FOURTH INTERNATIONAL

Molecular Diagnostics EUROPE

4-7 APRIL 2016

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LISBON, PORTUGAL

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CAMBRIDGE HEALTHTECH INSTITUTE'S FOURTH INTERNATIONAL

Molecular Diagnostics EUROPE

4-7 APRIL 2016 | SHERATON LISBOA HOTEL & SPA | LISBON, PORTUGAL

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Third Annual

Advances in Prenatal Molecular Diagnostics

Trends, Advances & Prospects

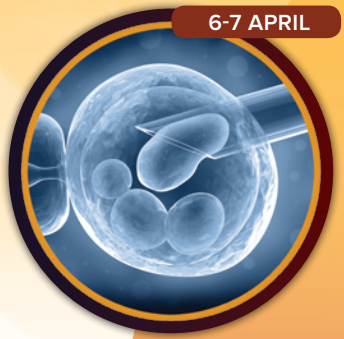
4-6 APRIL

Coverage Includes

- Ethical Issues in Prenatal Diagnostics
- Trends in Analysis of Invasively-Obtained Samples
- Clinical Implementation of Cell-Free DNA Tests
- Assays Based on Arrays, Sequencing and PCR
- Detection of Aneuploidies, Microdeletions and Monogenic Disorders
- The Commercial Potential of DNA from Isolated Fetal Cells

KEYNOTE PRESENTATION
Aim and Scope of Prenatal Screening: The Autonomy Paradigm and Its Limits
Wybo Dondorp, Ph.D., Associate Professor, Biomedical Ethics, Health, Ethics & Society, Maastricht University, The Netherlands

6-7 APRIL



Inaugural

Reproductive Genetic Diagnostics

Advances in Carrier Screening, Preimplantation Diagnostics, and POC Testing

Coverage Includes

- Next-Generation Sequencing for PGD and PGS
- Novel Diagnostic Approaches
- Prospects for Non-Invasive Diagnostic Methods
- Embryo Preparation, Treatment, and Assessment
- Ethical Implications of Advanced Testing Technologies

KEYNOTE PRESENTATION
25 Years of Chromosomal PGD and Counting
Darren K. Griffin, Ph.D., D.Sc., FIBiol, FRCPath, FRSA, Professor, Genetics, School of Biosciences, University of Kent, United Kingdom

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CONFERENCE AT-A-GLANCE

	Monday, 4 April	Tuesday, 5 April	Wednesday, 6 April	Thursday, 7 April
AM				
PM	Advances in Prenatal Molecular Diagnostics		Reproductive Genetic Diagnostics	
AM		Advanced Diagnostics for Infectious Disease		
PM			Point-of-Care Diagnostics	
AM		Circulating Tumour Cells		
PM			Circulating Cell-Free DNA	

About the Event

Novel molecular-based tools are rapidly entering the clinic and creating a new paradigm in healthcare. The **Fourth International Molecular Diagnostics Europe** event will return to Lisbon this spring and feature six tracks: *Prenatal Molecular Diagnostics*, *Reproductive Genetic Diagnostics*, *Circulating Tumour Cells*, *Circulating Cell-Free DNA*, *Advanced Diagnostics for Infectious Disease*, and *Point-of-Care Diagnostics*. As the prenatal diagnostics market has demonstrated, molecular diagnostics are being applied to the clinical setting for greater speed and accuracy of healthcare delivery, while paving the way for a new era in medicine.

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HOTEL & TRAVEL INFORMATION

Conference Hotel:

Sheraton Lisboa Hotel & Spa
Rua Latino Coelho, 1
1069-025 Lisbon, Portugal
Phone: (351)(21) 3120000

Reservations: Go to the travel page of **www.MolecularDxEurope.com**

Discounted Room Rate: €130 single/€150 double, includes breakfast
Discounted Room Rate Cut-off Date: 17 February 2016

Go to the travel page of www.MolecularDxEurope.com for additional info

Why Stay at the Sheraton Lisboa Hotel and Spa?

