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FOURTH ANNUAL

Advances in Prenatal Molecular Diagnostics

Trends & Implications in a Rapidly Changing Landscape

NOVEMBER 29 - DECEMBER 1, 2016 | Boston Marriott Cambridge | Cambridge, MA

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SPECIAL KEYNOTE PRESENTATION:

Pushing Towards the Limit for Non-Invasive Prenatal Testing

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

SECOND ANNUAL

Reproductive Genetic Diagnostics

Advances in Preimplantation and Infertility Diagnostics

DECEMBER 1 - 2, 2016 | Boston Marriott Cambridge | Cambridge, MA

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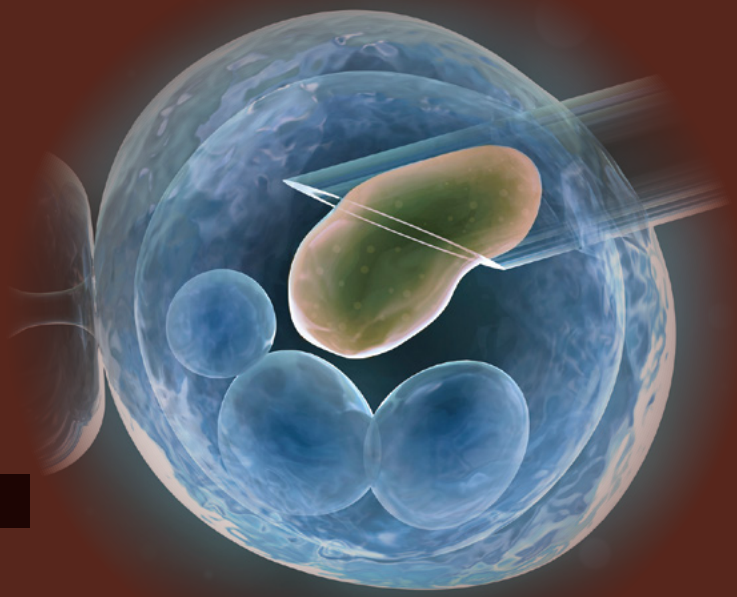

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The growing acceptance and availability of cell-free DNA-based screening from maternal blood is having a dramatic impact on prenatal testing. One result has been the steady decline in the number of women choosing invasive testing, with growing shift away from karyotyping in favor of arrays, or in some cases sequencing, for analysis of these samples.

The range of approaches being employed for cell-free DNA testing continues to grow, including the release of kits for in-house analysis and so-called second generation assays that provide much improved detection of sub-chromosomal anomalies. There has been a push to expand NIPT use beyond higher-risk pregnancies, as well as extending the range of genetic conditions reported from the tests, but such changes come at a price.

Research into testing based on isolation of fetal cells from maternal blood has been underway for decades, and the discussion has been couched in terms of "if this approach can be commercialized". At this meeting last year, for the first time, the conversation shifted to "when this approach is commercialized" as numerous groups reported substantial progress in achieving reliable isolation that would make this possible. Updated comparisons and examination of these issues, and the implementation of these different approaches, will again make for lively discussion and insight into where this field is headed and ways for researchers, test providers, clinicians and clinics to take these developments into consideration.

TUESDAY, NOVEMBER 29

7:30 am Registration and Morning Coffee

Overview

8:30 Chairperson's Remarks
Ronald Wapner, M.D., Director, Reproductive Genetics and Vice Chair, Research, Department of Obstetrics & Gynecology, Columbia University Medical Center

8:40 Trends and Implications for Fetal Anomalies: Beyond the Karyotype
Ronald Wapner, M.D., Director, Reproductive Genetics and Vice Chair, Research, Department of Obstetrics & Gynecology, Columbia University Medical Center
When a fetal structural anomaly is identified, the prognosis and subsequent counseling and treatment are dependent on understanding the underlying etiology. Although ultrasound imaging can define the structural alterations, it is incapable, in most cases, of determining the neurocognitive impact. Chromosomal microarray (CMA) and Whole Exome Sequencing (WES) now allow detailed understanding of the genomic cause of many of the anomalies. In cases with a normal karyotype, CMA will identify a causative copy number variant in 5-10% of cases. Early studies evaluating WES suggest that between 10 and 30% of anomalies may be due to a de-novo mutation.

9:10 Comparing Differing Diagnostic Values and Differing Types of Risk for Prenatal Diagnostic Options
Joe Leigh Simpson, M.D., Senior Vice President, Research & Global Programs, March of Dimes Foundation
Each of the three current options (universal invasive procedure, serum analyte/NT screening, cell-free DNA screening) should be offered in the context of differing patient desires for learning the presence of either just traditional autosomal trisomy or additional karyotypic abnormalities or copy number variants.

