**Seneca Hackathon 2021 Roche Project**

**Problem Statement:** It is estimated that there are over 300 million people worldwide living with one or more of ~7,000 identified rare diseases. While specific definitions can vary, a rare disease is an illness that affects a small percentage of the population. In Canada, about 1 in 12 Canadians, or ~3 million Canadians, are affected by a rare disease. The exact cause of many rare diseases is unknown, but most are caused by genetic changes often appearing at birth or emerging in early childhood. Rare diseases can also be caused by infections, allergies and environmental causes, or are degenerative and proliferative.

Rare disease patient management faces a number of challenges and comes with a significant burden on individuals, their families, and society as a whole. A key challenge faced is that small patient populations can ultimately lead to limited data and greater uncertainty around the understanding of a disease. There is constant research ongoing to better healthcare knowledge of rare diseases in order to optimize patient care. Leveraging patient registry data is one area that could help improve the diagnosis and care of patients with rare diseases.

**Foundational Objective:** How can open-registry clinical data be used to help Healthcare Professionals (HCPs) and the Healthcare System in patient care decision-making for rare diseases?

**Focus Areas:** Rare Diseases listed in Canada (as listed in Epidemiology Dataset below)

**Datasets:**

* Open dataset provided by Orphanet (<http://www.orphadata.org/cgi-bin/index.php>)
  + Genes Associated with Rare Diseases (<http://www.orphadata.org/data/xml/en_product6.xml>),
  + Phenotypes Associated with Rare Disorders (<http://www.orphadata.org/data/xml/en_product4.xml>),
  + Rare Disease Epidemiology (Filter for Canada) (<http://www.orphadata.org/data/xml/en_product9_prev.xml>),
  + Rare Diseases and Functional Consequences (<http://www.orphadata.org/data/xml/en_funct_consequences.xml>)
  + Groups free to explore other Orphadata as well
* Reference - Orphanet: an online rare disease and orphan drug database. © INSERM 1999. Available on http://www.orpha.net. *Accessed Date*.

**Project Thought-Starters:**

* How could this data be packaged, or visualized, in a way that could further support the Healthcare Ecosystem, including individual HCPs and patient organizations?
* Based on phenotypes, could this data help a HCP move efficiently towards a faster diagnosis or appropriate gene testing?
* Are there common genes that could be readily tested as a part of practice for Rare Diseases?
* What are some top considerations in relation to functional consequences of Rare Diseases?
* Based on findings, do we believe different groups (e.g. socioeconomic, location, background) will encounter different challenges with the burden of rare diseases?

**General Evaluation Criteria:**

* *Solution:* The level at which the solution addresses the foundational objective.
* *Findings:* Quality of insights gathered from the datasets.
* *Feasibility:* Ability to implement the proposed solution into the Healthcare System.
* *Creativity:* Is a unique approach taken to develop the solution and/or uncover insights.
* *Presentation:* Are the key messages delivered in a clear and concise way.