THIRD INTERNATIONAL
Molecular Diagnostics EUROPE
13-16 APRIL 2015
Marriott Hotel, Lisbon Portugal

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CAMBRIDGE HEALTHTECH INSTITUTE'S THIRD INTERNATIONAL

Molecular Diagnostics EUROPE

13-16 April 2015
MARRIOTT HOTEL | LISBON, PORTUGAL

13-15 APRIL

Second Annual
Prenatal Molecular Diagnostics
TRENDS, ADVANCES & PROSPECTS

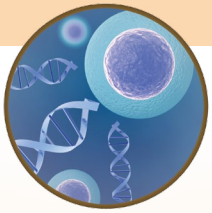
14-15 APRIL

Inaugural
Cancer Sequencing
SOLUTIONS FOR DISTRIBUTION AND ANALYSIS OF NGS DATA

15-16 APRIL

Inaugural
Advanced Diagnostics for Infectious Disease
LATEST TECHNOLOGIES AND IMPACT ON CLINICAL OUTCOME

15-16 APRIL



Inaugural
Circulating Cell-Free Nucleic Acids
PUSHING THE LIMITS OF SENSITIVITY IN CANCER DIAGNOSTICS

KEYNOTE PRESENTERS

Dennis Lo, M.D., Ph.D.
Chinese University of Hong Kong

Herman Goossens, Ph.D.
University Hospital Antwerp

Hans Lehrach, Ph.D.
Max Planck Institute for Molecular Genetics

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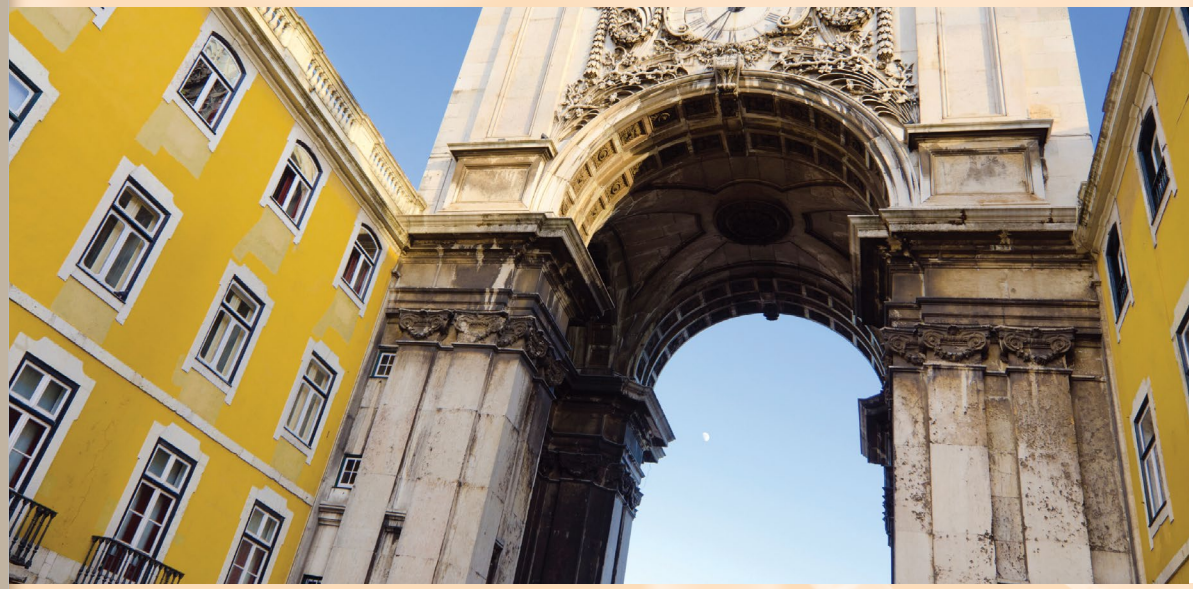
ABOUT THE EVENT

Novel molecular-based tools are rapidly entering the clinic and creating a new paradigm in healthcare. The Third International **Molecular Diagnostics Europe** event will feature four tracks: Prenatal Molecular Diagnostics, Cancer Sequencing, Advanced Diagnostics for Infectious Disease, and Circulating Cell-Free Nucleic Acids. These represent the hottest areas for applying molecular diagnostics to the clinical setting and will lead to greater speed and accuracy of healthcare delivery while paving the way for a new era in medicine.



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HOTEL AND TRAVEL INFORMATION

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Avenida Combatentes, 45
1600-042 Lisbon, Portugal
Phone: +(351) 21 723 54 00

Website:
www.MolecularDxEurope.com

Discounted Room Rate:
€85 single/€95 double,
includes breakfast

Discounted Room Rate Cut-off Date: 4 March 2015

Please visit the hotel and travel page of **www.MolecularDxEurope.com** or call the hotel directly to reserve your sleeping accommodations. You will need to identify yourself as a Cambridge Healthtech Institute conference attendee to receive the discounted room rate with the host hotel. Reservations made after the cut-off date or after the group room block has been filled (whichever comes first) will be accepted on a space- and rate-availability basis. Rooms are limited, so please book early.

We understand that you have many choices when making your travel arrangements. Please understand that reserving your room in the CHI room block at the conference hotel allows you to take full advantage of the conference sessions, events and networking opportunities, and ensures that our staff will be available to help should you have any issues with your accommodations.

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Second Annual

Prenatal Molecular Diagnostics

13-15 April 2015

Trends, Advances & Prospects

MONDAY, 13 APRIL

PRE-CONFERENCE SHORT COURSE*

09:00 – 12:00 Preimplantation Genetic Diagnostics

In Vitro Fertilization (IVF) offers the opportunity for a range of genetic diagnostics to be carried out prior to the selection of fertilized eggs for implantation. In some cases a major reason for choosing the IVF route is because of the risk of serious recessive genetic disorders, which can be detected by targeted screening. Screening and de-selection of eggs with aneuploidies is also a way of improving the success rate of IVF. Experience with arrays and with next-generation sequencing will be discussed, as will recommendations for increasing the occurrence of singleton births.

Instructors:

Joyce Harper, FRCPath, Genetics, Embryology and IVF Group, Institute for Womens Health, University College London, United Kingdom

Jorish Vermeesch, Ph.D., Department of Human Genetics, Catholic University of Leuvan, Belgium

Karsten R. Held, M.D., Medical Director, Reprogenetics Germany GmbH

Francesco Fiorentino, Ph.D., Chief Executive Officer, GENOMA Group, Italy

Tony Gordon, Ph.D., Laboratory Director (UK) and Managing Director (USA), Genesis Genetics

** Separate registration required for short courses.*

12:00 – 13:00 Registration

ANALYSIS OF INVASIVELY-OBTAINED SAMPLES

13:00 Chairperson's Opening Remarks

13:05 Current Status, Issues and Challenges for Molecular Analysis of Invasively-Obtained Samples

Marta Rodriguez de Alba, Ph.D., Genetics, Fundacion Jimenez Diaz, Spain

The uptake of different types of molecular analysis for invasively obtained prenatal samples has varied considerably depending on technology and location. QF-PCR rapidly replaced FISH analysis for rapid aneuploidy diagnosis. The transition to Chromosomal Microarray Analysis (CMA) has been slower, but recently has become much more widely accepted. Differences in the incorporation and reimbursement of CMA across countries in Europe will be presented. Some of the challenges for CMA, and approaches for addressing these challenges, will also be discussed.

13:35 Prenatal Diagnosis Using Software-Targeted Array CGH: Robust, Rapid and Unequivocal

Joo Wook Ahn, Ph.D., Guy's and St. Thomas' NHS Foundation Trust

Prenatal testing of invasively-obtained samples should demonstrate robust performance and rapid turn-around times and should be affordable within a state-funded health service. Known pathogenic CNVs should be captured, and, ideally, equivocal results should be avoided to minimize parental anxiety and delays in reporting. We have implemented a prenatal array CGH service using software-targeting, designed to detect CNVs of greater than 3Mb, and imbalance in regions of known pathogenicity. All our results are therefore unequivocal and can be reported immediately. As new regions of pathogenicity are published, these can be added to the software-targeting, as can any loci relevant to specific ultrasound anomalies. This service started in March 2012; the results of over 700 samples will be presented and discussed.