9:40 The Impact of Health Economic Evidence on the Coverage and Reimbursement of New Prenatal Diagnostics
Mark Girardi, Vice President, Market Access, GfK Health
In order to maximize coverage and adoption for new diagnostic technologies in prenatal testing, payers are requiring companies to demonstrate the potential economic as well as clinical benefits of using the new technology. Budget impact and clinical effectiveness models, which are an effective way to capture and articulate the potential value of a new technology, will be discussed.

10:10 Networking Coffee Break

10:30 Perspectives and Challenges to Genetic Counseling in the Prenatal Setting
Eugene Pergament, M.D., Ph.D., FACMG, Professor, Obstetrics and Gynecology, Northwestern; Attending, Northwestern University Medical School Memorial Hospital
The broad spectrum of genetic screening and diagnostic tests currently available presents unparalleled challenges to informing and counseling the prenatal patient. Genetic counseling now requires an unprecedented level of patient information and of understanding with regard to the implications and consequences of different forms of prenatal genetic testing. Various models of genetic counseling in the prenatal setting will be presented, addressing key counseling requirements in the era of molecular genetics.

11:00 Implementation of Patient-Like Seraseq™ Aneuploidy Reference Materials for NIPT and PGS Applications
Sponsored by seracare
Russell Garlick, CSO, SeraCare Life Sciences
As regulatory oversight increases, there is great demand for robust, patient-like reference materials that can be used to expedite development, perform analytical validation, and monitor daily run performance across the entire workflow. In this presentation, we describe our aneuploidy reference material technology, as well as summarize field performance for both NIPT and PGS applications.

11:30 Questions and Answers Related to Zika Virus Infection during Pregnancy
Suresh Boppana, Ph.D., University of Alabama, Birmingham
A large number of babies with microcephaly and other birth defects are born to women with Zika virus infection (ZIKV) during pregnancy during the ongoing epidemic of ZKV in Brazil and other Latin American countries. Both the WHO and CDC have concluded that there is a causal link between ZIKV during pregnancy and microcephaly. However, significant gaps including the lack of reliable, rapid and simple diagnostic methods remain in our understanding of the natural history and pathogenesis of ZIKV in pregnant women and its impact on the developing fetus. The state-of-the art knowledge on the diagnosis and monitoring of ZIKV during pregnancy and the similarities and differences with other congenital infections will be discussed.

12:00 pm Luncheon Presentation *(Sponsorship Opportunity Available)* **or Enjoy Lunch on Your Own**

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Isolation and Analysis of Fetal Cells from Maternal Blood

1:30 Chairperson's Remarks

Art Beaudet, M.D., Department of Molecular & Human Genetics, Baylor College of Medicine

1:35 High Precision Single-Cell Genomics for PGD/PGS and NIPT

Xiaoliang "Sunney" Xie, Ph.D., Department of Chemistry and Chemical Biology, Harvard University

Single-cell whole genome sequencing is becoming increasingly important for PGD/PGS and NIPT. A pre-requisite for sequencing is single-cell whole genome amplification (WGA), which currently lacks amplification uniformity, hence exhibiting low genome coverage and sequence-dependent bias. We have developed a new method for single-cell WGA which offers, to the best of our knowledge, the highest precision for copy number variation and single nucleotide variation. Most noticeably, it allows for detection of kilobase-deletions not detectable by previous single-cell WGA methods.

2:05 Validation Studies for the Launch of a CLIA-Compliant Fetal Cell-Based Noninvasive Prenatal Test

Art Beaudet, M.D., Department of Molecular & Human Genetics, Baylor College of Medicine

Steady progress has been achieved towards a fetal trophoblast-based form of NIPT. From 3-10 cells can be recovered at 8-14 weeks gestation in the majority of pregnancies. Individual cells are subjected to whole genome amplification and genotyping followed by copy number analysis using whole genome shotgun next-generation sequencing. Numerous sub-chromosomal abnormalities have been detected.

2:35 Genome Analyses Utilizing DNA from Fetal Cells in Pregnant Women's Blood – Making a Case for Cell-Based NIPT (cbNIPT)

Ripudaman Singh, Ph.D., COO, ARCEDI Biotech Aps (Denmark)

Non-Invasive Prenatal Testing based on fetal cells in maternal blood (cbNIPT) has an advantage over the cell-free NIPT (cfNIPT) in that the fetal DNA is not contaminated with the maternal DNA, and hence holds a potential for larger genomic coverage, higher detection rate and lower false positive rate of prenatal genetic testing. We present a high throughput method for enriching fetal cells from maternal blood, subsequent amplification of the fetal genome and detection of chromosomal and subchromosomal variations in the genome.

