EURORDIS-RARE DISEASES EUROPE POSITION ON THE EUROPEAN HEALTH DATA SPACE





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EURORDIS-Rare Diseases Europe is a unique, non-profit alliance of over 1000 rare disease patient organisations from 74 countries that work together to improve the lives of over 300 million people living with a rare disease globally.

By connecting patients, families and patient groups, as well as by bringing together all stakeholders and mobilising the rare disease community, EURORDIS strengthens the patient voice and shapes research, policies and patient services.

EXECUTIVE SUMMARY

Sharing health data to advance scientific research and improve clinical practice is of particular importance to the 30 million people living with a rare disease in Europe, where knowledge and expertise are limited, patient populations are geographically dispersed, and health data is scattered.

95% of people living with a rare disease are willing to share their health data. This a community where data equates to hope: an opportunity to access specialist healthcare in other European countries, the path to an earlier diagnosis or the chance to understand more about their little-understood disease through research.

However, this must be done in a way that protects the rights of this population. There is a clear need to find the right balance between ensuring that patient data is safe and secure and allowing this data to be made available for the development of new therapies and treatments. In particular, we must prevent data from being used to enable discrimination in insurance, loans and work.

The European Health Data Space Regulation, as proposed by the European Commission, is an unprecedented opportunity for Europe to unlock the potential of health data in Europe for advancing health and research, especially in the field of rare diseases, while implementing robust standards to ensure secure, ethical and responsible data sharing.

EURORDIS-Rare Diseases Europe, alongside its 1,000 patient organisation members, is calling on Europe to ensure that the rights and hopes of the rare disease community are embedded in this legislation.

The community calls for the European Health Data Space to:

I.Optimise electronic health records

Millions of rare disease patients do not have electronic health records or easy access to them. As many need to travel to another country to receive adequate treatment and healthcare or for professional and personal reasons, safe and

timely sharing of their health data across countries is key. Harmonisation and interoperability of electronic health records across Europe should be encouraged by introducing mandatory standards for electronic health record (EHR) systems.

II. Ensure the ethical use of secondary health data

Secondary use should enable health providers, research organisations and regulatory bodies to have access to health data for the purposes of research, innovation, policy making, educational activities, patient safety, regulatory activities, or personalised health care. In reality, divergent national rules hamper the development of cross-border data exchange on rare diseases.

Access to cross-border health data should become possible within a trusted governance framework based on clear rules and standards, with guidance from the European Reference Networks. Such a framework would further facilitate the creation of a strong cross-border data management system and corresponding rules for health data exchange.

IIII. Increase digital health literacy

Current levels of digital skills and literacy are low. The rare disease community are often faced with decisions on highly sensitive data categories, such as genetic data and cross-border uses of data through established European Reference Networks. It is therefore essential that the new system has educational programmes for citizens and healthcare professionals to ensure informed choices for patients and citizens.

IV. Encourage patient and public partnership

To understand what people living with a rare disease expect from rare disease research and data sharing, it is important to engage them all along the process while making sure their wishes and needs are embedded within research and healthcare delivery design. The new legislation should reflect patients' needs – both in terms of developing robust standards to ensure secure, ethical and responsible data sharing and allowing health data to be seamlessly shared across borders to benefit every person living with a rare disease in Europe.

INTRODUCTION

On 3 May 2022, the European Commission launched a proposal for a Regulation on the European Health Data Space (EHDS) [1]. Once adopted, the legal act will become a fundamental game changer for the digital transformation of the health sector in the European Union (EU), with far-reaching consequences for the rare disease community. The draft proposal aims to:

- give individuals increased digital access to and control over their personal health data;
- define common mandatory standards for electronic medical record systems to ensure their security and interoperability;
- create a consistent framework for the secondary use of health data for research, innovation, policy making, patient safety and other regulatory activities.

This legislation is of utmost importance for people affected by a rare disease, as it addresses the direct needs of patients, e.g. facilitating their access to health care, as well as some of the other salient issues related to rare diseases, such as the scarcity of available knowledge and expertise.

Given that there are over 30 million people across Europe affected by one or more of the 6,000 rare diseases and that less than 6% of them have an approved treatment, European action in the area of digital health could improve the provision of care, especially in regard to cross-border health care, and drive research and innovation.