Be in the Heart of it All - The Sheraton Lisboa Hotel and Spa is located right in the heart of the city!

Get Here Quickly - Located just minutes from Lisbon Airport, it's a quick trip in and out.

Energize - Enjoy a complimentary delicious breakfast, visit the gym, or swim in the pool (weather permitting).

Be Productive - Complimentary wifi in all attendee guest rooms lets you get work done on YOUR time.

Relax and Enjoy - Restaurants, shops and the historic sites of the beautiful city of Lisbon are just a short walk away.

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- Purely social
- Focus group
- Reception style
- Plated dinner with specific conversation focus

Exhibit

Exhibitors will enjoy facilitated networking opportunities with qualified delegates. Speak face-to-face with prospective clients and showcase your latest product, service, or solution.

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Third Annual

Advances in Prenatal Molecular Diagnostics

4-6 April 2016

Trends, Advances & Prospects

MONDAY, 4 APRIL

12:00 – 13:00 Registration

13:00 Chairperson's Opening Remarks

Marta Rodriguez de Alba, Ph.D., Genetics, Fundación Jiménez Díaz, Spain

13:10 KEYNOTE PRESENTATION: AIM AND SCOPE OF PRENATAL SCREENING: THE AUTONOMY PARADIGM AND ITS LIMITS

Wybo Dondorp, Ph.D., Associate Professor, Biomedical Ethics, Health, Ethics & Society, Maastricht University, The Netherlands

In the light of the thrust of the dynamics in prenatal screening towards genome wide & non-invasive testing, the received ethical framework with its emphasis on facilitating autonomous reproductive choice is in need of qualification, taking account of a) the 'paradox of choice', b) the informational privacy interests of the future child and c) the fact that increasingly, prenatal screening will serve prevention-aimed outcomes in addition to reproductive choice.

INVASIVELY-OBTAINED SAMPLES

13:55 Molecular-Cytogenetic Analysis of Invasively-Obtained Samples, Are We Getting More Comfortable with It? The Lab-Side Point of View

Marta Rodriguez de Alba, Ph.D., Genetics, Fundación Jiménez Díaz, Spain

The incorporation of the different emerging technologies applied to the field of prenatal diagnosis has varied throughout Europe. It has been mainly dependent on economic issues but also on the expertise of providers. Since genetic analysis of prenatal samples is usually performed in Services also offering postnatal diagnosis, providers have acquired the expertise testing those samples and therefore started to feel more comfortable with the interpretation of results in prenatal samples. The reasons for referral vary and although many articles support the idea that array-CGH technology should only be applied in specific cases, in many occasions the sample is referred to the Genetic Department with a mild indication and therefore, many concerns arise.

14:25 Prenatal Array Testing – How Unexpected Is the Unexpected?

Margorzata Srebniak, Ph.D., Laboratory Specialist, Clinical Genetics, Erasmus Medical Center, The Netherlands

Microarray is still not widely used in diagnostic laboratories across Europe, although it is already recommended for fetuses with congenital anomalies and may also be done in other cases referred for invasive testing. Unexpected diagnoses and incidental findings (in parental samples) are arguments against microarray especially in pregnancies without ultrasound anomalies. We would like to show all unexpected diagnoses as well as incidental findings that we encountered since 2010, to show that whole genome analysis with array may be favorable in prenatal diagnosis and to answer the question how unexpected is the unexpected.

14:55 Refreshment Break

15:30 Why Should We Implement aCGH as the First-Tier Test in All Invasive Prenatal Samples?

Julian Nevado, Ph.D., MBA, Genetics, Hospital Universitario La Paz (IdiPAZ), Spain

We evaluated the effectiveness of whole-genome array comparative genomic hybridization (aCGH) in prenatal diagnosis in a routine genetic laboratory. Array CGH was performed on 315 samples recruited prospectively as the first-tier test study during the last 3 years. In addition, 77 prenatal samples with abnormal fetal ultrasound findings found to have normal karyotypes were analyzed as a retrospective study using a custom Agilent-based 60K oligonucleotide array. In both cases, aCGH offered higher yields than conventional karyotype. Thus, for cost-effectiveness reasons among others that we discussed, we proposed aCGH in all invasive prenatal diagnosis as a first-tier test in combination with QFPCR to exclude common aneuploidies, triploidies and maternal cell contamination.

16:00 Invasive Confirmatory Procedure after a High-Risk cfDNA Test Result: May the Type of Detected Chromosomal Abnormality Influence the Choice of Diagnostic Procedure?

Francesca Romana Grati, Ph.D., Director, R&D, TOMA Advanced Biomedical Assays SpA, Italy

Cell-free DNA (cfDNA) screening tests use cell-free fetal DNA sequences isolated from maternal blood samples to evaluate the presence of fetal chromosome anueploidies and microdeletions. Currently, there is debate about the most appropriate confirmatory invasive method. The present study is aimed to discuss about the criteria for choice of i) the diagnostic invasive procedure basing on the chromosome specific likelihood of fetoplacental mosaicism and of ii) the laboratory diagnostic test that should be applied on invasively collected sample/s basing on the type of chromosome abnormality for which the cfDNA testing provided a high risk result.

16:30 Roundtable Breakout Discussions

Topics to be covered:

- Challenges of Ethics and Genetic Counseling
- Trends with Analysis of Invasively-Obtained Samples
- Clinical Implementation Issues with Cell-Free DNA Testing
- Testing beyond Common Aneuploidies
- Commercialization Potential for Isolated Fetal Cells
- Biomarkers for Preeclampsia and Pre-Term Labor

17:30 Close of Day One

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TUESDAY, 5 APRIL

NON-INVASIVE TESTING BASED ON CELL-FREE DNA

8:00 Registration and Morning Coffee

8:45 Chairperson's Remarks
Brigitte Faas, Ph.D., Human Genetics, Radbound University, The Netherlands

8:50 The Role of NGS in Reproductive Care
Brigitte Faas, Ph.D., Human Genetics, Radbound University, The Netherlands
In many laboratories and countries, the next generation sequencing (NGS) technique has already been implemented in the routine postnatal setting, by means of exome sequencing. With the introduction of Noninvasive Prenatal Testing (NIPT), NGS has also found its way to the prenatal setting. However, the widespread introduction of NGS in preconceptional and prenatal care is still very challenging. This presentation will give an overview of the advances of NGS application in reproductive care and discuss the challenges it goes along with.

9:20 Cell-Free DNA as a First Line Test vs. Contingent Screening: Perspective from Patients
Dick Oepkes, Professor, Obstetrics and Fetal Therapy, Obstetrics, Leiden University Medical Center, The Netherlands
Screening for chromosomal abnormalities in the fetus is a service to (anxious) pregnant women. For professionals, optimal quality is often equal to highest test performance. For patients, or actually pregnant women since they are not ill themselves, safety, short waiting time and avoidance of living in uncertainty for weeks are equally important. Although on paper, contingent screening may have benefits in terms of reasonably high accuracy against reasonable costs, the prolonged waiting time and especially the 'intermediate' bad news message that further testing is needed before we can reassur

9:50 Development and Validation of a Novel Whole Genome Sequencing Pipeline for Non-Invasive Prenatal Detection of Fetal Submicroscopic Chromosome Anomalies
Lingqian Wu, M.D., Ph.D., Deputy Director, State Key Laboratory of Medical Genetics of China, Central South University, China
To expand the clinical utility of NIPT beyond common chromosomal aneuploidies, we developed a PCR free whole genome sequencing NIPT pipeline and applied to 1500 prospective NIPT samples for clinical validation. We concluded that our novel method was reliable and accurate for detection of a range of fetal and maternal microdeletion/microduplication CNVs varying in size and genomic location.

10:05 Presentation to be Announced
Sponsored by CARTAGENIA

10:20 Implementation of Seraseq™ Aneuploidy Reference Materials for Non-Invasive Prenatal Screening
Sponsored by seracare
Ram Santhanam, MBA, Director, Market Development, Reproductive Health, SeraCare Life Sciences, United States

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

11:15 VENDOR PANEL DISCUSSION: Cell-Free DNA
Alex Helm, Product Manager, Illumina, United States
Solomon Moshkevich, Vice President, Product & Strategy, Natera, United States
Maximillian Schmid, M.D., Associate Director, Medical Affairs, Ariosa, United States
Daniel Grosu, M.D., MBA, CMO, Sequenom, Inc., United States
Philippos Patsalis, Ph.D., Provost, Cyprus School of Molecular Medicine and Chief Executive Medical Director, The Cyprus Institute of Neurology & Genetics, NIPD Genetics, Cyprus
Michael Lutz, Ph.D., CEO, LifeCodexx, Germany
Stephen Little, Ph.D., CEO, Premaitha Health, United States

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

13:15 Session Break

14:15 Chairperson's Remarks
Philippos Patsalis, Ph.D., Provost, Cyprus School of Molecular Medicine and Chief Executive Medical Director, The Cyprus Institute of Neurology & Genetics, Cyprus

14:20 Multiplexed Parallel Analysis of Targeted Genomic Regions for Non-Invasive Prenatal Testing
Philippos Patsalis, Ph.D., Provost, Cyprus School of Molecular Medicine and Chief Executive Medical Director, The Cyprus Institute of Neurology & Genetics, Cyprus
A novel targeted assay for the detection of fetal aneuploides of chromosomes 21, 18 and 13 has been developed by NIPD Genetics. It is based on the capture and analysis of selected genomic regions of interest. An advanced fetal fraction estimation and aneuploidy determination algorithm has also been developed. The analytical performance of this assay will be reviewed. The potential impact of this assay as an accurate and cost-effective option for non-invasive aneuplody detection will be discussed.

14:50 Development of an NGS Data Analysis and Algorithms for Prenatal Diagnosis
Myung L. Kim, Ph.D., Senior Vice President, In Vitro Diagnostics, SK Telecom, United States
Our *In Vitro* Diagnostics unit has developed an automated total solution for NGS data analysis. This solution provides a flexible and easily expendable analysis framework that can be used for genetic tests such as non-invasive prenatal tests (NIPT) and cancer panel or for biological annotation of identified mutations. As initial application, we implemented NIPT solution with a novel algorithm for chromosomal aneuploidy detection. In addition, we developed an independent algorithm for calling causal mutations of an X-linked monogenic disorder. Application to patients' data is presented.

15:20 The IONA Test: CE-IVD – Reliable, Simple, Standardised
Sponsored by premaithahealth
Mike Risley, Ph.D., Chief Development Officer, Premaitha
Focusing on the technical aspects of the IONA® test and the background to why the test has been developed using certain innovative methods. These include a dynamic fetal fraction assessment method and the result output of a likelihood ratio, ideal for prenatal screening as it can be combined with prior risks. Additional features include Premaitha Workflow Manager, world class Technical Support and bespoke analysis software.

15:35 Presentation to be Announced
Sponsored by sequenom
Mathias Ehrich, M.D., Senior Vice President, Research & Development, Sequenom, Inc., United States

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

16:30 Advances in the Non-Invasive Prenatal Diagnosis of Mendelian Disorders using Digital-PCR

Ana Bustamante Aragones, Ph.D., Assistant Head, Genetics, Fundación Jiménez Díaz, Spain

Non-invasive prenatal diagnosis (NIPD) based on the analysis of maternal blood is currently offered worldwide in prenatal diagnosis units. However, to date NIPD for Mendelian disorders is only being offered for paternal and *de novo* mutation exclusion and the study of the maternal inheritance remains challenging. This work shows a validation study of digital PCR (ddPCR) technology for the analysis of both paternally and maternally inherited fetal alleles.