14:05 Confirmatory Invasive Testing after a Positive NIPT Result: Chorionic Villus Sampling or Amniocentesis?

Diane Van Opstal, Ph.D., Clinical Genetics, Erasmus Medical Center, The Netherlands

Non-invasive prenatal testing (NIPT) for fetal trisomy detection already revealed that there is a small chance of a false positive and false negative result. This is partly due to the fact that the fetal DNA present in the cell-free maternal plasma fraction is derived from the cytotrophoblast of chorionic villi (CV). From cytogenetic studies in CV we know that the cytotrophoblast is not always representative for the fetus due to chromosomal mosaicism. Therefore, a positive NIPT should always be confirmed with invasive testing in order to investigate the fetal karyotype. The fact that NIPT can be performed from the 10th week of gestation on makes CV sampling, routinely applied between 11-14 weeks of gestation, a more suitable technique for confirmation studies than amniocentesis, mostly carried out after 15,5 gestational weeks. Based on our experience with cytogenetic investigations in CV, the choice for CV sampling or amniocentesis will highly depend on the chromosome aberration involved. For trisomy 13, 18 and 21, we can recommend confirmation studies in CV, provided that these studies include the cytogenetic investigation of both the cytotrophoblast (for confirmation of the NIPT result) and the mesenchymal core (for verification of the fetal karyotype). The protocol for other chromosome aberrations will be shown as well.

14:35 Utilization of a SNP Microarray for High-Resolution Prenatal Studies Over 15,000 Samples

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Stuart Schwartz, Ph.D., Strategic Director, Cytogenetics, Laboratory Corporation of America

The findings of over 15,000 prenatal tests utilizing SNP microarray analysis will be reviewed. This data illustrates the utilization not only for ultrasound abnormalities, but also for patients referred for AMA and patients identified with chromosomal anomalies that need better clarification. These studies have not only illustrated the importance of the detection of the gain or loss of chromosomal material, but also

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the importance of copy-neutral loss of heterozygosity (uniparental disomy or identity by descent). It has also demonstrated the usefulness of examining the SNP alleles for detection of maternal cell contamination, twin-twin contamination, as well as mosaicism and whole genome homozygosity.

15:05 Refreshment Break

FETAL CELL ISOLATION AND ANALYSIS

15:50 Advances in the Isolation and Analysis of Trophoblastic Cells for Non-Invasive Prenatal Diagnosis

Patrizia Paterlini-Brechot, Ph.D., Cellular and Molecular Biology, University of Paris Descartes, France

Trophoblastic cells can be isolated non-invasively from blood and from the cervix at very early terms of pregnancy. The DNA of these fetal cells is not mixed with maternal DNA and can be used efficiently for prenatal, early and non-invasive prenatal diagnosis. Results with consistent isolation and recovery of these cells, and subsequent genetic analysis of DNA from them will be presented. The advantages and remaining challenges for this approach will also be discussed.

16:20 Enrichment of Fetal Nucleated Red Blood Cells from Maternal Blood Using Novel Immunomagnetic Technology (CEPir) for Prenatal Diagnosis

Mahmoud Abuelhija, Ph.D., Research & Development, BioCEP Ltd., Israel

BioCEP has implemented a new method for Fetal Nucleated Red Blood Cells enrichment (fNRBC) from maternal blood, obtained at 8-14 weeks gestation. The new procedure is designed as an alternative to invasive prenatal testing, or to cell-free DNA testing. This approach offers lower risk than invasive approaches, while offering better access to fetal DNA than is achieved with cell-free DNA. Challenges and opportunities for commercialization of this approach in the near future will be discussed.

16:50 ARCEDI - a Rapid Method for Isolation of Fetal Cells from Maternal Blood

Steen Kolvraa, M.D., Chief Scientific Officer, Arcedi Biotech ApS, Denmark

A method for isolation of fetal cells, presumably extravillous trophoblasts, will be presented. The method involves initial PFA fixation of full blood followed by immunomagnetic enrichment of fetal cells using endothelial markers, smearing on slides and identification of fetal cells by ectodermal markers. The total method takes three days. In normal pregnancies we find around ten cells on 30 ml maternal blood.

17:20 Sponsored Presentation *(Opportunity Available)*

17:50 Close of Day One

TUESDAY, 14 APRIL

8:00 Registration and Morning Coffee

NIPT

9:00 Chairperson's Remarks

» 9:05 KEYNOTE PRESENTATION:
New Developments in Plasma DNA Sequencing for Non-Invasive Prenatal Testing

Y. M. Dennis Lo, M.D., Ph.D., Chairman, Chemical Pathology, Director, Li Ka Shing Institute of Health Sciences, The Chinese University of Hong Kong, China

The current generation of noninvasive prenatal tests for fetal aneuploidies is based on the counting of DNA molecules in maternal plasma. We have developed a new approach based on the measurement of molecular size, a method that exploits the fact that fetal DNA in maternal plasma is shorter than its maternal counterpart. Other exciting recent developments include the elucidation of the fetal methylome and transcriptome from maternal plasma. Such developments have expanded the spectrum of research and diagnostic applications of noninvasive prenatal testing.

9:35 Challenges Associated with the Implementation of Whole Genome NIPT

Brigitte Faas, Ph.D., Human Genetics, Radboud University, The Netherlands

The implementation of whole genome NIPT goes along with several challenges, either from a political, counseling or technical point-of-view. In this presentation these challenges will be illustrated by examples and results from the NIPT implementation study in the Netherlands and the Radboud University Nijmegen laboratory.

10:05 Implementing Cell-Free DNA Prenatal Testing into Clinical Practice in a Public Health Service

Suzanne Drury, Ph.D., Post-doctoral Scientist, Translational Research, NE Thames Regional Molecular Genetics Service, Great Ormond Street Hospital, NHS Foundation Trust

Implementing prenatal diagnosis based on cell free fetal DNA into public sector maternity care requires evaluation of laboratory performance, clinical utility and validity, health economics as well as patient and health professional opinions. In the UK NIPD for selected monogenic disorders is now part of standard care, and the costs, benefits, uptake and overall impact of NIPT for aneuploidy in the NHS is being assessed.

10:35 Challenges of Developing a Regulated *in vitro* Product for NIPT

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Stephen Little, Ph.D., CEO, Premaitha Health

Understanding the technical hurdles for launching a quality standard prenatal screening test to given consistency, reliability and robustness in a distrubuted environment. Importance of QA, QC, verification and validation to ensure rapid and reliable uptake.

10:50 NIPT Performance in a Large General Population and Comparison Between High-Risk and Low-Risk Pregnancies

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Grover Cong YU, Ph.D., CEO, BGI-Europe & Africa, BGI Diagnostics

NIPT has been widely used to screen for T21, T18 and T13 in the past few years, yet large clinical data has been limited and the validation of NIPT for low-risk mothers is still incomplete. We initiated a multi-center prospective study to collect such data in China. Sensitivity and specificity were calculated with the NIPT positive cases confirmed by karyotyping results, and NIPT negative results confirmed by neonatal tests and other follow-up. The study population was divided into high-risk and low-risk groups based on maternal age, previous Down's screening results, ultrasound findings and pregnancy history. Results from this study and the performance of NIPT in the two groups will be presented and compared.

