Advances in Prenatal Molecular Diagnostics

Trends & Implications in a Rapidly Changing Landscape

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Inaugural Reproductive Genetic Diagnostics

Advances in Carrier Screening, Preimplantation Diagnostics, and POC Testing

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Cambridge Healthtech Institute,
250 First Avenue, Suite 300,
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Third Annual
Advances in Prenatal Molecular Diagnostics
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November 16-18, 2015
Boston, MA | Omni Parker House Hotel

CONFERENCE INCLUDES:
• Trends and Best Practices for Invasively-Obtained Samples
• Evaluation and Implementation of NIPT
• Representatives from Eight NIPT Providers
• Expanded Coverage of Fetal Cell Isolation & Analysis Advances
• Biomarkers for Fetal and Maternal Health
• More than Ten Roundtable Breakout Discussions

Program Advisors:
Arthur Beaudet, M.D., Chair, Department of Molecular & Human Genetics, Baylor College of Medicine
Subhashini Chandrasekharan, Ph.D., Research Assistant Professor, Institute for Genome Sciences & Policy, Duke University
David H. Ledbetter, Ph.D., FACMG, Executive Vice President and CSO, Geisinger Health System
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 for Research and Global Programs, March of Dimes Foundation
Ronald J. Wapner, M.D., Director, Reproductive Genetics & Vice Chair, Research, Department of Obstetrics & Gynecology, Columbia University Medical Center
Innovative educational strategies are required to face the challenges of rapidly changing knowledge, at the intersections of research and clinical medicine, in the multidisciplinary field of prenatal diagnosis.

11:45 Challenges of Genetic Counseling: Traveling the Prenatal Diagnosis Road with our Patients
Mary-Frances Garber, MS, CGC, Private Practice: Listening, Reflecting, Healing
With the advancements in screening and testing options, patients may have a menu they can select from in an attempt to gain reassurance about the health of their baby, or to obtain a prenatal diagnosis of a certain condition. Genetic counselors have always had the responsibility of educating patients regarding the specifics surrounding these testing options, but perhaps more importantly travelling with and supporting couples on their prenatal diagnosis journey. Some of the ways genetic counseling has changed, as well as specific cases to illustrate counseling challenges, will be presented.

12:15 pm Luncheon Presentation: Seased™ Aneuploidy Reference Materials for Non-Invasive Prenatal Screening (NIPS)
Russell Garlick, Ph.D., CSO, SeraCare Life Sciences
The detection of circulating cell-free fetal DNA (cfDNA) in maternal blood by next-generation sequencing is becoming the preferred method to screen for fetal aneuploidy. As the market rapidly moves from high risk screening to “population” screening there is a need for reliable controls. The use of proper controls, standards and reference materials has always been critical in ensuring diagnostic laboratory quality management systems in order to ensure accurate results are reported. This presentation will focus on a new generation of NGS aneuploidy controls for non-invasive prenatal screening assays.

12:45 Session Break

NON-INVASIVE PRENATAL DIAGNOSTICS (NIPD)
1:45 Chairperson’s Remarks
Joe Leigh Simpson, M.D., Senior Vice President, Research and Global Programs, March of Dimes Foundation

1:50 Economic Analysis of Prenatal Diagnosis
Aaron Caughey, Ph.D., Professor and Chair, Department for Women’s Health Research & Policy, Oregon Health & Science University
There are a number of important economic considerations related to prenatal diagnosis. An analysis of both short- and long-term costs, as well as the impact on quality-adjusted life years related to prenatal diagnostic decision making, will be presented. Specific analytic approaches related to cost-effectiveness will also be explored.

2:20 The Pros and Cons of Extending NIPT for All Pregnancies, Not Just Those at Higher Risk
Joe Leigh Simpson, M.D., Senior Vice President, Research and Global Programs, March of Dimes Foundation
Detection of fetal aneuploidy through cell-free fetal DNA in maternal blood unequivocally is superior to traditional serum analyte/nuchal translucency. Sensitivity for tested aneuploids is higher (trisomy 21 > 99% versus at best 92-93%) and false rates much lower (0.1% versus 5%). Even greater value will be gained when NIPT can routinely yield information comparable to array CGH or at least karyotypes (5-7 Mb aberrations). Ability to extend further into transcriptomics will allow monitoring fetal development beyond traditional fetal genetic disorders.
2:50 Cell-Free Fetal DNA Screening in a Large Integrated Health Care System
Jeffrey Greenberg, MS, Genetic Screening Program Director, Genetic Services, SC Permanente Medical Group
The workflow, uptake, performance and outcomes of cell-free fetal DNA prenatal screening in a large integrated health care system will be presented. Prenatal screening in California is unique in its standardization under the Department of Public Health, which mandates the offering of, and oversees the testing and follow-up for, statewide analyte screening. Statistics for 1.5 years of cell-free DNA testing in a high-risk population will also be discussed.

3:20 Refreshment Break in the Exhibit Hall with Poster Viewing

4:00 Implementation of NIPT: A Case Study of Technology Evolution
Gautam Kollu, Head, Market Development, Reproductive and Genetic Health, Illumina
Since its introduction in 2012, NIPT has become the fastest adopted test in the history of molecular diagnostics and is currently the most widely used NGS clinical test. In the past two years, various labs have introduced panels and technological advancements that have expanded the clinical applications of NIPT. This talk will cover the evolution of NIPT from a focused test to its current state, with an eye on how the testing infrastructure has changed globally from a few California labs to labs worldwide doing this test.

