as well as international institutions, namely the European Parliament and the United Nations. The total score of the rating scales is divided by the number of valid responses to make the indexes ranging from 0 to 10, with higher scores indicating higher levels of trust.

The participants' views on the importance of trust in research host institutions and teams in decisions regarding data sharing were also assessed using a 5-point Likert scale ranging from "not important" to "very important" (range: 1-5) for the question: "There are some aspects people consider important to decide if they will share their health data for scientific research. If you had to make such decision, how important would you rate the following aspects: 1) trust in the institution hosting the research; 2) trust in the team conducting the research". For the analysis, the answers were dichotomized as "very important" and "other" (all other answers).

3.4 Data analysis

Data was described as counts and proportions for categorical variables, and medians and interquartile range (P25-P75) for non-normally distributed continuous variables. Means were compared with the Mann-Whitney test, and differences in frequencies and proportions of categorical variables were assessed using the Chi-square test or the Fisher exact test, as appropriate. Statistical significance was set at a value of $p < 0.01$. The statistical analyses were performed using the IBM Statistical Package for the Social Sciences (SPSS), version 27.0, Armonk, NY, USA.

3.5 Ethics

The study protocol of the larger project from which data for this study derives, received ethical approval from the Ethics Committee for Health from the UHCSJ / Faculty of Medicine of University of Porto (Ref. 99/19). All participants in the project were provided with detailed information about: the research objectives; the voluntary nature of participation in the study, stating that there will be no penalty or negative consequences for those who refuse to participate; the expected duration of the questionnaires; confidentiality and anonymity of all the information provided by the participants; privacy and data protection procedures; participants' entitlement to obtaining additional information and clarification of any aspect related to the research project; participants' right to freely withdraw participation from the study at any moment prior to publication of results, without any negative consequences; and the option to be informed about research results. This information was provided orally and by writing through an Information Sheet.

After clarifying any arising doubts, participants who took part in the questionnaire were given a consent form and asked to sign two copies (one for them to keep and another for the researcher). Underage participants gave verbal consent for participation and the consent form was signed by their legal guardians. Patients who were unable to give consent due to impairment or any other reason were not included in the study. There were no incentives for participation in the research project. All the research data collected, stored, processed, and analysed was pseudo-anonymised. To guarantee confidentiality and data protection, the questionnaires were filled in a private place.

Questionnaires do not disclose participants' identification and only the research team have access to identification data. Personal contacts, consent forms and questionnaires.

4 JOURNAL PAPER

Ngozi Nwebonyi, Susana Silva, Cláudia de Freitas. Public views about involvement in decision-making on health data sharing, access, use and reuse: the importance of trust in science and other institutions [Submitted]

Public views about involvement in decision-making on health data sharing, access, use and reuse: the importance of trust in science and other institutions

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ABSTRACT

Background: Data-intensive and needs-driven research can deliver substantial health benefits. However, concerns with privacy loss, undisclosed surveillance, and discrimination are on the rise due to mounting data breaches. This can undermine the trustworthiness of data processing institutions and reduce people's willingness to share their data. Involving the public in health data governance can help to address this problem by imbuing data processing frameworks with societal values. This study assesses public views about involvement in individual-level decisions concerned with health data and their association with trust in science and other institutions.

Methods: Cross-sectional study with 162 patients and 489 informal carers followed at two reference centers for rare diseases in an academic hospital in Portugal (June 2019-March 2020). Participants rated the importance of involvement in decision-making concerning health data sharing, access, use, and reuse from "not important" to "very important". Its association with sociodemographic characteristics, interpersonal trust, trust in national and international institutions, and the importance of trust in research teams and host institutions was tested.

Results: Most participants perceived involvement in decision-making about data sharing (85.1%), access (87.1%), use (85%) and reuse (79.9%) to be important or very important. Participants who ascribed a high degree of importance to trust in research host institutions were significantly more likely to value involvement in such decisions. A similar position was expressed by participants who valued trust in research teams for data sharing, access, and use. Participants with low levels of trust in national and international institutions and with lower levels of education attributed less importance to being involved in decisions about data use.

Conclusion: The high value attributed by participants to involvement in individual-level data governance stresses the need to broaden opportunities for public participation in health data decision-making, namely by introducing more dynamic approaches to consent. The important role played by trust in science and in other institutions in shaping participants' views about involvement highlights the relevance of pairing such dynamic consent approaches with the provision of transparent information about the implications of data sharing, the resources needed to make informed choices and the development of harm mitigation tools and redress.

INTRODUCTION

Health care quality improvement can be bolstered by data-intensive and needs-driven research (1). The use of big health data promises to transform biomedical and health care research and to deliver substantial public health benefits that range from disease risk prediction and prevention to the discovery of new therapies for untreatable health conditions, as are many rare diseases (2,3). However, mounting reports of data breaches and mismanagement have generated concern for privacy loss, undisclosed surveillance, and discrimination (4–6). These concerns can undermine public trust in data processing organizations (e.g., governmental, care and research institutions), which is key in shaping public attitudes towards data sharing and use (7,8). Perceived lack of institutional trustworthiness can limit people's willingness to share their data for research and to concede to its (re)use (9–11). There is, therefore, an imminent need for optimizing governance strategies to promote safe, acceptable, and beneficial uses of data in health research.

International policy agencies recommend the involvement of the public to ensure that data processing frameworks are consistent with societal values and individuals' expectations for the protection and use of their data (12,13). Public involvement is also substantiated by ethical arguments that center on the fair distribution of the benefits arising from data use (14,15). Furthermore, it has been argued that public involvement exercises can help foster authentic dialogue between researchers and publics, enhance accountability among data stewards on the governance chain and increase research trustworthiness, all of which are vital for ensuring and sustaining public trust in science (7).

Public involvement in health data governance entails awareness raising, consultation, partnering with and/or empowering of members of the public to participate in research and governance practices (16) and it can be set in motion through a variety of methods including deliberative polls, citizen juries, participatory appraisals, scenario-based workshops, and focus groups (16). Data holders can also participate via participant-led data cooperatives (e.g. Open Humans, DNA.Land, MIDATA) that enable them to share and aggregate their data while keeping control over its uses (17–20). At the individual level, public involvement can be fostered by enabling lay people to participate in decisions about particular aspects of data governance, including whether they want to share their data (*data sharing*), with whom they want to share their data with (*data access*), for what purposes it can be used (*data use*) and whether data can be shared for purposes other than those for which they were originally collected (*data reuse*). These individual-level decisions are typically enacted through different

types of informed consent procedures (21,22). Broad consent offers data donors limited opportunities for decision-making beyond the initial decision of sharing data. Specific consent enables data donors to play a more active role in deciding who uses their data and for what purposes. And finally, dynamic consent enables the highest degree of involvement in decision-making by allowing data donors to define and modify consent preferences over time, including decisions about the possible reuse of their data (23,24). The latter implies the creation of interactive platforms that enable data subjects to be notified of requests to use their data and re-contacted to proceed with making a decision regarding consent (25,26).

Opportunities for public involvement have expanded substantially in the past decade and there is a growing interest in understanding whether patients, and other members of the public, value involvement in individual-level decisions about health data sharing, access, use and reuse (27–34). Ludman et al. (2010) found that research participants wanted to decide whether their previously shared data could be submitted to a new database through active engagement in reconsenting procedures despite 'their extraordinary trust in the research team'. Similarly, another study showed that patients would like to be re-contacted to decide on the reuse of their data and that not being given the opportunity to reconsent would be perceived as a threat to individual and group autonomy (29). Courbier et al. (2019) found that patients and their family members would like to keep control over their shared data and that about half would not delegate the decision about whom their data will be shared with to an ethics committee. And a study involving research participants in four European countries showed they were supportive of de-identified data reuse if they were involved in decision-making about data sharing and access, namely by retaining control to withdraw their data at any time (29).

Most existing studies address the multiple aspects of individual-level data governance independently and few have explored how trust in research initiatives influences the value bestowed by different publics on involvement in data decision-making (28,32). In this study, we assess the views of rare disease patients and their informal carers about being involved in decisions regarding data sharing, access, use and reuse with a focus on the role played by trust in science and other institutions. Most rare diseases have no treatment and specific rare disease populations are very small and scattered geographically (28,35). Data sharing within and between countries is therefore essential for enabling research that can advance the development of accurate diagnoses and therapies (36). However, this type of research requires a combination of genetic and phenotypic information which presents a high privacy risk for these patients and their relatives. Assessing rare disease patients and carers' views about involvement in decision-making concerned with their data can help in designing a data governance structure suitable to

meet their needs and expectations from biomedical and health care research and to enhance the trustworthiness of institutions involved in research (14,37).

MATERIALS AND METHODS

Participants

This observational and cross-sectional study is part of a mixed methods project focusing on public involvement in health data governance whose protocol is described elsewhere (38). Participants include people with rare diseases and their informal carers who were consecutively recruited from two Reference Centers for Rare Diseases at the University Hospital Centre S. João (UHCSJ), in Portugal, between June 2019 and March 2020. Following a consultation, patients aged 12 years and above and their carers were handed a study information leaflet by a health professional. Subsequently, they were invited to participate in the study by a researcher who clarified any arising doubts or questions. Those who decided to participate were accompanied to a private setting, where they read and signed the informed consent. Underage participants who agreed to participate gave verbal consent and the informed consent form was signed by their legal representatives. All participants were asked to fill in a self-administered questionnaire individually.

Of the 728 people invited, 77 refused to take part in the study due to unwillingness to participate (n=37), lack of time (n=34), lack of consent from the legal tutor (n=3), limited literacy (n=2) and emotional distress following diagnosis (n=1). In total, 651 people (162 patients and 489 carers) agreed to participate (response rate: 89.4%).

Data collection

The structured questionnaire was developed by the research team based on a review of literature and existing instruments related to the research topic. The questionnaire was pretested by specialists with combined experience as professionals, informal carers and researchers (social and health sciences) and subsequently piloted by a group of patients and carers.

The assessment of the importance attributed by participants to involvement in decisions about health data sharing, access, use and reuse was based on the analysis of answers to four questions: 1) how important is it that you decide whether your data is shared for research purposes (*data sharing*); 2) how important is it that you decide whom your data is shared with (*data access*); 3) how important is it that you decide for what purposes your data is used for (*data use*); and, 4) how important is it that you decide whether your data can be used for

purposes other than those for which it was initially collected (*data reuse*). The level of importance was rated using a 5-point Likert scale, ranging from "very important" to "not important" (range 1–5). For this analysis, the variables were categorized into "important" (including participants who answered "important" and "very important") and "other" (including "not important", "slightly important" and "moderately important"). This study included 637 participants (159 patients and 478 carers), with available data on all the above-mentioned outcomes.

Data on sociodemographic characteristics (sex, age, educational level, marital status, occupation, and perceived income adequacy), as well as participants' involvement with patient organizations were collected. Occupations were classified according to the Portuguese Classification of Occupations 2011 (39) and grouped into four categories: 1) upper-white-collar, including executive civil servants, industrial directors and executives, professionals and scientists, middle management and technicians; 2) lower-white-collar, including administrative and related workers, service and sales workers; 3) blue-collar, which includes farmers and skilled agricultural workers, fisheries workers, skilled workers, craftsmen and similar, machine operators and assembly workers, and unskilled workers; and 4) other, including students, unemployed, domestic workers, participants on disability pension or on paid/unpaid leave, retired and informal carers or members of a foster family. Perceived income adequacy was measured through the question "When thinking of your household income, would you say that your household is able to make ends meet?". Participants could check one of the following answer categories: insufficient, caution with expenses, enough to make ends meet, and comfortable.