3:05 Refreshment Break in the Exhibit Hall with Poster Viewing

3:45 NanoVelcro Chips for Isolation and Characterization of Circulating Fetal Nucleated Cells – Toward Noninvasive Prenatal Diagnostics

Hsian-Rong Tseng, Ph.D., Professor, Molecular & Medical Pharmacology, University of California, Los Angeles

In contrast to the existing rare-cell sorting approaches, our joint research team at UCLA pioneered the concept of "NanoVelcro" cell-affinity assay, in which a capture antibody-coated nanosubstrate substantially enhances the performance of rare cell enrichment from blood. The ways in which different generations of NanoVelcro Chips were employed in streamlined workflows to isolate and characterize single circulating fetal nucleated cells (CFNCs, including both trophoblast and fetal nucleated red blood cells) in maternal blood will be presented. Using maternal blood samples collected from expectant mothers who carried single fetuses, the CFNC-derived CGH microarray data were able to detect fetal genders and chromosomal aberrations, which had been confirmed by standard clinical practice. In addition to sharing our latest research progress in developing CFNC-based noninvasive prenatal diagnostic (NIPD) solutions, the challenges that still need to be resolved will be presented.

4:15 Advances in the Isolation of Fetal Cells from Maternal Blood

Brynn Levy, MSc (Med), Ph.D., FACMG, Professor of Pathology & Cell Biology, Columbia University Medical Center; Director, Clinical Cytogenetics Laboratory, Co-Director, Division of Personalized Genomic Medicine, College of Physicians and Surgeons

4:45 Technical Insights into Highly Sensitive Isolation and Genetic Characterization of Trophoblastic Cells for Prenatal Non-Invasive Diagnostics of Genetic Disorders

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

Isolation of rare trophoblastic cells from blood or from cervix is a technical challenge with impact on the number of collected fetal cells and on the quality of their DNA. By using the ISET (Isolation by Size of Epithelial Tumor/Trophoblastic cells) system, we have developed different isolation protocols and analyzed the number of collected trophoblastic cells and the quality of their DNA at the single cell level. Our results show that ISET allows the consistent recovery of trophoblastic cells from blood and cervical samples and their complete genetic analysis. They also show that high throughput protocols can be developed aiming the use of trophoblastic cells collected non invasively for prenatal diagnosis of genetic disorders.

» 5:15 KEYNOTE PRESENTATION: Pushing towards the Limit for Non-Invasive Prenatal Testing

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

Non-invasive prenatal testing using circulating fetal DNA in maternal plasma has been introduced globally over the past 5 years. My group is working on extracting diagnostic information that is hidden in the plasma DNA pool. One area is research concerning the tissue of origin of plasma nucleic acids. Hence, using thousands of methylation markers exhibiting tissue-associated methylation signatures, we are able to provide new insights into the origin of plasma DNA and to attribute an observed aberration in plasma DNA to its source. These new results and other emerging developments in non-invasive prenatal testing will be discussed.

6:00 Close of Day

WEDNESDAY, NOVEMBER 30

8:00 am Breakfast Breakout Roundtable Discussions

- Can or Should Any Current Prenatal Testing Components Be Replaced?
- Ethical and Genetic Counselling Challenges for Prenatal Testing
- Intellectual Property Issues with New Prenatal Testing Technologies
- Economics of Prenatal Testing Options and Reimbursement
- Implications for Expanding Cell-Free DNA Screening for Lower Risk Mothers

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- Commercial Potential for Non-Invasively Obtained Fetal Cells
- Prenatal Testing beyond Common Aneuploidies
- Bioinformatics Issues for Sequence-Based Testing
- Developing Biomarkers for Preeclampsia and Pre-Term Birth
- Cell-Free DNA Screening

Cell-Free DNA Screening

9:00 Chairperson's Remarks
Joe Leigh Simpson, M.D., Senior Vice President, Research & Global Programs, March of Dimes Foundation

9:05 Resource and Policy Implications of Extending NIPS
Megan Allyse, Ph.D., Assistant Professor of Biomedical Ethics, The Mayo Clinic
There are two major directions for more expanded application of NIPS. One direction involves greater depth of testing with higher risk pregnancies, by including microdeletions and other sub-chromosomal genetic conditions. The other direction involves additional patients, specifically more general population pregnancies. Both of these cases create additional public health burdens that will need to be addressed systemically if NIPS is to succeed as a routine medical procedure. Specific examples and budgetary implications will be discussed.

9:35 What Cell-Free DNA Screening Can't Do
Mark Evans, M.D., President, Fetal Medicine Foundation of America; Professor of Obstetrics and Gynecology, Mt. Sinai School of Medicine; Comprehensive Genetics
Major advances in technology have vastly improved the scope and reliability of cell free fetal DNA. However, simultaneously similar improvements to microarray techniques now show clinically abnormal CNVs in about 1% of all CVS or amniocentesis specimens on low risk patients. Particularly for younger women the rate of finding an abnormal CNV is 10x that of Down syndrome. Thus, the effort to abandon procedures in favor of cffDNA not only costs substantially more, but detects far less. In my program we offer CVS and microarray to all patients regardless of age because of the yield of 1% which approximates the expected traditional rate of abnormalities in a 38 year old – far higher than the threshold typically used in prenatal diagnosis for the past 4 decades.