Figures show that 95% of people living with a rare disease are ready to share their health data for primary and secondary purposes [2]. The EHDS can, therefore, offer new opportunities to ultimately improve the lives of people with rare diseases through enhanced and safe access to data as well as through

^[1] European Commission (2022), Proposal for the European Health Data Space:

https://health.ec.europa.eu/publications/proposal-regulation-european-health-data-space_en [accessed October 2022] [2] EURORDIS (2019) Share and Protect our Health Data, https://www.eurordis.org/publications/share-and-protect-ourhealth-data [accessed October 2022]

advancing diagnosis, treatment, care and research for rare disease patients. This, however, should take into account patient rights, data protection risks and ethical considerations of big data uses such as discrimination or profiling.

Rare disease patients, regardless of the severity of their disease and their socio-demographic profile are willing to share their data:

97%

97%

better understand the mechanisms and causes of their disease to develop new treatments for their disease 97%

to improve diagnosis of their disease

95%

to receive additional specialist advice on their care to improve research on diseases other than theirs

95%

The goal of the paper is to provide recommendations for the four key areas of the European Health Data Space with high relevance for the rare disease community:

- Primary health data uses (i.e. related to providing healthcare services to individuals)
- Secondary health data uses (e.g. for purposes of research, innovation, policy making, educational activities, patient safety, regulatory activities, or personalised health care)
- Digital health literacy
- Public and patient partnership

EURORDIS would like to thank all those who contributed to this position paper. This work is the result of extensive consultation with people living with a rare disease and those around them, EURORDIS members, and volunteers in May-October 2022.

I. OPTIMISE ELECTRONIC HEALTH RECORDS

Primary use pertains to the processing of personal data related to providing healthcare services to individuals, including through electronic health record (EHR) systems. These systems are an essential tool to support health care, as they facilitate primary health data uses by processing personal electronic health data for the provision of health services to assess, maintain and care for the state of health of a concerned person. As many of those living with a rare disease need to travel to another country to receive adequate treatment and health care or for professional and personal reasons, safe and timely sharing of their health data across countries is key.

EHR systems are also highly relevant for doctors, as they allow doctors to easily share health information with different specialists. This is often the case in the context of cross-border healthcare services for rare disease patients, when doctors must consult specialists from different healthcare units within their own country or abroad. As such, wellestablished EHR systems would lead to better health outcomes for people with rare diseases, making it easier to share



data with different healthcare professionals and in different settings to enable the delivery of healthcare provision.

However, major issues persist. There is still a challenge of divergent systems, languages and practices. In some Member States, e.g. Ireland and Bulgaria, there are no developed EHRs.[3]

[3] European Commission (2021) 'Interoperability of Electronic Health Records in the EU' https://digitalstrategy.ec.europa.eu/en/library/interoperability-electronic-health-records-eu [accessed October 2022] For patients to fully benefit from European electronic health records, we call upon the policy makers to:

• Ensure the implementation and use of ICD-11, ORPHA codes, ICD-O3 in EHR systems

Due to their rarity and complexity, rare diseases are under-represented in healthcare coding systems. This further exacerbates the already existing lack of recognition of their importance and hinders the collection of high-quality data needed for healthcare and research purposes.[4] Consequently, this leads to delayed diagnosis, treatment and care.

The International Classification of Diseases (ICD) has long been the main basis for the comparability of statistics on the causes of mortality and morbidity across places and over time. In 2019, the World Health Assembly adopted a revised version of this Classification, ICD-11, which has been significantly more expressive and comprehensive than historical versions and includes rare diseases, though only to a certain extent.

Rare disease patients are more inclined to share their data than the general population. (Rare Barometer, 2020)

To enhance data collection and its quality for healthcare purposes and beyond, ICD-11 should be mandatory within EHR systems, as well as within other databases collecting data about rare diseases, in conjunction with the use of Orphanet nomenclature (i.e. the ORPHA codes). This is essential to ensure patients' visibility within national health and social systems, building thereby a robust and accurate longitudinal care record on rare diseases. Member States should, therefore, work towards the creation of national rare disease registries, based on the ORPHA codes.

Furthermore, the implementation of ICD-11 should go hand-in-hand with training and other capacity-building activities to make sure that those involved have the necessary knowledge to use the classification.

^[4] Aymé, S. et al. (2015) Rare diseases in ICD11: making rare diseases visible in health information systems through appropriate coding. Orphanet J Rare Dis 10, 35, https://doi.org/10.1186/s13023-015-0251-8 [accessed October 2022]

 Secure EHR systems through the adoption of high data processing, encryption, and storage standards, in line with the GDPR, NIS2 and other relevant laws

Data protection is a fundamental condition to ensure citizens' trust in EHR. It is also vital to guarantee necessary investment and ensure consistent oversight and enforcement of data protection rules. There is a need to allocate resources at both national and European levels for the supervision of data processing within EHRs and ensure the compliance of supervision with national and EU laws.

