Patient Registry Design Guide for Rare Disease Foundations

by

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The authors of this guide have a unique connection to this topic because they have professional careers in the healthcare and life sciences arenas, and they have personally dealt with the effects of rare disease in their own lives.

A patient registry is a collection of observed data collected around a specific disease, indication, or other qualifier.[1] Patient registries can be used in conjunction with various aspects of a patient population. A large use of registries in rare disease communities is to collect natural history data. Natural history data is data collected over time that can show the evolution of a rare disease for a patient and can bring into focus the complete picture of that disease. Natural history data collection can be invaluable in many ways. For example, during an interventional drug trial, the natural history data can replace the need for a placebo arm so that all patients can receive the drug.

Additionally, patient registries for rare disease communities can be used as a means of engaging the rare disease population, obtaining their opinions and perspectives, and providing a list of contacts (Contact Registry) for clinical trial opportunities. Each of these areas can be vitally important to the goals of a rare disease foundation. Thus, it is critical that the goals and objectives of a registry are understood and clearly communicated to all stakeholders.

Patients often place enormous hope and trust into their participation in research.[2] Patient registries can have higher participation and be much more successful if the participants see themselves as true partners and feel that their time and efforts are valued and respected. Collaborative relationships between researchers, patient foundations, and patient community along with clear and transparent communication are critical in creating and maintaining a thriving registry.

Often, multiple patient foundations in the same disease area create their own registries. However, if the ultimate goal is to have valuable data that will lead to treatments and eventually cures, it is optimal to have as much data as possible collected together, either within same registry or within different registries using the same registry platform. A registry platform that allows for multiple organizations to contribute data to the same registry, while each maintaining the ownership of their data, can be an immense benefit for all involved. Collaboration between organizations is integral in deciding the best way to collect and store useful data.

Some features that are important to consider when selecting a patient registry platform are:

- Can data be entered by patients, clinicians, or both?
- What data standards are used, and will the platform comply with regulatory agencies such as FDA and EMA?
- How will data be analyzed?

- Should data be entered and collated into the same registry by multiple organizations that support the same disease?
- How is ownership of the data decided and governed?
- Is there a need to accept electronic health records?
- Will international support and compliance be needed?
- Is a caregiver role available, and can a caregiver input data on behalf of a patient?
- Is informed consent capability included?
- What types of devices (phone, tablet, laptop, desktop, etc.) can be used?
- Are alerts and reminders available to prompt data collection?

Rare disease foundations, in some cases, do not realize how much time and technical expertise need to be initially invested to obtain meaningful data. This initial investment will produce a much better data set, will provide better validation, and will entice researchers to have interest in the data immediately. The collection of usable data is also a springboard for drug development. Conversely, without an initial investment in collecting meaningful data, there is a possibility that the data collected is of little or no use to researchers.

The following are some of the design and data issues that commonly occur and should be considered as the registry is designed:

- Confusion between "patient" and "caregiver" in the wording of the questions should be avoided. This is especially important to consider in the area of rare disease, where surveys are often answered by caregivers. It is a good practice to initially establish the relationship between the patient and the person filling out the survey and to phrase the rest of the questions in a way that would make it clear whether the question refers to the patient with the disease or the caregiver. Using data standards, such as CDISC standards, can be very helpful in this process.[2,3]

- Collection forms that are too long or too detailed can create an unnecessary burden for patients and can lead to inaccurate or missing data; in turn, this creates problems when data is analyzed and interpreted. Because of this, it is important to invest initially in deciding which information is necessary before any forms are created. If numerous questions need to be asked, it may be best to have several short and focused surveys.[1,2]

- The same or similar questions asked more than once can cause inaccuracies. For example, asking for the date of birth during registration and later asking for age can lead to inconsistencies and should be avoided. The best design is to ask for the date of birth once and then use that to derive the patient's age.[2]

- If participants are unclear on whose patient profile they are answering questions about, there can be significant data issues. For example, in a case where the same account holder has profiles for several family members with the same disease, showing each patient's first and last name and date of birth on each screen of the survey can be helpful.[2]

- Realizing a problem exists after a great deal of data has already been collected can be detrimental. It is important to initially test surveys on a small group of patient volunteers and to analyze the data to decide if improvements need to be made before sending the survey to the entire group. Once data collection starts with the larger group of patients, it is equally important to check it at regular intervals to catch any other problems that may have been initially missed.[2]

A patient registry is a powerful tool that can help patient foundations engage their patient and medical community, learn about the patient's perspectives and experiences, identify candidates for clinical trials, and collect data to use for research and drug development. A registry, however, will only be successful if the appropriate time and expertise are invested in order to understand the goals of the registry, select the optimal platform to be used, design the most appropriate surveys for the population, and continue to stay involved to address any concerns. Collaboration, transparency, and open communication between patient foundations and patients who contribute data are crucial to the long-term success of the patient registry.

About the Authors:

Jason Colquitt

Jason Colquitt lives in Carrollton, Georgia, and has been blessed to meet and work with many amazing people and organizations across his 20+ years in the healthcare technology field. His work has caused positive disruption within the healthcare industry as he has partnered with many organizations ranging from small start-ups to some of the world's largest companies. He has worked directly with patients, caregivers, physicians, regulators, and researchers. These past experiences and relationships have led him to focus as CEO of a company he founded in 2012 called Across Healthcare (https://acrosshealthcare.com/). Jason was diagnosed with Carnitine Palmitoyltransferase II Deficiency (CPT II), a rare mitochondrial disease, and believes he has been called to use his journey and technical background to help the rare disease community.
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Sophia Zilber lives in Boston, MA. Sophia has been working for 15+ years in the pharmaceutical industry and is currently working in Pfizer as a Statistical Programming Lead in Rare Disease Research Unit. Sophia's newborn daughter Miriam died from Leigh Syndrome, a form of the Mitochondrial Disease, in 2017. Following this tragic event, Sophia volunteered a lot of her time and professional and personal experience to help rare disease community. She has done a thorough data analysis of mitochondrial disease patient registry stewarded by United Mitochondrial Disease Foundation (UMDF) and was able to give a voice to the patients and caregivers who shared their comments in the registry as well as summarize multiple valuable lessons that can be helpful to others working with patient registries. Sophia also shared her personal experiences through blog posts and articles published in American Academy of Pediatrics Palliative Care Newsletter, KevinMD blog, Courageous Parents Network, and Global Genes. Sophia serves on the boards of Rare New England and New England Regional Genetics Group.
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