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11:05 Coffee Break in the Exhibit Hall with Poster Viewing

11:45 Sequencing of Cell-Free DNA as a Marker of Risk for Common Aneuploidies, Pre-Term Labor and Preeclampsia

Leona Poon, MRCOG, M.D., Consultant in Fetal Medicine and Obstetrics, Division of Women's Health, King's College London, United Kingdom

Clinical implementation of cell-free DNA testing can either be as routine general screening or as contingent screening, based on other results, such as the first trimester combined test. A comparison of these two approaches, and considerations for interpretation of cell-free DNA results, will be presented. We have also examined the potential for making use of cell-free DNA screening to detect increased risk of preeclampsia or spontaneous preterm labor. Early results and challenges with both of these testing approaches will also be discussed.

12:15 Comparison of Genetic Signatures to Identify Prognostic Markers for the Risk of Pre-Term Birth

Joe Vockley, Ph.D., Chief Scientific Officer, Inova Translational Medical Institute, Inova Fairfax Medical Center, Pediatrics, Virginia Commonwealth University School of Medicine, United States

The Inova Translational Medicine Institute has utilized whole genome sequences and other genomic data from thousands of families to develop a model for predicting the risk of pre-term birth. Data are generated from mother/father/baby trios from a pre-term cohort and a full-term cohort to include families from over 100 countries. RNA markers were found in the peripheral blood of mothers that correlate to pre-term birth but not to common causes of pre-term birth such as premature rupture of membranes and pre-eclampsia. These markers may be critical to the development of a tool for the prediction of preterm birth.

12:45 Luncheon Presentation: Advances in Next-Generation Sequencing for Non-Invasive Prenatal Testing

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Alex Helm, MBA, Senior Product Manager, Illumina, Inc.

13:15 Session Break

14:15 Chairperson's Remarks

14:20 Recovering High Fetal Fractions during Sample Preparation for NIPT: The Potential Role of Point-of-Care Technologies

Maiwenn Kersandy-Kerhoas, Ph.D., School of Engineering and Physical Sciences, Hariot Watt University, United Kingdom

Low fetal fraction of cell-free circulating DNA in the maternal circulation is the primary limitation met by most current prenatal molecular diagnostic technologies and results in sample rejection or lower test reliability. To circumvent this limitation and preserve high fetal fraction, strict sample collection and preparation guidelines have been advocated by clinicians and analysts. Commercial cell-preservation solutions have entered the market, however this area may benefit from a paradigm shift and high fetal fraction preservation and enrichment may lie in on-site sample preparation technologies. A comparative study revealing a 4-fold enrichment of fetal fractions after on-chip maternal blood plasma extraction compared to conventional centrifugation will be presented.

14:50 Clinical Experience Using Targeted Microarrays for NIPT

Tuba Gunel, Ph.D., Molecular Biology and Genetics, Istanbul University, Turkey

Analysis of maternal blood plasma cell-free fetal (cff) DNA using array-CGH allows for detection of whole chromosome differences between test and reference DNA. Such arrays are commonly used to analyze amniotic samples and pre-implantation embryos. We hypothesized that extraction, fluorescent labeling, hybridization, and analysis of cffDNA could be used to simultaneously screen for aneuploidy across every chromosome. This technology may also aid the identification of minor genetic aberrations, such as micro-deletions and micro-duplications, which could enhance prenatal genetic diagnostics. NIPT using microarrays provided faster and more accurate cell-free DNA (cfDNA) measurements than sequencing.

15:20 ANGELAB, Developing Lab-on-a-Chip-Based Systems for Epigenetic Non-Invasive Prenatal Diagnosis

Jesus M. Ruano-Lopez, Ph.D., IK4-Ikerian, Spain

The European Project called ANGELAB, which is developing a family of *In Vitro* Diagnostic Systems, will be presented. Each LabonaChip runs fetal DNA sample preparation in combination with real-time or Digital PCR. The methodology and system architecture used to transfer epigenetic biological protocols into modules, then into LabonaChip, and then into integrated systems will be described. The strategy, challenges and progress made over the first two years of this project will also be covered.

15:50 Refreshment Break in the Exhibit Hall with Poster Viewing

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16:30 PANEL DISCUSSION: NIPT Providers

How are the different providers of next-gen sequencing NIPT differentiating themselves, including the coverage of results for micro-deletions and other non-aneuploidy disorders? How do some of the newer approaches, based on arrays or being developed as kits to allow on-site analysis, compare with the more established approach? Will the emphasis continue to be on testing for higher-risk pregnancies, or will NIPT likely be extended and offered to all pregnancies?

Moderator: *Phillips Kuhl, President, Cambridge Healthtech Institute, United States*

Panelists: *Maximilian Schmid, MD, Associate Director, Medical Affairs, Ariosa Diagnostics, Inc.*

Michael Lutz, Ph.D., Chief Executive Officer, LifeCodexx AG, Germany

Solomon Moshkevich, Vice President, Marketing and Medical Education, Natera, United States

John Anson, Ph.D., Executive Vice President, Research & Development, Oxford Gene Technology, United Kingdom

Stephen Little, Chief Executive Officer, Premaitha Health, United Kingdom

Daniel Grosu, MD, Vice President, Chief Medical Officer, Sequenom Inc.

Alex Helm, Product Manager, Illumina Inc., United States

18:00 Welcome Reception in the Exhibit Hall with Poster Viewing

19:00 Close of Day Two

WEDNESDAY, 15 APRIL

8:00 Registration and Morning Coffee

NIPT

8:40 Chairperson's Remarks

8:45 NIPD for Monogenic Disorders without Using Next-Generation Sequencing

Jessica van den Oever, MSc, Department of Clinical Genetics, Laboratory for Diagnostic Genome Analysis (LDGA); Department of Clinical Genetics, Leiden University Medical Center

Next-generation sequencing (NGS) is the main method used for non-invasive prenatal testing and diagnosis. However this method is still quite costly and may not be accessible to all laboratories. Moreover, although the approach is very successful for detection of fetal aneuploidies it is also has its limitations. Noninvasive detection of fetal repeat expansions or paternally inherited mutations in certain regions of the genome is currently difficult, if not impossible, by using NGS. A sensitive, fast and low cost alternative for NGS-based targeted mutation scanning will be presented, as well as a validation study for detection of polymorphic paternally inherited CAG repeats for fetuses at risk of Huntington Disease.

9:15 Development of Quantitative PCR for NIPD of Single Gene Disorders

Claire Guissart, Ph.D., Laboratory of Genetic Medicine, University of Montpellier, France

In recent years, non-invasive prenatal diagnosis (NIPD) has found new applications in monogenic disease diagnostics for paternally inherited mutations. This presentation discusses the application of a qPCR-based mutant enrichment technique for NIPD of single gene disorder using Cystic Fibrosis as a clinical application model. In this respect, we have also repurposed a commercial mini-STR kit — frequently used in forensic STR analysis — as an external quality control to confirm the presence of cell-free fetal DNA.

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9:45 Experience with over 5000 NIPT Samples: Overcoming Technical and Biological Challenges

Joris R. Vermeesch, Ph.D., Professor, Center for Human Genetics, UZ Leuven / KU Leuven

NIPT for fetal aneuploidy detection is increasingly being offered in the clinical setting. We present an analysis pipeline that enables the differentiation of fetal trisomies from local maternal CNVs and the detection of non-classical aneuploidies as well as segmental imbalances. We present the results from its clinical application on over 4000 prospective pregnancies. Interestingly, our analysis pipeline also identified a pregnant woman with early-stage nodular sclerosis Hodgkin lymphoma.

10:15 Coffee Break in the Exhibit Hall with Poster Viewing

10:45 The Diagnostic Accuracy of Non-Invasive Fetal RHD Typing

Florentine Thurik, MD, Experimental Immunohematology, Sanquin Research

In the Netherlands non-invasive fetal RHD screening was implemented in July 2011. The evaluation of this screening program shows the high diagnostic accuracy of this approach; only 9 false negative results on > 26.000 serologically confirmed tests. Since the fetal RHD screening is based on a quantitative RHD PCR, we now have access to a huge data set on quantitative fetal DNA concentrations, showing the large biological variability. The causes of false positivity and negativity will be discussed, and the lessons to be learned from this analysis for other non-invasive tests.

