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Abstract

An overview of rare disease characteristics on research methodology

Danielle W. Hampel
University of California, USA.

Abstract:
Background About 30 million individuals in the United States are living with a rare disease, which by definition have a prevalence of 200,000 or fewer cases in the United States. Disease heterogeneity and geographic dispersion add to the difficulty of completing robust studies in small populations. Improving the ability to conduct research on rare diseases would have a significant impact on population health. The purpose of this paper is to raise awareness of methodological approaches that can address the challenges to conducting robust research on rare diseases. ApproachWe conducted a landscape review of available methodological and analytic approaches to address the challenges of rare disease research. Our objectives were to: 1. identify algorithms for matching study design to rare disease attributes and the methodological approaches applicable to these algorithms; 2. draw inferences on how research communities and infrastructure can contribute to the efficiency of research on rare diseases; and 3. to describe methodological approaches in the rare disease portfolio of the Patient-Centered Outcomes Research Institute (PCORI), a funder promoting both rare disease research and research infrastructure. Results. We identified three algorithms for matching study design to rare disease or intervention characteristics (Gagne, et.al, BMJ 349:g6802, 2014); (Gupta, et.al, J Clin Epidemiol 64:1085-1094, 2011); (Cornu, et. al, Orphet J Rare Dis 8:48,2012) and summarized the applicable methodological and analytic approaches. From this literature we were also able to draw inferences on how an effective research infrastructure can set an agenda, prioritize studies, accelerate accrual, catalyze patient engagement and terminate poorly performing studies. Of the 24 rare disease projects in the PCORI portfolio, 11 are randomized controlled trials (RCTs) using standard designs. Thirteen are observational studies using case-control, prospective cohort, or natural history designs. PCORI has supported the development of 9 Patient-Powered Research Networks (PPRNs) focused on rare diseases. Conclusion Matching research design to attributes of rare diseases and interventions can facilitate the completion of RCTs that are adequately powered. An effective research infrastructure can improve efficiency and avoid waste in rare disease research. Our review of the PCORI research portfolio demonstrates that it is feasible to conduct RCTs in rare disease. However, most of these studies are using standard RCT designs. This suggests that use of a broader array of methodological approaches to RCTs –such as adaptive trials, cross-over trials, and early escape designs can improve the productivity of robust research in rare diseases.

Treatment involves intake of hormones such as cortisol and aldosterone.

Biography:

Danielle W. Hampel is currently working as a lecturer in University of California, USA. She is having teaching experience. She is interested in Sample Preparation, SDS-PAGE, Cell Culture and Molecular Biology.

Recent Publications:

1. Association of maternal diet, micronutrient status, and milk volume with milk micronutrient concentrations in Indonesian mothers at 2 and 5 months postpartum, Danielle W. Hampel
2. Micronutrient supplementation of lactating Guatemalan women acutely increases infants' intake of riboflavin, thiamin, pyridoxal, and cobalamin, but not niacin, in a randomized crossover trial, Danielle W. Hampel
3. High-Throughput Analysis of Water-Soluble Choline and Related Metabolites in Human Milk, Danielle W. Hampel
4. The Associations Between Light Exposure During Pumping and Holder Pasteurization and the Macronutrient and Vitamin Concentrations in Human Milk, Danielle W. Hampel

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