The Value of Patient Registries: Common Needs for Rare Diseases

Nicole Mittman, Oliver Varette

Real-World Data (RWD) for decision-making sources include medico-administrative databases, billing and insurance claims, reporting from patients and clinicians, and observational research methods. Rare Disease Registries (RDR) platforms serve an important function in assembling and linking RWD for a broader population of patients—they collect RWD to evaluate natural histories of disease, outcome measures, standards of care, and drug safety and effectiveness. Patients with a rare disease are often sparsely populated throughout the country, with individual provinces and territories largely regulating the provision of health care in their own jurisdictions.

Rare Diseases Globally

There are approximately 350 million people living with rare diseases globally, with about 7,000 rare diseases identified. Of these diseases, more than 80% have a known genetic cause, yet only 10% have an approved therapy available to patients. In total, 75% of the diseases are pediatric and 30% of these impacted children do not live to the age of 5.[1] Research for new therapeutic options for rare diseases, which necessitates assembling large and representative patient cohorts to assess drug safety and effectiveness, can be challenging due to small and dispersed patient populations. RWD could be a complementary data source outside the clinical trial space that could be used to inform and support the work off clinicians, researchers, and decision-makers.

Real-World Evidence and Data Impact

Real-world evidence (RWE) incorporates various streams of RWD and may offer new perspectives to decision-makers in Canada and across the globe. RWE centres on the delivery of health care to inclusive, real-life populations to bridge existing evidence gaps related to drug safety, effectiveness, and value to ultimately promote public health. In recent years, there has been an increased appetite for exploring complementary sources of high-quality data to address current barriers and knowledge gaps.

RDR Challenges and Examples of Best Practices

There are some challenges that limit the use of RDRs in health technology assessment and other related fields of health research; these limits converge on uncertainties with data quality and an ongoing need to develop tools and guidance.

The European Network for Health Technology Assessment (EUnetHTA) has developed a Registry Evaluation and Quality Standards Tool (REQueST), which outlines universal and essential elements of good practice and evidence quality that can help guide the use of registries for heath technology assessment and decision-making. CADTH is using REQueST to understand the quality, accuracy, governance, and utility of data housed in RDRs.

To that end, *Reflections on the Canadian Bleeding Disorders Registry: Lessons Learned and Future Perspectives* published in the *Journal of Health Technologies* by Iorio and colleagues provides a comprehensive overview of the development and utility of a national database for hemophilia and other rare inherited bleeding disorders in Canada.[2] The CBDR project was initially launched in the 1970s and has since developed into a national resource that has contributed to driving advances in care standards, treatment availability, tools for engaging patients, and ultimately, the quality of health care available to patients. In particular, the CBDR has served as a source of RWD to support research and decision-makers, facilitate patient access to treatment, and serve as a connecting hub that unites clinicians, researchers, and patients. The article provides insights on networks, structure, and functions across a range of bleeding disorder indications.

Conclusion

Several barriers remain that impede the widespread use of RDRs for RWD, including concerns over data quality, governance, and implementation of the assembled RWE for decision-making purposes. This article is the first in a series of case studies that CADTH is pursuing that focus on registries for rare diseases, with the goal of leveraging the power of registries to improve the portrait of care for several rare diseases and support the decision-makers responsible for enacting these changes.

References

1. Urv T, Wong-Rieger D, Merkel PA, Macchia F, Sanders S. 7,000 challenges: the basis and burden of rare diseases. *Science and Life* [Webinar]. 2021; https://www.science.org/content/webinar/7000-challenges-basis-and-burden-rare-diseases?msclkid=b344688cd13c11ec9b130bbde9756fca. Accessed 2022 May 10.

2. Iorio A, Grenier S, Page D, et al. Reflections on the Canadian Bleeding Disorders Registry: lessons learned and future perspectives. *Can J Health Technol.* 2022; 2 (7).