Interpersonal trust, trust in national institutions and trust in international institutions were measured through ten self-administered questions based on the European Social Survey (ESS) rated on a scale from 0 to 10. Interpersonal trust was measured by three questions: "Generally speaking, would you say that most people can be trusted or that you can't be too careful in dealing with people?"; "Do you think that most people would try to take advantage of you if they got the chance, or would they try to be fair?"; and "Would you say that most of the time people try to be helpful or that they are mostly looking out for themselves?". As reported in another study (40) principal component analysis to these three questions produced a single component, explaining 70% of the variance. Institutional trust was measured by asking participants how they trusted national institutions such as a country's parliament, the legal system, the police, politicians, and political parties, as well as international institutions, namely the European Parliament and the United Nations. Principal component analysis of the dataset

shows that the variables are well suited for constructing two indexes, one for trust in national institutions and another for trust in international institutions. The total score of the rating scales is divided by the number of valid responses to make the indexes ranging from 0 to 10, with higher scores indicating higher levels of trust.

The views of patients and carers about the importance of trust in research host institutions and in research teams in decisions regarding data sharing were assessed using a 5-point Likert scale ranging from "not important" to "very important" (range: 0-4) for the question: “There are some aspects people consider important to decide if they will share their health data for scientific research. If you had to make such decision, how important would you rate the following aspects: 1) trust in the institution hosting the research; 2) trust in the team conducting the research”. For this analysis, the answers were dichotomized as "very important" and "other" (all other answers).

Data analysis

Categorical variables are presented as counts and proportions, while continuous variables were summarized as medians and interquartile range (P25-P75). The Chi-square test or the Fisher exact test, as well as the Mann-Whitney test were used, as appropriate, to assess the associations and mean differences between the explanatory variables and the outcomes. Statistical significance was set at a value of $p < 0.01$. The statistical analyses were performed using the software IBM SPSS Statistics for Windows, version 27.0 (IBM Corp., Armonk, N.Y., USA).

RESULTS

The characteristics of the participants and their views about involvement in decision-making on health data sharing, access, use, and reuse are presented in Tables 1 and 2, respectively. Most participants attained 12 or less years of education (75.6%) and were not involved with patient’s organizations (94.9%). Almost 80% of the carers were female, while over 53% of the patients were male. Carers were older (> 30 years) than patients (87.2% vs. 15.1%) and more frequently married or living with a partner (78.0% vs. 9.5%). More than half of carers perceived their income as insufficient (56.2%), while 64.6% of patients considered it comfortable/enough to make ends meet. About three quarters of the carers and two thirds of the patients perceived trust in research host institutions and trust in research teams as very important issues when making decisions about sharing data. Participants presented low levels of trust in national institutions (Median [P25-P75] 3.5 [1.8-5.2]), increasing slightly for trust in international

institutions (Median [P25-P75] 5.0 [2.5-7.0]) and interpersonal trust (Median [P25-P75] 4.7 [3.0-6.7]).

Most participants considered it important or very important to be involved in decisions concerned with health data sharing (85.1%), access (87.1%), use (85%) and reuse (79.9%). This trend was observed among both patients and carers (Table 2).

Carers and older participants stated more frequently the importance of being involved in decision-making regarding data sharing and data access (Table 3). More educated participants revealed a statistically significant tendency to attribute more importance to participation in decisions about data use, while participants with the lowest levels of trust in national and international institutions (Median[P25-P75]: 2.2[0.8-4.0]) and 3.0[1.0-5.0], respectively) were less likely to value such type of involvement. Participants who considered trust in research host institutions as very important rated higher the importance of being involved in decisions about data sharing, data access, data use, and data reuse. A similar position was primarily expressed by participants who valued trust in research teams for data sharing, data access, and data use.

DISCUSSION

The majority of people affected by rare diseases who were surveyed placed a high value on opportunities for involvement in decisions about health data sharing, access, use and reuse (ranging between 80-87%). These views differ from those of other publics such as people with diabetes among whom less than 50% considered important to decide what type of data can be shared and with whom (27). However, they are echoed by rare diseases communities across Europe who expressed a strong desire in keeping control over their shared data throughout the data processing cycle (80%) (28). Difficulties in obtaining diagnoses, the absence of cures, and oftentimes of treatment, inspire a firm commitment on the part of rare disease patients and their carers towards advancing research, which is further strengthened by a perceived need to optimise the use of scarce biospecimens and research resources (28,30,34,36,41). These challenges may explain rare diseases participants' eagerness to engage in decisions about how their data should be governed. Playing an active role in deciding what data can be shared, with whom and for which purposes can help to not only reorient governance frameworks to become more commensurate with their values and preferences, but also directly impact their lives, and those of future generations, by driving research and care to meet their specific needs (28,34).

Our findings also show a strong positive association between the value attributed to trust in science and the value attributed to public involvement in data governance. Participants who ascribed a high degree of importance to trust in research institutions when choosing whether to share their data were significantly more likely to value involvement across the full spectrum of aspects related with individual-level data governance (data sharing, access, use and reuse). A similar pattern was found for trust in researchers and involvement in decisions about data sharing, access and use. These findings resonate with Aitken and colleagues' (7) argument that public involvement is one out a set of institutional arrangements that are central in ensuring the trustworthiness of research, which, in turn, is required to foster public trust in science. Scientific initiatives guided by participatory ideals privilege reciprocity and acknowledge participants' expectations, needs and agency, not least by facilitating a dynamic approach to consent (24). Dynamic consent approaches afford participants an ongoing opportunity to decide the conditions in which their data can be shared, accessed, used and reused, over time and across a range of research initiatives and settings. These approaches also contribute to the establishment of ongoing communication with, and feedback from, researchers that can give rise to more substantive participatory initiatives (e.g. public deliberation exercises; public engagement in data access committees) (24,42–45). Such participatory initiatives carry potential to increase research transparency and to promote accountability by enabling researchers and diverse publics to come together and build dialogic relationships that are essential for uncovering existing concerns and imbuing systems of data governance with public values and the mechanisms needed to ensure oversight and redress for misconduct (7,46). However, while public involvement can enhance research trustworthiness, (7,47,48), a minimum level of public trust in science has to be present for public involvement to unfold (43). Our study corroborates these findings by showing that rare diseases patients and their carers are significantly more likely to value involvement in health data governance when they hold trust in science in high regard.

Following a wider international trend (49–51) , the Portuguese population has reported relatively high levels of trust in science (49). Yet, its level of trust in other institutions, including the European Parliament, national government, and the legal and health care systems, tends to be substantially lower (52–54). This was also observed among participants in our study who expressed low trust in national institutions and, to a lesser extent, in international institutions. Importantly, our study also shows that participants with the lowest levels of trust in national and international institutions attributed significantly less importance to getting involved in decision-making about how their data can be used. This trend may find explanation

in the idea that public involvement is unlikely to inspire reciprocal partnerships and lead to transformative change in institutions perceived to be opaque, irresponsive, and unaccountable (55). Effecting change that is transformative requires the development of trusting relationships between institutional stakeholders and lay members of the public, the ability to accommodate and build on different types of knowledge and expertise and a thorough commitment to attending to the needs, and responding to the concerns, of the various parties involved (56). Where institutions fail to cultivate trust, incentives for involvement may wane or disappear altogether (43). Participatory exercises demand time, skills, and the confidence that the efforts made are grounded on transparent information and can foster the change needed to engender meaningful partnerships and ensure accountability (7). When these conditions are not met the drive for participation tends to plummet.

Carers and older participants in our study were more prone to value involvement in decisions about data sharing and data access. These findings align with those of an international survey carried out with people affected by rare diseases that found that participants identifying as patient representatives and older respondents were both more likely to perceive health-related information as sensitive and to want to retain control over who accesses their information, how and why (28).

Finally, our study shows that participants with lower levels of education attributed significantly less importance to involvement in decision-making about the purposes for which their data can be used. This finding may be pointing to an unequal distribution of the resources needed to make informed decisions about data use. Big data, machine learning and artificial intelligence have contributed to expand the purposes of biomedical and healthcare research to a multitude of fast-evolving fields (57). Increasingly, research endeavours focus on issues that lay people may not be familiar with and feel wary to express opinions about (e.g. gene therapy) (58). Disregard for the needs of publics who are less equipped to assess the value and risks of cutting-edge research can contribute to avert their participation. Moreover, it can reinforce a long-lasting pattern of exclusion found across the European Region where minority and socioeconomically disadvantaged groups have been systematically under-represented in health research as well as in the participatory spaces created to involve lay people in its design and implementation (15,60).

Assessing and attending to consent preferences and offering time and support to anyone expected to make informed decisions is essential (61,62). However, with the exponential growth of data sources and data uses, informal support may not be sufficient to enable informed consent (38). As argued by Fiske et al. (2019b), it is necessary to make way for a new group

of professionals – health information counsellors – who can advise on the far-reaching implications of data decisions and assist in addressing arising ethical, legal and social challenges and dilemmas that often extend beyond the individual sphere (e.g. the right to choosing not to know and, thus, to decline the return of incidental research findings that may identify a predisposition for late-onset genetic diseases with implications for the offspring) (63). Health information counselling services may be especially relevant for decisions concerned with the use of one's data for purposes other than those for which it was originally collected. The reuse of health data can occur in contexts with norms and values different from those upheld in research and care settings and which are more often subject to "data trust deficits" (64). Commercial settings such as direct-to-consumer genetic testing companies are one such example where values such as transparency and reciprocity may be overridden by economic interest (e.g. patenting consumer data that was first shared under the pretense that it would be used to democratise genomics) (see 65). Elucidating on the ethical, legal and social implications of sharing data for research, care, commercial and other secondary purposes is of critical importance to reduce resource gaps, inform lay people's expectations, empower them to make informed decisions and promote the trustworthiness of data processing organisations.

Strengths and limitations

This study offers three major contributions. First, it is one of a few studies to assess public views about involvement in all key dimensions of individual-level data governance and to enable the identification of differences in the importance attributed to participation in decision-making concerned with health data sharing, access, use, and reuse. Another major contribution relates to the examination of its association with various types of trust and sociodemographic variables. Finally, data collection was carried out over an extended recruitment period of 10 months and participants were consecutively invited to participate at two reference centers for rare diseases located in an academic hospital center that oversees patients from the entire Northern Health Region of Portugal. Nevertheless, recruitment in one region limits the generalizability of the results and thus inferences for the general rare diseases population should be performed with caution. Furthermore, the value attributed to opportunities for involvement in decisions about health data sharing, access, use and reuse may be overestimated in this particular setting, as the reference centers have a strong academic orientation and are involved with rare diseases European Reference Networks. Many of the patients and carers surveyed have been involved in data sharing for national and international research projects

and are experienced in decision-making concerned with their health data. The recruitment of participants in non-academic and in private settings would enable an enriching comparison. Finally, further qualitative and quantitative research is warranted to uncover participants' motivations and expectations regarding involvement in individual-level data governance.