• Make publicly certified systems the only method of identifying individuals when they use EHR systems and other health information systems

To strengthen and enhance data protection, publicly certified identification systems should become a legally mandatory method for connecting to EHR and other health systems, as opposed to some private initiatives (e.g. Facebook or Gmail login) to enhance both user and system protection. One example of such public identification system could be eID,[5] a set of services provided by the European Commission to enable the mutual recognition of national electronic identification schemes (eID) across borders.

This would allow Europeans living with a rare disease to use their national eIDs when accessing online services from other European countries. Consequently, increased access and data sharing in such a cross-border environment would also result in enhanced research opportunities, thereby building capacity for faster diagnosis and better management of rare conditions.



[5] Regulation (EU) No 910/2014 of the European Parliament and of the Council of 23 July 2014 on electronic identification and trust services for electronic transactions in the internal market

• EHR systems and healthcare providers' data protection policies must be developed in line with the following rights:

The right to immediate access to personal electronic health records & the right to have a copy of personal electronic health records

Providing people with rare diseases with easy access to their health information empowers them to be more in control of decisions regarding their health and well-being [6].

Immediate access to personal health records and easily obtainable copies of information could facilitate condition monitoring, adherence to treatment plans, and progress tracking, in addition to significantly contributing to saving costs and speeding up diagnostic processes. In this case, rare disease patients would not need to repeat unnecessary tests when moving from one healthcare specialist/provider to another, which is of the utmost importance as patients often seek healthcare services from multiple specialists [7].

A copy of medical records should be free of charge for a patient.

The right to data portability

The right to portability enables individuals to move certain personal data provided from one platform to another offering similar services. It is essential to allow people with rare diseases to decide what should happen to the generated data when it is shared.

Moving health data across different services, regions and countries in a safe way should be simple for anyone affected by a rare disease. To date, this right has been largely neglected in health care due to the low interoperability of EHR systems.

^[6] Individuals' Right under HIPAA to Access their Health Information 45 CFR § 164.524 https://www.hhs.gov/hipaa/forprofessionals/privacy/guidance/access/index.html [accessed October 2022]

^[7] Ong, R. Y. C., & Sabapathy, S. (2020). Enhancing patient privacy protection under Hong Kong's Electronic Health Record Sharing System. Common Law World Review, 49(1), 4–30

The right to allow or restrict personal electronic health data sharing with a selected healthcare professional

Individuals should be able to restrict access to certain parts of their records, e.g. when an individual visits a dentist, they should be able to restrict access to information about their abortion. However, such a right should be exercised/granted only in situations when restricting access to information would not put at risk an individual's health and well-being. Emergency situations should be excluded.

People with rare diseases should be informed every time a healthcare professional who is not directly treating them requests access to their health records and should be asked to authorise access to the whole file, parts of it, or no access at all before the information is accessed. An affected person could also have an option to grant access to their data to a certain healthcare professionals' group, e.g. members of an ERN.

The right to data rectification

People living with a rare disease should have the right to request data rectification and correct possible errors, misdiagnoses or other inconsistencies in their medical records or medical prescriptions. This should only be done with the approval of a healthcare professional.

The right to information about who, and when someone, has accessed a patient's personal electronic health record

To minimise the risks of unauthorised access, it should be visible in EHR systems who has accessed an EHR or its parts, and when the EHR was accessed.

The right to be forgotten

The right to be forgotten, also known as the right to erasure, is a right to request that a data controller deletes personal data. Such a right is crucial, for example, for rare cancer survivors, as often individuals with a cancer history find difficulties in seeking loans or insurance packages, or in the search for certain employment. Medical records of a past disease might lead to discrimination against cancer survivors by companies, employers and different services.

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However, based on the General Data Protection Regulation (GDPR), it is not an absolute right, as it can only be exercised under specific circumstances (see GDPR Article 17[8]). In addition, an organisation's right to process someone's data might overrule the individual's right to be forgotten, e.g. the data is being processed for public health purposes, when the data is necessary to perform preventative or occupational medicine or when it represents important public or scientific research interest. Even though the conditions to the application of this right apply, it is important to consider each individual case, and where possible, fulfil the request of an individual.

Right to file a complaint to the National (Health) Data Protection Authority

Individuals should be sufficiently informed about their GDPR-granted right to file a complaint to the National (Health) Data Protection Authority. HCPs and other involved stakeholders should take a proactive role in informing patients about their rights.