4:30 Healthcare Economics in a General Screening Population
Sabah Oney, Ph.D., Director, Business Development, Operations, Ariosa Diagnostics, Inc.

5:00 Clinical Performance of the IONA® Test (CE-IVD)
William (Pepper) Denman, M.D., CMO, Premaitha Health
The IONA® test (CE-IVD) offers accurate non-invasive prenatal screening for Trisomies 21, 18, and 13 with a significant reduction in time to report results. Clinical performance to date of the IONA® test and the advantages offered to the clinical team will be highlighted.

5:15 Development and Validation of the Veracity Non-Invasive Prenatal Test
Philippos C. Patsalis, Ph.D., Head of Translational Genetics, The Cyprus Institute of Neurology and Genetics
The Veracity is a new Non-Invasive Prenatal Test, which was developed and validated to serve as an accurate and affordable screening test for trisomies 21, 18, and 13. Veracity is a novel targeted NIPT approach following the design of specific regions on chromosomes 13, 18, 21, X, and Y, capture and alignment of DNA fragments, and count and sophisticated analysis to achieve binary risk classification. The new test was validated by two blind independent clinical studies for the total of 706 samples, providing outstanding accuracy for trisomies 21, 18, and 13 gender determination. The Veracity test is available internationally as an affordable and very accurate screening test as of May 2015.

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

6:30 Close of Day

TUESDAY, NOVEMBER 17

8:00 Breakout Discussions with Continental Breakfast
Topics Include:
- Best Practices for Microarray and Sequence Analysis of Invasively-Obtained Samples
- The Economics of Prenatal Testing
- Ethical and Genetic Counseling Issues for Prenatal Diagnostics
- The Value of Determining Fetal Fraction with NIPT
- Pros and Cons of Offering NIPT for Lower-Risk Pregnancies
- Bioinformatics for NIPT Interpretation
- Testing for Single-Gene Disorders
- Evaluation of Microdeletions, Insertions, Copy-Number Variations and Translocations
- Challenges of Whole Genome Amplification for Small Samples of Fetal Cells
- Challenges and Prospects for Commercialization of Fetal Cell NIPT
- Biomarkers for Assessing the Risk of Preeclampsia and Pre-Term Labor

NON-INVASIVE PREGNANT DIAGNOSTICS (NIPD)
(Cont.)

8:55 Chairperson's Remarks
David H. Ledbetter, Ph.D., FACMG, Executive Vice President & CSO, Geisinger Health Systems

9:00 Advancement of Molecular Diagnostics in the Field of Non-Invasive Prenatal Diagnosis of Single Gene Disorders: Present Experiences and Future Developments
Michael Parks, Ph.D., Developmental Scientist, Regional Genetics, Birmingham Women’s NHS Foundation Trust (United Kingdom)
NIPT of aneuploidies using cell free fetal DNA is now an established method offered in many countries, including the US and the UK. Although research has proven that non-invasive prenatal diagnosis (NIPD) of single gene disorders is also possible, the relevant costs for these tests have been high. We have developed a molecular diagnostic method for NIPD of single gene disorders which has proven to be both accurate and affordable. After having successfully tested numerous patients at risk of bearing a child affected by Duchenne/Becker muscular dystrophy, we are now adapting our method to test patients for other single gene disorders. By being accurate, affordable and easily adaptable to detect most single gene disorders, our method could allow clinical genetics laboratories to offer safe and accurate NIPD of single gene disorders to their patients.

9:30 Prenatal Testing for Single Gene Disorders using Large Panels: A Case for Couple Screening
Glenn E. Palomaki, Ph.D., Associate Professor, Pathology and Laboratory Medicine, Women & Infants Hospital and the Alpert Medical School at Brown University
The field of prenatal screening is experiencing a rapidly expanding ability to simultaneously test for many single gene disorders. The introduction of such tests is hampered, however, by several implementation issues that may have at least a partial solution in offering such testing to couples rather than the mother then father in a long-standing sequential model. The methods and history of couple screening will be reviewed. Some of the issues relating to implementation
of newer approaches will also be discussed. Specific examples from currently offered tests will be given.

10:00 Beyond Aneuploidy: Prenatal Detection and Interpretation of Copy Number Variants (CNVs) That Cause Brain Disorders
David H. Ledbetter, Ph.D., FACMG, Executive Vice President & CSO, Geisinger Health Systems
Although individually rare, pathogenic CNVs are collectively common, occurring in ~1% of the general population and in low risk pregnancies. Many of these newly described CNV disorders are associated with significant cognitive (intellectual disability), behavioral (e.g., autism) and psychiatric (e.g., schizophrenia) manifestations and are therefore important for consideration in counseling regarding prenatal diagnostic options.