CONCLUSION

The high value attributed by participants to involvement in individual-level data governance stresses the need to rethink opportunities for public participation in health data decision-making. Broadening the consent options currently on offer to people affected by rare diseases to include mechanisms (e.g. digital communication interface) (24) through which they can engage more directly with research and express preferences regarding data sharing, access, use and reuse deservers thorough consideration. Trust in science and other institutions played an important role in shaping our participants' views about involvement. Accordingly, the adoption of a more dynamic approach to consent would likely need to be accompanied by the provision of transparent information about the implications of data sharing, assistance with obtaining the resources needed to make informed choices and the development of harm mitigation tools and redress.

Conflict of Interest

The authors declare that the research was conducted in the absence of any commercial or financial relationships that could be construed as a potential conflict of interest.

Author Contributions

CDF conceptualized the study. SS and CDF designed the data analysis strategy. NN performed data analysis. NN and CDF wrote the first draft of the manuscript. NN, SS and CDF contributed to the interpretation of data, critically reviewed previous versions of the manuscript and approved the final version.

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Ethics

Ethical approval was granted by the Ethics Committee for Health from the UHCSJ/Faculty of Medicine of University of Porto (Ref. 99/19). The study followed the norms of ethical research practice laid out in the Declaration of Helsinki. All research data collected, stored, processed and analyzed have been pseudo anonymized.

References

1. Rumsfeld JS, Joynt KE, Maddox TM. Big data analytics to improve cardiovascular care: promise and challenges. Nat Rev Cardiol (2016) 13:6. doi: 10.1038/nrcardio.2016.42
2. Flores M, Glusman G, Brogaard K, Price ND, Hood L. P4 medicine: how systems medicine will transform the healthcare sector and society. P Med (2013) 10:6. doi: 102217/pme1357
3. Benson M. Clinical implications of omics and systems medicine: focus on predictive and individualized treatment. J Int Med (2016) 279:3. doi: 10.1111/joim.12412
4. Prainsack B. The political economy of digital data: introduction to the special issue. Pol Stud. (2020) 41:5. doi: 10.1080/01442872.2020.1723519
5. Pollock AM, Roderick P. Trust in the time of markets: protecting patient information. The Lancet (2014) 383:9928. doi: 10.1016/S0140-6736(14)60727-3
6. Wadmann S, Hoeyer K. Dangers of the digital fit: Rethinking seamlessness and social sustainability in data-intensive healthcare. Big Data & Society (2018) 5:1. doi: 10.1177/2053951717752964
7. Aitken M, Cunningham-Burley S, Pagliari C. Moving from trust to trustworthiness: Experiences of public engagement in the Scottish Health Informatics Program. Sci and Pub Pol (2016) 43:5. doi: 10.1093/scipol/scv075
8. Trinidad SB, Fullerton SM, Bares JM, Jarvik GP, Larson EB, Burke W. Genomic research, and wide data sharing: Views of prospective participants. Genet Med (2010) 12:8. doi: 10.1097/GIM.0b013e3181e38f9e
9. Oderkirk J, Ronchi E. Governing data for better health and healthcare. OECD iLibrary (2018). doi: 10.1787/6b33a920-en [Accessed on December 10, 2021]
10. MORI I. The One-Way Mirror: Public attitudes to commercial access to health data. Wellcome Trust (2015). doi: 10.6084/m9.figshare.5616448.v1
11. Milne R, Morley KI, Almarri MA, Anwer S, Atutornu J, Baranova EE, et al. Demonstrating trustworthiness when collecting and sharing genomic data: public views across 22 countries. Gen Med (2021) 13:1. doi: 10.1186/s13073-021-00903-0
12. OECD. Recommendation of the Council on Health Data Governance. OECD (2016). https://www.oecd.org/health/health-systems/Recommendation-of-OECD-Council-on-Health-Data-Governance-Booklet.pdf [Accessed on December 10, 2021]

13. OECD. Health data governance: privacy, monitoring and research – policy brief. OECD (2015). https://www.oecd.org/health/health-systems/Health-Data-Governance-Policy-Brief.pdf [Accessed on December 10, 2021]
14. Kaye J, Terry SF, Juengst E, Coy S, Harris JR, Chalmers D, et al. Including all voices in international data sharing governance. Hum Gen (2018) 12:13. doi: 10.1186/s40246-018-0143-9
15. Fiske A, Prainsack B, Buyx A. Meeting the needs of underserved populations: setting the agenda for more inclusive citizen science of medicine. J. Med Ethics (2019) 45:9. doi: 10.1136/medethics-2018-105253
16. Aitken M, Tully MP, Porteous C, Denegri S, Cunningham-Burley S, Banner N, et al. Consensus Statement on Public Involvement and Engagement with Data Intensive Health Research. Int J Pop Data Sci (2019) 4:1. doi: 10.23889/ijpds.v4i1.586
17. Kariotis T, Ball MP, Greshake Tzovaras B, Dennis S, Sahama T, Johnston C, et al. Emerging health data platforms: From individual control to collective data governance. Data Pol (2020) 2. doi: 10.1017/dap.2020.14
18. Buyx A, Savio L del, Prainsack B, Vo H. Every participant is a PI. Citizen science and participatory governance in population studies. Int J Epid (2017) 46:2. doi: 10.1093/ije/dyw204
19. Vayena E, Blasimme A. Biomedical big data: New models of control over access, use and governance. J Bioethic Inquiry (2017) 14:4 doi: 10.1007/s11673-017-9809-6
20. Hafen E, Kossmann D, Brand A. Health data cooperatives - Citizen empowerment. Methods Inf Med (2014) 53:2 doi: 10.3414/ME13-02-0051
21. Beier K, Schweda M, Schicktanz S. Taking patient involvement seriously: A critical ethical analysis of participatory approaches in data-intensive medical research. BMC Med Inform Decis Mak (2019) 19:90. doi: 10.1186/s12911-019-0799-7
22. Kaye J, Curren L, Anderson N, Edwards K, Fullerton SM, Kanellopoulou N, et al. From patients to partners: participant-centric initiatives in biomedical research. Nat Rev Genet (2012) 13:5. doi: 10.1038/nrg3218
23. Budin-Ljøsne I, Teare HJA, Kaye J, Beck S, Bentzen HB, Caenazzo L, et al. Dynamic Consent: A potential solution to some of the challenges of modern biomedical research. BMC Med Ethics (2017) 18:1. doi: 10.1186/s12910-016-0162-9
24. Kaye J, Whitley EA, Lund D, Morrison M, Teare H, Melham K. Dynamic consent: A patient interface for twenty-first century research networks. Eur J Hum Genet (2015) 23:2. doi: 10.1038/ejhg.2014.71

25. Teare HJA, Morrison M, Whitley EA, Kaye J. Towards 'Engagement 2.0': Insights from a study of dynamic consent with biobank participants. Digital Health (2015) 1. doi: 101177/2055207615605644
26. Thiel DB, Platt J, Platt T, King SB, Fisher N, Shelton R, et al. Testing an online, dynamic consent portal for large population biobank research. Pub Health Gen (2015) 18:1. doi: 10.1159/000366128
27. Shah N, Coathup V, Teare H, Forgie I, Nicola G, Tue G, et al. Motivations for data sharing — views of research participants from four European countries: A DIRECT study. Eur J Hum Genet (2019) 27. doi: 10.1038/s41431-019-0344-2
28. Courbier S, Dimond R, Bros-facer V. Share and protect our health data: an evidence-based approach to rare disease patients' perspectives on data sharing and data protection - quantitative survey and recommendations. Orphanet J Rare Dis (2019) 14:1. doi:10.1186/s13023-019-1123-4
29. Shah N, Coathup V, Teare H, Forgie I, Giordano GN, Hansen TH, et al. Sharing data for future research — engaging participants' views about data governance beyond the original project: A DIRECT Study. Genet in Med (2019) 21:5. doi: 10.1038/s41436-018-0299-7
30. McCormack P, Kole A, Gainotti S, Mascalzoni D, Molster C, Lochmüller H, et al. "You should at least ask". The expectations, hopes and fears of rare disease patients on large-scale data and biomaterial sharing for genomics research. Eur J Hum Genet (2016) 24:10. doi: 10.1038/ejhg.2016.30
31. Riordan F, Papoutsi C, Reed JE, Marston C, Bell D, Majeed A. Patient and public attitudes towards informed consent models and levels of awareness of Electronic Health Records in the UK. Int J Med Inf (2015) 84:4. doi: 10.1016/j.ijmedinf.2015.01.008
32. Ludman EJ, Fullerton SM, Spangler L, Trinidad SB, Fujii MM, Jarvik GP, et al. Glad you asked: Participants' opinions of re-consent for DBGAP data submission. J Emp Res Hum Ethics (2010) 5:3. doi: /10.1525/jer.2010.5.3.9
33. Bell EA, Ohno-Machado L, Grando MA. Sharing my health data: A Survey of data sharing preferences of healthy individuals. AMIA Annu Symp Proc (2014) 14.
34. Darquy S, Moutel G, Lapointe AS, D'Audiffret D, Champagnat J, Guerroui S, et al. Patient/family views on data sharing in rare diseases: study in the European LeukoTreat project. Eur J Hum Genet (2015) 24:3. doi: 10.1038/ejhg.2015.115

35. Boulanger V, Schlemmer M, Rossov S, Seebald A, Gavin P. Establishing patient registries for rare diseases: Rationale and challenges. Pharmaceut Med (2020) 34:3. doi: 10.1007/s40290-020-00332-1
36. De Freitas C, dos Reis V, Silva S, Videira PA, Morava E, Jaeken J. Public and patient involvement in needs assessment and social innovation: A people-centred approach to care and research for congenital disorders of glycosylation. BMC Health Serv Res (2017) 17:1. doi: 10.1186/s12913-017-2625-1
37. Thorogood A. International data sharing and rare disease: The importance of ethics and patient involvement. In: Rare Diseases. (2020) IntechOpen, London. doi: 10.5772/intechopen.91237
38. De Freitas C, Amorim M, MacHado H, Leão Teles E, Baptista MJ, Renedo A, et al. Public and patient involvement in health data governance (DATAGov): protocol of a people-centred, mixed-methods study on data use and sharing for rare diseases care and research. BMJ Open (2021) 11:3. doi: 10.1136/bmjopen-2020-044289
39. Instituto Nacional Estatística - Classificação Portuguesa das Profissões. (2010). https://www.ine.pt/xurl/pub/107961853 [Accessed on December 11, 2021
40. Zmerli S, Newton K. Social trust and attitudes toward democracy. Public Opinion Quarterly (2008) 72:4. doi: 10.1093/poq/nfn054
41. Dwyer AA, Quinton R, Morin D, Pitteloud N. Identifying the unmet health needs of patients with congenital hypogonadotropic hypogonadism using a web-based needs assessment: Implications for online interventions and peer-to-peer support. Orphanet J Rare Dis (2014) 9:1. doi: 10.1186/1750-1172-9-83
42. Murtagh MJ, Blell MT, Butters OW, Cowley L, Dove ES, Goodman A, et al. Better governance, better access: Practising responsible data sharing in the METADAC governance infrastructure. Hum Genomics (2018) 12:1. doi: 10.1186/s40246-018-0154-6
43. Kraft SA, Cho MK, Gillespie K, Halley M, Varsava N, Ormond KE, et al. Beyond consent: Building trusting relationships with diverse populations in precision medicine research. Am J Bioeth (2018) 8:4. doi: 101080/1526516120181431322
44. Spencer K, Sanders C, Whitley, Edgar A, Lund D, Kaye J, William, et al. Patient perspectives on sharing anonymized personal health data using a digital system for dynamic consent and research feedback: A aualitative study. JMIR (2016) 8:4. doi: 10.2196/jmir.5011