10:05 Current Status of Expanded NIPS for Aneuploidy
Peter Benn, Ph.D., Department of Genomics and Genome Sciences, University of Connecticut Health Center
Non-invasive prenatal screening (NIPS) based on cell-free DNA in maternal plasma has expanded to include additional chromosome abnormalities beyond those involving chromosomes 21, 18, 13, X and Y. This includes unbalanced chromosome rearrangements, marker chromosomes, rare autosomal aneuploidies and also sets of specific microdeletion syndromes. In this presentation, I will review the current status of this testing, discuss clinical utility, and present some of the interpretation issues associated with expanded NIPT.

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

11:15 Genome-Wide Prenatal Cell Free DNA Testing: Clinical Relevance and Laboratory Experience
Sponsored by sequenom
Daniel S. Grosu, M.D., MBA, CMO, Clinical and Medical Affairs, Sequenom, Inc.
A significant proportion of chromosomal and sub-chromosomal abnormalities in the prenatal setting are not detectable by conventional cfDNA testing. Most of this informational gap can be bridged through a genome-wide approach that reports on copy number variation at a karyotype level of resolution (≥7 Mb in size) across the entire genome, in addition to select microdeletions less than 7 Mb in size. Clinical experience with genome-wide NIPT findings in a cohort of over 10,000 patients will be reviewed, alongside the clinical relevance of such findings.

11:30 Optimizing the Preanlytical Workflow for Circulating Cell-Free DNA using PAXgene Blood ccfDNA System
Sponsored by QIAGEN
Thomas Briggs, MS, MBA, Key Account and Business Development Manager, PreAnalytiX GmbH
Selection of preanalytical processing protocols can impact both performance and workflow efficiency when analyzing ccfDNA. The novel PAXgene Blood ccfDNA System addresses several of the common challenges in blood collection and stabilization of the ccfDNA profile during collection, storage and transport, while providing an integration system for quality ccfDNA isolation.

11:45 IP Issues in the Cell-Free DNA Testing Space
Konstantin Linnik, Ph.D., Partner, Intellectual Property, Nutter, McClennan & Fish LLP
Three recent U.S. Supreme Court decisions (Mayo, Myriad, and Alice) have produced a landslide change in IP protection for diagnostics. As a result, patent applicants and patent holders have been battled at the US Patent Office and the US courts. Many players are forced out of patent protection altogether. The currently pending case Ariosa v. Sequenom is seminal in attempting to re-dress the issues: Current state of IP protection in the U.S. and abroad for diagnostics, substance of Ariosa v. Sequenom case and how it relates to prior Supreme Court decisions, industry position, and potential future developments and practical approaches.

12:15 pm Luncheon Presentation *(Sponsorship Opportunity Available)*
or Enjoy Lunch on Your Own

1:45 Strategies and Structures Needed to Ensure Patients' Informed Access to NIPS
Ruth Farrell, M.D., Department of Obstetrics & Gynecology, Cleveland Clinic Foundation
There are a range of factors that have an impact on patient access to NIPS. One aspect of that involves patients learning about NIPS, having healthcare benefits in place to financially afford NIPT, and obtaining the resources to make an informed choice about if, when, and how to utilize it. Another involves providers, for which proper workflow is a key aspect. It is also about providers' education and skills to help patients make informed choices about NIPT in the context of their other screening and diagnostic testing options. Experience with these issues in a clinical setting will be presented.

2:15 Fragment Endpoints Reveal the Tissues of Origin of Circulating DNA
Matthew Snyder, Ph.D., University of Washington

2:45 Sponsored Presentation *(Opportunity Available)*

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3:15 Refreshment Break in the Exhibit Hall with Poster Viewing

4:00 Panel Discussion with Cell-Free DNA Screening Providers

Panelists:
Marcia Eisenberg, Ph.D., CSO, Labcorp
Solomon Moshkevich, Vice President, Product & Strategy, Natera
Peter Collins, CCO, Premaitha Health
Douglas Rabin, M.D., Medical Director, Women's Health, Quest Diagnostics
Daniel Grosu, M.D., MBA, CMO, Sequenom

5:30 Networking Reception in the Exhibit Hall with Poster Viewing

7:00 Close of Day

THURSDAY, DECEMBER 1

8:00 am Morning Coffee

Biomarkers of Preeclampsia and Pre-Term Birth

9:00 Chairperson's Remarks

9:05 Early Pregnancy Screening for Pre-Eclampsia Using Serum and Biophysical Markers

Howard Cuckle, Ph.D., OBGYN, Tel Aviv University, (Israel) and OBGYN, Columbia University Medical Center

One reason for retaining maternal serum markers and opting for contingent cfDNA rather than universal cfDNA testing is the value of maternal markers for detecting adverse outcomes such as pre-eclampsia. The use of a combination of two maternal serum markers and two biophysical markers can provide a useful means for early pregnancy (11-13 weeks) screening for pre-eclampsia. With such screening, aspirin can be effective in preventing pre-eclampsia if taken early enough.

