# Beyond exome sequencing – causal diversity of genetic diseases
# Arbeitsgemeinschaft für Gen-Diagnostik e.V.

**Program of the Annual AGD Meeting 2014, Potsdam Oct. 10-11**

Friday, October 10

**12:00-13:30** **New tools in genomics – Sponsors' Satellite Session**
Light refreshments and food will be served

*Session Chair:* *Udo Schimmel, Alphametrix Biotech GmbH, Rödermark*

- 12:00-12:15 **Frank Schacherer** (Biobase/QIAGEN, Wolfenbüttel/Hilden)
  "Genomic variant interpretation tools for translational research"
- 12:15-12:30 **Cougar Hao Hu** (Max-Planck Institute for Molecular Genetics, Berlin)
  "SNP-tagging in NGS studies: handling a devil in the details"
- 12:30-12:45 **Inga Nagel** (Universitätsklinikum Schleswig-Holstein, Campus Kiel)
  "Detection of a recurrent 11q-gain/loss-pattern in high malignant MYC-negative Burkitt-like lymphoma using the Affymetrix OncoScan® FFPE Assay Kit"
- 12:45-13:00 **Götz Frommer** (Agilent, Waldbronn)
  "Agilent's Clinical Research and Inherited Disease Panel for NGS"
- 13:00-13:15 **Kathrin Hoffmann** (PerkinElmer, Mainz)
  "Automated electrophoresis - LabChip GX Touch for high and medium throughput"
- 13:15-13:30 **Wieland Keilholz** (NuGEN Technologies, San Carlos, CA)
  "NuGEN's SPET technology: target enrichment system of unparalleled flexibility and performance"

**13:30-14:00** **Coffee Break, Industrial Exhibition and Poster Viewing**

**The following companies' sponsorship is gratefully acknowledged**

Advanced Analytical Technologies GmbH
Affymetrix UK Ltd.
Agilent Technologies Inc.
ATLAS Biolabs GmbH
Covaris Ltd.
Diagenode SA
DNA Genotek Inc.
Illumina GmbH
LGC Genomics GmbH
Life Technologies GmbH
NuGEN Technologies Inc.
PerkinElmer Inc.
QIAGEN GmbH
Steinbrenner Laborsysteme GmbH
Sysmex Suisse AG
TIB MOLBIOL GmbH

---

Venue – SEMINARIS Seehotel, An der Pirschheide 40, 14471 Potsdam, Tel. 0331-9090-0
Registration – Online at http://www.agdev.de/anmeldung.html, on-site registration starts on Friday at 11 am.
Fees – AGD Members 65 €, Non AGD Members 125 €, Students 35 €; late registration surcharge is 25 €.

# Beyond exome sequencing – causal diversity of genetic diseases
# Arbeitsgemeinschaft für Gen-Diagnostik e.V.

**Program of the Annual AGD Meeting 2014, Potsdam Oct. 10-11**

Friday, October 10

**14:00-14:15** **Welcome and Opening of Symposium**

*Peter Nürnberg, President of the AGD*
*Stefan Mundlos, Chair of the Program Committee*

**14:15-16:30** **Session 1 – Mutations in regulatory elements**

*Session Chair:* *Stefan Mundlos, Institute for Medical and Human Genetics, Berlin*

- 14:15-15:00 **Axel Visel** (Lawrence Berkeley National Laboratory, Berkeley, CA)
  "Enhancers in biology and disease"
- 15:00-15:30 **Graham Ritchie** (Wellcome Trust Sanger Institute, Hinxton, Cambridge)
  "Functional annotation of noncoding sequence variants"
- 15:30-16:00 **Lude Franke** (University Medical Center Groningen, The Netherlands)
  "Downstream expression effects of (rare) genetic variants"
- 16:00-16:30 **Michael Eberle** (Illumina Cambridge Ltd, Little Chesterford, Essex)
  "Building a better medical genome"

**16:30-17:00** **Coffee Break, Industrial Exhibition and Poster Viewing**

**17:00-18:30** **Session 2 – Structural variation and disease**

*Session Chair:* *Peter Nürnberg, Cologne Center for Genomics, Köln*

- 17:00-17:30 **Jan Korbel** (EMBL, Heidelberg)
  "Phenotypic impact of genomic structural variation"
- 17:30-18:00 **Francois Spitz** (EMBL, Heidelberg)
  "The architecture of gene expression"
- 18:00-18:30 **Dario Lupianez** (MPI for Molecular Genetics, Berlin)
  "Enhancer shuffling as a disease-causing process"

