Title: **Canada's path forward for rare diseases: Discovery to translation**

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Kym Boycott is a Medical Geneticist at the Children's Hospital of Eastern Ontario (CHEO) and Investigator at the CHEO Research Institute. She is an Associate Professor and holds a Tier II Research Chair in Neurogenetics from the Faculty of Medicine at the University of Ottawa. She completed her PhD, MD and FRCPC training in Medical Genetics at the University of Calgary. Dr. Boycott's research, bridging clinical medicine to basic research, is focused on elucidating the molecular pathogenesis of rare genetic diseases using next-generation sequencing approaches. She has been the recipient of the Canadian Institutes of Health Research Clinical Investigatorship Award from the Institute of Genetics, the SickKids Foundation Young Investigator Award and the Basil O'Connor March of Dimes Young Investigator Award. She was the Lead Investigator of the Genome Canada and CIHR funded 'Finding of Rare Disease Genes in Canada' (FORGE Canada) project, which investigated the molecular etiology of more than 250 rare pediatric diseases, identifying the cause in more than 55% and making 67 novel disease gene discoveries. She currently leads of the Genome Canada and CIHR funded large-scale project 'Enhanced CARE for RARE Genetic Diseases in Canada' which is focused on improving the clinical care of patients and families by expanding and improving the diagnosis and treatment of rare diseases.

Abstract: Work over the past 25 years has resulted in the identification of genes for approximately half of the estimated 7,000 rare monogenic diseases; it is predicted that most of the remaining disease genes will be identified by the year 2020. This marked acceleration is the result of next-generation sequencing (NGS) and analysis. However, as many as half of children affected with a rare genetic disease are still without a molecular diagnosis. In addition, effective therapies for rare genetic diseases are themselves comparatively rare. For many rare diseases where there may exist a therapeutic opportunity, the diversity and number of rare diseases combined with the small numbers of patients for each disorder effectively precludes, for all but a fraction of conditions, traditional costly drug discovery approaches. This presentation will highlight the expanding diagnostic and therapeutic challenges and opportunities for rare diseases as well as Canada's national collaborative platforms responding to the evolving rare disease space. Beginning in 2011, Canada established FORGE (Finding of Rare Disease Genes); a large-scale network configured to identify causal genes for

undiagnosed rare diseases. This program was followed by its successor, Canadian Care4Rare, undertaking both diagnoses as well as pre-clinical rare disease therapy development. The PhenomeCentral database was then developed as a repository for the country's standardized phenotypic and genotypic RD data, enabling data sharing for unsolved cases nationally and with international partners. To address the need for biological context for most rare disease genes, we have established the Rare Diseases Models and Mechanisms Network to mobilize the Canadian biomedical community of laboratory scientists and clinicians to work together to provide functional insights into rare disease genes. Finally, to generate research tools and data to facilitate drug discovery for rare genetic diseases, we have established a public-private-partnership with industry partners. Taken together, these platforms will accelerate scientific discovery and downstream clinical translation for patients and families affected by rare diseases.