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

11:10 NIPT Vendors Panel Discussion
Moderator: Phillips Kuhl, President, Cambridge Healthtech Institute Panelists:
Sabah Oney, Ph.D., Director, Business Development, Ariosa Diagnostics
Geoffrey Henno, NIPT Market Development Manager, Illumina
Solomon Moshkevich, Vice President, Product & Strategy, Natera
Philippos C. Patsalis, Ph.D., Head, Translational Genetics, The Cyprus Institute of Neurology and Genetics on behalf of NIPT Genetics Ltd.
Peter Collins, Chief Commercial Officer, Premama Health
Douglas Rabin, M.D., Medical Director, Women's Health, Quest Diagnostics
Mathias Ehrich, Ph.D., Vice President, Research & Development, Sequenom

12:30 pm Luncheon Presentation (Sponsorship Opportunity Available) or Enjoy Lunch on Your Own
THE PROMISE AND PROSPECTS FOR FETAL CELLS FROM MATERNAL BLOOD

1:45 Chairperson’s Remarks
Arthur L. Beaudet, M.D., Chair, Department of Molecular & Human Genetics, Baylor College of Medicine

1:50 TRIC: A New Window into the Developing Placenta and Fetal Genome
D. Randall Arman, Ph.D., Professor, Obstetrics and Gynecology, Wayne State University School of Medicine
Safe access to fetal tissue during pregnancies is the “Holy Grail” of noninvasive prenatal testing. Trophoblast Retrieval and Isolation from the Cervix (TRIC) is a safe, noninvasive procedure that captures fetal placental cells as early as 3 weeks after conception, and holds promise for fetal genetic testing and assessment of maternal risk for obstetric complications. Characterization and performance of the process, which provides intact human genome for molecular analysis, will be presented. Because developmental errors in extravillous trophoblast cells contribute to preeclampsia, fetal growth restriction and miscarriage, genetic analysis of these cells from the first trimester can provide very informative diagnostic results.

2:20 Fetal Cell Capture for Using Micro-Fabricated MEMS Device for Non-Invasive Prenatal Diagnostics
Fanqing Chen, Ph.D., Chief Scientific Advisor, R&D, Basera, Inc.
Fetal cell isolation from maternal blood for non-invasive prenatal diagnosis presents various challenges due to the rarity of such cells. Various approaches have been attempted to extract and analyze such cells for downstream genetic analysis and diagnostic assays, but with limited and variable success. Results related to fetal cell capture using a micro fabricated MEMS device and integrated microfluidic chip as an alternative strategy to non-invasive prenatal diagnostics.

2:50 Recovery and Analysis of Intact Fetal Cells:
Non-Invasive Prenatal Diagnosis using the DEPArray
Farideh Bischoff, Ph.D., Executive Director, Scientific Affairs, Silicon Biosystems, Inc.

3:20 Refreshment Break in the Exhibit Hall with Poster Viewing

4:00 Enrichment and Genetic Analysis of Intact Circulating Fetal Cells (CFC) for Non-Invasive Prenatal Genetic Testing
David Deng, M.D., Ph.D., Chief Scientist, Next Generation Sequencing (NGS), Daan Gene Co., Ltd. of Sun Yat-Sen University
While NIPT based on sequencing of cell-free DNA has proven to be an effective non-invasive means to detect trisomies, using it for detection of many single gene or complex genetic diseases has been much more challenging. By applying unique cell stabilization reagents, intact fetal cells, which contain the entire and pure fetal genome, have been successfully isolated from all pre-term maternal samples tested as early as 6 ½ weeks of gestation. Preliminary clinical testing showed that intact CFCs were enriched from all samples tested, and tests indicated these cells are of fetal origin. The potential of using analysis of isolated fetal cells as an alternative or in addition cell-free DNA sequencing will be discussed.

4:30 Cell-Based Noninvasive Prenatal Diagnosis I: Recovery of Cells
Steen Kalvraa, M.D., Ph.D., ARCDI Biotech ApS (Denmark)
Many groups have over the years tried to develop methods for isolating sufficient fetal cells from maternal blood to perform NIPT, but lack of suitable markers has hampered these attempts. We have performed expression array analyses on isolated fetal cells and in this way indicated that a frequent fetal cell type in maternal blood is the endovascular trophoblast. We have used this data to identify two markers that identify fetal cells in maternal blood with very high specificity, facilitating efficient isolation of such fetal cells for non-invasive prenatal testing.

5:00 Cell-Based Noninvasive Prenatal Diagnosis II: Analysis of Cells
Arthur L. Beaudet, M.D., Chair, Department of Molecular & Human Genetics, Baylor College of Medicine
We are focused on developing a fetal cell-based method of NIPT as a routine clinical test. In collaboration with multiple companies, we are now able to molecularly confirm recovery of fetal cells on a regular basis. Using whole genome amplification of 1-3 cells, it is possible to perform array CGH with these fetal cells.

5:30 Progress in Isolation and Analysis of Fetal Nucleated Red Blood Cells
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

6:00 Close of Day
THE PROMISE AND PROSPECTS FOR FETAL CELLS FROM MATERNAL BLOOD (Cont.)

8:00 am Breakfast Presentation (Sponsorship Opportunity Available) or Morning Coffee

8:30 Chairperson’s Remarks

8:35 Targeting Pure Fetal DNA from Circulating Trophoblastic Cells for the Development of Prenatal Non-Invasive Diagnostics of Genetic Disorders: Advantages and Technical Aspects
Patrizia Paterlini-Brechot, Ph.D., Cellular and Molecular Biology, University of Paris Descartes, (France)

Next Generation Sequencing (NGS) of DNA from circulating trophoblastic cells is a rapid and cheaper approach for the direct and non-invasive prenatal diagnosis of aneuploidy and for exploring non-invasively a wide range of genetic disorders. Our team has shown the consistency of circulating trophoblastic cells recovery by using ISET and the interest of using the pure fetal DNA extracted from them for non-invasive prenatal diagnosis (NIPND). We show here that the application of NGS analysis to circulating trophoblastic cells opens new avenues and hopes for non-invasive prenatal diagnosis.