11:15 PANEL DISCUSSION: Predicting the Landscape for Prenatal Molecular Diagnostics in Europe

The current landscape for prenatal molecular diagnostics varies across Europe from one country to another. How might changes such as array-based testing have an impact on invasively-obtained and possibly non-invasively obtained samples? How soon might the isolation of fetal cells from maternal blood become commercially viable, and what would the implications of this shift be for NIPT? Are kit-based tests for NIPT more likely to have an impact on NIPT in the short run? What other factors are likely to be important for changes in the clinical implementation of prenatal molecular diagnostics?

Panelists:

Marta Rodriguez de Alba, Ph.D., Genetics, Fundacion Jimenez Diaz, Spain

Patrizia Paterlini-Brechot, Ph.D., Cellular and Molecular Biology, University of Paris Descartes, France

Brigitte Faas, Ph.D., Human Genetics, Radboud University, The Netherlands

11:45 Close of Conference

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Inaugural

14-15 April 2015

Cancer Sequencing

Solutions for Distribution and Analysis for NGS Data

TUESDAY, 14 APRIL

8:00 Registration and Morning Coffee

9:00 Chairperson's Remarks

Andrew M. Hudson, MBChB BSc (Hons), MRCP, FRCR, Clinical Research Fellow, Cancer Research UK Manchester Institute, United Kingdom

» 9:05 KEYNOTE PRESENTATION: From NGS to a Truly Personalised Therapy

Hans Lehrach, Ph.D., Professor, Director Emeritus, Max Planck Institute for Molecular Genetics, Germany

We are in the middle of a revolution in Biology and Medicine, triggered primarily by the enormous increase in sequencing power (and concomitant decrease in sequencing cost) due to next generation sequencing techniques. While it took us in an international consortium more than ten years and between 1 and 3 billion US-Dollars to sequence the first human genome, we are now embarking on projects to sequence the genome of every cancer patients, as well as the genome and transcriptome of their tumors. To be able to use this flood of data we can generate on every patient to provide every patient with individually optimised medical care, we can use these data to model the patient and its tumour as interacting molecular models, which can be virtually 'treated' with 'virtual drugs' to be able to predict the effect and the side effects of ever possible therapy on the individual, to develop individually optimised therapies for every oncology patient. The development of such computer models of individual patients does however also offer new, revolutionary possibilities for virtualising drug development, increasing the number of drugs reaching the market, accelerating development, cutting costs, and cutting risk in the drug development, in turn, increasing the treatment options for patients.

SEQUENCING DATA INTERPRETATION

9:35 Drivers, Passengers, and Biomarkers via Network Enrichment Analysis of Tumor Molecular Profiles

Andrey Alexeyenko, Ph.D., Research Scientist, Science for Life Laboratory, BILS, Karolinska Institute, SciLifeLab, Sweden

The characterization of molecular landscapes benefits from elevating analyses at the pathway level, i.e., quantifying alterations in functional modules (pathways) rather than those of individual genes. Our novel methods detect activation of such modules in a robust and unbiased way. Molecular features of cancer tumors are functionally interpreted via their network positions. This enables distinguishing driver mutations and discovering network-anchored biomarkers for specific phenotypes.

10:05 Inflammation Promotes Liver Cancer through Genomic Modifications Different from those Determined by Other Etiological Factors

Fabio Iannelli, Ph.D., Postdoc, IFOM Foundation – The FIRC Institute of Molecular Oncology Foundation, Italy

This presentation will illustrate how we used acquired genomic alterations, namely somatic point mutations and copy number variants (CNVs), to show that genetic heterogeneity of liver cancers correlates with their distinct pathogenesis and leads to different mechanisms of tumor progression. It will also provide evidence that solid tumors are not necessarily associated with mutational instability. Other key points like current challenges in CNV detection from exome sequencing data will be discussed.

10:35 Enriching Nucleic Acids for Next-Generation Sequencing Analyses of SNPs, CNVs, Gene Fusions and More

Sponsored by NuGEN imagine more from less

Joe Don Heath, Ph.D., Vice President, Market Development-Diagnostics, NuGEN Technologies, Inc.

The novel Single Primer Enrichment Technology (SPET) and how it differs from existing target enrichment methods will be described. Sensitive variant detection from genomic DNA derived from fresh and FFPE tissues using 344 cancer-related genes will be demonstrated as well as utilization of SPET as a rapid, cost-effective screening tool for discovery of novel fusions and detection of known fusions with a panel of 500 cancer genes implicated in fusions events.

11:05 Coffee Break in the Exhibit Hall with Poster Viewing

11:45 Complete Pipeline for Clinical Reporting Of BRCA1/2 Massive Parallel Sequencing

Ettore Capoluongo, Ph.D., Professor, Clinical Biochemistry & Clinical Molecular Biology, Laboratory of Clinical Molecular Diagnostics, Laboratory Medicine, Policlinico Universitario "A. Gemelli", Cattolica Del Sacro Cuore, Italy

12:15 Sponsored Presentation *(Opportunity Available)*

12:45 Luncheon Presentation: ACCUSEQ: Innovative Tools for Precision Cancer Diagnostics

Sponsored by SeraCare LIFE SCIENCES

Trevor W. Brown, Vice President, Precision Medicine, SeraCare

The promise of precision medicine depends upon accurate, precise and dependable use of next generation diagnostic techologies such as NGS and ddPCR. Adequate tools for assay development, validation and monitoring simply do not readily exist. SeraCare is developing new tools to help laboratories robustly characterize NGS assays and their analysis pipelines and to monitor routine performance, both within laboratories and between laboratories.

13:15 Session Break

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Molecular Diagnostics EUROPE
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14:15 Chairperson's Remarks

Daniëlle A.M. Heideman, Ph.D., Clinical Scientist; Molecular Pathology; Associate Professor, Pathology, VU University Medical Center, The Netherlands

14:20 Performance of Amplicon-Based Next-Generation DNA Sequencing for Diagnostic Gene Mutation Profiling in Oncopathology

Daniëlle A.M. Heideman, Ph.D., Clinical Scientist; Molecular Pathology; Associate Professor, Pathology, VU University Medical Center, The Netherlands

Next-generation sequencing (NGS) is an important technological advance which has helped in the identification of genetic determinants of cancer and the discovery of new diagnostic, prognostic and therapeutic biomarkers. NGS is now maturing to the point where it is being considered by many pathology laboratories for routine diagnostic use. I will present a comprehensive evaluation of amplicon-based NGS for diagnostic mutation profiling in oncopathology, with particular focus on FFPE specimens.

SEQUENCING DATA INTEGRATION

14:50 DNA Methylation in Human Variation, Disease Risk and Cancer

Holger Heyn, Ph.D., Researcher, Cancer Epigenetics and Biology Program (PEBC), Bellvitge Institute for Biomedical Research (IDIBELL), Spain

DNA methylation patterns are important to establish phenotypes, but little is known about their contribution to human variation and disease risk. We identified distinctly methylated genes with impact on susceptibility to certain diseases. DNA methylation differences could be traced back to genetic variation and we propose that interrogating the interplay between genetic and epigenetic code is providing new information about the biology of diseases.

15:20 Bioinformatics for Precision Medicine

Philippe Hupé, Ph.D., Deputy Director, Bioinformatics, Institut Curie

A seamless information system developed at Institut Curie which facilitates the data integration of genomics and clinical and tracks in real-time the processing of individual samples will be presented.