45. Burgess MM. From “trust us” to participatory governance: Deliberative publics and science policy. Pub Understanding of Sci (2014) 23:1. doi: 10.1177/0963662512472160
46. O’Doherty KC, Burgess MM, Edwards K, Gallagher RP, Hawkins AK, Kaye J, et al. From consent to institutions: Designing adaptive governance for genomic biobanks. Soc Sci & Med. (2011) 73:3. doi: 10.1016/j.socscimed.2011.05.046
47. Kerasidou A. Trust me, I’m a researcher!: The role of trust in biomedical research. Med Health Care and Philos. (2016) 20:1. doi: 10.1007/s11019-016-9721-6
48. Moodley K, Singh S. “It’s all about trust”: reflections of researchers on the complexity and controversy surrounding biobanking in South Africa. BMC Med Ethics (2016) 17:1. doi: 10.1186/s12910-016-0140-2
49. Gallup. How does the world feel about science and health? Wellcome Global Monitor (2019). https://cms.wellcome.org/sites/default/files/wellcome-global-monitor-2018.pdf [Accessed on January 10, 2022]
50. Funk C, Hefferon M, Kennedy B, Johnson C. Trust and mistrust in Americans’ views of scientific experts. Pew Res Center (2019) 2.
51. Castell S, Charlton A, Clemence M, Pettigrew N, Pope S, Quigley A, et al. Public Attitudes to science. Ipsos Mori Soc Res Inst (2014) 194:28.
52. Asensio M. The Political Legitimacy of the healthcare system in Portugal: Insights from the European Social Survey. Healthcare (2021) 9:202. doi: 10.3390/healthcare9020202
53. Dotti Sani GM, Magistro B. Increasingly unequal? The economic crisis, social inequalities, and trust in the European Parliament in 20 European countries. Euro J Pol Res (2016) 55:2. doi: 10.1111/1475-6765.12126
54. Torcal M. The Decline of Political Trust in Spain and Portugal: Economic performance or political responsiveness? Am Behav Sci (2014) 58:12. doi: 10.1177/0002764214534662
55. Gaventa J. ‘Towards participatory governance: assessing the transformative possibilities’. In: Hickey S, Mohan G (eds) Towards participatory governance: assessing the transformative possibilities’ Participation: From tyranny to transformation. Zed Books Ltd, New York (2004) p. 25–41.
56. De Freitas C, Martin G. Inclusive public participation in health: Policy, practice, and theoretical contributions to promote the involvement of marginalised groups in healthcare. Soc Sci & Med (2015) 135:3. doi: 10.1016/j.socscimed.2015.04.019

57. Obermeyer Z, Emanuel EJ. Predicting the Future — Big data, machine learning, and clinical medicine. The N Eng J Med (2016) 375:13. doi: 10.1056/NEJMp1606181
58. Fiske A, Buyx A, Prainsack B. Health Information Counselors: A new profession for the age of big data. Acad Med (2019) 94:1. doi: 10.1097/ACM.0000000000002395
59. World Health Organisation. More than numbers — evidence for all. European Health report. (2018). https://www.euro.who.int/__data/assets/pdf_file/0003/380478/HEALTH_REPORT_HIGHLIGHTS_2018_EN.PDF [Accessed on December 22, 2021]
60. MacFarlane A, Ogoro M, de Freitas C, Niranjan V, Severoni S, Waagensen E. Migrants' involvement in health policy, service development and research in the WHO European Region: A narrative review of policy and practice. Trop Med & Inter Health (2021) 26:10. doi: 10.1111/tmi.13643
61. Baía I, de Freitas C, Samorinha C, Provoost V, Silva S. Dual consent? Donors' and recipients' views about involvement in decision-making on the use of embryos created by gamete donation in research. BMC Med Ethics (2019) 20:1. doi: 10.1186/s12910-019-0430-6
62. Gainotti S, Turner C, Woods S, Kole A, McCormack P, Lochmüller H, et al. Improving the informed consent process in international collaborative rare disease research: effective consent for effective research. Eur J Hum Genet (2016) 24:9. doi: 10.1038/ejhg.2016.2
63. Kleiderman E, Knoppers BM, Fernandez C, Boycott KM, Ouellette G, Wong-Rieger D, et al. Returning incidental findings from genetic research to children: views of parents of children affected by rare diseases. J Med Ethics (2014) 40:10. doi: 10.1136/medethics-2013-101648
64. Shah N, Johansson JV, Haraldsdóttir E, Bentzen HB, Coy S, Mascalzoni D, et al. Governing health data across changing contexts: A focus group study of citizen's views in England, Iceland, and Sweden. Int J Med Inf (2021) 156:138. doi: 10.1016/j.ijmedinf.2021.104623
65. Sterckx S, Cockbain J, Howard H, Huys I, Borry P. "Trust is not something you can reclaim easily": patenting in the field of direct-to-consumer genetic testing. Genet Med (2013) 15:5. doi: 10.1038/gim.2012.143

Supplementary Material

None.

Data Availability Statement

The datasets generated and analyzed for this study are not publicly available due to a confidentiality agreement securing participants' privacy and anonymity, but they are available from the corresponding author upon reasonable request.

Table 1. Characterization of the participants, stratified by people with rare diseases and their informal carers

Participants	Total (N=637)	Patients (n=159)	Carers (n=478)
Sex, n (%)			
Female	453 (71.1)	75 (47.2)	378 (79.1)
Male	184 (28.9)	84 (52.8)	100 (20.9)
Age (years), n (%)			
<18	92 (14.6)	92 (57.9)	-
18-30	103 (16.4)	43 (27.0)	60 (12.8)
>30	434 (69.0)	24 (15.1)	410 (87.2)
Educational level (years), n (%)			
≤ 12	476 (75.6)	151 (95.6)	325 (68.9)
> 12	154 (24.4)	7 (4.4)	147 (31.1)
Marital status, n (%)			
Married/living with partner	384 (60.9)	15 (9.5)	369 (78.0)
Other	247 (39.1)	143 (90.5)	104 (22.0)
Occupation, n (%)			
Upper white-collar	148 (24.6)	5 (3.2)	143 (32.3)
Lower white-collar	116 (19.3)	8 (5.1)	108 (24.4)
Blue-collar	87 (14.5)	9 (5.7)	78 (17.6)
Other	250 (41.6)	136 (86.1)	114 (25.7)
Perceived income adequacy, n (%)			
Insufficient/Caution with expenses	315 (51.3)	51 (35.4)	264 (56.2)
Enough to make ends meet/comfortable	299 (48.7)	93 (64.6)	206 (43.8)
Involvement in patient organizations, n (%)			
No	598 (94.9)	154 (98.1)	444 (93.9)
Yes	32 (5.1)	3 (1.9)	29 (6.1)
Trust in research host institution, n (%)			
Very important	461 (73.6)	107 (67.7)	354 (75.6)
Other	165 (26.4)	51 (32.3)	114 (24.4)
Trust in research team, n (%)			
Very important	448 (71.5)	103 (65.6)	345 (73.4)
Other	179 (28.5)	54 (34.4)	125 (26.6)
Trust in national institutions, Md (P25-P75)	3.5 (1.8-5.2)	4.5 (2.0-6.0)	3.4 (1.8-5.0)
Trust in international institutions, Md (P25-P75)	5.0 (2.5-7.0)	6.5 (3.0-8.0)	5.0 (2.0-7.0)
Interpersonal trust, Md (P25-P75)	4.7 (3.0-6.7)	4.7 (2.4-6.7)	4.7 (3.0-6.4)

Note: In each variable, the total may not add 637 participants, 159 patients or 478 carers due to missing values. The proportions may not add 100 due to rounding.

Table 2. Participants' views about involvement in decision-making regarding health data sharing, access, use and reuse

Involvement in decision-making regarding:	Total (N=637) *n* (%)	Patients (n=159) *n* (%)	Carers (n=478) *n* (%)
Data sharing			
Not important	14 (2.2)	6 (3.8)	8 (1.7)
Slightly important	14 (2.2)	6 (3.8)	8 (1.7)
Moderately important	67 (10.5)	28 (17.6)	39 (8.2)
Important	286 (44.9)	62 (39.0)	224 (46.9)
Very important	256 (40.2)	57 (35.8)	199 (41.6)
Data access			
Not important	13 (2.0)	7 (4.4)	6 (1.3)
Slightly important	15 (2.4)	7 (4.4)	8 (1.7)
Moderately important	54 (8.5)	26 (16.4)	28 (5.9)
Important	265 (41.6)	55 (34.6)	210 (43.9)
Very important	290 (45.5)	64 (40.3)	226 (47.3)
Data use			
Not important	7 (1.1)	2 (1.3)	5 (1.0)
Slightly important	17 (2.7)	7 (4.4)	10 (2.1)
Moderately important	71 (11.1)	26 (16.4)	45 (9.4)
Important	271 (42.5)	62 (39.0)	209 (43.7)
Very important	271 (42.5)	62 (39.0)	209 (43.7)
Data reuse			
Not important	13 (2.0)	6 (3.8)	7 (1.5)
Slightly important	19 (3.0)	6 (3.8)	13 (2.7)
Moderately important	96 (15.1)	31 (19.5)	65 (13.6)
Important	255 (40.0)	67 (42.1)	188 (39.3)
Very important	254 (39.9)	49 (30.8)	205 (42.9)

Note: The proportions may not add 100 due to rounding.