9:05 NanoVelcro Microchips for Isolation and Characterization of Circulating Fetal Nucleated Cells (CFNCs)
Hsian-Rong Tseng, Ph.D., Professor, Molecular and Medical Pharmacology, University of California, Los Angeles

A new non-invasive prenatal diagnostic (NIPD) technology capable of not only monitoring dynamic changes of circulating fetal nucleated cells (CFNCs) but also isolating CFNCs for prenatal genetic testing at early-stage of pregnancy has been developed at UCLA. Results from clinical evaluation of our NanoVelcro CFNCs enumeration and CFNCs genetic testing will be presented, along with potential implications for the impact of this approach on the field of prenatal molecular diagnostics.

10:05 Specific Detection of Preeclampsia at the Point-of-Care
Wendy Davis, CEO, GestVision, Inc.

Diagnosis of preeclampsia still relies on symptoms that are nonspecific for the condition, making diagnosis challenging. Recent advances have led to a highly specific urine test providing physicians with rapid information for determining patient status regarding preeclampsia.

12:15 pm Close of Conference
About the Conference:
Rapidly evolving technologies are transforming the way reproductive genetic diagnostics and screening are performed, but the differences, limitations, and benefits of these new technologies remain unclear to some. Cambridge Healthtech Institute’s Inaugural Reproductive Genetic Diagnostics conference aims to examine the latest technologies, including next-generation sequencing (NGS) and quantitative PCR, and their benefits and applications in carrier screening, preimplantation diagnostics, and product of conception (POC) testing. As these technologies develop and become widespread, we must understand their capabilities, where each technology is best applied, and be sure that they have been analytically and clinically validated. Furthermore, as the technology outpaces practices, we must take a critical eye to the ethical implications of the applications of these technologies, including the regions of the genome that should be sequenced and the types and amount of sequence variation that is necessary for diagnosis.

Sessions Include:
- Advances in NGS and Other Technologies
- Clinical Applications for Advanced Testing Technologies
- Embryo Preparation, Assessment, and Treatment
- Best Practices and Ethics

Keynote Speaker:
Joe Leigh Simpson, M.D.,
Senior Vice President for Research and Global Programs, March of Dimes Foundation
**WEDNESDAY, NOVEMBER 18**

**ADVANCES IN NGS AND OTHER TECHNOLOGIES**

12:30 pm Registration

2:00 Chairperson’s Remarks

Mark Umbarger, Ph.D., Director, Research and Development, Good Start Genetics

▷ 2:05 KEYNOTE PRESENTATION: Current and Expanding Indications for Preimplantation Genetic Diagnosis (PGD)

Joe Leigh Simpson, M.D., President for Research and Global Programs, March of Dimes Foundation

Where does PGD fit within the broader spectrum of prenatal genetic diagnosis? Sometimes either technology could be chosen, but in other circumstances PGD is uniquely appropriate. As desire increases to limit multiple gestations in ART, PGD to exclude aneuploid embryos and verify normalcy for euploid embryos will become progressively applicable.

2:35 Next-Generation Sequencing: Its Role in Reproductive Medicine

Brynn Levy, Professor, Pathology & Cell Biology at CUMC; Director, Clinical Cytogenetics Laboratory; Co-Director, Division of Personalized Genomic Medicine, College of Physicians and Surgeons, Columbia University Medical Center, and the New York Presbyterian Hospital

The introduction of microarrays into the clinical arena has shifted the way we look at chromosomes to a genomics-based view, offering greater resolution and new diagnostic categories such as UPD. NGS has rapidly become a part of the clinical testing menu, especially in pediatrics. However, its clinical utility in reproductive medicine remains an active area of investigation. This talk will focus on the benefits of the newer cytogenomic technologies that are being utilized for diagnostics in both the preimplantation and fetal stages of development.

3:05 CCS Without WGA

Nathan Treff, Director, Molecular Biology Research, Reproductive Medicine Associates of New Jersey; Associate Professor, Department of Obstetrics, Gynecology, and Reproductive Sciences, Rutgers-Robert Wood Johnson Medical School; Adjunct Faculty Member, Department of Genetics, Rutgers-The State University of New Jersey

It is well-established that WGA introduces artifacts when applied to human embryo biopsies for comprehensive chromosome screening (CCS). This presentation will describe an alternative strategy involving targeted multiplex qPCR which has undergone the most rigorous validation of any CCS method currently available. Comparison with WGA-based methods will also be presented demonstrating superiority in both preclinical accuracy and in the ability to combine single gene disorders and microdeletions and duplications with CCS.

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

4:15 Concurrent PGD for Single Gene Disorders and Aneuploidy on a Single Trophoderm Biopsy

Rebekah S. Zimmerman, Ph.D., FACMG, Director, Clinical Genetics, Foundation for Embryonic Competence

Many methods of comprehensive chromosome screening (CCS) involve whole genome amplification (WGA), making it difficult to obtain reliable PGD data for a single gene disorder (SGD) in parallel from a single biopsy. This study presents validation and clinical experience with an alternative approach involving multiplex qPCR.