15:50 Refreshment Break in the Exhibit Hall with Poster Viewing

CONSIDERATIONS FOR CLINICAL IMPLEMENTATION

16:25 Chairperson's Remarks

16:30 Whole Genome Sequencing from FFPE-Derived DNA Samples

Joanne Mason, Ph.D., Lead Scientist, BRC/NHS Oxford Molecular Diagnostics Centre, Oxford University Hospitals, United Kingdom

The easy availability of Formalin-Fixed Paraffin Embedded (FFPE) tissue makes it an ideal resource for molecular diagnostics. However, despite its stability and ability to preserve morphological information it is challenging to get high quality genetic material suitable for next-generation sequencing from FFPE samples. I will discuss how FFPE processing and extraction can be optimized to enable FFPE-derived DNA to be used for molecular diagnostics including for whole genome sequencing.

17:00 Evolution of Colorectal Cancer inferred from Deep-Sequencing Data

Francesca Ciccarelli, Ph.D., Associate Professor, Division of Cancer Studies, King's College London, United Kingdom

In my talk I will present our method to rebuild the evolutionary history of cancer clones based on DNA deep sequencing data. I will then show its application to the reconstruction of the history of a set of colorectal cancers. Finally, I will highlight how this analysis can help in interpreting the biology of the single lesion and to help in the choice of the therapeutic intervention.

17:30 Discrepancies in Cancer Genomic Sequencing Highlight Opportunities for Driver Mutation Discovery

Andrew M. Hudson, MBChB BSc (Hons), MRCP, FRCR, Clinical Research Fellow, Cancer Research UK Manchester Institute, United Kingdom

Comparing genomics data from CCLE and COSMIC revealed marked discrepancies in the detection of missense mutations in identical cell lines (57.38% conformity). Reasons for this discrepancy include difficult sequencing of GC-rich regions (sequencing cold-spots), passaging effects and variation of dbSNP filtering. These data highlight that significant opportunities in discovering oncogenic mutations exist by optimizing the sequencing of GC-rich regions

18:00 Welcome Reception in the Exhibit Hall with Poster Viewing

19:00 Close of Day One

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WEDNESDAY, 15 APRIL

8:00 Registration and Morning Coffee

CONSIDERATIONS FOR CLINICAL IMPLEMENTATION (cont'd)

8:40 Chairperson's Remarks
Kristin Pothier, Partner, Ernst & Young LLP, United States

8:45 Characterization of Pathways Involved in Colorectal Cancer Resistance to Neoadjuvant Chemoradiotherapy
Fernanda Koyama, Ph.D., Research Scientist, Molecular Oncology Center, Ludwig Institute for Cancer Research at Hospital Sirio-Libanes, Brazil
A number of works trying to classify patients have failed when approaching differential gene expression data as the only source of biological information. As the gene signatures rely on the sample set, there is an increasing need for a biological understanding to produce a signature in terms of pathways and biological processes. In this regard we promote a change in the way we look for sequencing data, specifically RNA-Seq, in an attempt to identify rectal cancer patients that are refractory to neoadjuvance treatment by identifying biological pathways involved in tumor resistance.

9:15 A SNP-Based Massively-Multiplexed PCR Approach for Detection of SNAs and CNAs in Plasma, Tumor Tissues, and Single Cells
Sponsored by natera
Matthew Hill, Ph.D., Vice President, Research and Development, Natera Inc.
Sensitive detection of large numbers of single nucleotide and copy number alterations (SNAs and CNAs) in cancer tissues and plasma has proven challenging. Leveraging methods proven in noninvasive prental testing, we demonstrate the ability to detect multiple SNAs with sensitivities to 0.01% and CNAs to 0.45% on multiple chromosome segments. In addition to plasma, this technology can be applied to fresh-frozen and FFPE tissue, as well as single cells.

9:45 Global Innovation and Marketing in NGS
Kristin Pothier, Partner, Ernst & Young LLP, United States
As NGS transitions from the research bench to the clinic, NGS companies will need to develop new marketing strategies while pharmaceutical companies will need to proactively develop partnerships in order to fully maximize the potential of personalized medicine. Lastly, established and emerging companies will need to innovate rapidly as next-next generation sequencing comes to fruition.

10:15 Coffee Break in the Exhibit Hall with Poster Viewing

10:45 PANEL DISCUSSION:
Global Innovation and Marketing in NGS
Moderator: *Kristin Pothier, Partner, Ernst & Young LLP, United States*
The NGS space is evolving rapidly, with new tests and new technologies coming online at an ever-accelerating pace. New, more expansive partnerships with drug companies are being inked and, according to Richard Klausner, Chief Medical Officer of Illumina, we are moving out of the era of companion diagnostics and into the era of companion therapeutics.
Panelists:
Sarah T. Bobulsky, Vice President, Life Sciences, Parthenon-EY
Jamie L. Platt, Ph.D., Vice President, Genomic Solutions, Geneuity (an MPLN company)

11:45 Close of Conference

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Advanced Diagnostics for Infectious Disease

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Latest Technologies and Impact on Clinical Outcome

WEDNESDAY, 15 APRIL

OVERVIEW SESSION

13:00 Chairperson's Opening Remarks

Gyorgy Abel, M.D., Ph.D., Director, Molecular Diagnostics, Immunology & Clinical Chemistry, Laboratory Medicine, Lahey Hospital & Medical Center, United States

» 13:05 KEYNOTE PRESENTATION: A Paradigm Shift of Diagnostic Medical Microbiology: Will it Finally Happen?

Herman Goossens, Ph.D., Professor, Medical Microbiology, University Hospital Antwerp, The Netherlands

A more personalized approach identifying those patients who really need treatment is needed. Unfortunately, many of the currently available diagnostic methods are too slow. We have seen a technological revolution with the development of numerous complex diagnostic assays. These systems can decrease the time required for detection of biomolecules, like proteins and nucleic acids, from a few hours to a few minutes and should greatly improve medical diagnostics. However most of these technologies have not yet permeated clinical diagnostic laboratories. Is the paradigm shift likely to happen?

13:35 New European IVD Regulations – Update and Analysis

David E. Barton, Ph.D., Chief Scientist, National Centre for Medical Genetics, Ireland

The IVD Directive regulates the market for diagnostic tests within the EU. The current system is inflexible, unresponsive and does not do a good job of protecting patients. New regulations are being developed, which will radically alter the landscape for IVDs in Europe. The regulations will have major implications for IVD manufacturers and for laboratory-developed tests. This presentation will bring the audience up to date on the latest developments.

14:05 The ESGMD/ESCMID Roadmap of Bringing New Technologies into the Clinic

Eric C.J. Claas, Ph.D., Associate Professor, Molecular Medical Microbiologist, Medical Microbiology, Leiden University Medical Center, The Netherlands

14:35 Solid Phase Multiplexed Electrochemical Quantitative Isothermal Amplification

Ciara K. O'Sullivan, Ph.D., ICREA Research Professor, Chemical Engineering, Universitat Rovira i Virgili, Spain

An overview of different approaches for electrochemical quantitative isothermal amplification will be provided. The use of post-amplification detection vs. real-time detection will be compared using a model system of Francisella tularensis. The optimised parameters applied to multiplexed detection of bioterrorist agents will then be outlined and future potential discussed.

15:05 Refreshment Break in the Exhibit Hall with Poster Viewing

INTEGRATION AND ADOPTION

15:45 Challenges of Integrating Functionalities in Diagnostic Devices for the Detection and Management of Infectious Diseases

Jesús M. Ruano-López, Ph.D., International Project Manager, Microsystems, IK4-Ikerlan and CIC microGUNE, Spain

This talk will describe barriers and future opportunities of *In Vitro* Diagnostic (IVD) devices from the microfluidic innovation point of view in the field of infectious diseases. The microfluidic community is also facing opportunities: an incipient technological maturity in microfluidic strategies for infectious diseases, the impact of the new IVD regulation on Laboratory Developed Tests (LDT), and the niche market left between liquid handling robotic systems and the point-of-care solutions. This presentation will unfold these IVD hurdles and chances.

