# Preface to the AHRQ Supplement

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According to a 2010 Institute of Medicine (IOM) report,[1] an estimated 5,000 to 8,000 diseases are categorized as rare and together affect millions of Americans. While some rare diseases have a very small patient population, others affect many thousands of people in the United States and worldwide. As the IOM report states, "rare diseases are not rare, at least in the aggregate"[1] and are responsible for an enormous impact on the lives of patients, their families, and health care systems. For many rare diseases, there are no diagnostic tests or treatments; and there is a lack of basic knowledge about their natural history, diagnosis, and how best to provide health care to those who are affected. Yet, for other rare diseases, despite the growing number of diagnostic tests and treatment options, we have insufficient evidence on their long-term effects, comparative effectiveness, and safety. Often research is also limited in its ability to assess how health care delivery affects patient outcomes or how medical practice can be improved to address the unique needs of patients living with rare diseases. The importance of outcomes research for rare diseases was recognized in the initial national priorities for comparative effectiveness research[2] and in the Patient Protection and Affordable Care Act, which required the Patient-Centered Outcomes Research Institute to appoint an expert advisory panel for rare disease research.

Despite the large knowledge gaps, there is tremendous potential for supporting a new generation of health outcomes studies in rare diseases through a growing number of patient registries, databases, and new methodological approaches. The Agency for Healthcare Research and Quality (AHRQ), through its Developing Evidence to Inform Decisions about Effectiveness (DEcIDE) Research Network, sponsored this supplement to present various strategies in the design, analysis, and conduct of health outcomes studies relevant to rare diseases. The purpose of this supplement is to disseminate illustrative examples of research methods that can be applied to understand health outcomes and to potentially stimulate new patient-centered outcomes studies for rare diseases. The inventive approaches used by researchers to study specific rare diseases could provide models for other researchers to emulate or adapt when designing health outcomes studies. While this supplement is not intended to endorse particular research methods or address all the challenges of conducting research in rare diseases, it does present a wide range of methods for studying health outcomes in this field. With the exponential growth in health data and the blurring of lines between observational research and clinical trials, new opportunities abound for initiating innovative research that directly helps improve health care for patients with rare diseases.

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## REFERENCES

1. Institute of Medicine Committee on Accelerating Rare Diseases Research and Orphan Product Development, Field MJ, Boat TF, eds. Rare Diseases and Orphan Products: Accelerating Research and Development. Washington (DC): National Academies Press (US); 2010. Available from: http://www.ncbi.nlm.nih.gov/books/NBK56189/.
2. Institute of Medicine. Initial National Priorities for Comparative Effectiveness Research. Washington, DC: The National Academies Press; 2009. http://www.effectivehealthcare.ahrq.gov/tools-and-resources/researcher-resources/.