IGC MSc Project

Title: Establishment of genetic markers with predictive capacity for the severity and prognosis of Early Onset Type 1 Diabetes (EOT1D).

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Abstract

Type 1 Diabetes (T1D) is a chronic disease where the immune system destroys the insulin-producing pancreatic beta cells. The disease emerges particularly in children and the young, imposing life-long dependence on insulin. In the last few decades we’ve witnessed an increase in T1D prevalence at all age groups, particularly in pre-school children. The disease, in these young children referred as Early Onset Type 1 Diabetes (EOT1D), is frequently associated with severe metabolic decompensation and abrupt loss of pancreatic beta cell activity. The severity of the disease and its emergence at an early age strongly imply the role of genetic factors in the pathogenesis of PT1D.

The main goal for this project is to identify genetic variants causing EOT1D through whole exome sequencing of a cohort of 100 children with the disease. So far, 23 samples have been sequenced (with a coverage >100x). Initial variant calling and variant functional annotation of these samples has already been performed. The candidate will apply variant prioritization methods to choose candidates for further experimental functional studies. Initially, this analysis will be limited to a set of candidate genes, and later extend the process to the whole dataset. The candidate will also explore GWAS-like approaches, comparing our cohort with publicly available genotypes of healthy individuals. Finally, we will also explore other complementary methods, such as HLA typing and estimation of copy number variation.

This project should allow the establishment of genetic markers for EOT1D that are predictive of disease severity and prognosis.