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Bardet–Biedl syndrome: A model for translational research in rare diseases,,

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Abstract

Bardet-Biedl syndrome (BBS) is a rare, multisystemic, genetic disease and member of a group of disorders called ciliopathies. This syndrome provides a mechanistic model for ciliopathies that may also extend to common disorders with complex inheritance patterns, including diabetes mellitus and obesity. Dysregulation of signaling pathways altering the cellular response to the extracellular environment is primary to the ciliopathies and characteristic of BBS. As BBS-centered translational research moves forward, innovative advances provide opportunities to improve the care of individuals with BBS and other rare diseases as well as common related conditions. This review aims to highlight the current understanding of the mechanisms underlying BBS and opportunities for advancing the care of individuals with rare diseases. Focal points: Bedside: understanding the multi-dimensional manifestations of ciliopathies, specifically Bardet- Biedl Syndrome (BBS) as a model ciliopathy, will accelerate research into therapeutic targets for ciliopathies, allowing for improved therapies for individuals with these debilitating disorders. Benchside: elucidating the molecular mechanisms of BBS is likely to increase the chance of discovering novel therapeutic approaches that may be generalizable to other ciliopathies and perhaps to common related disorders, such as obesity and diabetes mellitus. Industry: application of known drugs to new indications, or drug repositioning, and development of novel therapeutics, including gene therapies in BBS, may open new avenues for therapeutic discovery and development. Community: rare diseases affect millions of individuals throughout the world with significant impact on quality of life and longevity. The development of multidisciplinary clinics for BBS and effective implementation of a rare disease registry provides a model for advancing the care of individuals with rare diseases. Government and Regulatory Agencies: the importance of rare disease research and the impact of that research on common disorders should be supported with adequate funding and resources. Understanding the molecular pathways underlying ciliopathies, such as BBS, and advancement of translational medicine in ciliopathies will have far reaching societal benefits

Keywords: