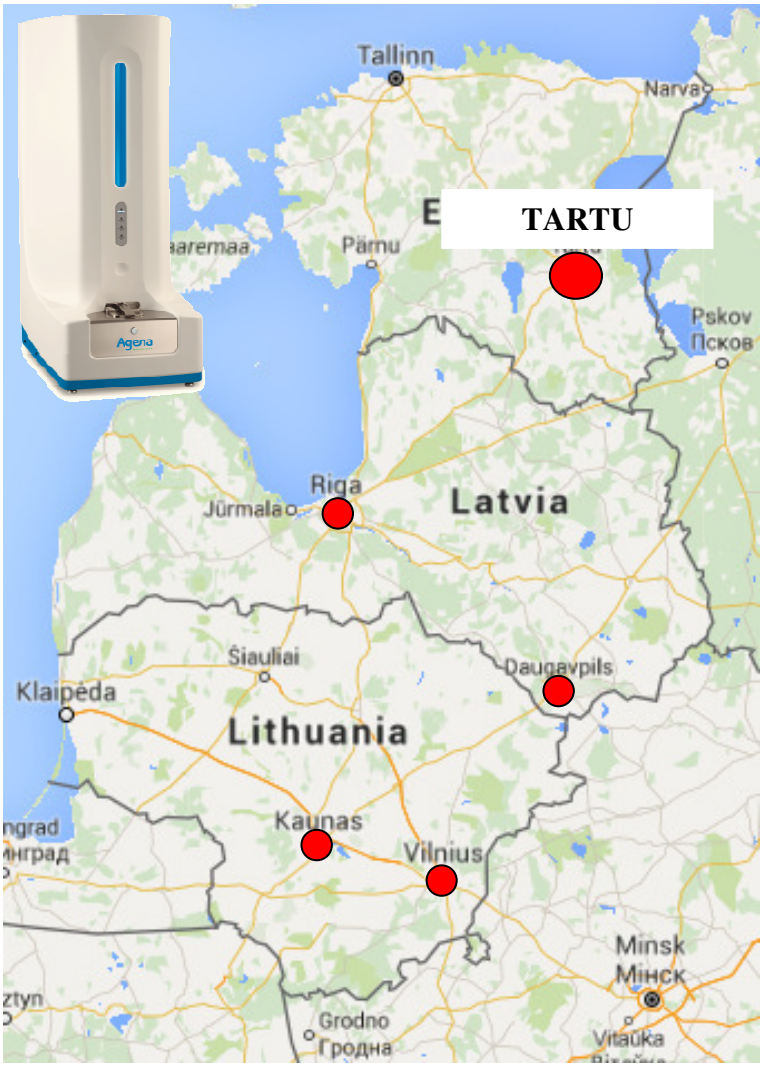
DNA mass spectrometry roadshow: Baltica 2016

Effective Genomics: Using mass spectrometry technology in modern science and diagnostics

22 September 2016



Estonian Biocentre, Riia 23b, 51010 Tartu, Estonia

Scientific & Organizing Committee

Charles Cantor Ph.D
Piotr Grochowski Ph.D
Tadeusz Malewski Ph.D
Wieslaw Kawczynski Ph.D

The DNA mass spectrometry roadshow is a project that takes leading speakers into scientists to talk about the latest advances in genetics. Topics include DNA mass spectrometry; SNP profiling; methylation profiling; gene expression analysis; and further bioinformatics analysis.
Roadshow events are delivered free of charge.

Roadshow programme:

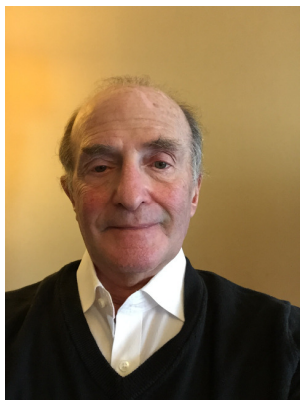
10:00 Lecture 1. Piotr Grochowski - *Introduction to DNA mass spectrometry*
10:30 Lecture 2. Charles Cantor - *The future of nucleic acids diagnostics*
11:30 Lecture 3. Tadeusz Malewski - *From SNP to function, from function to treatment*
12:30 Discussion

Roadshow timetable:

19.09.2016 Vilnius University, Vilnius, Lithuania
20.09.2016 Lithuanian University of Health Science, Kaunas, Lithuania
21.09.2016 Institute of Ecology, Daugavpils University, 13 Vienības, Daugavpils, Latvia
22.09.2016 Estonian Biocentre, Riia 23b, 51010 Tartu, Estonia
23.09.2016 Rigas Stradins University, Riga, Latvia

Charles Cantor

The future of nucleic acids diagnostics



Nucleic acid mass spectrometry (MS) is an ideal tool for the sensitive, quantitative analysis of DNA and RNA samples of intermediate complexity - it fills a niche between whole genome or exome sequencing and conventional real time PCR. MS can examine between 10 and 50 loci simultaneously depending on the sensitivity required in a particular assay. It has high sensitivity for minor alleles and can easily detect these down to a part per thousand in multiplexed assays. Among the most popular and powerful applications of this technology are complex germ line genetic assays, profiling if somatic mutations in solid and liquid cancer biopsies, detection of RNA transcripts from gene fusions or translocations and quantitative analysis of DNA methylation. Particularly in liquid biopsies extreme sensitivity is needed because so few analyte molecules are present- a number of examples of such applications will be presented. Other topics include lecture: germ line genotyping; somatic mutations in cancer biopsies; liquid biopsies. Can stochastic noise be overcome to allow non-invasive pre-symptomatic disease detection?

Tadeusz Malewski

From SNP to function, from function to treatment

Museum and Institute of Zoology, PAS, Warsaw, Poland

iPLEX module deliver plenty information about SNP but what are SNPs function? Comprehensive sites (eg. **Ensembl** www.ensembl.org; **dbSNP** www.ncbi.nlm.nih.gov/SNP) host an organised, collective resource linking out to various tools providing information about gene structure, splice variants, regulatory elements, SNPs effect on protein function, phenotypic effects itd.. Assessing splicing variants is of extreme importance when dealing with eukaryotic genomes, primarily due to their direct relation with candidate gene transcription, and also the acute sensitivity of splicing sites to SNP variations.

Direct mapping of SNPs to a particular disease / pathways is possible by **Gene set Analysis Toolkit** http://bioinfo.vanderbilt.edu/webgestalt/. Uploading list of SNP identifiers researchers gives KEGG_Pathway's with colored genes\proteins sorted by count of genes that each of them contains.

Agena Bioscience offers several panels with carefully chosen SNP set. OncoCarta, LungCarta, MelaCarta and other panels provide a comprehensive family of oncogene and tumor suppressor SNP panels for profiling somatic mutations, enabling cancer researchers to profile genetic changes rapidly for basic research, clinical research, and/or drug development studies. Targeted molecular therapeutics disrupting intracellular signaling pathways are increasingly used for the treatment of cancer. These strategies are based on our increasing understanding of the genes that are causally implicated in cancer and clinical observations that alterations in cancer genomes strongly influence patient response to drugs. Determination of the profiles of somatic genetics in individual patients can be used to direct anticancer therapy for the disease. **My Cancer Genome** www.mycancergenome.org is a personalized cancer medicine knowledge resource for physicians, patients, caregivers and researchers is a one-stop tool that matches tumor mutations to therapies, making information accessible and convenient for clinicians. **Genomics of Drug Sensitivity in Cancer** www.cancerrxgene.org collects data from large-scale drug screen incorporating detailed genomic and gene expression analyses to systematically identify drug response biomarkers. This information can be used to inform the optimal clinical application of cancer drugs, as well as having significant effects on the design, cost and ultimate success of new cancer drug development.

SNPs occurring in genes regulatory regions can change their spaciotemporal expression pattern. **QGE module** (Quantitative Gene Expression) combines competitive PCR with MALDI-TOF mass spectrometry and is the ideal complement to fine mapping studies and solution for post-array validation. This methodology is more sensitive than real-time quantitative PCR and permits very closely related genes to be assayed reliably and quantitatively.

Important issue of regulation of gene expression - DNA methylation can also be analysed on the MassARRAY system. **EpiTYPER** module is a tool for the discovery and quantitative analysis of DNA methylation. Using the speed and accuracy of the MassARRAY system, this method is ideal for discovery of methylation, for discrimination between methylated and non-methylated samples, and for quantifying the methylation levels of DNA. Information presented in the mass spectrum can be used to assess the degree of methylation for each CpG unit independently or to estimate the average methylation for the entire target region. Using this method, both hypermethylation and hypomethylation can be detected in samples. The MassARRAY system is able to detect the methylation level in a mixture as low as 5%.