Table 3. Factors influencing participants' views about involvement in decision-making regarding health data sharing, access, use and reuse

		Data sharing		Data access		Data use		Data reuse	
	Total	**Important[a] n (%)**	**Other[b] n (%)**	**Important[a] n (%)**	**Other[b] n (%)**	**Important[a] n (%)**	**Other[b] n (%)**	**Important[a] n (%)**	**Other[b] n (%)**
	637	542 (85.1)	95 (14.9)	555 (87.1)	82 (12.9)	542 (85.1)	95 (14.9)	509 (79.9)	128 (20.1)
Type of participant									
Patient	159	119 (74.8)*	40 (25.2)*	119 (74.8)*	40 (25.2)*	124 (78.0)	35 (22.0)	116 (73.0)	43 (27.0)
Carer	478	423 (88.5)*	55 (11.5)*	436 (91.2)*	42 (8.8)*	418 (87.4)	60 (2.6)	393 (82.2)	85 (17.8)
Sex									
Female	453	393 (86.8)	60 (13.2)	405 (89.4)	48 (10.6)	394 (87.0)	59 (13.0)	371 (81.9)	82 (18.1)
Male	184	149 (81.0)	35 (19.0)	150 (81.5)	34 (18.5)	148 (80.4)	36 (19.6)	138 (75.0)	46 (25.0)
Age (years)									
<18	92	67 (72.8)*	25 (27.2)*	66 (71.7)*	26 (28.3)*	69 (75.0)	23 (25.0)	69 (75.0)	23 (25.0)
18-30	103	86 (83.5)*	17 (16.5)*	93 (90.3)*	10 (9.7)*	88 (85.4)	15 (14.6)	85 (82.5)	18 (17.5)
>30	434	384 (88.5)*	50 (11.5)*	389 (89.6)*	45 (10.4)*	379 (87.3)	55 (12.7)	350 (80.6)	84 (19.4)
Educational level (years)									
≤12	476	397 (83.4)	79 (16.6)	404 (84.9)	72 (15.1)	389 (81.7)*	87 (18.3)*	372 (78.2)	104 (21.8)
>12	154	141 (91.6)	13 (8.4)	145 (94.2)	9 (5.8)	148 (96.1)*	6 (3.9)*	133 (86.4)	21 (13.6)
Marital status									
Married/living with partner	384	342 (89.1)	42 (10.9)	348 (90.6)	36 (9.4)	338 (88.0)	46 (12.0)	318 (82.8)	66 (17.2)
Others	247	197 (79.8)	50 (20.2)	202 (81.8)	45 (18.2)	200 (81.0)	47 (19.0)	188 (76.1)	59 (23.9)
Occupation									
Upper white-collar	148	137 (92.6)	11 (7.4)	137 (92.6)	11 (7.4)	138 (93.2)	10 (6.8)	119 (80.4)	29 (19.6)
Lower white-collar	116	99 (85.3)	17 (14.7)	102 (87.9)	14 (12.1)	98 (84.5)	18 (15.5)	91 (78.4)	25 (21.6)
Blue-collar	87	69 (79.3)	18 (20.7)	76 (87.4)	11 (12.6)	70 (80.5)	17 (19.5)	68 (78.2)	19 (21.8)
Other	250	204 (81.6)	46 (18.4)	205 (82.0)	45 (18.0)	205 (82.0)	45 (18.0)	199 (79.6)	51 (20.4)
Perceived income adequacy									
Insufficient/Caution with expenses	315	265 (84.1)	50 (15.9)	281 (89.2)	34 (10.8)	265 (84.1)	50 (15.9)	253 (80.3)	62 (19.7)
Enough to make ends meet/comfortable	299	259 (86.6)	40 (13.4)	253 (84.6)	46 (15.4)	258 (86.3)	41 (13.7)	238 (79.6)	61 (20.4)
Involvement with patients' organisations									
No	598	508 (84.9)	90 (15.1)	522 (87.3)	76 (12.7)	508 (84.9)	90 (15.1)	477 (79.8)	121 (20.2)
Yes	32	31 (96.9)	1 (3.1)	29 (90.6)	3 (9.4)	31 (96.9)	1 (3.1)	29 (90.6)	3 (9.4)
Trust in research host institution									
Very important	461	412 (89.4)*	49 (10.6)*	423 (91.8)*	38 (8.2)*	421 (91.3)*	40 (8.7)*	385 (83.5)*	76 (16.5)*
Other	165	121 (73.3)*	44 (26.7)*	121 (73.3)*	44 (26.7)*	111 (67.3)*	54 (32.7)*	115 (69.7)*	50 (30.3)*
Trust in research team									
Very important	448	401 (89.5)*	47 (10.5)*	410 (91.5)*	38 (8.5)*	405 (90.4)*	43 (9.6)*	369 (82.4)	79 (17.6)
Other	179	132 (73.7)*	47 (26.3)*	135 (75.4)*	44 (24.6)*	128 (71.5)*	51 (28.5)*	131 (73.2)	48 (26.8)
Trust in national institutions, Md (P25-P75)	3.5 (1.8-5.2)	3.8 (1.8-5.4)	2.4 (1.0-3.8)	3.8 (1.8-5.3)	2.6 (1.3-5.0)	3.8 (2.0-5.4)*	2.2 (0.8-4.0)*	3.6 (1.8-5.2)	3.2 (1.6-5.3)
Trust in international institutions, Md (P25-P75)	5.0 (2.5-7.0)	5.0 (2.5-7.0)	3.5 (1.0-5.5)	5.0 (2.5-7.0)	3.0 (1.0-7.0)	5.0 (2.5-7.0)*	3.0 (1.0-5.0)*	5.0 (2.5-7.0)	4.5 (2.0-7.0)
Interpersonal trust, Md (P25-P75)	4.7 (3.0-6.7)	4.7 (3.0-6.7)	3.7 (2.0-6.0)	4.7 (3.0-6.3)	5.0 (2.3-6.7)	4.7 (3.0-6.3)	3.7 (2.0-6.7)	4.7 (3.0-6.3)	**4.8 (3.0-6.7)**

[a] Includes participants who answered "important" and "very important"; [b] includes participants who answered, "not important", "slightly important" and "moderately important"; *p<0.001
Note: In each variable, the total may not add 637 participants, 159 patients or 478 carers due to missing values. The proportions may not add 100 due to rounding.

5 CONCLUSION

This dissertation assessed public views about involvement in all key aspects of individual-level data governance and enabled the identification of differences in the levels of importance attributed by rare diseases patients and their carers to participation in decision-making concerning health data sharing, access, use and reuse. It also examined how trust in science and other institutions influenced their views, and its association with sociodemographic characteristics.

The majority of participants attributed a high degree of importance to involvement in decision-making about health data sharing, access, use, and reuse. Participants who considered trust in research host institutions as very important rated higher the importance of being involved in decisions across the four aspects of data governance assessed. A similar position was expressed by participants who valued trust in research teams for participation in data sharing, data access, and data use. Individuals with a low level of trust in national and international institutions were less likely to value involvement in decisions about data use. Carers and older participants stated more frequently the importance of getting involved in decisions about data sharing and access, while more educated individuals were more likely to value involvement in decision-making about data use.

These findings highlight the need to consider the incorporation of more opportunities for involvement in individual-level data governance for rare disease communities. Such opportunities include dynamic consent mechanisms, which provide a digital interactive platform for more personalized and direct communication between researchers and participants. These mechanisms also enable rare disease patients to manage their consent preferences over time, especially in cases of data reuse. The influence of trust in our participants' views calls for more transparency in research and governance. This can be achieved by providing more transparent information to participants, for instance, regarding the implications of data sharing, access use and reuse, and by establishing accountability measures for the oversight of data breaches and mismanagement.

Further qualitative research is needed to understand the motivations, concerns, and expectations regarding involvement in individual-level data governance of people affected by rare diseases. It is also necessary to assess their preferences regarding different types of consent (broad, specific, dynamic). And finally, it would be important to inquire about their needs regarding involvement in decision-making about health data sharing, access, use and reuse, including whether they require support from their relatives or professionals in making informed decisions.

REFERENCES

Aitken, M., Tully, M. P., Porteous, C., Denegri, S., Cunningham-Burley, S., Banner, N., (…) Willison, D. J. (2019). Consensus Statement on Public Involvement and Engagement with Data Intensive Health Research. *International Journal of Population Data Science*, *4*(1). https://doi.org/10.23889/IJPDS.V4I1.586

Aitken, Mhairi, Cunningham-Burley, Sarah, & Pagliari, Claudia (2016). Moving from trust to trustworthiness: Experiences of public engagement in the Scottish Health Informatics Programme. *Science and Public Policy*, *43*(5). https://doi.org/10.1093/scipol/scv075

Annalijn, Conklin, Zoë Slote, Morris, & Ellen, Nolte. (2010). Involving the public in healthcare policy. An update of the research evidence and proposed evaluation framework. *Santa Monica, CA: RAND Corporation,* Retrieved from https://www.rand.org/content/dam/rand/pubs/technical_reports/2010/RAND_TR850.pd

Asha, Saxena (2019). The Role of Big Data Analytics and AI in the Future of Healthcare. Retrieved from https://www.dataversity.net/the-role-of-big-data-analytics-and-ai-in-the-future-of-healthcare/

Beier, Katharina, Schweda, Mark, & Schicktanz, Silke (2019). Taking patient involvement seriously: A critical ethical analysis of participatory approaches in data-intensive medical research. *BMC medical informatics and decision making*, *19*(1). https://doi.org/10.1186/s12911-019-0799-7

Benson, Mikael (2016). Clinical implications of omics and systems medicine: focus on predictive and individualized treatment. *Journal of Internal Medicine*, *279*(3). https://doi.org/10.1111/joim.12412

Boulanger, Vanessa, Schlemmer, Marissa, Rossov, Suzanne, Seebald, Allison, & Gavin, Pamela (2020). Establishing Patient Registries for Rare Diseases: Rationale and Challenges. *Pharmaceutical Medicine*, *34*(3). https://doi.org/10.1007/s40290-020-00332-1

Brasil, Sandra, Pascoal, Carlota, Francisco, Rita, Ferreira, Vanessa Dos Reis, Videira, Paula A., Videira & Valadão, Gonçalo (2019). Artificial Intelligence (AI) in Rare Diseases: Is the Future Brighter? *Genes*, *10*(12). https://doi.org/10.3390/GENES10120978

Braunack-Mayer, Annette, Fabrianesi, Belinda, Street, Jackie, Carter, Stacy M, Engelen, Lina, Carolan, Lucy, (…) Sproston, Kylie (2021). Sharing Government Health Data with the Private Sector: Community Attitudes Survey. *Journal of Medical Internet Research*, *23*(10). https://doi.org/10.2196/24200

Budin-Ljøsne, Isabelle, Teare, Harriet J.A., Kaye, Jane, Beck, Stephan, Bentzen, Heidi Beate, Caenazzo, Luciana, (…) Mascalzoni, Deborah (2017). Dynamic Consent: A

potential solution to some of the challenges of modern biomedical research. *BMC Medical Ethics*, *18*(1). https://doi.org/10.1186/s12910-016-0162-9

Buyx, Alena, Savio, Lorenzo del, Prainsack, Barbara, & Vo, Henry (2017). Every participant is a PI. Citizen science and participatory governance in population studies. *International Journal of Epidemiology*, *46*(2). https://doi.org/10.1093/ije/dyw204

Courbier, Sandra, Dimond, Rebecca, & Bros-facer, Virginie (2019). Share and protect our health data: an evidence-based approach to rare disease patients' perspectives on data sharing and data protection - quantitative survey and recommendations. *Orphanet Journal of Rare Diseases, 14*(1). https://doi.org/10.1186/s13023-019-1123-4

Cunningham-Burley, Sarah (2006). Public Knowledge and Public Trust. *Public Health Genomics*, *9*(3). https://doi.org/10.1159/000092658

Darquy, Sylviane, Moutel, Grégoire, Lapointe, Anne Sophie, D'Audiffret, Diane, Champagnat, Julie, Guerroui, Samia, (…) Duchange, Nathalie (2015). Patient/family views on data sharing in rare diseases: study in the European LeukoTreat project. *European Journal of Human Genetics, 24*(3). https://doi.org/10.1038/ejhg.2015.115

De Freitas, Cláudia, Amorim, Mariana, Machado, Helena, Leão Teles, Elisa, Baptista, Maria João, Renedo, Alicia, (…) Silva, Susana (2021). Public and patient involvement in health data governance (DATAGov): protocol of a people-centred, mixed-methods study on data use and sharing for rare diseases care and research. *BMJ Open*, *11*(3). https://doi.org/10.1136/BMJOPEN-2020-044289