Most of the mentioned rights are provided to individuals by the GDPR. However, the existence of these rights does not mean that these rights are freely exercised. To ensure that these rights are respected within the EHDS, it is important to create an infrastructure (e.g. different interoperability between services is needed to ensure the right to data portability) and oversight mechanisms. There is also a need for guidance and programmes to educate all stakeholders on how these rights should be interpreted in the healthcare context.

[8] Regulation (EU) 2016/679 of the European Parliament and of the Council of 27 April 2016 on the protection of natural persons with regard to the processing of personal data and on the free movement of such data

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Patients' stories

"It is difficult to get the diagnosis. Doctors don't know about my disease because it is very rare. We need to share all the information about this disease – how it is manifested, how it progresses, all the experience the patients and doctors have." Rare disease patient

"We are only 6 families in [a country] affected by this disease. If we don't offer our database, I think it's impossible for someone to help us, to know much about us." Rare disease patient

.....

.....

"My son has an ultra-rare genetic syndrome. When he started showing symptoms, I was told I was imagining things to the point in which I became the problem. If these medical opinions had been recorded, with the same doctors being the approving party in my right to data rectification, I would have probably still been looking for a diagnosis for my child. While the existence of EHR will improve a lot of diagnosisrelated aspects, we have to make sure that they do not, unwillingly, prolong the diagnosis odyssey for a very vulnerable population, children living with rare diseases, by silencing the voice of their parents." Mother of a child with a rare disease

II. ENSURE THE ETHICAL USE OF Secondary Health Data

The rarity of rare diseases means information and data about them are scarce, as there are often too few people in a geographic location to gather the necessary knowledge. Poor understanding of the pathophysiology of rare diseases remains a significant issue, and this is not only a challenge when finding treatment for a disease: it also results in under- or misdiagnosis, endangers patient health, and causes downstream issues for clinical trial patient selection. Data is vital to progress knowledge on rare diseases and to address this important public health concern.

However, it is crucial that increased data sharing for research, innovation, policy making, planning and management, patient safety and other regulatory activities does not come at the price of patient rights. With the EHDS, it is likely that health and research data sharing will increase greatly. While increased access to large volumes of sensitive data could significantly improve knowledge on rare diseases, it also makes individuals more vulnerable. The nature of risk is no longer limited to physical or psychological harm, but also informational harm, such as privacy breaches, algorithmic discrimination, and profiling. In the context of genetic information, the risk is not only restricted to the individual, but also to their family and generations to come.

In instances where health data comprises personal data, Member States have quite diverse approaches to legal bases for data processing (based on the GDPR exemptions). In some Member States, data subjects are mandated to provide consent for research or other secondary purposes, while in others it is encouraged to rely on alternative bases for processing (without consent but with other safeguards). While it preserves the uniqueness of national healthcare systems, it is negatively impacting cross-border research and healthcare, as there are a lot of confusion and delays due to a lack of harmonised rules. The EHDS proposal aims to facilitate a more coherent approach to health data secondary uses by establishing bases for processing, safeguards for processing, and governance mechanisms for providing access to health data. While

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it preserves the uniqueness of national healthcare systems, it is negatively impacting cross-border research and healthcare, as there are a lot of confusion and delays due to a lack of harmonised rules. The EHDS proposal aims to facilitate a more coherent approach to health data secondary uses by establishing bases for processing, safeguards for processing, and governance mechanisms for providing access to health data. While establishing consistency and achieving the right balance between research, other needs and an individual's risk, it is important to consider the following:

• Establishing data governance models for individuals to exercise control over their re-purposed data

Data governance refers to the exercise of authority and control over the management of data.[9] Informed consent and medical confidentiality have traditionally been used as mechanisms of data control in the health sector. However, in today's data-intense environment they seem to offer only a limited amount of control over the production, collection, use, and circulation of health data. Consent procedures often fall short of adequately informing data subjects about the terms of use of their data, and in the context of data, patients may experience a substantial lack of control over the flow of their data. Furthermore, traditional mechanisms such as informed consent are of limited value when it comes to the evaluation of big data research due to the quantity of data. For instance, it may not be realistic to obtain informed consent on a retrospective study involving millions of individuals.

Thus, in addition to exploring novel consent models such as dynamic consent or meta consent, there is a need to create participatory health data governance schemes within the EHDS to ensure secure, ethical data access and a safe environment while providing clarity and means to control health data uses to patients. It is important to note that the introduction of new elements for data control does not exclude the use of the 'traditional' methods: different models should be explored depending on the situation, setting and individual preferences.

