



# Copy Number Variation (CNV)

and genomic alterations  
in health and disease

## PROGRAM

Friday, November 28, 2008

09:00-09:20 *Patrinou GP (Rotterdam, NL)*

*Chrousos GP (Athens, GR)*

Welcome addresses

Session I      Techniques (Moderator: *Patrinou GP, Rotterdam, NL*)

09:20-09:40 *Speicher MR, (Graz, AT)*

Molecular cytogenetics

09:40-10:00 *Ylstra B (Amsterdam, NL)*

Stratification by DNA microarrays in oncology: From technique to therapeutic target

10:00-10:20 *Veltman JA (Nijmegen, NL)*

Diagnostic genome profiling by Single Nucleotide Polymorphism (SNP) arrays

10:20-10:40 *Patsalis PC (Nicosia, CY)*

X chromosome-specific microarrays for targeted locus copy number assessment

Company Lecture      (*Agilent*)

10:40-11:10 *Polten A (Santa Clara, CA, USA)*

Introducing a new high resolution array-CGH platform for CNV detection and discovery: A brief update on technology and applications

COFFEE BREAK

Online REGISTRATION:

<http://www.biomedgen.org/symposia/golden-helix-symposia.html>



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## Session II Chromosomal Disorders (Moderator: *Petersen MB, Athens, GR*)

11:30-11:50 *Perez-Jurado LA (Barcelona, ES)*

**Copy Number Variation: cause and susceptibility factor for recurrent genomic disorders**

11:50-12:10 *Stankiewicz P, (Houston, TX, USA)*

**Copy Number Variation in mental retardation: The Baylor experience**

12:10-12:30 *Zuffardi O (Pavia, IT)*

**Array-CGH in visible chromosome rearrangements**

12:30-12:50 *Knuutila S (Helsinki, FI)*

**Gene copy number alterations in human malignancies**

12:50-13:10 *Romano C (Troina, IT)*

**Clinical profile of patients admitted to array-CGH analysis**

13:10-13:30 *de Vries BB (Nijmegen, NL)*

**New syndromes detected by array-CGH**

## LUNCH BREAK

## Session III Other Disorders (Moderator: *Metaxotou C, Athens, GR*)

14:30-14:50 *Vissers LE (Nijmegen, NL)*

**Molecular karyotyping: linking gene dosage alterations to disease phenotypes**

14:50-15:10 *McCarthy SE (Cold Spring Harbor, NY, USA)*

**Rare structural variants in schizophrenia**

15:10-15:30 *Estivill X (Barcelona, ES)*

**Contribution of genomic structural variation to psoriasis susceptibility**

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Company Lecture      *(ROCHE-NimbleGen)*

15:30-16:00      Fiegler H (*West Sussex, UK*)  
Chromosomes in disarray

## COFFEE BREAK

Session IV      Population Genomics (*Moderator: Antonarakis SE, Geneva, CH*)

16:20-16:40      Redon R (*Cambridge, UK*)  
Copy number variation in the human genome

16:40-17:00      Veltman JA (*Nijmegen, NL*)  
Linking Copy Number Variation to subtle phenotypes in healthy individuals

17:00-17:20      Vermeesch JR (*Leuven, BE*)  
Mendelian Copy Number Variation

17:20-17:40      Dermitzakis ET (*Cambridge, UK*)  
Copy Number Variation and gene expression

## Keynote Lecture (Introduction: Patrinos GP, Rotterdam, NL)

17:45-18:30      Antonarakis SE (*Geneva, CH*)  
The Renaissance of Aneuploidy

## WELCOME RECEPTION

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## EVENING

FORMAL DINNER (Lecturers and moderators: by invitation only)

Dressing code: Smart casual

EVENING FREE (Participants)

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Saturday, November 29, 2008

Session V      Prenatal Diagnosis (*Moderator: Mavrou A, Athens, GR*)

09:00-09:20    *Le Caignec C (Nantes, FR)*

Genomic imbalances in fetuses with multiple malformations

09:20-09:40    *Tonnies H (Kiel, DE)*

Comparative Genomic Hybridization-based analyses of ancient DNA samples of malformed fetuses

09:40-10:00    *Geigl JB (Graz, AT)*

Array-CGH analysis of single cells

Company Lecture      (*Illumina*)

10:00-10:30    *Arnold HP (Eberfing, DE)*

Illumina SNP arrays: Expanding screening capabilities of cytogenetics laboratories

## COFFEE BREAK

Session VI      Quality Control (*Moderator: Kanavakis E, Athens, GR*)

11:00-11:20    *Zollino M (Rome, IT)*

Checklist of clinical signs to select patients for array-CGH and to validate the results

11:20-11:40    *Schoumans J (Stockholm, SE)*

Validation of different platforms

11:40-12:00    *Crolla JA (Salisbury, UK)*

Uniform and evidence based constitutional diagnostic oligonucleotide array-CGH: An International Consortium approach to design, implementation and interpretation

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