De Freitas, Cláudia, dos Reis, Vanessa, Silva, Susana, Videira, Paula A., Morava, Eva, & Jaeken, Jaak (2017). Public and patient involvement in needs assessment and social innovation: A people-centred approach to care and research for congenital disorders of glycosylation. *BMC Health Services Research*, *17*(1). https://doi.org/10.1186/S12913-017-2625-1/TABLES/3

Decherchi, Sergio, Pedrini, Elena, Mordenti, Marina, Cavalli, Andrea, & Sangiorgi, Luca (2021). Opportunities and Challenges for Machine Learning in Rare Diseases. *Frontiers in Medicine*, *8*. https://doi.org/10.3389/FMED.2021.747612/BIBTEX

European Commission (2019). *Support for the setting-up of registries of patients affected by rare diseases available for all the ERNs*. Retrieved from https://ec.europa.eu/newsroom/sante/items/654655/default

European Organization for Rare Diseases; EURORDIS (2012*). EURORDIS-NORD-CORD Joint Declaration of 10 Key Principles for Rare Disease Patient Registries*. Retrieved from http://www.effectivehealthcare.ahrq.gov/ehc/products/74/531/Registries%202nd%20ed%20final%20to%20Eisenberg%209-15-

Flores, Mauricio, Glusman, Gustavo, Brogaard, Kristin, Price, Nathan D., & Hood, Leroy (2013). P4 medicine: how systems medicine will transform the healthcare sector and society. *Personalized Medicine, 10*(6). https://doi.org/10.2217/PME.13.57

Gil, Press (2020). 6 Predictions About Data In 2020 And the Coming Decade. *Forbes*. Retrieved from https://www.forbes.com/sites/gilpress/2020/01/06/6-predictions-about-data-in-2020-and-the-coming-decade/?sh=4b5f3aea4fc3

Health Information and Quality Authority; HIQA (2021*). Findings from the National Public Engagement on Health Information*. Retrieved from https://www.hiqa.ie/sites/default/files/2021-09/Findings-from-the-National-Public-Engagement-on-Health-Information.pdf

Instituto Nacional Estatística (2010). *Classificação Portuguesa das Profissões. Lisbon: INE*. Retrieved from https://www.ine.pt/xurl/pub/107961853. ISBN 978-989-25-0010-2

Jo, Suyeon, & Nabatchi, Tina (2020). Different Processes, Different Outcomes? Assessing the Individual-Level Impacts of Public Participation. *Public Administration Review*, *81*(1) https://doi.org/10.1111/puar.13272

Jones, Linda A., Nelder, Jenny R., Fryer, Joseph M., Alsop, Philip H., Geary, Michael R., Prince, Mark, & Cardinal, Rudolf N. (2021). Public opinion on sharing data from UK health services for clinical and research purposes without explicit consent. *MedRxiv*, https://doi.org/10.1101/2021.07.19.21260635

Kariotis, Timothy, Ball, Mad Price, Greshake Tzovaras, Bastian, Dennis, Simon, Sahama, Tony, Johnston, Carolyn, (…) Borda, Ann (2020). Emerging health data platforms: From individual control to collective data governance. *Data & Policy*, *2*. https://doi.org/10.1017/dap.2020.14

Kaye, Jane, Terry, Sharon F., Juengst, Eric, Coy, Sarah, Harris, Jennifer R., Chalmers, Don, (…) Cambon-Thomsen, Anne (2018). Including all voices in international data sharing governance. *Human Genomics*, *12*(1). https://doi.org/10.1186/s40246-018-0143-9

Kaye, Jane, Whitley, Edgar A., Lund, David, Morrison, Michael, Teare, Harriet, & Melham, Karen (2015). Dynamic consent: A patient interface for twenty-first century research networks. *European Journal of Human Genetics*, *23*(2). https://doi.org/10.1038/ejhg.2014.71

Kerasidou, Angeliki (2016). Trust me, I'm a researcher!: The role of trust in biomedical research. *Medicine, Health Care and Philosophy*, *20*. https://doi.org/10.1007/s11019-016-9721-6

Kleiderman, Erika, Knoppers, Bartha Maria, Fernandez, Conrad v., Boycott, Kym M., Ouellette, Gail, Wong-Rieger, Durhane, (…) Avard, Denise (2014). Returning incidental findings from genetic research to children: views of parents of children affected by rare diseases. *Journal of Medical Ethics*, *40*(10). https://doi.org/10.1136/MEDETHICS-2013-101648

Knoppers, Bartha Maria, & Thorogood, Adrian Mark (2017). Ethics and Big Data in health. *Current Opinion in Systems Biology*, *4*. https://doi.org/10.1016/j.coisb.2017.07.001

Klynveld Peat Marwick Goerdeler; KPMG report (2018). *Data governance: Driving value in healthcare*. Retrieved from https://home.kpmg/content/dam/kpmg/xx/pdf/2018/06/data-governance-driving-value-in-health.pdf

Ludman, Evette J., Fullerton, Stephanie M., Spangler, Leslie, Trinidad, Susan Brown, Fujii, Monica M., Jarvik, Gail P., … Burke, Wylie (2010). Glad you asked: Participants' opinions of re-consent for DBGAP data submission. *Journal of Empirical Research on Human Research Ethics*, *5*(3). https://doi.org/10.1525/jer.2010.5.3.9

MacFarlane, Anne, Ogoro, Mamobo, De Freitas, Claudia, Niranjan, Vikram, Severoni, Santino, & Waagensen, Elisabeth (2021). Migrants' involvement in health policy, service development and research in the WHO European Region: A narrative review of policy and practice. *Tropical Medicine & International Health*, *26*(10). https://doi.org/10.1111/TMI.13643

Machado, Helena; Silva, Susana. (2015) "Public participation in genetic databases: Crossing the boundaries between biobanks and forensic DNA databases through the principle of solidarity". Journal of Medical Ethics, 41(10): 820-824. http://dx.doi.org/10.1136/medethics-2014-102126

McCormack, Pauline, Kole, Anna, Gainotti, Sabina, Mascalzoni, Deborah, Molster, Caron, Lochmüller, Hanns, & Woods, Simon (2016). "You should at least ask". the expectations, hopes and fears of rare disease patients on large-scale data and biomaterial sharing for genomics research. *European Journal of Human Genetics*, *24*(10). https://doi.org/10.1038/ejhg.2016.30

Murtagh, Madeleine J., Blell, Mwenza T., Butters, Olly W., Cowley, Lorraine, Dove, Edward S., Goodman, Alissa, … Burton, Paul R. (2018). Better governance, better access: Practising responsible data sharing in the METADAC governance infrastructure. *Human Genomics*, *12*(1). https://doi.org/10.1186/S40246-018-0154-6/TABLES/2

Oderkirk, Jillian, & Ronchi, Elettra (2018). Governing data for better health and healthcare. *OECD Observer*. https://doi.org/10.1787/6b33a920-en

Organization for Economic Co-operation and Development; OECD (2015). *Health Data Governance: Privacy, Monitoring and Research-Policy Brief*. Retrieved from https://www.oecd.org/health/health-systems/Health-Data-Governance-Policy-Brief.pdf

Organization for Economic Co-operation and Development; OECD (2016). *Recommendation of the Council on Health Data Governance, OECD/LEGAL/0433*. Retrieved from https://www.oecd.org/health/health-systems/Recommendation-of-OECD-Council-on-Health-Data-Governance-Booklet.pdf

Powles, Julia, & Hodson, Hal (2017). Google DeepMind and healthcare in an age of algorithms. *Health and Technology, 7*(4). https://doi.org/10.1007/S12553-017-0179-1

Public Health Data Agency (2021). *Pan-Canadian Health Data Strategy: Building Canada's Health Data Foundation*. Retrieved from https://www.canada.ca/content/dam/phac-aspc/documents/corporate/mandate/about-agency/external-advisory-bodies/list/pan-canadian-

health-data-strategy-reports-summaries/expert-advisory-group-report-02-building-canada-health-data-foundation/expert-advisory-group-report-02-building-canada-health-data-foundation.pdf

Resnik, David. B. (2011). Scientific research and the public trust. *Science and engineering ethics*, *17*(3). https://doi.org/10.1007/s11948-010-9210-x

Riordan, Fiona, Papoutsi, Chrysanthi, Reed, Julie E., Marston, Cicely, Bell, Derek, & Majeed, Azeem (2015). Patient and public attitudes towards informed consent models and levels of awareness of Electronic Health Records in the UK. *International Journal of Medical Informatics*, *84*(4). https://doi.org/10.1016/J.IJMEDINF.2015.01.008

Shah, Nisha, Coathup, Victoria, Teare, Harriet, Forgie, Ian, Giordano, Giuseppe Nicola, Hansen, Tue Haldor, (…) Kaye, Jane (2018). Sharing data for future research — engaging participants' views about data governance beyond the original project: a DIRECT Study. *Genetics in Medicine*, *0*(0. https://doi.org/10.1038/s41436-018-0299-7

Shah, Nisha, Coathup, Victoria, Teare, Harriet, Forgie, Ian, Nicola, Giuseppe, Tue, Giordano, (…) Kaye, Jane (2019). Motivations for data sharing — views of research participants from four European countries: A DIRECT study. *European Journal of Human Genetics*, *27*(5). https://doi.org/10.1038/s41431-019-0344-2

Serviços Partilhados do Ministério da Saúde; SPMS (2019). *From big data to smart health: putting data to work for the public´s health. Data Strategy for Next Generation Portuguese National Health Service*. Retrieved from https://www.spms.min-saude.pt/wp-content/uploads/2020/07/Data-Strategy_VERSAOFINAL_07.01.2020.pdf

Sterckx, Sigrid, Rakic, Vojin, Cockbain, Julian, & Borry, Pascal (2016). "You hoped we would sleepwalk into accepting the collection of our data": controversies surrounding the UK care.data scheme and their wider relevance for biomedical research. *Medicine, Health Care and Philosophy, 19*(2). https://doi.org/10.1007/S11019-015-9661-6

Supraha Goreta, Sandra, Dabelic, Sanja, & Dumic, Jerka (2012). Insights into complexity of congenital disorders of glycosylation. *Biochemia Medica*, *22*(2). https://hrcak.srce.hr/83220

Thorogood, Adrian (2020). International Data Sharing and Rare Disease: The Importance of Ethics and Patient Involvement. In *Rare Diseases*, 221. https://doi.org/10.5772/intechopen.91237

Trinidad, Susan Brown, Fullerton, Stephanie M., Bares, Julie M., Jarvik, Gail P., Larson, Eric B., & Burke, Wylie (2010). Genomic research and wide data sharing: Views of prospective participants. *Genetics in Medicine, 12*(8). https://doi.org/10.1097/gim.0b013e3181e38f9e

Understanding patient data (2022). *Learning data governance: a new approach to decisions about data*. Retrieved from https://understandingpatientdata.org.uk/learning-data-governance-new-approach-decisions-about-data

Van Staa, Tjeerd Pieter, Goldacre, Ben, Buchan, Iain, & Smeeth, Liam (2016). Big health data: the need to earn public trust. *BMJ*, *354*. https://doi.org/10.1136/BMJ.I3636

Williams, Gemma A, & Fahy, Nick (2019). Building and maintaining public trust to support the secondary use of personal health data. *Eurohealth, 25*(2).