17:00 Bringing NIPT to the Next Level: Detection of Fetal Trisomy based on Quantitative Real-Time PCR

Sponsored by lifecodexx

Michael Lutz, CEO, LifeCodexx, Germany

We will present data of the PrenaTest® based on a quantitative real-time PCR (qPCR) for the determination of fetal trisomy 21. From a total of 261 samples all 17 samples positive with trisomy 21 were correctly classified, resulting in a sensitivity and specificity of the new assay of 100%. While qPCR presents a more cost-efficient solution over NGS, the new assay will also be able to provide results in 72 hours or less.

17:30 Genomic Signatures Associated with Spontaneous Preterm Birth

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

Molecular markers associated with spontaneous premature birth (<37 weeks gestation) have been difficult to identify owing to heterogeneous clinical presentations and a multiplicity of pathways that regulate parturition. We analyzed genetic, molecular, and clinical data of expectant families to identify markers for longitudinal prenatal analysis and risk prediction using a big data machine learning analytics approach. Preterm birth was found to be associated with multiple markers and risk factors, which are potentially useful to predict gestational duration.

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

19:00 Close of Day Two

WEDNESDAY, 6 APRIL

FETAL CELL ISOLATION AND ANALYSIS

8:00 Registration and Morning Coffee

8:40 Chairperson's Remarks

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

8:45 Advances in the Use of Trophoblastic Cells for Prenatal Non-Invasive Diagnostics of Genetic Disorders

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

Non-Invasive Prenatal Diagnosis is technically bound to the challenge of analyzing rare fetal DNA sequences, extracted from blood along with maternal DNA sequences, or rare trophoblastic cells, extracted from blood or cervical samples along with maternal cells. Our results show that ISET allows the consistent recovery of trophoblastic cells from blood and cervical samples and the feasibility of using the trophoblast-derived fetal DNA for noninvasive prenatal diagnosis (NIPND). The advantages and limitations of using fetal cells versus those of using cell-free DNA for developing non-invasive prenatal diagnostic tests will also be discussed.

9:15 Recent Advances in the Enrichment and Characterization of Fetal Cells in Maternal Blood

Steen Kolvraa, Ph.D., CSO, ARCEDI Biotech, Denmark

Using expression array data, we have previously indicated that a major fraction of the fetal cells circulating in the blood of pregnant women are endovascular trophoblasts. Based on this, we had developed a method for the isolation of these fetal cells with the aim of doing cell-based NIPT. We have since then refined our procedure resulting in both higher fetal cell yield and better fetal cell specificity. These new results will be presented and discussed. Fetal DNA from circulating fetal cells is most likely of higher quality than free circulating fetal DNA and may therefore enable detection of more sub-chromosomal aberrations than NIPT based on circulating free fetal DNA. Knowledge on the present status on cell-based NIPT will also be presented.

9:45 Progress in Isolation and Analysis of Fetal Nucleated Red Blood Cells

Leonard Kellner, M.S., President, KellBenx, Inc., United States

Prenatal screening and diagnostics have changed forever. Karyotype (55 years) and MSS (35 years), since they were introduced, are feeling the pressures of the introduction of microarrays, next-gen and newer sequencing. The performance and reduced risk from non-invasive tests account for a significant drop in the level of invasive diagnostics. NIPT alone or as a reflex test in combination with MSS are being accepted around the world. With techniques used in pre-implantation genetics, and the ability to isolate fNRBC; the elusive diagnostic test using fetal cells may soon be realized.