9:35 Transcriptomic Clock during Pregnancy

Thuy Ngo, Ph.D., Department of Bioengineering, Stanford University (Stephen Quake's Lab)

Investigating and tracking the changes of the transcriptome during the course of pregnancy can help in the identification of abnormalities or adverse states readily. We have used next generation sequencing and quantitative PCR (qPCR) to analyze cell-free RNA in plasma from pregnant women at various time points across gestation. Differential expression analysis of cell-free RNA sequencing revealed distinct longitudinal patterns for several groups of genes modulated in pregnancy. These include immune modulation genes, placenta specific genes which we further monitored at high time resolution by qPCR measurements along with fetal specific genes from plasma collected weekly during pregnancy. Our results demonstrate that cell-free RNA can be used for noninvasive monitoring of both maternal responses and fetal development during pregnancy.

10:05 Data-Driven Development and Validation of Longitudinal Prediction Models for Preterm Birth from Electronic Medical Records

Iya Khalil, Ph.D., Executive Vice President and Co-Founder, GNS Healthcare

Maternal risk for preterm delivery is multifactorial and evolves dynamically throughout pregnancy. We have synthesized a longitudinal Bayesian causal model of baseline and prenatal risk factors contributing to Preterm Birth (PTB) using Electronic Medical Records (EMR) from ITMI's Preterm Birth research study. Causal models have achieved preliminary validation in ITMI's First 1,000 Days of Life observational study cohort. In conjunction with ITMI, we are developing partnerships to prospectively validate the causal model in external populations and deploy an integrated physician dashboard for patient risk assessment and clinical decision support. Using these tools, the risk of preterm delivery can be readily estimated from comprehensive electronic records to provide an individualized assessment for clinical case management throughout pregnancy.

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

11:15 Closing Panel: Predicting the Landscape for Prenatal Molecular Diagnostics: The Next Few Years

Mark Evans, M.D., Professor, Obstetrics & Gynecology, Mt. Sinai School of Medicine, and Comprehensive Genetics
Cynthia Morton, Ph.D., William Lambert Richardson Professor of Obstetrics, Gynecology & Reproductive Biology and Professor of Pathology, Harvard Medical School; Director, Cytogenetics, Brigham & Women's Hospital
Joe Leigh Simpson, M.D., Senior Vice President, Research & Global Programs, March of Dimes Foundation
Ronald Wapner, M.D., Director, Reproductive Genetics and Vice Chair, Research, Department of Obstetrics & Gynecology, Columbia University Medical Center

12:15 pm Close of Prenatal Molecular Diagnostics Conference

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Couples experiencing infertility or failed pregnancy can now take advantage of a number of molecular diagnostic options to determine the health and wellness of their embryos as well as their own fertility. Cambridge Healthtech Institute's Second Annual Reproductive Genetic Diagnostics conference examines the latest in preimplantation diagnostic and screening technologies, including their applications in assessing mosaicism, determining embryo transfer, and evolution in CCS. The meeting will also examine the potential for less- and non-invasive PGD, being developed in response to the potential effect biopsy and vitrification may have on the development of the embryo. There will be discussion on PGD for donor eggs, such as how much or how often testing should be done. The meeting will also cover advances in CRISPR, including current research and capabilities, as well as how this technology may or may not be used in the future. New this year, special attention will be paid to molecular diagnostics for finding infertility biomarkers.

THURSDAY, DECEMBER 1

1:00 pm Registration

Mosaicism

2:00 Chairperson's Remarks
Mark Umbarger, Ph.D., Director, Research and Development, Good Start Genetics

2:05 Mechanisms of Epigenetic Regulation in Early Development
Alexander Meissner, Ph.D., Professor, Stem Cell & Regenerative Biology, Harvard University
In mammals, cytosine methylation is predominantly restricted to CpG dinucleotides and stably distributed across the genome, with local, cell-type-specific regulation directed by DNA binding factors. This comparatively static landscape is in marked contrast with the events of fertilization, where most of the genomic methylation is erased. We will present the latest advances in our understanding of this critical window in mammalian development.

2:35 The Challenge of Embryonic Mosaicism in Preimplantation Genetic Screening
Jason Franasiak, M.D., FACOG, Obstetrics and Gynecology, Division of Reproductive Endocrinology and Infertility, Reproductive Medicine Associates of New Jersey and Thomas Jefferson University
The very nature of embryonic mosaicism presents diagnostic, analytical, and clinical challenges. Understanding distribution of chromosome compliments throughout the embryo, the complex bioinformatics involved in characterizing mosaicism in a single embryonic biopsy, and the varied clinical outcomes of embryos designated as mosaic are fundamental to implementing this in clinical care as it pertains to preimplantation genetic screening.