Young, Andrea, Menon, Devidas, Street, Jackie, Al-Hertani, Walla, & Stafinski, Tania (2018). Engagement of Canadian Patients with Rare Diseases and Their Families in the Lifecycle of Therapy: A Qualitative Study. *Patient-Patient centred outcome research*, *11*(3). https://doi.org/10.1007/s40271-017-0293-1Zmerli, Sonja, & Newton, Ken (2008). Social Trust and Attitudes Toward Democracy. *Public Opinion Quarterly*, *72*(4). https://doi.org/10.1093/POQ/NFN054

ANNEXES

ANNEX 1 – INFORMATION LEAFLET FOR PARTICIPANTS

Informação sobre o estudo

Estamos a desenvolver um estudo sobre as opiniões de doentes, cuidadores informais, profissionais de saúde, gestores, técnicos e pessoal administrativo quanto aos benefícios e riscos associados à partilha de informação de saúde e à participação dos cidadãos na sua governação, no âmbito das doenças raras.

Gostaríamos de contar com a sua participação.

Antes de decidir, é importante que saiba mais acerca deste estudo e do que lhe é pedido se aceitar participar.

Por favor, leia atentamente este folheto informativo e coloque todas as perguntas que achar necessário.

Por que queremos falar consigo?

A finalidade deste estudo é perceber de que forma as necessidades, preferências e valores das pessoas com doenças raras, cuidadores informais, profissionais de saúde, gestores, técnicos e pessoal administrativo podem ser incluídas na governação de informação de saúde.

Quando falamos de informação de saúde estamos a referir-nos a todo o tipo de informação direta ou indiretamente ligada à saúde de uma pessoa e à sua história clínica e familiar (por exemplo, registos clínicos, resultados de análises e outros exames complementares, intervenções e diagnósticos).

Serão convidadas a participar pessoas com mais de 12 anos de idade seguidas nos centros de referência para as Doenças Hereditárias do Metabolismo e de Cardiopatias Congénitas do Centro Hospitalar Universitário de São João (CHUSJ), e seus cuidadores informais, assim como profissionais de saúde, gestores, técnicos e pessoal administrativo da equipa alargada.

Em que consiste a sua participação?

Gostaríamos que respondesse a um questionário num local reservado nas instalações do CHUSJ, com uma duração prevista de 20 minutos. Estamos disponíveis para esclarecer todas as dúvidas e questões que possam surgir.

Quais serão os benefícios da sua participação?

Será participante de um estudo inovador que procura compreender os benefícios e os riscos associados ao envolvimento dos cidadãos na governação de informação de saúde, contribuindo para:
- Identificar os desafios inerentes à partilha de informação no âmbito das doenças raras e seus benefícios;
- Incentivar o debate em torno das respostas aos desafios que enfrenta a proteção de informação de saúde;
- Adaptar a governação de informação de saúde às necessidades, preferências e valores de todas as pessoas envolvidas.

A informação é confidencial?

Sim, nos termos exigidos pela lei. Toda a informação que partilhar connosco será vista somente pelos membros da equipa de investigação.
A informação será armazenada de forma segura. Sempre que as informações recolhidas forem utilizadas, nunca será usado o seu verdadeiro nome.

Sou obrigado/a a participar?

Não. Caso decida não participar, esta decisão não terá quaisquer desvantagens nem influenciará os cuidados de saúde. Mesmo depois de aceitar, poderá desistir em qualquer altura e sem justificação.

Quem coordena o estudo?

A coordenação é da responsabilidade do Instituto de Saúde Pública da Universidade do Porto (ISPUP), sendo a investigadora principal Cláudia de Freitas.

Este estudo é financiado pela **Fundação para a Ciência e a Tecnologia**.

Como será usada a informação?

Os resultados deste estudo serão divulgados de diversas formas (relatórios, artigos científicos e comunicações orais), junto da comunidade científica e de pessoas que podem tomar decisões em relação às políticas que regulam a proteção de dados de saúde.

Obrigado por ter lido este folheto!

A sua participação será muito valiosa.

A aplicação do questionário apenas prosseguirá depois de colocadas todas as questões pelo participante e após assinatura do consentimento informado.

Ser-lhe-á dado este folheto informativo e uma cópia do consentimento informado.

Para qualquer dúvida, sugestão ou comentário, por favor entre em contacto connosco:

Investigadoras:

Cláudia de Freitas | Susana Silva

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FOLHETO DE INFORMAÇÃO AO PARTICIPANTE

Participação dos cidadãos na governação de informação de saúde: partilha e proteção de dados em doenças raras

ANNEX 2 - QUESTIONNAIRE

ID ☐☐☐☐☐

QUESTIONÁRIO

Governação de informação de saúde

Este estudo pretende conhecer as opiniões de doentes, cuidadores informais e profissionais ligados à saúde quanto à participação dos cidadãos na governação de informação de saúde, no âmbito de doenças raras. A governação de informação de saúde refere-se às políticas, procedimentos e práticas relacionadas com a recolha, proteção, partilha e uso de informações de saúde.

Não há respostas certas nem erradas. Interessa-nos conhecer a sua opinião sincera. Agradecemos a sua colaboração e o tempo que irá disponibilizar para responder a este questionário.

Data de preenchimento: |__|__|/|__|__|/|__|__|__|__|
(dia) (mês) (ano)

1. **É portador de alguma doença rara?**
   - Não ☐0
   - Sim ☐1 Qual? _____

2. **É cuidador informal de uma pessoa com uma doença rara?**
   - Não ☐0
   - Sim ☐1 Qual a doença da pessoa de quem cuida? _____

3. **Neste momento, está a preencher este questionário na qualidade de:** (por favor assinale apenas uma opção de resposta)
   - Doente ☐1
   - Cuidador Informal ☐2
   - Profissional de saúde ☐3 Categoria profissional: _____
   - Outra ☐4 Qual? _____

4. O Regulamento Geral sobre a Proteção de Dados (RGPD) da União Europeia entrou em vigor a 25 de Maio de 2018. **Como se sente em relação à informação que tem acerca desse Regulamento?**
   - **-** Nada informado ☐0 (por favor passe para a pergunta 6)
   - Pouco informado ☐1
   - Mais ou menos informado ☐2
   - Informado ☐3
   - Muito informado ☐4

5. **Indique, por favor, quais foram as <u>duas principais fontes</u> onde obteve informação acerca do Regulamento Geral sobre a Proteção de Dados?**
   - Televisão ☐1
   - Jornais ☐2
   - Rádio ☐3
   - Internet ☐4
   - Profissionais de saúde ☐5
   - Associação de doentes ☐6
   - Familiares, amigos ou colegas ☐7
   - Outra
     Qual?_____ ☐8

Predisposição para partilhar informação, com quem e com que finalidades

6. Há diversas informações de saúde que podem ser usadas em investigação científica. **Em que medida estaria disponível para <u>partilhar</u> as seguintes informações para fins de investigação:**

	Não disponível				Sempre disponível
- Dados pessoais (ex. nível de escolaridade, idade)	☐0	☐1	☐2	☐3	☐4
- História clínica individual	☐0	☐1	☐2	☐3	☐4
- História clínica familiar	☐0	☐1	☐2	☐3	☐4
- Amostras biológicas (ex. sangue, urina)	☐0	☐1	☐2	☐3	☐4
- Informação genética (ex. ADN)	☐0	☐1	☐2	☐3	☐4

   6.1. É importante para nós compreender melhor a sua resposta. **Explique-nos, por favor, a sua posição.**

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7. **Para si, em que medida é importante poder decidir sobre a <u>partilha</u> das suas informações de saúde para fins de investigação?**
   - **-** Nada importante ☐0
   - Pouco importante ☐1
   - Mais ou menos importante ☐2
   - Importante ☐3
   - Muito importante ☐4

8. A investigação científica pode ser realizada por diversas instituições. **Em que medida estaria disponível para partilhar as suas informações de saúde com as seguintes <u>instituições</u>:**

	Não disponível				Sempre disponível
- Instituições de saúde onde recebo cuidados	☐0	☐1	☐2	☐3	☐4
- Universidades portuguesas	☐0	☐1	☐2	☐3	☐4
- Universidades fora de Portugal	☐0	☐1	☐2	☐3	☐4
- Empresas comerciais portuguesas (ex. farmacêuticas, seguradoras)	☐0	☐1	☐2	☐3	☐4
- Empresas comerciais fora de Portugal (ex. farmacêuticas)	☐0	☐1	☐2	☐3	☐4
- Associações de doentes portuguesas	☐0	☐1	☐2	☐3	☐4
- Associações de doentes internacionais	☐0	☐1	☐2	☐3	☐4

9. **Para si, em que medida é importante poder decidir <u>com quem</u> as suas informações de saúde são partilhadas?**
   - **-** Nada importante ☐0
   - Pouco importante ☐1
   - Mais ou menos importante ☐2
   - Importante ☐3
   - Muito importante ☐4

10. **Em que medida estaria disponível para partilhar as suas informações de saúde para as seguintes <u>finalidades</u>?**

	Não disponível				Sempre disponível
- Investigação sem fins lucrativos	☐0	☐1	☐2	☐3	☐4
- Investigação com fins lucrativos	☐0	☐1	☐2	☐3	☐4
- Bases de dados nacionais	☐0	☐1	☐2	☐3	☐4
- Bases de dados internacionais	☐0	☐1	☐2	☐3	☐4
- Uso de companhias de seguros	☐0	☐1	☐2	☐3	☐4
- Uso de empresas farmacêuticas	☐0	☐1	☐2	☐3	☐4

11. **Para si, em que medida é importante poder decidir as <u>finalidades</u> para as quais as suas informações de saúde são partilhadas?**
   - **-** Nada importante ☐0
   - Pouco importante ☐1
   - Mais ou menos importante ☐2
   - Importante ☐3
   - Muito importante ☐4

12. Suponha que autorizou o uso das suas informações de saúde em projetos de investigação. **Se as suas informações forem utilizadas para esse fim, gostaria de <u>ser notificado/a</u> sobre isso?**
   - Sim, sempre ☐1
   - Sim, uma vez por mês ☐2
   - Sim, uma vez por ano ☐3
   - Não ☐4

13. Existem alguns aspetos que as pessoas podem considerar importantes para decidir se vão partilhar as suas informações de saúde para investigação. **Se tivesse que tomar essa decisão, qual o grau de importância que atribuiria aos seguintes <u>aspetos</u>:**

	Nada importante				Muito importante
- Conhecer os objetivos da investigação	☐0	☐1	☐2	☐3	☐4
- Garantir a privacidade das minhas informações	☐0	☐1	☐2	☐3	☐4
- Garantir a segurança das minhas informações	☐0	☐1	☐2	☐3	☐4
- Confiar na organização que acolhe a investigação	☐0	☐1	☐2	☐3	☐4
- Confiar na equipa que vai realizar a investigação	☐0	☐1	☐2	☐3	☐4
- Poder retirar o consentimento a qualquer momento	☐0	☐1	☐2	☐3	☐4
- Ter acesso aos resultados após a conclusão do estudo	☐0	☐1	☐2	☐3	☐4

14. As informações de saúde recolhidas para tratamento médico ou para fins de investigação podem ser reutilizadas para outras finalidades (por exemplo, avaliação da qualidade e planeamento de serviços ou políticas). **Para si, em que medida é importante poder decidir se as suas informações de saúde podem ser <u>reutilizadas</u> para finalidades diferentes daquela para a qual foram recolhidas inicialmente?**

- Nada importante ☐0
- Pouco importante ☐1
- Mais ou menos importante ☐2
- Importante ☐3
- Muito importante ☐4

Posições sobre consentimento

15. A partilha de informações de saúde para investigação só pode ocorrer mediante consentimento prévio dos seus titulares. **Por favor indique qual a modalidade de <u>consentimento</u> que prefere relativamente à partilha de informação genética e não genética:**

15.1. Um **<u>consentimento geral</u>** (dado uma única vez para qualquer projeto de investigação) ou um **<u>consentimento específico</u>** (dado para cada projeto de investigação específico).