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

10:45 Image-Based Single Cell Sorting to Identify and Recover Fetal Cells Using the Deparray Platform

Farideh Bishoff, Ph.D., Executive Director, Scientific Affairs, Silicon Biosystems, United States

DEPArray™ is an innovative technology platform capable of sorting and isolating 100% pure single or pooled cells through a digitally controlled Dielectrophoretic field using a semiconductor chip. Single target cells can be isolated from enriched blood while pools of tens to hundreds of pure cells can be recovered from fixed tissue blocks. Thus, the DEPArray offers the potential for pre-analytical cell-type purification for downstream molecular analysis, which is a major step forward for precision medicine in oncology and prenatal genetics. Applications for recovery of fetal cells from maternal blood, placental tissue (to address heterogeneity and confined mosaicism) and products of conception will be addressed.

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

Marta Rodriguez de Alba, Ph.D., Genetics, Fundación Jiménez Díaz, Spain
Brigitte Faas, Ph.D., Human Genetics, Radbound University, The Netherlands
Wybo Dondorp, Ph.D., Associate Professor, Biomedical Ethics, Health, Ethics & Society, Maastricht University, The Netherlands
Patrizia Paterlini-Brechot, M.D., Ph.D., Professor, Cellular and Molecular Biology, University of Paris Descartes, France

11:45 Close of Conference

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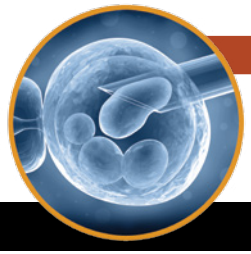

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Reproductive Genetic Diagnostics

6-7 April 2016

Advances in Carrier Screening, Preimplantation Diagnostics, and POC Testing

WEDNESDAY, 6 APRIL

12:00 – 13:00 Registration

NEXT-GENERATION SEQUENCING FOR PGD AND PGS

13:00 Chairperson's Opening Remarks
Bert Smeets, Ph.D., Professor, Clinical Genomics, Mitochondrial Diseases, Maastricht University Medical Center, The Netherlands

» 13:05 KEYNOTE PRESENTATION: 25 YEARS OF CHROMOSOMAL PGD AND COUNTING

Darren K. Griffin, Ph.D., D.Sc., FIBiol, FRCPath, FRSA, Professor, Genetics, School of Biosciences, University of Kent, United Kingdom

The talk will chronicle the chromosomal aspects of PGS and its early beginnings, to clinical applications in the last 25+ years. The rise, fall, and rise again of PGS will be covered from a scientific perspective with many a cautionary tale along the way, and the presentation will conclude with the implementation of novel technologies (such as Karyomapping) for universal PGD.

13:35 Shallow Whole Genome Sequencing is Well Suited for the Detection of Chromosomal Aberrations in Human Blastocysts
Björn Menten, Ph.D., Center for Medical Genetics, Ghent University, Ghent University Hospital, Belgium

Recent advances in *in vitro* fertilization techniques such as vitrification and trophoblast biopsy, as well as the advent of massive parallel sequencing, open up new possibilities for better preimplantation genetic diagnosis and screening. I will discuss the benefits of day 5 biopsy combined with next-generation sequencing for the detection of aneuploidy and smaller copy number aberrations in human embryos.

14:05 Evaluating PGS in the Laboratory
Sebastiaan Mastenbroek, Center for Reproductive Medicine, Academic Medical Center, University of Amsterdam, The Netherlands

Rating quality of evidence and grading strength of recommendations are cornerstones of evidence-based medicine. Careful analysis of currently available trials on PGS shows that there is not enough evidence to justify the current use of PGS in routine clinical practice.

14:35 Refreshment Break in the Exhibit Hall with Poster Viewing

15:15 Genetic Counselling Challenges in a Rapidly Evolving Diagnostic Arena: Spotlight on Preimplantation Genetic Screening (PGS)
Karen A. Sage, MSc, GC, Genetic Service Manager, CARE Fertility; The Bridge Centre; The London Women's Clinic, United Kingdom

The transitions to Next-Generation Sequencing (NGS) and whole genome amplification (WGA) are leading to increased challenges for clinicians at the front line. How do we discuss these unknowns with the patient/couples undergoing this treatment? How do we counsel patients prior to embryo transfer and what are we offering patients post transfer? What is the consensus for PGS embryo transfer and follow up? What are the current guidelines and are they appropriate and current? This talk will illustrate some of the challenges faced by clinicians today offering PGS and PGD in practice.