Advances and Practical Applications in PGD and PGS

3:05 Novel Application of NGS at NLRP7 to Permit Single Embryo Transfer for Patient with Multiple Consecutive Gestational Trophoblastic Disease (Molar Pregnancy)
E. Scott Sills, M.D., Ph.D., Medical Director, Reproductive Research Office, Center for Advanced Genetics
Gestational trophoblastic disease (molar pregnancy) occurs in about 1 of 1000 conceptions and is thought to result from fertilization of an abnormal oocyte which leads to an embryo with an irregular chromosomal constituency. Although the etiology of the condition remains incompletely understood, recent research has implicated a specific mutation (NLRP7) identified in some women with molar pregnancy. Here, we present our experience with genetic testing of both parents and blastocysts derived from IVF in a couple with five consecutive molar pregnancies. Data from PGS & gene sequencing are discussed to contribute new information concerning the genetics of molar pregnancy.

3:35 Refreshment Break in the Exhibit Hall with Poster Viewing

4:15 Optimal Euploid Embryo Transfer Strategy Following PGS with NGS: A Randomized Controlled Trial
Alison Coates, Embryology Laboratory Director, Oregon Reproductive Medicine
The two strategies currently used in clinical practice are frozen thawed or fresh transfer. The frozen thawed strategy involves cryopreservation of all blastocysts in the cohort to await PGS results in preparation for a frozen embryo transfer. The fresh strategy involves biopsy of only day 5 blastocysts and rush testing overnight for a fresh embryo transfer of euploid embryos on day 6. There are benefits and challenges to each approach.

4:45 Developments in Comprehensive Chromosome Screening
Nathan R. Treff, Ph.D., Director, Molecular Biology Research, Reproductive Medicine Associates of New Jersey
Comprehensive chromosome screening (CCS) for preimplantation embryonic aneuploidy has now demonstrated the ability to improve clinical outcomes for patients with infertility in multiple randomized controlled trials. Platforms for testing include SNP array, array CGH, qPCR, and next generation sequencing (NGS). Prior to the use of these various downstream quantitation methods is the critical step of DNA amplification from limited amounts of starting material obtained from an embryo biopsy. One strategy, whole genome amplification, introduces artefacts often not overcome by highly parallel methods of quantitation and can be over interpreted as mosaicism and segmental aneuploidy. Targeted methods of amplification reduce technical artefacts and provide proven accuracy and clinical predictive values in rigorous preclinical trials. These and other aspects of contemporary CCS methodologies will be discussed.

5:15 Sponsored Presentation *(Opportunity Available)*

6:00 Networking Reception in the Exhibit Hall with Poster Viewing

7:00 Close of Day

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FRIDAY, DECEMBER 2

7:30 am Breakfast Breakout Roundtable Discussions

Prospects for Less- and Non-Invasive Diagnostics

8:25 Chairperson's Remarks

E. Scott Sills, M.D., Ph.D., Medical Director, Reproductive Research Office, Center for Advanced Genetics

8:30 The Potential Use of Blastocoel Fluid from Differentiated Blastocysts to Conduct Preimplantation Genetic Screening

Kyle Tobler, M.D., Assistant Professor, Faculty, Reproductive Endocrinology and Infertility, Obstetrics and Gynecology, Womack Army Medical Center, Fort Bragg North Carolina

Blastocoel fluid is potentially under-utilized. This fluid may prove useful for further analysis of blastocysts as part of PGS and PGD, which would allow the genetic analysis to be completed without conducting an embryo biopsy. There are few and conflicting reports on the utility of blastocoel fluid for this purpose. This presentation will review the past research conducted on blastocoel fluid and possible future directions in the application of blastocoel fluid analysis to PGS and PGD.

9:00 Corona Cell RNA Sequencing from Individual Oocytes Revealed Transcripts and Pathways Linked to Euploid Oocyte Competence and Live Birth

Mandy Katz-Jaffe, Ph.D., Scientific and Genetics Director, Colorado Center for Reproductive Medicine

Corona cells surround the oocyte and maintain a close relationship through transzonal processes and gap junctions, making them an ideal source for the assessment of oocyte competence. In this study, individual corona cell transcriptomes were successfully generated using RNA-sequencing and reproductive outcomes analyzed following a frozen euploid blastocyst transfer. Numerous enriched pathways were associated with live birth including Wnt and MAP kinase signaling, highlighting novel non-invasive biomarkers of embryo viability.

9:30 Non-Invasive Testing of Gametes and Embryos for IVF

Denny Sakkas, Ph.D., Scientific Director, Boston IVF

The non-invasive assessment of preimplantation embryos has been largely limited to the use of morphology and has become the primary tool of the embryologist for screening gamete and embryo quality. The advent of "Omics" technologies has allowed us to broaden our assessment of gametes and embryos. This lecture will discuss how we can now assess the media surrounding the embryo using both proteomic or metabolomics based analysis. In addition, the use of advanced imaging systems that allow us to obtain relevant information linked to gamete and embryo quality will be discussed.

