Final Weeks to Reserve Your Place!

Cell-free DNA-based screening and the potential for cell-based screening has upended the field of prenatal testing. While cell-free tests are being used more and more in the clinic, cell-based tests are still on the cusp of commercialization. With both types of tests still in need of improvements in order to truly replace invasive tests, the near-term challenge is determining where the field heads once both options are viable. This meeting will discuss updated comparisons and examine the implementation, insurance, and clinical challenges associated with each test.

Furthermore, with advances in DNA amplification and sequencing comes greater opportunity to garner more information. Much discussion needs to take place in regards to the role of preconception counseling, expanded carrier screening, whole genome versus whole exome sequencing, and the effect these and any prenatal testing may have on the decision to continue a pregnancy or perform fetal therapy.

Final Agenda

Day 1 | Day 2 (http://www.healthtech.com/prenatal-diagnostics/#Day2) | Speaker Biographies (/pndx_pidx_Content.aspx?id=161197#PNDX) | Download Brochure (http://www.healthtech.com/pndx_pidx_Content.aspx?ekfrm=181183)

TUESDAY, NOVEMBER 28

8:00 am Registration and Morning Coffee

FETAL WHOLE EXOME SEQUENCING

9:00 Chairperson's Remarks
Art Beaudet, M.D., Department of Molecular & Human Genetics, Baylor College of Medicine

9:05 Prenatal DNA Sequencing: Clinical, Counseling, and Diagnostic Laboratory Considerations

Ahmad N. Abou Tayoun, Ph.D., Assistant Professor, Pathology and Laboratory Medicine, Children's Hospital of Philadelphia, University of Pennsylvania Perelman School of Medicine
Whole genome and exome sequencing on fetal is starting to be offered clinically in specialized centers, but it has not yet become routine practice. The technical, interpretation, and ethical challenges are greatest in the area of prenatal medicine because the fetus has a limited health history, and the physical examination is only indirectly available via prenatal sonography. This talk will describe an overview of these challenges and highlight the clinical utility, reporting, and counseling issues associated with prenatal DNA sequencing, as well as future considerations.

9:35 Prenatal Exome Sequencing in Anomalous Fetuses: New Opportunities and Challenges

Neeta Vora, M.D., Associate Professor, Department of OB GYN, Division of Maternal-Fetal Medicine, University of North Carolina Chapel Hill

(/Neeta_Vora_Podcast/)

Exome sequencing has utility in determining an underlying molecular etiology when performed on fetal specimens from pregnancies with structural abnormalities where standard genetic testing (karyotype and microarray) do not provide a diagnosis. Challenges related to genetics literacy and variant interpretation must be addressed by highly tailored pre- and post-test genetic counseling.

10:05 Implementation Considerations for Fetal Whole Exome Sequencing

Ignatia B. Van den Veyver, M.D., Professor, Obstetrics & Gynecology and Molecular Human Genetics, Baylor College of Medicine
Fetal diagnostic exome sequencing has the potential to significantly improve the identification of the genetic cause of fetal abnormalities detected on ultrasound imaging and in high-risk families. The responsible and effective implementation of fetal diagnostic exome sequencing into prenatal care is complex and still at early stages. Overview of current knowledge, research needs and case studies will be presented to highlight ethical, practical and counseling issues that must be considered.

10:35 Networking Coffee Break

10:55 WES for Recurrent Pregnancy Loss

Evica Rajcan-Separovic, Clinical Professor, University of British Columbia, Fellow Canadian College of Medical Genetics (Cytogenetics); Laboratory Scientist, Pathology and Laboratory Medicine, Children's and Women's Hospital of British Columbia
Epidemiological evidence suggests that genetic factors play a significant role in pathogenesis of miscarriage, and that both the fetal/placental and the parental genotypes are involved. The majority of miscarriages are sporadic; however, ~3-5% of couples trying to have children experience recurrent miscarriage. My talk will highlight advances in approaches to help diagnosis of recurrent miscarriage by identifying genetic abnormalities in miscarriages and couples using high resolution genomic technologies.