	Consentimento geral	Consentimento específico
- Informação genética (ex. ADN)	☐1 (p.f. passe para a pergunta 16)	☐2
- Informação não genética (ex. dados pessoais)	☐1 (p.f. passe para a pergunta 16)	☐2

15.2. Um consentimento específico para **<u>todas as dimensões</u>** do projeto de investigação ou um consentimento específico para **<u>cada uma das dimensões</u>** do projeto de investigação (por exemplo, uso e proteção de dados).

	Todas as dimensões	Cada uma das dimensões
- Informação genética (ex. ADN)	☐1	☐2
- Informação não genética (ex. dados pessoais)	☐1	☐2

16. **Indique os <u>dois riscos</u> mais importantes que, na sua opinião, podem associar-se à partilha de informação genética para fins de investigação.**

- Falta de segurança e controlo no acesso à informação ☐1
- Restrição dos direitos à privacidade e autonomia dos cidadãos ☐2
- Possibilidade de retirar informação que ultrapasse os objetivos de investigação ☐3
- Realização de estudos genéticos que possam discriminar negativamente os cidadãos ☐4
- Outro. Qual? _____ ☐5

17. **Indique os <u>dois benefícios</u> mais importantes que, na sua opinião, podem associar-se à partilha de informação genética para fins de investigação.**

- Descoberta de uma cura para doenças sem tratamento ☐1
- Desenvolvimento de estratégias para controlar a propagação de doenças ☐2
- Desenvolvimento de novos medicamentos e terapêuticas ☐3
- Desenvolvimento de tratamentos personalizados, tendo em conta as características de cada doente ☐4
- Outro. Qual? _____ ☐5

18. **Se precisasse de tomar uma <u>decisão</u> quanto à partilha das suas informações de saúde para fins de investigação, gostaria de:** (p.f. assinale <u>apenas uma</u> opção de resposta)

- Decidir sozinho/a ☐1 (p.f. passe para a pergunta 19)
- Decidir com o apoio de outra/s pessoa/s ☐2
- Delegar a decisão noutra/s pessoa/s ☐3

18.1. **Que pessoa/s?**

- Em familiares ou amigos ☐1
- Num/a profissional especificamente formado/a para prestar aconselhamento sobre informações de saúde ☐2
- Num/a profissional de saúde não especificamente formado/a para prestar aconselhamento sobre informações de saúde ☐3
- Outro. Qual? _____ ☐4

Opiniões sobre participação

Existem diversas formas pelas quais os cidadãos podem participar na governação de informação de saúde, por exemplo, através da presença em reuniões nas quais se planeiam procedimentos para a recolha, proteção, partilha e uso de informações de saúde, em colaboração com responsáveis pela gestão de informação e outros cidadãos.

19. **Se tivesse todas as condições para participar na governação de informação de saúde no âmbito de um hospital público, estaria disponível para <u>participar</u> em**:

	Sim	Talvez	Não
- Reuniões periódicas (ex. a cada 3 ou 4 meses), dando opiniões	☐1	☐2	☐3
- Reuniões periódicas (ex. a cada 3 ou 4 meses), colaborando na tomada de decisões	☐1	☐2	☐3
- Reuniões esporádicas, dando opiniões	☐1	☐2	☐3
- Reuniões esporádicas, colaborando na tomada de decisões	☐1	☐2	☐3

20. Na sua opinião, que **<u>vantagens</u>** podem resultar da **participação dos cidadãos na governação de informação de saúde num hospital público**?

_____

_____

_____

21. Na sua opinião, que **<u>desvantagens</u>** podem resultar da **participação dos cidadãos na governação de informação de saúde num hospital público**?

_____

_____

_____

22. Supondo que deu autorização para o uso das suas informações de saúde num projeto de investigação, **indique, por favor, em que medida seria importante para si <u>participar</u>…**

	Nada importante				Muito importante
- Na definição dos objetivos	☐0	☐1	☐2	☐3	☐4
- No recrutamento de participantes	☐0	☐1	☐2	☐3	☐4
- Na recolha de dados	☐0	☐1	☐2	☐3	☐4
- Na discussão dos resultados	☐0	☐1	☐2	☐3	☐4
- Na redação do relatório do projeto	☐0	☐1	☐2	☐3	☐4
- Na disseminação dos resultados	☐0	☐1	☐2	☐3	☐4

23. A recolha de informação de saúde pode ser efetuada pela própria pessoa através, por exemplo, de uma aplicação no seu telemóvel, tablet ou computador. **Estaria disponível para <u>participar na recolha</u> da sua própria informação de saúde?**

- Não ☐1 (p.f. passe para a questão 24)
- Talvez ☐2
- Sim ☐3

23.1. **Em que medida estaria disponível para <u>partilhar</u> a informação de saúde que recolheria para as seguintes finalidades**:

	Não disponível				Sempre disponível
- Contribuir para o diagnóstico e tratamento da sua doença, tendo em conta as suas características e necessidades pessoais	☐0	☐1	☐2	☐3	☐4
- Realizar investigação sem fins lucrativos relacionada:					
1) com a sua doença	☐0	☐1	☐2	☐3	☐4
2) com outras doenças	☐0	☐1	☐2	☐3	☐4
- Realizar investigação com fins lucrativos relacionada:					
1) com a sua doença	☐0	☐1	☐2	☐3	☐4
2) com outras doenças	☐0	☐1	☐2	☐3	☐4

24. **Qual a confiança pessoal que tem em cada uma das instituições abaixo indicadas?**

	Nenhuma confiança										Toda a confiança	Não responde	Não sabe
- Assembleia da República	☐0	☐1	☐2	☐3	☐4	☐5	☐6	☐7	☐8	☐9	☐10	☐	☐
- Sistema jurídico	☐0	☐1	☐2	☐3	☐4	☐5	☐6	☐7	☐8	☐9	☐10	☐	☐
- Polícia	☐0	☐1	☐2	☐3	☐4	☐5	☐6	☐7	☐8	☐9	☐10	☐	☐
- Políticos	☐0	☐1	☐2	☐3	☐4	☐5	☐6	☐7	☐8	☐9	☐10	☐	☐
- Partidos políticos	☐0	☐1	☐2	☐3	☐4	☐5	☐6	☐7	☐8	☐9	☐10	☐	☐
- Parlamento Europeu	☐0	☐1	☐2	☐3	☐4	☐5	☐6	☐7	☐8	☐9	☐10	☐	☐
- Nações Unidas	☐0	☐1	☐2	☐3	☐4	☐5	☐6	☐7	☐8	☐9	☐10	☐	☐

25. **De uma forma geral, acha que todo o cuidado é pouco quando se lida com as pessoas ou acha que se pode confiar na maioria das pessoas?**

Todo o cuidado é pouco										A maioria das pessoas é de confiança	Não responde	Não sabe
☐0	☐1	☐2	☐3	☐4	☐5	☐6	☐7	☐8	☐9	☐10	☐	☐

26. **Acha que a maior parte das pessoas tenta aproveitar-se de si sempre que pode, ou pensa que a maior parte das pessoas é honesta?**

Tenta aproveitar-se de mim										É honesta	Não responde	Não sabe
☐0	☐1	☐2	☐3	☐4	☐5	☐6	☐7	☐8	☐9	☐10	☐	☐

27. **Acha que, na maior parte das vezes, as pessoas estão preocupadas com elas próprias ou tentam ajudar outros?**

As pessoas estão preocupadas com elas próprias										As pessoas tentam ajudar os outros	Não responde	Não sabe
☐0	☐1	☐2	☐3	☐4	☐5	☐6	☐7	☐8	☐9	☐10	☐	☐

28. **Sexo:**

- Feminino ☐0
- Masculino ☐1

29. **Ano de nascimento:** |__|__|__|__|

30. **De onde é natural?**

- Portugal ☐1
- Outro país ☐2

  Qual?

_____

31. **Onde reside atualmente?** Distrito:

_____

32. **Qual é o seu estatuto marital?**

- Solteiro/a	☐1	- Viúvo/a	☐4
- Casado/a	☐2	- Divorciado/a	☐5
- União de facto	☐3	- Separado/a (casado/a mas não vive com o cônjuge)	☐6

33. **Qual o grau de escolaridade mais elevado que completou?**

- Nenhum, mas sabe ler e/ou escrever	☐1	- Bacharelato	☐6
- 1º Ciclo do ensino básico (4º ano)	☐2	- Licenciatura	☐7
- 2º Ciclo do ensino básico (6º ano)	☐3	- Mestrado/Mestrado Integrado	☐8
- 3º Ciclo do ensino básico (9º ano)	☐4	- Doutoramento	☐9
- Ensino secundário (12º ano)	☐5		

34.**Neste momento, qual é a sua principal situação profissional?** (p.f. assinale <u>apenas uma</u> opção de resposta)

- Empregado/a a tempo inteiro	☐1	- Estudante/na escola/em formação profissional	☐5
- Empregado/a a tempo parcial	☐2	- Doméstico/a/ocupa-se das tarefas do lar	☐6
- Desempregado/a	☐3	- Outra. Qual? _____	☐7
- Reformado/a ou pré-reformado/a	☐4		

35. **Qual é a sua profissão atual?** (se na questão anterior assinalou desempregado/a, doméstico/a, reformado/a ou outra, p.f. considere a sua última profissão)

_____

_____

36. **Considera que os rendimentos do seu agregado familiar são:**

- Insuficientes	☐1
- Tem de ter cuidado com os gastos	☐2
- Chega para as suas necessidades	☐3
- Confortáveis	☐4

37. **Está envolvido em alguma associação de doentes?**

- Não	☐0 (p.f. passe para a pergunta 38)
- Sim, estou envolvido…	
Numa associação local ou nacional	☐1
Em várias associações locais ou nacionais	☐2
Numa associação internacional	☐3
Em várias associações internacionais	☐4

37.1. **Há quanto tempo iniciou o seu envolvimento numa associação de doentes?**

- Menos de 1 ano ☐1
- Entre 1 a 5 anos ☐2
- Mais de 5 anos ☐3

38. **Até que ponto está satisfeito/a com a sua saúde?**

- Muito insatisfeito ☐0
- Insatisfeito ☐1
- Nem satisfeito nem insatisfeito ☐2
- Satisfeito ☐3
- Muito satisfeito ☐4

Agradecemos a sua colaboração!