10:00 Sponsored Presentation *(Opportunity Available)*

10:30 Coffee Break in the Exhibit Hall with Poster Viewing

Impact of Testing on Embryo Development

11:00 Chairperson's Remarks

Stephen A. Krawetz, BSc, Ph.D., Associate Director, C.S. Mott Center for Human Growth and Development, Charlotte B. Failing Professor of Fetal Therapy and Diagnosis, Ob/Gyn & Molecular Medicine & Genetics, Wayne State University School of Medicine

11:05 The Impact of Biopsy on Human Embryo Developmental Potential during Preimplantation Genetic Diagnosis

Danilo Cimadomo, MSc, Ph.D. Student, GENERA Center for Reproductive Medicine, Clinica Valle Giulia

The biopsy strategy represents a critical aspect not to affect embryos' intrinsic possibilities to result in a healthy full-term birth during PGD/PGD-A cycles. Single/double blastomere biopsy at the cleavage stage has been demonstrated detrimental for embryo viability. For polar body biopsy, no class I data have been produced to properly assess its value. Blastocyst stage biopsy has been instead established as the safest approach, whose implementation is indeed increasing worldwide.

Molecular Diagnostics for Infertility and Implantation Failure

11:35 Decoding the Genetic Basis of Human Fertility Potential and Infertility Conditions

Piraye Yurttas Beim, Ph.D., Founder, CEO, Celmatix

Over one-quarter of genes in the human genome have some impact on a woman's fertility. For the past seven years, Celmatix has conducted research to vet, validate and discover novel genetic biomarkers that will dramatically impact the diagnosis and treatment of infertility. Having this genetic lens on fertility goes beyond a woman's ability to conceive; a woman's reproductive health impacts every other aspect of her health including cardiovascular disease, osteoporosis, and cancer risk.

12:05 pm Sperm RNAs: Biomarkers of Male Fitness and the Birth of a Healthy Child

Stephen A. Krawetz, BSc, Ph.D., Associate Director, C.S. Mott Center for Human Growth and Development, Charlotte B. Failing Professor of Fetal Therapy and Diagnosis, Ob/Gyn & Molecular Medicine & Genetics, Wayne State University School of Medicine

The utility of sperm RNA elements to assess the male contribution as a function of the idiopathic infertile couple is presented. Derived from sperm RNAs, these elements can identify those idiopathic infertile males who are likely to father a healthy child without the use of ART and those who would benefit from ART. Perhaps an objective assay to assess the impact of the dad's contribution for the idiopathic infertile couple is foreshadowed.

12:35 Mitochondrial DNA as a Biomarker for *in vitro* Fertilization Outcome

Emre Seli, M.D., Professor, Obstetrics, Gynecology, and Reproductive Sciences, Yale School of Medicine

Mitochondrial function has been associated with oocyte function and embryo competence, with implications for reproductive aging. As such, testing of mitochondrial DNA content or function provides a potential target for assessment of viability of euploid embryos.

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1:05 Luncheon Presentation *(Sponsorship Opportunity Available)* **or Enjoy Lunch on Your Own**

Beyond PGD: Carrier Screening and POC Testing

2:15 Chairperson's Remarks

Jason Franasiak, M.D., FACOG, Obstetrics and Gynecology, Division of Reproductive Endocrinology and Infertility, Reproductive Medicine Associates of New Jersey and Thomas Jefferson University

2:20 Carrier Screening in the Era of Expanding Genetic Technology

Aishwarya Arjunan, Faculty, Center for Genetic Medicine, Northwestern University Feinburg School of Medicine

The Center for Jewish Genetics provides genetic education and carrier screening to individuals of Jewish descent. Carrier screening has traditionally been performed by targeted mutation analysis for founder mutations with an enzyme assay for Tay-Sachs carrier detection. The development of next-generation sequencing (NGS) allows for higher detection rates regardless of ethnicity. Here, we explore differences in carrier detection rates between genotyping and NGS in a primarily Jewish population.

2:50 Spectrum of Cytogenomic Abnormalities Revealed by Array-CGH in Products of Conception Culture Failure and Normal Karyotype Samples

Peining Li, Ph.D., Associate Professor, Genetics, Yale School of Medicine

Array comparative genomic hybridization (aCGH) analysis was performed on product of conception culture failure (POC-CF) and normal karyotype (POC-NK) samples. Compiled results from our study and reported case series demonstrated an abnormality detection rate of 35% for chromosomal abnormalities in POC-CF, 3.7% for pathogenic CNVs in POC-CF, and 4.6% for pathogenic CNVs in POC-NK. Further Ingenuity Pathway Analysis on gene content from the pathogenic CNVs revealed gene networks causing miscarriages.