11:25 Effect of Maternal Cell Contamination on Prenatal NGS Testing

Heather Mason-Suares, Ph.D., FACMG, Assistant Professor, Associate Director, Pathology, Laboratory for Molecular Medicine and Brigham & Women's Hospital, Cytogenetics Laboratory
Maternal cell contamination (MCC) poses a significant risk for prenatal misdiagnosis in molecular diagnostics. However, the effect of MCC on the interpretation of NGS results is not well studied. Such characterization is extremely important as NGS is rapidly becoming the standard of care in prenatal molecular diagnostics for high risk pregnancies. This talk examines how MCC may confound NGS testing, causing erroneous interpretation of clinical results and affecting pregnancy management.

(https://www.qiagen.com/us/)11:55 Introduction of Process Automation for the Quality Improvement of NIPT as Exemplified by the PrenaTest®

Wera Hofmann, CSO, LifeCodexx
The worldwide increasing use of non-invasive prenatal testing (NIPT) in clinical practice, the growing regulatory requirements and the national efforts for reimbursement are enforcing the need for further improvements of the applied methods. Pre-analytical laboratory processes such as automated solutions for cfDNA extraction to allow higher throughput and improved reproducibility of the method at lower cost are of high importance. The example of PrenaTest® describes such a successful development.

12:25 pm Luncheon Presentation *(Sponsorship Opportunity Available)* or Enjoy Lunch on Your Own

12:55 Session Break

CELL-FREE DNA SCREENING

1:55 Chairperson's Remarks
Patrizia Paterlini-Brechot, Ph.D., M.D., Cellular & Molecular Biology, University Paris Descartes

2:00 An Examination of PreSeek, a Non-Invasive Multi-Gene Sequencing Screen

Christine Eng, M.D., CMO, Chief Quality Officer, Baylor Genetics; Professor, Molecular and Human Genetics, Baylor College of Medicine
The PreSeek development team carefully selected genes for this non-invasive single gene detection platform by a thorough curation process focused on the detection of de novo variants in single gene disorders affecting the skeletal, cardiac, and neurological systems. Although traditional NIPT detects abnormalities that increase in risk with advanced maternal age, PreSeek is the first non-invasive test to detect disorders that may become more prevalent with advanced paternal age. Early clinical experience with this test demonstrates the use of this test in several different clinical situations including fetuses with ultrasound abnormalities.

2:30 (Mis)adventures in NIPT Confirmatory Testing

Stephen R. Moore, MBA, Ph.D., FACMG, Lab Director, Cytogenetics and Molecular Diagnostics; Assistant Professor, Molecular and Medical Genetics, Knight Diagnostics Labs, Oregon Health and Science University
Current recommendations are that all positive non-invasive prenatal testing (NIPT) be confirmed by one of two invasive tests, chorionic villi sampling or amniocentesis. There are many factors, technical and biological, that may lead to discordance between NIPT results and the result of the confirmatory test. This talk will outline such factors and provide examples from our own experience as a confirmatory testing center.

3:00 Extending the Scope of Prenatal Diagnosis for Monogenic Disorders: Non-Invasive Prenatal Diagnosis

Lyn Chitty, Ph.D., MBBS, MRCOG, Professor, Genetics and Genomic Medicine, UCL Great Ormond Street Institute of Child Health and North-East Thames Regional Genetics Service, Great Ormond Street Hospital for Children NHS Foundation Trust
Traditional prenatal diagnosis has involved invasive tests. The analysis of cell free DNA in maternal plasma has led to with widespread introduction of less invasive testing for aneuploidy, but less so for monogenic disorders. In this presentation I will explain how, in our accredited public service genetics laboratory, we have developed a comprehensive diagnostic service for the non-invasive prenatal diagnosis of monogenic disorders. Such that we now deliver >30% of genetic diagnosis using NIPD and for more than 50 different genetic conditions.