4:45 Live Birth of Two Healthy Babies with Monogenic Diseases and Chromosome Abnormality Simultaneously Avoided by MALBAC-Based Combined PGD and PGS

Xiaoliang Sunney Xie, Ph.D., Mallinckrodt Professor, Chemistry and Chemical Biology, Harvard University

Preimplantation genetic diagnosis (PGD) and preimplantation genomic screening (PGS) help patients to select embryos without monogenic disorders or chromosome abnormalities. Our MALBAC work has proved that a normal embryo can be identified and selected by one-step genome sequencing to eliminate both chromosomal abnormality and point mutations causing monogenic diseases. Furthermore, we report here the first successful MALBAC babies using an improved method with significantly reduced false positives and false negatives.

5:15 Analytical Validation of a Novel NGS-Based Preimplantation Genetic Screening Technology

Mark Umbarger, Ph.D., Director, Research and Development, Good Start Genetics

We have developed and implemented a novel NGS-based PGS technology that utilizes a single PCR reaction to amplify repetitive elements on each chromosome while simultaneously attaching sequencing adapters and sample-specific barcodes for multiplexed NGS. In this talk, we will compare and contrast the workflow of our approach to that of other NGS-based PGS approaches, and will outline the results of an analytical validation study that evaluated the accuracy of our approach relative to array comparative genomic hybridization (aCGH).

5:45 Welcome Reception in the Exhibit Hall with Poster Viewing

6:45 Close of Day

**THURSDAY, NOVEMBER 19**

7:30 am Morning Coffee

**CLINICAL APPLICATIONS FOR ADVANCED TESTING TECHNOLOGIES**

7:55 Chairperson’s Remarks

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

8:00 Expanded Carrier Screening for Monogenic Disorders

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

Highly accurate, low-cost methods for the identification of mutations have facilitated identification of carriers of monogenic disorders. This presentation will review current recommendations, discuss the advantages of expanded carrier screening, and consider future prospects.

8:30 Oocyte Mitochondrial Function and Testing: Implications for Assisted Reproduction

Emre Seli, M.D., Yale School of Medicine

Mitochondrial function has been associated with oocyte function, 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.

9:00 Preventing the Transmission of Mitochondrial Diseases through Germline Genome Editing

Alejandro Ocampo, Ph.D., Research Associate, Gene Expression Laboratory – Belmont, Salk Institute for Biological Studies
We have recently developed a novel strategy towards preventing the germline transmission of mitochondrial diseases through the selective elimination of mutated mtDNA using mitochondria targeted restriction endonucleases or TALENs. We are now evaluating the human safety and efficacy of this technology to prevent the transmission of human mitochondrial diseases.

9:30 Recovery and Analysis of Single (Fetal) Cells: DEPArray-Based Strategy to Examine CPM and POC

Farideh Bischoff, Ph.D., Executive Director, Scientific Affairs at Silicon Biosystems, Inc.

9:45 Sponsored Presentation (Opportunity Available)

10:00 Coffee Break in the Exhibit Hall with Poster Viewing

10:40 Numerical Chromosomal Abnormalities after PGS and D&C

Tanmoy Mukherjee, M.D., Assistant Clinical Professor, Obstetrics, Gynecology and Reproductive Science, Mount Sinai Hospital

This review provides an analysis of the most commonly identified numerical chromosome abnormalities following PGS and first trimester D&C samples in an infertile population utilizing ART. Although monosomies comprised >50% of all cytogenetic anomalies identified following PGS, there were few very identified in the post D&C samples. This suggests that while monosomies occur frequently in the IVF population, they commonly do not implant.

EMBRYO PREPARATION, ASSESSMENT, AND TREATMENT

11:10 Chairperson’s Remarks

Catherine Racowsky, Professor, Department of Obstetrics, Gynecology & Reproductive Biology, Harvard Medical School; Director, IVF Laboratory, Brigham & Women’s Hospital

11:15 Guidelines and Standards for Embryo Preparation: Embryo Culture, Growth, and Biopsy Guidelines for Successful Genetic Diagnosis

Michael A. Lee, MS, TS, ELD (ABB), Director of Laboratories, Fertility Solutions

This presentation will discuss the basics of state-of-the-art in vitro fertilization and embryo culture and embryology laboratory techniques. We will review laboratory conditions to maximize oocyte fertilization and embryo culture to produce optimum embryos for biopsy, as well as preparation of embryos for biopsy and post-biopsy culture and vitrification of techniques and protocols.

11:45 Current Status of Time-Lapse Imaging for Embryo Assessment and Selection in Clinical IVF

Catherine Racowsky, Professor, Department of Obstetrics, Gynecology & Reproductive Biology, Harvard Medical School; Director, IVF Laboratory, Brigham & Women’s Hospital

It is well established that conventional morphological assessment is by no means a perfect method for predicting viability of human embryos. This talk will assess the utility of time-lapse imaging as an alternative approach for embryo assessment. The benefits and limitations of current time-lapse data will be reviewed and the current status of this imaging technology for selecting the most viable embryo for transfer in clinical IVF will be considered.

12:15 pm The Curious Case of Fresh Versus Frozen Transfer

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

The lecture will discuss the historical differences in outcomes between fresh versus frozen transfers, including how outcomes and, in particular, how live birth weights differ. A rationale of when it is safe to perform a fresh or frozen transfer will also be discussed.

