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PRESENTATION TITLE: Drugging the human genome – The use of germline genomics in drug discovery and precision drug development

ABSTRACT: Massive investments in human genetics and genomics were anticipated to result in many innovative therapies. Using conservative criteria, we identified 40 germline genetic observations that directly led to new targets and, subsequently, to novel non-cancer therapies approved for rare (n=36) or common (n=4) conditions. Most genetically-driven therapeutics for rare diseases compensate for disease-causing loss-of-function mutations in target genes. Therapeutics approved for common conditions are inhibitors designed to pharmacologically mimic the natural disease-protective effects of rare loss-of-function variants. Genetics and genomics are presently enriching pharmaceutical pipelines with a wealth of newly discovered, robustly validated drug targets. Moreover, as they are used to select patients most susceptible to respond to investigational therapeutics, genetics and genomics have the potential to improve success rates and accelerate the clinical development of upcoming therapeutics. This type of "precision drug development" requires more investments into large, diverse, disease-based cohorts of patients deeply phenotyped and appropriately consented for such trials. During this presentation, I will describe the efforts we are doing in the CERC to capture this opportunity.

BIOGRAPHY: Vincent Mooser MD holds since 2019 the Canada Excellence Research Chair in Genomic Medicine at McGill entitled "From Genes to Innovative Therapies". He has a broad experience in internal medicine, human genetics and genomics and pharmaceutical sciences. His work over the past 25 years (including 10 years in industry) focuses on the exploitation of natural variations of the human genome to inform the discovery and development of novel therapeutics.