3:30 Refreshment Break in Exhibit Hall with Poster Viewing

4:10 Cell-Based Non-Invasive Prenatal Diagnosis by Capturing Cytotrophoblasts and Fetal Nucleated RBC by Nanostructured Microfluidics and Its Comparison with In-House Developed cfDNA Testing

Ming Chen, M.D., Ph.D., CEO, Department of Genomic Science and Technology, Changhua Christian Hospital Healthcare System, Taiwan; Adjunct Associate Professor, Department of Obstetrics and Gynecology, National Taiwan University, Taiwan; Honorary Co-Founder, Cytoaurora Biotechnologies, Inc., Hsinchu Science Park, Taiwan; Honorary CSO, Golden Meditech Holdings Limited (HKSE)
We collaborated with the semiconductor and AI sectors to produce an automated system based on nanostructure, microfluidics, and user-friendly computer analysis software, of which the following steps are automatically processed: capturing both the fetal nucleated RBCs and cytotrophoblasts, machine learning identification, followed by the isolation of the captured cells. The isolated cells can then be subjected to WGA and the subsequent aCGH or NGS analyses. The *in situ* captured cells can be subjected to FISH analysis. We also compared the results of this cell-based system to our in-house devised cfDNA testing (the algorithm called "GWNS") in our CAP-certified core NIPT lab. The advantage of this "Cell Reveal" system is it may solve the problem of fetoplacental mosaicism, and can possibly revive the field of traditional cytogenetics.

4:40 Panel Discussion: Cell-Free vs. Cell-Based NIPT
Art Beaudet, M.D., Department of Molecular & Human Genetics, Baylor College of Medicine
Niels Uldbjerg, PhD, Clinical Professor, Obstetrics & Gynecology, Aarhus University
Ming Chen, M.D., Ph.D., CEO, Department of Genomic Science and Technology, Changhua Christian Hospital Healthcare System, Taiwan; Adjunct Associate Professor, Department of Obstetrics and Gynecology, National Taiwan University, Taiwan; Honorary Co-Founder, Cytoaurora Biotechnologies, Inc., Hsinchu Science Park, Taiwan; Honorary CSO, Golden Meditech Holdings Limited (HKSE)
With cell-based noninvasive prenatal testing coming closer and closer to commercialization, those in industry and in the clinic need to consider how these tests differ, their advantages and limitations, and which is the best course to take for patients. Panelists will discuss scientific, insurance and reimbursement, and ethical considerations to take into account.

5:40 Networking Reception in Exhibit Hall with Poster Viewing

7:10 Close of Day

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WEDNESDAY, NOVEMBER 29

8:00 am Breakfast Breakout Roundtable Discussions

Fetal Exome Sequencing
Ignatia B. Van den Veyver, MD, Professor, Obstetrics & Gynecology and Molecular Human Genetics, Baylor College of Medicine
- Should it already be done clinically, and if so, when, where and by whom?
- What are the pre-test and post-test counseling requirements?
- What types of results should and should not be reported?

NIPT in the Asian Market
Ming Chen, MD, PhD CEO, Dept. Genomic Science and Technology, Changhua Christian Hospital Healthcare System, Taiwan; Adjunct Associate Professor, Dept. Obstetrics and Gynecology, National Taiwan University, Taiwan; Honorary Co-Founder, Cytoaurora Biotechnologies Inc., Hsinchu Science Park, Taiwan; Honorary CSO, Golden Meditech Holdings Limited (HKSE)
- Differences between the US and Asian markets: societal acceptance/understanding
- How these differences affect research and new discoveries
- How these differences affect clinical use

Biomarkers for Preeclampsia
Rachel Kelly BSc(Hons) MPH PhD, Research Fellow, Channing Division of Network Medicine, Brigham and Women's Hospital Harvard Medical School
- Is there a role for molecular biomarkers in preeclampsia diagnosis?
- How could novel biomarkers impact mortality and morbidity?
- Where are biomarkers needed most: prediction, diagnosis, prognosis, or endotyping?

Education & Counseling for NIPT
Katie Stoll, MS, LGC, Director, Clinical Services, Genetic Support Foundation
- What are the greatest challenges in providing proper education and counseling for patients?
- How can commercial companies contribute – or not contribute – to proper education?
- What are some strategies you have implemented or would like to see implemented in your practice?