16:15 Evaluation of Novel Commercial Assays for Molecular Diagnosis of Infectious Disease

Peter Muir, Ph.D., FRCPath, Consultant Clinical Scientist, Specialist Virology Centre, Public Health, United Kingdom

Prior to adoption into clinical service, it is important to confirm that new commercial assays are fit for purpose. This presentation will describe some case studies in performance evaluation, which aims to demonstrate adequate analytical performance, and clinical evaluation, which seeks to determine if a new assay can deliver a significant clinical or economic benefit when compared to existing practice.

16:45 Networking Reception in the Exhibit Hall with Poster Viewing

17:45 Close of Day

THURSDAY, 16 APRIL

8:00 Registration

8:30 Breakfast Presentation *(Sponsorship Opportunity Available)* **or Morning Coffee**

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POINT-OF-CARE TESTING IN INFECTIOUS DISEASES

9:00 Chairperson's Remarks
Till T. Bachmann, Ph.D., University of Edinburgh, United Kingdom

9:05 Molecular Diagnostics of Antimicrobial Resistance at Point-of-Care
Till T. Bachmann, Ph.D., Reader, Personalised Medicine in Infectious Disease; Deputy Head, Division of Infection and Pathway Medicine, College of Medicine and Veterinary Medicine, University of Edinburgh, United Kingdom
Antimicrobial resistance has become a threat of global dimension for healthcare systems and societies as it substantially diminishes our chances to fight bacterial infections with the current portfolio of drugs. Rapid diagnostics and especially Point-of-Care Testing can help tackling this problem by giving healthcare professionals the needed information to choose the best therapeutic interventions where they most need them. Ultimately, the aim is to tackle antimicrobial resistance by personalised approaches to administer the right treatments at the right time to the right patients as well as facilitating new drug developments using companion diagnostics for antibiotic therapies.

9:35 POC Molecular Testing in Resource-Limited Settings: The Landscape, The Opportunities, The Challenges
Maurine Murtagh, CEO, International Diagnostics Center, London School of Hygiene & Tropical Medicine
Access to diagnostic testing in resource-limited settings is often limited, especially outside of urban centers. Centralized testing has been a limiting factor. As a result, there is great interest in implementing molecular tests that can be used at or near the point of patient care, and there is a robust pipeline for such tests. However, implementation of POC testing platforms in resource-limited settings can be difficult. The challenges must be overcome in order to maximize the potential of POC molecular testing.

10:05 Improving Infectious Disease Diagnostics: A Simple Biochip-Based Approach for Rapid, Targeted Multiplex Detection
Sponsored by RANDOX MOLECULAR
Scott McKeown, Ph.D., Product Manager, Molecular, Randox Biosciences
Randox has developed multiplex microbiology panels for the detection of common infections from a single patient specimen. The technology has positive implications for the patient treatment pathway; appropriating antibiotic use and improving use of resources within the healthcare system.

10:20 Current Requirements in the World of Point of Care Applications: A Short Overview
Sponsored by QIAGEN
Roberto Spricigo, Ph.D., OEM Manager, QIAGEN Lake Constance
Healthcare professionals are looking for faster patient evaluation and efficient diagnosis. Rapid point-of-care tests are therefore becoming increasingly important as they provide precise quantitative results. The presentation will cover the requirements for POC devices for lateral flow and isothermal amplification applications.

10:35 Coffee Break in the Exhibit Hall with Poster Viewing

11:15 Global Perspective on Clinical Utility and Cost-Effectiveness of New Technologies
Gyorgy Abel, M.D., Ph.D., Director, Molecular Diagnostics, Immunology & Clinical Chemistry, Laboratory Medicine, Lahey Hospital & Medical Center, United States
The main drivers of point of care testing (POCT) are clinical demand and utility, and technological advances in molecular chemistry, microfabrication, miniaturization, microfluidics, nanochips, biosensors, lateral flow, paper-based devices, and wireless communication. POCT in major hospitals is resource intensive and expensive, which may be offset by savings due to better outcomes and lower cost of overall care. Advancements in POCT are most spectacular in the fight against infectious diseases, both in industrialized nations and resource-poor developing countries. The presentation highlights the major global technological and utilization trends in infectious disease POCT, and the related costs and clinical benefit.

11:45 Point-of-Care Microdevices for Virus Analysis
Siyang Zheng, Ph.D., Assistant Professor, Biomedical Engineering, The Pennsylvania State University, United States
Point-of-care microdevices can provide sensitive, accurate, rapid and low-cost virus analysis for a large population. As a useful platform, it must not only pursue single performance characteristics, but also excel at multiple performance parameters. Throughout the past decades, tremendous progress has been made in point-of-care biomedical microdevices. The focus of the talk will be on lab-on-a-chip systems for viral infectious diseases.

12:15 Session Break

12:45 Luncheon Presentation *(Sponsorship Opportunity Available)* **or Enjoy Lunch on Your Own**

13:15 Session Break

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CLINICAL SEQUENCING OF PATHOGENS

14:15 Chairperson's Remarks

14:20 The Diagnosis of Infectious Diseases by Whole Genome Next-Generation Sequencing

Marc Eloit, Ph.D., Chairman and CSO, PathoQuest; Head, Laboratory for Pathogen Discovery, Institut Pasteur, France

The availability of next-generation sequencing (NGS) techniques is about to revolutionize diagnostics of infectious diseases by using a single test, based on microbe whole-genome NGS (WG-NGS) , which takes advantage of improvement in sample preparation, availability of sequencers and optimized pipeline for taxonomic assignation. The presentation will show power and pitfalls of WG-NGS, and will illustrate its use in routine diagnosis.

14:50 Infectious Disease Applications of Next-Generation Sequencing

Vanya Gant, Ph.D., FRCP, FRCPath, Divisional Clinical Director, Infection, Microbiology, UCLH NHS Foundation Trust, United Kingdom

There remains a significant gap between what is possible with NGS for detection of infectious diseases, and what is implemented at the front line of clinical service. This talk will explore some reasons why this might be, and offer some possible solutions.

15:20 Full Genome Virus Deep-Sequencing for Detection and Monitoring Transmission

Matthew Cotten, Ph.D., Senior Staff Scientist, Virus Genomics, Wellcome Trust Sanger Institute, United Kingdom

The presentation will cover topics such as: the methods of deep sequencing of RNA viruses, virus discovery methods and the utility of virus full genome data for diagnostics, for transmission studies and for monitoring zoonotic risks. Examples will be provided from the work of our group on MERS coronavirus, rotavirus, norovirus and RSV transmission.

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15:50 Designing Point of Care Instruments: Challenges and Proven Tools for Success

David James, Senior Vice President, Diagnostics, Invetech

Developing market-leading instruments and consumables to meet the demanding requirements of Point of Care infectious disease markets requires specialist skills and experience. Product development stakeholders with competing priorities are often challenged to find a method to agree on the "critical few" requirements that will provide the best product solutions. In this session you will learn about valuable tools and processes that are critical to first defining a winning design and them delivering a robust and competitive final product in the shortest possible time to market.

16:20 Refreshment Break in the Exhibit Hall with Poster Viewing

MASS SPEC FOR INFECTIOUS DISEASE DETECTION

17:00 Technical Features of MALDI-TOF Mass Spectrometry Systems for the Identification of Bacterial, Mycobacterial and Fungal Pathogens

Christine C. Ginocchio, Ph.D., MT (ASCP), Professor of Medicine, Hofstra North Shore-LIJ School of Medicine; Vice President, Global Microbiology Affairs, bioMérieux, United States

Technical features and performance characteristics of the bioMérieux VITEK MS and Bruker MALDI BioTyper CA Systems for detection of bacteria, yeast and mycobacteria will be presented. Database development, methods for spectral analysis, sample isolate preparation and results interpretation will be explained. Direct specimen testing from positive blood cultures will be reviewed. Impact of rapid results on laboratory workflow and clinical decision making will be discussed.