[9] Rene A. et al. (2019) Data governance: A conceptual framework, structured review, and research agenda, International Journal of Information Management, Volume 49, ISSN 0268-4012 Establishing public engagement (multi-directional communication, consultation, and participation) in health data governance is a necessary ethical requirement. It can also contribute to ensuring the transparency of the EHDS, promoting accountability, and fostering trust.

It is also necessary to formally involve patients in the governance structure to ensure that individual data is used with respect and in the best public interest. Health data access bodies should provide detailed information on the uses of the data, so individuals have all the information required to exercise their rights. It is also essential to adopt provisions allowing re-evaluation of consent given by a parent for a child when the child becomes an adult.

However, health data comes from many sources, takes different formats, is stored in multiple systems falls under diverse access policies, and includes diverse uses and users. What is more, health data concepts are complex and involve technical, medical and legal jargon. Therefore, there may not be one model that fits all when it comes to health data governance and meaningful patient engagement. See Part IV of the paper for the proposed model.

There is no single solution for consent: in certain situations, the best solution will be broad consent. In other situations, dynamic consent may be required, and in some cases, the consent will be overridden by the public interest needs. In certain situations, there might also be a need for deferred consent which is used to recruit patients in emergency research when informed consent cannot be obtained prior to enrolment. The environment should remain flexible yet safe to respect diverse preferences and needs.

What is dynamic consent?

Dynamic consent is a personalised, participant-centred communication interface to enable research participants to be at the centre of the decision-making on what happens with their data. This approach is 'dynamic' as it allows interactions over time, allowing participants to consent to new projects, withdraw their consent or make other choices in real-time as their circumstances change. The dynamic consent model does not restrict participants to the opportunity to give broad consent only but allows them to provide different types of consent depending on the kind of study. These consent preferences travel securely with

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their samples or data so that third parties know the scope of the consent that applies. Dynamic consent goes beyond informed consent and could help individuals to control their data and online presence. For the dynamic consent model to be successfully implemented it should be specified when consent decisions are required when decisions should be reviewed, which data are held and stored, and where and how the consent decisions relate to data. [10]

What is meta consent?

Meta consent denotes the idea that people should be asked how and when they would like to be presented with a request for consent. That is, people should be asked to design how they in the future would like to provide consent to the use of their personal health data and biological material. By expressing a preference for how and when to provide consent, people can be said to provide consent on a meta-level. This is the defining idea in the model of meta-consent.[11]

• European Research Infrastructures and European Reference Networks (ERNs) should offer guidance and advice on the feasibility of the EDHS to enable data uses for secondary purposes

It is crucial to gather further evidence on the state of play to assess how any new obligations and models will be implemented in practice, as well as setting realistic timelines to comply with these new obligations. Through a structured dialogue with the co-legislators, European Research Infrastructures and ERNs could serve to gather additional evidence to assess the feasibility, time and resources needed for data holders across the EU to enable secondary uses and comply with the obligations to respond to single data requests as required by the draft EHDS Regulation and refine these obligations accordingly. In addition, it would be critical to bringing in non-University hospitals that have a less mature IT and data infrastructure than the university hospitals that are members of the ERNs. This would help to establish reasonable transition periods, better estimate

[11] Ploug T, Holm S. (2016) Meta Consent - A Flexible Solution to the Problem of Secondary Use of Health Data. Bioethics. Nov;30(9):721-732. doi: 10.1111/bioe.12286. [accessed October 2022]

^[10] Kaye, J. et al. (2015) Dynamic consent: a patient interface for twenty-first century research networks. Eur J Hum Genet 23, 141–146. https://doi.org/10.1038/ejhg.2014.71 [accessed October 2022]

the human and financial resources needed to comply with the new obligations and inform the future implementing and delegated acts.

• Ensuring strong standards for data anonymisation, pseudonymisation and additional safeguards for both personal and non-personal sensitive data categories

Anonymisation makes a dataset non-identifiable by removing personal identifiers (full anonymisation) or by replacing them with keys that the original data controller can use to re-identify the data (pseudonymisation).[12] Anonymised data is considered non-personal data and falls out of the GDPR's scope, while pseudonymised data is considered personal data and are subject to the GDPR regime.

While both techniques are considered data privacy-enhancing mechanisms, it is worth noting that anonymisation technologies do not always quarantee full privacy, since increasing capabilities in data analytics might render the data re-identifiable. Therefore, the absence of standards for clear strona anonymisation may contribute to creating a false sense of security and ultimately a false sense of control.