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

2:05 Why Does IVF Fail? Finding a Single Euploid Embryo is Harder than You Think

Jamie Grifo, M.D., Ph.D., Program Director, New York University Fertility Center; Professor, New York University Langone Medical Center

This talk will focus on chromosomal abnormalities in embryos, the different factors that affect them, and how they contribute to IVF failure. Dr. Grifo will review the published literature as well his own and describe an optimal approach to IVF that limits risk and maximizes benefit.

BEST PRACTICE AND ETHICS

2:30 Chairperson’s Remarks

Mache Seibel, M.D., Professor, OB/GYN, University of Massachusetts Medical School; Editor, My Menopause Magazine; Author, The Estrogen Window
2:35 Genetic Counseling Bridges the Gap between Complex Genetic Information and Patient Care
Mary Ann W. Campion, EdD, MS, CGC, Director, Master’s Program in Genetic Counseling; Assistant Dean, Graduate Medical Sciences; Assistant Professor, Obstetrics and Gynecology, Boston University School of Medicine

In this domain, ethical issues abound, including barriers to informed consent, duty to warn, associated costs (to the healthcare system and to the patient), and controversial indications for testing.

3:05 Ethical Issues of Next-Generation Sequencing and Beyond
Eugene Pergament, M.D., Ph.D., FACMG, Professor, Obstetrics and Gynecology, Northwestern; Attending, Northwestern University Medical School Memorial Hospital

This presentation on the ethical considerations of next-generation sequencing and related technologies will address the current status and future prospects of three critical issues. Does the introduction of these technologies into clinical practice in the United States: 1) Raise new ethical issues concerning preimplantation genetic testing? 2) Facilitate preimplantation genetic therapies? And, if so, 3) What should be the roles and responsibilities of local, state, and federal governments, of various medical societies, and of individual programs providing preimplantation genetic services?

3:35 Session Break

3:45 Closing Panel: The Future of Reproductive Genetic Diagnostics: Is Reproductive Technology Straining the Seams of Ethics?

Moderator: Mache Seibel, M.D., Professor, OB/GYN, University of Massachusetts Medical School; Editor, My Menopause Magazine; Author, The Estrogen Window
Panelists: Rebekah S. Zimmerman, Ph.D., FACMG, Director, Clinical Genetics, Foundation for Embryonic Competence
Denny Sakkas, Ph.D., Scientific Director, Boston IVF
Michael A. Lee, MS, TS, ELD (ABB), Director of Laboratories, Fertility Solutions
Nicholas Collins, MS, CGC, Manager, Reproductive Health Specialists, Counsyl

Benjamin Franklin said, “An ounce of prevention is worth a pound of cure.” Reproductive genetic diagnostic tools and tests are evolving at the speed of light. Are we able to keep up with the practical and ethical implications of this technology? Join this panel of experts who will grapple with this question and others such as:

- Where is this technology going? What is the next evolutionary step?
- What are the biggest challenges scientists, clinicians, and counselors face with diagnostic tools – and the information we gather – today?
- Where do our responsibilities lie in the treatment of embryos before and after treatment?

4:30 Close of Conference

HOTEL & TRAVEL INFORMATION

CONFERENCE VENUE AND HOST HOTEL:
Omni Parker House Hotel
60 School Street
Boston, MA 02108
Phone: 1-800-THE-OMNI

Discounted Room Rate: $255 s/d
Discounted Cut-off Date: October 16, 2015

Reservations: Go to the travel page of healthtech.com/prenatal-diagnostics

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

Why Stay at the Omni Parker House?
This grand luxury hotel has been symbolic to Boston’s rich history and culture since 1855. Old-World charm and elegance are accompanied by all of the modern conveniences of a world-class establishment. Nestled in the heart of downtown Boston, Omni Parker House is located along the Freedom Trail and at the foot of Beacon Hill, Boston Common, Quincy Market and Faneuil Hall marketplace. Omni Parker House is just 2.3 miles from Logan International Airport.
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<tr>
<th>WHO CAN YOU EXPECT TO NETWORK WITH IN BOSTON?</th>
<th>Partial list of past attendees</th>
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### AGENDA

- **Inaugural Reproductive Genetic Diagnostics**
  - Childrens Hospitals & Clinics, Exec Dir & VP of Operations
  - Cleveland Clinic Foundation, Obstetrics & Gynecology
  - ClinicalValues, Head
  - Colorado Ctr for Reproductive Medicine, Genetic Counselor
  - Columbus Univ, Prof, Obstetrics & Gynecology
  - Columbus Regional Healthcare System, MFM
  - CombiMatrix Diagnostics, Director, Cytogenetics
  - CombiMatrix Diagnostics, President & CEO
  - Complete Genomics Inc, Sr Staff Scientist
  - Coombe Womens Hospital, Histopathology
  - Cornell Univ, Assoc Prof, Pathology & Lab Medicine
  - Cornell Univ, Genetic Counselor
  - Counsyl Inc, Genetic Counselor
  - Counsyl Inc, Natl Dir Genetic Counselors & Mgr
  - Ctr for Medical Genetics, Medical Dir
  - Ctr Hospitalier Univ, Head, Lab & Human Genetics
  - CytoGenX, Clinical Lab Dir
  - Dartmouth Hitchcock Medical Ctr, Genetic Counselor
  - Devsyr AB, CEO
diaDexus Inc, CEO
Drexel Univ, Prof, Obstetrics & Gynecology
Duke Univ, Asst Research Prof
Enterprise Partners Venture Capital, Managing Dir Life Sciences
Erasmus Univ, Clinical Genetics
FDA CDER, Supv Biologist, OIR CDRH
FDA CDRH, Scientist, OIR
Fertiliy, President & CEO
Fetal Life Science Ctr Co Ltd, Emeritus Prof, Obstetrics & Gynecology
Fetal Medicine Foundation USA, Dir of Re Accreditation
Fiocruz, IBMP
Foundation for Embryonic Competence
Foundation Medicine Inc, Advisor
Fox Rothschild LLP, Counsel, Intellectual Property
Fundacion Jimenez Diaz, Asst to Head

