Application number/Title: 25205 - Identification and characterization of functional variants with a causal role in chronic cardiopulmonary diseases

Applicant PI: Dr Sébastien Thériault

Application Institution:
IUCPQ, 2725 Chemin Ste-Foy, Quebec City, G1V 4G5, Canada

Keywords provided by the Applicant PI to describe the research project:

Cardiopulmonary diseases, genetic, causal variants, targets

Application Lay Summary:

1a: Our goal is to characterize genetic determinants involved in chronic cardiopulmonary diseases, including cardiovascular disease (CVD) and aortic valve stenosis as well as respiratory diseases most particularly asthma, chronic obstructive pulmonary disease (COPD) and lung cancer. Our project comprises two aims: 1) Identify genetic variants and genes causally involved in these conditions by leveraging UK Biobank and other publicly available data as well as data from our institution; 2) Identify potential therapeutic targets for interventions and evaluate their risk of adverse effects.

1b: Cardiopulmonary diseases contribute significantly to morbidity and mortality worldwide and in the UK. Genetic variants have been associated with these conditions but their functionality remains unknown. Our research will determine the role and causality of these variants and could therefore contribute to significant improvements in prevention, screening and diagnostic procedures. Identification of specific genes and biological pathways involved in the pathogenesis of cardiopulmonary diseases could also lead to the development of new therapeutic agents.
1c: We will first use genetic and clinical data from UK Biobank to identify novel genetic variants associated with cardiopulmonary diseases and their risk factors and replicate previous findings. We will then examine the impact of those variants and others identified locally on a wide range of phenotypes to gain insight on the mechanisms explaining their association and identify variants more likely to be functional and causal. Finally, we will use this information to identify potential therapeutic targets for these life-threatening conditions.

1d: We are requesting data from all available participants. Data from the whole

cohort will be required to obtain sufficient power to detect genetic associations. We have experience in analyzing large datasets and genomic data.