In particular, given the fact that technological advancements make it easier to de-identify individuals. Furthermore, mixed datasets including both personal and non-personal data may increase the risk of re-identification and full data anonymisation does not enable individual control over the data used for specific purposes, whereas some of such purposes may not be desirable to an individual due to ethical reasons. This is particularly important in the context of self-learning algorithms, where the chain of responsibility may not be as straightforward due to the constantly changing nature of the algorithm.

[12] AEPD-EDPS (2021) joint paper on 10 misunderstandings related to anonymisation

https://edps.europa.eu/system/files/2021-04/21-04-27_aepd-edps_anonymisation_en_5.pdf [accessed October 2022]

In any case, researchers frequently need to use pseudonymised health data, rather than anonymised data, to respond to certain research questions.

For these reasons, there is a need to put additional safeguards and data control mechanisms to manage both anonymised and pseudonymised health datasets. The following elements might be considered:

- Data stewardship, specifying the roles and responsibilities around data management and accountability for each personal data controller from each patient's route within the national and European health data system/space.
- Data policies and procedures to manage datasets, including enforcing authentication and access rights to data as well as the organisational measures and policies to ensure the quality, accuracy and security of the data and regulatory compliance. Tools to help preserve the autonomy and rights of individuals to control their data.
- Data standards, specifications and rules for the definition, creation, storage, and usage of data.[13]
- Defining the clear role of the Research Ethics Committees (RECs) in the use of health data analytics for research

Alongside traditional methods to analyse health data for research, data analytics technologies are increasingly used by researchers. In light of this evolution, there is a need to redefine and adjust the role of the Research Ethics Committees (RECs). The main function of RECs is to protect research participants by identifying ethical issues posed by research involving human subjects.[14] However, the mandate of RECs is not clear when it comes to assessing the risks and benefits of research projects involving big data and analytics. For instance, it is up to each EU Member State to decide whether research based on anonymised data should seek ethical review.[15] Whereas, traditionally the use of anonymised data is considered to be a lower risk for an

^[13] EURORDIS (2020) No time to lose: Building a data strategy for the European Reference Networks hhttp://l.eurordis.org/no-time-to-lose-ERNs [accessed October 2022]

^[14] Hunter, D. (2007). The Roles of Research Ethics Committees: Implications for Membership. Research Ethics, 3(1), 24–26. https://doi.org/10.1177/174701610700300110 [accessed October 2022]

^[15] Ferretti A. et al.(2021) Ethics review of big data research: What should stay and what should be reformed? BMC Med Ethics.. doi: 10.1186/s12910-021-00616-4. PMID: 33931049; PMCID: PMC8085804. [accessed October 2022]

individual, as a such concept of harm has only been linked to vulnerability in data protection. However, the risks of anonymised/aggregated big data are not only limited to an individual's privacy; such risks also involve group discrimination (e.g. anonymised data reveal health patterns of a certain sub-group) or dignitary harm[16]. In addition, as mentioned previously, anonymised data does not offer a bulletproof guarantee of re-identification. Especially when it comes to rare disease health data, which due to its scarcity may sometimes not even be anonymised. Given the complexities of technological advancements, as well as the emergence of new risks and harms linked with the use of data analytics in health research, there is a need to clearly define the mandate of RECs and develop a common methodology to assess big data projects both in case of personal and non-personal data uses. In addition, there is a need to ensure that RECs have sufficient expertise in the domain of big data research and their expertise is based on the relevant code of ethics at the European and national levels.

• Making rare disease patient data FAIR

Rare disease patient data are typically sensitive, present in multiple registries controlled by different data controllers, and non-interoperable. Making these data Findable, Accessible, Interoperable, and Reusable (FAIR) for humans and machines at source enables federated discovery and analysis across data controllers, processors, and holders. This facilitates accurate diagnosis, optimal clinical management, and personalised treatments.

Four principles of data FAIRification [17]

- Findability: Digital resources should be easy to find for both humans and computers. Extensive machine-actionable metadata is essential for the

^[16] Article 7 of the Charter of Fundamental Rights of the European Union protects the dignity of persons by regulating inappropriate communications that threaten to degrade, humiliate, or mortify them. Dignitary privacy follows a

normative logic designed to prevent harm to personality caused by the violation of civility rules.