Commercialization Challenges for Fetal Cell-Based NIPT
Patrizia Paterlini-Brechot, Ph.D., M.D., Cellular & Molecular Biology, University Paris Descartes
- What are the scientific and technical roadblocks to developing a commercial cell-based NIPT?
- What are the competitive requirements and expected benefit ?
- What are the insurance and/or regulatory barriers?
- What are the clinical implications for when a cell-based NIPT is commercialized?

ISOLATION AND ANALYSIS OF FETAL CELLS FROM MATERNAL BLOOD

9:00 Chairperson's Remarks
Heather Mason-Suares, Ph.D., FACMG, Associate Director, Pathology, Laboratory for Molecular Medicine and Brigham & Women's Hospital, Cytogenetics Laboratory

9:05 Reasons for the Elusiveness of Cell-Based NIPT

Art Beaudet, M.D., Department of Molecular & Human Genetics, Baylor College of Medicine
Researchers have struggled to develop clinical testing in the form of cell-based NIPT. The rarity of fetal cells in the mother's blood is perhaps the biggest challenge. At least three forms of testing are desirable: 1) detection of inherited Mendelian disorders, 2) genome-wide detection of copy number abnormalities at the highest possible resolution, and 3) genome-wide detection of *de novo* point mutations. Various combinations of methods will be required to achieve all of these goals.

9:35 Technical Advances for Isolation and Genetic Analysis of Circulating Trophoblastic Cells

Patrizia Paterlini-Brechot, Ph.D., M.D., Cellular & Molecular Biology, University Paris Descartes
Circulating fetal cells offer an interesting opportunity to analyze fetal DNA not mixed with maternal DNA aiming to develop a non-invasive approach for prenatal genetic diagnosis (NI-PND). Critical issues for this goal are the number of fetal cells which can be recovered from a blood sample, the purity of cell recovery, the quality of the recovered fetal cells DNA and the assay workflow allowing to develop a high-throughput analysis generating reliable results at a very affordable price. We will show results using the ISET patented method to isolate trophoblastic cells without the use of antibodies and analyze their DNA for non-invasive prenatal diagnosis. We will discuss the different critical issues and the possible solutions in order to bring to the market a new test for NIPND.

10:05 Advances in Cell-Based Non-Invasive Prenatal Diagnosis

Ripudaman Singh, Ph.D., COO, ARCEDI Biotech Aps
In the last few years, advances in fetal cell enrichment and detection technologies have invigorated interest in using these rare cells for cell-based non-invasive prenatal diagnosis (cbNIPD). By using a proprietary technology, we have shown that we can isolate fetal cells from every pregnant sample and use the DNA from isolated fetal cells to detect chromosomal and sub-chromosomal changes in the fetal genome. The results from the cbNIPD were verified by the results from chorionic villi sampling. Having performed a preliminary study for implementing our method in a clinical setup, we are in the process of launching a cell-based clinical test in Denmark. In this test, results from the cell-based prenatal analyses on high risk pregnancies will be compared with cell-free non-invasive prenatal testing (cfNIPT). The aim of this clinical test will be to replace cfNIPT with a more superior alternative, based on fetal cells from maternal blood.

10:35 Coffee Break in Exhibit Hall with Poster Viewing

11:15 Imprinted NanoVelcro Microchips for Isolation and Characterization of Circulating Fetal Trophoblasts – Toward Noninvasive Prenatal Diagnostics

Shuang Hou, Ph.D., Senior Research Scientist, Department of Surgery, University of California, Los Angeles
Circulating fetal nucleated cells (CFNCs) in maternal blood offer an ideal source of fetal genomic DNA for noninvasive prenatal diagnostics (NIPD). We developed a new class of NanoVelcro Microchips to effectively enrich a subcategory of CFNCs, i.e., circulating trophoblasts (cTBs) from maternal blood. Our results support the use of NanoVelcro Microchips for cTB-based noninvasive prenatal genetic testing, which holds potential for further development toward future NIPD solution.