17:30 The RADICAL Trial: How the Iridica PCR/MS Device Performs in Critical Care

David Brealey, Ph.D., MRCP, FRCA, FFICM, Consultant in Intensive Care Medicine, University College London Hospitals NHS Foundation Trust, United Kingdom

The management of sepsis has changed little over the decades; multiple potential therapies have come and gone. Management still relies on rapid identification of the pathogen and initiation of appropriate antibiotics. Unfortunately current techniques for bacterial detection are slow and frequently negative. The newer molecular techniques offer us an opportunity to change all this. The RADICAL study assessed a new PCR + Electrospray ionization mass spectrometry device against standard hospital culture techniques in a critically ill population. We discuss the trial and whether the results are applicable.

18:00 Close of Conference

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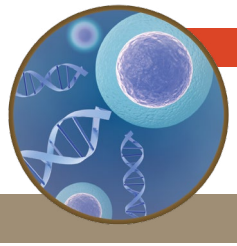

Inaugural

Circulating Cell-Free Nucleic Acids

15-16 April 2015

Pushing the Limits of Sensitivity in Cancer Diagnostics

WEDNESDAY, 15 APRIL

CIRCULATING TUMOR DNA (ctDNA) IN LIQUID BIOPSY

13:00 Chairperson's Opening Remarks

Catherine Alix- Panabières, Ph.D., Director, Laboratory Rare Human Circulating Cells, Cell and Tissue Biopathology of Tumors, University Medical Center of Montpellier, France

» 13:05 KEYNOTE PRESENTATION: GENOME-WIDE PLASMA DNA SEQUENCING FOR CANCER DETECTION

Y. M. Dennis Lo, M.D.,Ph.D., Chairman, Chemical Pathology, Director, Li Ka Shing Institute of Health Sciences, The Chinese University of Hong Kong, China

We have explored the use of genomewide plasma DNA sequencing for cancer detection. We have shown that tumor-derived copy number aberrations, single nucleotide variations and methylation changes can be detected using this approach. This approach can be used to explore tumoral heterogeneity using plasma nucleic acids. This method can be used for multiple tumor types and has potential clinical applications for cancer detection and monitoring.

13:35 Real-Time Liquid Biopsy in Cancer Patients: Circulating Tumor Cells vs. Circulating Tumor DNA

Klaus Pantel, M.D., Professor and Founding Director, Institute of Tumor Biology, University Medical Center Hamburg-Eppendorf, University of Hamburg, Germany

The analysis of therapeutic targets and drug resistance–conferring gene mutations on circulating tumor cells (CTCs) and cell-free circulating tumor DNA (ctDNA) released into the peripheral blood is now feasible. Both CTCs and ctDNA provide complementary information on assessing new drugs or drug combinations. The liquid biopsy concept will contribute to a better understanding and clinical management of drug resistance in patients with cancer.

14:05 Detection and Characterization of Viable Circulating Tumor Cells as Liquid Biopsy for Cancer

Catherine Alix- Panabières, Ph.D., Director, Laboratory Rare Human Circulating Cells, Cell and Tissue Biopathology of Tumors, University Medical Center of Montpellier, France

The enumeration and characterization of circulating tumor cells (CTCs) in the peripheral blood may provide important prognostic information and might help to monitor efficacy of therapy. Since current assays cannot distinguish between apoptotic and viable CTCs, it is now possible to apply the fluoroEPISPOT assay that detects proteins secreted/released/shed from single epithelial cancer cells. We applied this technology in breast, colon, prostate, head & neck and ovarian cancer as well in melanoma.

14:35 Circulating Tumor DNA for Noninvasive Cancer Diagnostics

Dana W.Y. Tsui, Ph.D., Postdoctoral Research Fellow, Cancer Research UK Cambridge Institute, United Kingdom

This talk will discuss recent advances in the use of circulating tumor DNA for non-invasive study of cancer genomics, with particular focus on non-small cell lung cancer. Clinical cases will be presented to demonstrate its potential clinical utility for molecular stratification, monitoring disease progression and detecting acquire resistance to therapies.

15:05 Refreshment Break in the Exhibit Hall with Poster Viewing

15:45 Blood-Based Genotyping of Colorectal Cancer Patients

Giulia Siravegna, Oncology, University of Torino; IRCC-Candiolo Cancer Institute, Italy

Liquid biopsy and cfDNA analysis allow genotyping of colorectal cancer (CRC) patients using a blood sample. CRC patients represent a model to assess whether blood analyses could in principle be used to perform diagnosis, to guide clinical decisions and to monitor the efficacy of therapies, establishing proof of principle that genotyping of cancer alleles in the patients' blood allows clinically valuable longitudinal assessment for patients.

16:15 ctDNA in Triple Negative Breast Cancer Patients in a Targeted Therapy Multicentric Clinical Trial

Jean-Yves Pierga, Ph.D., Professor, Medical Oncology, Circulating Biomarkers Lab, Institut Curie & Université Paris Descartes, France

16:45 Networking Reception in the Exhibit Hall with Poster Viewing

17:45 Close of Day

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THURSDAY, 16 APRIL

8:00 Registration

8:30 Breakfast Presentation *(Sponsorship Opportunity Available)* **or Morning Coffee**

CIRCULATING TUMOR DNA FOR PATIENT MANAGEMENT

9:00 Chairperson's Remarks

Klaus Pantel, M.D., Professor and Founding Director, Institute of Tumor Biology, University Medical Center Hamburg-Eppendorf, University of Hamburg, Germany

9:05 Circulating Cell-Free DNA as a Strong Multimarker Diagnostic, Theranostic and Prognostic Tool for Metastatic Colorectal Cancer Patients Management Care

Alain R. Thierry, Ph.D., Senior Investigator, Institute of Research on Oncology of Montpellier, INSERM, France

Circulating cell-free DNA (ccfDNA) appears as a liquid biopsy and a breakthrough tool in cancer diagnostics. We designed a test enabling multimarker ccfDNA analysis. We recently presented first clinical validation of the analysis of circulating DNA in oncology. Potential of examining qualitative (mutational status) and quantitative (concentration of ccfDNA various species) is investigated in regards to diagnostics, prognosis and patients follow up.

9:35 Picoliter Droplet-Based Digital PCR for the Quantitative Analysis of Circulating Tumor DNA for Cancer Patient Follow Up

Valerie Taly, Ph.D., Group Leader, UMR S1147, University of Paris Descartes, CNRS, France

Picoliter droplet-based digital PCR represents a highly efficient tool for cancer research, allowing unprecedented sensitivity and accuracy for rare sequences detection. We will illustrate its pertinence for overcoming actual clinical oncology challenges by presenting the results of different retrospective and prospective studies. In particular, the detection of circulating tumor DNA and its potential use for patient treatment management will be demonstrated.

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Biological Dynamics
Rapid answers for clinical treatment

10:05 Quantification of Circulating Biomarkers from Plasma and Serum Using AC Electrokinetics

David J. Charlot, Ph.D., President and CTO, Biological Dynamics, Inc.

Interest in the isolation, quantification, and analysis of cell-free biomarkers directly from blood has grown significantly. Biological Dynamics has developed proprietary platforms for isolating and quantifying large circulating biomarkers from physiological solutions using AC Electrokinetics (ACE). Biomarkers, such as necrotic cell-free DNA (ncfDNA), have been established as indicators of cancer, and the ability to detect and track these biomarkers unlocks a new era in early disease diagnosis and treatment response monitoring.