NOVEL DIAGNOSTIC APPROACHES

15:45 Haplarithmis Enables Both Copy Number Profiling and Haplotyping of Single Cells and Improves Preimplantation Genetic Diagnosis
Joris Vermeesch, Ph.D.Ir, Professor, Molecular Cytogenetics and Genome Research, KU Leuven, Belgium

We developed a novel method, enabling concurrent copy number profiling and haplotyping, which improves preimplantation diagnosis. The approach and analysis pipeline, the validation and results from its clinical implementation will be presented.

16:15 Molecular Diagnostic and Reproductive Challenges in mtDNA Disease
Bert Smeets, Ph.D., Professor, Clinical Genomics, Mitochondrial Diseases, Maastricht University Medical Center, The Netherlands

The mitochondrial DNA (mtDNA) is a circular, maternally transmitted multicopy genome, located within the mitochondria Mutations in the mtDNA are a frequent cause of severe metabolic disorders. This presentation will focus on NGS protocols to identify mtDNA mutations, bioinformatics tools to classify the pathogenicity, and reproductive options, like PGD, to prevent the transmission of mtDNA diseases to future generations.

16:45 Close of Day One

THURSDAY, 7 APRIL

PROSPECTS FOR NON-INVASIVE DIAGNOSTIC METHODS

8:00 Registration

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

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9:00 Chairperson's Remarks
Darren K. Griffin, Ph.D., D.Sc., FIBiol, FRCPath, FRSA, Professor, Genetics, School of Biosciences, University of Kent, United Kingdom

9:05 Preimplantation Genetic Diagnosis and Screening: Now and the Future
Simone Palini, Ph.D., Senior Clinical Embryologist, Lab Director, IVF Unit, "Cervesi" Hospital Cattolica, Italy
Recent works describe the possibility of a non-invasive diagnosis on the blastocoele fluid and culture media, but its use has yet to be demonstrated in the clinic. This talk will discuss the use of PGS for the detection of aneuploid embryos, due to the presence of mosaicism in the embryo, and to discuss the limits that label an embryo as healthy. The future goal is to complement PGD and PGS with the analysis of other non-invasive matrices.

9:35 The Potential of Extracellular Embryo DNA for Preimplantation Genetic Testing
Luca Galluzzi, Ph.D., Research Fellow, Biomolecular Sciences, School of Biotechnology, University of Urbino, Italy
Preimplantation genetic diagnosis and screening currently rely on biopsy of one or few embryo cells. To avoid or limit this invasive procedure, the presence of embryo genomic DNA has been evaluated in extracellular matrices such as blastocoele fluid and embryo culture medium. The potential use of this extracellular DNA in genotyping applications has been then investigated.

10:05 Sponsored Presentation *(Opportunity Available)*

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

11:15 Novel Correlates of Embryo Viability
Gabor L. Kovacs, M.D., Ph.D., DSc, Professor, Laboratory Medicine, Szentágothai Research Centre of the University of Pécs, Hungary
A novel polypeptide marker was found to differentiate between viable and nonviable embryos during *in vitro* fertilization. This molecule was identified with MS as the α-1 fragment of human haptoglobin. Further questions are if there is any correlation between the amount of the haptoglobin fragment in spent embryo culture medium with embryo morphology and cell free nucleic acid release into the medium.

EMBRYO PREPARATION, TREATMENT, AND ASSESSMENT

11:45 How the *in vitro* Environment Shapes Human Preimplantation Embryo Development
Sjoerd Repping, Head, Center for Reproductive Medicine, Academic Medical Center, University of Amsterdam, The Netherlands
During medically assisted reproduction, preimplantation embryos are exposed to an *in vitro* environment. This talk will shed light on how this environment affects the competence and development of human embryos, how these effects are mediated and what the potential consequences of these effects are for MAR success rates and health of offspring.

