



WHITE PAPER:

**A Patient-Centric
Approach to Consent**



A Patient-CENTRIC Approach to Consent

RARE-X is committed to transforming rare disease by ensuring that patient communities, clinicians, researchers, and drug developers have access to the right data at the right time. At the heart of RARE-X's approach is a belief that patients should own their data. In July 2021, RARE-X launched its first set of rare disease pilot programs on its data collection platform supported by data governance and consent that will ensure participants who spend the time to enter their data are able to share it with those stakeholders they choose.

RARE-X understood that to achieve this vision of enabling data sharing to accelerate research, improve the understanding of rare diseases, and speed diagnoses; it would need to take a novel approach. This approach included creating governance and a patient-centric consent process supported by technology designed for the future life of data. This includes leveraging existing technology platforms for the widespread sharing of data developed at the Broad Institute of MIT and Harvard that follows federated data-sharing principles and enables access to structured data in a semi-automated manner. The RARE-X platform strives for the data collection program to be supported through collaborations with technology providers with the strongest security and data oversight.

The organization has invested significant resources in developing its governance and consent. RARE-X Research and Data Governance Lead Vanessa Vogel-Farley and data governance expert Joy Pritts spearheaded an 18-month effort with the RARE-X governance and compliance, data standards, and patient engagement teams. An extensive network of scientific advisors, survey developers, attorneys, and rare disease patient communities generously shared their time and insights in helping RARE-X forge its approach to governance and consent.

By capitalizing on technology, RARE-X has taken an approach to governance and consent that is forward-looking and provides needed flexibility to ensure data lives beyond its initial and expected use, maximizing potential usage of this data to understand disease and develop therapies. These efforts also aim to avoid requiring participants to enter the same data repeatedly over time.

What RARE-X did

Consent documents can be long, difficult to understand, and overwhelming for research participants to work through. Often consent agreements do not contemplate or allow for the future use of data beyond the immediate intended use. To minimize the burden on participants while ensuring understanding of the consent document, RARE-X breaks down the consent process into short, readable pieces. By using straightforward language that can be understood by people of any education level, the consent process can be completed without requiring a significant investment of time or sacrificing participants' understanding of what they are agreeing to do. Also, RARE-X recognizes that different participants may have different interests and needs with regards to what may be considered effective data to collect. As such, the consent process supports all types of participants and participant relationships for the consent process. And, the organization has done this with an eye toward enabling multi-stakeholder data sets to support cross-disease research.

RARE-X has taken a modular approach to its surveys and designed them to be short and easy to complete. Based on the participants' responses, they will be invited to answer other short surveys, and



they are not asked to complete surveys that do not apply to them. The organization has applied this same approach to its consent agreement. RARE-X provides a simple consent form that is understandable to a broad set of participants. The consents include summaries of each consent topic for participants to quickly review the important consent aspects, and additional details in an easy-to-digest, Q and A format to provide a more granular understanding. In essence, the consent document allows rare disease patients and/or their parents or legal guardians to agree to provide the participants data for rare disease data-collection purposes. They will later be able to determine with whom they share their data and for what uses through a separate data-sharing preference survey.

Consent agreements are legal, binding documents that do not change over time. They remain in force until they are revoked or replaced with a new consent agreement. RARE-X has turned the traditional consent process on its head. By restricting the consent document to the agreement of data collection and using a separate data sharing preference survey, it allows the participant to easily change their data sharing preference without altering their initial consent agreement. This separate survey allows participants to choose with whom they are willing to share their data and how it may be used. It allows them to choose to do such things as participate in biospecimen collections, share their data with rare disease drug developers, or share it with patient communities. Their responses to the data preference survey create a data-sharing ontology that codes their data. RARE-X then uses that ontology to automatically manage who can access the data based on the data-sharing selections tied to it.

All of this is enabled by technology that can easily track and allow sharing according to a participant's stated preferences. As an example of the power of this flexible approach, a participant may not want to share their data with industry today, but if a company reaches out to RARE-X in the future because of a new clinical study for which a participant would qualify, RARE-X is able to reach out to notify the participant of the opportunity. There is no need to go through a new consent process. All the participant needs to do is update his or her data-sharing preferences, if desired.