Gene Therapy: Research and Potential Uses

3:20 Panel Discussion: The Role of Preimplantation Genetic Diagnosis in Germline Gene Therapy

Moderator: Eugene Pergament, M.D., Ph.D., FACMG, Professor, Obstetrics and Gynecology, Northwestern; Attending, Northwestern University Medical School Memorial Hospital

Jason Franasiak, M.D., FACOG, Obstetrics and Gynecology, Division of Reproductive Endocrinology and Infertility, Reproductive Medicine Associates of New Jersey and Thomas Jefferson University

Peter Benn, Professor, Genetics and Genome Sciences, University of Connecticut Health Center

Germline gene therapy has now become a technical reality and the field of preimplantation genetic diagnosis will be directly involved and responsible. Unresolved are issues directly related to the ethical, moral, social and economic consequences of the application of germline gene therapy to human gametes and preimplantation embryos. Objective insights into the positive and negative implications of germline gene therapy are the focus of this panel discussion.

4:20 Close of Reproductive Genetic Diagnostics Conference

All speakers were outstanding. Meeting content was diverse and enlightening. I am looking forward to next year's meeting.

-Associate Director, Medical Affairs, Ferring Pharmaceuticals

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Cambridge Healthtech Institute encourages attendees to gain further exposure by presenting their work in the poster sessions. To secure a poster board and inclusion in the conference materials, your abstract must be submitted, approved and your registration paid in full by **October 21, 2016**.

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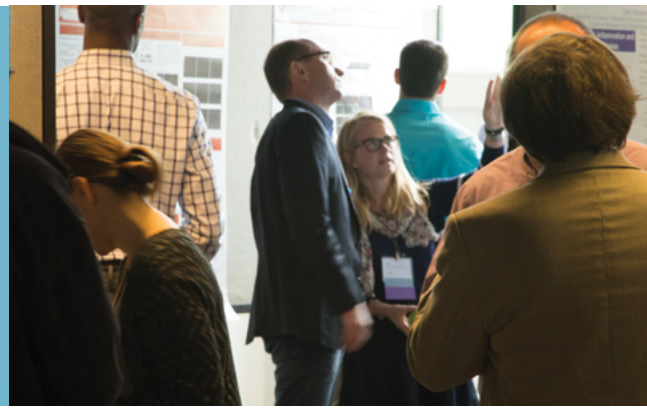
Poster inquiries: jring@healthtech.com



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For sponsorship and exhibit information, please contact:

Ilana Quigley
Sr Business Development Manager
781-972-5457
iquigley@healthtech.com

Hotel & Travel Information

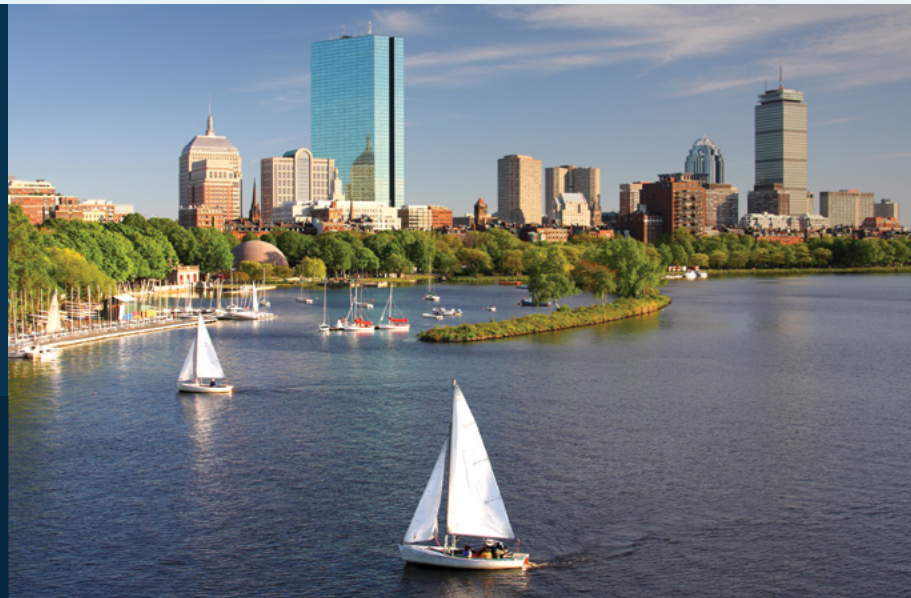
CONFERENCE VENUE AND HOTEL:

Boston Marriott Cambridge
50 Broad Street
Cambridge, MA 02142
(617) 494-6600

Discounted Room Rate: $259 s/d
Discounted Cut-off Date: November 7, 2016

RESERVATIONS AND ADDITIONAL TRAVEL INFORMATION:

Go to the travel page of **healthtech.com/prenatal-diagnostics** for additional info



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Advance registration until October 21	$2549	$1099
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(Includes access to Prenatal Molecular Diagnostics only)

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(Includes access to Reproductive Genetic Diagnostics only)

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Poster Submission - Discount ($50 Off): Poster abstracts are due by October 21, 2016. 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.

Group Discounts: Discounts are available for multiple attendees from the same organization. For more information on group rates contact Jeff Knight at +1-781-247-6264 or jknight@healthtech.com

Want to Register by Phone? Contact our Registration Department at 781-972-5400 or Toll-free in the US at 888-999-6288.

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