12:15 Sponsored Presentation *(Opportunity Available)*

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

13:15 Session Break

14:15 Dessert Break in the Exhibit Hall with Poster Viewing

14:55 Chairperson's Remarks
Simone Palini, Ph.D., Senior Clinical Embryologist, Lab Director, IVF Unit, "Cervesi" Hospital Cattolica, Italy

15:00 Embryo Development with Regard to Its Chromosomal Status
Olga Chaplia, Embryologist, Cytogeneticist, IVF Laboratory, Medical Centre Reproductive Genetics Clinic Victoria, Ukraine
As genetic component of embryo significantly affects its implantation capacity, certain developmental patterns may reflect the chromosome status of embryo. The use of proper morphologic criteria indirectly advances selection of euploid embryos for transfer if genetic testing is not feasible.

15:30 Time-Lapse and PGS: Better Together?
Belén Ramos, Ph.D., Clinical Embryologist, IVF Spain – Alicante, Spain
Some groups suggest markers such as mitochondrial DNA levels in order to select the most viable embryo among the euploid ones. In our PGS program,

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we hypothesized that time-lapse could be as an additional viability marker to the euploidy in order to increase implantation. This talk will discuss our findings in a study focused on morphokinetic parameters, including P2 and P3, and how they correlate to blastocyst formation, euploidy, and implantation rates.

16:00 Vitrification of Human Embryos and Blastocysts: As Clear as a Glass

Etienne van Den Abbeel, Ph.D., Professor, Ghent University, IVF Laboratory Director, Centre for Reproductive Medicine, University Hospital Ghent, Belgium

The aim of the presentation will be to review the current status of the vitrification of biopsied and non-biopsied human blastocysts and to discuss the advantages of vitrification techniques and extended culture in preimplantation genetic diagnosis and screening improving the chances of achieving a viable pregnancy, not only free of undesired single-gene defects but also aneuploidy.

16:30 Sponsored Presentation *(Opportunity Available)*

17:00 Refreshment Break

ETHICAL IMPLICATIONS OF ADVANCED TESTING TECHNOLOGIES

17:15 Reproductive Genetic Testing: Dynamics and Ethics

Guido de Wert, Ph.D., Professor, Biomedical Ethics, Faculty of Health, Medicine and Life Sciences, Department of Health, Ethics & Society, Maastricht University, The Netherlands

Reproductive genetic testing, including both screening and diagnosis, and applied in the context of preconception care, medically assisted reproduction and during pregnancy, may help to avoid serious harms and contribute to human welfare. At the same time, such testing raises substantive and procedural normative questions. What are the proper aims of such testing in these different contexts? Which ethical principles and ethical frameworks should be guiding? And how to handle possible conflicts between different stakeholders?

17:45 Panel Discussion: The Future of Preimplantation Genetic Diagnostics and Screening: Ethical Implications and Future Prospects

Moderator: György Kosztolányi, Ph.D., President, Human Reproduction Committee of Scientific Health Council, Professor Emeritus, University of Pécs, Hungary

Panelists: Sjoerd Repping, Head, Center for Reproductive Medicine, Academic Medical Center, University of Amsterdam, The Netherlands

Juliet Tizzard, Director, Strategy and Corporate Affairs, Human Fertilisation & Embryology Authority, United Kingdom

Diagnostic tools and technologies are rapidly evolving across the world, changing and challenging the way reproductive genetic diagnosis and screening are performed. We must continue to examine and debate the ethics, efficacy, and implications of preimplantation genetic diagnosis and screening across the world, as well as keep an eye toward regulatory challenges across Europe and the world.

18:30 Close of Conference

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Monday-Wednesday, 4-6 April	Wednesday-Thursday, 6-7 April
Advances in Prenatal Molecular Diagnostics	Reproductive Genetic Diagnostics
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Advanced Diagnostics for Infectious Disease	Point-of-Care Diagnostics
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