- **Final Weeks to Register**
  - Aetna Inc, Sr Medical Dir & Head
  - Affymetrix Inc, COO & Exec VP
  - Affymetrix Inc, Sr VP Global Clinical Strategic Marketing
  - Agilent Technologies Inc, Dir R&D
  - Agilent Technologies Inc, Dir Sales
  - American Hospital of Paris, Prenatal Diagnosis
  - Ariosa Diagnostics Inc, Assoc Dir Medical Affairs
  - Ariosa Diagnostics Inc, CEO
  - Ariosa Diagnostics Inc, Lead Medical Science Liaison
  - ARUP Labs, Medical Lab Dir Cytogenetics
  - Bain Capital LLC
  - Baylor College of Medicine, Chairman & Prof
  - Baylor College of Medicine, Prof Obstetrics & Gynecology
  - Beijing United Family Hospital, OB & GYN
  - Berry Genomics Co Ltd, CEO
  - Bonei Olam, Dir Genetic Counseling
  - BORN Ontario, Screening Specialist
  - Boston Maternal Fetal Medicine, Obstetrician
  - Boston Univ, Dir Genetic Counseling Program
  - Brandeis Univ, Dir Genetic Counseling Program
  - Bridgeport Hospital, Dir Maternal Fetal Medicine
  - Brigham & Womens Hospital, Prof, Obstetrics Gynecology & Reproductive Biology
  - Carilion Clinic Maternal Fetal Medicine, Genetic Counselor
  - Carmenta Bioscience, CEO
  - Cartagenia Inc, Product Marketing Dir
  - Cellscape Corp, Chief Geneticist
  - Cellscape Corp, CMO
  - Cellscape Corp, Founder & CTO
  - CEMIC, Geneticist
  - Chemical & Engineering News, Sr Editor
  - Childrens Hospital Boston, Telemedicine & Telehealth
  - Childrens Hospital of Eastern Ontario, Clinical Geneticist
  - Childrens Hospital of Philadelphia, Asst Clinical Prof OB & GYN
  - Geisinger Health System, Dir & Sr Investigator
  - Geisinger Health System, Dir Policy & Education
  - Geisinger Health System, Exec VP & CSO
  - GenePeeks, CEO
  - GenePeeks, Chief Scientific Officer
  - Genesis Genetics Asia Corp, CEO
  - Genesis Genetics LLC, Lab Founder
  - Genetics & IFV Institute, Genetic Counselor
  - Genia Technologies, COO
  - GenoLogics Life Sciences Software Inc, Mgr
  - GenomeWeb LLC, Editor
  - Genomic Health Inc, Sr Dir Patient Advocacy
  - Ghenr Univ, Pharmaceutical Sciences
  - GNS Healthcare, CEO & President & Co Founder
  - Good Start Genetics Inc, Founder VP Research & Technology
  - Good Start Genetics Inc, Medical Dir & VP Lab Operations
  - Guangdong Women & Children Hospital, Physician, Prenatal Diagnosis Ctr
  - Guangdong Women & Children Hospital, Prof & Head, Clinical Genetic Ctr
  - Hangzhou GeonaX Biotech Co, CEO
  - Harvard Univ, Assoc Dir
  - Harvard Univ, Mallinckrodt Prof
  - Healthcor Partners Mgmt LP, Managing Director
  - Hellenic Open Univ, Researcher
  - High Risk Pregnancy Ctr
  - Hitachi High Technologies, Dir Life Science Div
  - Hitachi Solutions America Ltd, Dir Life Science
  - Hogan Lovells US LLP, Partner
  - Hospital Foch, Prof Hematology
  - Hvidovre Hospital, Assoc Prof & Consultant, OB GYN
  - Illumina Inc, Dir Strategic Partnerships
  - Illumina Inc, Translational & Consumer Genomics
  - Illumina Inc, VP Sales & Market Dev
  - Inova Translational Medicine Institute, Postdoc Research Fellow
  - Insight Medical Genetics, President & Managing Member