**18:30-19:30** **Keynote lecture by Michael Skinner**
**(School of Biological Sciences at Washington State University)**
"Epigenetic transgenerational inheritance of disease susceptibility"

**19:30-20:30** **Buffet, Industrial Exhibition and Poster Viewing**

**20:30-21:30** **Concert "...aus alten Märchen winkt es...."**
zauberhafte Klaviermusik zu vier Händen und Duette aus Oper und Oratorium

---

Venue – SEMINARIS Seehotel, An der Pirschheide 40, 14471 Potsdam, Tel. 0331-9090-0
Registration – Online at http://www.agdev.de/anmeldung.html, on-site registration starts on Friday at 11 am.
Fees – AGD Members 65 €, Non AGD Members 125 €, Students 35 €; late registration surcharge is 25 €.

# Beyond exome sequencing – causal diversity of genetic diseases
# Arbeitsgemeinschaft für Gen-Diagnostik e.V.

**Program of the Annual AGD Meeting 2014, Potsdam Oct. 10-11**

Saturday, October 11

**8:30-10:30 Session 3 – Understanding the role of non-coding RNAs for disease**

*Session Chair: Peter Nürnberg, Cologne Center for Genomics, Köln*

- 08:30-09:00 **Andre Franke** (Christian-Albrechts-University, Kiel)
  "The blood-borne miRNome of human disease"
- 09:00-09:30 **Thomas Hansen** (iNANO, Aarhus University, Aarhus)
  "Natural RNA circles"
- 09:30-10:00 **Bernd Matzenbach** (Life Technologies, Darmstadt)
  "A strategy to implement an NGS RNA-panel into clinical labs"
- 10:00-10:30 **Guillaume Durin** (Covaris Ltd., Brighton)
  "Adaptive Focused Acoustics (AFA) technology for superior chromatin shearing"

**10:30-11:00 Coffee Break, Industrial Exhibition and Poster Viewing**

**11:00-13:00 Session 4 – Epigenetic disease etiologies**

*Session Chair: Stefan Mundlos, Institute for Medical and Human Genetics, Berlin*

- 11:00-11:30 **Marc Bleischwitz** (Diagenode SA, Liège)
  "Enabling solutions for reproducible ChIP-sequencing and epigenetic analyses"
- 11:30-12:00 **Melanie Ehrlich** (Tulane Cancer Center, New Orleans, LA)
  "Fine-tuning developmental genes: DNA methylation and hydroxymethylation"
- 12:00-12:30 **Jörn Walter** (University of the Saarland, Saarbrücken)
  "Epigenomics - from sequence to interpretation"
- 12:30-13:00 **Hans van Bokhoven** (Nijmegen Medical Center, Nijmegen)
  "Genetic and epigenetic networks in intellectual disabilities"

**13:00-13:30 Oral presentations of poster prize winners**

**13:30-14:30 Buffet, Industrial Exhibition and Poster Viewing**

**14:30-16:00 AGD General Assembly**

**16:00 End of Meeting**

---

Venue – SEMINARIS Seehotel, An der Pirschheide 40, 14471 Potsdam, Tel. 0331-9090-0
Registration – Online at http://www.agdev.de/anmeldung.html, on-site registration starts on Friday at 11 am.
Fees – AGD Members 65 €, Non AGD Members 125 €, Students 35 €; late registration surcharge is 25 €.

# Beyond exome sequencing – causal diversity of genetic diseases

## Arbeitsgemeinschaft für Gen-Diagnostik e.V.

**Program of the Annual AGD Meeting 2014, Potsdam Oct. 10-11**



Venue – SEMINARIS Seehotel, An der Pirschheide 40, 14471 Potsdam, Tel. 0331-9090-0
Registration – Online at http://www.agdev.de/anmeldung.html, on-site registration starts on Friday at 11 am.
Fees – AGD Members 65 €, Non AGD Members 125 €, Students 35 €; late registration surcharge is 25 €.