LUXEMBOURG INSTITUTE OF HEALTH
RESEARCH DEDICATED TO LIFE

CHL Centre Hospitalier de Luxembourg

FROM BIOBANKING TO PRECISION MEDICINE AND BEYOND

The Estonian experience: State-of-the-art in the integration of routine clinical data into research/biobanks

Abstract

The Estonian Biobank was founded in 2000 as a population-based biobank. 15 years later, the biobank includes a collection of health and genetics data of around 5 percent of the adult population of Estonia. All participants of the biobank have gone through a standardized health examination; donated blood samples for purification of DNA, white blood cells, and plasma. A significant part of the cohort (n=20,000) has been genotyped using genome-wide genotyping arrays the rest will be done in 2017. The Human Genes Research Act allows regular updating of data through linkage to national registries enabling long-term follow-up of the cohort and to re-contact the gene donors. Pilot projects for returning the data to the gene donors are underway. In the past few years increasing amount of attention has been placed on translating the results of genetic research to improve public health. A nationwide technical infrastructure (X-road) for the secure electronic exchange of medical data has also been established and is maintained by the state. As of 2010, medical data from health insurance fund, hospitals, primary care physicians and pharmacies (digital prescription records) are all accessible through the X-road in a strictly regulated manner. This allows creating the disease (or life!) trajectories on all gene donors from the birth in the Estonian Biobank, where all contacts with the medical systems incl. ICD-10 diagnoses, prescriptions, lab data and EMR are included. Recently, we have completed the sequencing of the deep (~30X coverage, PCR-Free) whole genomes of 2,500 gene donors in addition to 2500 full exomes. This cohort has been characterized for sequence variations and imputed to GWAS sample. The first data are truly very promising. The function of the rare variants can be elucidated in some cases and they might be potentially very good targets for further development together with industrial partners.

Biosketch

Andres Metspalu, full professor of Biotechnology and Director of the Estonian Genome Center of the University of Tartu, was graduated from the University of Tartu in 1976 with MD, received PhD in 1979. He was as a postdoc at Colombia University and Yale University in 1981-1982. His main scientific interests are human genomics, genetics of complex diseases and population based biopanks and application of the precision medicine in health care. He has published more than 300 papers and chapters in international peer review journals and books. H-index: 51.

This conference is organised in the framework of the "Journée Médicale de la recherche 2016" and will be followed by the prize giving ceremony of the "Prix de la Recherche".

Registration and information: www.jrm.lu

SAVE THE DATE

26th Oct 2016
4pm - 6pm

VENUE

Centre Hospitalier Luxembourg (CHL)

Amphithéâtre
4, rue Barblé
L-1210 Luxembourg

SPEAKER

Prof Andres METSPALU

Professor of Biotechnology
& Director of the Estonian Genome Center, University of Tartu.

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