Why RARE-X did this

Over the years, traditional consent forms have become constructed to include more legal jargon to serve the legal and ethical review requirements of the institutions collecting the data rather than focusing on the participant's understanding of the study procedures. As such, traditional consent agreements focus on the immediate purpose for which the data is being collected. They are not concerned with addressing potential future uses for the data that may arise or realizing the broadest potential benefits from the data being collected.

RARE-X knows there are many barriers to data-sharing. It believes the best way to remove obstacles to sharing data is to enable patients to control who can access their data and use it. Academic researchers may not want to share data ahead of publishing. Drug companies may not want to share data because of competitive reasons. Rare disease organizations working within the same disease area may refuse to share data with other organizations they view as competitors. Even well-intentioned data owners who wish to share data may be prohibited from doing so because of narrowly written consent agreements. RARE-X wants to ensure that patients have control and can share their data with whomever they like. RARE-X's governance and consent forms have been built for this purpose. It allows patients to decide who they share their data with, and it allows them to change their minds over time.



Why it's different

RARE-X knows that to truly drive progress in rare disease forward, data needs to have a life and purpose after its collection for its original, intended use. Unfortunately, in some cases, competition or previous lack of vision has resulted in data being locked away in silos rather than helping advance our understanding of rare diseases, improve diagnosis, and speed the development of new therapies.

The RARE-X consent and governance processes capitalize on advances in information technology that allow us to create a flexible, changeable, and interoperable process. Most importantly, it recognizes patients playing an ownership role with their own data and gives them, rather than a researcher, patient organization, or corporation, the power to determine who has access to their data. RARE-X also knows that current patients may not be able to decide how their data is used in the future because of a loss of cognitive abilities or death. Rather than lose the use of that data, the structure RARE-X has created ensures the longevity of the data, and a caregiver or legally authorized representative can set the data-sharing preferences. Similarly, young patients who today may not be able to provide informed consent can take control of their data once they reach an age where they can make their own decisions about data-sharing preferences. This also allows their original data to live on under their control.

The vision and the need

Increasingly, researchers in rare disease have come to recognize that the breakthrough to treatments for one disorder may lie in the insights provided in a different disorder. To enable these types of insights, data needs to be structured to ensure that it can be connected and interoperable. Unfortunately, too often rare disease researchers have their hands tied with their own protocols and well-intentioned protections of participants ensuring they would not share the data they collect.

RARE-X knows rare diseases know no borders. It also knows that rare and ultra-rare diseases can be heterogeneous and that it's critical to get data from a large enough sample of participants to provide meaningful insights. Too often, research studies based on small patient samples are not representative of the true broad population with a given condition. As the patient population grows, the insights are not updated quickly enough to provide the newest insights. RARE-X wants to ensure it's reaching as many patients as possible and opening up the research realm by helping scientists connect to patients they would not otherwise know existed or did not have access to.

RARE-X believes that by lowering barriers to participation for rare disease patients while protecting their privacy and allowing them to retain ownership of their own data, RARE-X will discover patients where they are (location, education level, engagement abilities), rather than relying on an academic center of excellence to identify them. This will help researchers access a larger pool of patients with a given disorder, accelerate the enrollment of clinical trials, and help entice biopharmaceutical companies to pursue treatment for disorders by helping them better understand the size of a patient population and the nature of a condition. RARE-X's goal is to get to as many patients as possible that may never have been identified or able to otherwise say, "I am here, and I would like to give data about myself and what it's like to live with this condition."

With an estimated 9,500 rare diseases, looking at them one at a time will not be the best approach to yielding the answers patients seek. Looking across rare disease can make it possible for researchers to



understand correlations between diseases and may reveal common biologic pathways, shared features, or common mechanisms underlying different conditions.

The RARE-X platform makes this possible. The governance of the data it houses enables patients to choose to allow researchers to leverage RARE-X housed data across diseases to help find patterns and answers that might not otherwise be possible to pursue

RARE-X has built a data-collection program with a technical platform and governance that enables a new generation of research that moves beyond an isolated researcher looking at a single disease to a collaborative and cross-disciplinary approach where biomedical researchers, bioinformaticians, computer scientists, data scientists, mathematicians, and others can contribute their unique talents to solving challenges of diagnosing, treating, and curing rare diseases. The status quo won't support the future of research. RARE-X will continue to work to leverage technology in innovative ways to accelerate advances that only a few years ago would have been unimaginable.





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