10:35 Coffee Break in the Exhibit Hall with Poster Viewing

11:15 PANEL DISCUSSION: Next Steps for Clinical Advancement of Circulating Biomarkers

Moderator: *Klaus Pantel, M.D., Professor and Founding Director, Institute of Tumor Biology, University Medical Center Hamburg-Eppendorf, University of Hamburg, Germany*

Panelists: *Alain R. Thierry, Ph.D., Senior Investigator, Institute of Research on Oncology of Montpellier, INSERM, France*

Valerie Taly, Ph.D., Group Leader, UMR S1147, University of Paris Descartes, CNRS, France

Jean-Yves Pierga, Ph.D., Professor, Medical Oncology, Circulating Biomarkers Lab, Institut Curie & Université Paris Descartes, France

12:15 Sponsored Presentation *(Opportunity Available)*

12:45 Luncheon Presentation *(Sponsorship Opportunity Available)* **or Enjoy Lunch on Your Own**

13:15 Session Break

TECHNOLOGY TO CAPTURE AND CHARACTERIZE

14:15 Chairperson's Remarks

Fred R. Kramer, Ph.D., Professor, Microbiology, Biochemistry & Molecular Genetics, Rutgers University, United States

14:20 Multiplex Detection of Extremely Rare Mutant Sequences Associated with Cancer

Fred R. Kramer, Ph.D., Rutgers University, United States

Extraordinarily specific "SuperSelective" PCR primers enable the simultaneous quantitation of extremely rare mutations that occur in different cells, even though they are located within the same or adjacent codons. Consequently, multiplex assays for the early detection of mutations associated with cancer should enable therapy to be guided by the results of periodic "liquid biopsies" that analyze DNA fragments present in plasma samples.

15:20 cfDNA Profiling in Large Human Melanoma Blood and Tissue Bank

William A. Robinson, Ph.D., M.D., Rella and Monroe Rifkin Endowed Chair & Professor, Medical Oncology, University of Colorado Denver, Anschutz Medical Campus, United States

We have examined serum and plasma samples for mutated cfDNA in a large number of human melanoma samples and correlated the findings with staging, clinical course and response to various treatment(s). This includes serial samples from individual patients. Our data suggests that patients can be monitored for recurrence and response to treatment using cfDNA.

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15:50 LiquidBiopsy™ Platform: A Molecular Platform that Supports Next-Generation Sequence Analysis of Circulating Tumor Cells and Circulating Tumor DNA

Sponsored by ThermoFisher SCIENTIFIC

Paul W. Dempsey, Ph.D., CSO, Cynvenio Biosystems

Circulating tumor cells (CTC) and cell-free DNA (cfDNA) represent important and distinct templates that can support the analysis of tumor-associated mutations by next generation sequencing (NGS). The two templates present distinct advantages for clinical research analysis and provide complementary information from a blood sample. Support for molecular analysis of CTC and cfDNA requires technology to overcome both limiting template and very high signal to noise ratios. Data will be presented describing how the LiquidBiopsy™ Platform supports analysis of CTC and cfDNA from a blood sample by pairing novel CTC enrichment technologies with high sensitivity sequencing in a single Ion PGM™ System based analysis.

16:20 Refreshment Break in the Exhibit Hall with Poster Viewing

17:00 Single-Tube Enrichment of Mutations in Cancer Gene Panels from Circulating DNA, Using COLD-PCR Prior to Targeted Amplicon Re-Sequencing

G. Mike Makrigiorgos, Ph.D., Professor, Radiation Oncology, Dana Farber and Harvard Medical School, United States

We present a newly developed method via which mutations in numerous amplicons are first enriched via COLD-PCR in a single-tube reaction, prior to targeted re-sequencing. Using this approach, mutations of 0.01-0.1% abundance can be detected via next-generation sequencing.

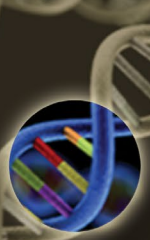
17:30 From Serum to Saliva Diagnostics – Comparative Studies on Circulating Free Nucleic Acids

Christa Noehammer, Ph.D., Senior Scientist, Molecular Diagnostics, AIT Austrian Institute of Technology, Austria

The aim of our research activities at AIT is to define reliable biomarkers suitable for early and non-invasive disease diagnosis and prognosis. We have particularly focused on the establishment and optimization of a whole range of high throughput technologies (e.g. planar -and bead microarrays, microfluidic quantitative PCR, Luminex bead technology) to meet the special demands and challenges of diagnostic biomarker discovery - and validation in body fluids. Using this specific technology expertise e.g. we successfully discovered autoantibody- and DNA methylation -based diagnostic marker panels for the big 4 cancer entities (breast, colon, prostate, lung) in serum or plasma. Based on these success stories and the evident advantages of saliva as a diagnostic matrix our special interest was now to go for saliva diagnostics and to evaluate saliva among others for its suitability for circulating cell-free nucleic acid-based diagnostics by comparing the reliability and performance of a DNA-methylation and microRNA biomarkers in serum and saliva of breast cancer patients.

18:00 Close of Conference

Cambridge Healthtech Institute,
250 First Avenue, Suite 300,
Needham, MA 02494
www.healthtech.com



Molecular Diagnostics EUROPE

How to Register: www.MolecularDxEurope.com

reg@healthtech.com • P: 781.972.5400 or Toll-free in the U.S. 888.999.6288

Please use keycode MDXE F when registering

PRICING AND REGISTRATION INFORMATION

SHORT COURSE PRICING

	Commercial	Academic, Government, Hospital-Affiliated
Preconference Short Course	€549	€319

SC1: Preimplantation Genetic Diagnostics (13 April)

CONFERENCE PACKAGE PRICING

EVENT PRICING *BEST VALUE*
(Includes access to all four conferences, does not include short courses)

	Commercial	Academic, Government, Hospital-Affiliated
Registrations after 13 March 2015 and on-site	€1929	€949

BASIC PRICING

(Includes access to one conference, does not include short courses)

	Commercial	Academic, Government, Hospital-Affiliated
Registrations after 13 March 2015 and on-site	€1399	€639

CONFERENCE SELECTIONS

Monday-Wednesday, 13-15 April	Wednesday-Thursday, 15-16 April
Prenatal Molecular Diagnostics	Advanced Diagnostics for Infectious Disease
Tuesday-Wednesday, 14-15 April	**Wednesday-Thursday, 15-16 April**
Cancer Sequencing	Circulating Cell-Free Nucleic Acids

CONFERENCE DISCOUNTS

Poster Submission Discount (€45 Off) Poster abstracts are due by 13 March 2015. Once your registration has been fully processed, we will send an email containing a unique link allowing you to submit your poster abstract. If you do not receive your link within 5 business days, please contact jring@healthtech.com. *CHI reserves the right to publish your poster title and abstract in various marketing materials and products.

Alumni Discount SAVE 20%: Cambridge Healthtech Institute appreciates your past participation at Molecular Diagnostics Europe. As a result of the great loyalty you have shown us, we are pleased to extend to you the exclusive opportunity to save an additional 20% off the registration rate.

Register 3 - 4th is Free! Individuals must register for the same conference or conference combination and submit completed registration form together for discount to apply.

Group Discounts are Available! Special rates are available for multiple attendees from the same organization. For more information on group discounts contact Uma Patel at 781-972-5447

***Alumni, Twitter, LinkedIn, Facebook or any other promotional discounts cannot be combined.** Discounts not applicable on Event Short Courses.

(Alumni and Register 3 - 4th is free discounts cannot be combined)

If you are unable to attend but would like to purchase the Molecular Diagnostics Europe CD for €600 (plus shipping), please visit MolecularDxEurope.com. Massachusetts delivery will include sales tax.

ADDITIONAL REGISTRATION DETAILS

Each registration includes all conference sessions, posters and exhibits, food functions, and access to the conference proceedings link.

Handicapped Equal Access: In accordance with the ADA, Cambridge Healthtech Institute is pleased to arrange special accommodations for attendees with special needs. All requests for such assistance must be submitted in writing to CHI at least 30 days prior to the start of the meeting.

To view our Substitutions/ Cancellations Policy, go to http://www.healthtech.com/regdetails

Video and or audio recording of any kind is prohibited onsite at all CHI events.