 ^[17] Jacobsen A. et al. (2020) FAIR Principles: Interpretations and Implementation Considerations. Data Intelligence 2020;
 2 (1-2): 10-29. doi: https://doi.org/10.1162/dint_r_00024 [accessed October 2022]

 ^[17] Jacobsen A. et al. (2020) FAIR Principles: Interpretations and Implementation Considerations. Data Intelligence 2020;
 2 (1-2): 10-29. doi: https://doi.org/10.1162/dint_r_00024 [accessed October 2022]

automatic discovery of relevant datasets and services.

- Accessibility: Protocols for retrieving digital resources should be made explicit, for both humans and machines, including well-defined mechanisms to obtain authorisation for access to protected data.

- Interoperability: When two or more digital resources are related to the same topic or entity, it should be possible for machines to merge the information into a richer, unified view of that entity. Similarly, when a digital entity is capable of being processed by an online service, a machine should be capable of automatically detecting this compliance and facilitating the interaction between the data and that tool. This requires that the meaning (semantics) of each participating resource – be the data and/or services service – is clear.

- Reusability: Digital resources are sufficiently well described for both humans and computers, such that a machine is capable of deciding: if a digital resource should be reused (i.e. is it relevant to the task at hand?); if a digital resource can be reused, and under what conditions (i.e. do I fulfil the conditions of reuse?); and whom to credit if it is reused.

Ensuring public return on data investment

Given the high value of health data, which would be increasingly shared by individuals, it is important not to reduce it to a commodity that does not bring a societal benefit. While direct payments to individuals raise a number of ethical concerns and may lead to unfair practices, it is also problematic not to have any 'return' of investment on the commercialised gains of data. One of the options to guarantee such a return would be to 'pay back' to society, instead of an individual gain. For instance, if thanks to data made available through the EHDS for research, medicine or treatment are developed and used commercially, conditionalities should apply. A conditionality may include a requirement to allow research results use to other parties and not 'lock in' the acquired knowledge within one entity, independently if research led to the desired outcome or not. There could also be a conditionality to make more affordable those medicines that are developed thanks to the use of public data, thus acknowledging the societal contribution of data as a valuable input into the research outcome.

III. INCREASE DIGITAL HEALTH LITERACY

Digital health literacy is a critical component for people with rare diseases, healthcare professionals, researchers, and other involved stakeholders to have the necessary skills to be able to meaningfully participate in a newly created system, as well as fully benefit from it. Digital health literacy can be considered as the convergence of both digital literacy and health literacy. However, certain competencies of digital health literacy may not be covered by either digital literacy or health literacy; therefore, it is important to make a distinction.[18] The Transactional Model of Digital Health literacy outlines competence levels of digital health literacy [19]:

- Functional: the ability to successfully read and write about health using technological devices;
- Communicative: the ability to control, adapt and collaborate in communication about health with others in online social environments.
- Critical: the ability to evaluate the relevance, trustworthiness, and risks of sharing and receiving health-related information through the digital ecosystem;
- Translational: the ability to apply health-related information from the digital ecosystem in different contexts.

It is necessary to include these elements in educational programmes targeting specific groups. Development of the EHDS must go together with the digital education of healthcare professionals and patient communities to ensure a successful, patient-centred implementation of the EHDS. The proposal should put in place mechanisms to ensure educational and capacity-building assistance to patients to better understand their rights and obligations on how to manage their health data when it comes to uses of both primary and secondary health data. It is a crucial component to ensure a well-functioning data-sharing infrastructure. It should also be user-friendly and intuitive at the EU level,

^[18] van Kessel R et al. (2022) Digital health literacy as a super determinant of health: More than simply the sum of its parts. Internet Interv. 7;27:100500. doi: 10.1016/j.invent.2022.100500. PMID: 35242586; PMCID: PMC8861384.
[19] Paige SR et al. (2018O Proposing a Transactional Model of eHealth Literacy: Concept Analysis. J Med Internet Res. Oct 2;20(10):e10175. doi: 10.2196/10175. PMID: 30279155; PMCID: PMC6231800.

without the sufficient digital literacy levels of end users, the system put in place will not be properly used. The rare disease community might need specificallydesigned educational programmes due to the unique link with the ERNs and frequent uses of highly sensitive data categories, such as genetic data. A supportive profession specialised in healthcare digitalisation should be present to provide expertise to both healthcare professionals and patients where needed.