11:45 TRIC: Safe Prenatal Testing with Pap Smears to Interrogate the Fetal Genome and Pregnancy Health

D. Randall Armant, Ph.D., Professor, Obstetrics and Gynecology, Wayne State University School of Medicine
Trophoblast Retrieval and Isolation from the Cervix (TRIC) is a safe, noninvasive procedure that captures fetal cells migrating from the placenta as early as three weeks post-conception. TRIC holds promise for prenatal genetic testing and risk assessment of obstetrical complications. Isolated trophoblast cells provide fetal DNA for comprehensive analysis of the fetal genome. Additionally, their molecular profiles are associated with subsequent onset of preeclampsia, fetal growth restriction and miscarriage.

EXPANDED CARRIER SCREENING

12:15 pm Expanded Carrier Screening: Diagnostic Yield and Unexpected Findings
Lisa Edelmann, Ph.D., FACMG, Associate Professor, Department of Genetics and Genomic Sciences at the Icahn School of Medicine at Mount Sinai

Expanded carrier screening has the highest yield when a sequence-based approach is used to interrogate the coding regions of genes with supplementation by additional methodologies for regions of the genome in which sequencing is not adequate. The infrastructure necessary to build a high-throughput NGS- based carrier screen includes automation, extensive sequencing capacity and a multifaceted bioinformatics solution that allows for batch analysis, export and reporting. Data on over 150,000 individuals will be presented.

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

1:15 Session Break

PRENATAL DIAGNOSTICS IN THE CLINIC

2:10 Chairperson's Remarks

Ignatia B. Van den Veyver, M.D., Professor, Obstetrics & Gynecology and Molecular Human Genetics, Baylor College of Medicine

2:15 Delivering a Prenatal Diagnosis of Down Syndrome: Lessons Learned from Evidence-Based Literature

Brian Skotko, M.D., MPP, Co-Director, Down Syndrome Program, Massachusetts General Hospital

In this presentation, Dr. Brian Skotko reviews the evidence-based research on how physicians can effectively deliver a prenatal diagnosis of Down syndrome. When should the diagnosis be given? How should the news be best delivered? Who should convey the information? What should be mentioned in that initial conversation? The presentation includes highlights from his publications in Pediatrics, American Journal of Obstetrics and Gynecology, and American Journal of Medical Genetics.

2:45 Meeting the Balance of Supply and Demand of Genetic Counselors

Katie Stoll, MS, LGC, Director, Clinical Services, Genetic Support Foundation

Genetic counselors are instrumental to the successful application of new genetic technologies into clinical practice. They help to ensure that genetic testing is used appropriately and also translate complex results into meaningful information for patients and other healthcare providers. The dramatic expansion of genetic testing technology has created many new opportunities for genetic counselors and has also created workforce challenges. In this presentation, we will consider the changing employment landscape of genetic counselors and the implications for genetic services. Alternative delivery models and innovative tools for supporting patient education and informed decision making will also be presented.

BIOMARKERS FOR PREECLAMPSIA

3:15 Integrative Omics in the Study of Preeclampsia

Rachel Kelly, BSc(Hons), MPH, Ph.D., Research Fellow, Channing Division of Network Medicine, Brigham and Women's Hospital Harvard Medical School

Omic technologies including metabolomics, transcriptomics and proteomics represent novel methods for the development of predictive, diagnostic and prognostic biomarkers of preeclampsia, as well as a means of identifying preeclampsia endotypes. Integration of multiple omic-based biomarkers representing different hierarchical stages of the central biological dogma, additionally provides a global systems biology view of the pathogenesis of this disorder. In this talk, we will demonstrate the utility of integrative omic analyses in the study of and management of preeclampsia.

3:45 Refreshment Break in Exhibit Hall with Poster Viewing

CLOSING SESSION

4:30 Closing Panel: Predicting the Landscape for Prenatal Molecular Diagnostics: The Next Few Years

There are a number of advancements that the prenatal field will pursue: cell-based NIPT, NIPT for microdeletions, biomarkers for preeclampsia and preterm birth, and ultimately patient and physician education. This panel will discuss future directions for the field and potential directions for these areas.

5:30 Close of Advances in Prenatal Molecular Diagnostics

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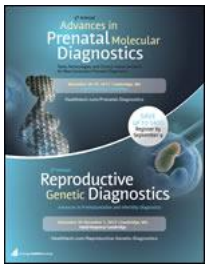

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