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Institute of Prenatal Diagnosis & Reproductive Genetics, Maternal Fetal Medicine
Integrated Genetics, Sr VP & GM
Intermountain Healthcare, Genetic Counselor
Intl Peace Maternity & Child Health Hospital, Assoc Prof, Prenatal Diagnosis Ctr
Ironwood Pharmaceuticals Inc, Assoc Dir Bus Dev & Licensing
Istanbul Univ, Prof
IWK Health Ctr, Cytogeneticist & Div Head
Kaiser Permanente, Obstetrics & Gynecology
Kazusa DNA Research Institute, Dir
KellBenzx Inc, CEO
KellBenzx Inc, President & Chairman
KingMed Diagnostics, Dean & Dir
Kyoto Univ, Prof
LabCorp, President
LabCorp, Strategic Dir Cytogenetics
LabPLUS, Prenatal Team & Diagnostic Genetics
London Health Sciences Ctr, Clinical Biochemist
Long Island Jewish Medical Ctr, Obstetrics & Gynecology
Luminex Corp, Sr Marketing Mgr
March of Dimes, Sr VP Research & Global Programs
Marshfield Clinic, Dir Medical Genetics
Massachusetts Down Syndrome Congress, Family Support Dir
Massachusetts General Hospital, Asst Prof Neurology
Mayo Clinic & Foundation, Co Dir Molecular Genetics Lab & Assoc Prof
MedStar Washington Hospital Ctr, Fellow, Maternal Fetal Medicine
Metabolomic Diagnostics Inc, VP R&D
Minnesota Perinatal Physicians PA
Mizmedi Hospital, Obstetrics & Gynecology
Montefiore Medical Ctr, Pathologist Asst
Mount Sinai Hospital, Genetic Counselor
Mount Sinai Hospital, Maternal Fetal Medicine
NantHealth
Natera Inc, CMO
Natera Inc, Sr Dir R&D & Intellectual Property
Natera Inc, Sr VP Commercial Operations
Natl Univ of Singapore, Obstetrics & Gynaecology
NERGG Inc, Exec Dir
New England BioLabs Inc, Assoc Dir Bus Dev
New England Obstetrics & Gynecology Assocs, Genetic Counselor
Newton Wellesley Hospital, Certified Genetic Counselor
Northwestern Univ, Prof & Dir
Oxford Gene Technology, Exec VP R&D
Oxford Gene Technology, VP
PacGenomics, CEO & President
Pacific Ctr for Reproductive Medicine
PCRM, Physician
Path Group, Dir Cytogenetics & Array Svcs
Pathology Associates Medical Labs PAML, Genomics Mgr
Peking Union Medical College Hospital, Prof, OB & GYN
Perinatal Associates of Northern Nevada, Genetic Counselor
PerkinElmer, Dir, Molecular Cytogenetics
PerkinElmer, Sr Scientist, Molecular Diagnostics
PerkinElmer Life & Analytical Sciences, VP
Maternal Fetal Health
Phoenix Childrens Hospital, Sr VP & Chief Research Officer
Prenatal Medizin Muenchen, Obstetrics & Gynecology
Preeclampsia Foundation, Exec Dir
Prenal Health, CEO
Prenal Health, CMO
Prenatal Diagnosis of Northern California, Medical Dir
Progenity Inc, CMO
Pura Vida Investments, Analyst
Quest Diagnostics Inc, Dir Marketing
Quest Diagnostics Inc, Exec Dir Bus Dev
QUIDEL Corp, CSO
RA Capital Mgmt LLC, Analyst
Radboud Univ Nijmegen, Clinical Cytogeneticist
RainDance Technologies Inc, Dir Genomic Applications
RARECELLS, Founder & CSO
RareCyte Inc, CMO
Reproductive Genetic Innovations LLC, President & PGD & IVF Dir
Rosetta Genomics Inc, CMO
Sonic Healthcare, VP Accounts & Payor Strategies
SEQUENOM Inc, Sr Dir MDx Dev & VP
SEQUENOM Inc, Sr VP R&D
SeraCare Life Sciences Inc, CSO
SeraCare Life Sciences Inc, VP Precision Medicine
Shire Pharmaceuticals, Sr Dir Process Optimization
Siemens Healthcare Diagnostics, Diagnostic & Technology Transfer
Silicon Biosystems, Dir Marketing Dev
Silicon Biosystems, Project Leader
SingleBio, VP Assay Dev
Siriraj Hospital, OB & GYN
SK Telecom, Project Leader
South Shore Healthcare, Scientific Dir, Prenatal Testing
South Shore Hospital, Chairman, Obstetrics & Gynecology
South Shore Hospital, Perinatal Counselor
Southern General Hospital, Medical Genetics
St Elizabeth Healthcare, OB GYN
Swedish Perinatal Medicine Clinic, Medical Dir
TessArae LLC, Dir
Thermo Fisher Scientific Inc, Head Clinical Alliances
Toho Univ, Obstetrics & Gynecology
Trillium Health Partners, Medical Dir
Tufts Medical Ctr, Prof, Pediatrics
Univ of Adelaide, Sr Research Fellow, Obstetrics & Gynecology
Univ of Auckland, Prof, Maternal Fetal Medicine
Univ of Florida Gainesville, Prof & Dir, Obstetrics & Gynecology
Univ Of Hong Kong, Prof, Biochemistry
Univ of Indonesia, Head, Fetomaternal Div
Univ of Iowa, Program Manager Medical Screening
Univ of Kansas, Krantz Chair, OB & GYN
Univ of Miami, Assoc Chief Genetics & Genomics
Univ of Montreal, Dir Ethics & Health Branch & Assoc Prof
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Ilana Quigley  |  Sr. Business Development Manager
781-972-5457 | iquigley@healthtech.com
### PRICING & REGISTRATION

#### CONFERENCE PRICING

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#### PREGNATAL MOLECULAR DIAGNOSTICS CONFERENCE PRICING

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#### REPRODUCTIVE GENETIC DIAGNOSTICS CONFERENCE PRICING

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