In addition, given the unique placement of patient organisations and civil society organisations, these groups can play a trusted offline role to coordinate data sharing and management, enable informed choices for patients about sharing their health-related data, and support the implementation of the European Health Data Space. Moreover, patient and civil society representatives can play a significant role in the development of new tools to control data use through the participatory arrangement of data models – e.g. acting as data cooperatives.[20]

[20] A health data cooperative is a collective where health-related data are integrated, stored, used, and shared under the control of the cooperative members.

IV. ENCOURAGE PATIENTS AND PUBLIC PARTNERSHIPS

A patient partnership is a mutual relationship between persons living with a rare disease and other stakeholders where input from people living with a rare disease or caring for someone with a rare disease routinely and formally informs policy reflections and decisions. Patient partnership implies going beyond empowerment and engagement and considering people living with a rare disease and their advocates as equal partners and actors in policy and programme design and evaluation.

Patient partnership within the EHDS system could significantly increase public understanding of the created health data space, help them to navigate through complex issues, and allow individuals to ensure that their data is used with respect to the set rules and in the best public interest. What is more, it would ensure a better understanding of data sharing pros and cons, weigh them, and manage expectations accordingly.

The EHDS will establish several governance structures at the national level, such as health data access bodies, digital health authorities, and the European level, through the European Health Data Space Board. We call for the involvement of patients and civil society representatives in each structure. The following potential partnership models should be explored:

• National Public Advisory Board for Health Data embedded in the Health Data Access bodies' governance structure

This board would be comprised of individuals or carers of people whose data is made available by the Health Data Access Body, with the goal of consulting and providing feedback to the Health Data Access Boards on individual-centred data sharing. Some of these board members could be actively involved in Health Data Access Bodies' decision-making groups (e.g. Access Review Committee, Ethics Advisory Committee).

• Digital Health Citizen Panel

Citizen Panels composed of citizens randomly selected could serve as an advisory body to the national Digital Health Authorities to provide feedback on diverse matters linked with national healthcare system digitalisation. Sampling methodologies would need to ensure that the panels are representative of each country's local communities in terms of geographic origin, gender, age, socioeconomic background and level of education.

• European Societal Advisory Board

A European Societal Advisory Board embedded in the EHDS Board governance structure, composed of 27 national citizen representatives could play an advisory role to the European Health Data Space Board. Sub-groups may be created to focus on different topics – e.g. a Chair that is a member of the EHDS Board executive board and reports directly to the EHDS Executive Board, and some other members who would be actively involved in the EHDS Board's working groups.

Key functions of patient/civil society consultative bodies



Oversee data access process



Share experiences and advice



Protect participants' interests in research and decision-making



Listen and respond to societal feedback on the EHDS elements

GLOSSARY

Personal data

means any information relating to an identified or identifiable natural person ('data subject'); an identifiable natural person is one who can be identified, directly or indirectly, in particular by reference to an identifier such as a name, an identification number, location data, an online identifier or to one or more factors specific to the physical, physiological, genetic, mental, economic, cultural or social identity of that natural person.[21]

Non-personal data

means data other than personal data.

Health data

means personal data related to the physical or mental health of a natural person, including the provision of healthcare services, which reveal information about his or her health status.[22]

Genetic data

means personal data relating to the inherited or acquired genetic characteristics of a natural person which give unique information about the physiology or the health of that natural person and which result, in particular, from an analysis of a biological sample from the natural person in question.[23]

Primary use of health data

means the processing of personal electronic health data for the provision of health services to assess, maintain or restore the state of health of the natural person to whom that data relates, including the prescription, dispensation and provision of medicinal products and medical devices, as well as for relevant social security, administrative or reimbursement services.[24]

^[21] The General Data Protection Regulation[22] Ibid[23] Ibid[24] Ibid

'Secondary use of health data

means the processing of personal data for purposes other than those for which the personal data were initially collected.[25]

EHR (electronic health record)

means a collection of electronic health data related to a natural person and collected in the health system, processed for healthcare purposes.[26]

EHR system (electronic health record system)

means any appliance or software intended by the manufacturer to be used for storing, intermediating, importing, exporting, converting, editing or viewing electronic health records.[27]

Centres of expertise

healthcare units highly specialised in the management and care of people living with a rare disease, which aim at providing the highest standards of care to deliver a timely diagnosis, appropriate treatments, and follow-up. Each Centre of Expertise is specialised in a single rare disease or in a group of rare diseases.

European Reference Networks (ERNs)

virtual networks involving healthcare providers across Europe. ERNs aim at facilitating discussion on complex or rare diseases and conditions that require highly specialised treatment, concentrated knowledge and resources.

^[25] The General Data Protection Regulation

^[26] Ibid

^[27] Ibid

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