PERSIMUNE SCIENTIFIC ANNUAL REPORT 2017

Annual highlights:

Professor Lars Fugger and colleagues contributed to an analysis comparing classified phenotypes against findings from analysis of host genomics and report an improvement of more than 20% increase in power to detect genetic effects over other approaches and identifies new associations between classical human leukocyte antigen (HLA) alleles and common immune-mediated diseases (IMDs). The study emphasises that a more careful system biological characterisation (in this instance host genome bioinformatics analysis) contributes to a better and biologically-determined classification of causes of disease. (https://www.ncbi.nlm.nih.gov/pubmed/28759005). In personalised medicine, it is critical to biologically correctly classify the various phenotypes when exploring which biological mechanisms are causing them – the central focus in PERSIMUNE.

The START study continues to generate important new results. Post doc Jemma O-Connor and colleagues published in Lancet HIV (https://www.ncbi.nlm.nih.gov/pubmed/28063815) that the number of circulating CD4+ lymphocytes (key cell in coordinating the specific immune system) greatly affects the risk of severe bacterial infections in HIV-infected persons, and further that immediate initiation of antiretroviral therapy reduced the risk of these clinical outcomes. This benefit was seen because of reduction of HIV replication, and consequently increasing circulating number of these cells; observed improved number of neutrophils (a cell of the innate immune system), conversely, did not lead to apparent clinical benefit. Diverse profiles of severe bacterial infections were observed ranging from tuberculosis to bacterial pneumonia. These data have implications also beyond HIV for patient groups where impaired CD4+ lymphocyte function is observed.

One of the main features of PERSIMUNE's infrastructure is the creation of a datawarehouse (or data lake), where all available data elements for the cohort of patients under study (approximately 160,000 persons) are combined within one system. This allows for combined analyses of each and all of the data elements using biostatistical, bioinformatics and artificial intelligence analyses techniques. This datawarehouse was generated to enable PERSIMUNE to perform its research. However, during 2017, it has become clear that the features hereof are novel and unique and potentially usable on a larger scale as part of strategies to implement personalised medicine as part of routine practise. Consequently, PERSIMUNE has been approached by multiple national and international stakeholders with interest to better understand the features of this infrastructure.