

## Beyond Best Practices, Measuring Success in Rare Disease Registries

"Rare diseases impact less than 200,000 individuals in the United States. There are nearly 7,000 such conditions that impact nearly 30 million people in the US. Rare disease patients often face challenges getting an accurate diagnosis, finding medical professionals familiar with their condition, and finding effective treatment options. Patients and families often feel isolated and desperate for additional research on their condition.

Registries are tools used to collect data. Registries can be used to perform natural history studies and collect social, economic, quality of life, and clinical data. They can increase healthcare planning and improve the coordination of patient care. They can also assist researchers to ensure a sufficient sample size for clinical trials or other research initiatives. Rare disease registries can focus on a specific disease, a group of diseases, on a specific treatment, or on a collection of diseases, this is often dependent on the sponsor. Data collectors usually include physicians caregivers, and patients.

An established framework for evaluating successful implementation and the long-term organizational goals associated with launching patient registries does not currently exist. The focus on current assessment measures in registries focus on the outcomes discussed in the patient reported outcomes research and the common data elements to populate the registry. To guide a more comprehensive assessment plan, the CDC Framework for Program Evaluation in Public Health was identified. The purpose is to investigate the entire process through the steps of the program and apply the standards at each phase. The steps are engage stakeholders, describe the program, focus the evaluation design, gather credible evidence, justify conclusions, and ensure use and share lessons learned. Registry specific metrics were then applied to the model.

Some measures are specific to the implementation process and should be considered short-term objectives, such as 1.a. Establish and document clear roles for each of the primary stakeholders. However, other objectives are tied to long-term goals related to health outcomes and research objectives, such as 6.c. An increase in the number of published research studies. Evaluation should occur as key project implementation milestones are met and then on an annual basis. An associated generic implementation timeline was created that includes key project deliverables and tasks that should be completed prior to the launch of a registry."

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